東北メディカル・メガバンク計画　バイオバンク利用による主要業績リスト

作成日：2025 年 12 月 1 日

種類		新規件数 2025/5/31 -2025/10/1		これまでの件数 2015 – 2025/5/30		合計
		件数	文献番号	件数	文献番号	
原著論文（利用した試料・情報別）	1.　ゲノム情報					
	1_1. 頻度情報	68 件	(1-68)	1157 件	(69--1225)	1225 件
	1_2. 標準ゲノム	0 件		11 件	(760, 779, 874, 962, 1226-1232)	11 件
	1_3. 全ゲノム	2 件	(37, 1233)	8 件	(660, 891, 1234-1239)	10 件
	1_4. SNP Array データ					
	1_4_1. Japonica Array	4 件	(1240-1243)	112 件	(131, 148, 149, 161, 195, 240, 264, 413, 416, 534, 660, 666, 678, 860, 929, 974, 1040, 1059, 1235-1239, 1244-1332)	116 件
	1_4_2. OmniExpressExome	0 件		10 件	(660, 1298, 1333-1340)	10 件
	1_5. iMETHYL	1 件	(1341)	38 件	(666, 1342-1378)	39 件
	1_6. その他のゲノム	0 件		1 件	(1379)	1 件
	2.　メタゲノム	0 件		1 件	(1380)	1 件
	3.　メタボローム情報	2 件	(1381-1382)	7 件	(534, 1312,1383-1387)	9 件
	4.　GWAS レポジトリ	0 件		5 件	(1298, 1388-1391)	5 件
	4. 試料					
	4_1. 細胞	0 件		1 件	(198)	1 件
	4_2. 血清・血漿	1 件	(1392)	3 件	(1393-1395)	4 件
	4_3. DNA	0 件		4 件	(1295, 1379, 1396, 1397)	4 件
	4_4. 母乳	0 件		1 件	(1398)	1 件
	5. 健康調査情報					
	5_1. 調査票・検体検査情報等	4 件	(37, 1233, 1392, 1399)	40 件	(660, 860, 891, 974, 1040, 1239, 1281, 1295, 1298, 1312, 1313, 1325, 1379, 1385, 1386, 1395, 1396, 1398, 1400-1421)	44 件
	5_2. MRI 検査情報	0 件		1 件	(1414)	1 件
	6. 施設、技術・方法、データベース	1 件	(1422)	27 件	(357, 860, 1423-1447)	28 件
	小計（※重複を除いた件数）	**79** 件		**1368** 件		**1447** 件

知財申請	1. ゲノム情報	0 件		2 件	(1448, 1449)	2 件
	2. 健康調査情報	0 件		2 件	(1449, 1450)	2 件
	3. 血清	0 件		1 件	(1450)	1 件
	小計（※重複を除いた件数）	0 件	—	**3** 件		**3** 件
引用・レビュー・学会抄録等	1. 論文・本での引用	2 件	(1451, 1452)	300 件	(1453-1752)	302 件
	2. レビュー	3 件	(1753-1755)	164 件	(1756-1919)	167 件
	3. 学会抄録	0 件		58 件	(1920-1977)	58 件
	4. 学位論文	4 件	(1978-1981)	33 件	(1982-2014)	37 件
	小計（※重複を除いた件数）	**9** 件		**555** 件		**564** 件
合計（※重複を除いた件数）		**88** 件	—	**1926** 件	—	**2014** 件

分譲利用による成果

種類		新規件数 2025/5/31 -2025/10/1		これまでの件数 – 2025/5/30		合計
		件数	文献番号	件数	文献番号	
原著論文（利用した試料・情報別）	ゲノム情報					
	頻度情報	0 件		2 件	(660, 974)	2 件
	全ゲノムデータ	0 件		2 件	(660, 1239)	2 件
	SNP Array データ	0 件		4 件	(660, 974, 1239, 1295)	4 件
	その他のゲノムデータ （ミトコンドリアゲノム）	0 件		1 件	(1379)	1 件
	メタボローム情報	0 件		2 件	(1385, 1386)	2 件
	健康調査情報					
	調査票情報、検体検査情報、特定健診情報、生理機能検査情報	1 件	(1392)	17 件	(660, 974, 1239, 1295, 1379, 1385, 1386, 1395, 1396, 1402-1404, 1411, 1414. 1416, 1418-1420)	18 件
	MRI 検査情報	0 件		1 件	(1414)	1 件
	試料:血清	1 件	(1392)	1 件	(1395)	2 件
	試料:DNA	0 件		3 件	(1295, 1379, 1396)	3 件
	小計（※重複を除いた件数）	**1** 件		**18** 件		**19** 件
知財申請	ゲノム情報	0 件		1 件	(1449)	1 件
	健康調査情報	0 件		2 件	(1449, 1450)	2 件
	試料：血清	0 件		1 件	(1450)	1 件
	小計（※重複を除いた件数）	**0** 件	—	**2** 件		**2** 件
合計（※重複を除いた件数）		**1** 件	—	**20** 件	—	**21** 件

検索方法：

① Google Scholar を用いて次のキーワードで検索した。キーワード：1KJPN、2KJPN、3.5KJPN、4.7KJPN、8.3KJPN、14KJPN、38KJPN、54KJPN、iJGVD、Integrative Japanese Genome Variation、JG1、JG2、JRGA、Japonica Array、jMorp、iMethyl、Tohoku Medical Megabank Project、Tohoku Medical Megabank Organization、ToMMo。

② ToMMo 所属の先生によって発表された論文を PubMed でひき、Cited by にリストされた論文を検索した。

③ 分譲申請による利用者から提出された「研究実施経過報告書」に記載されていた発表論文等を検索した。

除外基準： first author、corresponding author もしくは last author が岩手医科大学いわて東北メディカル・メガバンク機構もしくは東北大学東北メディカル・メガバンク機構に所属している論文。

検索結果のなかで、明らかに該当しない論文（例えば、言語名 Tommo So についての論文等）。

■文献（年毎にアルファベット順）

ゲノム頻度情報（1-1225）

1. Hiruma, T., Inoue, S., Ko, T., Nomura, S., Abe, R., Bujo, C. *et al.* (2024) PRKAG2 Syndrome Caused by a Novel Missense Variant Mimicked Sporadic Hypertrophic Cardiomyopathy Until Its Progression to Burned-Out Phase Circ Heart Fail **17**, e012047 10.1161/circheartfailure.124.012047
2. Aida, R., Watanabe, H., Shiiya, T., Sakurazawa, C., Otsuka, T., Goto, S. *et al.* (2025) Dual Renal and Cardiac Phenotypes Associated with Rare Variants Inherited from Both Parents Intern Med 10.2169/internalmedicine.6003-25
3. Ajmeriya, S., Chatterjee, B., andKarmakar, S. (2025) Identification of variants from gene expression data of opioid-addicted patients J Appl Genet 10.1007/s13353-025-00989-x
4. Ando, M., Higuchi, Y., Yuan, J. H., Yoshimura, A., Yano, C., Hobara, T. *et al.* (2025) Nationwide Characterization of MFN2-Related CMT in 176 Japanese Patients: Clinical and Genetic Insights Ann Clin Transl Neurol 10.1002/acn3.70218
5. Aoki, T., Inui, A., Ogata, Y., Igarashi, A., Yanagi, K., Yamamori, M. *et al.* (2025) Functional analysis of novel and recurrent RINT1 variants in patients with infantile liver dysfunction Journal of human genetics 10.1038/s10038-025-01404-3
6. Asagai, Y., Tanaka, Y., Hanafusa, H., Nagano, C., Horinouchi, T., Ishimori, S. *et al.* (2025) Clinical characteristics of patients with SALL1-related disorder Pediatr Nephrol **40**, 3407-3414 10.1007/s00467-025-06878-z
7. Cho, E. H., Lee, M., Ki, C. S., Seol, C. A., andJang, M. A. (2025) Genetic epidemiology of epithelial-stromal TGFBI dystrophies in a large Korean population Sci Rep **15**, 25360 10.1038/s41598-025-08189-7
8. Endo, Y., Koga, T., Mohamed, L., Tsuji, Y., Umeda, M., Hayashi, H. *et al.* (2025) Mediterranean fever gene variants may prevent the development of lupus nephritis in Japanese patients with systemic lupus erythematosus Front Immunol **16**, 1571208 10.3389/fimmu.2025.1571208
9. Fujita, K., Kimura, T., Yamakawa, A., Niida, S., Ozaki, K., Sakurai, T. *et al.* (2025) Genetic background and multidomain interventions in mild cognitive impairment Alzheimers Res Ther **17**, 130 10.1186/s13195-025-01764-0
10. German, H. M., Zaki, M. S., Usmani, M. A., Karagoz, I., Efthymiou, S., Abdel-Hamid, M. S. *et al.* (2025) Comprehensive genotypic, phenotypic, and biochemical characterization of GOT2 deficiency: A progressive neurodevelopmental disorder with epilepsy and abnormal movements Genet Med **27**, 101587 10.1016/j.gim.2025.101587

11. Gotoh, N., Munkh-Erdene, T., Hayashi, A., Kasamatsu, T., Kitamura, M., Kojima, A. *et al.* (2025) Impact of inflammatory cytokine polymorphisms on the progression of myelodysplastic syndromes Ann Hematol **104**, 5111-5123 10.1007/s00277-025-06614-5
12. Hamanaka, K., Fujita, A., Miyatake, S., Misawa, K., Koshimizu, E., Uchiyama, Y. *et al.* (2025) Genome sequencing provides high diagnostic yield and new etiological insights for intellectual disability and developmental delay NPJ Genom Med **10**, 60 10.1038/s41525-025-00521-4
13. Hidaka, K., Hayashi, S., Nakakuki, M., Naruse, S., andIshiguro, H. (2025) Progressive Pulmonary Lesion due to Cystic Fibrosis Transmembrane Conductance Regulator Dysfunction: A Case Study From Japan Cureus **17**, e86774 10.7759/cureus.86774
14. Higa, N., Hayashi, T., Mizobuchi, K., Kuniyoshi, K., Kondo, H., andNakano, T. (2025) A novel deletion-insertion variant of RS1 in X-linked retinoschisis Ophthalmic Genet 1-4 10.1080/13816810.2025.2523473
15. Hiraide, T., Hayashi, T., Yamoto, K., Masunaga, Y., Asahina, M., Ogata, T. *et al.* (2025) An atypical case of macrocephaly and severe intellectual disability associated with a missense variant in the guanine nucleotide exchange factor-1 domain of TRIO Brain Dev **47**, 104405 10.1016/j.braindev.2025.104405
16. Hirano, Y., Shimura, K., Mitani-Konno, M., Fukushima, N., Hasegawa, T., andNarumi, S. (2025) Airway compression in a neonate with thyroid dyshormonogenesis due to SLC26A7 defect Eur J Endocrinol **193**, K16-K21 10.1093/ejendo/lvaf198
17. Hobara, T., Ando, M., Higuchi, Y., Yuan, J. H., Yoshimura, A., Saito, T. *et al.* (2025) Charcot-Marie-Tooth-like presentation in giant axonal neuropathy: clinical variability and prevalence in a large Japanese case series J Neurol **272**, 514 10.1007/s00415-025-13243-5
18. Honda, M., Narumi, S., Hasegawa, K., Goto, Y., Kawashima, Y., Ohara, O. *et al.* (2025) Coproporphyrinogen oxidase deficiency causes primary adrenal insufficiency and 46,XY DSD J Clin Endocrinol Metab 10.1210/clinem/dgaf329
19. Hu, Q., Kimura, Y., Ikeda, S., Tanaka, Y., Nakanoko, T., Ota, M. *et al.* (2025) Circulating tumor DNA monitoring detects minimal residual disease and predicts outcomes in patients with esophageal adenocarcinoma or squamous cell carcinoma after esophagectomy BJC Rep **3**, 52 10.1038/s44276-025-00158-x
20. Igarashi, A., Makita, Y., Kobayashi, N., Abe, Y., Aoki, T., Iida, T. *et al.* (2025) Novel biallelic splicing and deletion variants of ADAMTS3 found in adult patients with Hennekam lymphangiectasia-lymphedema syndrome 3 BMJ Connections Clinical Genetics and Genomics **2**, 10.1136/bmjccgg-2024-000006
21. Ishikawa, T., Kimoto, H., Seki, A., Shirai, M., Uto, K., Makiyama, T. *et al.* (2025) Phenotypic spectrum of cardiac conduction disturbance and cardiomyopathy linked to titin canonical splice-site variants Cardiovasc Res **121**, 1712-1721 10.1093/cvr/cvaf135
22. Jang, M. A., Heo, W. Y., Lee, J. K., Kim, J. W., Kim, S. M., Jang, J. H. *et al.* (2025) Carrier Frequency and Prevalence of Citrin Deficiency in East Asians and Koreans Based on Comprehensive Analysis of Pathogenic SLC25A13 Variants Ann Lab Med **45**, 530-538 10.3343/alm.2024.0631
23. Joo, S. Y., Jang, S. H., Kim, J. A., Kim, S. J., Choi, J. Y., Jung, J. *et al.* (2025) Leveraging underrepresented population data improves interpretation of genetic variants associated with hearing loss Sci Rep **15**, 33693 10.1038/s41598-025-18852-8
24. Kakumoto, T., Matsukawa, T., Tokimura, R., Tsuboyama, Y., Hayashi, Y., Mitsutake, A. *et al.* (2025) Vanishing White Matter Disease With EIF2B2 c.254 >A Variant: Mild Clinical and MRI Findings Neurol Genet **11**, e200293 10.1212/NXG.0000000000200293

25. Kakuta, R., Takemoto, A., Tanimoto, K., Noji, R., Kudo, R., Kano, Y. *et al.* (2025) Characterization of a novel comprehensive genomic profiling test with better detection of heterozygous deletions and RNA-based gene fusion analysis Oncologist **30**, oyaf056 10.1093/oncolo/oyaf056
26. Kamatani, T., Umeda, K., Iwasawa, T., Miya, F., Matsumoto, K., Mikami, S. *et al.* (2025) Clonal diversity shapes the tumour microenvironment leading to distinct immunotherapy responses in metastatic urothelial carcinoma Nat Commun **16**, 7995 10.1038/s41467-025-63309-1
27. Kashimada, A., Mizuno, T., Tanaka, E., Hosokawa, S., Udagawa, T., Hiraoka, Y. *et al.* (2025) De novo variant in RING finger protein 213 causes systemic vasculopathy JCI Insight **10**, 10.1172/jci.insight.190094
28. Katoh-Fukui, Y., Saito, D., Narumi, H., Hattori, A., Igarashi, M., Uehara, E. *et al.* (2025) DHX37 variants in patients with 46,XY disorders or differences of sex development Hum Genome Var **12**, 18 10.1038/s41439-025-00322-2
29. Kawamoto, R., Mori, T., Chiga, M., Fujimaru, T., Nanamatsu, A., Fujiki, T. *et al.* (2025) Importance of family history and health checkup for school-aged children for type IV collagen-associated nephropathy in hereditary kidney disease J Nephrol **38**, 1909-1916 10.1007/s40620-025-02355-w
30. Kido, J., Mizukami, T., Misumi, Y., Sugawara, K., Kusunoki, S., Nishimura, N. *et al.* (2025) Osteogenesis imperfecta, intellectual disability and recurrent infections in a male with a pathogenic SASH3 variant Hum Genome Var **12**, 19 10.1038/s41439-025-00323-1
31. Kim, J., Cho, S., Jeong, K. H., Ha, W. S., Kim, K. M., Chu, M. K. *et al.* (2025) TMEM14A Gene Affects Hippocampal Sclerosis in Mesial Temporal Lobe Epilepsy J Clin Med **14**, 3810 10.3390/jcm14113810
32. Makutani, Y., Iwamoto, M., Daito, K., Tokoro, T., Ueda, K., andKawamura, J. (2025) Two cases of genetic testing for familial adenomatous polyposis without a family history Int Cancer Conf J 1-9 10.1007/s13691-025-00782-x
33. Matsunaga, M., Maruyama, S., Nakamura, T., Yonee, C., Yonekawa, T., Higuchi, Y. *et al.* (2025) Early onset respiratory failure in two pairs of unrelated Japanese siblings with SELENON-related myopathy Brain and Development Case Reports **3**, 100099 10.1016/j.bdcasr.2025.100099
34. Moriura, Y., Nishio, Y., Ichimura, S., Noda, H., Tanahashi, Y., Yamamoto, H. *et al.* (2025) De novo CDKN1C variant in Beckwith-Wiedermann spectrum with atypical complications Hum Genome Var **12**, 9 10.1038/s41439-025-00316-0
35. Murase, A., Mishima, H., Aoki, S., Hara, S., Kubiura-Ichimaru, M., Ohba, T. *et al.* (2025) Variants of NLRP genes encoding subcortical maternal complex components are linked to biparental placental mesenchymal dysplasia Hum Genomics **19**, 94 10.1186/s40246-025-00814-w
36. Muto, M., Gohda, T., Takagi, M., Kobayashi, T., Kihara, M., Saeki, H. *et al.* (2025) A Novel PHEX Mutation in X-Linked Hypophosphataemic Rickets With Reduced Expression of NaPi-IIa and NaPi-IIc in the Proximal Tubules: A Case Report Nephrology (Carlton) **30**, e70104 10.1111/nep.70104
37. Nagino, K., Akasaki, Y., Fuse, N., Ogishima, S., Shimizu, A., Uruno, A. *et al.* (2025) Integration of Digital Phenotyping and Genomics for Dry Eye Disease: Protocol for a Prospective Cohort Study JMIR Res Protoc **14**, e67862 10.2196/67862
38. Nakagawa, H., Aoyama, S., Yokobori, J., Furusawa, H., Mitsumura, T., Takemoto, A. *et al.* (2025) A novel fumarate hydratase frameshift mutation in lung adenocarcinoma: A case report Cancer Genet **298-299**, 16-19 10.1016/j.cancergen.2025.08.006
39. Nakamura, K., Sato, Y., Hashimoto, M., Matsumoto, N., Nitta, S., Okazaki, Y. *et al.* (2025) Mitochondrial DNA

variants in normal skins: Insights into prevalent pathogenic variants and quality control surveillance Mitochondrion **85**, 102081 ARTN 102081 10.1016/j.mito.2025.102081

40. Nakashima, D., Tosaki, T., Sasaki, T., Honda, Y., Yokote, S., Mori, T. *et al.* (2025) Combined Alport Syndrome Type 3A and Mitochondrial Disease Presenting with a Thin Base Membrane and Overt Albuminuria: A Case Report Intern Med 10.2169/internalmedicine.5838-25
41. Nishina, S., Torii, K., Ishitani, S., Yoshida, T., Fukami, M., Kurosawa, K. *et al.* (2025) Novel biallelic CDK9 variants are associated with retinal dystrophy without CHARGE-like malformation syndrome Journal of human genetics 10.1038/s10038-025-01395-1
42. Nishioka, M., Tanei, Z. I., Saito, Y., Morishima, M., Hino, M., Mori, K. *et al.* (2025) Mosaic variants of neurodevelopmental and mitochondrial genes in postmortem paraventricular thalamus in bipolar disorder detected by deep exome sequencing Psychiatry and clinical neurosciences **79**, 653-666 10.1111/pcn.13871
43. Odate, T., Kondo, T., Katoh, R., Ito, K., Ueno, T., Yatabe, Y. *et al.* (2025) Progression of Follicular Thyroid Carcinomas to Anaplastic Thyroid Carcinomas: Molecular and Clinicopathologic Characteristics with Comparison to Papillary Thyroid Carcinoma-Derived Anaplastic Thyroid Carcinomas Endocr Pathol **36**, 30 10.1007/s12022-025-09875-y
44. Ogawa, A., Watanabe, S., Ozerova, I., Tsai, A. Y., Kuchitsu, Y., Chong, H. B. *et al.* (2025) Adenosine kinase and ADAL coordinate detoxification of modified adenosines to safeguard metabolism Cell **188**, 6151-6169 e6124 10.1016/j.cell.2025.07.041
45. Okamura, K., Saito, T., Ohori, S., Takada, F., Nomura, T., Oba, E. *et al.* (2025) Deep intronic variant in TYR causes OCA1A in Japanese families through pseudoexon activation J Dermatol Sci **120**, 77-79 10.1016/j.jdermsci.2025.09.003
46. Orimo, K., Matsukawa, T., Shiomi, K., Goto, R., Mitsutake, A., Kuromi, Y. *et al.* (2025) Two Japanese families with adult-onset leukoencephalopathy caused by pathogenic variants in CST3 J Neurol Sci **478**, 123708 10.1016/j.jns.2025.123708
47. Pan, Y., Iejima, D., Yoshitake, K., Tsunoda, K., Iwata, T., andJapan Eye Genetics, C. (2025) Clinical features and molecular mechanisms of RP1L1 variants causing occult macular dystrophy HGG Adv **6**, 100461 10.1016/j.xhgg.2025.100461
48. Saeki, S., Otsuki, J., Enatsu, N., Wei, X. Q., Mukaida, N., Okamoto, R. *et al.* (2025) Association of the RNASE9 c.1+1G>A splice site variant with decreased sperm motility in Japanese men Gene Reports **41**, 102366 ARTN 102366 10.1016/j.genrep.2025.102366
49. Sawada, T., Kido, J., Tsukimura, T., Sugawara, K., Shiga, T., Saito, S. *et al.* (2025) Evaluation of GLA variants detected in newborn screening for Fabry disease using biomarker analysis Mol Genet Metab Rep **44**, 101245 10.1016/j.ymgmr.2025.101245
50. Semba, R., Eguchi, H., Takatsu, M., Hashizume, T., Moteki, H., Maeno, K. *et al.* (2025) Two-hit events occurred independently in bilateral breast cancers in a germline double heterozygous carrier for BRCA1 and BRCA2 Breast Cancer **32**, 1472-1479 10.1007/s12282-025-01740-4
51. Shinkawa, K., Ishii, A., Kimura, T., Toriu, N., Nakagawa, N., Wada, T. *et al.* (2025) A Case of Siblings with End-Stage Kidney Disease and Retinal Degeneration Suggestive of Partial Alstrom Syndrome Nephron 1-11 10.1159/000547625
52. Shinno, K., Kubota, M., Kosaki, R., Kosaki, K., andIshiguro, A. (2025) Atypical gradual progression in rapid-onset dystonia-parkinsonism with a rare ATP1A3 variant and the long-term effectiveness of botulinum toxin therapy

Brain and Development Case Reports **3**, 100092 10.1016/j.bdcasr.2025.100092

53. Shiraishi, K., Tsuburaya, R. S., Mukaida, S., Itomi, S., Kajimoto, S., Yano, N. *et al.* (2025) Morphological erythrocyte abnormalities as potential biomarker of uridine-responsive epileptic encephalopathy Brain and Development Case Reports **3**, 100081 10.1016/j.bdcasr.2025.100081
54. Sonehara, K., Watanabe, R., Matsumura, Y., Mitsui, Y., Ogawa, Y., Odomari, K. *et al.* (2025) Whole-genome sequencing reveals rare and structural variants contributing to psoriasis and identifies CERCAM as a risk gene Cell Genom **5**, 100978 10.1016/j.xgen.2025.100978
55. Sugisawa, C., Uehara, E., Tanase-Nakao, K., Inoue, K., Iida, T., Otsuka, F. *et al.* (2025) Clinical and Genetic Characteristics of Adult Nonautoimmune Hypothyroidism: A Single-Institution Study J Clin Endocrinol Metab 10.1210/clinem/dgaf352
56. Suzuki, E., Shima, H., Ueda, A., Nakabayashi, K., Matsubara, K., Kuroki, Y. *et al.* (2025) De Novo Splice Site Variant of TCF12 in a Boy With Isolated Kallmann Syndrome Case Rep Endocrinol **2025**, 2350842 10.1155/crie/2350842
57. Suzuki, S., Seino, M., Sato, H., Saito, Y., Saito, K., Yamada, Y. *et al.* (2025) Potential Resistance to Oxaliplatin-Based Regimens in Gastric Cancer Patients with ERBB2 R678Q Mutation: Evidence from a National Genomic Database Curr Issues Mol Biol **47**, 10.3390/cimb47060430
58. Takahashi, T., Sawada, K., Yamashita, T., Yamamoto, W., Iijima, Y., Adachi, A. *et al.* (2025) Genetic Profiling Reveals the Distinctions Among MTX-Associated DLBCL, EBV-Positive Mucocutaneous Ulcer, and EBV + DLBCL Cancer Sci **116**, 2306-2316 10.1111/cas.70111
59. Takahashi-Kobayashi, M., Sawai, T., Nagano, C., Kawanishi, K., Nakajima, K., Suzuki, H. *et al.* (2025) De novo C3 Glomerulonephritis in a Kidney Transplant Recipient Associated With a Rare CFH Variant of Unknown Significance Kidney Int Rep **10**, 4094-4097 10.1016/j.ekir.2025.08.035
60. Tanaka, M., Shimomura, M., Ashihara, K., Urayama, K., andKarakawa, S. (2025) Early-Onset Chronic Drug-Induced Cardiomyopathy in a Pediatric Patient With Ewing Sarcoma Cureus **17**, e90840 10.7759/cureus.90840
61. Tanikawa, W., Okamoto, S., Ohara, O., Masunaga, Y., Yamoto, K., Fujisawa, Y. *et al.* (2025) Comprehensive Molecular Studies in 88 Japanese Patients With Congenital Hypogonadotropic Hypogonadism J Clin Endocrinol Metab 10.1210/clinem/dgaf548
62. Uehara, E., Sugihara, K., Hata, I., Akiba, K., Hattori, A., Kageyama, I. *et al.* (2025) KCNJ11 readthrough variant in a patient with congenital hyperinsulinism Clin Pediatr Endocrinol **34**, 260-263 10.1297/cpe.2025-0025
63. Yamamoto, R., Takeshita, Y., Takayama, H., Tsujiguchi, H., Kannon, T., Sato, T. *et al.* (2025) The SELENOP Polymorphism rs7579 Predicts Hepatic Steatosis in Females With Insulin Resistance in the General Population J Endocr Soc **9**, bvaf144 10.1210/jendso/bvaf144
64. Yonamine, M., Koyanagi, R., Ishibashi, N., Aita, Y., andTakekoshi, K. (2025) Germline Multigene Panel Testing in Japanese Patients with Pheochromocytoma and Paraganglioma: Technical Feasibility and Clinical Utility Genet Test Mol Biomarkers **29**, 290-293 10.1177/19450265251382504
65. Yoshiji, S., Hasebe, M., Tanaka, D., Shimizu, M., Soma, Y., Kawaguchi, T. *et al.* (2025) Genetic and Clinical Characteristics of Monogenic Diabetes in Japan: A Nationwide Study by the Japan Diabetes Society J Clin Endocrinol Metab dgaf478 10.1210/clinem/dgaf478
66. Yunoki, T., Matsuoka, C., Osakada, Y., Fukui, Y., Takemoto, M., Morihara, R. *et al.* (2025) Two Cases of Autosomal Recessive Spinocerebellar Ataxia-8 Showing Two Novel Variants of SYNE1 in Japanese Families Intern Med 10.2169/internalmedicine.5602-25

67. Zuo, S., Zhang, K., Dai, J., Fent, K., Huang, A. Y., andZhao, Y. (2025) Genetic Polymorphisms in Nuclear Receptors Drive Variations in Responses to Environmental Exposures Environ Sci Technol **59**, 19733-19744 10.1021/acs.est.5c08097
68. 赤羽智子, 平沢晃, 井本逸勢, 増田健太, 杉森弥生, 荻島大貴 *et al.* (2025) 日本人卵巣癌例のがん遺伝子パネル検査を契機に検出された BRCA1 生殖細胞系列の希少病的 variant 遺伝性腫瘍 **25**, 49-55,
69. Gouda, N., Shiwa, Y., Akashi, M., Yoshikawa, H., Kasahara, K., andFurusawa, M. (2016) Distribution of human single-nucleotide polymorphisms is approximated by the power law and represents a fractal structure Genes Cells **21**, 396-407 10.1111/gtc.12344
70. Hayano, T., Matsui, H., Nakaoka, H., Ohtake, N., Hosomichi, K., Suzuki, K. *et al.* (2016) Germline Variants of Prostate Cancer in Japanese Families PLoS One **11**, e0164233 10.1371/journal.pone.0164233
71. Ichikawa, M., Aiba, T., Ohno, S., Shigemizu, D., Ozawa, J., Sonoda, K. *et al.* (2016) Phenotypic Variability of ANK2 Mutations in Patients With Inherited Primary Arrhythmia Syndromes Circ J **80**, 2435-2442 10.1253/circj.CJ-16-0486
72. Ishizuka, K., Kimura, H., Yoshimi, A., Banno, M., Kushima, I., Uno, Y. *et al.* (2016) Investigation of single-nucleotide variants in MBD5 associated with autism spectrum disorders and schizophrenia phenotypes Nagoya J Med Sci **78**, 465-474 10.18999/nagjms.78.4.465
73. Kawase, K., Sugiura, T., Nagaya, Y., Yamada, T., Sugimoto, M., Ito, K. *et al.* (2016) Single nucleotide polymorphisms in AGTR1, TFAP2B, and TRAF1 are not associated with the incidence of patent ductus arteriosus in Japanese preterm infants Pediatrics international : official journal of the Japan Pediatric Society **58**, 461-466 10.1111/ped.12861
74. Kohda, M., Tokuzawa, Y., Kishita, Y., Nyuzuki, H., Moriyama, Y., Mizuno, Y. *et al.* (2016) A Comprehensive Genomic Analysis Reveals the Genetic Landscape of Mitochondrial Respiratory Chain Complex Deficiencies PLoS Genet **12**, e1005679 10.1371/journal.pgen.1005679
75. Kohmoto, T., Tsuji, A., Morita, K., Naruto, T., Masuda, K., Kashimada, K. *et al.* (2016) A novel COL11A1 missense mutation in siblings with non-ocular Stickler syndrome Hum Genome Var **3**, 16003 10.1038/hgv.2016.3
76. Masamune, A., Nakano, E., Niihori, T., Hamada, S., Nagasaki, M., Aoki, Y. *et al.* (2016) Variants in the UBR1 gene are not associated with chronic pancreatitis in Japan Pancreatology **16**, 814-818 10.1016/j.pan.2016.06.662
77. Mitsui, S. N., Yasue, A., Masuda, K., Naruto, T., Minegishi, Y., Oyadomari, S. *et al.* (2016) Novel human mutation and CRISPR/Cas genome-edited mice reveal the importance of C-terminal domain of MSX1 in tooth and palate development Sci Rep **6**, 38398 10.1038/srep38398
78. Watanabe, M., Hayabuchi, Y., Ono, A., Naruto, T., Horikawa, H., Kohmoto, T. *et al.* (2016) Detection of 1p36 deletion by clinical exome-first diagnostic approach Hum Genome Var **3**, 16006 10.1038/hgv.2016.6
79. Watanabe, M., Nakagawa, R., Naruto, T., Kohmoto, T., Suga, K., Goji, A. *et al.* (2016) A novel missense mutation of COL5A2 in a patient with Ehlers-Danlos syndrome Hum Genome Var **3**, 16030 10.1038/hgv.2016.30
80. Yamaguchi, K., Nagayama, S., Shimizu, E., Komura, M., Yamaguchi, R., Shibuya, T. *et al.* (2016) Reduced expression of APC-1B but not APC-1A by the deletion of promoter 1B is responsible for familial adenomatous polyposis Sci Rep **6**, 26011 10.1038/srep26011
81. Yamaguchi, K., Urakami, K., Nagashima, T., Shimoda, Y., Ohnami, S., Ohnami, S. *et al.* (2016) Prevalence of low-penetrant germline TP53 D49H mutation in Japanese cancer patients Biomed Res **37**, 259-264 10.2220/biomedres.37.259

82. Akiyama, M., Okada, Y., Kanai, M., Takahashi, A., Momozawa, Y., Ikeda, M. *et al.* (2017) Genome-wide association study identifies 112 new loci for body mass index in the Japanese population Nature genetics **49**, 1458-1467 10.1038/ng.3951

83. Ando, M., Hashiguchi, A., Okamoto, Y., Yoshimura, A., Hiramatsu, Y., Yuan, J. *et al.* (2017) Clinical and genetic diversities of Charcot-Marie-Tooth disease with MFN2 mutations in a large case study Journal of the peripheral nervous system : JPNS **22**, 191-199 10.1111/jns.12228

84. Ando, M., Okamoto, Y., Yoshimura, A., Yuan, J. H., Hiramatsu, Y., Higuchi, Y. *et al.* (2017) Clinical and mutational spectrum of Charcot-Marie-Tooth disease type 2Z caused by MORC2 variants in Japan Eur J Neurol **24**, 1274-1282 10.1111/ene.13360

85. Christophersen, I. E., Rienstra, M., Roselli, C., Yin, X., Geelhoed, B., Barnard, J. *et al.* (2017) Large-scale analyses of common and rare variants identify 12 new loci associated with atrial fibrillation Nature genetics **49**, 946-952 10.1038/ng.3843

86. Eriguchi, Y., Kuwabara, H., Inai, A., Kawakubo, Y., Nishimura, F., Kakiuchi, C. *et al.* (2017) Identification of candidate genes involved in the etiology of sporadic Tourette syndrome by exome sequencing American journal of medical genetics Part B, Neuropsychiatric genetics : the official publication of the International Society of Psychiatric Genetics **174**, 712-723 10.1002/ajmg.b.32559

87. Hamada, N., Negishi, Y., Mizuno, M., Miya, F., Hattori, A., Okamoto, N. *et al.* (2017) Role of a heterotrimeric G-protein, Gi2, in the corticogenesis: possible involvement in periventricular nodular heterotopia and intellectual disability J Neurochem **140**, 82-95 10.1111/jnc.13878

88. Hamanaka, T., Kimura, M., Sakurai, T., Ishida, N., Yasuda, J., Nagasaki, M. *et al.* (2017) A Histologic Categorization of Aqueous Outflow Routes in Familial Open-Angle Glaucoma and Associations With Mutations in the MYOC Gene in Japanese Patients Invest Ophthalmol Vis Sci **58**, 2818-2831 10.1167/iovs.16-20646

89. Hirasawa, A., Imoto, I., Naruto, T., Akahane, T., Yamagami, W., Nomura, H. *et al.* (2017) Prevalence of pathogenic germline variants detected by multigene sequencing in unselected Japanese patients with ovarian cancer Oncotarget **8**, 112258-112267 10.18632/oncotarget.22733

90. Ikeda, F., Yoshida, K., Toki, T., Uechi, T., Ishida, S., Nakajima, Y. *et al.* (2017) Exome sequencing identified RPS15A as a novel causative gene for Diamond-Blackfan anemia Haematologica **102**, e93-e96 10.3324/haematol.2016.153932

91. Isaka, M., Serizawa, M., Kenmotsu, H., Koh, Y., Takahashi, S., Maniwa, T. *et al.* (2017) Comparison of Clinically Relevant Mutation Profiles Between Preoperative Biopsy and Corresponding Surgically Resected Specimens in Japanese Patients With Non-Small-cell Lung Cancer by Amplicon-based Massively Parallel Sequencing Clin Lung Cancer **18**, 519-526 e511 10.1016/j.cllc.2016.11.022

92. Ishizuka, K., Fujita, Y., Kawabata, T., Kimura, H., Iwayama, Y., Inada, T. *et al.* (2017) Rare genetic variants in CX3CR1 and their contribution to the increased risk of schizophrenia and autism spectrum disorders Transl Psychiatry **7**, e1184 10.1038/tp.2017.173

93. Iwama, K., Mizuguchi, T., Takanashi, J. I., Shibayama, H., Shichiji, M., Ito, S. *et al.* (2017) Identification of novel SNORD118 mutations in seven patients with leukoencephalopathy with brain calcifications and cysts Clin Genet **92**, 180-187 10.1111/cge.12991

94. Iwanaga, A., Okubo, Y., Yozaki, M., Koike, Y., Kuwatsuka, Y., Tomimura, S. *et al.* (2017) Analysis of clinical symptoms and ABCC6 mutations in 76 Japanese patients with pseudoxanthoma elasticum J Dermatol **44**, 644-650 10.1111/1346-8138.13727

95. Kaneko, Y., Nakano, A., Arimoto, Y., Nara, K., Mutai, H., andMatsunaga, T. (2017) The first sporadic case of DFNA11 identified by next-generation sequencing Int J Pediatr Otorhinolaryngol **100**, 183-186 10.1016/j.ijporl.2017.07.007

96. Kasahara, T., Ishiwata, M., Kakiuchi, C., Fuke, S., Iwata, N., Ozaki, N. *et al.* (2017) Enrichment of deleterious variants of mitochondrial DNA polymerase gene (POLG1) in bipolar disorder Psychiatry and clinical neurosciences **71**, 518-529 10.1111/pcn.12496

97. Kawasaki, Y., Oda, H., Ito, J., Niwa, A., Tanaka, T., Hijikata, A. *et al.* (2017) Identification of a High-Frequency Somatic NLRC4 Mutation as a Cause of Autoinflammation by Pluripotent Cell-Based Phenotype Dissection Arthritis & rheumatology (Hoboken, NJ) **69**, 447-459 10.1002/art.39960

98. Kawashima, M., Hitomi, Y., Aiba, Y., Nishida, N., Kojima, K., Kawai, Y. *et al.* (2017) Genome-wide association studies identify PRKCB as a novel genetic susceptibility locus for primary biliary cholangitis in the Japanese population Human molecular genetics **26**, 650-659 10.1093/hmg/ddw406

99. Kusakabe, M., Nagai, M., Nakano, E., Jitsukawa, O., Nishigori, C., andYamanishi, K. (2017) A Japanese Case of Ichthyosiform Erythroderma with a Novel Mutation in NIPAL4/Ichthyin Acta Derm Venereol **97**, 397-398 10.2340/00015555-2550

100. Lee, S., Seo, J., Park, J., Nam, J. Y., Choi, A., Ignatius, J. S. *et al.* (2017) Korean Variant Archive (KOVA): a reference database of genetic variations in the Korean population Sci Rep **7**, 4287 10.1038/s41598-017-04642-4

101. Matsudate, Y., Naruto, T., Hayashi, Y., Minami, M., Tohyama, M., Yokota, K. *et al.* (2017) Targeted exome sequencing and chromosomal microarray for the molecular diagnosis of nevoid basal cell carcinoma syndrome J Dermatol Sci **86**, 206-211 10.1016/j.jdermsci.2017.02.282

102. Matsuura, K., Sawai, H., Ikeo, K., Ogawa, S., Iio, E., Isogawa, M. *et al.* (2017) Genome-Wide Association Study Identifies TLL1 Variant Associated With Development of Hepatocellular Carcinoma After Eradication of Hepatitis C Virus Infection Gastroenterology **152**, 1383-1394 10.1053/j.gastro.2017.01.041

103. Mishima, E., Mori, T., Sohara, E., Uchida, S., Abe, T., andIto, S. (2017) Inherited, not acquired, Gitelman syndrome in a patient with Sjogren's syndrome: importance of genetic testing to distinguish the two forms CEN Case Rep **6**, 180-184 10.1007/s13730-017-0271-4

104. Morimoto, Y., Ono, S., Imamura, A., Okazaki, Y., Kinoshita, A., Mishima, H. *et al.* (2017) Deep sequencing reveals variations in somatic cell mosaic mutations between monozygotic twins with discordant psychiatric disease Hum Genome Var **4**, 17032 10.1038/hgv.2017.32

105. Murakami, A., Yasuhira, S., Mayama, H., Miura, H., Maesawa, C., andSatoh, K. (2017) Characterization of PAX9 variant P20L identified in a Japanese family with tooth agenesis PLoS One **12**, e0186260 10.1371/journal.pone.0186260

106. Nakagomi, H., Hirotsu, Y., Okimoto, K., Sakamoto, I., Amemiya, K., Nakagomi, S. *et al.* (2017) PALB2 mutation in a woman with bilateral breast cancer: A case report Mol Clin Oncol **6**, 556-560 10.3892/mco.2017.1189

107. Nishio, S. Y., andUsami, S. I. (2017) The Clinical Next-Generation Sequencing Database: A Tool for the Unified Management of Clinical Information and Genetic Variants to Accelerate Variant Pathogenicity Classification Hum Mutat **38**, 252-259 10.1002/humu.23160

108. Nishiyama, A., Niihori, T., Warita, H., Izumi, R., Akiyama, T., Kato, M. *et al.* (2017) Comprehensive targeted next-generation sequencing in Japanese familial amyotrophic lateral sclerosis Neurobiol Aging **53**, 194 e191-194 e198 10.1016/j.neurobiolaging.2017.01.004

109. Ohnami, S., Nagashima, T., Urakami, K., Shimoda, Y., Kamada, F., Saito, J. *et al.* (2017) Whole exome sequencing detects variants of genes that mediate response to anticancer drugs J Toxicol Sci **42**, 137-144, <Go to ISI>://WOS:000398651800003

110. Ohto, T., Enokizono, T., Tanaka, R., Tanaka, M., Suzuki, H., Sakai, A. *et al.* (2017) A novel BBS10 mutation identified in a patient with Bardet-Biedl syndrome with a violent emotional outbreak Hum Genome Var **4**, 17033 10.1038/hgv.2017.33

111. Okada, A., Kohmoto, T., Naruto, T., Yokota, I., Kotani, Y., Shimada, A. *et al.* (2017) The first Japanese patient with mandibular hypoplasia, deafness, progeroid features and lipodystrophy diagnosed via POLD1 mutation detection Hum Genome Var **4**, 17031 10.1038/hgv.2017.31

112. Okamoto, N., Kohmoto, T., Naruto, T., Masuda, K., Komori, T., andImoto, I. (2017) Novel CLCN7 compound heterozygous mutations in intermediate autosomal recessive osteopetrosis Hum Genome Var **4**, 17036 10.1038/hgv.2017.36

113. Okamoto, N., Tsuchiya, Y., Miya, F., Tsunoda, T., Yamashita, K., Boroevich, K. A. *et al.* (2017) A novel genetic syndrome with STARD9 mutation and abnormal spindle morphology Am J Med Genet A **173**, 2690-2696 10.1002/ajmg.a.38391

114. Okamoto, N., Watanabe, M., Naruto, T., Matsuda, K., Kohmoto, T., Saito, M. *et al.* (2017) Genome-first approach diagnosed Cabezas syndrome via novel CUL4B mutation detection Hum Genome Var **4**, 16045 10.1038/hgv.2016.45

115. Okazaki, M., Suzuki, H., Takahashi, Y., Ishiura, H., Goto, J., Hirano, M. *et al.* (2017) Novel mutation in the SOD1 gene in a patient with early-onset, rapidly progressive amyotrophic lateral sclerosis Neurology and Clinical Neuroscience **5**, 189-191 10.1111/ncn3.12153

116. Oshima, Y., Yamamoto, T., Ishikawa, T., Mishima, H., Matsusue, A., Umehara, T. *et al.* (2017) Postmortem genetic analysis of sudden unexpected death in infancy: neonatal genetic screening may enable the prevention of sudden infant death Journal of human genetics **62**, 989-995 10.1038/jhg.2017.79

117. Seki, A., Ishikawa, T., Daumy, X., Mishima, H., Barc, J., Sasaki, R. *et al.* (2017) Progressive Atrial Conduction Defects Associated With Bone Malformation Caused by a Connexin-45 Mutation J Am Coll Cardiol **70**, 358-370 10.1016/j.jacc.2017.05.039

118. Soma, N., Higashimoto, K., Imamura, M., Saitoh, A., Soejima, H., andNagasaki, K. (2017) Long term survival of a patient with Perlman syndrome due to novel compound heterozygous missense mutations in RNB domain of DIS3L2 Am J Med Genet A **173**, 1077-1081 10.1002/ajmg.a.38111

119. Suzuki, O., Eguchi, H., Chika, N., Sakimoto, T., Ishibashi, K., Kumamoto, K. *et al.* (2017) Prevalence and clinicopathologic/molecular characteristics of mismatch repair-deficient colorectal cancer in the under-50-year-old Japanese population Surg Today **47**, 1135-1146 10.1007/s00595-017-1486-x

120. Tobita, T., Nomura, S., Morita, H., Ko, T., Fujita, T., Toko, H. *et al.* (2017) Identification of MYLK3 mutations in familial dilated cardiomyopathy Sci Rep **7**, 17495 10.1038/s41598-017-17769-1

121. Yamaki, M., Saito, H., Isono, K., Goto, T., Shirakawa, H., Shoji, N. *et al.* (2017) Genotyping Analysis of Bitter-Taste Receptor Genes TAS2R38 and TAS2R46 in Japanese Patients with Gastrointestinal Cancers J Nutr Sci Vitaminol (Tokyo) **63**, 148-154 10.3177/jnsv.63.148

122. Yamamoto, G., Kikuchi, M., Kobayashi, S., Arai, Y., Fujiyoshi, K., Wakatsuki, T. *et al.* (2017) Routine genetic testing of lung cancer specimens derived from surgery, bronchoscopy and fluid aspiration by next generation sequencing Int J Oncol **50**, 1579-1589 10.3892/ijo.2017.3935

123. Yamamoto, N., Mutai, H., Namba, K., Morita, N., Masuda, S., Nishi, Y. *et al.* (2017) Prevalence of TECTA mutation in patients with mid-frequency sensorineural hearing loss Orphanet J Rare Dis **12**, 157 10.1186/s13023-017-0708-z

124. Yasukochi, Y., Sakuma, J., Takeuchi, I., Kato, K., Oguri, M., Fujimaki, T. *et al.* (2017) Longitudinal exome-wide association study to identify genetic susceptibility loci for hypertension in a Japanese population Exp Mol Med **49**, e409; 10.1038/emm.2017.209

125. 矢本香織, 川原央好, and 漆原直人 (2017) 次世代シークエンサーにより ACTG2 遺伝子変異が同定されたヒルシュスプルング病類縁疾患の 1 例 日本小児外科学会雑誌 **53**, 1210-1214,

126. Abe, S., Takeda, H., Nishio, S. Y., andUsami, S. I. (2018) Sensorineural hearing loss and mild cardiac phenotype caused by an EYA4 mutation Hum Genome Var **5**, 23 10.1038/s41439-018-0023-9

127. Arai, E., Miura, F., Totoki, Y., Yamashita, S., Tian, Y., Gotoh, M. *et al.* (2018) Epigenome mapping of human normal purified hepatocytes: personal epigenome variation and genome-epigenome correlation Epigenomics **10**, 955-979 10.2217/epi-2017-0111

128. Arita, K., Niimi, H., Yamagishi, N., Ueno, T., Kitajima, I., andSugiyama, T. (2018) Factor X heterozygous mutation in a patient with potential risk of bleeding: A case report Medicine (Baltimore) **97**, e10950 10.1097/MD.0000000000010950

129. Belal, H., Nakashima, M., Matsumoto, H., Yokochi, K., Taniguchi-Ikeda, M., Aoto, K. *et al.* (2018) De novo variants in RHOBTB2, an atypical Rho GTPase gene, cause epileptic encephalopathy Hum Mutat **39**, 1070-1075 10.1002/humu.23550

130. Chen, C. J., Lu, T. P., Lin, L. Y., Liu, Y. B., Ho, L. T., Huang, H. C. *et al.* (2018) Impact of Ancestral Differences and Reassessment of the Classification of Previously Reported Pathogenic Variants in Patients With Brugada Syndrome in the Genomic Era: A SADS-TW BrS Registry Front Genet **9**, 680 10.3389/fgene.2018.00680

131. Chiba, H., Kakuta, Y., Kinouchi, Y., Kawai, Y., Watanabe, K., Nagao, M. *et al.* (2018) Allele-specific DNA methylation of disease susceptibility genes in Japanese patients with inflammatory bowel disease PLoS One **13**, e0194036 10.1371/journal.pone.0194036

132. Chinen, Y., Nakamura, S., Ganaha, A., Hayashi, S., Inazawa, J., Yanagi, K. *et al.* (2018) Mild prominence of the Sylvian fissure in a Bainbridge-Ropers syndrome patient with a novel frameshift variant in ASXL3 Clin Case Rep **6**, 330-336 10.1002/ccr3.1361

133. Dateki, S., Kitajima, T., Kihara, T., Watanabe, S., Yoshiura, K. I., andMoriuchi, H. (2018) Novel compound heterozygous variants in the LARP7 gene in a patient with Alazami syndrome Hum Genome Var **5**, 18014 10.1038/hgv.2018.14

134. Dobashi, A., Togashi, Y., Tanaka, N., Yokoyama, M., Tsuyama, N., Baba, S. *et al.* (2018) TP53 and OSBPL10 alterations in diffuse large B-cell lymphoma: prognostic markers identified via exome analysis of cases with extreme prognosis Oncotarget **9**, 19555-19568 10.18632/oncotarget.24656

135. Eisfeldt, J., Nilsson, D., Andersson-Assarsson, J. C., andLindstrand, A. (2018) AMYCNE: Confident copy number assessment using whole genome sequencing data PLoS One **13**, e0189710 10.1371/journal.pone.0189710

136. Fujimaru, T., Mori, T., Sekine, A., Mandai, S., Chiga, M., Kikuchi, H. *et al.* (2018) Kidney enlargement and multiple liver cyst formation implicate mutations in PKD1/2 in adult sporadic polycystic kidney disease Clin Genet **94**, 125-131 10.1111/cge.13249

137. Fukuda, H., Imagawa, E., Hamanaka, K., Fujita, A., Mitsuhashi, S., Miyatake, S. *et al.* (2018) A novel missense SNAP25b mutation in two affected siblings from an Israeli family showing seizures and cerebellar ataxia Journal

of human genetics **63**, 673-676 10.1038/s10038-018-0421-3

138. Hiraide, T., Nakashima, M., Yamoto, K., Fukuda, T., Kato, M., Ikeda, H. *et al.* (2018) De novo variants in SETD1B are associated with intellectual disability, epilepsy and autism Hum Genet **137**, 95-104 10.1007/s00439-017-1863-y
139. Hishinuma, E., Narita, Y., Saito, S., Maekawa, M., Akai, F., Nakanishi, Y. *et al.* (2018) Functional Characterization of 21 Allelic Variants of Dihydropyrimidine Dehydrogenase Identified in 1070 Japanese Individuals Drug Metab Dispos **46**, 1083-1090 10.1124/dmd.118.081737
140. Horikawa, Y., Hosomichi, K., Enya, M., Ishiura, H., Suzuki, Y., Tsuji, S. *et al.* (2018) No novel, high penetrant gene might remain to be found in Japanese patients with unknown MODY Journal of human genetics **63**, 821-829 10.1038/s10038-018-0449-4
141. Horiuchi, Y., Lin, J., Shinojima, Y., Fujiwara, K., Moriyama, M., andNagase, H. (2018) Characterizing key nucleotide polymorphisms of hepatitis C virus-disease associations via mass-spectrometric genotyping Int J Oncol **52**, 441-452 10.3892/ijo.2017.4209
142. Hosono, K., Nishina, S., Yokoi, T., Katagiri, S., Saitsu, H., Kurata, K. *et al.* (2018) Molecular Diagnosis of 34 Japanese Families with Leber Congenital Amaurosis Using Targeted Next Generation Sequencing Sci Rep **8**, 8279 10.1038/s41598-018-26524-z
143. Ishige, T., Itoga, S., Utsuno, E., Nishimura, M., Yoshikawa, M., Kato, N. *et al.* (2018) Variant in C-terminal region of intestinal alkaline phosphatase associated with benign familial hyperphosphatasaemia J Med Genet **55**, 701-704 10.1136/jmedgenet-2017-104964
144. Ishige, T., Satoh, M., Itoga, S., Nishimura, M., Matsushita, K., andNomura, F. (2018) High-throughput genotyping of GC (vitamin D-binding protein) by melting analysis with locked nucleic acid-incorporating dual hybridization probe for improving mismatch discrimination Clin Chim Acta **487**, 126-132 10.1016/j.cca.2018.09.034
145. Ishizuka, K., Kimura, H., Kushima, I., Inada, T., Okahisa, Y., Ikeda, M. *et al.* (2018) Assessment of a glyoxalase I frameshift variant, p.P122fs, in Japanese patients with schizophrenia Psychiatr Genet **28**, 90-93 10.1097/YPG.0000000000000204
146. Iwama, K., Iwata, A., Shiina, M., Mitsuhashi, S., Miyatake, S., Takata, A. *et al.* (2018) A novel mutation in SLC1A3 causes episodic ataxia Journal of human genetics **63**, 207-211 10.1038/s10038-017-0365-z
147. Iwama, K., Osaka, H., Ikeda, T., Mitsuhashi, S., Miyatake, S., Takata, A. *et al.* (2018) A novel SLC9A1 mutation causes cerebellar ataxia Journal of human genetics **63**, 1049-1054 10.1038/s10038-018-0488-x
148. Jia, X., Horinouchi, T., Hitomi, Y., Shono, A., Khor, S. S., Omae, Y. *et al.* (2018) Strong Association of the HLA-DR/DQ Locus with Childhood Steroid-Sensitive Nephrotic Syndrome in the Japanese Population J Am Soc Nephrol **29**, 2189-2199 10.1681/ASN.2017080859
149. Kakuta, Y., Kawai, Y., Okamoto, D., Takagawa, T., Ikeya, K., Sakuraba, H. *et al.* (2018) NUDT15 codon 139 is the best pharmacogenetic marker for predicting thiopurine-induced severe adverse events in Japanese patients with inflammatory bowel disease: a multicenter study J Gastroenterol **53**, 1065-1078 10.1007/s00535-018-1486-7
150. Kakuta, Y., Kinouchi, Y., andShimosegawa, T. (2018) Pharmacogenetics of thiopurines for inflammatory bowel disease in East Asia: prospects for clinical application of NUDT15 genotyping J Gastroenterol **53**, 172-180 10.1007/s00535-017-1416-0
151. Katagiri, S., Hayashi, T., Yoshitake, K., Murai, N., Matsui, Z., Kubo, H. *et al.* (2018) Compound heterozygous splice site variants in the SCLT1 gene highlight an additional candidate locus for Senior-Loken syndrome Sci Rep

8, 16733 10.1038/s41598-018-35152-6

152. Kurata, K., Hosono, K., Hikoya, A., Kato, A., Saitsu, H., Minoshima, S. *et al.* (2018) Clinical characteristics of a Japanese patient with Bardet-Biedl syndrome caused by BBS10 mutations Jpn J Ophthalmol **62**, 458-466 10.1007/s10384-018-0591-8
153. Kurata, K., Hosono, K., andHotta, Y. (2018) Long-term clinical course of 2 Japanese patients with PRPF31-related retinitis pigmentosa Jpn J Ophthalmol **62**, 186-193 10.1007/s10384-017-0560-7
154. Kurata, K., Hosono, K., andHotta, Y. (2018) Clinical and genetic findings of a Japanese patient with RP1-related autosomal recessive retinitis pigmentosa Doc Ophthalmol **137**, 47-56 10.1007/s10633-018-9649-7
155. Matsumoto, A., Matsui, I., Mori, T., Sakaguchi, Y., Mizui, M., Ueda, Y. *et al.* (2018) Severe Osteomalacia with Dent Disease Caused by a Novel Intronic Mutation of the CLCN5 gene Intern Med **57**, 3603-3610 10.2169/internalmedicine.1272-18
156. Mikasa, M., Kanai, K., Li, Y., Yoshino, H., Mogushi, K., Hayashida, A. *et al.* (2018) COQ2 variants in Parkinson's disease and multiple system atrophy J Neural Transm (Vienna) **125**, 937-944 10.1007/s00702-018-1885-1
157. Momozawa, Y., Iwasaki, Y., Parsons, M. T., Kamatani, Y., Takahashi, A., Tamura, C. *et al.* (2018) Germline pathogenic variants of 11 breast cancer genes in 7,051 Japanese patients and 11,241 controls Nat Commun **9**, 4083 10.1038/s41467-018-06581-8
158. Morimoto, Y., Shimada-Sugimoto, M., Otowa, T., Yoshida, S., Kinoshita, A., Mishima, H. *et al.* (2018) Whole-exome sequencing and gene-based rare variant association tests suggest that PLA2G4E might be a risk gene for panic disorder Transl Psychiatry **8**, 41 10.1038/s41398-017-0088-0
159. Mutoh, H., Kato, M., Akita, T., Shibata, T., Wakamoto, H., Ikeda, H. *et al.* (2018) Biallelic Variants in CNPY3, Encoding an Endoplasmic Reticulum Chaperone, Cause Early-Onset Epileptic Encephalopathy Am J Hum Genet **102**, 321-329 10.1016/j.ajhg.2018.01.004
160. Nagasaki, K., Nishimura, G., Kikuchi, T., Nyuzuki, H., Sasaki, S., Ogawa, Y. *et al.* (2018) Nonsense mutations in FZD2 cause autosomal-dominant omodysplasia: Robinow syndrome-like phenotypes Am J Med Genet A **176**, 739-742 10.1002/ajmg.a.38623
161. Naito, T., Yokoyama, N., Kakuta, Y., Ueno, K., Kawai, Y., Onodera, M. *et al.* (2018) Clinical and genetic risk factors for decreased bone mineral density in Japanese patients with inflammatory bowel disease J Gastroenterol Hepatol **33**, 1873-1881 10.1111/jgh.14149
162. Nakamura, S., Yokoyama, K., Yusa, N., Ogawa, M., Takei, T., Kobayashi, A. *et al.* (2018) Circulating tumor DNA dynamically predicts response and/or relapse in patients with hematological malignancies Int J Hematol **108**, 402-410 10.1007/s12185-018-2487-2
163. Nishioka, M., Bundo, M., Ueda, J., Yoshikawa, A., Nishimura, F., Sasaki, T. *et al.* (2018) Identification of somatic mutations in monozygotic twins discordant for psychiatric disorders NPJ Schizophr **4**, 7 10.1038/s41537-018-0049-5
164. Ohmoto, A., Morizane, C., Kubo, E., Takai, E., Hosoi, H., Sakamoto, Y. *et al.* (2018) Germline variants in pancreatic cancer patients with a personal or family history of cancer fulfilling the revised Bethesda guidelines J Gastroenterol **53**, 1159-1167 10.1007/s00535-018-1466-y
165. Okamoto, N., Kohmoto, T., Naruto, T., Masuda, K., andImoto, I. (2018) Primary microcephaly caused by novel compound heterozygous mutations in ASPM Hum Genome Var **5**, 18015 10.1038/hgv.2018.15
166. Sasaki, H., Yanagi, K., Ugi, S., Kobayashi, K., Ohkubo, K., Tajiri, Y. *et al.* (2018) Definitive diagnosis of mandibular hypoplasia, deafness, progeroid features and lipodystrophy (MDPL) syndrome caused by a recurrent de novo

mutation in the POLD1 gene Endocr J **65**, 227-238 10.1507/endocrj.EJ17-0287

167. Sato, T., Samura, O., Kato, N., Taniguchi, K., Takahashi, K., Ito, Y. *et al.* (2018) Novel TFAP2A mutation in a Japanese family with Branchio-oculo-facial syndrome Hum Genome Var **5**, 5 10.1038/s41439-018-0004-z
168. Shimizu, D., Iwashima, S., Sato, K., Hayano, S., Fukami, M., Saitsu, H. *et al.* (2018) GATA4 variant identified by whole-exome sequencing in a Japanese family with atrial septal defect: Implications for male sex development Clin Case Rep **6**, 2229-2233 10.1002/ccr3.1851
169. Suzuki, A., Kawano, S., Mitsuyama, T., Suyama, M., Kanai, Y., Shirahige, K. *et al.* (2018) DBTSS/DBKERO for integrated analysis of transcriptional regulation Nucleic Acids Res **46**, D229-D238 10.1093/nar/gkx1001
170. Suzuki, H., Kurosawa, K., Fukuda, K., Ijima, K., Sumazaki, R., Saito, S. *et al.* (2018) Japanese pathogenic variant database: DPV Translational Science of Rare Diseases **3**, 133-137 10.3233/trd-180027
171. Suzuki-Muromoto, S., Wakusawa, K., Miyabayashi, T., Sato, R., Okubo, Y., Endo, W. *et al.* (2018) A case of new PCDH12 gene variants presented as dyskinetic cerebral palsy with epilepsy Journal of human genetics **63**, 749-753 10.1038/s10038-018-0432-0
172. Takahashi, K., Hayano, T., Sugimoto, R., Kashiwagi, H., Shinoda, M., Nishijima, Y. *et al.* (2018) Exome and copy number variation analyses of Mayer-Rokitansky-Kuster- Hauser syndrome Hum Genome Var **5**, 27 10.1038/s41439-018-0028-4
173. Takahashi, M., Hosomichi, K., Yamaguchi, T., Nagahama, R., Yoshida, H., Maki, K. *et al.* (2018) Whole-genome sequencing in a pair of monozygotic twins with discordant cleft lip and palate subtypes Oral Dis **24**, 1303-1309 10.1111/odi.12910
174. Takahashi, M., Hosomichi, K., Yamaguchi, T., Nagahama, R., Yoshida, H., Marazita, M. L. *et al.* (2018) Exploration of genetic factors determining cleft side in a pair of monozygotic twins with mirror-image cleft lip and palate using whole-genome sequencing and comparison of craniofacial morphology Arch Oral Biol **96**, 33-38 10.1016/j.archoralbio.2018.08.009
175. Takeda, K., Kou, I., Mizumoto, S., Yamada, S., Kawakami, N., Nakajima, M. *et al.* (2018) Screening of known disease genes in congenital scoliosis Mol Genet Genomic Med **6**, 966-974 10.1002/mgg3.466
176. Takenouchi, T., Inaba, M., Uehara, T., Takahashi, T., Kosaki, K., andMizuno, S. (2018) Biallelic mutations in NALCN: Expanding the genotypic and phenotypic spectra of IHPRF1 Am J Med Genet A **176**, 431-437 10.1002/ajmg.a.38543
177. Takeuchi, F., Akiyama, M., Matoba, N., Katsuya, T., Nakatochi, M., Tabara, Y. *et al.* (2018) Interethnic analyses of blood pressure loci in populations of East Asian and European descent Nat Commun **9**, 5052 10.1038/s41467-018-07345-0
178. Takeyari, S., Kubota, T., Miyata, K., Yamamoto, K., Nakayama, H., Yamamoto, K. *et al.* (2018) Japanese patient with Cole-carpenter syndrome with compound heterozygous variants of SEC24D Am J Med Genet A **176**, 2882-2886 10.1002/ajmg.a.40643
179. Tamai, K., Tada, K., Takeuchi, A., Nakamura, M., Marunaka, H., Washio, Y. *et al.* (2018) Fetal ultrasonographic findings including cerebral hyperechogenicity in a patient with non-lethal form of Raine syndrome Am J Med Genet A **176**, 682-686 10.1002/ajmg.a.38598
180. Tanabe, H., Higuchi, Y., Yuan, J. H., Hashiguchi, A., Yoshimura, A., Ishihara, S. *et al.* (2018) Clinical and genetic features of Charcot-Marie-Tooth disease 2F and hereditary motor neuropathy 2B in Japan Journal of the peripheral nervous system : JPNS **23**, 40-48 10.1111/jns.12252
181. Tobita, T., Nomura, S., Fujita, T., Morita, H., Asano, Y., Onoue, K. *et al.* (2018) Genetic basis of cardiomyopathy

and the genotypes involved in prognosis and left ventricular reverse remodeling Sci Rep **8**, 1998 10.1038/s41598-018-20114-9

182. Tokaji, N., Ito, H., Kohmoto, T., Naruto, T., Takahashi, R., Goji, A. *et al.* (2018) A rare male patient with classic Rett syndrome caused by MeCP2_e1 mutation Am J Med Genet A **176**, 699-702 10.1002/ajmg.a.38595
183. Uehara, T., Takenouchi, T., Kosaki, R., Kurosawa, K., Mizuno, S., andKosaki, K. (2018) Redefining the phenotypic spectrum of de novo heterozygous CDK13 variants: Three patients without cardiac defects Eur J Med Genet **61**, 243-247 10.1016/j.ejmg.2017.12.004
184. Veldore, V. H., Choughule, A., Routhu, T., Mandloi, N., Noronha, V., Joshi, A. *et al.* (2018) Validation of liquid biopsy: plasma cell-free DNA testing in clinical management of advanced non-small cell lung cancer Lung Cancer (Auckl) **9**, 1-11 10.2147/LCTT.S147841
185. Wardell, C. P., Fujita, M., Yamada, T., Simbolo, M., Fassan, M., Karlic, R. *et al.* (2018) Genomic characterization of biliary tract cancers identifies driver genes and predisposing mutations J Hepatol **68**, 959-969 10.1016/j.jhep.2018.01.009
186. Watanabe, S., Kyo, H., Kang, L., Eguchi, R., Altaf-Ul-Amin, M., Morita, A. *et al.* (2018) Data Intensive Study of Accessibility of Edible Species and Healthcare Across the Globe Japanese Journal of Complementary and Alternative Medicine **15**, 37-60,
187. Yamaji, F., Soeda, A., Shibata, H., Morikawa, T., Suzuki, K., Yoshida, S. *et al.* (2018) A new mutation of congenital methemoglobinemia exacerbated after methylene blue treatment Acute Med Surg **5**, 199-201 10.1002/ams2.335
188. Yamamoto, T., Yamamoto-Shimojima, K., Ueda, Y., Imai, K., Takahashi, Y., Imagawa, E. *et al.* (2018) Independent occurrence of de novo HSPD1 and HIP1 variants in brothers with different neurological disorders - leukodystrophy and autism Hum Genome Var **5**, 18 10.1038/s41439-018-0020-z
189. Yamanaka, M., Shiga, K., Fujiwara, S., Mizuguchi, Y., Yasuda, S., Ishizawa, K. *et al.* (2018) A Novel SDHB IVS2-2A>C Mutation Is Responsible for Hereditary Pheochromocytoma/Paraganglioma Syndrome Tohoku J Exp Med **245**, 99-105 10.1620/tjem.245.99
190. Yang, X., Li, J., Fang, Y., Zhang, Z., Jin, D., Chen, X. *et al.* (2018) Rho Guanine Nucleotide Exchange Factor ARHGEF17 Is a Risk Gene for Intracranial Aneurysms Circ Genom Precis Med **11**, e002099 10.1161/CIRCGEN.117.002099
191. Yoshikawa, A., Nishimura, F., Inai, A., Eriguchi, Y., Nishioka, M., Takaya, A. *et al.* (2018) Mutations of the glycine cleavage system genes possibly affect the negative symptoms of schizophrenia through metabolomic profile changes Psychiatry and clinical neurosciences **72**, 168-179 10.1111/pcn.12628
192. Yu, Y., Lin, Y., Takasaki, Y., Wang, C., Kimura, H., Xing, J. *et al.* (2018) Rare loss of function mutations in N-methyl-D-aspartate glutamate receptors and their contributions to schizophrenia susceptibility Transl Psychiatry **8**, 12 10.1038/s41398-017-0061-y
193. Yuan, J. H., Hashiguchi, A., Okamoto, Y., Yoshimura, A., Ando, M., Shiomi, K. *et al.* (2018) Clinical and mutational spectrum of Japanese patients with recessive variants in SH3TC2 Journal of human genetics **63**, 281-287 10.1038/s10038-017-0388-5
194. Yuge, K., Iwama, K., Yonee, C., Matsufuji, M., Sano, N., Saikusa, T. *et al.* (2018) A novel STXBP1 mutation causes typical Rett syndrome in a Japanese girl Brain Dev **40**, 493-497 10.1016/j.braindev.2018.02.002
195. Akimoto, S., Goto, C., andKuriki, K. (2019) Relationship between ethanol consumption and TBL2 rs17145738 on LDL-C concentration in Japanese adults: a four season 3-day weighed diet record study BMC Nutr **5**, 61

10.1186/s40795-019-0315-6

196. Akiyama, M., Ishigaki, K., Sakaue, S., Momozawa, Y., Horikoshi, M., Hirata, M. *et al.* (2019) Characterizing rare and low-frequency height-associated variants in the Japanese population Nat Commun **10**, 4393 10.1038/s41467-019-12276-5
197. Akiyoshi, T., Tanaka, N., Kiyotani, K., Gotoh, O., Yamamoto, N., Oba, K. *et al.* (2019) Immunogenomic profiles associated with response to neoadjuvant chemoradiotherapy in patients with rectal cancer Br J Surg **106**, 1381-1392 10.1002/bjs.11179
198. Amano, Y., Akazawa, Y., Yasuda, J., Yoshino, K., Kojima, K., Kobayashi, N. *et al.* (2019) A low-frequency IL4R locus variant in Japanese patients with intravenous immunoglobulin therapy-unresponsive Kawasaki disease Pediatr Rheumatol Online J **17**, 34 10.1186/s12969-019-0337-2
199. Aoto, S., Katagiri, S., Wang, Y., Pagnamenta, A. T., Sakamoto-Abutani, R., Toyoda, M. *et al.* (2019) Frequent retrotransposition of endogenous genes in ERCC2-deficient cells derived from a patient with xeroderma pigmentosum Stem Cell Res Ther **10**, 273 10.1186/s13287-019-1381-z
200. Aoyama, K. I., Kimura, M., Yamazaki, H., Uchibori, M., Kojima, R., Osawa, Y. *et al.* (2019) New PCNT candidate missense variant in a patient with oral and maxillofacial osteodysplasia: a case report BMC Med Genet **20**, 126 10.1186/s12881-019-0858-z
201. Asanomi, Y., Shigemizu, D., Miyashita, A., Mitsumori, R., Mori, T., Hara, N. *et al.* (2019) A rare functional variant of SHARPIN attenuates the inflammatory response and associates with increased risk of late-onset Alzheimer's disease Mol Med **25**, 20 10.1186/s10020-019-0090-5
202. Boda, H., Miyata, M., Inagaki, H., Shinkai, Y., Kato, T., Yoshikawa, T. *et al.* (2019) FOXA2 gene mutation in a patient with congenital complex pituitary hormone deficiency Eur J Med Genet **62**, 103570 10.1016/j.ejmg.2018.11.004
203. Borna, N. N., Kishita, Y., Kohda, M., Lim, S. C., Shimura, M., Wu, Y. *et al.* (2019) Mitochondrial ribosomal protein PTCD3 mutations cause oxidative phosphorylation defects with Leigh syndrome Neurogenetics **20**, 9-25 10.1007/s10048-018-0561-9
204. Chen, Y. A., Yogo, E., Kurihara, N., Ohno, T., Higuchi, C., Rokushima, M. *et al.* (2019) Assessing drug target suitability using TargetMine F1000Res **8**, 233 10.12688/f1000research.18214.2
205. Chinen, Y., Nakamura, S., Kaneshi, T., Nakayashiro, M., Yanagi, K., Kaname, T. *et al.* (2019) A novel nonsense SMC1A mutation in a patient with intractable epilepsy and cardiac malformation Hum Genome Var **6**, 23 10.1038/s41439-019-0053-y
206. Daida, K., Nishioka, K., Li, Y., Yoshino, H., Kikuchi, A., Hasegawa, T. *et al.* (2019) Mutation analysis of LRP10 in Japanese patients with familial Parkinson's disease, progressive supranuclear palsy, and frontotemporal dementia Neurobiol Aging **84**, 235 e211-235 e216 10.1016/j.neurobiolaging.2019.08.030
207. Dateki, S., Watanabe, S., Mishima, H., Shirakawa, T., Morikawa, M., Kinoshita, E. *et al.* (2019) A homozygous splice site ROBO1 mutation in a patient with a novel syndrome with combined pituitary hormone deficiency Journal of human genetics **64**, 341-346 10.1038/s10038-019-0566-8
208. Den, K., Kudo, Y., Kato, M., Watanabe, K., Doi, H., Tanaka, F. *et al.* (2019) Recurrent NUS1 canonical splice donor site mutation in two unrelated individuals with epilepsy, myoclonus, ataxia and scoliosis - a case report BMC Neurol **19**, 253 10.1186/s12883-019-1489-x
209. Fujii, R., Hishida, A., Suzuki, K., Imaeda, N., Goto, C., Hamajima, N. *et al.* (2019) Cluster of differentiation 36 gene polymorphism (rs1761667) is associated with dietary MUFA intake and hypertension in a Japanese

population Br J Nutr **121**, 1215-1222 10.1017/S0007114519000679

210. Fukumura, S., Yamauchi, K., Kawanabe, A., Yamamoto, A., Nakaza, M., Kubota, T. *et al.* (2019) Functional analysis of a double-point mutation in the KCNJ2 gene identified in a family with Andersen-Tawil syndrome J Neurol Sci **407**, 116521 10.1016/j.jns.2019.116521
211. Fukunaga, H., Prise, K. M., andTaki, Y. (2019) Updated understanding of WRN variants using the Japanese whole-genome reference panel 3.5KJPNv2 Geriatr Gerontol Int **19**, 961-962 10.1111/ggi.13724
212. Fukunaga, H., Taki, Y., andPrise, K. M. (2019) Diversity of ATM gene variants: a population-based genome data analysis for precision medicine Hum Genomics **13**, 38 10.1186/s40246-019-0234-2
213. Furukawa, H., Oka, S., Shimada, K., Hashimoto, A., Komiya, A., Matsui, T. *et al.* (2019) Role of Deleterious Rare Alleles for Acute-Onset Diffuse Interstitial Lung Disease in Collagen Diseases Clin Med Insights Circ Respir Pulm Med **13**, 1179548419866443 10.1177/1179548419866443
214. Gotoh, O., Sugiyama, Y., Takazawa, Y., Kato, K., Tanaka, N., Omatsu, K. *et al.* (2019) Clinically relevant molecular subtypes and genomic alteration-independent differentiation in gynecologic carcinosarcoma Nat Commun **10**, 4965 10.1038/s41467-019-12985-x
215. Gustafson, M. A., McCormick, E. M., Perera, L., Longley, M. J., Bai, R., Kong, J. *et al.* (2019) Mitochondrial single-stranded DNA binding protein novel de novo SSBP1 mutation in a child with single large-scale mtDNA deletion (SLSMD) clinically manifesting as Pearson, Kearns-Sayre, and Leigh syndromes PLoS One **14**, e0221829 10.1371/journal.pone.0221829
216. Hamaguchi, Y., Aoki, M., Watanabe, S., Mishima, H., Yoshiura, K. I., Moriuchi, H. *et al.* (2019) KAT6B-related disorder in a patient with a novel frameshift variant (c.3925dup) Hum Genome Var **6**, 54 10.1038/s41439-019-0085-3
217. Hamanaka, K., Miyatake, S., Koshimizu, E., Tsurusaki, Y., Mitsuhashi, S., Iwama, K. *et al.* (2019) RNA sequencing solved the most common but unrecognized NEB pathogenic variant in Japanese nemaline myopathy Genet Med **21**, 1629-1638 10.1038/s41436-018-0360-6
218. Hamanaka, K., Sugawara, Y., Shimoji, T., Nordtveit, T. I., Kato, M., Nakashima, M. *et al.* (2019) De novo truncating variants in PHF21A cause intellectual disability and craniofacial anomalies Eur J Hum Genet **27**, 378-383 10.1038/s41431-018-0289-x
219. Hashimoto, T., Harita, Y., Takizawa, K., Urae, S., Ishizuka, K., Miura, K. *et al.* (2019) In Vivo Expression of NUP93 and Its Alteration by NUP93 Mutations Causing Focal Segmental Glomerulosclerosis Kidney Int Rep **4**, 1312-1322 10.1016/j.ekir.2019.05.1157
220. Hata, Y., Hirono, K., Yamaguchi, Y., Ichida, F., Oku, Y., andNishida, N. (2019) Minimal inflammatory foci of unknown etiology may be a tentative sign of early stage inherited cardiomyopathy Mod Pathol **32**, 1281-1290 10.1038/s41379-019-0274-0
221. Hata, Y., Ichimata, S., Yamaguchi, Y., Hirono, K., Oku, Y., Ichida, F. *et al.* (2019) Clinicopathological and Genetic Profiles of Cases with Myocytes Disarray-Investigation for Establishing the Autopsy Diagnostic Criteria for Hypertrophic Cardiomyopathy J Clin Med **8**, 463-474 10.3390/jcm8040463
222. Higuchi, T., Oka, S., Furukawa, H., Nakamura, M., Komori, A., Abiru, S. *et al.* (2019) Role of deleterious single nucleotide variants in the coding regions of TNFAIP3 for Japanese autoimmune hepatitis with cirrhosis Sci Rep **9**, 7925 10.1038/s41598-019-44524-5
223. Hiraide, T., Hattori, A., Ieda, D., Hori, I., Saitoh, S., Nakashima, M. *et al.* (2019) De novo variants in SETD1B cause intellectual disability, autism spectrum disorder, and epilepsy with myoclonic absences Epilepsia Open **4**,

476-481 10.1002/epi4.12339

224. Hiraide, T., Kaba Yasui, H., Kato, M., Nakashima, M., andSaitsu, H. (2019) A de novo variant in RAC3 causes severe global developmental delay and a middle interhemispheric variant of holoprosencephaly Journal of human genetics **64**, 1127-1132 10.1038/s10038-019-0656-7

225. Hirono, K., Ichida, F., Nishio, N., Ogawa-Tominaga, M., Fushimi, T., Feichtinger, R. G. *et al.* (2019) Mitochondrial complex deficiency by novel compound heterozygous TMEM70 variants and correlation with developmental delay, undescended testicle, and left ventricular noncompaction in a Japanese patient: A case report Clin Case Rep **7**, 553-557 10.1002/ccr3.2050

226. Hori, M., Miyauchi, E., Son, C., andHarada-Shiba, M. (2019) Detection of the benign c.2579C>T (p.A860V) variant of the LDLR gene in a pedigree-based genetic analysis of familial hypercholesterolemia J Clin Lipidol **13**, 335-339 10.1016/j.jacl.2019.01.004

227. Hori, M., Ohta, N., Takahashi, A., Masuda, H., Isoda, R., Yamamoto, S. *et al.* (2019) Impact of LDLR and PCSK9 pathogenic variants in Japanese heterozygous familial hypercholesterolemia patients Atherosclerosis **289**, 101-108 10.1016/j.atherosclerosis.2019.08.004

228. Hosoda, Y., Miyake, M., Schellevis, R. L., Boon, C. J. F., Hoyng, C. B., Miki, A. *et al.* (2019) Genome-wide association analyses identify two susceptibility loci for pachychoroid disease central serous chorioretinopathy Commun Biol **2**, 468 10.1038/s42003-019-0712-z

229. Howles, S. A., Wiberg, A., Goldsworthy, M., Bayliss, A. L., Gluck, A. K., Ng, M. *et al.* (2019) Genetic variants of calcium and vitamin D metabolism in kidney stone disease Nat Commun **10**, 5175 10.1038/s41467-019-13145-x

230. Ichikawa, Y., Tanaka, M., Kurita, E., Nakajima, M., Tanaka, M., Oishi, C. *et al.* (2019) Novel SLC20A2 variant in a Japanese patient with idiopathic basal ganglia calcification-1 (IBGC1) associated with dopa-responsive parkinsonism Hum Genome Var **6**, 44 10.1038/s41439-019-0073-7

231. Ide, M., Ohnishi, T., Toyoshima, M., Balan, S., Maekawa, M., Shimamoto-Mitsuyama, C. *et al.* (2019) Excess hydrogen sulfide and polysulfides production underlies a schizophrenia pathophysiology EMBO Mol Med **11**, e10695 10.15252/emmm.201910695

232. Igeta, H., Watanabe, Y., Morikawa, R., Ikeda, M., Otsuka, I., Hoya, S. *et al.* (2019) Rare compound heterozygous missense SPATA7 variations and risk of schizophrenia; whole-exome sequencing in a consanguineous family with affected siblings, follow-up sequencing and a case-control study Neuropsychiatr Dis Treat **15**, 2353-2363 10.2147/NDT.S218773

233. Inoue, M., Uchino, S., Iida, A., Noguchi, S., Hayashi, S., Takahashi, T. *et al.* (2019) COX6A2 variants cause a muscle-specific cytochrome c oxidase deficiency Ann Neurol **86**, 193-202 10.1002/ana.25517

234. Ishihara, N., Inagaki, H., Miyake, M., Kawamura, Y., Yoshikawa, T., andKurahashi, H. (2019) A case of early onset life-threatening epilepsy associated with a novel ATP1A3 gene variant Brain Dev **41**, 285-291 10.1016/j.braindev.2018.10.008

235. Iso, M., Suzuki, M., Yanagi, K., Minowa, K., Sakurai, Y., Nakano, S. *et al.* (2019) The CFTR gene variants in Japanese children with idiopathic pancreatitis Hum Genome Var **6**, 17 10.1038/s41439-019-0049-7

236. Ito, T., Yamaguchi, T., Wakatsuki, T., Suzuki, T., Eguchi, H., Okazaki, Y. *et al.* (2019) The single-base-pair deletion, MSH2 c.2635-3delC affecting intron 15 splicing can be a cause of Lynch syndrome Jpn J Clin Oncol **49**, 477-480 10.1093/jjco/hyz031

237. Iwama, K., Mizuguchi, T., Takeshita, E., Nakagawa, E., Okazaki, T., Nomura, Y. *et al.* (2019) Genetic landscape

of Rett syndrome-like phenotypes revealed by whole exome sequencing J Med Genet **56**, 396-407 10.1136/jmedgenet-2018-105775

238. Iwanami, M., Oishi, A., Ogino, K., Seko, Y., Nishida-Shimizu, T., Yoshimura, N. *et al.* (2019) Five major sequence variants and copy number variants in the EYS gene account for one-third of Japanese patients with autosomal recessive and simplex retinitis pigmentosa Mol Vis **25**, 766-779, https://www.ncbi.nlm.nih.gov/pubmed/31814702
239. Kajii, T. S., Oka, A., Saito, F., Mitsui, J., andIida, J. (2019) Whole-exome sequencing in a Japanese pedigree implicates a rare non-synonymous single-nucleotide variant in BEST3 as a candidate for mandibular prognathism Bone **122**, 193-198 10.1016/j.bone.2019.03.004
240. Kakuta, Y., Kawai, Y., Naito, T., Hirano, A., Umeno, J., Fuyuno, Y. *et al.* (2019) A Genome-wide Association Study Identifying RAP1A as a Novel Susceptibility Gene for Crohn's Disease in Japanese Individuals J Crohns Colitis **13**, 648-658 10.1093/ecco-jcc/jjy197
241. Kameya, S., Fujinami, K., Ueno, S., Hayashi, T., Kuniyoshi, K., Ideta, R. *et al.* (2019) Phenotypical Characteristics of POC1B-Associated Retinopathy in Japanese Cohort: Cone Dystrophy With Normal Funduscopic Appearance Invest Ophthalmol Vis Sci **60**, 3432-3446 10.1167/iovs.19-26650
242. Kanzawa-Kiriyama, H., Jinam, T. A., Kawai, Y., Sato, T., Hosomichi, K., Tajima, A. *et al.* (2019) Late Jomon male and female genome sequences from the Funadomari site in Hokkaido, Japan Anthropological Science **127**, 83-108 10.1537/ase.190415
243. Kashimada, A., Hasegawa, S., Nomura, T., Shiraku, H., Moriyama, K., Suzuki, T. *et al.* (2019) Genetic analysis of undiagnosed ataxia-telangiectasia-like disorders Brain Dev **41**, 150-157 10.1016/j.braindev.2018.09.007
244. Kato, K., Miya, F., Hamada, N., Negishi, Y., Narumi-Kishimoto, Y., Ozawa, H. *et al.* (2019) MYCN de novo gain-of-function mutation in a patient with a novel megalencephaly syndrome J Med Genet **56**, 388-395 10.1136/jmedgenet-2018-105487
245. Kawabata, Y., Nishida, N., Awata, T., Kawasaki, E., Imagawa, A., Shimada, A. *et al.* (2019) Genome-Wide Association Study Confirming a Strong Effect of HLA and Identifying Variants in CSAD/lnc-ITGB7-1 on Chromosome 12q13.13 Associated With Susceptibility to Fulminant Type 1 Diabetes Diabetes **68**, 665-675 10.2337/db18-0314
246. Kiniwa, Y., Yasuda, J., Saito, S., Saito, R., Motoike, I. N., Danjoh, I. *et al.* (2019) Identification of genetic alterations in extramammary Paget disease using whole exome analysis J Dermatol Sci **94**, 229-235 10.1016/j.jdermsci.2019.03.006
247. Kishida, Y., Oishi, T., Sugino, T., Shiomi, A., Urakami, K., Kusuhara, M. *et al.* (2019) Associations Between Loss of ARID1A Expression and Clinicopathologic and Genetic Variables in T1 Early Colorectal Cancer Am J Clin Pathol **152**, 463-470 10.1093/ajcp/aqz062
248. Kitano, T., Kitajiri, S. I., Nishio, S. Y., andUsami, S. I. (2019) Detailed Clinical Features of Deafness Caused by a Claudin-14 Variant Int J Mol Sci **20**, 4579-4590 10.3390/ijms20184579
249. Kiyozumi, Y., Matsubayashi, H., Horiuchi, Y., Higashigawa, S., Oishi, T., Abe, M. *et al.* (2019) Germline mismatch repair gene variants analyzed by universal sequencing in Japanese cancer patients Cancer Med **8**, 5534-5543 10.1002/cam4.2432
250. Koh, K., Ichinose, Y., Ishiura, H., Nan, H., Mitsui, J., Takahashi, J. *et al.* (2019) PLA2G6-associated neurodegeneration presenting as a complicated form of hereditary spastic paraplegia Journal of human genetics **64**, 55-59 10.1038/s10038-018-0519-7

251. Kubo, A., Sasaki, T., Suzuki, H., Shiohama, A., Aoki, S., Sato, S. *et al.* (2019) Clonal Expansion of Second-Hit Cells with Somatic Recombinations or C>T Transitions Form Porokeratosis in MVD or MVK Mutant Heterozygotes J Invest Dermatol **139**, 2458-2466 e2459 10.1016/j.jid.2019.05.020
252. Kubota, D., Oishi, N., Gocho, K., Kikuchi, S., Yamaki, K., Igarashi, T. *et al.* (2019) Novel homozygous in-frame deletion of GNAT1 gene causes golden appearance of fundus and reduced scotopic ERGs similar to that in Oguchi disease in Japanese family Ophthalmic Genet **40**, 480-487 10.1080/13816810.2019.1686159
253. Kuroda, Y., Murakami, H., Enomoto, Y., Tsurusaki, Y., Takahashi, K., Mitsuzuka, K. *et al.* (2019) A novel gene (FAM20B encoding glycosaminoglycan xylosylkinase) for neonatal short limb dysplasia resembling Desbuquois dysplasia Clin Genet **95**, 713-717 10.1111/cge.13530
254. Kutsuma, T., Katagiri, S., Hayashi, T., Yoshitake, K., Iejima, D., Gekka, T. *et al.* (2019) Novel biallelic loss-of-function KCNV2 variants in cone dystrophy with supernormal rod responses Doc Ophthalmol **138**, 229-239 10.1007/s10633-019-09679-6
255. Lasher, D., Szabo, A., Masamune, A., Chen, J. M., Xiao, X., Whitcomb, D. C. *et al.* (2019) Protease-Sensitive Pancreatic Lipase Variants Are Associated With Early Onset Chronic Pancreatitis Am J Gastroenterol **114**, 974-983 10.14309/ajg.0000000000000051
256. Lee, J. S., Kanai, K., Suzuki, M., Kim, W. S., Yoo, H. S., Fu, Y. *et al.* (2019) Arylsulfatase A, a genetic modifier of Parkinson's disease, is an alpha-synuclein chaperone Brain **142**, 2845-2859 10.1093/brain/awz205
257. Lewis, J. D., Caldara, A. L., Zimmer, S. E., Stahley, S. N., Seybold, A., Strong, N. L. *et al.* (2019) The desmosome is a mesoscale lipid raft-like membrane domain Mol Biol Cell **30**, 1390-1405 10.1091/mbc.E18-10-0649
258. Low, S. K., Chin, Y. M., Ito, H., Matsuo, K., Tanikawa, C., Matsuda, K. *et al.* (2019) Identification of two novel breast cancer loci through large-scale genome-wide association study in the Japanese population Sci Rep **9**, 17332 10.1038/s41598-019-53654-9
259. Maeda-Katahira, A., Nakamura, N., Hayashi, T., Katagiri, S., Shimizu, S., Ohde, H. *et al.* (2019) Autosomal dominant optic atrophy with OPA1 gene mutations accompanied by auditory neuropathy and other systemic complications in a Japanese cohort Mol Vis **25**, 559-573, https://www.ncbi.nlm.nih.gov/pubmed/31673222
260. Maekawa, K., Nishio, S. Y., Abe, S., Goto, S. I., Honkura, Y., Iwasaki, S. *et al.* (2019) Mutational Spectrum and Clinical Features of Patients with LOXHD1 Variants Identified in an 8074 Hearing Loss Patient Cohort Genes (Basel) **10**, 735-754 10.3390/genes10100735
261. Makarious, M. B., Diez-Fairen, M., Krohn, L., Blauwendraat, C., Bandres-Ciga, S., Ding, J. *et al.* (2019) ARSA variants in alpha-synucleinopathies Brain **142**, e70 10.1093/brain/awz340
262. Miki, T., Yokota, O., Haraguchi, T., Ikeuchi, T., Zhu, B., Takenoshita, S. *et al.* (2019) Young adult-onset, very slowly progressive cognitive decline with spastic paraparesis in Alzheimer's disease with cotton wool plaques due to a novel presenilin1 G417S mutation Acta Neuropathol Commun **7**, 19 10.1186/s40478-019-0672-z
263. Minami, S. B., Nara, K., Mutai, H., Morimoto, N., Sakamoto, H., Takiguchi, T. *et al.* (2019) A clinical and genetic study of 16 Japanese families with Waardenburg syndrome Gene **704**, 86-90 10.1016/j.gene.2019.04.023
264. Misawa, K., Watanabe, H., Yokoseki, A., Wakasugi, M., Onodera, O., Narita, I. *et al.* (2019) Is the population of Sado Island genetically close to the population of western Japan? Hum Genome Var **6**, 26 10.1038/s41439-019-0058-6
265. Miura, S., Kosaka, K., Fujioka, R., Uchiyama, Y., Shimojo, T., Morikawa, T. *et al.* (2019) Spinocerebellar ataxia 27 with a novel nonsense variant (Lys177X) in FGF14 Eur J Med Genet **62**, 172-176 10.1016/j.ejmg.2018.07.005

266. Miyabayashi, T., Ochiai, T., Suzuki, N., Aoki, M., Inui, T., Okubo, Y. *et al.* (2019) A novel homozygous mutation of the TFG gene in a patient with early onset spastic paraplegia and later onset sensorimotor polyneuropathy Journal of human genetics **64**, 171-176 10.1038/s10038-018-0538-4

267. Miyado, M., Fukami, M., Takada, S., Terao, M., Nakabayashi, K., Hata, K. *et al.* (2019) Germline-Derived Gain-of-Function Variants of Gsalpha-Coding GNAS Gene Identified in Nephrogenic Syndrome of Inappropriate Antidiuresis J Am Soc Nephrol **30**, 877-889 10.1681/ASN.2018121268

268. Miyagawa, T., Hida, A., Shimada, M., Uehara, C., Nishino, Y., Kadotani, H. *et al.* (2019) A missense variant in PER2 is associated with delayed sleep-wake phase disorder in a Japanese population Journal of human genetics **64**, 1219-1225 10.1038/s10038-019-0665-6

269. Miyamichi, D., Nishina, S., Hosono, K., Yokoi, T., Kurata, K., Sato, M. *et al.* (2019) Retinal structure in Leber's congenital amaurosis caused by RPGRIP1 mutations Hum Genome Var **6**, 32 10.1038/s41439-019-0064-8

270. Miyamoto, S., Nakashima, M., Ohashi, T., Hiraide, T., Kurosawa, K., Yamamoto, T. *et al.* (2019) A case of de novo splice site variant in SLC35A2 showing developmental delays, spastic paraplegia, and delayed myelination Mol Genet Genomic Med **7**, e814 10.1002/mgg3.814

271. Miyamoto-Mikami, E., Miyamoto, N., Kumagai, H., Hirata, K., Kikuchi, N., Zempo, H. *et al.* (2019) COL5A1 rs12722 polymorphism is not associated with passive muscle stiffness and sports-related muscle injury in Japanese athletes BMC Med Genet **20**, 192 10.1186/s12881-019-0928-2

272. Mizobuchi, K., Katagiri, S., Hayashi, T., Yoshitake, K., Fujinami, K., Kuniyoshi, K. *et al.* (2019) Clinical findings of end-stage retinitis pigmentosa with a homozygous PDE6A variant (p.R653X) Am J Ophthalmol Case Rep **13**, 110-115 10.1016/j.ajoc.2018.12.019

273. Mizuguchi, T., Suzuki, T., Abe, C., Umemura, A., Tokunaga, K., Kawai, Y. *et al.* (2019) A 12-kb structural variation in progressive myoclonic epilepsy was newly identified by long-read whole-genome sequencing Journal of human genetics **64**, 359-368 10.1038/s10038-019-0569-5

274. Mizuno, K., Akamatsu, S., Sumiyoshi, T., Wong, J. H., Fujita, M., Maejima, K. *et al.* (2019) eVIDENCE: a practical variant filtering for low-frequency variants detection in cell-free DNA Sci Rep **9**, 15017 10.1038/s41598-019-51459-4

275. Mori, M., Hira, A., Yoshida, K., Muramatsu, H., Okuno, Y., Shiraishi, Y. *et al.* (2019) Pathogenic mutations identified by a multimodality approach in 117 Japanese Fanconi anemia patients Haematologica **104**, 1962-1973 10.3324/haematol.2018.207241

276. Morimoto, Y., Yoshida, S., Kinoshita, A., Satoh, C., Mishima, H., Yamaguchi, N. *et al.* (2019) Nonsense mutation in CFAP43 causes normal-pressure hydrocephalus with ciliary abnormalities Neurology **92**, e2364-e2374 10.1212/WNL.0000000000007505

277. Moriwaki, T., Yamazaki, N., So, T., Kosuga, M., Miyazaki, O., Narumi-Kishimoto, Y. *et al.* (2019) Normal early development in siblings with novel compound heterozygous variants in ASPM Hum Genome Var **6**, 56 10.1038/s41439-019-0088-0

278. Motoi, Y., Ito, Z., Suzuki, S., Takami, S., Matsuo, K., Sato, M. *et al.* (2019) FADS2 and ELOVL6 mutation frequencies in Japanese Crohn's disease patients Drug Discov Ther **13**, 354-359 10.5582/ddt.2019.01081

279. Mun, D. G., Bhin, J., Kim, S., Kim, H., Jung, J. H., Jung, Y. *et al.* (2019) Proteogenomic Characterization of Human Early-Onset Gastric Cancer Cancer Cell **35**, 111-124 e110 10.1016/j.ccell.2018.12.003

280. Nagahara, Y., Tsujikawa, M., Takigawa, T., Xu, P., Kai, C., Kawasaki, S. *et al.* (2019) A novel mutation in gelatinous drop-like corneal dystrophy and functional analysis Hum Genome Var **6**, 33 10.1038/s41439-019-0060-z

281. Nakashima, M., Tohyama, J., Nakagawa, E., Watanabe, Y., Siew, C. G., Kwong, C. S. *et al.* (2019) Identification of de novo CSNK2A1 and CSNK2B variants in cases of global developmental delay with seizures Journal of human genetics **64**, 313-322 10.1038/s10038-018-0559-z

282. Nan, H., Ichinose, Y., Tanaka, M., Koh, K., Ishiura, H., Mitsui, J. *et al.* (2019) UBAP1 mutations cause juvenile-onset hereditary spastic paraplegias (SPG80) and impair UBAP1 targeting to endosomes Journal of human genetics **64**, 1055-1065 10.1038/s10038-019-0670-9

283. Nikopoulos, K., Cisarova, K., Quinodoz, M., Koskiniemi-Kuendig, H., Miyake, N., Farinelli, P. *et al.* (2019) A frequent variant in the Japanese population determines quasi-Mendelian inheritance of rare retinal ciliopathy Nat Commun **10**, 2884 10.1038/s41467-019-10746-4

284. Nishiyama, T., Nakatochi, M., Goto, A., Iwasaki, M., Hachiya, T., Sutoh, Y. *et al.* (2019) Genome-wide association meta-analysis and Mendelian randomization analysis confirm the influence of ALDH2 on sleep durationin the Japanese population Sleep **42**, 10.1093/sleep/zsz046

285. Noda, Y., Kato, T., Kato, A., Nishizawa, H., Miyazaki, J., Ito, M. *et al.* (2019) Potentially effective method for fetal gender determination by noninvasive prenatal testing for X-linked disease Congenit Anom (Kyoto) **59**, 88-92 10.1111/cga.12302

286. Ohnishi, T., Balan, S., Toyoshima, M., Maekawa, M., Ohba, H., Watanabe, A. *et al.* (2019) Investigation of betaine as a novel psychotherapeutic for schizophrenia Ebiomedicine **45**, 432-446 10.1016/j.ebiom.2019.05.062

287. Ohtsuka, K., Ohnishi, H., Fujiwara, M., Morii, T., Matsushima, S., Ogura, W. *et al.* (2019) Predisposition to Lung Adenocarcinoma in a Family Harboring the Germline EGFR V843I Mutation JCO Precis Oncol **3**, 10.1200/PO.19.00104

288. Okano, S., Makita, Y., Katada, A., Harabuchi, Y., Kohmoto, T., Naruto, T. *et al.* (2019) Novel compound heterozygous CDH23 variants in a patient with Usher syndrome type I Hum Genome Var **6**, 8 10.1038/s41439-019-0037-y

289. Ono, S., Matsuda, J., Watanabe, E., Akaike, H., Teranishi, H., Miyata, I. *et al.* (2019) Novel neuroblastoma amplified sequence (NBAS) mutations in a Japanese boy with fever-triggered recurrent acute liver failure Hum Genome Var **6**, 2 10.1038/s41439-018-0035-5

290. Otomo, N., Mizumoto, S., Lu, H. F., Takeda, K., Campos-Xavier, B., Mittaz-Crettol, L. *et al.* (2019) Identification of novel LFNG mutations in spondylocostal dysostosis Journal of human genetics **64**, 261-264 10.1038/s10038-018-0548-2

291. Oyama, R., Kito, F., Takahashi, M., Sakumoto, M., Shiozawa, K., Qiao, Z. *et al.* (2019) Establishment and characterization of a novel dedifferentiated chondrosarcoma cell line, NCC-dCS1-C1 Hum Cell **32**, 202-213 10.1007/s13577-018-00232-2

292. Ozeki, M., Aoki, Y., Nozawa, A., Yasue, S., Endo, S., Hori, Y. *et al.* (2019) Detection of NRAS mutation in cell-free DNA biological fluids from patients with kaposiform lymphangiomatosis Orphanet J Rare Dis **14**, 215 10.1186/s13023-019-1191-5

293. Saiki, S., Sasazawa, Y., Fujimaki, M., Kamagata, K., Kaga, N., Taka, H. *et al.* (2019) A metabolic profile of polyamines in parkinson disease: A promising biomarker Ann Neurol **86**, 251-263 10.1002/ana.25516

294. Sakaguchi, A., Yamashita, Y., Ishii, T., Uehara, T., Kosaki, K., Takahashi, T. *et al.* (2019) Further evidence of a causal association between AGO1, a critical regulator of microRNA formation, and intellectual disability/autism spectrum disorder Eur J Med Genet **62**, 103537 10.1016/j.ejmg.2018.09.004

295. Sakoh, T., Sekine, A., Mori, T., Mizuno, H., Kawada, M., Hiramatsu, R. *et al.* (2019) A familial case of

pseudohypoaldosteronism type II (PHA2) with a novel mutation (D564N) in the acidic motif in WNK4 Mol Genet Genomic Med **7**, e705 10.1002/mgg3.705

296. Sekimizu, M., Yoshida, A., Mitani, S., Asano, N., Hirata, M., Kubo, T. *et al.* (2019) Frequent mutations of genes encoding vacuolar H(+) -ATPase components in granular cell tumors Genes Chromosomes Cancer **58**, 373-380 10.1002/gcc.22727
297. Sekine, S. I., Kaneko, M., Tanaka, M., Ninomiya, Y., Kurita, H., Inden, M. *et al.* (2019) Functional evaluation of PDGFB-variants in idiopathic basal ganglia calcification, using patient-derived iPS cells Sci Rep **9**, 5698 10.1038/s41598-019-42115-y
298. Shimizu, H., Watanabe, S., Kinoshita, A., Mishima, H., Nishimura, G., Moriuchi, H. *et al.* (2019) Identification of a homozygous frameshift variant in RFLNA in a patient with a typical phenotype of spondylocarpotarsal synostosis syndrome Journal of human genetics **64**, 467-471 10.1038/s10038-019-0581-9
299. Shimizu, M., Yoda, H., Nakakuki, K., Saso, A., Saito, I., Hishinuma, E. *et al.* (2019) Genetic variants of flavin-containing monooxygenase 3 (FMO3) derived from Japanese subjects with the trimethylaminuria phenotype and whole-genome sequence data from a large Japanese database Drug Metab Pharmacokinet **34**, 334-339 10.1016/j.dmpk.2019.06.001
300. Sugiyama, K., Moteki, H., Kitajiri, S. I., Kitano, T., Nishio, S. Y., Yamaguchi, T. *et al.* (2019) Mid-Frequency Hearing Loss Is Characteristic Clinical Feature of OTOA-Associated Hearing Loss Genes (Basel) **10**, 715-731 10.3390/genes10090715
301. Sunami, K., Ichikawa, H., Kubo, T., Kato, M., Fujiwara, Y., Shimomura, A. *et al.* (2019) Feasibility and utility of a panel testing for 114 cancer-associated genes in a clinical setting: A hospital-based study Cancer Sci **110**, 1480-1490 10.1111/cas.13969
302. Suzuki, H., Yoshida, T., Morisada, N., Uehara, T., Kosaki, K., Sato, K. *et al.* (2019) De novo NSF mutations cause early infantile epileptic encephalopathy Ann Clin Transl Neurol **6**, 2334-2339 10.1002/acn3.50917
303. Suzuki, K., Akiyama, M., Ishigaki, K., Kanai, M., Hosoe, J., Shojima, N. *et al.* (2019) Identification of 28 new susceptibility loci for type 2 diabetes in the Japanese population Nature genetics **51**, 379-386 10.1038/s41588-018-0332-4
304. Takata, A., Nakashima, M., Saitsu, H., Mizuguchi, T., Mitsuhashi, S., Takahashi, Y. *et al.* (2019) Comprehensive analysis of coding variants highlights genetic complexity in developmental and epileptic encephalopathy Nat Commun **10**, 2506 10.1038/s41467-019-10482-9
305. Takata, R., Takahashi, A., Fujita, M., Momozawa, Y., Saunders, E. J., Yamada, H. *et al.* (2019) 12 new susceptibility loci for prostate cancer identified by genome-wide association study in Japanese population Nat Commun **10**, 4422 10.1038/s41467-019-12267-6
306. Takayama, K., Ohno, S., Ding, W. G., Ashihara, T., Fukumoto, D., Wada, Y. *et al.* (2019) A de novo gain-of-function KCND3 mutation in early repolarization syndrome Heart Rhythm **16**, 1698-1706 10.1016/j.hrthm.2019.05.033
307. Takenaka, S., Kuroda, Y., Ohta, S., Mizuno, Y., Hiwatari, M., Miyatake, S. *et al.* (2019) A Japanese patient with RAD51-associated Fanconi anemia Am J Med Genet A **179**, 900-902 10.1002/ajmg.a.61130
308. Takenouchi, T., Shimada, H., Uehara, T., Kanai, Y., Takahashi, T., andKosaki, K. (2019) A paradoxical thrombogenic mutation in factor II at the target site of arthropod bleeding toxin Eur J Med Genet **62**, 93-95 10.1016/j.ejmg.2018.06.003
309. Takenouchi, T., Wei, F. Y., Suzuki, H., Uehara, T., Takahashi, T., Okazaki, Y. *et al.* (2019) Noninvasive diagnosis

of TRIT1-related mitochondrial disorder by measuring i(6) A37 and ms(2) i(6) A37 modifications in tRNAs from blood and urine samples Am J Med Genet A **179**, 1609-1614 10.1002/ajmg.a.61211

310. Terashima, T., Umemoto, K., Takahashi, H., Hosoi, H., Takai, E., Kondo, S. *et al.* (2019) Germline mutations in cancer-predisposition genes in patients with biliary tract cancer Oncotarget **10**, 5949-5957 10.18632/oncotarget.27224
311. Tozawa, Y., Abdrabou, S., Nogawa-Chida, N., Nishiuchi, R., Ishida, T., Suzuki, Y. *et al.* (2019) A deep intronic mutation of c.1166-285 T > G in SLC46A1 is shared by four unrelated Japanese patients with hereditary folate malabsorption (HFM) Clin Immunol **208**, 108256 10.1016/j.clim.2019.108256
312. Tsukamoto, M., Yamashita, M., Nishi, T., andNakagawa, H. (2019) A Human ABC Transporter ABCC4 Gene SNP (rs11568658, 559 G > T, G187W) Reduces ABCC4-Dependent Drug Resistance Cells **8**, 39 10.3390/cells8010039
313. Tsumura, K., Arai, E., Tian, Y., Shibuya, A., Nishihara, H., Yotani, T. *et al.* (2019) Establishment of permutation for cancer risk estimation in the urothelium based on genome-wide DNA methylation analysis Carcinogenesis **40**, 1308-1319 10.1093/carcin/bgz112
314. Udagawa, C., Horinouchi, H., Shiraishi, K., Kohno, T., Okusaka, T., Ueno, H. *et al.* (2019) Whole genome sequencing to identify predictive markers for the risk of drug-induced interstitial lung disease PLoS One **14**, e0223371 10.1371/journal.pone.0223371
315. Uehara, T., Suzuki, H., Okamoto, N., Kondoh, T., Ahmad, A., O'Connor, B. C. *et al.* (2019) Pathogenetic basis of Takenouchi-Kosaki syndrome: Electron microscopy study using platelets in patients and functional studies in a Caenorhabditis elegans model Sci Rep **9**, 4418 10.1038/s41598-019-40988-7
316. Ueno, S., Inooka, D., Meinert, M., Ito, Y., Tsunoda, K., Fujinami, K. *et al.* (2019) Three cases of acute-onset bilateral photophobia Jpn J Ophthalmol **63**, 172-180 10.1007/s10384-018-00649-0
317. Umeki, I., Niihori, T., Abe, T., Kanno, S. I., Okamoto, N., Mizuno, S. *et al.* (2019) Delineation of LZTR1 mutation-positive patients with Noonan syndrome and identification of LZTR1 binding to RAF1-PPP1CB complexes Hum Genet **138**, 21-35 10.1007/s00439-018-1951-7
318. Yamada, N., Asano, Y., Fujita, M., Yamazaki, S., Inanobe, A., Matsuura, N. *et al.* (2019) Mutant KCNJ3 and KCNJ5 Potassium Channels as Novel Molecular Targets in Bradyarrhythmias and Atrial Fibrillation Circulation **139**, 2157-2169 10.1161/CIRCULATIONAHA.118.036761
319. Yamoto, K., Saitsu, H., Nishimura, G., Kosaki, R., Takayama, S., Haga, N. *et al.* (2019) Comprehensive clinical and molecular studies in split-hand/foot malformation: identification of two plausible candidate genes (LRP6 and UBA2) Eur J Hum Genet **27**, 1845-1857 10.1038/s41431-019-0473-7
320. Yoshimura, A., Yuan, J. H., Hashiguchi, A., Ando, M., Higuchi, Y., Nakamura, T. *et al.* (2019) Genetic profile and onset features of 1005 patients with Charcot-Marie-Tooth disease in Japan J Neurol Neurosurg Psychiatry **90**, 195-202 10.1136/jnnp-2018-318839
321. Zar Kyaw, T., Yamaguchi, S., Imai, C., Uematsu, M., andSato, N. (2019) The utility of post-test newborn blood spot screening cards for epigenetic association analyses: association between HIF3A methylation and birth weight-for-gestational age Journal of human genetics **64**, 795-801 10.1038/s10038-019-0621-5
322. Zhang, C., Gao, Y., Ning, Z., Lu, Y., Zhang, X., Liu, J. *et al.* (2019) PGG.SNV: understanding the evolutionary and medical implications of human single nucleotide variations in diverse populations Genome Biol **20**, 215 10.1186/s13059-019-1838-5
323. Zhang, L., Liu, L., Ma, M., Cheng, S., Cheng, B., Li, P. *et al.* (2019) Integrative analysis of transcriptome-wide

association study data and mRNA expression profiles identified candidate genes and pathways associated with atrial fibrillation Heart Vessels **34**, 1882-1888 10.1007/s00380-019-01418-w

324. Alsabban, A. H., Morikawa, M., Tanaka, Y., Takei, Y., andHirokawa, N. (2020) Kinesin Kif3b mutation reduces NMDAR subunit NR2A trafficking and causes schizophrenia-like phenotypes in mice EMBO J **39**, e101090 10.15252/embj.2018101090
325. Borna, N. N., Kishita, Y., Abe, J., Furukawa, T., Ogawa-Tominaga, M., Fushimi, T. *et al.* (2020) NAD(P)HX dehydratase protein-truncating mutations are associated with neurodevelopmental disorder exacerbated by acute illness Brain **143**, e54 10.1093/brain/awaa130
326. Curry, J. N., Saurette, M., Askari, M., Pei, L., Filla, M. B., Beggs, M. R. *et al.* (2020) Claudin-2 deficiency associates with hypercalciuria in mice and human kidney stone disease J Clin Invest **130**, 1948-1960 10.1172/JCI127750
327. Daida, K., Funayama, M., Li, Y., Yoshino, H., Hayashida, A., Ikeda, A. *et al.* (2020) Identification of Disease-Associated Variants by Targeted Gene Panel Resequencing in Parkinson's Disease Front Neurol **11**, 576465 10.3389/fneur.2020.576465
328. Fujinami, K., Liu, X., Ueno, S., Mizota, A., Shinoda, K., Kuniyoshi, K. *et al.* (2020) RP2-associated retinal disorder in a Japanese cohort: Report of novel variants and a literature review, identifying a genotype-phenotype association Am J Med Genet C Semin Med Genet **184**, 675-693 10.1002/ajmg.c.31830
329. Fujinami, K., Oishi, A., Yang, L., Arno, G., Pontikos, N., Yoshitake, K. *et al.* (2020) Clinical and genetic characteristics of 10 Japanese patients with PROM1-associated retinal disorder: A report of the phenotype spectrum and a literature review in the Japanese population Am J Med Genet C Semin Med Genet **184**, 656-674 10.1002/ajmg.c.31826
330. Fujinami-Yokokawa, Y., Fujinami, K., Kuniyoshi, K., Hayashi, T., Ueno, S., Mizota, A. *et al.* (2020) Clinical and Genetic Characteristics of 18 Patients from 13 Japanese Families with CRX-associated retinal disorder: Identification of Genotype-phenotype Association Sci Rep **10**, 9531 10.1038/s41598-020-65737-z
331. Fujita, H., Sasaki, T., Miyamoto, T., Akutsu, S. N., Sato, S., Mori, T. *et al.* (2020) Premature aging syndrome showing random chromosome number instabilities with CDC20 mutation Aging Cell **19**, e13251 10.1111/acel.13251
332. Fukaishi, T., Minami, I., Masuda, S., Miyachi, Y., Tsujimoto, K., Izumiyama, H. *et al.* (2020) A case of generalized lipodystrophy-associated progeroid syndrome treated by leptin replacement with short and long-term monitoring of the metabolic and endocrine profiles Endocr J **67**, 211-218 10.1507/endocrj.EJ19-0226
333. Fukuda, T., Hiraide, T., Yamoto, K., Nakashima, M., Kawai, T., Yanagi, K. *et al.* (2020) Exome reports A de novo GNB2 variant associated with global developmental delay, intellectual disability, and dysmorphic features Eur J Med Genet **63**, 103804 10.1016/j.ejmg.2019.103804
334. Hashimoto, N., Dateki, S., Suzuki, E., Tsuchihashi, T., Isobe, A., Banno, S. *et al.* (2020) Compound heterozygous variants in the ABCG8 gene in a Japanese girl with sitosterolemia Hum Genome Var **7**, 25 10.1038/s41439-020-00112-y
335. Hayashi, H., Tanishima, S., Fujii, K., Mori, R., Okada, C., Yanagita, E. *et al.* (2020) Clinical impact of a cancer genomic profiling test using an in-house comprehensive targeted sequencing system Cancer Sci **111**, 3926-3937 10.1111/cas.14608
336. Hayashi, T., Hosono, K., Kubo, A., Kurata, K., Katagiri, S., Mizobuchi, K. *et al.* (2020) Long-term observation of a Japanese mucolipidosis IV patient with a novel homozygous p.F313del variant of MCOLN1 Am J Med Genet A **182**, 1500-1505 10.1002/ajmg.a.61575

337. Hayashi, T., Hosono, K., Kurata, K., Katagiri, S., Mizobuchi, K., Ueno, S. *et al.* (2020) Coexistence of GNAT1 and ABCA4 variants associated with Nougaret-type congenital stationary night blindness and childhood-onset cone-rod dystrophy Doc Ophthalmol **140**, 147-157 10.1007/s10633-019-09727-1

338. Hiraide, T., Kataoka, M., Suzuki, H., Aimi, Y., Chiba, T., Isobe, S. *et al.* (2020) Poor outcomes in carriers of the RNF213 variant (p.Arg4810Lys) with pulmonary arterial hypertension J Heart Lung Transplant **39**, 103-112 10.1016/j.healun.2019.08.022

339. Hiraide, T., Nakashima, M., Ikeda, T., Tanaka, D., Osaka, H., andSaitsu, H. (2020) Identification of a deep intronic POLR3A variant causing inclusion of a pseudoexon derived from an Alu element in Pol III-related leukodystrophy Journal of human genetics **65**, 921-925 10.1038/s10038-020-0786-y

340. Hiraide, T., Watanabe, S., Matsubayashi, T., Yanagi, K., Nakashima, M., Ogata, T. *et al.* (2020) A de novo TOP2B variant associated with global developmental delay and autism spectrum disorder Mol Genet Genomic Med **8**, e1145 10.1002/mgg3.1145

341. Hirose, S., Murakami, N., Takahashi, K., Kuno, I., Takayanagi, D., Asami, Y. *et al.* (2020) Genomic alterations in STK11 can predict clinical outcomes in cervical cancer patients Gynecol Oncol **156**, 203-210 10.1016/j.ygyno.2019.10.022

342. Hirotsu, Y., Schmidt-Edelkraut, U., Nakagomi, H., Sakamoto, I., Hartenfeller, M., Narang, R. *et al.* (2020) Consolidated BRCA1/2 Variant Interpretation by MH BRCA Correlates with Predicted PARP Inhibitor Efficacy Association by MH Guide Int J Mol Sci **21**, 3895 10.3390/ijms21113895

343. Hori, H., Itoh, M., Yoshida, F., Lin, M., Niwa, M., Hakamata, Y. *et al.* (2020) The BDNF Val66Met polymorphism affects negative memory bias in civilian women with PTSD Sci Rep **10**, 3151 10.1038/s41598-020-60096-1

344. Hori, M., Takahashi, A., Son, C., Ogura, M., andHarada-Shiba, M. (2020) The benign c.344G > A: p.(Arg115His) variant in the LDLR gene interpreted from a pedigree-based genetic analysis of familial hypercholesterolemia Lipids Health Dis **19**, 62 10.1186/s12944-020-01252-4

345. Horiuchi, Y., Ichikawa, T., Ohnishi, T., Iwayama, Y., Toriumi, K., Miyashita, M. *et al.* (2020) LDB2 locus disruption on 4p16.1 as a risk factor for schizophrenia and bipolar disorder Hum Genome Var **7**, 31 10.1038/s41439-020-00117-7

346. Hosseini, D. K., Ataikia, S., Hosseini, H. K., Han, B., andSun, H. (2020) Association of polymorphisms in ADAMTS-7 gene with the susceptibility to coronary artery disease - a systematic review and meta-analysis Aging (Albany NY) **12**, 20915-20923 10.18632/aging.104118

347. Igarashi, M., Masunaga, Y., Hasegawa, Y., Kinjo, K., Miyado, M., Saitsu, H. *et al.* (2020) Nonsense-associated altered splicing of MAP3K1 in two siblings with 46,XY disorders of sex development Sci Rep **10**, 17375 10.1038/s41598-020-74405-1

348. Ikeda, S., Akamatsu, C., Ijuin, A., Nagashima, A., Sasaki, M., Mochizuki, A. *et al.* (2020) Prenatal diagnosis of Fraser syndrome caused by novel variants of FREM2 Hum Genome Var **7**, 32 10.1038/s41439-020-00119-5

349. Ikeya, A., Nakashima, M., Yamashita, M., Kakizawa, K., Okawa, Y., Saitsu, H. *et al.* (2020) CCNB2 and AURKA overexpression may cause atypical mitosis in Japanese cortisol-producing adrenocortical carcinoma with TP53 somatic variant PLoS One **15**, e0231665 10.1371/journal.pone.0231665

350. Inoue, T., Nakamura, A., Iwahashi-Odano, M., Tanase-Nakao, K., Matsubara, K., Nishioka, J. *et al.* (2020) Contribution of gene mutations to Silver-Russell syndrome phenotype: multigene sequencing analysis in 92 etiology-unknown patients Clin Epigenetics **12**, 86 10.1186/s13148-020-00865-x

351. Ishigaki, K., Akiyama, M., Kanai, M., Takahashi, A., Kawakami, E., Sugishita, H. *et al.* (2020) Large-scale

genome-wide association study in a Japanese population identifies novel susceptibility loci across different diseases Nature genetics **52**, 669-679 10.1038/s41588-020-0640-3

352. Ishikawa, M., Tada, Y., Tanaka, H., Morii, W., Inaba, M., Takada, H. *et al.* (2020) A Family with Gitelman Syndrome with Asymptomatic Phenotypes while Carrying Reported SLC12A3 Mutations Case Rep Nephrol Dial **10**, 71-78 10.1159/000507845
353. Ishizuka, K., Yoshida, T., Kawabata, T., Imai, A., Mori, H., Kimura, H. *et al.* (2020) Functional characterization of rare NRXN1 variants identified in autism spectrum disorders and schizophrenia J Neurodev Disord **12**, 25 10.1186/s11689-020-09325-2
354. Iwahashi-Odano, M., Fujisawa, Y., Ogata, T., Nakashima, S., Muramatsu, M., andNarumi, S. (2020) Identification and functional characterization of a novel PAX8 mutation (p.His39Pro) causing familial thyroid hypoplasia Clin Pediatr Endocrinol **29**, 173-178 10.1297/cpe.29.173
355. Izumi, R., Takahashi, T., Suzuki, N., Niihori, T., Ono, H., Nakamura, N. *et al.* (2020) The genetic profile of dysferlinopathy in a cohort of 209 cases: Genotype-phenotype relationship and a hotspot on the inner DysF domain Hum Mutat **41**, 1540-1554 10.1002/humu.24036
356. Jia, X., Yamamura, T., Gbadegesin, R., McNulty, M. T., Song, K., Nagano, C. *et al.* (2020) Common risk variants in NPHS1 and TNFSF15 are associated with childhood steroid-sensitive nephrotic syndrome Kidney international **98**, 1308-1322 10.1016/j.kint.2020.05.029
357. Kakuta, Y., Izumiyama, Y., Okamoto, D., Nakano, T., Ichikawa, R., Naito, T. *et al.* (2020) High-resolution melt analysis enables simple genotyping of complicated polymorphisms of codon 18 rendering the NUDT15 diplotype J Gastroenterol **55**, 67-77 10.1007/s00535-019-01638-x
358. Kanamori, T., Sanada, M., Ri, M., Ueno, H., Nishijima, D., Yasuda, T. *et al.* (2020) Genomic analysis of multiple myeloma using targeted capture sequencing in the Japanese cohort Br J Haematol **191**, 755-763 10.1111/bjh.16720
359. Kanchanasevee, C., Sriwattanapong, K., Theerapanon, T., Thaweesapphithak, S., Chetruengchai, W., Porntaveetus, T. *et al.* (2020) Phenotypic and Genotypic Features of Thai Patients With Nonsyndromic Tooth Agenesis and WNT10A Variants Front Physiol **11**, 573214 10.3389/fphys.2020.573214
360. Kaneyasu, T., Mori, S., Yamauchi, H., Ohsumi, S., Ohno, S., Aoki, D. *et al.* (2020) Prevalence of disease-causing genes in Japanese patients with BRCA1/2-wildtype hereditary breast and ovarian cancer syndrome NPJ Breast Cancer **6**, 25 10.1038/s41523-020-0163-1
361. Kasamo, K., Nakamura, M., Daimou, Y., andSano, A. (2020) A PRIMPOL mutation and variants in multiple genes may contribute to phenotypes in a familial case with chronic progressive external ophthalmoplegia symptoms Neurosci Res **157**, 58-63 10.1016/j.neures.2019.07.006
362. Katagiri, S., Hayashi, T., Nakamura, M., Mizobuchi, K., Gekka, T., Komori, S. *et al.* (2020) RDH5-Related Fundus Albipunctatus in a Large Japanese Cohort Invest Ophthalmol Vis Sci **61**, 53 10.1167/iovs.61.3.53
363. Katagiri, S., Hosono, K., Hayashi, T., Murai, N., Wake, E., Miyata, I. *et al.* (2020) Novel biallelic splice-site BBS1 variants in Bardet-Biedle syndrome: a case report of the first Japanese patient Doc Ophthalmol **141**, 77-88 10.1007/s10633-020-09752-5
364. Kato, H., Kushima, I., Mori, D., Yoshimi, A., Aleksic, B., Nawa, Y. *et al.* (2020) Rare genetic variants in the gene encoding histone lysine demethylase 4C (KDM4C) and their contributions to susceptibility to schizophrenia and autism spectrum disorder Transl Psychiatry **10**, 421 10.1038/s41398-020-01107-7
365. Kawasaki, A., Namba, N., Sada, K. E., Hirano, F., Kobayashi, S., Nagasaka, K. *et al.* (2020) Association of TERT

and DSP variants with microscopic polyangiitis and myeloperoxidase-ANCA positive vasculitis in a Japanese population: a genetic association study Arthritis Res Ther **22**, 246 10.1186/s13075-020-02347-0

366. Kishita, Y., Shimura, M., Kohda, M., Akita, M., Imai-Okazaki, A., Yatsuka, Y. *et al.* (2020) A novel homozygous variant in MICOS13/QIL1 causes hepato-encephalopathy with mitochondrial DNA depletion syndrome Mol Genet Genomic Med **8**, e1427 10.1002/mgg3.1427
367. Koh, K., Ishiura, H., Shimazaki, H., Tsutsumiuchi, M., Ichinose, Y., Nan, H. *et al.* (2020) VPS13D-related disorders presenting as a pure and complicated form of hereditary spastic paraplegia Mol Genet Genomic Med **8**, e1108 10.1002/mgg3.1108
368. Kosaki, R., Kubota, M., Uehara, T., Suzuki, H., Takenouchi, T., andKosaki, K. (2020) Consecutive medical exome analysis at a tertiary center: Diagnostic and health-economic outcomes Am J Med Genet A **182**, 1601-1607 10.1002/ajmg.a.61589
369. Kubota, D., Matsumoto, K., Hayashi, M., Oishi, N., Gocho, K., Yamaki, K. *et al.* (2020) High-resolution photoreceptor imaging analysis of patients with autosomal dominant retinitis pigmentosa (adRP) caused by HK1 mutation Ophthalmic Genet **41**, 629-638 10.1080/13816810.2020.1810284
370. Kubota, T., Wu, F., Vicart, S., Nakaza, M., Sternberg, D., Watanabe, D. *et al.* (2020) Hypokalaemic periodic paralysis with a charge-retaining substitution in the voltage sensor Brain Commun **2**, fcaa103 10.1093/braincomms/fcaa103
371. Kuniyoshi, K., Hayashi, T., Kameya, S., Katagiri, S., Mizobuchi, K., Tachibana, T. *et al.* (2020) Clinical Course and Electron Microscopic Findings in Lymphocytes of Patients with DRAM2-Associated Retinopathy Int J Mol Sci **21**, 1331-1345 10.3390/ijms21041331
372. Kutsuwada, Y. (2020) Examination of method to detect silent allele on D19S433 locus Japanese Journal of Forensic Science and Technology **25**, 91-104 10.3408/jafst.766
373. Li, H., Yuan, S., Minegishi, Y., Suga, A., Yoshitake, K., Sheng, X. *et al.* (2020) Novel mutations in malonyl-CoA-acyl carrier protein transacylase provoke autosomal recessive optic neuropathy Human molecular genetics **29**, 444-458 10.1093/hmg/ddz311
374. Li, Y., Ikeda, A., Yoshino, H., Oyama, G., Kitani, M., Daida, K. *et al.* (2020) Clinical characterization of patients with leucine-rich repeat kinase 2 genetic variants in Japan Journal of human genetics **65**, 771-781 10.1038/s10038-020-0772-4
375. Liu, X., Fujinami, K., Kuniyoshi, K., Kondo, M., Ueno, S., Hayashi, T. *et al.* (2020) Clinical and Genetic Characteristics of 15 Affected Patients From 12 Japanese Families with GUCY2D-Associated Retinal Disorder Transl Vis Sci Technol **9**, 2 10.1167/tvst.9.6.2
376. Maeda, Y., Sasaki, A., Kasai, S., Goto, S., Nishio, S. Y., Sawada, K. *et al.* (2020) Prevalence of the mitochondrial 1555 A>G and 1494 C>T mutations in a community-dwelling population in Japan Hum Genome Var **7**, 27 10.1038/s41439-020-00115-9
377. Matsukawa, T., Shoji, H., Urasaki, Y., Ishiura, H., Mitsui, J., Oguri, S. *et al.* (2020) Novel variant of CSF1R in sporadic case with early-onset cognitive impairment Neurology and Clinical Neuroscience **8**, 430-432 10.1111/ncn3.12452
378. Matsunaga, H., Ito, K., Akiyama, M., Takahashi, A., Koyama, S., Nomura, S. *et al.* (2020) Transethnic Meta-Analysis of Genome-Wide Association Studies Identifies Three New Loci and Characterizes Population-Specific Differences for Coronary Artery Disease Circ Genom Precis Med **13**, e002670 10.1161/CIRCGEN.119.002670
379. Matsushita, I., Morita, H., andKondo, H. (2020) Autosomal dominant foveal hypoplasia without visible macular

abnormalities and PAX6 mutations Jpn J Ophthalmol **64**, 635-641 10.1007/s10384-020-00766-9

380. Mitsutake, A., Matsukawa, T., Porto, K. J. L., Sato, T., Katsumata, J., Seki, T. *et al.* (2020) A Japanese family with primary familial brain calcification presenting with paroxysmal kinesigenic dyskinesia - A comprehensive mutational analysis J Neurol Sci **418**, 117091 10.1016/j.jns.2020.117091
381. Mukai, M., Mizuta, I., Watanabe-Hosomi, A., Koizumi, T., Matsuura, J., Hamano, A. *et al.* (2020) Genotype-phenotype correlations and effect of mutation location in Japanese CADASIL patients Journal of human genetics **65**, 637-646 10.1038/s10038-020-0751-9
382. Murakami, H., Tamura, N., Enomoto, Y., Shimasaki, K., Kurosawa, K., andHanada, K. (2020) Intellectual disability-associated gain-of-function mutations in CERT1 that encodes the ceramide transport protein CERT PLoS One **15**, e0243980 10.1371/journal.pone.0243980
383. Nagasaki, K., Shibata, N., Nyuzuki, H., Sasaki, S., Ogawa, Y., Soda, S. *et al.* (2020) A Japanese Family with DICER1 Syndrome Found in Childhood-Onset Multinodular Goitre Horm Res Paediatr **93**, 477-482 10.1159/000511140
384. Nagasawa, S., Ikeda, K., Horie-Inoue, K., Sato, S., Takeda, S., Hasegawa, K. *et al.* (2020) Identification of novel mutations of ovarian cancer-related genes from RNA-sequencing data for Japanese epithelial ovarian cancer patients Endocr J **67**, 219-229 10.1507/endocrj.EJ19-0283
385. Nagashima, T., Yamaguchi, K., Urakami, K., Shimoda, Y., Ohnami, S., Ohshima, K. *et al.* (2020) Japanese version of The Cancer Genome Atlas, JCGA, established using fresh frozen tumors obtained from 5143 cancer patients Cancer Sci **111**, 687-699 10.1111/cas.14290
386. Nagata, M., Setoh, K., Takahashi, M., Higasa, K., Kawaguchi, T., Kawasaki, H. *et al.* (2020) Association of ALPL variants with serum alkaline phosphatase and bone traits in the general Japanese population: The Nagahama Study Journal of human genetics **65**, 337-343 10.1038/s10038-019-0712-3
387. Nakamura, R., Misawa, K., Tohnai, G., Nakatochi, M., Furuhashi, S., Atsuta, N. *et al.* (2020) A multi-ethnic meta-analysis identifies novel genes, including ACSL5, associated with amyotrophic lateral sclerosis Commun Biol **3**, 526 10.1038/s42003-020-01251-2
388. Nakamura, S., Chinen, Y., Satou, K., Tokashiki, T., Kumada, S., Yanagi, K. *et al.* (2020) A severe case of status dystonicus caused by a de novo KMT2B missense mutation Eur J Med Genet **63**, 104057 10.1016/j.ejmg.2020.104057
389. Nakamura, Y., Taniguchi, H., Ikeda, M., Bando, H., Kato, K., Morizane, C. *et al.* (2020) Clinical utility of circulating tumor DNA sequencing in advanced gastrointestinal cancer: SCRUM-Japan GI-SCREEN and GOZILA studies Nat Med **26**, 1859-1864 10.1038/s41591-020-1063-5
390. Nakashima, M., Kato, M., Matsukura, M., Kira, R., Ngu, L. H., Lichtenbelt, K. D. *et al.* (2020) De novo variants in CUL3 are associated with global developmental delays with or without infantile spasms Journal of human genetics **65**, 727-734 10.1038/s10038-020-0758-2
391. Nakaza, M., Kitamura, Y., Furuta, M., Kubota, T., Sasaki, R., andTakahashi, M. P. (2020) Analysis of the genetic background associated with sporadic periodic paralysis in Japanese patients J Neurol Sci **412**, 116795 10.1016/j.jns.2020.116795
392. Nanki, Y., Chiyoda, T., Hirasawa, A., Ookubo, A., Itoh, M., Ueno, M. *et al.* (2020) Patient-derived ovarian cancer organoids capture the genomic profiles of primary tumours applicable for drug sensitivity and resistance testing Sci Rep **10**, 12581 10.1038/s41598-020-69488-9
393. Nawa, Y., Kimura, H., Mori, D., Kato, H., Toyama, M., Furuta, S. *et al.* (2020) Rare single-nucleotide DAB1

variants and their contribution to Schizophrenia and autism spectrum disorder susceptibility Hum Genome Var **7**, 37 10.1038/s41439-020-00125-7

394. Nishiguchi, K. M., Kunikata, H., Fujita, K., Hashimoto, K., Koyanagi, Y., Akiyama, M. *et al.* (2020) Association of CRX genotypes and retinal phenotypes confounded by variable expressivity and electronegative electroretinogram Clin Exp Ophthalmol **48**, 644-657 10.1111/ceo.13743
395. Nozawa, A., Ozeki, M., Yasue, S., Endo, S., Kadowaki, T., Ohnishi, H. *et al.* (2020) Myelodysplastic syndromes in a pediatric patient with Cri du Chat syndrome with a ring chromosome 5 Int J Hematol **112**, 728-733 10.1007/s12185-020-02909-7
396. Numa, S., Oishi, A., Higasa, K., Oishi, M., Miyata, M., Hasegawa, T. *et al.* (2020) EYS is a major gene involved in retinitis pigmentosa in Japan: genetic landscapes revealed by stepwise genetic screening Sci Rep **10**, 20770 10.1038/s41598-020-77558-1
397. Odake, Y., Koh, K., Takiyama, Y., Ishiura, H., Tsuji, S., Yamada, M. *et al.* (2020) Identification of a novel mutation in ATP13A2 associated with a complicated form of hereditary spastic paraplegia Neurol Genet **6**, e514 10.1212/NXG.0000000000000514
398. Ohta, K., Ozawa, T., Fujinaka, H., Goto, K., andNakajima, T. (2020) Cerebral Small Vessel Disease Related to a Heterozygous Nonsense Mutation in HTRA1 Intern Med **59**, 1309-1313 10.2169/internalmedicine.4041-19
399. Oji, Y., Hatano, T., Ueno, S. I., Funayama, M., Ishikawa, K. I., Okuzumi, A. *et al.* (2020) Variants in saposin D domain of prosaposin gene linked to Parkinson's disease Brain **143**, 1190-1205 10.1093/brain/awaa064
400. Oka, Y., Hamada, M., Nakazawa, Y., Muramatsu, H., Okuno, Y., Higasa, K. *et al.* (2020) Digenic mutations in ALDH2 and ADH5 impair formaldehyde clearance and cause a multisystem disorder, AMeD syndrome Sci Adv **6**, eabd7197 10.1126/sciadv.abd7197
401. Okada, T., Washida, K., Irie, K., Saito, S., Noguchi, M., Tomita, T. *et al.* (2020) Prevalence and Atypical Clinical Characteristics of NOTCH3 Mutations Among Patients Admitted for Acute Lacunar Infarctions Front Aging Neurosci **12**, 130 10.3389/fnagi.2020.00130
402. Okano, T., Imai, K., Naruto, T., Okada, S., Yamashita, M., Yeh, T. W. *et al.* (2020) Whole-Exome Sequencing-Based Approach for Germline Mutations in Patients with Inborn Errors of Immunity J Clin Immunol **40**, 729-740 10.1007/s10875-020-00798-3
403. Ozaki, A., Sasaki, M., Hiraide, T., Sumitomo, N., Takeshita, E., Shimizu-Motohashi, Y. *et al.* (2020) A case of CLCN2-related leukoencephalopathy with bright tree appearance during aseptic meningitis Brain Dev **42**, 462-467 10.1016/j.braindev.2020.02.008
404. Sada, K., Hidaka, S., Imaishi, N., Shibata, K., Katashima, R., Noso, S. *et al.* (2020) Clinical and genetic analysis in a family with familial renal glucosuria: Identification of an N101K mutation in the sodium-glucose cotransporter 2 encoded by a solute carrier family 5 member 2 gene J Diabetes Investig **11**, 573-577 10.1111/jdi.13157
405. Sakai, K., Kuwana, M., Tanaka, H., Hosomichi, K., Hasegawa, A., Uyama, H. *et al.* (2020) HLA loci predisposing to immune TTP in Japanese: potential role of the shared ADAMTS13 peptide bound to different HLA-DR Blood **135**, 2413-2419 10.1182/blood.2020005395
406. Sakai, K., Tanikawa, C., Hirasawa, A., Chiyoda, T., Yamagami, W., Kataoka, F. *et al.* (2020) Identification of a novel uterine leiomyoma GWAS locus in a Japanese population Sci Rep **10**, 1197 10.1038/s41598-020-58066-8
407. Sasaki, T., Nishimoto, Y., Abe, Y., Takayama, M., Hirose, N., Okano, H. *et al.* (2020) Sex-Specific Effects of

Apolipoprotein epsilon4 Allele on Mortality in Very Old and Centenarian Japanese Men J Gerontol A Biol Sci Med Sci **75**, 1874-1879 10.1093/gerona/glz242

408. Sato, S., Morimoto, T., Tanaka, S., Hotta, K., Fujikado, T., Tsujikawa, M. *et al.* (2020) Novel mutation identified in Leber congenital amaurosis - a case report BMC Ophthalmol **20**, 313 10.1186/s12886-020-01577-9
409. Sato, T., Kojima, T., Samura, O., Kawaguchi, S., Nakamura, A., Nakajima, M. *et al.* (2020) Two unrelated pedigrees with achondrogenesis type 1b carrying a Japan-specific pathogenic variant in SLC26A2 Am J Med Genet A **182**, 735-739 10.1002/ajmg.a.61469
410. Sawada, J., Katayama, T., Tokashiki, T., Kikuchi, S., Kano, K., Takahashi, K. *et al.* (2020) The First Case of Spinocerebellar Ataxia Type 8 in Monozygotic Twins Intern Med **59**, 277-283 10.2169/internalmedicine.2905-19
411. Shibano, M., Watanabe, A., Takano, N., Mishima, H., Kinoshita, A., Yoshiura, K. I. *et al.* (2020) Target Capture/Next-Generation Sequencing for Nonsyndromic Cleft Lip and Palate in the Japanese Population Cleft Palate Craniofac J **57**, 80-87 10.1177/1055665619857650
412. Shibata, A., Kasai, M., Terashima, H., Hoshino, A., Miyagawa, T., Kikuchi, K. *et al.* (2020) Case-control association study of rare nonsynonymous variants of SCN1A and KCNQ2 in acute encephalopathy with biphasic seizures and late reduced diffusion J Neurol Sci **414**, 116808 10.1016/j.jns.2020.116808
413. Shimizu, S., Mimura, J., Hasegawa, T., Shimizu, E., Imoto, S., Tsushima, M. *et al.* (2020) Association of single nucleotide polymorphisms in the NRF2 promoter with vascular stiffness with aging PLoS One **15**, e0236834 10.1371/journal.pone.0236834
414. Shinagawa, J., Moteki, H., Nishio, S. Y., Noguchi, Y., andUsami, S. I. (2020) Haplotype Analysis of GJB2 Mutations: Founder Effect or Mutational Hot Spot? Genes (Basel) **11**, 250-259 10.3390/genes11030250
415. Suga, K., Imoto, I., Ito, H., Naruto, T., Goji, A., Osumi, K. *et al.* (2020) Next-generation sequencing for the diagnosis of patients with congenital multiple anomalies and / or intellectual disabilities J Med Invest **67**, 246-249 10.2152/jmi.67.246
416. Sugino, S., Konno, D., Kawai, Y., Nagasaki, M., Endo, Y., Hayase, T. *et al.* (2020) Long non-coding RNA MIR4300HG polymorphisms are associated with postoperative nausea and vomiting: a genome-wide association study Hum Genomics **14**, 31 10.1186/s40246-020-00282-4
417. Suzuki, A., Katoh, H., Komura, D., Kakiuchi, M., Tagashira, A., Yamamoto, S. *et al.* (2020) Defined lifestyle and germline factors predispose Asian populations to gastric cancer Sci Adv **6**, eaav9778 10.1126/sciadv.aav9778
418. Tada, Y., Kume, K., Matsuda, Y., Kurashige, T., Kanaya, Y., Ohsawa, R. *et al.* (2020) Genetic screening for potassium channel mutations in Japanese autosomal dominant spinocerebellar ataxia Journal of human genetics **65**, 363-369 10.1038/s10038-019-0717-y
419. Taguchi, M., Mishima, H., Shiozawa, Y., Hayashida, C., Kinoshita, A., Nannya, Y. *et al.* (2020) Genome analysis of myelodysplastic syndromes among atomic bomb survivors in Nagasaki Haematologica **105**, 358-365 10.3324/haematol.2019.219386
420. Takeda, Y., Kubota, D., Oishi, N., Maruyama, K., Gocho, K., Yamaki, K. *et al.* (2020) Novel GUCY2D Variant (E843Q) at Mutation Hotspot Associated with Macular Dystrophy in a Japanese Patient J Nippon Med Sch **87**, 92-99 10.1272/jnms.JNMS.2020_87-207
421. Takeshita, Y., Ohto, T., Enokizono, T., Tanaka, M., Suzuki, H., Fukushima, H. *et al.* (2020) Novel ARX mutation identified in infantile spasm syndrome patient Hum Genome Var **7**, 9 10.1038/s41439-020-0094-2
422. Takeuchi, I., Kawai, T., Nambu, M., Migita, O., Yoshimura, S., Nishimura, K. *et al.* (2020) X-linked inhibitor of

apoptosis protein deficiency complicated with Crohn's disease-like enterocolitis and Takayasu arteritis: A case report Clin Immunol **217**, 108495 10.1016/j.clim.2020.108495

423. Takizaki, N., Tsurusaki, Y., Katsumata, K., Enomoto, Y., Murakami, H., Muroya, K. *et al.* (2020) Novel CUL7 biallelic mutations alter the skeletal phenotype of 3M syndrome Hum Genome Var **7**, 1 10.1038/s41439-020-0090-6
424. Uchino, A., Nagai, M., Kanazawa, N., Ichinoe, M., Yanagisawa, N., Adachi, K. *et al.* (2020) An autopsy case of G(M1) gangliosidosis type II in a patient who survived a long duration with artificial respiratory support Neuropathology **40**, 379-388 10.1111/neup.12651
425. Uchiyama, H., Masunaga, Y., Ishikawa, T., Fukuoka, T., Fukami, M., Saitsu, H. *et al.* (2020) TSC1 intragenic deletion transmitted from a mosaic father to two siblings with cardiac rhabdomyomas: Identification of two aberrant transcripts Eur J Med Genet **63**, 104060 10.1016/j.ejmg.2020.104060
426. Udagawa, C., Sasaki, Y., Tanizawa, Y., Suemizu, H., Ohnishi, Y., Nakamura, Y. *et al.* (2020) Whole-exome sequencing of 79 xenografts as a potential approach for the identification of genetic variants associated with sensitivity to cytotoxic anticancer drugs PLoS One **15**, e0239614 10.1371/journal.pone.0239614
427. Uehara, T., Abe, K., Oginuma, M., Ishitani, S., Yoshihashi, H., Okamoto, N. *et al.* (2020) Pathogenesis of CDK8-associated disorder: two patients with novel CDK8 variants and in vitro and in vivo functional analyses of the variants Sci Rep **10**, 17575 10.1038/s41598-020-74642-4
428. Uehara, T., Yamada, M., Umetsu, S., Nittono, H., Suzuki, H., Fujisawa, T. *et al.* (2020) Biallelic Mutations in the LSR Gene Cause a Novel Type of Infantile Intrahepatic Cholestasis J Pediatr **221**, 251-254 10.1016/j.jpeds.2020.01.064
429. Wada, Y., Maekawa, M., Ohnishi, T., Balan, S., Matsuoka, S., Iwamoto, K. *et al.* (2020) Peroxisome proliferator-activated receptor alpha as a novel therapeutic target for schizophrenia Ebiomedicine **62**, 103130 10.1016/j.ebiom.2020.103130
430. Walls, W. D., Moteki, H., Thomas, T. R., Nishio, S. Y., Yoshimura, H., Iwasa, Y. *et al.* (2020) A comparative analysis of genetic hearing loss phenotypes in European/American and Japanese populations Hum Genet **139**, 1315-1323 10.1007/s00439-020-02174-y
431. Watabe, T., Kanzaki, S., Sato, N., Matsunaga, T., Muramatsu, M., andOgawa, K. (2020) Single nucleotide polymorphisms in tinnitus patients exhibiting severe distress Sci Rep **10**, 13023 10.1038/s41598-020-69467-0
432. Yakou, F., Suwanai, H., Ishikawa, T., Itou, M., Shikuma, J., Miwa, T. *et al.* (2020) A Novel Homozygous Mutation of Thyroid Peroxidase Gene Abolishes a Disulfide Bond Leading to Congenital Hypothyroidism Int J Endocrinol **2020**, 9132372 10.1155/2020/9132372
433. Yamada, M., Shiraishi, Y., Uehara, T., Suzuki, H., Takenouchi, T., Abe-Hatano, C. *et al.* (2020) Diagnostic utility of integrated analysis of exome and transcriptome: Successful diagnosis of Au-Kline syndrome in a patient with submucous cleft palate, scaphocephaly, and intellectual disabilities Mol Genet Genomic Med **8**, e1364 10.1002/mgg3.1364
434. Yamamoto, K., Sakaue, S., Matsuda, K., Murakami, Y., Kamatani, Y., Ozono, K. *et al.* (2020) Genetic and phenotypic landscape of the mitochondrial genome in the Japanese population Commun Biol **3**, 104 10.1038/s42003-020-0812-9
435. Yamamoto, T., Miyoshi, H., Kakizaki, F., Maekawa, H., Yamaura, T., Morimoto, T. *et al.* (2020) Chemosensitivity of Patient-Derived Cancer Stem Cells Identifies Colorectal Cancer Patients with Potential Benefit from FGFR Inhibitor Therapy Cancers (Basel) **12**, 2010 10.3390/cancers12082010

436. Yanagisawa, M., Namekata, K., Aida, T., Katou, S., Takeda, T., Harada, T. *et al.* (2020) EAAT1 variants associated with glaucoma Biochem Biophys Res Commun **529**, 943-949 10.1016/j.bbrc.2020.06.099

437. Yang, L., Fujinami, K., Ueno, S., Kuniyoshi, K., Hayashi, T., Kondo, M. *et al.* (2020) Genetic Spectrum of EYS-associated Retinal Disease in a Large Japanese Cohort: Identification of Disease-associated Variants with Relatively High Allele Frequency Sci Rep **10**, 5497 10.1038/s41598-020-62119-3

438. Yaoita, N., Satoh, K., Satoh, T., Shimizu, T., Saito, S., Sugimura, K. *et al.* (2020) Identification of the Novel Variants in Patients With Chronic Thromboembolic Pulmonary Hypertension J Am Heart Assoc **9**, e015902 10.1161/JAHA.120.015902

439. Yasuda, R., Yoshida, T., Mizuta, I., Watanabe, M., Nakano, M., Sato, R. *et al.* (2020) Adult-onset leukoencephalopathy with homozygous LAMB1 missense mutation Neurol Genet **6**, e442 10.1212/NXG.0000000000000442

440. Yasuda, T., Sanada, M., Nishijima, D., Kanamori, T., Iijima, Y., Hattori, H. *et al.* (2020) Clinical utility of target capture-based panel sequencing in hematological malignancies: A multicenter feasibility study Cancer Sci **111**, 3367-3378 10.1111/cas.14552

441. Yatsuka, Y., Kishita, Y., Formosa, L. E., Shimura, M., Nozaki, F., Fujii, T. *et al.* (2020) A homozygous variant in NDUFA8 is associated with developmental delay, microcephaly, and epilepsy due to mitochondrial complex I deficiency Clin Genet **98**, 155-165 10.1111/cge.13773

442. Yokoi, K., Nakajima, Y., Matsuoka, H., Shinkai, Y., Ishihara, T., Maeda, Y. *et al.* (2020) Impact of DPYD, DPYS, and UPB1 gene variations on severe drug-related toxicity in patients with cancer Cancer Sci **111**, 3359-3366 10.1111/cas.14553

443. Abe-Hatano, C., Iida, A., Kosugi, S., Momozawa, Y., Terao, C., Ishikawa, K. *et al.* (2021) Whole genome sequencing of 45 Japanese patients with intellectual disability Am J Med Genet A **185**, 1468-1480 10.1002/ajmg.a.62138

444. Aguilar-Ordonez, I., Perez-Villatoro, F., Garcia-Ortiz, H., Barajas-Olmos, F., Ballesteros-Villascan, J., Gonzalez-Buenfil, R. *et al.* (2021) Whole genome variation in 27 Mexican indigenous populations, demographic and biomedical insights PLoS One **16**, e0249773 10.1371/journal.pone.0249773

445. Andoh, A., Kawahara, M., Imai, T., Tatsumi, G., Inatomi, O., andKakuta, Y. (2021) Thiopurine pharmacogenomics and pregnancy in inflammatory bowel disease J Gastroenterol **56**, 881-890 10.1007/s00535-021-01805-z

446. Bai, S., Geng, Y., Duan, H., Xu, L., Yu, Z., Yuan, J. *et al.* (2021) A novel p.Pro871Leu missense mutation in SPECC1L gene causing craniosynostosis in a patient Orthod Craniofac Res **24**, 480-485 10.1111/ocr.12473

447. Balan, S., Iwayama, Y., Ohnishi, T., Fukuda, M., Shirai, A., Yamada, A. *et al.* (2021) A loss-of-function variant in SUV39H2 identified in autism-spectrum disorder causes altered H3K9 trimethylation and dysregulation of protocadherin beta-cluster genes in the developing brain Mol Psychiatry **26**, 7550-7559 10.1038/s41380-021-01199-7

448. Bhattacharjee, M. J., Lin, J. J., Chang, C. Y., Chiou, Y. T., Li, T. N., Tai, C. W. *et al.* (2021) Identifying Primate ACE2 Variants That Confer Resistance to SARS-CoV-2 Mol Biol Evol **38**, 2715-2731 10.1093/molbev/msab060

449. Bhattacharyya, C., Das, C., Ghosh, A., Singh, A. K., Mukherjee, S., Majumder, P. P. *et al.* (2021) SARS-CoV-2 mutation 614G creates an elastase cleavage site enhancing its spread in high AAT-deficient regions Infect Genet Evol **90**, 104760 10.1016/j.meegid.2021.104760

450. Daida, K., Nishioka, K., Li, Y., Yoshino, H., Shimada, T., Dougu, N. *et al.* (2021) PLA2G6 variants associated with the number of affected alleles in Parkinson's disease in Japan Neurobiol Aging **97**, 147 e141-147 e149

10.1016/j.neurobiolaging.2020.07.004

451. Danda, V. S. R., Paidipelly, S. R., Verepula, M., Lodha, P., Thaduri, K. R., Konda, C. *et al.* (2021) Exploring the Genetic Diversity of Isolated Hypogonadotropic Hypogonadism and Its Phenotypic Spectrum: A Case Series J Reprod Infertil **22**, 38-46 10.18502/jri.v22i1.4994
452. Endo, Y., Koga, T., Otaki, H., Furukawa, K., andKawakami, A. (2021) Systemic lupus erythematosus overlapping dermatomyositis owing to a heterozygous TREX1 Asp130Asn missense mutation Clin Immunol **227**, 108732 10.1016/j.clim.2021.108732
453. Endo, Y., Koga, T., Ubara, Y., Sumiyoshi, R., Furukawa, K., andKawakami, A. (2021) Mediterranean fever gene variants modify clinical phenotypes of idiopathic multi-centric Castleman disease Clin Exp Immunol **206**, 91-98 10.1111/cei.13632
454. Enomoto, Y., Tsurusaki, Y., Tominaga, M., Kobayashi, S., Inoue, M., Fujita, K. *et al.* (2021) A Recurrent Variant in POLR1B, c.3007C>T; p.Arg1003Cys, Associated with Atresia of the External Canal and Microtia in Treacher Collins Syndrome Type 4 Mol Syndromol **12**, 127-132 10.1159/000513224
455. Fujikura, K., andUesaka, K. (2021) Genetic variations in the human severe acute respiratory syndrome coronavirus receptor ACE2 and serine protease TMPRSS2 J Clin Pathol **74**, 307-313 10.1136/jclinpath-2020-206867
456. Fujishima, N., Kohmaru, J., Koyota, S., Kuba, K., Saga, T., Omokawa, A. *et al.* (2021) Clonal hematopoiesis in adult pure red cell aplasia Sci Rep **11**, 2253 10.1038/s41598-021-81890-5
457. Fukuda, A., Kodama, Y., Mashima, J., Fujisawa, T., andOgasawara, O. (2021) DDBJ update: streamlining submission and access of human data Nucleic Acids Res **49**, D71-D75 10.1093/nar/gkaa982
458. Fukunaga, K., Hishinuma, E., Hiratsuka, M., Kato, K., Okusaka, T., Saito, T. *et al.* (2021) Determination of novel CYP2D6 haplotype using the targeted sequencing followed by the long-read sequencing and the functional characterization in the Japanese population Journal of human genetics **66**, 139-149 10.1038/s10038-020-0815-x
459. Funato, M., Tsunematsu, Y., Yamazaki, F., Tamura, C., Kumamoto, T., Takagi, M. *et al.* (2021) Characteristics of Li-Fraumeni Syndrome in Japan; A Review Study by the Special Committee of JSHT Cancer Sci **112**, 2821-2834 10.1111/cas.14919
460. Futagawa, M., Yamamoto, H., Kochi, M., Urakawa, Y., Sogawa, R., Kato, F. *et al.* (2021) Retroperitoneal leiomyosarcoma in a female patient with a germline splicing variant RAD51D c.904-2A > T: a case report Hered Cancer Clin Pract **19**, 48 10.1186/s13053-021-00205-x
461. Georgiou, M., Robson, A. G., Fujinami, K., Leo, S. M., Vincent, A., Nasser, F. *et al.* (2021) KCNV2-Associated Retinopathy: Genetics, Electrophysiology, and Clinical Course-KCNV2 Study Group Report 1 Am J Ophthalmol **225**, 95-107 10.1016/j.ajo.2020.11.022
462. Goto, K., Kukita, Y., Honma, K., Ogawa, K., Nishida, H., Takai, T. *et al.* (2021) Signet-ring cell/histiocytoid carcinoma of the axilla: a clinicopathological and genetic analysis of 11 cases, review of the literature, and comparison with potentially related tumours Histopathology **79**, 926-939 10.1111/his.14436
463. Gowda, V. K., Vegda, H., Sugumar, K., Narayanappa, G., Srinivasan, V. M., Santhoshkumar, R. *et al.* (2021) Neuronal Ceroid Lipofuscinosis: Clinical and Laboratory Profile in Children from Tertiary Care Centre in South India J Pediatr Genet **10**, 266-273 10.1055/s-0040-1715575
464. Ha, T. W., Jung, H. U., Kim, D. J., Baek, E. J., Lee, W. J., Lim, J. E. *et al.* (2021) Association Between Environmental Factors and Asthma Using Mendelian Randomization: Increased Effect of Body Mass Index on

Adult-Onset Moderate-to-Severe Asthma Subtypes Front Genet **12**, 639905 10.3389/fgene.2021.639905

465. Hagio, K., Amano, T., Hayashi, H., Takeshita, T., Oshino, T., Kikuchi, J. *et al.* (2021) Impact of clinical targeted sequencing on endocrine responsiveness in estrogen receptor-positive, HER2-negative metastatic breast cancer Sci Rep **11**, 8109 10.1038/s41598-021-87645-6
466. Hamamoto, Y., Kukita, Y., Kitamura, M., Kurashige, M., Masaie, H., Fuji, S. *et al.* (2021) Bcl-2-negative IGH-BCL2 translocation-negative follicular lymphoma of the thyroid differs genetically and epigenetically from Bcl-2-positive IGH-BCL2 translocation-positive follicular lymphoma Histopathology **79**, 521-532 10.1111/his.14378
467. Hanafusa, H., Hidaka, Y., Yamaguchi, T., Shimojo, H., Tsukahara, T., Murase, T. *et al.* (2021) Heterozygous missense variant in TRPC6 in a boy with rapidly progressive infantile nephrotic syndrome associated with diffuse mesangial sclerosis Am J Med Genet A **185**, 2175-2179 10.1002/ajmg.a.62216
468. Hashizume, M., Gonzalez, G., Ono, C., Takashima, A., andIwasaki, M. (2021) Population-Specific ACE2 Single-Nucleotide Polymorphisms Have Limited Impact on SARS-CoV-2 Infectivity In Vitro Viruses **13**, 67 10.3390/v13010067
469. Hata, T., Mizuma, M., Motoi, F., Ishida, M., Ohtsuka, H., Nakagawa, K. *et al.* (2021) Germline DNA damage repair gene mutations in pancreatic cancer patients with personal/family histories of pancreas/breast/ovarian/prostate cancer in a Japanese population Ann Gastroenterol Surg **5**, 853-864 10.1002/ags3.12482
470. Hatano, M., Fukushima, H., Ohto, T., Ueno, Y., Saeki, S., Enokizono, T. *et al.* (2021) Variants in KIF2A cause broad clinical presentation; the computational structural analysis of a novel variant in a patient with a cortical dysplasia, complex, with other brain malformations 3 Am J Med Genet A **185**, 1113-1119 10.1002/ajmg.a.62084
471. Hayashi, K., Noguchi-Shinohara, M., Sato, T., Hosomichi, K., Kannon, T., Abe, C. *et al.* (2021) Effects of functional variants of vitamin C transporter genes on apolipoprotein E E4-associated risk of cognitive decline: The Nakajima study PLoS One **16**, e0259663 10.1371/journal.pone.0259663
472. Hayashi, T., Katagiri, S., Kubota, D., Mizobuchi, K., Ishiuji, Y., Asahina, A. *et al.* (2021) The first Japanese family of CDH3-related hypotrichosis with juvenile macular dystrophy Mol Genet Genomic Med **9**, e1688 10.1002/mgg3.1688
473. Hayashi, T., Kondo, H., Matsushita, I., Mizobuchi, K., Baba, A., Iida, K. *et al.* (2021) Homozygous single nucleotide duplication of SLC38A8 in autosomal recessive foveal hypoplasia: The first Japanese case report Doc Ophthalmol **143**, 323-330 10.1007/s10633-021-09842-y
474. Hayashi, T., Mizobuchi, K., Kameya, S., Yoshitake, K., Iwata, T., andNakano, T. (2021) A new PDE6A missense variant p.Arg544Gln in rod-cone dystrophy Doc Ophthalmol **143**, 107-114 10.1007/s10633-021-09826-y
475. Hayashida, A., Li, Y., Yoshino, H., Daida, K., Ikeda, A., Ogaki, K. *et al.* (2021) The identified clinical features of Parkinson's disease in homo-, heterozygous and digenic variants of PINK1 Neurobiol Aging **97**, 146 e141-146 e113 10.1016/j.neurobiolaging.2020.06.017
476. Hiraide, T., Fukumura, S., Yamamoto, A., Nakashima, M., andSaitsu, H. (2021) Familial periodic paralysis associated with a rare KCNJ5 variant that supposed to have incomplete penetrance Brain Dev **43**, 470-474 10.1016/j.braindev.2020.10.010
477. Hiraide, T., Tanaka, T., Masunaga, Y., Ohkubo, Y., Nakashima, M., Fukuda, T. *et al.* (2021) Global developmental delay, systemic dysmorphism and epilepsy in a patient with a de novo U2AF2 variant Journal of human genetics **66**, 1185-1187 10.1038/s10038-021-00948-4
478. Hiraide, T., Yamoto, K., Masunaga, Y., Asahina, M., Endoh, Y., Ohkubo, Y. *et al.* (2021) Genetic and phenotypic

analysis of 101 patients with developmental delay or intellectual disability using whole-exome sequencing Clin Genet **100**, 40-50 10.1111/cge.13951

479. Hiramatsu, K., Nishio, S. Y., Kitajiri, S. I., Kitano, T., Moteki, H., Usami, S. I. *et al.* (2021) Prevalence and Clinical Characteristics of Hearing Loss Caused by MYH14 Variants Genes (Basel) **12**, 10.3390/genes12101623
480. Hori, H., Itoh, M., Lin, M., Yoshida, F., Niwa, M., Hakamata, Y. *et al.* (2021) Childhood maltreatment history and attention bias variability in healthy adult women: role of inflammation and the BDNF Val66Met genotype Transl Psychiatry **11**, 122 10.1038/s41398-021-01247-4
481. Horiuchi, Y., Matsubayashi, H., Kiyozumi, Y., Nishimura, S., Higashigawa, S., Kado, N. *et al.* (2021) Disclosure of secondary findings in exome sequencing of 2480 Japanese cancer patients Hum Genet **140**, 321-331 10.1007/s00439-020-02207-6
482. Imoto, I., Saito, M., Suga, K., Kohmoto, T., Otsu, M., Horiuchi, K. *et al.* (2021) Functionally confirmed compound heterozygous ADAM17 missense loss-of-function variants cause neonatal inflammatory skin and bowel disease 1 Sci Rep **11**, 9552 10.1038/s41598-021-89063-0
483. Inagaki, Y., Ogawa, T., Tabata, M. J., Nagata, Y., Watanabe, R., Kawamoto, T. *et al.* (2021) Identification of OPN3 as associated with non-syndromic oligodontia in a Japanese population Journal of human genetics **66**, 769-775 10.1038/s10038-021-00903-3
484. Inano, T., Araki, M., Morishita, S., Imai, M., Kihara, Y., Okuda, M. *et al.* (2021) Cell-autonomous megakaryopoiesis associated with polyclonal hematopoiesis in triple-negative essential thrombocythemia Sci Rep **11**, 17702 10.1038/s41598-021-97106-9
485. Inoue, M., Saito, Y., Yonekawa, T., Ogawa, M., Iida, A., Nishino, I. *et al.* (2021) Causative variant profile of collagen VI-related dystrophy in Japan Orphanet J Rare Dis **16**, 284 10.1186/s13023-021-01921-2
486. Itai, T., Hamanaka, K., Sasaki, K., Wagner, M., Kotzaeridou, U., Brosse, I. *et al.* (2021) De novo variants in CELF2 that disrupt the nuclear localization signal cause developmental and epileptic encephalopathy Hum Mutat **42**, 66-76 10.1002/humu.24130
487. Itai, T., Miyatake, S., Taguri, M., Nozaki, F., Ohta, M., Osaka, H. *et al.* (2021) Prenatal clinical manifestations in individuals with COL4A1/2 variants J Med Genet **58**, 505-513 10.1136/jmedgenet-2020-106896
488. Ito, S., Sato, I., Mochizuki, M., Yamaguchi, K., Tamai, K., Minato, T. *et al.* (2021) Robustness of a Cancer Profiling Test Using Formalin-fixed Paraffin Embedded Tumor Specimens Anticancer Res **41**, 1341-1348 10.21873/anticanres.14891
489. Juang, J. J., Lu, T. P., Su, M. W., Lin, C. W., Yang, J. H., Chu, H. W. *et al.* (2021) Rare variants discovery by extensive whole-genome sequencing of the Han Chinese population in Taiwan: Applications to cardiovascular medicine J Adv Res **30**, 147-158 10.1016/j.jare.2020.12.003
490. Kadowaki, S., Hashimoto, K., Nishimura, T., Kashimada, K., Kadowaki, T., Kawamoto, N. *et al.* (2021) Functional analysis of novel A20 variants in patients with atypical inflammatory diseases Arthritis Res Ther **23**, 52 10.1186/s13075-021-02434-w
491. Kagawa, M., Kawakami, S., Yamamoto, A., Suzuki, O., Kamae, N., Eguchi, H. *et al.* (2021) Identification of Lynch syndrome-associated DNA mismatch repair-deficient bladder cancer in a Japanese hospital-based population Int J Clin Oncol **26**, 1524-1532 10.1007/s10147-021-01922-y
492. Kakizawa, K., Yamashita, M., Nakashima, M., Kawauchi, Y., Ikeya, A., Matsushita, A. *et al.* (2021) Retroperitoneal Paraganglioma With Asymptomatic Follicular Lymphoma: A Case Report J Endocr Soc **5**, bvab171 10.1210/jendso/bvab171

493. Kase, K., Saito, M., Nakajima, S., Takayanagi, D., Saito, K., Yamada, L. *et al.* (2021) ARID1A deficiency in EBV-positive gastric cancer is partially regulated by EBV-encoded miRNAs, but not by DNA promotor hypermethylation Carcinogenesis **42**, 21-30 10.1093/carcin/bgaa123

494. Kawachi, H., Kunimasa, K., Kukita, Y., Nakamura, H., Honma, K., Kawamura, T. *et al.* (2021) Atezolizumab with bevacizumab, paclitaxel and carboplatin was effective for patients with SMARCA4-deficient thoracic sarcoma Immunotherapy **13**, 799-806 10.2217/imt-2020-0311

495. Kawai, Y., Hitomi, Y., Ueta, M., Khor, S. S., Nakatani, K., Sotozono, C. *et al.* (2021) Mapping of susceptible variants for cold medicine-related Stevens-Johnson syndrome by whole-genome resequencing NPJ Genom Med **6**, 9 10.1038/s41525-021-00171-2

496. Kawano-Matsuda, F., Maeda, T., Kaname, T., Yanagi, K., andIhara, K. (2021) X-linked mental retardation and severe short stature with a novel mutation of the KDM5C gene Clin Pediatr Endocrinol **30**, 61-64 10.1297/cpe.30.61

497. Kikkawa, Y., Hashimoto, T., Takizawa, K., Urae, S., Masuda, H., Matsunuma, M. *et al.* (2021) Laminin beta2 variants associated with isolated nephropathy that impact matrix regulation JCI Insight **6**, 10.1172/jci.insight.145908

498. Kishita, Y., Shimura, M., Kohda, M., Fushimi, T., Nitta, K. R., Yatsuka, Y. *et al.* (2021) Genome sequencing and RNA-seq analyses of mitochondrial complex I deficiency revealed Alu insertion-mediated deletion in NDUFV2 Hum Mutat **42**, 1422-1428 10.1002/humu.24274

499. Koh, K., Takaki, R., Ishiura, H., Tsuji, S., andTakiyama, Y. (2021) SPG9A with the new occurrence of an ALDH18A1 mutation in a CMT1A family with PMP22 duplication: case report BMC Neurol **21**, 64 10.1186/s12883-021-02087-x

500. Kondo, A., Nagano, C., Ishiko, S., Omori, T., Aoto, Y., Rossanti, R. *et al.* (2021) Examination of the predicted prevalence of Gitelman syndrome by ethnicity based on genome databases Sci Rep **11**, 16099 10.1038/s41598-021-95521-6

501. Kondo, H., Matsushita, I., Nagata, T., Fujihara, E., Hosono, K., Uchio, E. *et al.* (2021) Retinal Features of Family Members With Familial Exudative Vitreoretinopathy Caused By Mutations in KIF11 Gene Transl Vis Sci Technol **10**, 18 10.1167/tvst.10.7.18

502. Kumamoto, T., Yamazaki, F., Nakano, Y., Tamura, C., Tashiro, S., Hattori, H. *et al.* (2021) Medical guidelines for Li-Fraumeni syndrome 2019, version 1.1 Int J Clin Oncol **26**, 2161-2178 10.1007/s10147-021-02011-w

503. Makabe, T., Yamagami, W., Hirasawa, A., Miyabe, I., Wakatsuki, T., Kikuchi, M. *et al.* (2021) Incidence of germline variants in Lynch syndrome-related genes among Japanese endometrial cancer patients aged 40 years or younger Int J Clin Oncol **26**, 1767-1774 10.1007/s10147-021-01953-5

504. Makitie, R. E., Pekkinen, M., Morisada, N., Kobayashi, D., Yonezawa, Y., Nishimura, G. *et al.* (2021) A Novel IFITM5 Variant Associated with Phenotype of Osteoporosis with Calvarial Doughnut Lesions: A Case Report Calcif Tissue Int **109**, 626-632 10.1007/s00223-021-00878-5

505. Masago, K., andFujita, S. (2021) Novel NR4A1 Arg293Ser Mutation in Patients With Familial Crohn's Disease In Vivo **35**, 2135-2140 10.21873/invivo.12483

506. Masunaga, Y., Fujisawa, Y., Muramatsu, M., Ono, H., Inoue, T., Fukami, M. *et al.* (2021) Insulin resistant diabetes mellitus in SHORT syndrome: case report and literature review Endocr J **68**, 111-117 10.1507/endocrj.EJ20-0291

507. Masunaga, Y., Kagami, M., Kato, F., Usui, T., Yonemoto, T., Mishima, K. *et al.* (2021) Parthenogenetic mosaicism:

generation via second polar body retention and unmasking of a likely causative PER2 variant for hypersomnia Clin Epigenetics **13**, 73 10.1186/s13148-021-01062-0

508. Masunaga, Y., Mochizuki, M., Kadoya, M., Wada, Y., Okamoto, N., Fukami, M. *et al.* (2021) Primary ovarian insufficiency in a female with phosphomannomutase-2 gene (PMM2) mutations for congenital disorder of glycosylation Endocr J **68**, 605-611 10.1507/endocrj.EJ20-0706
509. Matsumoto, A., Kojima, K., Miya, F., Miyauchi, A., Watanabe, K., Iwamoto, S. *et al.* (2021) Two cases of DYNC1H1 mutations with intractable epilepsy Brain Dev **43**, 857-862 10.1016/j.braindev.2021.05.005
510. Minato, T., Ito, S., Li, B., Fujimori, H., Mochizuki, M., Yamaguchi, K. *et al.* (2021) Liquid biopsy with droplet digital PCR targeted to specific mutations in plasma cell-free tumor DNA can detect ovarian cancer recurrence earlier than CA125 Gynecol Oncol Rep **38**, 100847 10.1016/j.gore.2021.100847
511. Mitsutake, A., Unuma, A., Kawai, M., Kubota, A., Ishiura, H., Sakuishi, K. *et al.* (2021) Severe dilated cardiomyopathy and ventricular arrhythmia in a patient with Emery-Dreifuss muscular dystrophy harboring a novel frameshift mutation in Neurology and Clinical Neuroscience **9**, 490-493 10.1111/ncn3.12552
512. Mizukami, M., Ishikawa, A., Miyazaki, S., Tsuzuki, A., Saito, S., Niihori, T. *et al.* (2021) A de novo CHD3 variant in a child with intellectual disability, autism, joint laxity, and dysmorphisms Brain Dev **43**, 563-565 10.1016/j.braindev.2020.12.004
513. Mori, T., Chiga, M., Fujimaru, T., Kawamoto, R., Mandai, S., Nanamatsu, A. *et al.* (2021) Phenotypic differences of mutation-negative cases in Gitelman syndrome clinically diagnosed in adulthood Hum Mutat **42**, 300-309 10.1002/humu.24159
514. Morimoto, N., Nagahama, K., Mori, T., Fujimaru, T., Tsuura, Y., Terai, A. *et al.* (2021) A Novel LMX1B Variant Identified in a Patient Presenting with Severe Renal Involvement and Thin Glomerular Basement Membrane Nephron **145**, 776-782 10.1159/000518423
515. Morimoto, Y., Ono, S., Yoshida, S., Mishima, H., Kinoshita, A., Tanaka, T. *et al.* (2021) A unique missense variant in the E1A-binding protein P400 gene is implicated in schizophrenia by whole-exome sequencing and mutant mouse models Transl Psychiatry **11**, 132 10.1038/s41398-021-01258-1
516. Murakami, N., Asami, Y., Yoshida, H., Takayanagi, D., Hirose, S., Kuno, I. *et al.* (2021) Distribution of genetic alterations in high-risk early-stage cervical cancer patients treated with postoperative radiation therapy Sci Rep **11**, 10567 10.1038/s41598-021-90139-0
517. Nagae, G., Yamamoto, S., Fujita, M., Fujita, T., Nonaka, A., Umeda, T. *et al.* (2021) Genetic and epigenetic basis of hepatoblastoma diversity Nat Commun **12**, 5423 10.1038/s41467-021-25430-9
518. Nakamura, R., Tohnai, G., Atsuta, N., Nakatochi, M., Hayashi, N., Watanabe, H. *et al.* (2021) Genetic and functional analysis of KIF5A variants in Japanese patients with sporadic amyotrophic lateral sclerosis Neurobiol Aging **97**, 147 e111-147 e117 10.1016/j.neurobiolaging.2020.07.010
519. Nakamura, Y., Okanishi, T., Yamada, H., Okazaki, T., Hosoda, C., Itai, T. *et al.* (2021) Progressive cerebral atrophies in three children with COL4A1 mutations Brain Dev **43**, 1033-1038 10.1016/j.braindev.2021.06.008
520. Naruse, H., Ishiura, H., Mitsui, J., Takahashi, Y., Matsukawa, T., Sakuishi, K. *et al.* (2021) Splice-site mutations in KIF5A in the Japanese case series of amyotrophic lateral sclerosis Neurogenetics **22**, 11-17 10.1007/s10048-020-00626-1
521. Nishiguchi, K. M., Miya, F., Mori, Y., Fujita, K., Akiyama, M., Kamatani, T. *et al.* (2021) A hypomorphic variant in EYS detected by genome-wide association study contributes toward retinitis pigmentosa Commun Biol **4**, 140 10.1038/s42003-021-01662-9

522. Nishina, S., Hosono, K., Ishitani, S., Kosaki, K., Yokoi, T., Yoshida, T. *et al.* (2021) Biallelic CDK9 variants as a cause of a new multiple-malformation syndrome with retinal dystrophy mimicking the CHARGE syndrome Journal of human genetics **66**, 1021-1027 10.1038/s10038-021-00909-x

523. Nishioka, M., Kazuno, A. A., Nakamura, T., Sakai, N., Hayama, T., Fujii, K. *et al.* (2021) Systematic analysis of exonic germline and postzygotic de novo mutations in bipolar disorder Nat Commun **12**, 3750 10.1038/s41467-021-23453-w

524. Odagiri, S., Kabata, D., Tomita, S., Kudo, S., Sakaguchi, T., Nakano, N. *et al.* (2021) Clinical and Genetic Characteristics of Patients with Mild Hyperphenylalaninemia Identified by Newborn Screening Program in Japan Int J Neonatal Screen **7**, 17 10.3390/ijns7010017

525. Oe, Y., Mishima, E., Mori, T., Okamoto, K., Honkura, Y., Nagasawa, T. *et al.* (2021) A Novel Mutation in LMX1B (p.Pro219Ala) Causes Focal Segmental Glomerulosclerosis with Alport Syndrome-like Phenotype Intern Med **60**, 2991-2996 10.2169/internalmedicine.6987-20

526. Ohashi, K., Fukuhara, S., Miyachi, T., Asai, T., Imaeda, M., Goto, M. *et al.* (2021) Comprehensive Genetic Analysis of Non-syndromic Autism Spectrum Disorder in Clinical Settings J Autism Dev Disord **51**, 4655-4662 10.1007/s10803-021-04910-3

527. Oishi, A., Fujinami, K., Mawatari, G., Naoi, N., Ikeda, Y., Ueno, S. *et al.* (2021) Genetic and Phenotypic Landscape of PRPH2-Associated Retinal Dystrophy in Japan Genes (Basel) **12**, 1817 10.3390/genes12111817

528. Ota, M., Nagafuchi, Y., Hatano, H., Ishigaki, K., Terao, C., Takeshima, Y. *et al.* (2021) Dynamic landscape of immune cell-specific gene regulation in immune-mediated diseases Cell **184**, 3006-3021 e3017 10.1016/j.cell.2021.03.056

529. Otsuka, T., Hori, H., Yoshida, F., Itoh, M., Lin, M., Niwa, M. *et al.* (2021) Association of CRP genetic variation with symptomatology, cognitive function, and circulating proinflammatory markers in civilian women with PTSD J Affect Disord **279**, 640-649 10.1016/j.jad.2020.10.045

530. Park, K. S. (2021) Two Approaches for a Genetic Analysis of Pompe Disease: A Literature Review of Patients with Pompe Disease and Analysis Based on Genomic Data from the General Population Children (Basel) **8**, 601 10.3390/children8070601

531. Pati, A., Padhi, S., Nayak, T. K. S., andPanda, A. K. (2021) Angiotensin-Converting Enzyme 2 Genetic Variants Are Associated With Coronavirus Disease 2019 J Infect Dis **224**, 2181-2183 10.1093/infdis/jiab531

532. Pooh, R. K., Machida, M., Imoto, I., Arai, E. N., Ohashi, H., Takeda, M. *et al.* (2021) Fetal Megalencephaly with Cortical Dysplasia at 18 Gestational Weeks Related to Paternal UPD Mosaicism with PTEN Mutation Genes (Basel) **12**, 358 10.3390/genes12030358

533. Rahman, M. M., andFatema, K. (2021) Genetic Diagnosis in Children with Epilepsy and Developmental Disorders by Targeted Gene Panel Analysis in a Developing Country J Epilepsy Res **11**, 22-31 10.14581/jer.21004

534. Sakaue, S., Kanai, M., Tanigawa, Y., Karjalainen, J., Kurki, M., Koshiba, S. *et al.* (2021) A cross-population atlas of genetic associations for 220 human phenotypes Nature genetics **53**, 1415-1424 10.1038/s41588-021-00931-x

535. Sakurai, Y., Watanabe, T., Abe, Y., Nawa, T., Uchida, T., Aoi, H. *et al.* (2021) Head titubation and irritability as early symptoms of Joubert syndrome with a homozygous NPHP1 variant Brain Dev **43**, 863-866 10.1016/j.braindev.2021.04.011

536. Segawa, K., Kikuchi, A., Noji, T., Sugiura, Y., Hiraga, K., Suzuki, C. *et al.* (2021) A sublethal ATP11A mutation associated with neurological deterioration causes aberrant phosphatidylcholine flipping in plasma membranes J

Clin Invest **131**, 10.1172/JCI148005

537. Shibata, Y., Matsushima, M., Matsukawa, T., Ishiura, H., Tsuji, S., andYabe, I. (2021) Adrenoleukodystrophy siblings with a novel ABCD1 missense variant presenting with phenotypic differences: a case report and literature review Journal of human genetics **66**, 535-537 10.1038/s10038-020-00866-x
538. Shimizu, M., Koibuchi, N., Mizugaki, A., Hishinuma, E., Saito, S., Hiratsuka, M. *et al.* (2021) Genetic variants of flavin-containing monooxygenase 3 (FMO3) in Japanese subjects identified by phenotyping for trimethylaminuria and found in a database of genome resources Drug Metab Pharmacokinet **38**, 100387 10.1016/j.dmpk.2021.100387
539. Shimizu, M., Mizugaki, A., Koibuchi, N., Sango, H., Uenuma, Y., andYamazaki, H. (2021) A series of simple detection systems for genetic variants of flavin-containing monooxygenase 3 (FMO3) with impaired function in Japanese subjects Drug Metab Pharmacokinet **41**, 100420 10.1016/j.dmpk.2021.100420
540. Shinya, Y., Hiraide, T., Momoi, M., Goto, S., Suzuki, H., Katsumata, Y. *et al.* (2021) TNFRSF13B c.226G>A (p.Gly76Ser) as a Novel Causative Mutation for Pulmonary Arterial Hypertension J Am Heart Assoc **10**, e019245 10.1161/JAHA.120.019245
541. Shirasawa, M., Yoshida, T., Shimoda, Y., Takayanagi, D., Shiraishi, K., Kubo, T. *et al.* (2021) Differential Immune-Related Microenvironment Determines Programmed Cell Death Protein-1/Programmed Death-Ligand 1 Blockade Efficacy in Patients With Advanced NSCLC J Thorac Oncol **16**, 2078-2090 10.1016/j.jtho.2021.07.027
542. Shirasawa, M., Yoshida, T., Takayanagi, D., Shiraishi, K., Yagishita, S., Sekine, K. *et al.* (2021) Activity and Immune Correlates of Programmed Death-1 Blockade Therapy in Patients With Advanced Large Cell Neuroendocrine Carcinoma Clin Lung Cancer **22**, 282-291 e286 10.1016/j.cllc.2021.02.003
543. Sugisawa, C., Ono, M., Kashimada, K., Hasegawa, T., andNarumi, S. (2021) Inactivation of a Frameshift TSH Receptor Variant Val711Phefs*18 is Due to Acquisition of a Hydrophobic Degron J Clin Endocrinol Metab **106**, e265-e272 10.1210/clinem/dgaa772
544. Suryamohan, K., Diwanji, D., Stawiski, E. W., Gupta, R., Miersch, S., Liu, J. *et al.* (2021) Human ACE2 receptor polymorphisms and altered susceptibility to SARS-CoV-2 Commun Biol **4**, 475 10.1038/s42003-021-02030-3
545. Suzuki, S., Goto, A., Nakatochi, M., Narita, A., Yamaji, T., Sawada, N. *et al.* (2021) Body mass index and colorectal cancer risk: A Mendelian randomization study Cancer Sci **112**, 1579-1588 10.1111/cas.14824
546. Takano, T., Ota, H., Ohishi, H., Hata, K., Furukawa, R., andNakabayashi, K. (2021) Adult acampomelic campomelic dysplasia and disorders of sex development due to a reciprocal translocation involving chromosome 17q24.3 upstream of the SOX9 gene Eur J Med Genet **64**, 104332 10.1016/j.ejmg.2021.104332
547. Takao, M., Yamaguchi, T., Eguchi, H., Yamada, T., Okazaki, Y., Tomita, N. *et al.* (2021) APC germline variant analysis in the adenomatous polyposis phenotype in Japanese patients Int J Clin Oncol **26**, 1661-1670 10.1007/s10147-021-01946-4
548. Takaragawa, M., Tobina, T., Shiose, K., Kakigi, R., Tsuzuki, T., Ichinoseki-Sekine, N. *et al.* (2021) Genotype Score for Iron Status Is Associated with Muscle Fiber Composition in Women Genes (Basel) **13**, 5 10.3390/genes13010005
549. Takizawa, N., Tanaka, S., Nishimoto, K., Sugiura, Y., Suematsu, M., Ohe, C. *et al.* (2021) Familial Hyperaldosteronism Type 3 with a Rapidly Growing Adrenal Tumor: An In Situ Aldosterone Imaging Study Curr Issues Mol Biol **44**, 128-138 10.3390/cimb44010010
550. Taniguchi, K., Inoue, M., Arai, K., Uchida, K., Migita, O., Akemoto, Y. *et al.* (2021) Novel TNFAIP3 microdeletion in a girl with infantile-onset inflammatory bowel disease complicated by a severe perianal lesion Hum Genome

Var **8**, 1 10.1038/s41439-020-00128-4

551. Taniguchi, T., Ando, M., Okamoto, Y., Yoshimura, A., Higuchi, Y., Hashiguchi, A. *et al.* (2021) Genetic spectrum of Charcot-Marie-Tooth disease associated with myelin protein zero gene variants in Japan Clin Genet **99**, 359-375 10.1111/cge.13881
552. Tsuyuki, S., Takeshima, H., Sekine, S., Yamagata, Y., Ando, T., Yamashita, S. *et al.* (2021) Comparable genetic alteration profiles between gastric cancers with current and past Helicobacter pylori infection Sci Rep **11**, 23443 10.1038/s41598-021-02761-7
553. Uchiyama, Y., Yamaguchi, D., Iwama, K., Miyatake, S., Hamanaka, K., Tsuchida, N. *et al.* (2021) Efficient detection of copy-number variations using exome data: Batch- and sex-based analyses Hum Mutat **42**, 50-65 10.1002/humu.24129
554. Uehara, D. T., Mitsubuchi, H., andInazawa, J. (2021) A missense variant in NUF2, a component of the kinetochore NDC80 complex, causes impaired chromosome segregation and aneuploidy associated with microcephaly and short stature Hum Genet **140**, 1047-1060 10.1007/s00439-021-02273-4
555. Uehara, T., Sanuki, R., Ogura, Y., Yokoyama, A., Yoshida, T., Futagawa, H. *et al.* (2021) Recurrent NFIA K125E substitution represents a loss-of-function allele: Sensitive in vitro and in vivo assays for nontruncating alleles Am J Med Genet A **185**, 2084-2093 10.1002/ajmg.a.62226
556. Umehara, H., Nakamura, M., Nagai, M., Kato, Y., Ueno, S. I., andSano, A. (2021) Positional cloning and comprehensive mutation analysis of a Japanese family with lithium-responsive bipolar disorder identifies a novel DOCK5 mutation Journal of human genetics **66**, 243-249 10.1038/s10038-020-00840-7
557. Ura, H., Togi, S., andNiida, Y. (2021) Targeted Double-Stranded cDNA Sequencing-Based Phase Analysis to Identify Compound Heterozygous Mutations and Differential Allelic Expression Biology (Basel) **10**, 256 10.3390/biology10040256
558. Usui, Y., Matsuo, K., Oze, I., Ugai, T., Koyanagi, Y., Maeda, Y. *et al.* (2021) Impact of PSCA Polymorphisms on the Risk of Duodenal Ulcer J Epidemiol **31**, 12-20 10.2188/jea.JE20190184
559. Watanabe, D., Yagasaki, H., Narusawa, H., Saito, T., Mitsui, Y., Miyake, K. *et al.* (2021) Screening of frequent variants associated with congenital hypothyroidism: a comparison with next generation sequencing Endocr J **68**, 1411-1419 10.1507/endocrj.EJ21-0353
560. Watanabe, K., Nakashima, M., Kumada, S., Mashimo, H., Enokizono, M., Yamada, K. *et al.* (2021) Identification of two novel de novo TUBB variants in cases with brain malformations: case reports and literature review Journal of human genetics **66**, 1193-1197 10.1038/s10038-021-00956-4
561. Yamaguchi, H., Nagase, H., Tokumoto, S., Tomioka, K., Nishiyama, M., Takeda, H. *et al.* (2021) Prevalence of Wilson disease based on genome databases in Japan Pediatrics international : official journal of the Japan Pediatric Society **63**, 918-922 10.1111/ped.14565
562. Yamamoto, J., Yamamoto, M., Takano, K., Okazaki, T., Arakawa, R., Hara, H. *et al.* (2021) Venous thromboembolism is caused by prothrombin p.Arg541Trp mutation in Japanese individuals Hum Genome Var **8**, 13 10.1038/s41439-021-00145-x
563. Yamazaki, H., Nakamura, T., Hosono, K., Yamaguchi, T., Hiratsuka, Y., Hotta, Y. *et al.* (2021) Sensorineural hearing loss and hypoplastic cochlea in Axenfeld-Rieger syndrome with FOXC1 mutation Auris Nasus Larynx **48**, 1204-1208 10.1016/j.anl.2020.07.006
564. Yokotsuka-Ishida, S., Nakamura, M., Tomiyasu, Y., Nagai, M., Kato, Y., Tomiyasu, A. *et al.* (2021) Positional cloning and comprehensive mutation analysis identified a novel KDM2B mutation in a Japanese family with

minor malformations, intellectual disability, and schizophrenia Journal of human genetics **66**, 597-606 10.1038/s10038-020-00889-4

565. Yoshida, K., Mushimoto, Y., Tanase-Nakao, K., Akiba, K., Ishii, K., Urakami, T. *et al.* (2021) A case report with functional characterization of a HNF1B mutation (p.Leu168Pro) causing MODY5 Clin Pediatr Endocrinol **30**, 179-185 10.1297/cpe.30.179
566. Zempo, H., Kim, S. J., Fuku, N., Nishida, Y., Higaki, Y., Wan, J. *et al.* (2021) A pro-diabetogenic mtDNA polymorphism in the mitochondrial-derived peptide, MOTS-c Aging (Albany NY) **13**, 1692-1717 10.18632/aging.202529
567. 加藤創生, 古家美菜絵, 大野久美子, 齋藤琢, and 伊東伸朗 (2021) 骨形成不全症による二次性骨粗鬆症に対するロモソズマブの効果 日本内分泌学会雑誌 **97**, 50-52,
568. 丸山慶子, and 小亀浩市 (2021) 公開データベースと発現実験を用いたアンチトロンビン, プロテイン C およびプロテイン S 遺伝子の病的バリアントの頻度推定 日本血栓止血学会誌 **32**, 635-637,
569. 前川素子, and 和田唯奈 (2021) 核内受容体 PPARα に着目した統合失調症病態メカニズムの解明 日本生物学的精神医学会誌 **32**, 144-148,
570. 中村憲一郎, 松田貴雄, 花岡拓哉, 後藤勝政, and 松原悦朗 (2021) 疾患関連遺伝子エクソーム解析によりLMNA遺伝子変異を認めたラミノパチーの 1 例 臨床神経学 **61**, 663-670 10.5692/clinicalneurol.cn-001610
571. Abe, A., Imoto, I., Tange, S., Nishimura, M., andIwasa, T. (2022) Prevalence of Pathogenic Germline BRCA1/2 Variants and Their Association with Clinical Characteristics in Patients with Epithelial Ovarian Cancer in a Rural Area of Japan Genes (Basel) **13**, 1085 10.3390/genes13061085
572. Abe, K., Kitago, M., Kosaki, K., Yamada, M., Iwasaki, E., Kawasaki, S. *et al.* (2022) Genomic analysis of familial pancreatic cancers and intraductal papillary mucinous neoplasms: A cross-sectional study Cancer Sci **113**, 1821-1829 10.1111/cas.15316
573. Al-Tuama, J. A. (2022) Lack of Association Between PTPN22 1858 C> T Gene Polymorphism and Susceptibility to Generalized Vitiligo in a Iraqi Population Iraqi journal of biotechnology **21**,
574. An, Y., andLee, C. (2022) Mixed model-based eQTL analysis reveals lncRNAs associated with regulation of genes involved in sex determination and spermatogenesis: The key to understanding human gender imbalance Comput Biol Chem **99**, 107713 10.1016/j.compbiolchem.2022.107713
575. Ando, M., Higuchi, Y., Okamoto, Y., Yuan, J., Yoshimura, A., Takei, J. *et al.* (2022) An NEFH founder mutation causes broad phenotypic spectrum in multiple Japanese families Journal of human genetics **67**, 399-403 10.1038/s10038-022-01019-y
576. Ando, M., Higuchi, Y., Takeuchi, M., Hashiguchi, A., andTakashima, H. (2022) The first case of infantile-onset multisystem neurologic, endocrine, and pancreatic disease caused by novel PTRH2 mutation in Japan Neurol Sci **43**, 2133-2136 10.1007/s10072-021-05817-8
577. Ando, M., Higuchi, Y., Yuan, J., Yoshimura, A., Taniguchi, T., Kojima, F. *et al.* (2022) Comprehensive Genetic Analyses of Inherited Peripheral Neuropathies in Japan: Making Early Diagnosis Possible Biomedicines **10**, 1546 10.3390/biomedicines10071546
578. Ando, M., Higuchi, Y., Yuan, J., Yoshimura, A., Taniguchi, T., Takei, J. *et al.* (2022) Novel heterozygous variants of SLC12A6 in Japanese families with Charcot-Marie-Tooth disease Ann Clin Transl Neurol **9**, 902-911 10.1002/acn3.51603
579. Ando, M., Higuchi, Y., Yuan, J. H., Yoshimura, A., Kitao, R., Morimoto, T. *et al.* (2022) Novel de novo POLR3B mutations responsible for demyelinating Charcot-Marie-Tooth disease in Japan Ann Clin Transl Neurol **9**, 747-

755 10.1002/acn3.51555

580. Asanomi, Y., Shigemizu, D., Akiyama, S., Miyashita, A., Mitsumori, R., Hara, N. *et al.* (2022) A functional variant of SHARPIN confers increased risk of late-onset Alzheimer's disease Journal of human genetics **67**, 203-208 10.1038/s10038-021-00987-x

581. Baba, K., Fukuda, T., Furuta, M., Tada, S., Imai, A., Asano, Y. *et al.* (2022) A Mild Clinical Phenotype with Myopathic and Hemolytic Forms of Phosphoglycerate Kinase Deficiency (PGK Osaka): A Case Report and Literature Review Intern Med **61**, 3589-3594 10.2169/internalmedicine.9221-21

582. Berry, V., Fujinami, K., Mochizuki, K., Iwata, T., Pontikos, N., Quinlan, R. A. *et al.* (2022) A recurrent variant in LIM2 causes an isolated congenital sutural/lamellar cataract in a Japanese family Ophthalmic Genet **43**, 622-626 10.1080/13816810.2022.2090010

583. Chen, H. D., Yu, C. C., Yang, I. H., Hung, C. C., Kuo, M. C., Tarng, D. C. *et al.* (2022) UMOD Mutations in Chronic Kidney Disease in Taiwan Biomedicines **10**, 10.3390/biomedicines10092265

584. Cong, P. K., Bai, W. Y., Li, J. C., Yang, M. Y., Khederzadeh, S., Gai, S. R. *et al.* (2022) Genomic analyses of 10,376 individuals in the Westlake BioBank for Chinese (WBBC) pilot project Nat Commun **13**, 2939 10.1038/s41467-022-30526-x

585. Daida, K., Nishioka, Y., Li, Y., Yoshino, H., Funayama, M., Hattori, N. *et al.* (2022) A complex form of hereditary spastic paraplegia harboring a novel variant, p.W1515*, in the SPG11 gene eNeurologicalSci **26**, 100391 10.1016/j.ensci.2021.100391

586. Daida, K., Shimonaka, S., Shiba-Fukushima, K., Ogata, J., Yoshino, H., Okuzumi, A. *et al.* (2022) alpha-Synuclein V15A Variant in Familial Parkinson's Disease Exhibits a Weaker Lipid-Binding Property Mov Disord **37**, 2075-2085 10.1002/mds.29162

587. Ebihara, T., Nagatomo, T., Sugiyama, Y., Tsuruoka, T., Osone, Y., Shimura, M. *et al.* (2022) Severe spinal cord hypoplasia due to a novel ATAD3A compound heterozygous deletion Mol Genet Metab Rep **33**, 100912 10.1016/j.ymgmr.2022.100912

588. Enomoto, N., Hayashi, T., Matsuura, T., Tanaka, K., Takeuchi, R., Tomita, G. *et al.* (2022) The second Japanese family with Malattia Leventinese/Doyne honeycomb retinal dystrophy Doc Ophthalmol **144**, 67-75 10.1007/s10633-021-09859-3

589. Enomoto, Y., Yokoi, T., Tsurusaki, Y., Murakami, H., Tominaga, M., Minatogawa, M. *et al.* (2022) Divergent variant patterns among 19 patients with Rubinstein-Taybi syndrome uncovered by comprehensive genetic analysis including whole genome sequencing Clin Genet **101**, 335-345 10.1111/cge.14103

590. Fan, L., Miura, S., Shimojo, T., Sugino, H., Fujioka, R., andShibata, H. (2022) A novel 1-bp deletion variant in DAG1 in Japanese familial asymptomatic hyper-CK-emia Hum Genome Var **9**, 4 10.1038/s41439-022-00182-0

591. Fujise, K., Okubo, M., Abe, T., Yamada, H., Takei, K., Nishino, I. *et al.* (2022) Imaging-based evaluation of pathogenicity by novel DNM2 variants associated with centronuclear myopathy Hum Mutat **43**, 169-179 10.1002/humu.24307

592. Fujita, S., Horitani, E., Miyashita, Y., Fujita, Y., Fukui, K., Kamada, Y. *et al.* (2022) Whole-exome Sequencing Analysis of a Japanese Patient With Hyperinsulinemia and Liver Dysfunction J Endocr Soc **6**, bvac008 10.1210/jendso/bvac008

593. Fujita, S., Nishizawa, H., Miyashita, Y., Imada, T., Yamaguchi, T., Murano, T. *et al.* (2022) Genetic assessment using whole-exome sequencing for a young hypertriglyceridemic patient with repeated acute pancreatitis Endocr J **69**, 1101-1108 10.1507/endocrj.EJ22-0024

594. Furukawa, H., Oka, S., Kondo, N., Nakagawa, Y., Shiota, N., Kumagai, K. *et al.* (2022) The Contribution of Deleterious Rare Alleles in ENPP1 and Osteomalacia Causative Genes to Atypical Femoral Fracture J Clin Endocrinol Metab **107**, e1890-e1898 10.1210/clinem/dgac022

595. Furukawa, S., Miyamoto, S., Fukumura, S., Kubota, K., Taga, T., Nakashima, M. *et al.* (2022) Two novel heterozygous variants in ATP1A3 cause movement disorders Hum Genome Var **9**, 7 10.1038/s41439-022-00184-y

596. Furuta, S., Aleksic, B., Nawa, Y., Kimura, H., Kushima, I., Ishizuka, K. *et al.* (2022) Investigation of OLIG2 as a candidate gene for schizophrenia and autism spectrum disorder Nagoya J Med Sci **84**, 260-268 10.18999/nagjms.84.2.260

597. Ganaha, A., Hishinuma, E., Kaname, T., Hiratsuka, M., Kondo, S., andTono, T. (2022) Rapid Genetic Diagnosis for Okinawan Patients with Enlarged Vestibular Aqueduct Using Single-Stranded Tag Hybridization Chromatographic Printed-Array Strip J Clin Med **11**, 1099 10.3390/jcm11041099

598. Goto, K., Kukita, Y., Honma, K., Ohike, N., Komori, T., Ishida, Y. *et al.* (2022) Sweat-gland carcinoma with neuroendocrine differentiation (SCAND): a clinicopathologic study of 13 cases with genetic analysis Mod Pathol **35**, 33-43 10.1038/s41379-021-00921-8

599. Green, T. E., Scheffer, I. E., Berkovic, S. F., andHildebrand, M. S. (2022) UNC13B and focal epilepsy Brain **145**, e10-e12 10.1093/brain/awab485

600. Gu, J. Q., Zhao, H., Guo, X. Y., Sun, H. Y., Xu, J. Y., andWei, Y. L. (2022) A high-performance SNP panel developed by machine-learning approaches for characterizing genetic differences of Southern and Northern Han Chinese, Korean, and Japanese individuals Electrophoresis **43**, 1183-1192 10.1002/elps.202100184

601. Hakui, H., Kioka, H., Miyashita, Y., Nishimura, S., Matsuoka, K., Kato, H. *et al.* (2022) Loss-of-function mutations in the co-chaperone protein BAG5 cause dilated cardiomyopathy requiring heart transplantation Sci Transl Med **14**, eabf3274 10.1126/scitranslmed.abf3274

602. Hama, Y., Date, H., Fujimoto, A., Matsui, A., Ishiura, H., Mitsui, J. *et al.* (2022) A Novel de novo KIF1A Mutation in a Patient with Ataxia, Intellectual Disability and Mild Foot Deformity Cerebellum 10.1007/s12311-022-01489-y

603. Hamanaka, K., Miyoshi, K., Sun, J. H., Hamada, K., Komatsubara, T., Saida, K. *et al.* (2022) Amelioration of a neurodevelopmental disorder by carbamazepine in a case having a gain-of-function GRIA3 variant Hum Genet **141**, 283-293 10.1007/s00439-021-02416-7

604. Hanafusa, H., Abe, S., Ohyama, S., Kyono, Y., Kido, T., Nakasone, R. *et al.* (2022) Influence of UGT1A1 Genetic Variants on Free Bilirubin Levels in Japanese Newborns: A Preliminary Study Int J Environ Res Public Health **19**, 13090 10.3390/ijerph192013090

605. Hara-Isono, K., Nakamura, A., Fuke, T., Inoue, T., Kawashima, S., Matsubara, K. *et al.* (2022) Pathogenic Copy Number and Sequence Variants in Children Born SGA With Short Stature Without Imprinting Disorders J Clin Endocrinol Metab **107**, e3121-e3133 10.1210/clinem/dgac319

606. Harada-Shiba, M., Ako, J., Hirayama, A., Nakamura, M., Nohara, A., Sato, K. *et al.* (2022) Familial Hypercholesterolemia in Patients with Acute Coronary Syndrome: Genetic Insights from EXPLORE-J J Atheroscler Thromb **29**, 1201-1212 10.5551/jat.62989

607. Hasegawa, M., Fukushima, H., Suzuki, R., Yamaki, Y., Hosaka, S., Inaba, M. *et al.* (2022) Effect of Germline MEFV Polymorphisms on the Prognosis of Japanese Children with Cancer: A Regional Analysis Oncology **100**, 376-383 10.1159/000524833

608. Hashiguchi, M., Monden, Y., Nozaki, Y., Watanabe, K., Nakashima, M., Saitsu, H. *et al.* (2022) A TUBB4A Met363Thr variant in pediatric hypomyelination without atrophy of the basal ganglia Hum Genome Var **9**, 19 10.1038/s41439-022-00198-6

609. Hatano, M., Udagawa, T., Kanamori, T., Sutani, A., Mori, T., Sohara, E. *et al.* (2022) A novel SLC5A2 heterozygous variant in a family with familial renal glucosuria Hum Genome Var **9**, 42 10.1038/s41439-022-00221-w

610. Hayakawa, M., Matsubara, T., Mochizuki, Y., Takeuchi, C., Minamitani, M., Imai, M. *et al.* (2022) An autopsied case report of spastic paraplegia with thin corpus callosum carrying a novel mutation in the SPG11 gene: widespread degeneration with eosinophilic inclusions BMC Neurol **22**, 2 10.1186/s12883-021-02514-z

611. Higashigawa, S., Matsubayashi, H., Kiyozumi, Y., Kado, N., Nishimura, S., Oishi, T. *et al.* (2022) Present status of germline findings in precision medicine for Japanese cancer patients: issues in the current system Jpn J Clin Oncol **52**, 599-608 10.1093/jjco/hyac046

612. Hirabayashi, Y., Katayama, K., Mori, M., Matsuo, H., Fujimoto, M., Joh, K. *et al.* (2022) Mutation Analysis of Thin Basement Membrane Nephropathy Genes (Basel) **13**, 10.3390/genes13101779

613. Hiraide, T., Shimizu, K., Miyamoto, S., Aoto, K., Nakashima, M., Yamaguchi, T. *et al.* (2022) Genome sequencing and RNA sequencing of urinary cells reveal an intronic FBN1 variant causing aberrant splicing Journal of human genetics **67**, 387-392 10.1038/s10038-022-01016-1

614. Hiraide, T., Suzuki, H., Momoi, M., Shinya, Y., Fukuda, K., Kosaki, K. *et al.* (2022) RNF213-Associated Vascular Disease: A Concept Unifying Various Vasculopathies Life (Basel) **12**, 555 10.3390/life12040555

615. Hiraide, T., Suzuki, H., Shinya, Y., Momoi, M., Inami, T., Katsumata, Y. *et al.* (2022) TET2 Variants in Japanese Patients With Pulmonary Arterial Hypertension CJC Open **4**, 416-419 10.1016/j.cjco.2021.11.008

616. Hiramatsu, Y., Okamoto, Y., Yoshimura, A., Yuan, J. H., Ando, M., Higuchi, Y. *et al.* (2022) Complex hereditary peripheral neuropathies caused by novel variants in mitochondrial-related nuclear genes J Neurol **269**, 4129-4140 10.1007/s00415-022-11026-w

617. Homma, H., Saito, M., Saito, A., Kozuma, A., Matsumoto, R., Matsumoto, S. *et al.* (2022) The Association between Total Genotype Score and Athletic Performance in Weightlifters Genes (Basel) **13**, 10.3390/genes13112091

618. Horckmans, M., Diaz Villamil, E., Verdier, C., Laurell, H., Ruidavets, J. B., De Roeck, L. *et al.* (2022) Loss-of-function N178T variant of the human P2Y(4) receptor is associated with decreased severity of coronary artery disease and improved glucose homeostasis Front Pharmacol **13**, 1049696 10.3389/fphar.2022.1049696

619. Hori, A., Migita, O., Isogawa, N., Takada, F., andHata, K. (2022) A novel TP63 variant in a patient with ankyloblepharon-ectodermal defect-cleft lip/palate syndrome and Rapp-Hodgkin syndrome-like ectodermal dysplasia Hum Genome Var **9**, 17 10.1038/s41439-022-00186-w

620. Hori, M., Takahashi, A., Hosoda, K., andHarada-Shiba, M. (2022) Identification of a novel large duplication (exon2_6dup): copy number variation in the LDLR gene in a large family with familial hypercholesterolemia by whole-genome sequencing J Clin Lipidol **16**, 167-172 10.1016/j.jacl.2022.01.007

621. Huang, D., Zhou, Y., Yi, X., Fan, X., Wang, J., Yao, H. *et al.* (2022) VannoPortal: multiscale functional annotation of human genetic variants for interrogating molecular mechanism of traits and diseases Nucleic Acids Res **50**, D1408-D1416 10.1093/nar/gkab853

622. Iio, K., Mori, T., Bessho, S., Imai, Y., Hatanaka, M., Omori, H. *et al.* (2022) Gitelman syndrome with a novel frameshift variant in SLC12A3 gene accompanied by chronic kidney disease and type 2 diabetes mellitus CEN Case Rep **11**, 191-195 10.1007/s13730-021-00652-4

623. Ishida, A., Mizuno, H., Aoyama, K., Sasaki, S., Negishi, Y., Arakawa, T. *et al.* (2022) Partial nephrogenic diabetes insipidus with a novel arginine vasopressin receptor 2 gene variant Clin Pediatr Endocrinol **31**, 44-49 10.1297/cpe.2021-0029

624. Ishigami, D., Koizumi, S., Miyawaki, S., Hongo, H., Teranishi, Y., Mitsui, J. *et al.* (2022) Symptomatic and Stenotic Developmental Venous Anomaly with Pontine Capillary Telangiectasia: A Case Report with Genetic Considerations NMC Case Rep J **9**, 139-144 10.2176/jns-nmc.2022-0022

625. Itoh, M., Okajima, M., Kittaka, Y., Yachie, A., Wada, T., andSaikawa, Y. (2022) Tertiary hyperparathyroidism in patients with pseudohypoparathyroidism type 1a Bone Rep **16**, 101569 10.1016/j.bonr.2022.101569

626. Jain, P. K., Jayappa, S., Sairam, T., Mittal, A., Paul, S., Rao, V. J. *et al.* (2022) Ribosomal protein S6 kinase beta-1 gene variants cause hypertrophic cardiomyopathy J Med Genet **59**, 984-992 10.1136/jmedgenet-2021-107866

627. Kaimori, J. Y., Kikkawa, Y., Motooka, D., Namba-Hamano, T., Takuwa, A., Okazaki, A. *et al.* (2022) A heterozygous LAMA5 variant may contribute to slowly progressive, vinculin-enhanced familial FSGS and pulmonary defects JCI Insight **7**, 10.1172/jci.insight.158378

628. Kakehi, R., Hori, H., Yoshida, F., Itoh, M., Lin, M., Niwa, M. *et al.* (2022) Hypothalamic-pituitary-adrenal axis and renin-angiotensin-aldosterone system in adulthood PTSD and childhood maltreatment history Front Psychiatry **13**, 967779 10.3389/fpsyt.2022.967779

629. Kanzaki, R., Morino, H., Mukaida, K., Kume, K., Ohsawa, R., Noda, Y. *et al.* (2022) Genetic screening for malignant hyperthermia and comparison of clinical symptoms in Japan Hiroshima Journal of Medical Sciences **71**, 31-38 10.24811/hjms.71.1-2_31

630. Kasugai, Y., Kohmoto, T., Taniyama, Y., Koyanagi, Y. N., Usui, Y., Iwase, M. *et al.* (2022) Association between germline pathogenic variants and breast cancer risk in Japanese women: The HERPACC study Cancer Sci **113**, 1451-1462 10.1111/cas.15312

631. Katata, Y., Uneoka, S., Saijyo, N., Aihara, Y., Miyazoe, T., Koyamaishi, S. *et al.* (2022) The longest reported sibling survivors of a severe form of congenital myasthenic syndrome with the ALG14 pathogenic variant Am J Med Genet A **188**, 1293-1298 10.1002/ajmg.a.62629

632. Katayanagi, S., Setoguchi, Y., Kitagawa, S., Okamoto, T., andMiyazaki, Y. (2022) Alternative Gene Expression by TOLLIP Variant Is Associated With Lung Function in Chronic Hypersensitivity Pneumonitis Chest **161**, 458-469 10.1016/j.chest.2021.08.052

633. Kato, H., Ansh, A. J., Lester, E. R., Kinoshita, Y., Hidaka, N., Hoshino, Y. *et al.* (2022) Identification of ENPP1 Haploinsufficiency in Patients With Diffuse Idiopathic Skeletal Hyperostosis and Early-Onset Osteoporosis J Bone Miner Res **37**, 1125-1135 10.1002/jbmr.4550

634. Kato, H., Koga, M., Kinoshita, Y., Hidaka, N., Hoshino, Y., Takashi, Y. *et al.* (2022) Utility of Multimodality Approach Including Systemic FGF23 Venous Sampling in Localizing Phosphaturic Mesenchymal Tumors J Endocr Soc **7**, bvac181 10.1210/jendso/bvac181

635. Kato, M., Kudo, Y., Hatase, M., Tsuchida, N., Takeyama, S., Sugiyama, T. *et al.* (2022) Moyamoya Disease Associated with a Deficiency of Complement Component 6 J Stroke Cerebrovasc Dis **31**, 106601 10.1016/j.jstrokecerebrovasdis.2022.106601

636. Kawashima, I., Oishi, N., Kasai, K., Inoue, T., Hosokawa, E., Nakadate, A. *et al.* (2022) Transdifferentiation of mantle cell lymphoma into sarcoma with limited neuromuscular differentiation after conventional chemotherapy Virchows Arch **480**, 1101-1105 10.1007/s00428-021-03148-9

637. Kimura, H., Nakatochi, M., Aleksic, B., Guevara, J., Toyama, M., Hayashi, Y. *et al.* (2022) Exome sequencing analysis of Japanese autism spectrum disorder case-control sample supports an increased burden of synaptic function-related genes Transl Psychiatry **12**, 265 10.1038/s41398-022-02033-6

638. Kobayashi Kato, M., Asami, Y., Takayanagi, D., Matsuda, M., Shimada, Y., Hiranuma, K. *et al.* (2022) Clinical impact of genetic alterations of CTNNB1 in patients with grade 3 endometrial endometrioid carcinoma Cancer Sci **113**, 1712-1721 10.1111/cas.15328

639. Kobayashi, Y., Ito, Y., Taniguchi, K., Harada, K., Yamamura, M., Sato, T. *et al.* (2022) Novel missense COL2A1 variant in a fetus with achondrogenesis type II Hum Genome Var **9**, 40 10.1038/s41439-022-00218-5

640. Komatsu, K., Fukumura, S., Minagawa, K., Nakashima, M., andSaitsu, H. (2022) A new case of concurrent existence of PRRT2-associated paroxysmal movement disorders with c.649dup variant and 16p11.2 microdeletion syndrome Brain Dev **44**, 474-479 10.1016/j.braindev.2022.03.008

641. Kondo, T., Hara, N., Koyama, S., Yada, Y., Tsukita, K., Nagahashi, A. *et al.* (2022) Dissection of the polygenic architecture of neuronal Abeta production using a large sample of individual iPSC lines derived from Alzheimer's disease patients Nat Aging **2**, 125-139 10.1038/s43587-021-00158-9

642. Kotan, L. D. (2022) Comparative Analyses of Turkish Variome and Widely Used Genomic Variation Databases for the Evaluation of Rare Sequence Variants in Turkish Individuals: Idiopathic Hypogonadotropic Hypogonadism as a Disease Model J Clin Res Pediatr Endocrinol **14**, 293-301 10.4274/jcrpe.galenos.2022.2022-3-11

643. Kurata, K., Hosono, K., Takayama, M., Katsuno, M., Saitsu, H., Ogata, T. *et al.* (2022) Retinitis pigmentosa with optic neuropathy and COQ2 mutations: A case report Am J Ophthalmol Case Rep **25**, 101298 10.1016/j.ajoc.2022.101298

644. Kurelac, I., Cavina, B., Sollazzo, M., Miglietta, S., Fornasa, A., De Luise, M. *et al.* (2022) NDUFS3 knockout cancer cells and molecular docking reveal specificity and mode of action of anti-cancer respiratory complex I inhibitors Open Biol **12**, 220198 10.1098/rsob.220198

645. Kurihara, S., Matsui, H., Ohtake, N., Aoki, M., Sekine, Y., Arai, S. *et al.* (2022) Variants in HOXB13, G132E and F127C, Are Associated With Prostate Cancer Risk in Japanese Men Cancer Diagn Progn **2**, 542-548 10.21873/cdp.10139

646. Li, B., Liu, Q., Chen, X., Chen, T., Dang, W., Zhao, J. *et al.* (2022) A Novel Idiopathic Atrial Calcification: Pathologic Manifestations and Potential Mechanism Front Cardiovasc Med **9**, 788958 10.3389/fcvm.2022.788958

647. Li, X., Peng, S., Wu, X., Guan, B., Tse, G., Chen, S. *et al.* (2022) C-reactive protein and atrial fibrillation: Insights from epidemiological and Mendelian randomization studies Nutr Metab Cardiovasc Dis **32**, 1519-1527 10.1016/j.numecd.2022.03.008

648. Lo, T., Kushima, I., Aleksic, B., Kato, H., Nawa, Y., Hayashi, Y. *et al.* (2022) Sequencing of selected chromatin remodelling genes reveals increased burden of rare missense variants in ASD patients from the Japanese population Int Rev Psychiatry **34**, 154-167 10.1080/09540261.2022.2072193

649. Lv, L., Sun, X., Liu, B., Song, J., Wu, D. J. H., Gao, Y. *et al.* (2022) Genetically Predicted Serum Albumin and Risk of Colorectal Cancer: A Bidirectional Mendelian Randomization Study Clin Epidemiol **14**, 771-778 10.2147/CLEP.S367547

650. Masunaga, Y., Nishimura, G., Takahashi, K., Hishiyama, T., Imamura, M., Kashimada, K. *et al.* (2022) Clinical and molecular findings in three Japanese patients with N-acetylneuraminic acid synthetase-congenital disorder of glycosylation (NANS-CDG) Sci Rep **12**, 17079 10.1038/s41598-022-21751-x

651. Matsumae, T., Kodama, T., Myojin, Y., Maesaka, K., Sakamori, R., Takuwa, A. *et al.* (2022) Circulating Cell-Free DNA Profiling Predicts the Therapeutic Outcome in Advanced Hepatocellular Carcinoma Patients Treated with Combination Immunotherapy Cancers (Basel) **14**, 3367 10.3390/cancers14143367

652. Matsumoto, A., Tsuda, H., Furui, S., Kawada-Nagashima, M., Anzai, T., Seki, M. *et al.* (2022) A case of congenital fiber-type disproportion syndrome presenting dilated cardiomyopathy with ACTA1 mutation Mol Genet Genomic Med **10**, e2008 10.1002/mgg3.2008

653. Matsumoto, H., Sasai, H., Kawamoto, N., Katsuyama, M., Minamiyama, M., Kuru, S. *et al.* (2022) Founder genetic variants of ABCC4 and ABCC11 in the Japanese population are not associated with the development of subacute myelo-optico-neuropathy (SMON) Mol Genet Genomic Med **10**, e1845 10.1002/mgg3.1845

654. Matsuo, T., Chaomulige, Miyaji, M., Hosoya, O., Saito, A., andNakazono, K. (2022) Candidate Genes in Testing Strategies for Linkage Analysis and Bioinformatic Sorting of Whole Genome Sequencing Data in Three Small Japanese Families with Idiopathic Superior Oblique Muscle Palsy Int J Mol Sci **23**, 8626 10.3390/ijms23158626

655. Matsushita, H. B., Hiraide, T., Hayakawa, K., Okano, S., Nakashima, M., Saitsu, H. *et al.* (2022) Compound heterozygous ADAMTS9 variants in Joubert syndrome-related disorders without renal manifestation Brain Dev **44**, 161-165 10.1016/j.braindev.2021.10.004

656. Matsuyama, M., Sawada, H., Inoue, S., Hishinuma, A., Sekiya, R., Sato, Y. *et al.* (2022) Goiter in a 6-year-old patient with novel thyroglobulin gene variant (Gly145Glu) causing intracellular thyroglobulin transport disorder: Correlation between goiter size and the free T3 to free T4 ratio Clinical Pediatric Endocrinology **31**, 185-191 10.1297/cpe.31.2022-0006

657. Mishra, R., Kulshreshtha, S., Mandal, K., Khurana, A., Diego-Alvarez, D., Pradas, L. *et al.* (2022) COASY related pontocerebellar hypoplasia type 12: A common Indian mutation with expansion of the phenotypic spectrum Am J Med Genet A **188**, 2339-2350 10.1002/ajmg.a.62768

658. Mitsuhashi, N., Toyo-Oka, L., Katayama, T., Kawashima, M., Kawashima, S., Miyazaki, K. *et al.* (2022) TogoVar: A comprehensive Japanese genetic variation database Hum Genome Var **9**, 44 10.1038/s41439-022-00222-9

659. Miura, R., Kawaoka, T., Imamura, M., Kosaka, M., Johira, Y., Shirane, Y. *et al.* (2022) Benign Recurrent Intrahepatic Cholestasis Type 1 with Novel Nonsense Mutations in the ATP8B1 Gene Case Rep Gastroenterol **16**, 110-115 10.1159/000522145

660. Miyagawa, T., Shimada, M., Honda, Y., Kodama, T., Tokunaga, K., andHonda, M. (2022) A variant in orexin receptor-2 is associated with self-reported daytime sleepiness in the Japanese population Journal of human genetics **67**, 377-380 10.1038/s10038-022-01015-2

661. Miyagawa, T., Tanaka, S., Shimada, M., Sakai, N., Tanida, K., Kotorii, N. *et al.* (2022) A rare genetic variant in the cleavage site of prepro-orexin is associated with idiopathic hypersomnia NPJ Genom Med **7**, 29 10.1038/s41525-022-00298-w

662. Miyamoto, S., Nakashima, M., Fukumura, S., Kumada, S., andSaitsu, H. (2022) An intronic GNAO1 variant leading to in-frame insertion cause movement disorder controlled by deep brain stimulation Neurogenetics **23**, 129-135 10.1007/s10048-022-00686-5

663. Miyashita, K., Matsuda, Y., Okajima, M., Toma, T., Yachie, A., andWada, T. (2022) Role of E148Q in familial Mediterranean fever with an exon 10 mutation in MEFV Pediatrics international : official journal of the Japan Pediatric Society **64**, e14696 10.1111/ped.14696

664. Mizobuchi, K., Hayashi, T., Matsuura, T., andNakano, T. (2022) Clinical characterization of autosomal dominant retinitis pigmentosa with NRL mutation in a three-generation Japanese family Doc Ophthalmol **144**, 227-235

10.1007/s10633-022-09874-y

665. Mizumoto, K., Kato, K., Fujinami, K., Sugita, T., Sugita, I., Hattori, A. *et al.* (2022) A Japanese boy with Bardet-Biedl syndrome caused by a novel homozygous variant in the ARL6 gene who was initially diagnosed with retinitis punctata albescens: A case report Medicine (Baltimore) **101**, e32161 10.1097/MD.0000000000032161

666. Mori, T., Ueno, K., Tokunaga, K., Kawai, Y., Matsuda, K., Nishida, N. *et al.* (2022) A single-nucleotide-polymorphism in the 5'-flanking region of MSX1 gene as a predictive marker candidate for platinum-based therapy of esophageal carcinoma Ther Adv Med Oncol **14**, 17588359221080580 10.1177/17588359221080580

667. Morikawa, R., Watanabe, Y., Igeta, H., Arta, R. K., Ikeda, M., Okazaki, S. *et al.* (2022) Novel missense SETD1A variants in Japanese patients with schizophrenia: Resequencing and association analysis Psychiatry Res **310**, 114481 10.1016/j.psychres.2022.114481

668. Munekane, A., Ohsawa, Y., Fukuda, T., Nishimura, H., Nishimatsu, S. I., Sugie, H. *et al.* (2022) Maximal Multistage Shuttle Run Test-induced Myalgia in a Patient with Muscle Phosphorylase B Kinase Deficiency Intern Med **61**, 1241-1245 10.2169/internalmedicine.8137-21

669. Murakami, H., Tanimoto, Y., Tanimoto, K., Inoue, S., Ishikawa, T., Makita, N. *et al.* (2022) Arrhythmogenic right ventricular cardiomyopathy in a Japanese patient with a homozygous founder variant of DSG2 in the East Asian population Hum Genome Var **9**, 28 10.1038/s41439-022-00206-9

670. Murakami, H., Uehara, T., Enomoto, Y., Nishimura, N., Kumaki, T., Kuroda, Y. *et al.* (2022) Persistent Hyperplastic Primary Vitreous with Microphthalmia and Coloboma in a Patient with Okur-Chung Neurodevelopmental Syndrome Mol Syndromol **13**, 75-79 10.1159/000517977

671. Nagata, Y., Watanabe, R., Eichhorn, C., Ohno, S., Aiba, T., Ishikawa, T. *et al.* (2022) Targeted deep sequencing analyses of long QT syndrome in a Japanese population PLoS One **17**, e0277242 10.1371/journal.pone.0277242

672. Nakagawa, R., Takasawa, K., Gau, M., Tsuji-Hosokawa, A., Kawaji, H., Murakawa, Y. *et al.* (2022) Two ovarian candidate enhancers, identified by time series enhancer RNA analyses, harbor rare genetic variations identified in ovarian insufficiency Human molecular genetics **31**, 2223-2235 10.1093/hmg/ddac023

673. Nakajima, K., Suzuki, H., Yamamoto, M., Yamamoto, T., Kawai, T., Nakabayashi, K. *et al.* (2022) A familial case of periodontal Ehlers-Danlos syndrome lacking skin extensibility and joint hypermobility with a missense mutation in C1R J Dermatol **49**, 714-718 10.1111/1346-8138.16372

674. Nakajima, T., Kawabata-Iwakawa, R., Tamura, S., Hasegawa, H., Kobari, T., Itoh, H. *et al.* (2022) Novel CACNA1C R511Q mutation, located in domain Ⅰ-Ⅱ linker, causes non-syndromic type-8 long QT syndrome PLoS One **17**, e0271796 10.1371/journal.pone.0271796

675. Nakano, T., Sasahara, Y., Kikuchi, A., Moriya, K., Niizuma, H., Niihori, T. *et al.* (2022) Novel POLE mutations identified in patients with IMAGE-I syndrome cause aberrant subcellular localisation and protein degradation in the nucleus J Med Genet **59**, 1116-1122 10.1136/jmedgenet-2021-108300

676. Nakano, Y., Taira, Y., Sasaki, R., Tadokoro, K., Yunoki, T., Nomura, E. *et al.* (2022) Novel ABCD1 mutation detected in a symptomatic female carrier of adrenoleukodystrophy Neurology and Clinical Neuroscience **11**, 58-60 10.1111/ncn3.12667

677. Nan, H., Mizuno, T., Arisaka, A., Sei, K., andTakiyama, Y. (2022) A p.Glu420Gln mutation in SPAST is associated with infantile onset spastic paraplegia complicated by cerebella ataxia, epilepsy, peripheral neuropathy, and hypoplasia of the corpus callosum Neurol Sci **43**, 2123-2126 10.1007/s10072-022-05879-2

678. Narumi, S., Opitz, R., Nagasaki, K., Muroya, K., Asakura, Y., Adachi, M. *et al.* (2022) GWAS of thyroid dysgenesis identifies a risk locus at 2q33.3 linked to regulation of Wnt signaling Human molecular genetics **31**, 3967-3974

10.1093/hmg/ddac093

679. Nezu, M., Hirotsu, Y., Amemiya, K., Katsumata, M., Watanabe, T., Takizawa, S. *et al.* (2022) A case of juvenile-onset pheochromocytoma with KIF1B p.V1529M germline mutation Endocr J **69**, 705-716 10.1507/endocrj.EJ21-0475

680. Nishimori, Y., Iida, A., Ogasawara, M., Okubo, M., Yonenobu, Y., Kinoshita, M. *et al.* (2022) TNNI1 Mutated in Autosomal Dominant Proximal Arthrogryposis Neurol Genet **8**, e649 10.1212/NXG.0000000000000649

681. Nishio, S. Y., andUsami, S. I. (2022) Frequency of the STRC-CATSPER2 deletion in STRC-associated hearing loss patients Sci Rep **12**, 634 10.1038/s41598-021-04688-5

682. Nishio, S. Y., andUsami, S. I. (2022) Prevalence and clinical features of autosomal dominant and recessive TMC1-associated hearing loss Hum Genet **141**, 929-937 10.1007/s00439-021-02364-2

683. Nishiyama, M., Takahashi, K., Morizumi, S., Takahashi, Y., Iwamura, S., Sumitomo, K. *et al.* (2022) Transient and Recurrent Pulmonary Infiltrations Associated with Familial Mediterranean Fever Intern Med **61**, 3415-3419 10.2169/internalmedicine.8951-21

684. Nohara, F., Tajima, G., Sasai, H., andMakita, Y. (2022) MCAD deficiency caused by compound heterozygous pathogenic variants in ACADM Hum Genome Var **9**, 2 10.1038/s41439-021-00177-3

685. Obara, K., Abe, E., andToyoshima, I. (2022) Dyggve-Melchior-Clausen Syndrome Caused by a Novel Frameshift Variant in a Japanese Patient Mol Syndromol **13**, 350-359 10.1159/000521516

686. Oda, I., Danno, D., Saigoh, K., Wolf, J., Kawashita, N., Hirano, M. *et al.* (2022) Hemiplegic migraine type 2 with new mutation of the ATP1A2 gene in Japanese cases Neurosci Res **180**, 83-89 10.1016/j.neures.2022.03.002

687. Ohkawa, T., Nishimura, A., Kosaki, K., Aoki-Nogami, Y., Tomizawa, D., Kashimada, K. *et al.* (2022) PAX3/7-FOXO1 fusion-negative alveolar rhabdomyosarcoma in Schuurs-Hoeijmakers syndrome Journal of human genetics **67**, 51-54 10.1038/s10038-021-00965-3

688. Ohnami, S., Naruoka, A., Isaka, M., Mizuguchi, M., Nakatani, S., Kamada, F. *et al.* (2022) Comparison of genetic susceptibility to lung adenocarcinoma and squamous cell carcinoma in Japanese patients using a novel panel for cancer-related drug-metabolizing enzyme genes Sci Rep **12**, 17928 10.1038/s41598-022-22914-6

689. Oishi, N., Segawa, T., Miyake, K., Mochizuki, K., andKondo, T. (2022) Incidence, clinicopathological features and genetics of in-situ follicular neoplasia: a comprehensive screening study in a Japanese cohort Histopathology **80**, 820-826 10.1111/his.14617

690. Okamoto, N., Miya, F., Tsunoda, T., Kanemura, Y., Saitoh, S., Kato, M. *et al.* (2022) Four pedigrees with aminoacyl-tRNA synthetase abnormalities Neurol Sci **43**, 2765-2774 10.1007/s10072-021-05626-z

691. Padhi, S., Sarangi, S., Nayak, N., Pati, A., andPanda, A. K. (2022) OAS1 rs1131454 genetic variant is associated with Alzheimer's disease: an epidemiological analysis Brain **145**, e61-e63 10.1093/brain/awac132

692. Pan, Y., Suga, A., Kimura, I., Kimura, C., Minegishi, Y., Nakayama, M. *et al.* (2022) METTL23 mutation alters histone H3R17 methylation in normal-tension glaucoma J Clin Invest **132**, 10.1172/JCI153589

693. Park, H., Hosomichi, K., Kim, Y. I., Hikita, Y., Tajima, A., andYamaguchi, T. (2022) Comprehensive Genetic Exploration of Fused Teeth by Whole Exome Sequencing Appl Sci-Basel **12**, 11899 ARTN 11899 10.3390/app122311899

694. Park, H., Hosomichi, K., Kim, Y. I., Tajima, A., andYamaguchi, T. (2022) Exploring the Genetic Basis of Dens Evaginatus Using Whole-Exome Sequencing Appl Sci-Basel **12**, 8962 ARTN 8962 10.3390/app12188962

695. Pati, A., Nayak, N., Sarangi, S., Barik, D., Kumar Nahak, S., Padhi, S. *et al.* (2022) CCL5 rs2107538 Variant Is Associated With Protection Against SARS-CoV-2 Infection and Related Mortality: A Population-Based Study J

Infect Dis **225**, 1865-1866 10.1093/infdis/jiac026

696. Saiki, R., Katayama, K., Kitano, M., Tsujimoto, K., Tanaka, F., Suzuki, Y. *et al.* (2022) A Perihilar Variant of Focal Segmental Glomerulosclerosis Due to De novo Branchio-oto-renal Syndrome Intern Med **61**, 2033-2038 10.2169/internalmedicine.8508-21
697. Saito, M., Ogasawara, M., Inaba, Y., Osawa, Y., Nishioka, M., Yamauchi, S. *et al.* (2022) Successful treatment of congenital myasthenic syndrome caused by a novel compound heterozygous variant in RAPSN Brain Dev **44**, 50-55 10.1016/j.braindev.2021.09.001
698. Saito, Y., Nakamura, K., Fukuda, T., Sugie, H., Hayashi, S., Noguchi, S. *et al.* (2022) Muscle biochemical and pathological diagnosis in Pompe disease J Neurol Neurosurg Psychiatry 10.1136/jnnp-2022-329085
699. Sano, S., Masunaga, Y., Kato, F., Fujisawa, Y., Saitsu, H., andOgata, T. (2022) Combined pituitary hormone deficiency in a patient with an FGFR1 missense variant: case report and literature review Clin Pediatr Endocrinol **31**, 172-177 10.1297/cpe.2022-0020
700. Sano, Y., Ota, S., Oishi, M., Honda, M., Omoto, M., Kawai, M. *et al.* (2022) A Japanese Patient with Hereditary Myopathy with Early Respiratory Failure Due to the p.P31732L Mutation of Titin Intern Med **61**, 1587-1592 10.2169/internalmedicine.7733-21
701. Sato, H., Suga, K., Suzue, M., Honma, Y., Hayabuchi, Y., Miyai, S. *et al.* (2022) Novel large deletion involving EVC and EVC2 in Ellis-van Creveld syndrome Hum Genome Var **9**, 15 10.1038/s41439-022-00190-0
702. Sato, S., Morimoto, T., Fujikado, T., Tanaka, S., Tsujikawa, M., andNishida, K. (2022) Extensive Macular Atrophy with Pseudodrusen in a Japanese Patient Evaluated by Wide-Field OCTA Case Rep Ophthalmol **13**, 847-854 10.1159/000526970
703. Seyama, R., Tsuchida, N., Okada, Y., Sakata, S., Hamada, K., Azuma, Y. *et al.* (2022) Two families with TET3-related disorder showing neurodevelopmental delay with craniofacial dysmorphisms Journal of human genetics **67**, 157-164 10.1038/s10038-021-00986-y
704. Shibano, M., Kubota, T., Kokubun, N., Miyaji, Y., Kuriki, H., Ito, Y. *et al.* (2022) Periodic paralysis due to cumulative effects of rare variants in SCN4A with small functional alterations Muscle & Nerve **66**, 757-761 10.1002/mus.27725
705. Shibuya, M., Yaoita, H., Kodama, K., Okubo, Y., Endo, W., Inui, T. *et al.* (2022) A patient with early-onset SMAX3 and a novel variant of ATP7A Brain Dev **44**, 63-67 10.1016/j.braindev.2021.08.004
706. Shigemizu, D., Asanomi, Y., Akiyama, S., Higaki, S., Sakurai, T., Ito, K. *et al.* (2022) Network-based meta-analysis and the candidate gene association studies reveal novel ethnicity-specific variants in MFSD3 and MRPL43 associated with dementia with Lewy bodies American journal of medical genetics Part B, Neuropsychiatric genetics : the official publication of the International Society of Psychiatric Genetics **189**, 139-150 10.1002/ajmg.b.32908
707. Shigemizu, D., Asanomi, Y., Akiyama, S., Mitsumori, R., Niida, S., andOzaki, K. (2022) Whole-genome sequencing reveals novel ethnicity-specific rare variants associated with Alzheimer's disease Mol Psychiatry **27**, 2554-2562 10.1038/s41380-022-01483-0
708. Shimizu, M., Hirose, N., Kato, M., Sango, H., Uenuma, Y., Makiguchi, M. *et al.* (2022) Further survey of genetic variants of flavin-containing monooxygenase 3 (FMO3) in Japanese subjects found in an updated database of genome resources and identified by phenotyping for trimethylaminuria Drug Metab Pharmacokinet **46**, 100465 10.1016/j.dmpk.2022.100465
709. Shimura, M., Onuki, T., Sugiyama, Y., Matsuhashi, T., Ebihara, T., Fushimi, T. *et al.* (2022) Development of Leigh

syndrome with a high probability of cardiac manifestations in infantile-onset patients with m.14453G > A Mitochondrion **63**, 1-8 10.1016/j.mito.2021.12.005

710. Shirazi, P. G., Heidari, A., andFarshadmoghadam, H. (2022) Novel Missense Variation in NDUFA9 Gene in an Iranian Patient with Fatal Leigh Syndrome Iranian Journal of Pediatrics **32**, ARTN e115845 10.5812/ijp-115845
711. Sonehara, K., Kimura, Y., Nakano, Y., Ozawa, T., Takahashi, M., Suzuki, K. *et al.* (2022) A common deletion at BAK1 reduces enhancer activity and confers risk of intracranial germ cell tumors Nat Commun **13**, 4478 10.1038/s41467-022-32005-9
712. Sonehara, K., Sakaue, S., Maeda, Y., Hirata, J., Kishikawa, T., Yamamoto, K. *et al.* (2022) Genetic architecture of microRNA expression and its link to complex diseases in the Japanese population Human molecular genetics **31**, 1806-1820 10.1093/hmg/ddab361
713. Suga, A., Yoshitake, K., Minematsu, N., Tsunoda, K., Fujinami, K., Miyake, Y. *et al.* (2022) Genetic characterization of 1210 Japanese pedigrees with inherited retinal diseases by whole-exome sequencing Hum Mutat **43**, 2251-2264 10.1002/humu.24492
714. Sugisawa, C., Taniyama, M., Sato, T., Takahashi, Y., Hasegawa, T., andNarumi, S. (2022) Biallelic PROKR2 variants and congenital hypogonadotropic hypogonadism: a case report and a literature review Endocr J **69**, 831-838 10.1507/endocrj.EJ21-0779
715. Suh, S., Lee, S., Gym, H., Yoon, S., Park, S., Cha, J. *et al.* (2022) A systematic review on papers that study on Single Nucleotide Polymorphism that affects coronavirus 2019 severity BMC Infect Dis **22**, 47 10.1186/s12879-022-07034-w
716. Suzuki, H., Aoki, K., Kurosawa, K., Imagawa, K., Ohto, T., Yamada, M. *et al.* (2022) De novo non-synonymous CTR9 variants are associated with motor delay and macrocephaly: human genetic and zebrafish experimental evidence Human molecular genetics **31**, 3846-3854 10.1093/hmg/ddac136
717. Suzumura, M., Matsuwaki, Y., andTakeuchi, K. (2022) A Case of Chronic Rhinosinusitis Caused by Primary Ciliary Dyskinesia Associated with Mutations of the DNAH8 Gene Nippon Jibiinkoka Tokeibugeka Gakkai Kaiho(Tokyo) **125**, 1384-1389 10.3950/jibiinkotokeibu.125.9_1384
718. Tabata, T., Kuramoto, Y., Ohtani, T., Miyawaki, H., Miyashita, Y., Sera, F. *et al.* (2022) Phospholamban p.Arg14del Cardiomyopathy: A Japanese Case Series Intern Med **61**, 1987-1993 10.2169/internalmedicine.8594-21
719. Tachibana, N., Hosono, K., Nomura, S., Arai, S., Torii, K., Kurata, K. *et al.* (2022) Maternal Uniparental Isodisomy of Chromosome 4 and 8 in Patients with Retinal Dystrophy: SRD5A3-Congenital Disorders of Glycosylation and RP1-Related Retinitis Pigmentosa Genes (Basel) **13**, 10.3390/genes13020359
720. Takai, E., Nakamura, H., Chiku, S., Kubo, E., Ohmoto, A., Totoki, Y. *et al.* (2022) Whole-exome Sequencing Reveals New Potential Susceptibility Genes for Japanese Familial Pancreatic Cancer Ann Surg **275**, e652-e658 10.1097/SLA.0000000000004213
721. Takamatsu, G., Yanagi, K., Koganebuchi, K., Yoshida, F., Lee, J. S., Toyama, K. *et al.* (2022) Haplotype phasing of a bipolar disorder pedigree revealed rare multiple mutations of SPOCD1 gene in the 1p36-35 susceptibility locus J Affect Disord **310**, 96-105 10.1016/j.jad.2022.04.150
722. Tanase-Nakao, K., Muroya, K., Adachi, M., Abe, K., Hasegawa, T., andNarumi, S. (2022) A patient with congenital hypothyroidism due to a PAX8 frameshift variant accompanying a urogenital malformation Clinical Pediatric Endocrinology **31**, 250-255 10.1297/cpe.31.2022-0030
723. Tanida, K., Shimada, M., Khor, S. S., Toyoda, H., Kato, K., Kotorii, N. *et al.* (2022) Genome-wide association study of idiopathic hypersomnia in a Japanese population Sleep and Biological Rhythms **20**, 137-148

10.1007/s41105-021-00349-2

724. Terashima, T., Morizane, C., Ushiama, M., Shiba, S., Takahashi, H., Ikeda, M. *et al.* (2022) Germline variants in cancer-predisposing genes in pancreatic cancer patients with a family history of cancer Jpn J Clin Oncol **52**, 1105-1114 10.1093/jjco/hyac110

725. Teruyama, F., Kuno, A., Murata, Y., Nakagawa, T., Shiba-Ishii, A., Yuguchi, S. *et al.* (2022) Mutational landscape of primary breast angiosarcoma with repeated resection and recurrence over a 15-year period: A case report Pathol Int **72**, 457-463 10.1111/pin.13257

726. Tohnai, G., Nakamura, R., Atsuta, N., Nakatochi, M., Hayashi, N., Ito, D. *et al.* (2022) Mutation screening of the DNAJC7 gene in Japanese patients with sporadic amyotrophic lateral sclerosis Neurobiol Aging **113**, 131-136 10.1016/j.neurobiolaging.2021.12.002

727. Tomar, S., Klinzing, D. C., Chen, C. K., Gan, L. H., Moscarello, T., Reuter, C. *et al.* (2022) Causative Variants for Inherited Cardiac Conditions in a Southeast Asian Population Cohort Circ Genom Precis Med **15**, e003536 10.1161/CIRCGEN.121.003536

728. Tong, D., Tanaka, M., Eguchi, H., Okazaki, Y., Muramatsu, M., andArai, T. (2022) COL17A1 germline variant p.Ser1029Ala and mucosal malignant melanoma: An autopsy study Mol Clin Oncol **16**, 32 10.3892/mco.2021.2465

729. Toyama, M., Takasaki, Y., Branko, A., Kimura, H., Kato, H., Nawa, Y. *et al.* (2022) Exome sequencing of Japanese schizophrenia multiplex families supports the involvement of calcium ion channels PLoS One **17**, e0268321 10.1371/journal.pone.0268321

730. Tsuchida, Y., Nagafuchi, Y., Uehara, T., Suzuki, H., Yamada, M., Kono, M. *et al.* (2022) Rheumatoid arthritis in a patient with compound heterozygous variants in the COL11A2 gene and progressive hearing loss: A case report Medicine (Baltimore) **101**, e28828 10.1097/MD.0000000000028828

731. Tsujikawa, K., Hamanaka, K., Riku, Y., Hattori, Y., Hara, N., Iguchi, Y. *et al.* (2022) Actin-binding protein filamin-A drives tau aggregation and contributes to progressive supranuclear palsy pathology Sci Adv **8**, eabm5029 10.1126/sciadv.abm5029

732. Ueta, Y., Aso, K., Haga, Y., Takahashi, H., andSatoh, M. (2022) Congenital hypogonadotropic hypogonadism complicated by neuroblastoma Clin Pediatr Endocrinol **31**, 159-162 10.1297/cpe.2021-0070

733. Usami, S. I., Isaka, Y., Miyagawa, M., andNishio, S. Y. (2022) Variants in CDH23 cause a broad spectrum of hearing loss: from non-syndromic to syndromic hearing loss as well as from congenital to age-related hearing loss Hum Genet **141**, 903-914 10.1007/s00439-022-02431-2

734. Wang, C., Horigane, S. I., Wakamori, M., Ueda, S., Kawabata, T., Fujii, H. *et al.* (2022) Identification of ultra-rare disruptive variants in voltage-gated calcium channel-encoding genes in Japanese samples of schizophrenia and autism spectrum disorder Transl Psychiatry **12**, 84 10.1038/s41398-022-01851-y

735. Watanabe, H., Yoshida, C., Hidaka, M., Ogawa, T., Tomita, T., andFutai, E. (2022) Specific Mutations in Aph1 Cause gamma-Secretase Activation Int J Mol Sci **23**, 507 10.3390/ijms23010507

736. Watanabe, K., Nakashima, M., Wakatsuki, R., Bunai, T., Ouchi, Y., Nakamura, T. *et al.* (2022) Cognitive Impairment in a Complex Family With AAGGG and ACAGG Repeat Expansions in RFC1 Detected by ExpansionHunter Denovo Neurol Genet **8**, e682 10.1212/NXG.0000000000000682

737. Yachida, S., Totoki, Y., Noe, M., Nakatani, Y., Horie, M., Kawasaki, K. *et al.* (2022) Comprehensive Genomic Profiling of Neuroendocrine Carcinomas of the Gastrointestinal System Cancer Discov **12**, 692-711 10.1158/2159-8290.CD-21-0669

738. Yakabe, A., Ikuse, T., Ito, N., Yamada, H., Saito, N., Kitamura, Y. *et al.* (2022) A COL4A1 variant in a neonate with multiple intracranial hemorrhages and congenital cataracts Hum Genome Var **9**, 24 10.1038/s41439-022-00199-5

739. Yamada, M., Suzuki, H., Futagawa, H., Takenouchi, T., Miya, F., Yoshihashi, H. *et al.* (2022) Phenotypic overlap between cardioacrofacial dysplasia-2 and oral-facial-digital syndrome Eur J Med Genet **65**, 104512 10.1016/j.ejmg.2022.104512

740. Yamamoto, K., Ohashi, K., Fujimoto, M., Ieda, D., Nakamura, Y., Hattori, A. *et al.* (2022) Long-term follow-up of a patient with autosomal dominant lower extremity-predominant spinal muscular atrophy-2 due to a BICD2 variant Brain Dev **44**, 578-582 10.1016/j.braindev.2022.04.006

741. Yokoi, S., Ito, T., Sahashi, K., Nakatochi, M., Nakamura, R., Tohnai, G. *et al.* (2022) The SYNGAP1 3'UTR Variant in ALS Patients Causes Aberrant SYNGAP1 Splicing and Dendritic Spine Loss by Recruiting HNRNPK J Neurosci **42**, 8881-8896 10.1523/JNEUROSCI.0455-22.2022

742. Yoshida, N., Ishikawa, H., Eguchi, H., Okazaki, Y., Hirose, R., Inoue, K. *et al.* (2022) Promotion Effects of Smoking in Polyp Development in Monozygotic Twins with Atypical Colorectal Polyposis Case Rep Gastroenterol **16**, 375-381 10.1159/000524944

743. Yoshino, H., Li, Y., Nishioka, K., Daida, K., Hayashida, A., Ishiguro, Y. *et al.* (2022) Genotype-phenotype correlation of Parkinson's disease with PRKN variants Neurobiol Aging **114**, 117-128 10.1016/j.neurobiolaging.2021.12.014

744. Yuan, J. H., Higuchi, Y., Ando, M., Matsuura, E., Hashiguchi, A., Yoshimura, A. *et al.* (2022) Multi-type RFC1 repeat expansions as the most common cause of hereditary sensory and autonomic neuropathy Front Neurol **13**, 986504 10.3389/fneur.2022.986504

745. Yuan, J. H., Higuchi, Y., Hashiguchi, A., Ando, M., Yoshimura, A., Nakamura, T. *et al.* (2022) Genetic spectrum and founder effect of non-dystrophic myotonia: a Japanese case series study J Neurol **269**, 6406-6415 10.1007/s00415-022-11305-6

746. Zhang, X., Wang, L., Ma, Y., Wang, Y., Liu, H., Liu, M. *et al.* (2022) CEP128 is involved in spermatogenesis in humans and mice Nat Commun **13**, 1395 10.1038/s41467-022-29109-7

747. 山下朋代, 前川加奈美, 若原珠美, 小出竜雄, 田中裕子, 三輪谷隆史 *et al.* (2022) 典型的な頭部 MRI 所見を呈した L-2 ヒドロキシグルタル酸尿症の小児例 脳と発達 **54**, 126-131,

748. 別府祥平, 池中建介, 藪本大紀, 藤堂謙一, 橋口昭大, 髙嶋博 *et al.* (2022) Senataxin (SETX) 遺伝子バリアントを認めた孤発性 amyotrophic lateral sclerosis (ALS) の 1 例 臨床神経学 cn-001675,

749. Abe-Hatano, C., Yokoi, T., Ida, K., andKurosawa, K. (2023) Mosaicism of a Truncating Variant of CASK Causes Congenital Heart Disease and Neurodevelopmental Disorder Mol Syndromol **13**, 517-521 10.1159/000524375

750. Adachi, K., Ohyama, K., Tanaka, Y., Nakano, H., Sato, T., Murayama, N. *et al.* (2023) Plasma and Hepatic Exposures of Celecoxib and Diclofenac Prescribed Alone in Patients with Cytochrome P450 2C9*3 Modeled after Virtual Oral Administrations and Likely Associated with Adverse Drug Events Reported in a Japanese Database Biol Pharm Bull **46**, 856-863 10.1248/bpb.b23-00189

751. Adachi, K., Ohyama, K., Tanaka, Y., Sato, T., Murayama, N., Shimizu, M. *et al.* (2023) High hepatic and plasma exposures of atorvastatin in subjects harboring impaired cytochrome P450 3A4 *16 modeled after virtual administrations and possibly associated with statin intolerance found in the Japanese adverse drug event report database Drug Metab Pharmacokinet **49**, 100486 10.1016/j.dmpk.2022.100486

752. Aihara, Y., Shirota, M., Kikuchi, A., Katata, Y., Abe, Y., Niihori, T. *et al.* (2023) A novel variant in the

transmembrane 4 domain of ANO3 identified in a two-year-old girl with developmental delay and tremor Journal of human genetics **68**, 51-54 10.1038/s10038-022-01082-5

753. Akbari, M., Ebrahimi Tapeh, Z., Zaersabet, M., Rahimi, H., andGanji, M. (2023) Novel pyrroline-5-carboxylate reductase 2 (PYCR2) mutation in an Iranian patient with hypomyelinating leukodystrophy: findings of molecular and in silico investigations Egyptian Journal of Medical Human Genetics **24**, 12 10.1186/s43042-023-00393-2
754. Akiyama, Y., Sonehara, K., Maeda, D., Katoh, H., Naito, T., Yamamoto, K. *et al.* (2023) Genome-wide association study identifies risk loci within the major histocompatibility complex region for Hunner-type interstitial cystitis Cell Rep Med **4**, 101114 10.1016/j.xcrm.2023.101114
755. Ansai, O., Hayashi, R., Nakamura, A., Sasaki, J., Hasegawa, A., Deguchi, T. *et al.* (2023) Deep dermatophytosis caused by Trichophyton rubrum in an elderly patient with CARD9 deficiency: A case report and literature review J Dermatol 10.1111/1346-8138.16995
756. Aoki, S., Watanabe, K., Kato, M., Konishi, Y., Kubota, K., Kobayashi, E. *et al.* (2023) Two novel cases of biallelic SMPD4 variants with brain structural abnormalities Neurogenetics 10.1007/s10048-023-00737-5
757. Arai, A., Yoshimitsu, M., Otsuka, M., Ito, Y., Miyazono, T., Nakano, N. *et al.* (2023) Identification of putative noncanonical driver mutations in patients with essential thrombocythemia Eur J Haematol **110**, 639-647 10.1111/ejh.13945
758. Azuma, K., Horisawa, S., Mashimo, H., Fukuda, M., Kumada, S., Kawamata, T. *et al.* (2023) Loss-of-function mutations in SGCE found in Japanese patients with myoclonus-dystonia Clin Genet **103**, 209-213 10.1111/cge.14233
759. Bae, H., Shim, K. H., Yoo, J., Yang, Y. S., An, S. S. A., andKang, M. J. (2023) Double Mutations in a Patient with Early-Onset Alzheimer's Disease in Korea: An APP Val551Met and a PSEN2 His169Asn Int J Mol Sci **24**, 7446 10.3390/ijms24087446
760. Daida, K., Funayama, M., Billingsley, K. J., Malik, L., Miano-Burkhardt, A., Leonard, H. L. *et al.* (2023) Long-Read Sequencing Resolves a Complex Structural Variant in PRKN Parkinson's Disease Mov Disord **38**, 2249-2257 10.1002/mds.29610
761. Fujihara, C., Hafiyyah, O. A., andMurakami, S. (2023) Identification of disease-associate variants of aggressive periodontitis using genome-wide association studies Jpn Dent Sci Rev **59**, 357-364 10.1016/j.jdsr.2023.09.001
762. Fujita, A., Kato, M., Sugano, H., Iimura, Y., Suzuki, H., Tohyama, J. *et al.* (2023) An integrated genetic analysis of epileptogenic brain malformed lesions Acta Neuropathol Commun **11**, 33 10.1186/s40478-023-01532-x
763. Fujitani, H., Eguchi, H., Kochi, Y., Arai, T., Muramatsu, M., andOkazaki, Y. (2023) Rare germline variants in pancreatic cancer and multiple primary cancers: an autopsy study Eur J Cancer Prev **32**, 286-297 10.1097/CEJ.0000000000000787
764. Fukuoka, K., Nakazawa, A., Hirato, J., Nobusawa, S., Itabashi, T., Arakawa, Y. *et al.* (2023) An infantile pineal embryonal tumor showing pathological features of a CNS ganglioneuroblastoma and the methylation profiling of an atypical teratoid/rhabdoid tumor and SMARCA4 mutation J Neuropathol Exp Neurol **82**, 664-667 10.1093/jnen/nlad033
765. Furuhata-Yoshimura, M., Yamaguchi, T., Izu, Y., andKosho, T. (2023) Homozygous splice site variant affecting the first von Willebrand factor A domain of COL12A1 in a patient with myopathic Ehlers-Danlos syndrome Am J Med Genet A **191**, 2631-2639 10.1002/ajmg.a.63328
766. Furukawa, S., Kato, M., Nomura, T., Sumitomo, N., Yoneno, S., Nakashima, M. *et al.* (2023) Novel compound heterozygous ATP1A2 variants in a patient with fetal akinesia/hypokinesia sequence Am J Med Genet A

10.1002/ajmg.a.63453

767. Furukawa, S., Kushima, I., Aleksic, B., andOzaki, N. (2023) Case reports of two siblings with autism spectrum disorder and 15q13.3 deletions Neuropsychopharmacol Rep **43**, 462-466 10.1002/npr2.12340

768. Gao, J., Makiyama, T., Yamamoto, Y., Kobayashi, T., Aoki, H., Maurissen, T. L. *et al.* (2023) Novel Calmodulin Variant p.E46K Associated With Severe Catecholaminergic Polymorphic Ventricular Tachycardia Produces Robust Arrhythmogenicity in Human Induced Pluripotent Stem Cell-Derived Cardiomyocytes Circ Arrhythm Electrophysiol **16**, e011387 10.1161/CIRCEP.122.011387

769. Goto, K., Yoshikawa, S., Takai, T., Tachibana, K., Honma, K., Isei, T. *et al.* (2023) Clinicopathologic and genetic characterization of invasive melanoma with BRAF V600K mutation: A study of 16 cases J Cutan Pathol **50**, 739-747 10.1111/cup.14470

770. Haga, S., Takeguchi, R., Tanaka, R., Satake, A., Makita, Y., Yanagi, K. *et al.* (2023) Clinical characteristics of muscle cramps in hereditary angiopathy with nephropathy, aneurysms, and muscle cramps syndrome associated with a novel COL4A1 pathogenic variant: A family case study Brain Dev **45**, 390-394 10.1016/j.braindev.2023.02.008

771. Hanafusa, H., Yamaguchi, H., Kondo, H., Nagasaka, M., Juan Ye, M., Oikawa, S. *et al.* (2023) Dravet syndrome and hemorrhagic shock and encephalopathy syndrome associated with an intronic deletion of SCN1A Brain Dev **45**, 317-323 10.1016/j.braindev.2023.01.008

772. Hata, Y., Ichimata, S., Yoshida, K., Yamaguchi, Y., Hirono, K., andNishida, N. (2023) Comprehensive pathological and genetic investigation of three young adult myotonic dystrophy type 1 patients with sudden unexpected death J Neurol 1-12 10.1007/s00415-023-11850-8

773. Hattori, Y., Sawada, T., Kido, J., Sugawara, K., Yoshida, S., Matsumoto, S. *et al.* (2023) Frequency of iduronate-2-sulfatase gene variants detected in newborn screening for mucopolysaccharidosis type II in Japan Mol Genet Metab Rep **37**, 101003 10.1016/j.ymgmr.2023.101003

774. Hayashi, T., Mizobuchi, K., Kameya, S., Ueno, S., Matsuura, T., andNakano, T. (2023) A mild form of POC1B-associated retinal dystrophy with relatively preserved cone system function Doc Ophthalmol **147**, 59-70 10.1007/s10633-023-09936-9

775. Hida, A., Iida, A., Ukai, M., Kadotani, H., Uchiyama, M., Ebisawa, T. *et al.* (2023) Novel CLOCK and NR1D2 variants in 64 sighted Japanese individuals with non-24-hour sleep-wake rhythm disorder Sleep **46**, 10.1093/sleep/zsad063

776. Higashiyama, M., Motoi, N., Yotsukura, M., Yoshida, Y., Nakagawa, K., Yagishita, S. *et al.* (2023) Clinicopathological characteristics and molecular analysis of lung cancer associated with ciliated muconodular papillary tumor/bronchiolar adenoma Pathol Int **73**, 188-197 10.1111/pin.13316

777. Higuchi, T., Oka, S., Furukawa, H., Shimada, K., Tsunoda, S., Ito, S. *et al.* (2023) Association of a Single Nucleotide Variant in TERT with Airway Disease in Japanese Rheumatoid Arthritis Patients Genes (Basel) **14**, 10.3390/genes14112084

778. Higuchi, T., Oka, S., Furukawa, H., Shimada, K., Tsunoda, S., Ito, S. *et al.* (2023) Association of a FAM13A variant with interstitial lung disease in Japanese rheumatoid arthritis RMD Open **9**, e002828 10.1136/rmdopen-2022-002828

779. Hijikata, M., Morimoto, K., Takekoshi, D., Shimoda, M., Wakabayashi, K., Miyabayashi, A. *et al.* (2023) Analysis of Aberrant Splicing Events and Gene Expression Outliers in Primary Ciliary Dyskinesia Am J Respir Cell Mol Biol **68**, 702-705 10.1165/rcmb.2022-0394LE

780. Hiraide, T., Akita, T., Uematsu, K., Miyamoto, S., Nakashima, M., Sasaki, M. *et al.* (2023) A novel de novo KCNB1 variant altering channel characteristics in a patient with periventricular heterotopia, abnormal corpus callosum, and mild seizure outcome Journal of human genetics **68**, 25-31 10.1038/s10038-022-01090-5

781. Hiraide, T., Shimizu, K., Okumura, Y., Miyamoto, S., Nakashima, M., Ogata, T. *et al.* (2023) A deep intronic TCTN2 variant activating a cryptic exon predicted by SpliceRover in a patient with Joubert syndrome Journal of human genetics **68**, 499-505 10.1038/s10038-023-01143-3

782. Honda, T., Seto, K., Endo, S., Takemoto, A., Tanimoto, K., Kobayashi, M. *et al.* (2023) The possibility of mutations of RAS signaling genes and/or TP53 in combination as a negative prognostic impact on pathological stage I non-small cell lung cancer Cancer Med **12**, 19406-19413 10.1002/cam4.6535

783. Hori, M., Takahashi, A., Hosoda, K., Ogura, M., andHarada-Shiba, M. (2023) A Low-Frequency APOB p.(Pro955Ser) Variant Contributes to the Severity of/Variability in Familial Hypercholesterolemia J Clin Endocrinol Metab **108**, 422-432 10.1210/clinem/dgac572

784. Horii, Y., Kuroda, Y., Saito, Y., Enomoto, Y., Naruto, T., andKurosawa, K. (2023) A CAMK2B variant associated with tetralogy of Fallot, developmental delay, and growth retardation Eur J Med Genet **66**, 104845 10.1016/j.ejmg.2023.104845

785. Ikeda, A., Kumaki, T., Tsuyusaki, Y., Tsuji, M., Enomoto, Y., Fujita, A. *et al.* (2023) Genetic and clinical features of pediatric-onset hereditary spastic paraplegia: a single-center study in Japan Front Neurol **14**, 1085228 10.3389/fneur.2023.1085228

786. Ikeda, S., Hu, Q., Natsugoe, K., Harima, T., Tanaka, Y., Kinoshita, I. *et al.* (2023) A pathological complete response after immunotherapy with pembrolizumab for distal duodenal adenocarcinoma caused by Lynch syndrome: a case report Int Cancer Conf J **12**, 279-284 10.1007/s13691-023-00622-w

787. Inoue, M., Noguchi, S., Inoue, Y. U., Iida, A., Ogawa, M., Bengoechea, R. *et al.* (2023) Distinctive chaperonopathy in skeletal muscle associated with the dominant variant in DNAJB4 Acta Neuropathol **145**, 235-255 10.1007/s00401-022-02530-4

788. Isa, K., Takada, S., Takeda, H., Tsuneyama, H., Ogasawara, K., Takahashi, D. *et al.* (2023) Two new JK silencing alleles identified by single molecule sequencing with 20-Kb long-reads Transfusion **63**, 1441-1446 10.1111/trf.17397

789. Ishiguro, T., Konno, T., Hara, N., Zhu, B., Okada, S., Shibata, M. *et al.* (2023) Novel partial deletions, frameshift and missense mutations of CSF1R in patents with CSF1R-related leukoencephalopathy Eur J Neurol **30**, 1861-1870 10.1111/ene.15796

790. Ito, N., Kudo, T., Eguchi, H., Jimbo, K., Furuhata, A., Okuno, T. *et al.* (2023) Attenuated Expression of SLCO2A1 Caused by DNA Methylation in Pediatric Inflammatory Bowel Disease Inflamm Bowel Dis izad116 10.1093/ibd/izad116

791. Ito, S., Tsurumi, K., Shindo, N., Soma, S., Yamaguchi, K., Tamai, K. *et al.* (2023) Multi-gene Liquid Biopsy to Detect Resistance to First-line Osimertinib in Patients With EGFR-mutated Lung Adenocarcinoma Anticancer Res **43**, 5031-5040 10.21873/anticanres.16702

792. Ito, T., Takayanagi, D., Sekine, S., Hashimoto, T., Shimada, Y., Matsuda, M. *et al.* (2023) Comparison of clinicopathological and genomic profiles in anal squamous cell carcinoma between Japanese and Caucasian cohorts Sci Rep **13**, 3587 10.1038/s41598-023-30624-w

793. Iwasa, T., Urasaki, A., Kakihana, Y., Nagata-Akaho, N., Harada, Y., Takeda, S. *et al.* (2023) Computational and Experimental Analyses for Pathogenicity Prediction of ACVRL1 Missense Variants in Hereditary Hemorrhagic

Telangiectasia J Clin Med **12**, 5002 10.3390/jcm12155002

794. Kage, H., Shinozaki-Ushiku, A., Ishigaki, K., Sato, Y., Tanabe, M., Tanaka, S. *et al.* (2023) Clinical utility of Todai OncoPanel in the setting of approved comprehensive cancer genomic profiling tests in Japan Cancer Sci **114**, 1710-1717 10.1111/cas.15717
795. Kananazawa, Y., Yamada, T., Yamaguchi, T., Saito, Y., Kakinuma, D., Masuda, Y. *et al.* (2023) A novel germline SMAD4 variant detected in a Japanese family with juvenile polyposis syndrome and hereditary hemorrhagic telangiectasia Jpn J Clin Oncol **53**, 275-279 10.1093/jjco/hyac189
796. Karimi Azam, F., Sohrabi, B., Rahimi, H., andGanji, M. (2023) Trio whole-exome sequencing reveals a novel de novo mutation in COL2A1 gene in an Iranian patient with hypochondroplasia Gene Reports **31**, 101754 10.1016/j.genrep.2023.101754
797. Kashiwagi, Y., Suzuki, S., Takahashi, R., Yamanaka, G., Hirai, Y., andKawashima, H. (2023) Association of the Mannose-Binding Lectin 2 BB Genotype with COVID-19-Related Mortality Life (Basel) **13**, 382 10.3390/life13020382
798. Kato, H., Braddock, D. T., andIto, N. (2023) Genetics of Diffuse Idiopathic Skeletal Hyperostosis and Ossification of the Spinal Ligaments Curr Osteoporos Rep 1-15 10.1007/s11914-023-00814-6
799. Kawai, T., Iwasaki, Y., Ogata-Kawata, H., Kamura, H., Nakamura, K., Hata, K. *et al.* (2023) Identification of a KDM6A somatic mutation responsible for Kabuki syndrome by excluding a conflicting KMT2D germline variant through episignature analysis Eur J Med Genet **66**, 104806 10.1016/j.ejmg.2023.104806
800. Kawamoto, N., Hamada, Y., Kobayashi, S., Naruse, H., Ishiura, H., Matsukawa, T. *et al.* (2023) Noncanonical splice-site variant in peripheral myelin protein 22 gene (PMP22) in a patient with hereditary neuropathy with liability to pressure palsies Journal of the peripheral nervous system : JPNS **28**, 513-517 10.1111/jns.12558
801. Kawamura, H., Ikawa, M., Hirono, K., Kimura, J., Okuno, T., Kawatani, M. *et al.* (2023) Low-frequency maternal novel MYH7 mosaicism mutation in recurrent fetal-onset severe left ventricular noncompaction: a case report Front Pediatr **11**, 1195222 10.3389/fped.2023.1195222
802. Khatter, S., Lall, M., Agrawal, S., Mahay, S. B., Dimri, N., Sharma, N. *et al.* (2023) The Impact of Isolated Increased Nuchal Translucency ≥95th Centile on Perinatal Outcome: A Prospective Cohort Study from a North Indian Genetic Center Journal of Fetal Medicine **10**, 009-015 10.1055/s-0043-57001
803. Kido, J., Egami, K., Misumi, Y., Sugawara, K., Tsuchida, N., Matsumoto, N. *et al.* (2023) X-linked intellectual disability related to a novel variant of KLHL15 Hum Genome Var **10**, 21 10.1038/s41439-023-00248-7
804. Kishita, Y., Sugiura, A., Onuki, T., Ebihara, T., Matsuhashi, T., Shimura, M. *et al.* (2023) Strategic validation of variants of uncertain significance in ECHS1 genetic testing J Med Genet 10.1136/jmg-2022-109027
805. Kobayashi, Y., Tohyama, J., Akasaka, N., Yamada, K., Hojo, M., Seki, E. *et al.* (2023) The HCN1 p.Ser399Pro variant causes epileptic encephalopathy with super-refractory status epilepticus Hum Genome Var **10**, 20 10.1038/s41439-023-00247-8
806. Kodama, K., Aoyama, H., Murakami, Y., Takanashi, J. I., Koshimizu, E., Miyatake, S. *et al.* (2023) A case of early-infantile onset, rapidly progressive leukoencephalopathy with calcifications and cysts caused by biallelic SNORD118 variants Radiol Case Rep **18**, 1217-1220 10.1016/j.radcr.2022.11.033
807. Komura, K., Hirosuna, K., Tokushige, S., Tsujino, T., Nishimura, K., Ishida, M. *et al.* (2023) The Impact of FGFR3 Alterations on the Tumor Microenvironment and the Efficacy of Immune Checkpoint Inhibitors in Bladder Cancer Mol Cancer **22**, 185 10.1186/s12943-023-01897-6
808. Kudo, H., Suzuki, R., Kondo, A., Nozu, K., Nakamura, Y., Mikami, H. *et al.* (2023) Association of Familial Fanconi

Syndrome with a Novel GATM Variant Tohoku J Exp Med **260**, 337-340 10.1620/tjem.2023.J046

809. Kumagai, H., Kaneko, T., Shintake, Y., Miyamoto-Mikami, E., Tomita, H., Fukuo, M. *et al.* (2023) Genetic polymorphisms related to muscular strength and flexibility are associated with artistic gymnastic performance in the Japanese population Eur J Sport Sci **23**, 955-963 10.1080/17461391.2022.2078741
810. Kuroda, Y., Saito, Y., Enomoto, Y., Naruto, T., Mitsui, J., andKurosawa, K. (2023) PHACES-like syndrome with TMEM260 compound heterozygous variants Am J Med Genet A **191**, 2215-2218 10.1002/ajmg.a.63245
811. Li, D., Johmura, Y., Morimoto, S., Doi, M., Nakanishi, K., Ozawa, M. *et al.* (2023) LONRF2 is a protein quality control ubiquitin ligase whose deficiency causes late-onset neurological deficits Nat Aging **3**, 1001-1019 10.1038/s43587-023-00464-4
812. Makiguchi, M., Shimizu, M., Yokota, Y., Shimamura, E., Hishinuma, E., Saito, S. *et al.* (2023) Variants of Flavin-Containing Monooxygenase 3 Found in Subjects in an Updated Database of Genome Resources Drug Metab Dispos **51**, 884-891 10.1124/dmd.123.001310
813. Makishima, H., Saiki, R., Nannya, Y., Korotev, S., Gurnari, C., Takeda, J. *et al.* (2023) Germ line DDX41 mutations define a unique subtype of myeloid neoplasms Blood **141**, 534-549 10.1182/blood.2022018221
814. Masson, E., Zou, W. B., Pu, N., Rebours, V., Genin, E., Wu, H. *et al.* (2023) Classification of PRSS1 variants responsible for chronic pancreatitis: An expert perspective from the Franco-Chinese GREPAN study group Pancreatology **23**, 491-506 10.1016/j.pan.2023.04.004
815. Matsubara, H., Miyoshi, H., Kakizaki, F., Morimoto, T., Kawada, K., Yamamoto, T. *et al.* (2023) Efficacious Combination Drug Treatment for Colorectal Cancer That Overcomes Resistance to KRAS G12C Inhibitors Mol Cancer Ther **22**, 529-538 10.1158/1535-7163.MCT-22-0411
816. Matsumoto, M., Fujihara, C., Nantakeeratipat, T., Kitagaki, J., Yamamoto, Y., Yamada, S. *et al.* (2023) Lipase-a single-nucleotide polymorphism rs143793106 is associated with increased risk of aggressive periodontitis by negative influence on the cytodifferentiation of human periodontal ligament cells J Periodontal Res **58**, 175-183 10.1111/jre.13079
817. Miura, S., Hiruki, S., Okada, T., Takei, S. I., Senzaki, K., Okada, Y. *et al.* (2023) Case report: Frontotemporal dementia and amyotrophic lateral sclerosis caused by a missense variant (p.Arg89Trp) in the valosin-containing protein gene Front Genet **14**, 1155998 10.3389/fgene.2023.1155998
818. Miyabe, S., Ito, S., Sato, I., Abe, J., Tamai, K., Mochizuki, M. *et al.* (2023) Clinical and genomic features of non-small cell lung cancer occurring in families Thorac Cancer **14**, 940-952 10.1111/1759-7714.14825
819. Miyamoto, S., Nakamura, K., Kato, M., Nakashima, M., andSaitsu, H. (2023) Identification of pathogenic deep intronic variant and exonic LINE-1 insertion in a patient with Meckel syndrome Ann Hum Genet **87**, 196-202 10.1111/ahg.12507
820. Morikawa, H., Nishina, S., Torii, K., Hosono, K., Yokoi, T., Shigeyasu, C. *et al.* (2023) A pediatric case of congenital stromal corneal dystrophy caused by the novel variant c.953del of the DCN gene Hum Genome Var **10**, 9 10.1038/s41439-023-00239-8
821. Morohashi, T., Hayashi, T., Mizobuchi, K., Nakano, T., andMorioka, I. (2023) Bardet-Biedl syndrome associated with novel compound heterozygous variants in BBS12 gene Doc Ophthalmol **146**, 165-171 10.1007/s10633-022-09915-6
822. Mounai, Y., Yoshida, T., Ito, S., Fukuda, K., Shimazu, K., Taguchi, D. *et al.* (2023) Pulmonary Artery Intimal Sarcoma in a Patient with Lynch Syndrome: Response to an Immune Checkpoint Inhibitor Case Rep Oncol **16**, 21-29 10.1159/000528682

823. Muromachi, K., Hosomichi, K., Park, H., Yamaguchi, T., andTani-Ishii, N. (2023) Identification of Candidate Genes of Familial Multiple Idiopathic Cervical Root Resorption J Endod **49**, 1537-1547 10.1016/j.joen.2023.09.008
824. Nakamura, Y., Kato, N., Tatematsu, Y., Arai, Y., Mori, N., Shibata, K. *et al.* (2023) Clinical characteristics of anti-GBM disease with thrombotic microangiopathy: a case report and literature review CEN Case Rep 1-8 10.1007/s13730-023-00797-4
825. Nakano, S., Yamaji, T., Shiraishi, K., Hidaka, A., Shimazu, T., Kuchiba, A. *et al.* (2023) Smoking and risk of colorectal cancer according to KRAS and BRAF mutation status in a Japanese prospective Study Carcinogenesis **44**, 476-484 10.1093/carcin/bgad046
826. Nakashima, M., Argilli, E., Nakano, S., Sherr, E. H., Kato, M., andSaitsu, H. (2023) De novo CLCN3 variants affecting Gly327 cause severe neurodevelopmental syndrome with brain structural abnormalities Journal of human genetics **68**, 291-298 10.1038/s10038-022-01106-0
827. Natsumoto, B., Shoda, H., Nagafuchi, Y., Ota, M., Okumura, T., Horie, Y. *et al.* (2023) Functional evaluation of rare OASL variants by analysis of SLE patient-derived iPSCs J Autoimmun **139**, 103085 10.1016/j.jaut.2023.103085
828. Nawaz, S., Hussain, S., Bilal, M., Syed, N., Liaqat, K., Ullah, I. *et al.* (2023) A variant in sperm-specific glycolytic enzyme enolase 4 (ENO4) causes human male infertility J Gene Med e3583 10.1002/jgm.3583
829. Niida, Y., Kobayashi, A., Togi, S., andUra, H. (2023) Recessive dystrophic epidermolysis bullosa caused by a novel COL7A1 variant with isodisomy Hum Genome Var **10**, 29 10.1038/s41439-023-00257-6
830. Niida, Y., Togi, S., andUra, H. (2023) Streamlining Genetic Diagnosis With Long-Range Polymerase Chain Reaction (PCR)-Based Next-Generation Sequencing for Type I and Type II Collagenopathies Cureus **15**, e50482 10.7759/cureus.50482
831. Nishioka, M., Takayama, J., Sakai, N., Kazuno, A. A., Ishiwata, M., Ueda, J. *et al.* (2023) Deep exome sequencing identifies enrichment of deleterious mosaic variants in neurodevelopmental disorder genes and mitochondrial tRNA regions in bipolar disorder Mol Psychiatry 1-13 10.1038/s41380-023-02096-x
832. Noda, Y., Kido, J., Misumi, Y., Sugawara, K., Ohori, S., Fujita, A. *et al.* (2023) Heterozygous c.175C>T variant in PURA gene causes severe developmental delay Clin Case Rep **11**, e7779 10.1002/ccr3.7779
833. Obara, K., Abe, E., Mamiya, S., andToyoshima, I. (2023) Cerebellar Hypoperfusion in Two Patients with Cornelia de Lange Syndrome with Novel NIPBL Variants Mol Syndromol **14**, 51-58 10.1159/000525681
834. Obara, K., Abe, E., andToyoshima, I. (2023) Whole-Exome Sequencing Identified a Novel DYRK1A Variant in a Patient With Intellectual Developmental Disorder, Autosomal Dominant 7 Cureus **15**, e33379 10.7759/cureus.33379
835. Obata, Y., Takayama, K., Nishikubo, H., Tobimatsu, A., Matsuda, I., Uehara, Y. *et al.* (2023) Combined pituitary hormone deficiency harboring CHD7 gene missense mutation without CHARGE syndrome: a case report BMC Endocr Disord **23**, 118 10.1186/s12902-023-01373-8
836. Ogawa, A., Ohira, S., Kato, Y., Ikuta, T., Yanagida, S., Mi, X. *et al.* (2023) Activation of the urotensin-II receptor by remdesivir induces cardiomyocyte dysfunction Commun Biol **6**, 511 10.1038/s42003-023-04888-x
837. Ohno, K., Suzuki, S., Akamatsu, N., NIishimata, S., Yamazaki, T., Kashiwagi, Y. *et al.* (2023) Pathology analysis of tonsils and exome analysis of Japanese patients with periodic fever, aphthous stomatitis, pharyngitis, and adenitis syndrome J Tokyo Med Univ **81**, 159-166,
838. Oishi, N., Satou, A., Miyaoka, M., Kawashima, I., Segawa, T., Miyake, K. *et al.* (2023) Genetic and immunohistochemical profiling of NK/T-cell lymphomas reveals prognostically relevant BCOR-MYC association

Blood Adv **7**, 178-189 10.1182/bloodadvances.2022007541

839. Okubo, Y., Shibuya, M., Nakamura, H., Kawashima, A., Kodama, K., Endo, W. *et al.* (2023) Neonatal developmental and epileptic encephalopathy with movement disorders and arthrogryposis: A case report with a novel missense variant of SCN1A Brain Dev **45**, 505-511 10.1016/j.braindev.2023.06.009
840. Orimo, K., Kakumoto, T., Hara, R., Goto, R., Ishiura, H., Mitsui, J. *et al.* (2023) A Japanese family with idiopathic basal ganglia calcification carrying a novel XPR1 variant J Neurol Sci **451**, 120732 10.1016/j.jns.2023.120732
841. Peng, H., Li, Y., Yoshino, H., Shimizu, M., Nishioka, K., Funayama, M. *et al.* (2023) Analysis of LIN28A variants in patients with Parkinson's disease Journal of human genetics **68**, 329-331 10.1038/s10038-022-01109-x
842. Peter, V. G., Kaminska, K., Santos, C., Quinodoz, M., Cancellieri, F., Cisarova, K. *et al.* (2023) The first genetic landscape of inherited retinal dystrophies in Portuguese patients identifies recurrent homozygous mutations as a frequent cause of pathogenesis PNAS Nexus **2**, pgad043 10.1093/pnasnexus/pgad043
843. Rajeswaran, P. K., Kumar, P. N., Balagurunathan, M., andArumugam, J. (2023) Interference of Red Cell Parameters by an Endogenous Substance: An Interesting Case Report Journal of Laboratory Physicians 10.1055/s-0043-1772461
844. Saito-Koyama, R., Tamai, K., Yasuda, J., Okamura, Y., Yamazaki, Y., Inoue, C. *et al.* (2023) Morphometric analysis of nuclear shape irregularity as a novel predictor of programmed death-ligand 1 expression in lung squamous cell carcinoma Virchows Arch 1-12 10.1007/s00428-023-03548-z
845. Sakamoto, T., Ajiro, M., Watanabe, A., Matsushima, S., Ueda, K., andHagiwara, M. (2023) Application of the CDK9 inhibitor FIT-039 for the treatment of KSHV-associated malignancy BMC Cancer **23**, 71 10.1186/s12885-023-10540-y
846. Sakurai, M., Ishitsuka, K., Ito, R., Wilkinson, A. C., Kimura, T., Mizutani, E. *et al.* (2023) Chemically defined cytokine-free expansion of human haematopoietic stem cells Nature **615**, 127-133 10.1038/s41586-023-05739-9
847. Sanai, H., Nakamura, Y., Koike, Y., Murota, H., Kosho, T., andSase, M. (2023) The detailed obstetric course of the first Japanese patient with AEBP1-related Ehlers-Danlos syndrome (classical-like EDS, type 2) J Obstet Gynaecol Res **49**, 1043-1047 10.1111/jog.15541
848. Sasaki, T., Nishimoto, Y., Hirata, T., Abe, Y., Hirose, N., Takayama, M. *et al.* (2023) Status and physiological significance of circulating adiponectin in the very old and centenarians: an observational study Elife **12**, 10.7554/eLife.86309
849. Satake, T., Kondo, S., Tanabe, N., Mizuno, T., Katsuya, Y., Sato, J. *et al.* (2023) Pathogenic Germline Variants in BRCA1/2 and p53 Identified by Real-world Comprehensive Cancer Genome Profiling Tests in Asian Patients Cancer Res Commun **3**, 2302-2311 10.1158/2767-9764.CRC-23-0018
850. Sato-Akuhara, N., Trimmer, C., Keller, A., Niimura, Y., Shirasu, M., Mainland, J. D. *et al.* (2023) Genetic variation in the human olfactory receptor OR5AN1 associates with the perception of musks Chem Senses **48**, 10.1093/chemse/bjac037
851. Seyama, R., Nishikawa, M., Uchiyama, Y., Hamada, K., Yamamoto, Y., Takeda, M. *et al.* (2023) A missense variant at the RAC1-PAK1 binding site of RAC1 inactivates downstream signaling in VACTERL association Sci Rep **13**, 9789 10.1038/s41598-023-36381-0
852. Shimizu, M., Makiguchi, M., Hishinuma, E., Saito, S., Hiratsuka, M., andYamazaki, H. (2023) Rare but impaired flavin-containing monooxygenase 3 (FMO3) variants reported in a recently updated Japanese mega-databank of genome resources Drug Metab Pharmacokinet **55**, 100539 10.1016/j.dmpk.2023.100539

853. Shimizu, M., Makiguchi, M., Yokota, Y., Shimamura, E., Matsuta, M., Nakamura, Y. *et al.* (2023) Simple confirmation methods for rare but impaired variants of human flavin-containing monooxygenase 3 (FMO3) found in an updated genome resource databank Drug Metab Pharmacokinet **53**, 100528 10.1016/j.dmpk.2023.100528

854. Shimomura, A., Yoshida, M., Kubo, T., Yamashita, S., Noguchi, E., Nagayama, A. *et al.* (2023) Clinicopathological features, genetic alterations, and BRCA1 promoter methylation in Japanese male patients with breast cancer Breast Cancer Res Treat **197**, 593-602 10.1007/s10549-022-06822-x

855. Shirasawa, M., Yoshida, T., Shiraishi, K., Goto, N., Yagishita, S., Imabayashi, T. *et al.* (2023) Tumor microenvironment-mediated immune profiles and efficacy of anti-PD-L1 antibody plus chemotherapy stratified by DLL3 expression in small-cell lung cancer Br J Cancer **129**, 2003-2013 10.1038/s41416-023-02427-3

856. Shirasawa, M., Yoshida, T., Shiraishi, K., Takigami, A., Takayanagi, D., Imabayashi, T. *et al.* (2023) Identification of inflamed-phenotype of small cell lung cancer leading to the efficacy of anti-PD-L1 antibody and chemotherapy Lung Cancer **179**, 107183 10.1016/j.lungcan.2023.107183

857. Shirota, H., Komine, K., Takahashi, M., Takahashi, S., Miyauchi, E., Niizuma, H. *et al.* (2023) Clinical decisions by the molecular tumor board on comprehensive genomic profiling tests in Japan: A retrospective observational study Cancer Med **12**, 6170-6181 10.1002/cam4.5349

858. Soma, K., Kitagawa, Y., Toki, T., Miura, F., Shimada, J., Sato, T. *et al.* (2023) Early-onset Marfan syndrome with a novel missense mutation: A case report J Cardiol Cases **27**, 283-286 10.1016/j.jccase.2023.02.019

859. Sone, J., Ueno, S., Akagi, A., Miyahara, H., Tamai, C., Riku, Y. *et al.* (2023) NOTCH2NLC GGC repeat expansion causes retinal pathology with intranuclear inclusions throughout the retina and causes visual impairment Acta Neuropathol Commun **11**, 71 10.1186/s40478-023-01564-3

860. Sugawara, Y., Hirakawa, Y., Nagasu, H., Narita, A., Katayama, A., Wada, J. *et al.* (2023) Genome-wide association study of the risk of chronic kidney disease and kidney-related traits in the Japanese population: J-Kidney-Biobank Journal of human genetics **68**, 55-64 10.1038/s10038-022-01094-1

861. Sugawara, Y., Kato, H., Nagasaki, M., Yoshida, Y., Fujisawa, M., Minegishi, N. *et al.* (2023) CFH-CFHR1 hybrid genes in two cases of atypical hemolytic uremic syndrome Journal of human genetics **68**, 427-430 10.1038/s10038-023-01129-1

862. Suzuki, R., Takahashi, H., Yoshida, C., Hidaka, M., Ogawa, T., andFutai, E. (2023) Specific Mutations near the Amyloid Precursor Protein Cleavage Site Increase gamma-Secretase Sensitivity and Modulate Amyloid-beta Production Int J Mol Sci **24**, 3970 10.3390/ijms24043970

863. Tajima, R., Okazaki, A., Sato, T., Ozaki, K., Motooka, D., Okazaki, Y. *et al.* (2023) Genetic Landscape of Masticatory Muscle Tendon-Aponeurosis Hyperplasia Genes (Basel) **14**, 10.3390/genes14091718

864. Takagi, M., Ono, S., Kumaki, T., Nishimura, N., Murakami, H., Enomoto, Y. *et al.* (2023) Complex congenital cardiovascular anomaly in a patient with AGO1-associated disorder Am J Med Genet A **191**, 882-892 10.1002/ajmg.a.63089

865. Takahashi, N., Mishima, T., Fujioka, S., Izumi, K., Ando, M., Higuchi, Y. *et al.* (2023) Siblings with Cockayne Syndrome B Type III Presenting with Slowly Progressive Cerebellar Ataxia Intern Med **62**, 2253-2259 10.2169/internalmedicine.0061-22

866. Takeuchi, I., Yanagi, K., Takada, S., Uchiyama, T., Igarashi, A., Motomura, K. *et al.* (2023) STAT6 gain-of-function variant exacerbates multiple allergic symptoms J Allergy Clin Immunol **151**, 1402-1409 e1406 10.1016/j.jaci.2022.12.802

867. Takeyari, S., Yamamoto, K., Fujiwara, M., Ohata, Y., Kitaoka, T., Kubota, T. *et al.* (2023) A family with brachydactyly mental retardation syndrome with a missense variant in HDAC4 Clin Pediatr Endocrinol **32**, 105-109 10.1297/cpe.2022-0076

868. Tamaoka, S., Fukuda, A., Nakabayashi, K., Matsubara, K., Ogata-Kawata, H., Muranishi, Y. *et al.* (2023) Rare sequence variants associated with the risk of non-syndromic biliary atresia Hepatol Res **53**, 1134-1141 10.1111/hepr.13946

869. Tamaoka, S., Saito, K., Yoshida, T., Nakabayashi, K., Tatsumi, K., Kawamura, T. *et al.* (2023) Exome-based genome-wide screening of rare variants associated with the risk of polycystic ovary syndrome Reprod Med Biol **22**, e12504 10.1002/rmb2.12504

870. Taura, Y., Tozawa, T., Isoda, K., Hirai, S., Chiyonobu, T., Yano, N. *et al.* (2023) Leigh-like syndrome with progressive cerebellar atrophy caused by novel HIBCH variants Hum Genome Var **10**, 23 10.1038/s41439-023-00251-y

871. Tomioka, Y., Sugimoto, S., Yamamoto, H., Tomida, S., Shiotani, T., Tanaka, S. *et al.* (2023) Identification of genetic loci associated with renal dysfunction after lung transplantation using an ethnic-specific single-nucleotide polymorphism array Sci Rep **13**, 8912 10.1038/s41598-023-36143-y

872. Tomofuji, Y., Kishikawa, T., Sonehara, K., Maeda, Y., Ogawa, K., Kawabata, S. *et al.* (2023) Analysis of gut microbiome, host genetics, and plasma metabolites reveals gut microbiome-host interactions in the Japanese population Cell Rep **42**, 113324 10.1016/j.celrep.2023.113324

873. Tomofuji, Y., Sonehara, K., Kishikawa, T., Maeda, Y., Ogawa, K., Kawabata, S. *et al.* (2023) Reconstruction of the personal information from human genome reads in gut metagenome sequencing data Nat Microbiol **8**, 1079-1094 10.1038/s41564-023-01381-3

874. Torii, K., Nishina, S., Morikawa, H., Mizobuchi, K., Takayama, M., Tachibana, N. *et al.* (2023) The Structural Abnormalities Are Deeply Involved in the Cause of RPGRIP1-Related Retinal Dystrophy in Japanese Patients Int J Mol Sci **24**, 13678 10.3390/ijms241813678

875. Totoki, Y., Saito-Adachi, M., Shiraishi, Y., Komura, D., Nakamura, H., Suzuki, A. *et al.* (2023) Multiancestry genomic and transcriptomic analysis of gastric cancer Nature genetics **55**, 581-594 10.1038/s41588-023-01333-x

876. Tsuneura, Y., Kawai, T., Yamada, K., Aoki, S., Nakashima, M., Eda, S. *et al.* (2023) A Novel Constitutively Active c.98G > C, p.(R33P) Variant in RAB11A Associated with Intellectual Disability Promotes Neuritogenesis and Affects Oligodendroglial Arborization Human Mutation **2023**, 8126544 Artn 8126544 10.1155/2023/8126544

877. Turan, B., Göktas, E., Sönmez, F. M., Aydin, H. I., Aydogdu, D., Zamani, A. G. *et al.* (2023) Cerebral creatine deficiency syndrome with a novel missense variant in gene Neurology and Clinical Neuroscience **11**, 286-288 10.1111/ncn3.12726

878. Uchida, N., Mizuno, Y., Seno, S., Koyama, Y., Takahashi, T., Shibata, H. *et al.* (2023) A novel missense variant of FGFR1 in a Japanese girl with Kallmann syndrome and holoprosencephaly Clin Pediatr Endocrinol **32**, 79-81 10.1297/cpe.2022-0060

879. Ueda, H., Tran, Q. T. H., Tran, L. N. T., Higasa, K., Ikeda, Y., Kondo, N. *et al.* (2023) Characterization of cytoskeletal and structural effects of INF2 variants causing glomerulopathy and neuropathy Sci Rep **13**, 12003 10.1038/s41598-023-38588-7

880. Uehara, D. T., Muramatsu, T., Ishii, S., Suzuki, H., Fukushima, K., Arasaki, Y. *et al.* (2023) Identification of a Biallelic Missense Variant in Gasdermin D (c.823G > C, p.Asp275His) in a Patient of Atypical Gorham-Stout

Disease in a Consanguineous Family Jbmr Plus e10784 10.1002/jbm4.10784

881. Utsunomiya, H., Akazawa, Y., Ueyama, H., Iwano, T., Yamamoto, M., Uchida, R. *et al.* (2023) Gastric Juvenile Polyposis with Intramucosal Cancer Diagnosed by Magnifying Endoscopy with Narrow-band Imaging Intern Med **62**, 3333-3339 10.2169/internalmedicine.1612-23

882. Wang, X., Hao, X. J., Dai, C. G., Ding, Y. J., Xiong, L., Deng, J. *et al.* (2023) Identification of 8 Rare Deleterious Variants in ADAMTS13 by Next-generation Sequencing in a Chinese Population with Thrombotic Thrombocytopenic Purpura Curr Med Sci **43**, 1043-1050 10.1007/s11596-023-2793-7

883. Welch, C. L., Aldred, M. A., Balachandar, S., Dooijes, D., Eichstaedt, C. A., Graf, S. *et al.* (2023) Defining the clinical validity of genes reported to cause pulmonary arterial hypertension Genet Med **25**, 100925 10.1016/j.gim.2023.100925

884. Xiao, S., Kai, Z., Murphy, D., Li, D., Patel, D., Bielowka, A. M. *et al.* (2023) Functional filter for whole-genome sequencing data identifies HHT and stress-associated non-coding SMAD4 polyadenylation site variants >5 kb from coding DNA Am J Hum Genet **110**, 1903-1918 10.1016/j.ajhg.2023.09.005

885. Xu, Y., Feng, G., Yano, T., Masuda, S., Nagao, M., Gotoh, S. *et al.* (2023) Characteristic genetic spectrum of primary ciliary dyskinesia in Japanese patients and global ethnic heterogeneity: population-based genomic variation database analysis Journal of human genetics **68**, 455-461 10.1038/s10038-023-01142-4

886. Yagasaki, H., Narusawa, H., Watanabe, D., Kobayashi, K., Mitsui, H., Asano, Y. *et al.* (2023) Novel TRPS1 frameshift variant in tricho-rhino-phalangeal syndrome type I accompanied by zinc deficiency Eur J Med Genet **66**, 104870 10.1016/j.ejmg.2023.104870

887. Yamada, E., Ishikawa, E., Miyazaki, T., Miki, S., Sugii, N., Kohzuki, H. *et al.* (2023) P53-negative status and gross total resection as predictive factors for autologous tumor vaccine treatment in newly diagnosed glioblastoma patients Neurooncol Adv **5**, vdad079 10.1093/noajnl/vdad079

888. Yamada, S., Ko, T., Ito, M., Sassa, T., Nomura, S., Okuma, H. *et al.* (2023) TEAD1 trapping by the Q353R-Lamin A/C causes dilated cardiomyopathy Sci Adv **9**, eade7047 10.1126/sciadv.ade7047

889. Yamaguchi, T., Hayashi, S., Hayashi, D., Matsuyama, T., Koitabashi, N., Ogiwara, K. *et al.* (2023) Comprehensive genetic screening for vascular Ehlers-Danlos syndrome through an amplification-based next-generation sequencing system Am J Med Genet A **191**, 37-51 10.1002/ajmg.a.62982

890. Yamaguchi, T., Hayashi, S., Nagai, S., Uchiyama, A., Motegi, S. I., Fujikawa, T. *et al.* (2023) Case report: further delineation of AEBP1-related Ehlers-Danlos Syndrome (classical-like EDS type 2) in an additional patient and comprehensive clinical and molecular review of the literature Front Genet **14**, 1102101 10.3389/fgene.2023.1102101

891. Yamaki, M., Saito, H., Mimori, T., Suzuki, Y., Nagasaki, M., Suzuki, K. *et al.* (2023) Analysis of Genetic Polymorphism of Bitter Taste Receptor TAS2R38 and TAS2R46, and Its Relationship with Eating and Drinking Habits in Japanese ToMMo Subjects J Nutr Sci Vitaminol (Tokyo) **69**, 347-356 10.3177/jnsv.69.347

892. Yamazawa, K., Sugano, K., Tanakaya, K., Inoue, S., Murakami, H., Nakashima, M. *et al.* (2023) The pathogenic role of the BRCA2 c.7847C>T (p.Ser2616Phe) variant in breast and ovarian cancer predisposition Cancer Sci **114**, 2993-3002 10.1111/cas.15799

893. Yanagita, E., Yamada, H., Kobayashi, T., Aimono, E., Nakamura, K., Hirasawa, A. *et al.* (2023) The DNA integrity number and concentration are useful parameters for successful comprehensive genomic profiling test for cancer using formalin-fixed paraffin embedded tissue Pathol Int **73**, 198-206 10.1111/pin.13318

894. Yorifuji, T., Watanabe, Y., Kitayama, K., Yamada, Y., Higuchi, S., Mori, J. *et al.* (2023) Targeted gene panel

analysis of Japanese patients with maturity-onset diabetes of the young-like diabetes mellitus: Roles of inactivating variants in the ABCC8 and insulin resistance genes J Diabetes Investig **14**, 387-403 10.1111/jdi.13957

895. Yoshida, A., Aoyama, K., Yamaguchi, N., Suzuki, A., Mizuno, H., Tada, H. *et al.* (2023) An infant with a heterozygous variant of ABCG5 presented with hypercholesterolemia only during breastfeeding Clin Pediatr Endocrinol **32**, 114-118 10.1297/cpe.2022-0075
896. Yoshida, T., Matsubara, K., Ogata-Kawata, H., Miyado, M., Ishiwata, K., Nakabayashi, K. *et al.* (2023) Variations in gender identity and sexual orientation of university students Sex Med **11**, qfad057 10.1093/sexmed/qfad057
897. Yoshida-Tanaka, K., Shimada, M., Honda, Y., Fujimoto, A., Tokunaga, K., Honda, M. *et al.* (2023) Narcolepsy type I-associated DNA methylation and gene expression changes in the human leukocyte antigen region Sci Rep **13**, 10464 10.1038/s41598-023-37511-4
898. Yoshihama, K., Mutai, H., Sekimizu, M., Ito, F., Saito, S., Nakamura, S. *et al.* (2023) Molecular basis of carotid body tumor and associated clinical features in Japan identified by genomic, immunohistochemical, and clinical analyses Clin Genet **103**, 466-471 10.1111/cge.14294
899. Yoshihara, K., Baba, T., Tokunaga, H., Nishino, K., Sekine, M., Takamatsu, S. *et al.* (2023) Homologous recombination inquiry through ovarian malignancy investigations: JGOG3025 Study Cancer Sci **114**, 2515-2523 10.1111/cas.15747
900. Yuan, J. H., Cheng, X., Matsuura, E., Higuchi, Y., Ando, M., Hashiguchi, A. *et al.* (2023) Genetic, electrophysiological, and pathological studies on patients with SCN9A-related pain disorders Journal of the peripheral nervous system : JPNS **28**, 597-607 10.1111/jns.12590
901. Yuan, J. H., Higuchi, Y., Hashiguchi, A., Ando, M., Yoshimura, A., Nakamura, T. *et al.* (2023) Gene panel analysis of 119 index patients with suspected periodic paralysis in Japan Front Neurol **14**, 1078195 10.3389/fneur.2023.1078195
902. Zheng, C., Wang, F., Sun, Y., Zhou, Z., You, Y., He, D. *et al.* (2023) Identification of Distinct Genetic Profiles of Palindromic Rheumatism Using Whole-Exome Sequencing Arthritis & rheumatology (Hoboken, NJ) **75**, 1947-1957 10.1002/art.42614
903. 伊藤あかね, 平出拓也, 古澤有花子, 松本由里, and 田冬季子 (2023) c. 116G> A, p.(Arg39His) ホモ接合性バリアントが同定された一過性眼振を伴う遊離シアル酸蓄積症の姉弟例 浜松医科大学小児科学雑誌 **1**, 36-44,
904. 勝部暢介, 佐治重衡, 岡野舞子, 石野淳, 高橋昌一, 後藤政広 *et al.* (2023) 血管肉腫の同胞発症がみられた 1 家系 遺伝性腫瘍 **22**, 106-111,
905. 名嘉山一郎 (2023) BRCA2 遺伝子に同一病的バリアントを認めた男性乳癌の 2 例 遺伝性腫瘍 **22**, 97-100,
906. Amano, N., Narumi, S., Aizu, K., Miyazawa, M., Okamura, K., Ohashi, H. *et al.* (2024) Single-Exon Deletions of ZNRF3 Exon 2 Cause Congenital Adrenal Hypoplasia J Clin Endocrinol Metab **109**, 641-648 10.1210/clinem/dgad627
907. Arai, Y., Okanishi, T., Okazaki, T., Awano, H., Seyama, R., Uchiyama, Y. *et al.* (2024) An adolescent case of ASXL3-related disorder with delayed onset of feeding difficulty BMC Pediatr **24**, 308 10.1186/s12887-024-04774-3
908. Araki, Y., Rajkovich, K. E., Gerber, E. E., Gamache, T. R., Johnson, R. C., Tran, T. H. N. *et al.* (2024) SynGAP regulates synaptic plasticity and cognition independently of its catalytic activity Science **383**, eadk1291 10.1126/science.adk1291
909. Barrios-Navas, A., Nguyen, T. L., Gallo, J. E., Mariño-Ramírez, L., Soto, J. M. S., Sánchez, A. *et al.* (2024)

Unveiling ancestral threads: Exploring CCR5 Δ32 mutation frequencies in Colombian populations for HIV/AIDS therapeutics Infect Genet Evol **125**, 105680 10.1016/j.meegid.2024.105680

910. Beryozkin, A., Sher, I., Ehrenberg, M., Zur, D., Newman, H., Gradstein, L. *et al.* (2024) Best Disease: Global Mutations Review, Genotype-Phenotype Correlation, and Prevalence Analysis in the Israeli Population Invest Ophthalmol Vis Sci **65**, 39 10.1167/iovs.65.2.39
911. Bosselmann, C. M., Leu, C., Brunger, T., Hoffmann, L., Baldassari, S., Chipaux, M. *et al.* (2024) Analysis of 1386 epileptogenic brain lesions reveals association with DYRK1A and EGFR Nat Commun **15**, 10429 10.1038/s41467-024-54911-w
912. Chen, J., Wang, M., Duan, S., Yang, Q., Liu, Y., Zhao, M. *et al.* (2024) Genetic history and biological adaptive landscape of the Tujia people inferred from shared haplotypes and alleles Hum Genomics **18**, 104 10.1186/s40246-024-00672-y
913. Doi, H., Kageyama, I., Katoh-Fukui, Y., Hattori, A., Fukami, M., andShimura, N. (2024) Homozygous 6-bp deletion of IGFALS in a prepubertal boy with short stature Hum Genome Var **11**, 27 10.1038/s41439-024-00285-w
914. Enomoto, Y., Naruto, T., Mitsui, J., Ueda, H., andKurosawa, K. (2024) A TMEM260 Biallelic Deletion Underlies Truncus Arteriosus Molecular Syndromology 10.1159/000543316
915. Fan, J., He, H. Y., Li, H. H., Wu, P. L., Tang, L., Deng, B. Y. *et al.* (2024) Associations between UGT1A1, SLCO1B1, SLCO1B3, BLVRA and HMOX1 polymorphisms and susceptibility to neonatal severe hyperbilirubinemia in Chinese Han population BMC Pediatr **24**, 82 10.1186/s12887-024-04537-0
916. Feng, X., Liu, S., Li, K., Bu, F., andYuan, H. (2024) NCAD v1.0: a database for non-coding variant annotation and interpretation J Genet Genomics **51**, 230-242 10.1016/j.jgg.2023.12.005
917. Fujii, E., Kato, M. K., Yamaguchi, M., Higuchi, D., Koyama, T., Komatsu, M. *et al.* (2024) Genomic profiles of Japanese patients with vulvar squamous cell carcinoma Sci Rep **14**, 13058 10.1038/s41598-024-63913-z
918. Fujimaru, T., Mori, T., Chiga, M., Mandai, S., Kikuchi, H., Ando, F. *et al.* (2024) Genetic Diagnosis of Adult Hemodialysis Patients With Unknown Etiology Kidney Int Rep **9**, 994-1004 10.1016/j.ekir.2024.01.027
919. Fujimaru, T., Mori, T., Sekine, A., Chiga, M., Mandai, S., Kikuchi, H. *et al.* (2024) Importance of IFT140 in Patients with Polycystic Kidney Disease Without a Family History Kidney Int Rep **9**, 2685-2694 10.1016/j.ekir.2024.06.021
920. Fujinami, K., Nishiguchi, K. M., Oishi, A., Akiyama, M., Ikeda, Y., andResearch Group on Rare, I. D. (2024) Specification of variant interpretation guidelines for inherited retinal dystrophy in Japan Jpn J Ophthalmol **68**, 389-399 10.1007/s10384-024-01063-5
921. Fujita, Y., Ogawa, S., Sumichika, Y., Saito, K., Yoshida, S., Matsumoto, H. *et al.* (2024) Elderly-onset familial Mediterranean Fever Carrying MEFV Exon 10 Variants in a Japanese Patient: A Case Report Intern Med 10.2169/internalmedicine.4463-24
922. Fussiger, H., Lima, P., Souza, P. V. S., Freua, F., Husny, A. S. E., Leão, E. *et al.* (2024) Clinicogenetic characterization of cerebrotendinous xanthomatosis in Brazil Clin Genet **106**, 721-732 10.1111/cge.14602
923. Gan, P., Hajis, M. I. B., Yumna, M., Haruman, J., Matoha, H. K., Wahyudi, D. T. *et al.* (2024) Development and validation of a pharmacogenomics reporting workflow based on the illumina global screening array chip Front Pharmacol **15**, 1349203 10.3389/fphar.2024.1349203
924. Goto, K., Koyanagi, Y., Akiyama, M., Murakami, Y., Fukushima, M., Fujiwara, K. *et al.* (2024) Disease-specific variant interpretation highlighted the genetic findings in 2325 Japanese patients with retinitis pigmentosa and

allied diseases J Med Genet **61**, 613-620 10.1136/jmg-2023-109750

925. Guo, F., Liu, Z., Long, G., Zhang, B., Liu, D., andYu, S. (2024) Performance and characterization of 94 identity-informative SNPs in Northern Han Chinese using ForenSeq ™ DNA signature prep kit J Forensic Leg Med **103**, 102678 10.1016/j.jflm.2024.102678
926. Guo, M., Lin, J., Cao, X., Zhou, J., Ben, S., Chen, S. *et al.* (2024) Genetic variants in hypoxia-inducible factor pathway are associated with colorectal cancer risk and immune infiltration J Cell Mol Med **28**, e18019 10.1111/jcmm.18019
927. Hanafusa, H., Yamaguchi, H., Morisada, N., Ye, M. J., Matsumoto, R., Nagase, H. *et al.* (2024) End-stage ADPKD with a low-frequency PKD1 mosaic variant accelerated by chemoradiotherapy Hum Genome Var **11**, 17 10.1038/s41439-024-00273-0
928. Hao, L., Liang, L., Gao, X., Zhan, X., Ji, W., Chen, T. *et al.* (2024) Screening of 1.17 million newborns for inborn errors of metabolism using tandem mass spectrometry in Shanghai, China: A 19-year report Mol Genet Metab **141**, 108098 10.1016/j.ymgme.2023.108098
929. Harada, S., Iida, M., Miyagawa, N., Hirata, A., Kuwabara, K., Matsumoto, M. *et al.* (2024) Study Profile of the Tsuruoka Metabolomics Cohort Study (TMCS) J Epidemiol **34**, 393-401 10.2188/jea.JE20230192
930. Harino, T., Tanaka, K., Motooka, D., Masuike, Y., Takahashi, T., Yamashita, K. *et al.* (2024) D-loop mutations in mitochondrial DNA are a risk factor for chemotherapy resistance in esophageal cancer Sci Rep **14**, 31653 10.1038/s41598-024-80226-3
931. He, J., Kou, S. H., Li, J., Ding, X., andWang, S. M. (2024) Pathogenic variants in human DNA damage repair genes mostly arose after the latest human out-of-Africa migration Front Genet **15**, 1408952 10.3389/fgene.2024.1408952
932. Higa, N., Hayashi, T., Mizobuchi, K., Iwasa, M., Kubota, S., Kuniyoshi, K. *et al.* (2024) A novel RPE65 variant p.(Ala391Asp) in Leber congenital amaurosis: a case report and literature review in Japan Front Med (Lausanne) **11**, 1442107 10.3389/fmed.2024.1442107
933. Higuchi, T., Oka, S., Furukawa, H., andTohma, S. (2024) The contributions of deleterious rare alleles in NLRP12 and inflammasome-related genes to polymyalgia rheumatica Sci Rep **14**, 490 10.1038/s41598-024-51320-3
934. Higuchi, T., Oka, S., Shimada, K., Tsunoda, S., Ito, S., Okamoto, A. *et al.* (2024) Association of rare single nucleotide variant MUC5B rs35705950 with interstitial lung disease in Japanese rheumatoid arthritis Rheumatology (Oxford) 10.1093/rheumatology/keae422
935. Hijikata, M., Morimoto, K., Ito, M., Wakabayashi, K., Miyabayashi, A., Yamada, H. *et al.* (2024) Genetic Variants Supporting the Diagnosis of Primary Ciliary Dyskinesia in Japan Clin Genet 10.1111/cge.14640
936. Hiraide, T., Hayashi, T., Ito, Y., Urushibata, R., Uchida, H., Kitagata, R. *et al.* (2024) Case Report: Novel compound heterozygous TPRKB variants cause Galloway-Mowat syndrome Front Pediatr **12**, 1360867 10.3389/fped.2024.1360867
937. Hiraide, T., Yoshioka, W., Ito, Y., Urushibata, R., Hayashi, T., Ishigaki, H. *et al.* (2024) Familial hyperCKemia with exercise-induced myalgia associated with a novel missense variant in RYR1 Brain and Development Case Reports **2**, 100025 10.1016/j.bdcasr.2024.100025
938. Hirano, Y., Kuroda, Y., Enomoto, Y., Naruto, T., Muroya, K., andKurosawa, K. (2024) Noonan syndrome-like phenotype associated with an ERF frameshift variant Am J Med Genet A **194**, e63652 10.1002/ajmg.a.63652
939. Hirono, K., Hata, Y., Ichimata, S., Nishida, N., Imamura, T., Asano, Y. *et al.* (2024) Sarcomere gene variants did not improve cardiac function in pediatric patients with dilated cardiomyopathy from Japanese cohorts Sci Rep

14, 30469 10.1038/s41598-024-77360-3

940. Hirono, K., Hata, Y., Imamura, T., Tsuboi, K., Takarada, S., Okabe, M. *et al.* (2024) Determination of Genotype and Phenotypes in Pediatric Patients With Biventricular Noncompaction J Am Heart Assoc **13**, e035614 10.1161/jaha.124.035614
941. Hitomi, Y., Ueno, K., Aiba, Y., Nishida, N., Kono, M., Sugihara, M. *et al.* (2024) A genome-wide association study identified PTPN2 as a population-specific susceptibility gene locus for primary biliary cholangitis Hepatology **80**, 776-790 10.1097/HEP.0000000000000894
942. Hobara, T., Higuchi, Y., Yoshida, M., Suehara, M., Ando, M., Yuan, J. H. *et al.* (2024) Genetic and pathophysiological insights from autopsied patient with primary familial brain calcification: novel MYORG variants and astrocytic implications Acta Neuropathol Commun **12**, 136 10.1186/s40478-024-01847-3
943. Honda, S., Yamaguchi, H., Aimono, E., Hara, S., Minamiguchi, S., Norose, T. *et al.* (2024) High-grade Solid Pseudopapillary Neoplasms of the Pancreas: Distinct Clinicopathological Malignant Features With Intriguing Gene Alterations through a Comparison With the Conventional Type Am J Surg Pathol **48**, 353-363 10.1097/PAS.0000000000002177
944. Hsu, J. S., Wu, D. C., Shih, S. H., Liu, J. F., Tsai, Y. C., Lee, T. L. *et al.* (2024) Complete genomic profiles of 1496 Taiwanese reveal curated medical insights J Adv Res **66**, 197-207 10.1016/j.jare.2023.12.018
945. Ibe, M., Tamura, S., Kosako, H., Yamashita, Y., Ishii, M., Tanaka, M. *et al.* (2024) Familial schwannomatosis carrying LZTR1 variant p.R340X with brain tumor: A case report Mol Genet Metab Rep **40**, 101107 10.1016/j.ymgmr.2024.101107
946. Idogawa, M., Mariya, T., Tanaka, Y., Saito, T., Nakase, H., Tokino, T. *et al.* (2024) The frequency and pathogenicity of BRCA1 and BRCA2 variants in the general Japanese population Journal of human genetics **69**, 225-230 10.1038/s10038-024-01233-w
947. Ikuta, T., Suzuki, R., andInoue, A. (2024) The repertoire of G-protein-coupled receptor variations in the Japanese population 54KJPN Genes Cells **29**, 1026-1036 10.1111/gtc.13164
948. Inagaki, N., Okano, T., Kobayashi, M., Fujii, M., Yazaki, Y., Takei, Y. *et al.* (2024) Pediatric hypertrophic cardiomyopathy caused by a novel TNNI3 variant Hum Genome Var **11**, 14 10.1038/s41439-024-00272-1
949. Inamori, K. I., Nakamura, K., Shishido, F., Hsu, J. C., Nagafuku, M., Nitta, T. *et al.* (2024) Functional evaluation of novel variants of B4GALNT1 in a patient with hereditary spastic paraplegia and the general population Front Neurosci **18**, 1437668 10.3389/fnins.2024.1437668
950. Inoue, M., Sebastian, W. A., Sonoda, S., Miyahara, H., Shimizu, N., Shiraishi, H. *et al.* (2024) Biallelic variants in LARS1 induce steatosis in developing zebrafish liver via enhanced autophagy Orphanet J Rare Dis **19**, 219 10.1186/s13023-024-03226-6
951. Inoue, S., Ko, T., Shindo, A., Nomura, S., Yamada, T., Jimba, T. *et al.* (2024) Association Between Clonal Hematopoiesis and Left Ventricular Reverse Remodeling in Nonischemic Dilated Cardiomyopathy JACC Basic Transl Sci **9**, 956-967 10.1016/j.jacbts.2024.04.010
952. Inoue, T., Takase, R., Uchida, K., Kodo, K., Suda, K., Watanabe, Y. *et al.* (2024) The c.1617del variant of TMEM260 is identified as the most frequent single gene determinant for Japanese patients with a specific type of congenital heart disease Journal of human genetics **69**, 215-222 10.1038/s10038-024-01225-w
953. Ishihara, E., Matsubayashi, H., Nishimura, S., Isaka, M., Konno, H., Goto, S. *et al.* (2024) Four cancer cases with pathological germline variant RAD51D c.270_271dup J Obstet Gynaecol Res **50**, 1742-1747 10.1111/jog.16045
954. Ito, M., Morimoto, K., Ohfuji, T., Miyabayashi, A., Wakabayashi, K., Yamada, H. *et al.* (2024) FOXJ1 Variants

Causing Primary Ciliary Dyskinesia with Hydrocephalus: A Case Report from Japan Intern Med **63**, 1433-1437 10.2169/internalmedicine.2565-23

955. Ito, S., Ando, M., Aoki, S., Soma, S., Zhang, J., Hirano, N. *et al.* (2024) Usefulness of multigene liquid biopsy of bile for identifying driver genes of biliary duct cancers Cancer Sci 10.1111/cas.16365
956. Itonaga, T., Maeda, M., Koga, H., Hasegawa, Y., andIhara, K. (2024) Asymptomatic 3-methylglutaconic aciduria type 1 detected by high C5-OH on newborn screening Mol Genet Metab Rep **38**, 101024 10.1016/j.ymgmr.2023.101024
957. Iwasaki, T., Shirota, H., Sasaki, K., Ouchi, K., Nakayama, Y., Oshikiri, H. *et al.* (2024) Specific cancer types and prognosis in patients with variations in the KEAP1-NRF2 system: A retrospective cohort study Cancer Sci **115**, 4034-4044 10.1111/cas.16355
958. Ji, X., Zhao, N., Liu, H., Wu, Y., andLiu, L. (2024) Case report: Two novel compound heterozygous variant of SLC12A3 gene in a gitelman syndrome family and literature review Front Genet **15**, 1391015 10.3389/fgene.2024.1391015
959. Kanamori, H., Yamada, Y., Ito, Y., Shirosaki, K., Yamagishi, S., Maeda, Y. *et al.* (2024) Noninvasive graft monitoring using donor-derived cell-free DNA in Japanese liver transplantation Hepatol Res **54**, 300-314 10.1111/hepr.13978
960. Kashiwa, A., Itoh, H., Makiyama, T., Wada, Y., Ozawa, J., Kato, K. *et al.* (2024) Clinical characterization of type 1 long QT syndrome caused by C-terminus Kv7.1 variants Heart Rhythm **21**, 1113-1120 10.1016/j.hrthm.2024.02.021
961. Kato, M. K., Fujii, E., Asami, Y., Momozawa, Y., Hiranuma, K., Komatsu, M. *et al.* (2024) Clinical features and impact of p53 status on sporadic mismatch repair deficiency and Lynch syndrome in uterine cancer Cancer Sci **115**, 1646-1655 10.1111/cas.16121
962. Kawakami, R., Hiraide, T., Watanabe, K., Miyamoto, S., Hira, K., Komatsu, K. *et al.* (2024) RNA sequencing and target long-read sequencing reveal an intronic transposon insertion causing aberrant splicing Journal of human genetics **69**, 91-99 10.1038/s10038-023-01211-8
963. Kiko, T., Asano, R., Ishibashi, T., Endo, H., Nishi, N., Hayashi, H. *et al.* (2024) Prevalence and Clinical Characteristics of Heterozygous RNF213 p.Arg4810Lys Variant Carriers Diagnosed With Chronic Thromboembolic Pulmonary Hypertension J Am Heart Assoc **13**, e035009 10.1161/jaha.124.035009
964. Kimura, T., Fujita, K., Sakurai, T., Niida, S., Ozaki, K., andShigemizu, D. (2024) Whole-genome sequencing to identify rare variants in East Asian patients with dementia with Lewy bodies NPJ Aging **10**, 52 10.1038/s41514-024-00180-2
965. Kimura-Yoshida, A., Sato, T., Ichihashi, Y., Wasa, M., Narumi, S., Ishii, T. *et al.* (2024) A novel SOX2 frameshift pathogenic variant located in the transactivation domain in a male infant with hypogonadotropic hypogonadism Clin Pediatr Endocrinol **33**, 244-248 10.1297/cpe.2024-0013
966. Kishida, D., Nakamura, A., Yazaki, M., Tsuchiya-Suzuki, A., Ichikawa, T., Shimojima, Y. *et al.* (2024) Variables for differential diagnosis of familial Mediterranean fever: Multiple correspondence analysis of a large Japanese cohort Mod Rheumatol **34**, 784-790 10.1093/mr/road084
967. Kishio, N., Iwama, K., Nakanishi, S., Shindo, R., Yasui, M., Nicho, N. *et al.* (2024) A deletion variant in LMX1B causing nail-patella syndrome in Japanese twins Hum Genome Var **11**, 10 10.1038/s41439-024-00266-z
968. Koga, T., Kita, K., Okumura, J., Yoshiura, K. I., andKawakami, A. (2024) A novel frameshift mutation in ADCK1 identified in a case of chronic fatigue syndrome successfully treated with oral 5-ALA/SFC Immunol Med 1-5

10.1080/25785826.2024.2445399

969. Komaki, S., Kubota, A., Katsuse, K., Kitamura, A., Maeda, M., Matsukawa, T. *et al.* (2024) A Case of a Patient with Calpainopathy Carrying Compound Heterozygous Mutations of a de novo Pathogenic Variant of c.1333G>A and a Novel Variant of c.1331C>T in CAPN3 Intern Med 10.2169/internalmedicine.3435-23

970. Komatsu, K., Kato, M., Kubota, K., Fukumura, S., Yamada, K., Hori, I. *et al.* (2024) Identifying pathogenic variants in rare pediatric neurological diseases using exome sequencing Sci Rep **14**, 24746 10.1038/s41598-024-75020-0

971. Kondo, H., Tsukahara-Kawamura, T., Matsushita, I., Nagata, T., Hayashi, T., Nishina, S. *et al.* (2024) Familial Exudative Vitreoretinopathy With and Without Pathogenic Variants of Norrin/beta-Catenin Signaling Genes Ophthalmol Sci **4**, 100514 10.1016/j.xops.2024.100514

972. Konecna, K., Zapletalova, P., Freiberger, T., andTichy, L. (2024) LDLR gene rearrangements in Czech FH patients likely arise from one mutational event Lipids Health Dis **23**, 36 10.1186/s12944-024-02013-3

973. Koshimizu, E., Kato, M., Misawa, K., Uchiyama, Y., Tsuchida, N., Hamanaka, K. *et al.* (2024) Detection of hidden intronic DDC variant in aromatic L-amino acid decarboxylase deficiency by adaptive sampling Journal of human genetics **69**, 153-157 10.1038/s10038-023-01217-2

974. Koyama, S., Liu, X., Koike, Y., Hikino, K., Koido, M., Li, W. *et al.* (2024) Population-specific putative causal variants shape quantitative traits Nature genetics **56**, 2027-2035 10.1038/s41588-024-01913-5

975. Kubota, K., Hiraide, T., Nakama, M., Adachi, M., Nakashima, M., Saitsu, H. *et al.* (2024) GRIN2A-related disorder causes profound developmental delay and a disorder affecting movement Brain and Development Case Reports **2**, 100034,

976. Kubota, Y., Sato, T., Matsuyama, M., Maruo, Y., Narumi, S., Ishii, T. *et al.* (2024) Siblings of eonatal hyperbilirubinemia with double issense variants Clinical Pediatric Endocrinology **33**, 195-198 10.1297/cpe.33.2024-0028

977. Kuribayashi, S., Fukuhara, S., Kitakaze, H., Tsujimura, G., Imanaka, T., Okada, K. *et al.* (2024) KEAP1-NRF2 system regulates age-related spermatogenesis dysfunction Reprod Med Biol **23**, e12595 10.1002/rmb2.12595

978. Kurihara, Y., Honda, T., Takemoto, A., Seto, K., Endo, S., Tanimoto, K. *et al.* (2024) Immunohistochemistry of p53 surrogates TP53 mutation as an accurate predictor for early-relapse of surgically resected stage I-III lung adenocarcinoma JTCVS Open **20**, 183-193 10.1016/j.xjon.2024.06.005

979. Kuroda, Y., Ikegawa, T., Kato, A., Aida, N., Naruto, T., andKurosawa, K. (2024) Biallelic TXNDC15 variants associated with Joubert syndrome-related molar tooth sign and forebrain malformation Journal of human genetics 10.1038/s10038-024-01290-1

980. Kuroda, Y., Naruto, T., Tsuyusaki, Y., Kato, A., Aida, N., andKurosawa, K. (2024) Role of TOE1 variants at the nuclear localization motif in pontocerebellar hypoplasia 7 Journal of human genetics **69**, 349-355 10.1038/s10038-024-01244-7

981. Kuroda, Y., Saito, Y., Enomoto, Y., Naruto, T., andKurosawa, K. (2024) A Japanese patient with Teebi hypertelorism syndrome and a novel CDH11 EC1 domain variant Am J Med Genet A **194**, 94-99 10.1002/ajmg.a.63386

982. Lin, Y., Xu, J., andYang, T. (2024) [Genetic and phenotypic analysis of MYO15A rare variants associated with autosomal recessive hearing loss] Lin Chuang Er Bi Yan Hou Tou Jing Wai Ke Za Zhi **38**, 38-43 10.13201/j.issn.2096-7993.2024.01.006

983. Lu, H. F., Chou, C. H., Lin, Y. J., Uchiyama, S., Terao, C., Wang, Y. W. *et al.* (2024) The genome-wide association

study of serum IgE levels demonstrated a shared genetic background in allergic diseases Clin Immunol **260**, 109897 10.1016/j.clim.2024.109897

984. Madrid, N. Y., andGiraldo, L. J. M. (2024) Reverse phenotyping and artificial intelligence in identifying novel and De Novo variants in the RAB39B gene associated with X-linked epilepsy and intellectual disability Journal of Artificial Intelligence & Robotics,

985. Maeda, T., Yanagi, T., Kitamura, S., Nishihara, H., Ono, Y., Mizukami, Y. *et al.* (2024) A familial case of extramammary Paget disease: Analysis of whole-exome sequencing EJC Skin Cancer **2**, 100025,

986. Makino, K., Ishii, T., Ogiso, S., Nakakura, A., Nishio, T., Fukumitsu, K. *et al.* (2024) Combination of risk alleles of PNPLA3, TM6SF2, and HSD17B13 of donors can predict recurrence of steatotic liver disease after liver transplantation Hepatol Res 10.1111/hepr.14086

987. Masuda, L., Hasegawa, A., Kamura, H., Hasegawa, F., Yamamura, M., Taniguchi, K. *et al.* (2024) Missense BICD2 variants in fetuses with congenital arthrogryposis and pterygia Hum Genome Var **11**, 32 10.1038/s41439-024-00290-z

988. Masunaga, Y., Ono, H., Fujisawa, Y., Taniguchi, K., Saitsu, H., andOgata, T. (2024) Sotos syndrome with marked overgrowth in three Japanese patients with heterozygous likely pathogenic NSD1 variants: case reports with review of literature Endocr J **71**, 75-81 10.1507/endocrj.EJ23-0502

989. Matsubayashi, H., Kiyozumi, Y., Harada, R., Mukaigawa, T., Sugiura, T., Ishiwatari, H. *et al.* (2024) A Japanese Family Meeting the Clinical Diagnostic Criteria for MEN1 with a MEN1 Variant of Uncertain Significance Intern Med **63**, 1119-1123 10.2169/internalmedicine.2300-23

990. Matsui, H., andHirata, M. (2024) Evaluation of the pathogenic potential of germline DDX41 variants in hematopoietic neoplasms using the ACMG/AMP guidelines Int J Hematol **119**, 552-563 10.1007/s12185-024-03728-w

991. Matsumoto, H., Sasai, H., Kawamoto, N., Katsuyama, M., Minamiyama, M., Kuru, S. *et al.* (2024) Loss-of-function polymorphisms in NQO1 are not associated with the development of subacute myelo-optico-neuropathy Mol Genet Genomic Med **12**, e2470 10.1002/mgg3.2470

992. Matsumoto, M., Oyake, M., Itonaga, T., Maeda, M., Suenobu, S., Sato, D. *et al.* (2024) Characteristic phenotypes of ADH5/ALDH2 deficiency during childhood Eur J Med Genet **69**, 104939 10.1016/j.ejmg.2024.104939

993. Matsuoka, T., Yoshida, T., Kora, K., Yano, N., Taura, Y., Nakamura, T. *et al.* (2024) A mild case of Cockayne syndrome with a novel start-loss variant of ERCC8 Hum Genome Var **11**, 39 10.1038/s41439-024-00297-6

994. Matsushima, N., Shibata, S., Leu, J. H., Vermeulen, A., Duffner, J., Ling, L. E. *et al.* (2024) Pharmacokinetics and Pharmacodynamics of Nipocalimab, a Neonatal Fc Receptor Blocker, in Healthy Japanese Volunteers Clin Drug Investig **44**, 587-599 10.1007/s40261-024-01380-0

995. McFarland, K. N., Tiwari, A., Hashem, V., Zhang, L., Zeng, D., Vincent, J. *et al.* (2024) Extended haplotype with rs41524547-G defines the ancestral origin of SCA10 Human molecular genetics ddae092 10.1093/hmg/ddae092

996. Mishima, Y., Tsuruya, K., Tazawa, Y., Arase, Y., Hirose, S., Shiraishi, K. *et al.* (2024) An Adult Case of Benign Recurrent Intrahepatic Cholestasis Due to MYO5B Deficiency Tokai J Exp Clin Med **49**, 133-136, https://www.ncbi.nlm.nih.gov/pubmed/39182182

997. Mitsutake, A., Kawai, M., Orimo, K., Matsukawa, T., Ishiura, H., Mitsui, J. *et al.* (2024) A Japanese Family with a Novel Pathogenic Variant in KIF1A Presenting with Spastic Paraparesis, Cerebellar Ataxia, and Intellectual Disability Cerebellum **24**, 20 10.1007/s12311-024-01782-y

998. Mitsutake, A., Matsukawa, T., Naito, T., Ishiura, H., Mitsui, J., Harada, H. *et al.* (2024) A Novel De Novo Variant in KCNH5 in a Patient with Refractory Epileptic Encephalopathy Intern Med 3999-3924 10.2169/internalmedicine.3999-24

999. Miura, A., Yamamoto, T., Funayama, K., Koyama, A., Takatsuka, H., Sato, T. *et al.* (2024) Postmortem Identification of Genetic Variations Associated with Sudden Unexpected Death in Young People Int Heart J **65**, 55-62 10.1536/ihj.23-252

1000. Miwa, M., Kitagawa, M., Asami, Y., Kobayashi-Kato, M., Watanabe, T., Ogasawara, A. *et al.* (2024) Prevalence and outcomes of germline pathogenic variants of homologous recombination repair genes in ovarian cancer Cancer Sci **115**, 3952-3962 10.1111/cas.16367

1001. Miyamoto, H., Kawakami, F., Abe, S., Sugita, H., andMatsui, H. (2024) Comprehensive Cancer Genomic Profiling of Liver Metastasis Led to the Unexpected Identification of Colorectal Cancer Intern Med **63**, 63-70 10.2169/internalmedicine.1845-23

1002. Miyanohara, I., Ohori, J., Tabuchi, M., Nishio, S. Y., Yamashita, M., andUsami, S. I. (2024) Comprehensive Genetic Evaluation in Patients with Special Reference to Late-Onset Sensorineural Hearing Loss Genes (Basel) **15**, 571 10.3390/genes15050571

1003. Miyatake, S., Doi, H., Yaguchi, H., Koshimizu, E., Kihara, N., Matsubara, T. *et al.* (2024) Complete nanopore repeat sequencing of SCA27B (GAA-FGF14 ataxia) in Japanese J Neurol Neurosurg Psychiatry 10.1136/jnnp-2024-333541

1004. Mizobuchi, K., Hayashi, T., Tanaka, K., Kuniyoshi, K., Murakami, Y., Nakamura, N. *et al.* (2024) Genetic and Clinical Features of ABCA4-Associated Retinopathy in a Japanese Nationwide Cohort Am J Ophthalmol **264**, 36-43 10.1016/j.ajo.2024.03.007

1005. Momoi, M., Katsumata, Y., Hiraide, T., Shinya, Y., Goto, S., Inami, T. *et al.* (2024) Chronic Thromboembolic Pulmonary Hypertension as RNF213-Associated Vascular Disease JACC Asia **4**, 1017-1019 10.1016/j.jacasi.2024.09.012

1006. Momoi, M., Katsumata, Y., Kunimoto, H., Inami, T., Miya, F., Anzai, A. *et al.* (2024) Clonal Hematopoiesis in Chronic Thromboembolic Pulmonary Hypertension J Am Heart Assoc **13**, e035498 10.1161/jaha.124.035498

1007. Morooka, N., Kido, J., Ueno, H., Misumi, Y., Sugawara, K., Kameyama, S. *et al.* (2024) A pathogenic missense variant, c.2149G>A (p.Gly717Arg), in CDK13 in a female patient with CDK13-related disorder: A case report and literature review of 112 cases Brain and Development Case Reports **2**, 100023 10.1016/j.bdcasr.2024.100023

1008. Munenaga, T., Ichihashi, Y., Nakano, S., Shibata, H., Sato, T., Asanuma, H. *et al.* (2024) Characteristic external genitalia in male neonates with 5alpha-reductase deficiency Endocr J EJ23-0740 10.1507/endocrj.EJ23-0740

1009. Muranishi, Y., Itonaga, T., Ihara, K., Katoh-Fukui, Y., Tamaoka, S., Hattori, A. *et al.* (2024) PTPN11 and FLNA variants in a boy with ambiguous genitalia, short stature, and non-specific dysmorphic features Clin Pediatr Endocrinol **33**, 169-173 10.1297/cpe.2023-0074

1010. Muranishi, Y., Kobori, Y., Katoh-Fukui, Y., Tamaoka, S., Hattori, A., Osaka, A. *et al.* (2024) Systematic molecular analyses for 115 karyotypically normal men with isolated non-obstructive azoospermia Hum Reprod **39**, 1131-1140 10.1093/humrep/deae057

1011. Nagao, T., Inoue, M., Ito, Y., Kunihara, T., Kawame, H., Samura, O. *et al.* (2024) Loeys-Dietz syndrome with a novel in-frame SMAD3 deletion diagnosed as a result of postpartum aortic dissection: A case report Taiwan J Obstet Gynecol **63**, 225-228 10.1016/j.tjog.2024.01.018

1012. Nagashima, T., Yamaguchi, K., Urakami, K., Shimoda, Y., Ohnami, S., Ohshima, K. *et al.* (2024) Evaluation of whole genome sequencing utility in identifying driver alterations in cancer genome Sci Rep **14**, 23898 10.1038/s41598-024-74272-0

1013. Nagayama, S., Takahashi, H., Hasegawa, F., Hori, A., Kizami, S., Furukawa, R. *et al.* (2024) A novel variant in IFT122 associated with a severe phenotype of cranioectodermal dysplasia Congenit Anom (Kyoto) **64**, 177-181 10.1111/cga.12569

1014. Nakajima, E., Yokohama, Y., Sugiyama, S., Taketazu, M., Mitsube, K., Yamada, T. *et al.* (2024) Unclassifiable short-rib thoracic dysplasia diagnosed using targeted gene panel sequencing Hum Genome Var **11**, 44 10.1038/s41439-024-00302-y

1015. Nakama, M., Miwa, Y., Manabe, S., Shimamoto, S., andOhnishi, H. (2024) Novel variant of FBN2 in a patient with congenital contractual arachnodactyly Hum Genome Var **11**, 7 10.1038/s41439-024-00264-1

1016. Nakamura, A., Naruse, H., Mitsutake, A., Mitsui, J., Morishita, S., Iwakoshi, M. *et al.* (2024) A novel de novo disease-causing variant in a pediatric patient with spastic paraplegia Neurology and Clinical Neuroscience 10.1111/ncn3.12860

1017. Nakamura, H., Kikuchi, A., Sakai, H., Kamimura, M., Watanabe, Y., Onuma, R. *et al.* (2024) Case Report: Identification of a CARD8 variant in all three patients with PFAPA syndrome complicated with Kawasaki disease Front Pediatr **12**, 1340263 10.3389/fped.2024.1340263

1018. Nakamura, S., Ibi, K., Tanaka, H., Takami, H., Okada, K., Takasugi, N. *et al.* (2024) Ventriculosubgaleal shunt placement for hydrocephalus in osteogenesis imperfecta with novel compound heterozygous CRTAP variants Hum Genome Var **11**, 16 10.1038/s41439-024-00274-z

1019. Nakamura, W., Hirata, M., Oda, S., Chiba, K., Okada, A., Mateos, R. N. *et al.* (2024) Assessing the efficacy of target adaptive sampling long-read sequencing through hereditary cancer patient genomes NPJ Genom Med **9**, 11 10.1038/s41525-024-00394-z

1020. Nakamura, Y., Shimada, I. S., Maroofian, R., Falabella, M., Zaki, M. S., Fujimoto, M. *et al.* (2024) Biallelic null variants in PNPLA8 cause microcephaly by reducing the number of basal radial glia Brain awae185 10.1093/brain/awae185

1021. Nakashima, M., Shiroshima, T., Fukaya, M., Sugawara, T., Sakagami, H., andYamazawa, K. (2024) C-terminal truncations in IQSEC2: implications for synaptic localization, guanine nucleotide exchange factor activity, and neurological manifestations Journal of human genetics **69**, 119-123 10.1038/s10038-023-01210-9

1022. Nakashima, T., Yamamoto, R., Ohno, M., Sugino, H., Takahashi, M., Funakoshi, Y. *et al.* (2024) Development of a rapid and comprehensive genomic profiling test supporting diagnosis and research for gliomas Brain Tumor Pathol **41**, 50-60 10.1007/s10014-023-00476-3

1023. Nakato, D., Yasue, Y., Matsubara, K., Suzuki, H., Kosaki, R., Takenouchi, T. *et al.* (2024) De novo variants in UPF1 associated with intellectual disabilities: Human genetic and functional evidences using Drosophila model Eur J Med Genet **72**, 104983 10.1016/j.ejmg.2024.104983

1024. Narumi, S. (2024) Deciphering the mystery of CHNG3 Ann Pediatr Endocrinol Metab **29**, 279-283 10.6065/apem.2448186.093

1025. Narumi, S., Nagasaki, K., Kiriya, M., Uehara, E., Akiba, K., Tanase-Nakao, K. *et al.* (2024) Functional variants in a TTTG microsatellite on 15q26.1 cause familial nonautoimmune thyroid abnormalities Nature genetics **56**, 869-876 10.1038/s41588-024-01735-5

1026. Narusawa, H., Ogawa, T., Yagasaki, H., Nagasaki, K., Urakawa, T., Saito, T. *et al.* (2024) Comprehensive study

on central precocious puberty: molecular and clinical analyses in 90 patients J Clin Endocrinol Metab 10.1210/clinem/dgae666

1027. Naruse, H., Ishiura, H., Esaki, K., Mitsui, J., Satake, W., Greimel, P. *et al.* (2024) SPTLC2 variants are associated with early-onset ALS and FTD due to aberrant sphingolipid synthesis Ann Clin Transl Neurol **11**, 946-957 10.1002/acn3.52013

1028. Niida, Y., Fujita, W., Togi, S., andUra, H. (2024) Investigation of a novel PROS1 splicing variant in a patient with protein S deficiency Hum Genome Var **11**, 28 10.1038/s41439-024-00286-9

1029. Niizeki, H., Tanaka, R., Nomura, T., Seki, A., Miyasaka, M., Matsumoto, Y. *et al.* (2024) Lack of cutis verticis gyrata is associated with c.1279_1290del12 of SLCO2A1 in 43 Japanese patients with pachydermoperiostosis J Dermatol Sci **114**, 86-88 10.1016/j.jdermsci.2024.03.008

1030. Nishi, E., Yanagi, K., Kaname, T., andOkamoto, N. (2024) Clinical details of individuals with Rauch-Steindl syndrome due to NSD2 truncating variants Mol Genet Genomic Med **12**, e2396 10.1002/mgg3.2396

1031. Nishi, E., Yanagi, K., Okamoto, N., andKaname, T. (2024) Clinical Features of a Japanese Girl With Radio-Tartaglia Syndrome due to a SPEN Truncating Variant Am J Med Genet A e63910 10.1002/ajmg.a.63910

1032. Nishida, T., Naito, Y., Takahashi, T., Saito, T., Hisamori, S., Manaka, D. *et al.* (2024) Molecular and clinicopathological features of KIT/PDGFRA wild-type gastrointestinal stromal tumors Cancer Sci **115**, 894-904 10.1111/cas.16058

1033. Nishino, T., Yumura, M., Sunami, K., Kubo, T., Ichikawa, H., Yasuda, T. *et al.* (2024) Leveraging non-coding regions to guarantee the accuracy of small-sized panel-based tumor mutational burden estimates Cancer Sci 10.1111/cas.16342

1034. Nishiyama, A., Niihori, T., Suzuki, N., Izumi, R., Akiyama, T., Kato, M. *et al.* (2024) Updated Genetic Analysis of Japanese Familial ALS Patients Carrying SOD1 Variants Revealed Phenotypic Differences for Common Variants Neurol Genet **10**, e200196 10.1212/NXG.0000000000200196

1035. Noguchi, A., Tezuka, T., Okuda, H., Kobayashi, H., Harada, K. H., Yoshida, T. *et al.* (2024) Genetic Analysis of SCN11A, SCN10A, and SCN9A in Familial Episodic Pain Syndrome (FEPS) in Japan and Proposal of Clinical Diagnostic Criteria Int J Mol Sci **25**, 6832 10.3390/ijms25136832

1036. Ochiai, K., Ohashi, T., Mori, H., Saitsu, H., andTakanashi, J. I. (2024) Chloride Voltage-Gated Channel 2 (CLCN2)-Related Leukoencephalopathy Exhibiting Reduced Choline Levels on Magnetic Resonance Spectroscopy Cureus **16**, e61716 10.7759/cureus.61716

1037. Odate, T., Satomi, K., Kubo, T., Matsushita, Y., Ueno, T., Kurose, A. *et al.* (2024) Inflammatory Rhabdomyoblastic Tumor: Clinicopathologic and Molecular Analysis of 13 Cases Mod Pathol **37**, 100359 10.1016/j.modpat.2023.100359

1038. Ohhara, Y., Tomaru, U., Kinoshita, I., Hatanaka, K. C., Noguchi, T., Hatanaka, Y. *et al.* (2024) Polymorphisms of the PD-L1 gene 3'-untranslated region are associated with the expression of PD-L1 in non-small cell lung cancer Genes Chromosomes Cancer **63**, e23216 10.1002/gcc.23216

1039. Ohira, M., Saitsu, H., Nakashima, M., Sato, N., Inoue, K., andTakao, M. (2024) CLCN2-related leukoencephalopathy with novel compound heterozygous variants followed with magnetic resonance imaging over 17 years: a case report BMC Neurol **24**, 412 10.1186/s12883-024-03919-2

1040. Ojima, T., Namba, S., Suzuki, K., Yamamoto, K., Sonehara, K., Narita, A. *et al.* (2024) Body mass index stratification optimizes polygenic prediction of type 2 diabetes in cross-biobank analyses Nature genetics **56**, 1100-1109 10.1038/s41588-024-01782-y

1041. Okamoto, N., Yoshida, S., Ogitani, A., Etani, Y., Yanagi, K., andKaname, T. (2024) Biallelic loss-of-function variants of EZH1 cause a novel developmental disorder with central precocious puberty Am J Med Genet A e63726 10.1002/ajmg.a.63726

1042. Okamoto, T., Mizuta, R., Takahashi, Y., Otani, Y., Sasaki, E., Horio, Y. *et al.* (2024) Genomic landscape of glioblastoma without IDH somatic mutation in 42 cases: a comprehensive analysis using RNA sequencing data J Neurooncol **167**, 489-499 10.1007/s11060-024-04628-z

1043. Okubo, S., Naruse, H., Ishiura, H., Sudo, A., Esaki, K., Mitsui, J. *et al.* (2024) Genetic and functional analyses of SPTLC1 in juvenile amyotrophic lateral sclerosis J Neurol **272**, 36 10.1007/s00415-024-12776-5

1044. Orimo, K., Mitsui, J., Matsukawa, T., Tanaka, M., Nomoto, J., Ishiura, H. *et al.* (2024) Association study of GBA1 variants with MSA based on comprehensive sequence analysis -Pitfalls in short-read sequence analysis depending on the human reference genome Journal of human genetics 1-9 10.1038/s10038-024-01266-1

1045. Osawa, K., Yamamoto, S., Yamano, Y., Kita, A., Okamoto, K., Kato, N. *et al.* (2024) Overlapping Atypical Hemolytic Uremic Syndrome and C3 Glomerulopathy with Mutation in CFI in a Japanese Patient: A Case Report Intern Med **63**, 1777-1782 10.2169/internalmedicine.2713-23

1046. Osawa, Y., Ichiwata, N., Kenmotsu, J., Okada, T., Masunaga, Y., Ogata, T. *et al.* (2024) Diabetes mellitus with severe insulin resistance in a young male patient with a heterozygous pathogenic IRS1 frameshift variant Clin Pediatr Endocrinol **33**, 87-93 10.1297/cpe.2023-0081

1047. Otgonbayar, G., Lo, T., Hayashi, Y., Furuta, S., Aleksic, B., Nawa, Y. *et al.* (2024) Study of the genetic association between selected 3q29 region genes and schizophrenia and autism spectrum disorder in the Japanese population Nagoya J Med Sci **86**, 216-222, https://www.ncbi.nlm.nih.gov/pmc/articles/PMC11219234/pdf/2186-3326-86-0216.pdf

1048. Ozaki, K., Yatsuka, Y., Oyazato, Y., Nishiyama, A., Nitta, K. R., Kishita, Y. *et al.* (2024) Biallelic GGGCC repeat expansion leading to NAXE-related mitochondrial encephalopathy NPJ Genom Med **9**, 48 10.1038/s41525-024-00429-5

1049. Ozawa, S. W., Inomata, S., Hata, Y., Takarada, S., Okabe, M., Nakaoka, H. *et al.* (2024) Novel MYH7 Variant in the Neonate of a Mother with Gestational Diabetes Mellitus Showing Left Ventricular Hypertrophy and Noncompaction Genes (Basel) **15**, 10.3390/genes15030381

1050. Park, J. E., Kang, M. C., Lee, T., Cho, E. H., Jang, M. A., Won, D. *et al.* (2024) Prevalence Estimation of the PALB2 Germline Variant in East Asians and Koreans through Population Database Analysis Cancers (Basel) **16**, 10.3390/cancers16193318

1051. Sadamitsu, K., Yanagi, K., Hasegawa, Y., Murakami, Y., Low, S. E., Ooshima, D. *et al.* (2024) A novel homozygous variant of the PIGK gene caused by paternal disomy in a patient with neurodevelopmental disorder, cerebellar atrophy, and seizures Journal of human genetics 1-11 10.1038/s10038-024-01264-3

1052. Saijo, N., Yaoita, H., Takayama, J., Ota, C., Kawai, E., Kimura, M. *et al.* (2024) A Prevalent TMEM260 Deletion Causes Conotruncal Heart Defects, Including Truncus Arteriosus Am J Med Genet A e63906 10.1002/ajmg.a.63906

1053. Saito, S., Saito, Y., Sato, S., Aoki, S., Fujita, H., Ito, Y. *et al.* (2024) Gene-specific somatic epigenetic mosaicism of FDFT1 underlies a non-hereditary localized form of porokeratosis Am J Hum Genet **111**, 896-912 10.1016/j.ajhg.2024.03.017

1054. Sako, S., Oishi, K., Ida, H., andImagawa, E. (2024) Allele frequency of pathogenic variants causing acid sphingomyelinase deficiency and Gaucher disease in the general Japanese population Hum Genome Var **11**, 24

10.1038/s41439-024-00282-z

1055. Sakuma, N., Nishio, S. Y., Goto, S. I., Honkura, Y., Oda, K., Takeda, H. *et al.* (2024) Detailed Clinical Features of PTPRQ-Associated Hearing Loss Identified in a Large Japanese Hearing Loss Cohort Genes (Basel) **15**, 10.3390/genes15040489

1056. Samejima, M., Nakashima, M., Shibasaki, J., Saitsu, H., andKato, M. (2024) Splicing variant of WDR37 in a case of Neurooculocardiogenitourinary syndrome Brain Dev **46**, 154-159 10.1016/j.braindev.2023.11.007

1057. Sano, S., Iwamoto, S., Matsushita, R., Masunaga, Y., Fujisawa, Y., andOgata, T. (2024) A novel GNAS-Gsalpha splice donor site variant in a girl with pseudohypoparathyroidism type 1A and her mother with pseudopseudohypoparathyroidism Clin Pediatr Endocrinol **33**, 66-70 10.1297/cpe.2023-0065

1058. Sato, M., Sato, A., Saiki, H., Kato, K., Ohno, S., andHorie, M. (2024) Vasospastic angina preceding diagnosis of arrhythmogenic cardiomyopathy in a young athlete HeartRhythm Case Rep **10**, 577-581 10.1016/j.hrcr.2024.05.017

1059. Sato, S., Iino, C., Sasada, T., Soma, G., Furusawa, K., Yoshida, K. *et al.* (2024) Epidemiological Study on the Interaction between the PNPLA3 (rs738409) and Gut Microbiota in Metabolic Dysfunction-Associated Steatotic Liver Disease Genes (Basel) **15**, 10.3390/genes15091172

1060. Sato, T., Ichihashi, Y., Sugie, H., Ishii, T., andHasegawa, T. (2024) A novel 2.4-kb PHKA2 deletion in a boy with glycogen storage disease type IXa Congenit Anom (Kyoto) **64**, 63-65 10.1111/cga.12555

1061. Setoguchi, A., Kawano, H., Okano, S., Honda, T., Kato, T., Dateki, S. *et al.* (2024) An Iron-chelating Agent Improved the Cardiac Function in a Patient with Severe Heart Failure Due to Hereditary Hemochromatosis Intern Med **63**, 253-258 10.2169/internalmedicine.1809-23

1062. Seyama, K., Sato, T., Sato, S., Hirai, T., Intractable Respiratory, D., Pulmonary Hypertension Research Group, M. o. H. L. *et al.* (2024) Allelic frequency of pathogenic alpha(1)-antitrypsin variants in the Japanese population: Results from a survey of open Japanese genetic variation databases Respir Investig **62**, 794-797 10.1016/j.resinv.2024.07.001

1063. Shibata, N., Numakura, C., Hamajima, T., Miyako, K., Fujiwara, I., Mori, J. *et al.* (2024) Clinical and molecular analyses of isolated central congenital hypothyroidism based on a survey conducted in Japan Endocr J **71**, 471-480 10.1507/endocrj.EJ23-0391

1064. Shiga, Y., Hashimoto, K., Fujita, K., Maekawa, S., Sato, K., Kubo, S. *et al.* (2024) Identification of OPTN p.(Asn51Thr): A novel pathogenic variant in primary open-angle glaucoma Genetics in Medicine Open **2**, 100839 10.1016/j.gimo.2023.100839

1065. Shimura, K., Ichihashi, Y., Nakano, S., Sato, T., Hamajima, T., Numasawa, K. *et al.* (2024) DHX37 Variant is One of Common Genetic Causes in Japanese Patients with Testicular Regression Syndrome / Partial Gonadal Dysgenesis without Mullerian Derivatives Horm Res Paediatr 10.1159/000537761

1066. Shirai, H., Saito-Sato, N., Horiuchi, E., Kikuchi, H., Kadowaki, S., Ohnishi, H. *et al.* (2024) Case report: Adult case of A20 haploinsufficiency suspected as neuro-Behcet disease Front Immunol **15**, 1508307 10.3389/fimmu.2024.1508307

1067. Shiraishi, T., Katayama, Y., Nishiyama, M., Shoji, H., Miyakawa, T., Mizoo, T. *et al.* (2024) The complex etiology of autism spectrum disorder due to missense mutations of CHD8 Mol Psychiatry 1-16 10.1038/s41380-024-02491-y

1068. Shitara, K., Muro, K., Watanabe, J., Yamazaki, K., Ohori, H., Shiozawa, M. *et al.* (2024) Baseline ctDNA gene alterations as a biomarker of survival after panitumumab and chemotherapy in metastatic colorectal cancer Nat

Med **30**, 730-739 10.1038/s41591-023-02791-w

1069. Shoji, T., Yamauchi, I., Kawasaki, H., Iwanaga, K., Hakata, T., Tanaka, D. *et al.* (2024) Case report: Duplication of the GCK gene is a novel cause of nesidioblastosis: evidence from a case with Silver-Russell syndrome-like phenotype related to chromosome 7 Front Endocrinol (Lausanne) **15**, 1431547 10.3389/fendo.2024.1431547
1070. Shoji, Y., Hayashida, S., Masuda, H., Tachibana, E., andOkumura, Y. (2024) A case report of familial catecholaminergic polymorphic ventricular tachycardia with a novel mutation in the ryanodine receptor 2 Eur Heart J Case Rep **8**, ytae652 10.1093/ehjcr/ytae652
1071. Sole-Navais, P., Juodakis, J., Ytterberg, K., Wu, X., Bradfield, J. P., Vaudel, M. *et al.* (2024) Genome-wide analyses of neonatal jaundice reveal a marked departure from adult bilirubin metabolism Nat Commun **15**, 7550 10.1038/s41467-024-51947-w
1072. Sonehara, K., Yano, Y., Naito, T., Goto, S., Yoshihara, H., Otani, T. *et al.* (2024) Common and rare genetic variants predisposing females to unexplained recurrent pregnancy loss Nat Commun **15**, 5744 10.1038/s41467-024-49993-5
1073. Suga, A., Mizobuchi, K., Inooka, T., Yoshitake, K., Minematsu, N., Tsunoda, K. *et al.* (2024) A homozygous structural variant of RPGRIP1 is frequently associated with achromatopsia in Japanese patients with IRD Genetics in Medicine Open **2**, 101843 10.1016/j.gimo.2024.101843
1074. Sumida, T., Ogawa, S., Higo, S., Kuramoto, Y., Eto, R., Ikeda, Y. *et al.* (2024) Four cardiomyopathy patients with a heterozygous DSG2 p.Arg119Ter variant Hum Genome Var **11**, 47 10.1038/s41439-024-00304-w
1075. Suzuki, H., Muramatsu, Y., Miya, F., Asada, H., Yamada, M., Nishimura, G. *et al.* (2024) Biallelic loss-of-function variants in the centriolar protein CCP110 leads to a ciliopathy-like phenotype Eur J Med Genet **70**, 104955 10.1016/j.ejmg.2024.104955
1076. Suzuki, S., andSaito, Y. (2024) Genomic Analysis of Advanced Phyllodes Tumors Using Next-Generation Sequencing and Their Chemotherapy Response: A Retrospective Study Using the C-CAT Database Medicina (Kaunas) **60**, 10.3390/medicina60111898
1077. Suzuki, S., Saito, Y., Saito, K., Yamada, Y., Takahashi, K., Kumanishi, R. *et al.* (2024) Limited Efficacy of Anti-EGFR Monoclonal Antibodies in Colorectal Cancer Patients with Rare RAS Variants: Analysis of the C-CAT Database Curr Issues Mol Biol **46**, 14476-14486 10.3390/cimb46120869
1078. Suzuki, T., Kurano, M., Isono, A., Uchino, T., Sayama, Y., Tomomitsu, H. *et al.* (2024) Genetic and biochemical analysis of severe hypertriglyceridemia complicated with acute pancreatitis or with low post-heparin lipoprotein lipase mass Endocr J **71**, 447-460 10.1507/endocrj.EJ23-0438
1079. Suzuki, Y., Iemura, R., Sutani, A., Mizuno, Y., Adachi, E., Ushiama, M. *et al.* (2024) Familial and early recurrent pheochromocytoma in a child with a novel in-frame duplication variant of VHL Clin Pediatr Endocrinol **33**, 229-237 10.1297/cpe.2024-0020
1080. Takaiso, N., Imoto, I., Matsumoto, T., andYoshimura, A. (2024) Novel MLH1 nonsense variant in a patient with suspected Lynch syndrome Hum Genome Var **11**, 36 10.1038/s41439-024-00294-9
1081. Takaiso, N., Imoto, I., Yoshimura, A., Ouchi, A., Komori, K., Iwata, H. *et al.* (2024) BARD1 deletion in a patient with suspected hereditary colorectal cancer Hum Genome Var **11**, 11 10.1038/s41439-024-00267-y
1082. Takamatsu, S., Hillman, R. T., Yoshihara, K., Baba, T., Shimada, M., Yoshida, H. *et al.* (2024) Molecular classification of ovarian high-grade serous/endometrioid carcinomas through multi-omics analysis: JGOG3025-TR2 study Br J Cancer **131**, 1340-1349 10.1038/s41416-024-02837-x
1083. Takami, K., Tsuji, S., Jokoji, R., Okubo, Y., andHigashiyama, M. (2024) Paediatric pustulotic arthro-osteitis patient

with an IL36RN variant, heterozygous c.115+6T>C, who was successfully treated with tonsillectomy: A case report and literature review Exp Dermatol **33**, e15016 10.1111/exd.15016

1084. Takao, A., Yamaguchi, T., Eguchi, H., Okazaki, Y., Ishikawa, H., Akagi, K. *et al.* (2024) Genetic Testing of Japanese Patients with Serrated Polyposis Syndrome: A Multicentric Study J Anus Rectum Colon **8**, 348-355 10.23922/jarc.2024-030

1085. Takase, K., Susa, S., Sato, H., Hada, Y., Nagaoka, K., Takakubo, N. *et al.* (2024) Identification of causative gene variants for patients with known monogenic diabetes using a targeted next-generation sequencing panel in a single-center study Diabetol Int **15**, 203-211 10.1007/s13340-023-00669-3

1086. Takeda, Y., Ueki, M., Matsuhiro, J., Walinda, E., Tanaka, T., Yamada, M. *et al.* (2024) A de novo dominant-negative variant is associated with OTULIN-related autoinflammatory syndrome J Exp Med **221**, 10.1084/jem.20231941

1087. Takegami, N., Matsukawa, T., Hamada, M., Tanifuji, S., Tamura, T., Yamaguchi-Takegami, N. *et al.* (2024) Adrenomyeloneuropathy with Later Development of Cerebral Form Caused by a Hemizygous Splice-site Variant in ABCD1 Intern Med **63**, 999-1004 10.2169/internalmedicine.2240-23

1088. Takeuchi, I., Tanase-Nakao, K., Ogawa, A., Sugawara, T., Migita, O., Kashima, M. *et al.* (2024) Congenital anaemia associated with loss-of-function variants in DNA polymerase epsilon 1 J Med Genet **61**, 239-243 10.1136/jmg-2023-109444

1089. Tanaka, H., Fukawa, Y., Yamamoto, K., Tanimoto, K., Takemoto, A., Mori, T. *et al.* (2024) Prognostic Impact and Genomic Backgrounds of Renal Parenchymal Infiltration or Micronodular Spread in Nonmetastatic Clear Cell Renal Cell Carcinoma Mod Pathol **37**, 100590 10.1016/j.modpat.2024.100590

1090. Tanase-Nakao, K., Iwahashi-Odano, M., Sugisawa, C., Abe, K., Muroya, K., Yamamoto, Y. *et al.* (2024) Genotype–Phenotype Correlations in 30 Japanese Patients With Congenital Hypothyroidism Attributable to TG Defects The Journal of Clinical Endocrinology & Metabolism **109**, 2358-2365 10.1210/clinem/dgae098

1091. Targovnik, H. M., Barh, D., Papendieck, P., Adrover, E., Gallo, A. M., Chiesa, A. *et al.* (2024) A novel pathogenic DICER1 gene variant is associated with hereditary multinodular goiter in an Argentine family as evidenced by clinical, biochemical and molecular genetic analysis Endocrine 10.1007/s12020-024-04098-3

1092. Tezuka, T., Ishiguro, M., Taniguchi, D., Osogaguchi, E., Shiba-Fukushima, K., Ogata, J. *et al.* (2024) Clinical characteristics and pathophysiological properties of newly discovered LRRK2 variants associated with Parkinson's disease Neurobiol Dis **199**, 106571 10.1016/j.nbd.2024.106571

1093. Tokunaga, S., Shimomura, H., Taniguchi, N., Yanagi, K., Kaname, T., Okamoto, N. *et al.* (2024) A novel DLG4 variant causes DLG4-related synaptopathy with intellectual regression Hum Genome Var **11**, 1 10.1038/s41439-023-00260-x

1094. Tomofuji, Y., Edahiro, R., Sonehara, K., Shirai, Y., Kock, K. H., Wang, Q. S. *et al.* (2024) Quantification of escape from X chromosome inactivation with single-cell omics data reveals heterogeneity across cell types and tissues Cell Genom **4**, 100625 10.1016/j.xgen.2024.100625

1095. Tomomasa, D., Lee, B. H., Hirata, Y., Inoue, Y., Majima, H., Imanaka, Y. *et al.* (2024) Inherited CARD9 Deficiency Due to a Founder Effect in East Asia J Clin Immunol **44**, 121 10.1007/s10875-024-01724-7

1096. Tonouchi, E., Morita, K. I., Harazono, Y., Hoshino, K., andYoda, T. (2024) NF1 with 47,XYY mosaicism diagnosed by mandibular neurofibromas Hum Genome Var **11**, 22 10.1038/s41439-024-00279-8

1097. Torazawa, S., Miyawaki, S., Imai, H., Hongo, H., Ishigami, D., Shimizu, M. *et al.* (2024) RNF213 p.Arg4810Lys Wild Type is Associated with De Novo Hemorrhage in Asymptomatic Hemispheres with Moyamoya Disease Transl

Stroke Res **15**, 729-738 10.1007/s12975-023-01159-z

1098. Tsuchida, M., Goto, S., Watanabe, H., Goto, S., Yamaguchi, H., andNarita, I. (2024) Defective C3d caused by C3 p.W1034R in inherited atypical hemolytic uremic syndrome Mol Genet Genomic Med **12**, e2288 10.1002/mgg3.2288
1099. Tsuda, S., Sakamoto, A., Kawaguchi, H., Uchiyama, T., Kaname, T., Yanagi, K. *et al.* (2024) Novel biallelic GNE variants identified in a patient with chronic thrombocytopenia without any symptoms of myopathy Ann Hematol **103**, 5945-5950 10.1007/s00277-024-06104-0
1100. Tsujikawa, T., Ohno, K., Morita, K. I., Saburi, S., Mitsuda, J., Yoshimura, K. *et al.* (2024) Clinical, genomic and immune microenvironmental determinants of nivolumab response in head and neck squamous cell carcinoma Front Immunol **15**, 1390873 10.3389/fimmu.2024.1390873
1101. Tsujimoto, T., Ou, Y., Suzuki, M., Murata, Y., Inubushi, T., Nagata, M. *et al.* (2024) Compromised actin dynamics underlie the orofacial cleft in Baraitser-Winter Cerebrofrontofacial syndrome with a variant in ACTB Human molecular genetics **33**, 1975-1985 10.1093/hmg/ddae133
1102. Uchida, N., Ishii, T., Nishimura, G., Sato, T., Kuratsuji, G., Nagasaki, K. *et al.* (2024) RMRP-related short stature: A report of six additional Japanese individuals with cartilage hair hypoplasia and literature review Am J Med Genet A **194**, e63562 10.1002/ajmg.a.63562
1103. Uehara, E., Hori, N., Tanase-Nakao, K., Akiba, K., Sueoka, H., Matsubara, K. *et al.* (2024) Congenital Hypothyroidism with Thyroid in situ: A Case Report with NKX2-1 and DUOX2 Hypomorphic Variants Horm Res Paediatr 1-7 10.1159/000538895
1104. Uehara, T., Seki, E., Nonoda, Y., Kumaki, T., Tsuyusaki, Y., Aida, N. *et al.* (2024) Two siblings with acute necrotizing encephalopathy associated with variants of LARS1 Am J Med Genet A e63803 10.1002/ajmg.a.63803
1105. Usami, S. I., Nishio, S. Y., Gavilan, J., Van de Heyning, P., Mertens, G., Karltorp, E. *et al.* (2024) HEARRING group genetic marker study: genetic background of CI patients Acta Otolaryngol **144**, 542-551 10.1080/00016489.2024.2416082
1106. Utsumi, T., Tsumura, M., Yashiro, M., Kato, Z., Noma, K., Sakura, F. *et al.* (2024) Exclusive Characteristics of the p.E555K Dominant-Negative Variant in Autosomal Dominant E47 Deficiency J Clin Immunol **44**, 167 10.1007/s10875-024-01758-x
1107. Utsuno, Y., Hamada, K., Hamanaka, K., Miyoshi, K., Tsuchimoto, K., Sunada, S. *et al.* (2024) Novel missense variants cause intermediate phenotypes in the phenotypic spectrum of SLC5A6-related disorders Journal of human genetics **69**, 69-77 10.1038/s10038-023-01206-5
1108. Waman, V. P., Ashford, P., Lam, S. D., Sen, N., Abbasian, M., Woodridge, L. *et al.* (2024) Predicting human and viral protein variants affecting COVID-19 susceptibility and repurposing therapeutics Sci Rep **14**, 14208 10.1038/s41598-024-61541-1
1109. Wang, L., Du, R., Han, L., Yang, R., andLi, Y. (2024) A new missense mutation c.1240A>G in fumarate hydratase gene leads to uterine leiomyoma associated hereditary leiomyomatosis and renal cell cancer (HLRCC) syndrome in Chinese Transl Oncol **45**, 101963 10.1016/j.tranon.2024.101963
1110. Wang, Z., Kometani, M., Zeitlin, L., Wilnai, Y., Kinoshita, A., Yoshiura, K. I. *et al.* (2024) Heterozygous mutations in the straitjacket region of the latency-associated peptide domain of TGFB2 cause Camurati-Engelmann disease type II Journal of human genetics **69**, 599-605 10.1038/s10038-024-01274-1
1111. Watanabe, D., Okamoto, N., Kobayashi, Y., Suzuki, H., Kato, M., Saitoh, S. *et al.* (2024) Biallelic structural

variants in three patients with ERCC8-related Cockayne syndrome and a potential pitfall of copy number variation analysis Sci Rep **14**, 19741 10.1038/s41598-024-70831-7

1112. Watanabe, D., Tsujioka, Y., Nakato, D., Yamada, M., Suzuki, H., Ohnishi, T. *et al.* (2024) Digital clubbing without hypoxia for lysinuric protein intolerance Eur J Med Genet **71**, 104967 10.1016/j.ejmg.2024.104967
1113. Watanabe, D., Yagasaki, H., Narusawa, H., andInukai, T. (2024) Coinheritance of HNF1A and glucokinase variants in maturity-onset diabetes of the young Endocrinol Diabetes Metab Case Rep **2024**, 10.1530/EDM-23-0100
1114. Watanabe, K., Bunai, T., Sakamoto, M., Ishigaki, S., Iwakura, T., Ohashi, N. *et al.* (2024) A family with neuronal intranuclear inclusion disease with focal segmental glomerulosclerosis J Neurol 1-11 10.1007/s00415-024-12593-w
1115. Watanabe, K., Nishio, S. Y., Usami, S. I., andDeafness Gene Study, C. (2024) The prevalence and clinical features of MYO7A-related hearing loss including DFNA11, DFNB2 and USH1B Sci Rep **14**, 8326 10.1038/s41598-024-57415-1
1116. Wu, X., Yoshino, T., Maeda-Minami, A., Ishida, S., Tanaka, M., Nishi, A. *et al.* (2024) Exploratory study of cold hypersensitivity in Japanese women: genetic associations and somatic symptom burden Sci Rep **14**, 1918 10.1038/s41598-024-52119-y
1117. Yagi, S., Kadota, M., Bando, R., Miyamoto, R., Morino, H., Kakutani, A. *et al.* (2024) Pulmonary Arterial Hypertension in Neurofibromatosis Type 1: A Case with a Novel NF1 Gene Mutation Intern Med 3856-3824 10.2169/internalmedicine.3856-24
1118. Yamada, S., Umehara, T., Sonehara, K., Kijima, N., Kawabata, S., Takano, K. *et al.* (2024) Genome-wide association study on meningioma risk in Japan: a multicenter prospective study J Neurooncol **169**, 281-286 10.1007/s11060-024-04727-x
1119. Yamaguchi, T., Ikegami, M., Aruga, T., Kanemasa, Y., Horiguchi, S. I., Kawai, K. *et al.* (2024) Genomic landscape of comprehensive genomic profiling in patients with malignant solid tumors in Japan Int J Clin Oncol **29**, 1417-1431 10.1007/s10147-024-02554-8
1120. Yamamoto, K., Lee, Y., Masuda, T., Ozono, K., Iwatani, Y., Watanabe, M. *et al.* (2024) Functional landscape of genome-wide postzygotic somatic mutations between monozygotic twins DNA Res **31**, 10.1093/dnares/dsae028
1121. Yamamura, Y., Fukami, M., Matsuyama, M., andSawada, H. (2024) A novel variant of IGSF1 in siblings with congenital central hypothyroidism whose diagnosis was prompted by school health checkups Clin Pediatr Endocrinol **33**, 17-22 10.1297/cpe.2023-0046
1122. Yamamura-Miyazaki, N., Sakakibara, N., Nozu, K., Shima, Y., Satomura, K., Yamamoto, S. *et al.* (2024) Identification of CUBN variants in triplets with a 20-year history of proteinuria CEN Case Rep 1-6 10.1007/s13730-024-00919-6
1123. Yamasaki, S., Shintani, T., Ando, T., Miyauchi, M., andYanamoto, S. (2024) Transformation of an odontogenic keratocyst into a solid variant of odontogenic keratocyst/keratoameloblastoma during long-term follow-up: A case report Mol Med Rep **29**, 10.3892/mmr.2024.13168
1124. Yamoto, K., Kato, F., Yamoto, M., Fukumoto, K., Shimizu, K., Saitsu, H. *et al.* (2024) TBX5 pathogenic variant in a patient with congenital heart defect and tracheal stenosis Congenit Anom (Kyoto) **64**, 23-27 10.1111/cga.12548
1125. Yaoita, H., Kawai, E., Takayama, J., Iwasawa, S., Saijo, N., Abiko, M. *et al.* (2024) Genetic etiology of truncus

arteriosus excluding 22q11.2 deletion syndrome and identification of c.1617del, a prevalent variant in TMEM260, in the Japanese population Journal of human genetics **69**, 177-183 10.1038/s10038-024-01223-y

1126. Yoneno, S., Yamamoto, K., Tabata, K., Shimizu-Motohashi, Y., Tomita, A., Hayashi, T. *et al.* (2024) A novel heterozygous TMEM63A variant in a familial case with early onset nystagmus, severe hypomyelination, and a favorable adult prognosis Journal of human genetics 1-5 10.1038/s10038-024-01268-z
1127. Zaitsu, S., Aoyagi, Y., Nishida, H., Nakamura, K., Yano, M., andKobayashi, E. (2024) Genetic Profiling of Sebaceous Carcinoma Arising from an Ovarian Mature Teratoma: A Case Report Int J Mol Sci **25**, 10.3390/ijms25126351
1128. Zhu, J., Zhao, Z., Li, S., Zhou, Y., Kong, L., Fu, X. *et al.* (2024) High-Resolution Haplotyping of the PAH Gene Enables Early Gestation Noninvasive Prenatal Diagnosis of Phenylketonuria and Evolution Analysis of Recurrent Pathogenic Variations Prenat Diagn **44**, 1198-1209 10.1002/pd.6645
1129. Zukeran, H., Akiba, K., Higuchi, S., Mori, J., Yorifuji, T., andHasegawa, Y. (2024) A case of diffuse congenital hyperinsulinism in which continuous glucose monitoring contributed to the choice of a treatment strategy following a subtotal pancreatectomy Clinical Pediatric Endocrinology **33**, 174-180 10.1297/cpe.33.2023-0086
1130. 石塚由美子, 江口英孝, 堀本義哉, 佐々木律子, 岡﨑康司, 新井正美 *et al.* (2024) 男性乳癌における生殖細胞系列病的バリアント探索の試み 遺伝性腫瘍 **24**, 128-135,
1131. 中村善胤, 佐藤秀則, 垣内謙祐, 宮野佑樹, 細川隆史, and 荒若繁樹 (2024) CLCN1 遺伝子に新規変異 p. F343C を同定した先天性ミオトニアの 1 家系 臨床神経学 **64**, 344-348,
1132. 田村友紀, 落合恵理子, 山崎一樹, and 大澤資樹 (2024) 日本人集団において個人を識別するのに必要な STR と SNP の最小座位数 日本法科学技術学会誌 **29**, 25-33,
1133. 田中智大, 村上智美, 江間達哉, 奥村良法, 清水健司, 村山圭 *et al.* (2024) Leigh 脳症様の経過を呈したモリブデン補酵素欠損症 A 型の 1 例 脳と発達 **56**, 443-445,
1134. Aiba, T., Ohno, S., Takegami, M., Kato, Y., Sakaguchi, H., Shimamoto, K. *et al.* (2025) Clinical Impact of Genetic Testing for Long QT Syndrome - Evidence From a Nationwide LQTS Registry in Japan Circ J **89**, 835-844 10.1253/circj.CJ-25-0105
1135. Akiba, K., Matsubara, K., Hattori, A., andFukami, M. (2025) Intragenic duplication of PHEX in a girl with X-linked hypophosphatemia: a case report with review of literature Endocr J **72**, 413-419 10.1507/endocrj.EJ24-0355
1136. Akimoto, T., Inaba, H., Ogishima, S., Kumada, K., Mitsuhashi, A., Miyashita, R. *et al.* (2025) Comparison of genotypes and phenotypes for von Willebrand factor gene variants using Japanese genome database Blood Vessels, Thrombosis & Hemostasis **2**, 100070 https://doi.org/10.1016/j.bvth.2025.100070
1137. Ando, M., Higuchi, Y., Yuan, J. H., Yoshimura, A., Yano, C., Hobara, T. *et al.* (2025) SOD1-related inherited peripheral neuropathies in a Japanese cohort: genetic variants and clinical insights J Neurol **272**, 191 10.1007/s00415-025-12925-4
1138. Asano, R., Okazawa, M., Ishibashi, T., Ding, X., Ohta-Ogo, K., Akaki, K. *et al.* (2025) A Novel Missense Variant of ZC3H12A in Pulmonary Arterial Hypertension Circ Rep **7**, 207-211 10.1253/circrep.CR-25-0007
1139. Bai, G. Y., Shan, K. S., Li, C. S., Wang, X. H., Feng, M. Y., andGao, Y. (2025) Gastric gastrointestinal stromal tumor in a patient with neurofibromatosis type I presenting with anemia: A case report World J Gastrointest Oncol **17**, 99304 10.4251/wjgo.v17.i3.99304
1140. Bayam, E., Tilly, P., Collins, S. C., Rivera Alvarez, J., Kannan, M., Tonneau, L. *et al.* (2025) Bi-allelic variants in WDR47 cause a complex neurodevelopmental syndrome EMBO Mol Med **17**, 129-168 10.1038/s44321-024-00178-z

1141. Danno, D., Tada, H., Oda, I., Kawashita, N., Hirano, M., Kitamura, S. *et al.* (2025) Expanding the Genetic and Clinical Spectrum of SCN1A-Related Hemiplegic Migraine: Analysis of Mutations in Japanese Int J Mol Sci **26**, 10.3390/ijms26041426

1142. Ebata, A., Takeichi, T., Nishida, K., Chretien, B., Miyazaki, A., Yoshikawa, T. *et al.* (2025) Estimating the proportions of allele frequencies for SERPINA12 pathogenic variants in Japanese patients with Nagashima-type palmoplantar keratosis/keratoderma Br J Dermatol **193**, 184-185 10.1093/bjd/ljaf111

1143. Fujii, K., Ueki, M., Morishita, M., Ikushima, H., Isago, H., Watanabe, K. *et al.* (2025) Clinical utility of comprehensive genomic profiling in non-small cell lung cancer: An analysis of a nation-wide database Lung Cancer **200**, 108099 10.1016/j.lungcan.2025.108099

1144. Fujita, A., Suenaga, Y., Takeshita, E., Takahashi, Y., Suzuki, Y., Ohori, S. *et al.* (2025) Triple mosaic variants of PURA in a patient with multiple congenital anomalies Journal of human genetics **70**, 227-230 10.1038/s10038-024-01315-9

1145. Fukushima, T., Nakamura, K., Terai, H., Ohgino, K., Kawano, R., Ishikawa, M. *et al.* (2025) Clinical Sequence Revealed the Prevalence and Biological Significance of Somatic Pathogenic Variants in Thoracic Cancer: Implications for Germline Status Clin Lung Cancer **26**, 407-419 e420 10.1016/j.cllc.2025.03.012

1146. Furutani, M., Kimura, T., Fukunaga, K., Suganuma, M., Takemura, M., Matsui, Y. *et al.* (2025) Identification of a Risk Allele at SLC41A3 and a Protective Allele HLA-DPB1*02:01 Associated with Sarcopenia in Japanese Gerontology **71**, 376-387 10.1159/000545298

1147. Ghasemi, M.-R., Mirahmadi, M., Bayat, H., Miryounesi, M., Mirfakhraie, R., Salehpour, S. *et al.* (2025) Unraveling and Expanding the Genotypic Spectrum of Kabuki Syndrome with Identification of de Novo Protein-Truncating Mutations in the KMT2D Gene: Insights into the Role of Premature Stop Codons in the Etiology of the Disorder OBM Genetics **09**, 294 10.21926/obm.genet.2502294

1148. Ghasemi, M.-R., Mirahmadi, M., Bayat, H., Nooshabadi, M. S., Jamshidi, S., Salehpour, S. *et al.* (2025) Exploring the Genetic Landscape of Knobloch Syndrome: Novel Variant Identification and Literature Review OBM Genetics **09**, 291 10.21926/obm.genet.2502291

1149. Goto, Y., Niihori, T., Mizuno, S., Okamoto, N., Ogata, T., Kurosawa, K. *et al.* (2025) Missense and truncated variants in ERF in individuals with a Noonan-like phenotype without craniosynostosis Sci Rep **15**, 15179 10.1038/s41598-025-89719-1

1150. Hadi, A. F., Arta, R. K., Kushima, I., Egawa, J., Watanabe, Y., Ozaki, N. *et al.* (2025) Association Analysis of Rare CNTN5 Variants With Autism Spectrum Disorder in a Japanese Population Neuropsychopharmacol Rep **45**, e12527 10.1002/npr2.12527

1151. Hasegawa, Y., Miura, S., Kagami, M., Dateki, S., andMiura, K. (2025) Diagnosis and Genetic Counseling Before and After the Birth of Children With Joubert Syndrome and Beckwith-Wiedemann Syndrome Cureus **17**, e80677 10.7759/cureus.80677

1152. Hasegawa, Y., Miura, S., Nagata, A., Yoshiura, K. I., andMiura, K. (2025) Repeated Pregnancies With Full Trisomy 21: A Case Report and Literature Review Cureus **17**, e83275 10.7759/cureus.83275

1153. Hashiba, T., Sugawara, Y., Hirakawa, Y., Sato, D., Inagi, R., andNangaku, M. (2025) Pathogenic variants prevalence patients with diabetic kidney disease in Japan: A descriptive study J Diabetes Investig **16**, 1263-1273 10.1111/jdi.70041

1154. Hida, T., Idogawa, M., Ishikawa, A., Okura, M., Sasaki, S., Tokino, T. *et al.* (2025) A case of familial progressive hyperpigmentation with or without hypopigmentation presenting with hypopigmented striae along the lines of

Blaschko J Dermatol **52**, 159-162 10.1111/1346-8138.17459

1155. Hijikata, M., Morimoto, K., Ito, M., Wakabayashi, K., Miyabayashi, A., andKeicho, N. (2025) Robust detection of pathogenic HYDIN variants that cause primary ciliary dyskinesia using RNA-seq of nasal mucosa J Med Genet **62**, 180-184 10.1136/jmg-2024-110400
1156. Hokmabadi, M., Asadifard, E., Arbabi-Bidgoli, S., Naghavi, A., andHasanzad, M. (2025) Analysis of CYP2D6 Gene Variant Frequencies in Iranian Population Iran J Med Sci **50**, 351-358 10.30476/ijms.2024.102645.3570
1157. Huong, Q. T. T., Truc, L. T. N., Ueda, H., Fukui, K., Higasa, K., Sato, Y. *et al.* (2025) Nerve Enlargement in Patients with INF2 Variants Causing Peripheral Neuropathy and Focal Segmental Glomerulosclerosis Biomedicines **13**, 127 10.3390/biomedicines13010127
1158. Iijima, H., Imai-Okazaki, A., Kishita, Y., Sugiura, A., Shimura, M., Murayama, K. *et al.* (2025) Role of BOLA3 in the mitochondrial Fe-S cluster clarified by metabolomic analysis Mol Genet Metab **145**, 109113 10.1016/j.ymgme.2025.109113
1159. Ito, M., Morimoto, K., Hijikata, M., Hasegawa, H., Wakabayashi, K., Miyabayashi, A. *et al.* (2025) Severe bronchiectasis and chronic rhinosinusitis due to homozygous WFDC2 Variants: The first three cases reported from Japan Respir Med Case Rep **55**, 102214 10.1016/j.rmcr.2025.102214
1160. Iyama, S., Chi, S., Idogawa, M., Ikezoe, T., Fukushima, K., Utsu, Y. *et al.* (2025) Prognostic impact of TET2 mutations in patients with acute myeloid leukemia: HM-SCREEN-Japan 01 and 02 study Ann Hematol **104**, 275-284 10.1007/s00277-025-06227-y
1161. Kaimori, J. Y., Matsuda, J., Jan, A., Kawano, Y., Kawaoka, T., Asahina, Y. *et al.* (2025) A Novel Heterozygous and Pathogenic Variant of the HNF1B Gene Associated with Autosomal Dominant Tubulointerstitial Kidney Disease with a Broad Spectrum of Extrarenal Phenotypes: A Case Report Intern Med 10.2169/internalmedicine.4548-24
1162. Kato, C., Morimoto, S., Takahashi, S., Namba, S., Wang, Q. S., Okada, Y. *et al.* (2025) Spinal cord motor neuron phenotypes and polygenic risk scores in sporadic amyotrophic lateral sclerosis: deciphering the disease pathology and therapeutic potential of ropinirole hydrochloride J Neurol Neurosurg Psychiatry **96**, 199-201 10.1136/jnnp-2024-333690
1163. Khosravi, S., Amini, E., Emamikhah, M., Alavi, A., Lang, A. E., andRohani, M. (2025) Motor Neuron Involvement in Two ATP13A2-Related Families: ALS And HSP-Like Phenotypes Mov Disord Clin Pract **12**, 852-857 10.1002/mdc3.70027
1164. Kojima, D., Shibata, M., Shikano, H., Maruo, Y., andFujii, H. (2025) Identification of a novel missense variant in the AVP gene in a Japanese pedigree with familial neurohypophyseal diabetes insipidus Clin Pediatr Endocrinol **34**, 77-82 10.1297/cpe.2024-0067
1165. Kokame, K. (2025) Estimating rare genetic disease prevalence: a challenging task Blood Adv **9**, 2968-2969 10.1182/bloodadvances.2024015546
1166. Kongphan, O., Lert-Itthiporn, W., Cha'on, U., Anutrakulchai, S., Nahok, K., Artkaew, N. *et al.* (2025) Alleles of CYP3A5 and their association with renal function in chronic kidney disease PeerJ **13**, e19424,
1167. Kozuma, A., Miyamoto-Mikami, E., Saito, M., Homma, H., Deguchi, M., Matsumoto, S. *et al.* (2025) Association of the GALNTL6 rs558129 polymorphism with muscle strength in Japanese athletes Biology of Sport **42**, 161-167 10.5114/biolsport.2025.147010
1168. Kubota, T., Nagata, M., Takagi, K., Ishihara, Y., Kojima, K., Uchikura, Y. *et al.* (2025) Hydrops fetalis due to loss of function of hNav1.4 channel via compound heterozygous variants Journal of human genetics **70**, 3-8

10.1038/s10038-024-01284-z

1169. Kuroda, Y., Nagai, K., Kawai, Y., Naruto, T., Saijou, H., Morikawa, S. *et al.* (2025) Genotype-phenotype correlations and phenotypic expansion in a case series of ReNU syndrome associated with RNU4-2 variants J Med Genet 10.1136/jmg-2024-110604

1170. Luo, Y., Funayama, M., Hatano, T., Li, Y., Yoshino, H., Yamashita, S. *et al.* (2025) Identification of DAGLB variants in Japanese early-onset Parkinson's disease J Neural Transm (Vienna) **132**, 1023-1030 10.1007/s00702-025-02926-y

1171. Maki, H., Sakai, N., Kataoka, M., Fujii, K., Kageyama, Y., Hayama, T. *et al.* (2025) Family study of bipolar disorder with comorbid anxiety disorder points to THSD7A with possible role of parent-of-origin effect PCN Rep **4**, e70071 10.1002/pcn5.70071

1172. Masunaga, Y., Kinjo, K., Murai, Y., Fujisawa, Y., Saitsu, H., andOgata, T. (2025) Insulin resistant diabetes mellitus in a girl with mild Rabson-Mendenhall syndrome: efficacy of sodium glucose co-transporter 2 inhibitor Diabetol Int **16**, 182-187 10.1007/s13340-024-00773-y

1173. Matsukawa, T., Sudo, A., Kakumoto, T., Hao, A., Kainaga, M., Chang, H. *et al.* (2025) In-frame deletion variant of ABCD1 in a sporadic case of adrenoleukodystrophy Hum Genome Var **12**, 5 10.1038/s41439-025-00309-z

1174. Minami, S., Kida, A., Inoue, S., Murakami, H., Morita, N., Takagi, A. *et al.* (2025) Auditory neuropathy spectrum disorder and related auditory features in patients with hearing loss associated with the MT-TS1 m.7471dup variant Mitochondrion **84**, 102056 10.1016/j.mito.2025.102056

1175. Mitsutake, A., Matsukawa, T., Hino, R., Fujino, G., Sakai, Y., Mitsui, J. *et al.* (2025) Clinical and Genetic Analyses of SPG7 in Japanese Patients with Undiagnosed Ataxia Intern Med **advpub**, 10.2169/internalmedicine.4767-24

1176. Mizobuchi, K., Hayashi, T., Kameya, S., Ohta, Y., Kuribayashi, K., andShinoda, K. (2025) Relatively preserved retinal function in RPE65-associated retinopathy: a case report Doc Ophthalmol **150**, 97-104 10.1007/s10633-025-10007-4

1177. Nakagawa, S., Sato, T., Ohashi, E., Kajita, M., Miya, F., Yamamoto, K. *et al.* (2025) An organoid library of human esophageal squamous cell carcinomas (ESCCs) uncovers the chemotherapy-resistant ESCC features Commun Biol **8**, 507 10.1038/s42003-025-07869-4

1178. Nakato, D., Morisada, N., Iwatani, S., Nishida, C., Watanabe, D., Yamada, M. *et al.* (2025) RHOA-associated disorder can be non-mosaic European Journal of Medical Genetics **75**, 105019 ARTN 105019 10.1016/j.ejmg.2025.105019

1179. Nakatochi, M., Kushima, I., Aleksic, B., Kimura, H., Kato, H., Inada, T. *et al.* (2025) Copy number variations in RNF216 and postsynaptic membrane-associated genes are associated with bipolar disorder: a case-control study in the Japanese population Psychiatry and clinical neurosciences **79**, 12-20 10.1111/pcn.13752

1180. Nishi, K., Matsumoto, H., Sunadome, H., Nagasaki, T., Oguma, T., Tashima, N. *et al.* (2025) IL1RL1 variant may affect the response to type 2 biologics in patients with severe asthma ERJ Open Res **11**, 10.1183/23120541.00448-2024

1181. Nishimura, N., Mizuguchi, T., Hamada, K., Yuge, K., Sakamoto, M., Tsuchida, N. *et al.* (2025) Hemizygous SMARCA1 variants cause X-linked intellectual disability Journal of human genetics **70**, 359-363 10.1038/s10038-025-01346-w

1182. Nishioka, K. (2025) Erratum to: Homozygous alpha-synuclein p.A53V in familial Parkinson's disease Neurobiol Aging **146**, 74-76 10.1016/j.neurobiolaging.2024.09.001

1183. Noda, Y., Kido, J., Sawada, T., Tanaka, K., Kumeda, K., Yoshida, S. *et al.* (2025) Newborn screening for

hypophosphatasia: development of a high-throughput tissue nonspecific alkaline phosphatase activity assay using dried blood spots JBMR Plus **9**, ziae172 10.1093/jbmrpl/ziae172

1184. Nomura, Y., Ishikawa, T., Ohno, S., Makita, N., Horie, M., Naruse, H. *et al.* (2025) Atypical Holt-Oram syndrome: Early-onset sick sinus syndrome in a Japanese family with a novel TBX5 mutation, Q469 Journal of Cardiology Cases **32**, 10-14,
1185. Nozaki, H., Sakakibara, N., Hanafusa, H., Inoki, Y., Tanaka, Y., Kitakado, H. *et al.* (2025) The first case of Al-Raqad syndrome in Japan is associated with a homozygous DCPS exonic variant resulting in aberrant splicing Brain Dev **47**, 104366 10.1016/j.braindev.2025.104366
1186. Okubo, S., Matsukawa, T., Kawamoto, N., Tsujita, M., Orimo, K., Naruse, H. *et al.* (2025) Elevated serum autotaxin levels and multiple system atrophy-like presentation in a patient with PLA2G6-associated neurodegeneration Journal of human genetics **70**, 381-384 10.1038/s10038-025-01342-0
1187. Orimo, K., Matsukawa, T., Mitsutake, A., Cho, T., Naruse, H., Sakiyama, Y. *et al.* (2025) Clinical, neuroimaging and genetic findings in the Japanese case series of CLCN2-related leukoencephalopathy J Neurol Sci **472**, 123486 10.1016/j.jns.2025.123486
1188. Osakada, Y., Yunoki, T., Matsuoka, C., Fukui, Y., Tsunoda, K., Deguchi, K. *et al.* (2025) Long-Term Follow-Up of a Patient With SPG11 Neurology and Clinical Neuroscience **13**, 198-200 10.1111/ncn3.12874
1189. Sakai, T., Furutani, M., Nakashima, M., Ishibashi, N., Maeda, J., Oguri, N. *et al.* (2025) Genome-Wide Association Study of Atrial Fibrillation Recurrence After Radiofrequency Catheter Ablation in a Japanese Population J Cardiovasc Electrophysiol **36**, 1494-1503 10.1111/jce.16674
1190. Sasaki, T., Hiraki, H., Yashima-Abo, A., Nagashima, H., Endo, F., Yaegashi, M. *et al.* (2025) Comprehensive Genome Profiling-Initiated Tumor-Informed Circulating Tumor DNA Monitoring for Patients With Advanced Cancer Cancer Sci **116**, 764-774 10.1111/cas.16446
1191. Sase, M., Sato, T., Sato, H., Miya, F., Zhang, S., Haeno, H. *et al.* (2025) Comparative analysis of tongue cancer organoids among patients identifies the heritable nature of minimal residual disease Dev Cell **60**, 396-413 e396 10.1016/j.devcel.2024.10.007
1192. Semba, R., Eguchi, H., Takatsu, M., Hashizume, T., Moteki, H., Maeno, K. *et al.* (2025) Two-hit events occurred independently in bilateral breast cancers in a germline double heterozygous carrier for BRCA1 and BRCA2 Breast Cancer 10.1007/s12282-025-01740-4
1193. Shimomura, M., Mizoguchi, Y., Kajihara, K., Sakura, F., Noma, K., Fujii, T. *et al.* (2025) The Co-Occurrence of Low-Frequency Pathogenic Variants in TBXA2R Exacerbating the Hemorrhagic Symptoms in Siblings with Hemophilia B Semin Thromb Hemost 10.1055/a-2563-9899
1194. Shimomura, R., Kihara, Y., Yanagishita, T., Ishiguro, K., Shichiji, M., Sato, T. *et al.* (2025) Challenges in genetic counseling for RYR1-related myopathies Brain Dev **47**, 104363 10.1016/j.braindev.2025.104363
1195. Shimura, K., Ichihashi, Y., Abe, K., Ishii, T., Hasegawa, T., andNarumi, S. (2025) Familial Non-Autoimmune Hyperthyroidism Caused by an Extracellular Domain Variant (p.Leu267Phe) of the TSH Receptor Horm Res Paediatr 1-9 10.1159/000544836
1196. Shirota, H., Miyake, A., Kawamura, M., Suzuki, S., Saito, K., Yasuda, J. *et al.* (2025) Regional Differences in the Frequency of BRCA1 and BRCA2 Variants in Northeastern Japan: A Cohort Study Cancer Med **14**, e70443 10.1002/cam4.70443
1197. Sonehara, K., Uwamino, Y., Saiki, R., Takeshita, M., Namba, S., Uno, S. *et al.* (2025) Germline variants and mosaic chromosomal alterations affect COVID-19 vaccine immunogenicity Cell Genom **5**, 100783

10.1016/j.xgen.2025.100783

1198. Sugiura, K., Kubo, T., Inoue, S., Nomura, S., Yamada, T., Tobita, T. *et al.* (2025) Unveiling Clinical and Genetic Distinctions in Pure-Apical Versus Distal-Dominant Apical Hypertrophic Cardiomyopathy J Am Heart Assoc **14**, e038208 10.1161/JAHA.124.038208

1199. Takeji, Y., Tada, H., Takamura, M., Tomura, A., andHarada-Shiba, M. (2025) Prevalence and Clinical Characteristics of Familial Hypercholesterolemia in Patients with Acute Coronary Syndrome according to the Current Japanese Guidelines: Insight from the EXPLORE-J study J Atheroscler Thromb **32**, 23-33 10.5551/jat.64972

1200. Tanikawa, W., Saitsu, H., Nakamura, Y., Shirafuta, Y., Fujisawa, Y., Fukami, M. *et al.* (2025) Homozygous FIGLA missense variant in two Japanese sisters with primary ovarian insufficiency: Case reports and literature review Reprod Med Biol **24**, e12635 10.1002/rmb2.12635

1201. Terayama, S., Kobayashi, M., Suemitsu, T., Samura, O., andOishi, K. (2025) Long-Term Outcomes of Early Enzyme Replacement Therapy With Asfotase Alfa in Perinatal Benign Hypophosphatasia: Amelioration of Bone Deformities in a Young Child Cureus **17**, e78473 10.7759/cureus.78473

1202. Tomomasa, D., Suzuki, T., Takeuchi, I., Goto, K., Hagiwara, S. I., Keino, D. *et al.* (2025) Successful Allogeneic Hematopoietic Cell Transplantation for Patients with IL10RA Deficiency in Japan Journal of Clinical Immunology **45**, 6 ARTN 6 10.1007/s10875-024-01795-6

1203. Torazawa, S., Miyawaki, S., Imai, H., Hongo, H., Ishigami, D., Shimizu, M. *et al.* (2025) Association of rare variants in RNF213 with severe progression of intracranial artery stenosis in quasi-moyamoya disease J Neurosurg **142**, 394-403 10.3171/2024.6.JNS24202

1204. Uehara, E., Akiba, K., Matsubara, K., Fukami, M., andTanase-Nakao, K. (2025) Amplicon-based targeted next-generation sequencing using dried blood spots for 46,XY differences/disorders of sex development: Aiming for diagnosis by minimally invasive testing Clin Pediatr Endocrinol **34**, 172-179 10.1297/cpe.2024-0081

1205. Uehara, S., Hirai, K., Shirai, T., Otaki, H., Akamatsu, T., andItoh, K. (2025) Vitamin D Receptor rs2228570 Gene Polymorphism Is Associated with Asthma Severity and Exacerbations Biol Pharm Bull **48**, 86-92 10.1248/bpb.b24-00684

1206. Wakasa, M., Nakagawa, C., Takamura, T. A., Fujibayashi, K., Akao, H., Kitayama, M. *et al.* (2025) A male patient with pseudoxanthoma elasticum caused by isodisomy of chromosome 16 containing a nonsense variant of the ABCC6 gene: A quarter-century treatment experience Atheroscler Plus **60**, 1-5 10.1016/j.athplu.2025.03.001

1207. Walsh, R., Mauleekoonphairoj, J., Mengarelli, I., Bosada, F. M., Verkerk, A. O., van Duijvenboden, K. *et al.* (2025) A Rare Noncoding Enhancer Variant in SCN5A Contributes to the High Prevalence of Brugada Syndrome in Thailand Circulation **151**, 31-44 10.1161/CIRCULATIONAHA.124.069041

1208. Watanabe, D., Hasebe, Y., Yagasaki, H., Nakato, D., Yamada, M., Suzuki, H. *et al.* (2025) Brain calcification in congenital heart defects and ectodermal dysplasia (CHDED) Eur J Med Genet **73**, 104992 10.1016/j.ejmg.2024.104992

1209. Watanabe, Y., Nishioka, M., Morikawa, R., Takano-Isozaki, S., Igeta, H., Mori, K. *et al.* (2025) Rare nonsynonymous germline and mosaic de novo variants in Japanese patients with schizophrenia Psychiatry and clinical neurosciences **79**, 37-44 10.1111/pcn.13758

1210. Wei, X., Otsuki, J., Fujii, M., Kodera, K., Yamada, A., Hayashi, N. *et al.* (2025) SURF1 p.D202H variant is not associated with Leigh syndrome but may contribute to impaired pregnancy outcomes Gene Reports **40**, 102240 10.1016/j.genrep.2025.102240

1211. Xu, X., Manabe, N., Ohno, S., Komatsu, S., Fujimura, T., andYamaguchi, Y. (2025) 3D structural insights into the effect of N-glycosylation in human chitotriosidase variant G102S Biochim Biophys Acta Gen Subj **1869**, 130730 10.1016/j.bbagen.2024.130730

1212. Yamaguchi, M., Akabane, S., Niitsu, H., Nakahara, H., Toshida, A., Mochizuki, T. *et al.* (2025) The usefulness of comprehensive genome profiling test in screening of Lynch syndrome independent of the conventional clinical screening or microsatellite instability tests Journal of human genetics 10.1038/s10038-025-01345-x

1213. Yamin, M. B., Hatta, F. H. M., Aazmi, M. S., Syafruddin, S. E., Lee, M. Y. S., Ramzi, N. H. *et al.* (2025) UNRAVELLING PUTATIVE ETHNIC-SPECIFIC OBESITY LOCI IN MALAYSIAN YOUTHS: A PRELIMINARY GENOME-WIDE ASSOCIATION STUDY The Southeast Asian Journal of Tropical Medicine and Public Health **56**, 253-290,

1214. Yang, W., Jin, S., Jiang, J., Ji, W. J., andHe, Q. (2025) A Novel Missense Mutation at EDA2R Gene Identified in a Case Study Associated with Hypohidrotic Ectodermal Dysplasia Regenerative Medicine and Dentistry **2**, 2 10.53941/rmd.2025.100002

1215. Yata, T., Sato, G., Ogawa, K., Naito, T., Sonehara, K., Saiki, R. *et al.* (2025) Contribution of germline and somatic mutations to risk of neuromyelitis optica spectrum disorder Cell Genom **5**, 100776 10.1016/j.xgen.2025.100776

1216. Yazaki, S., Hori, M., Aiba, H., Momozawa, Y., Yoshida, M., Shiino, S. *et al.* (2025) Impact of germline variants on breast and ovarian cancer risk in Japanese women: an original cohort study and meta-analysis Ebiomedicine **116**, 105758 10.1016/j.ebiom.2025.105758

1217. Ye, S., Eura, Y., Matsumoto, M., andKokame, K. (2025) Genetic analysis using long-read sequencing to overcome the difficulties in VWF gene Res Pract Thromb Haemost **9**, 102888 10.1016/j.rpth.2025.102888

1218. 河合泰寛, 竹下絵里, 須貝研司, 山本薫, 馬場信平, 住友典子 *et al.* (2025) 緩徐進行性に痙性四肢麻痺を来した ADAR バリアントによる遅発型の Aicardi-Goutières 症候群 脳と発達 **57**, 34-38,

1219. 宮田世羽, 小須賀基通, 加藤光広, 渡邊一樹, 中島光子, 才津浩智 *et al.* (2025) WDR81 遺伝子の新規複合ヘテロ接合性バリアントを認めた小滑脳症の 1 例 脳と発達 **57**, 59-61,

1220. 宮冬樹 (2025) バリアントデータを用いた次世代シーケンス手法の分析的妥当性の検証 日本遺伝子診療学会誌 **2**, 2-6 10.69246/jsgdt.2.1_2

1221. 三浦崇徳, 水本洋, 阿水利沙, 園田真理, 遠藤耕介, and 要匡 (2025) 迅速網羅的遺伝子解析により診断した ACTG2 visceral myopathy の 1 例 日本周産期・新生児医学会雑誌 **61**, 180-184 10.34456/jjspnm.61.1_180

1222. 山田莉沙, 谷村知繁, 戸川泰子, 杉本真里, 戸川貴夫, 杉浦崇浩 *et al.* (2025) P3H1 遺伝子に新規の病原性変異を認めた骨形成不全症 Sillence III 型の 1 例 日本周産期・新生児医学会雑誌 **61**, 223-228 10.34456/jjspnm.61.1_223

1223. 小田いつき, 團野大介, 多田陽香, 池川敦子, 中村朱美, 北村重和 *et al.* (2025) 片麻痺性片頭痛(HM)におけるチャネル病遺伝子のバリアント解析 日本遺伝子診療学会誌 **2**, 7-11 10.69246/jsgdt.2.1_7

1224. 馬場悠輔, 前田明子, 村松恭祐, 富永健太, 上田洲裕, 小巻奨吾 *et al.* (2025) 幼児期に側弯と呼吸不全を来し成人移行に際する再評価により SELENON 関連ミオパチーと診断した 33 歳女性例 臨床神経学 **65**, 139-145 10.5692/clinicalneurol.cn-002046

1225. 木村聡一郎, 加藤創生, 星野良朋, 渡部創, 日髙尚子, 岡田慶太 *et al.* (2025) 骨形成不全症 35 例における COL1A1, COL1A2 バリアントと臨床像の検討 日本内分泌学会雑誌 **101**, 36-38,

標準ゲノム情報（1226-1232）

1226. Okayama, T., Hashiguchi, Y., Kikuyama, H., Yoneda, H., andKanazawa, T. (2018) Next-generation sequencing analysis of multiplex families with atypical psychosis Transl Psychiatry **8**, 221 10.1038/s41398-018-0272-x

1227. Llamas, B., Narzisi, G., Schneider, V., Audano, P. A., Biederstedt, E., Blauvelt, L. *et al.* (2019) A strategy for

building and using a human reference pangenome F1000Res **8**, 1751 10.12688/f1000research.19630.2

1228. Ishikura, S., Nakabayashi, K., Nagai, M., Tsunoda, T., andShirasawa, S. (2020) ZFAT binds to centromeres to control noncoding RNA transcription through the KAT2B-H4K8ac-BRD4 axis Nucleic Acids Res **48**, 10848-10866 10.1093/nar/gkaa815

1229. Kim, S. J., Sota, Y., Naoi, Y., Honma, K., Kagara, N., Miyake, T. *et al.* (2021) Determining homologous recombination deficiency scores with whole exome sequencing and their association with responses to neoadjuvant chemotherapy in breast cancer Transl Oncol **14**, 100986 10.1016/j.tranon.2020.100986

1230. Oku, H., Watanabe, A., Tokuda, Y., Nakayama, T., Tashiro, K., Sotozono, C. *et al.* (2022) Efficient DNA/RNA extraction from tarsal plates by SK mill, a freeze-crush apparatus J Biochem **172**, 385-394 10.1093/jb/mvac078

1231. Lopes, M., Louzada, S., Ferreira, D., Verissimo, G., Eleuterio, D., Gama-Carvalho, M. *et al.* (2023) Human Satellite 1A analysis provides evidence of pericentromeric transcription BMC Biol **21**, 28 10.1186/s12915-023-01521-5

1232. Ishikawa, R., Yamada, H., Saitsu, H., Miyazaki, R., Takahashi, J., Takinami, R. *et al.* (2025) Immunohistochemical and molecular evolutionary features of jejunoileal adenocarcinoma unveiled through comparative analysis with colorectal adenocarcinoma Neoplasia **66**, 101180 10.1016/j.neo.2025.101180

全ゲノム情報（1233-1239）

1233. Lin, M. R., Wu, I. W., Chou, W. H., Lin, Y. F., Hung, K. Y., Kojima, K. *et al.* (2025) Whole exome sequencing and polygenic risk assessment for kidney functions and clinical management in both hospital-based cohort and population-based Asian cohorts J Biomed Sci **32**, 72 10.1186/s12929-025-01168-0

1234. Sun, Y., Irwanto, A., Toyo-Oka, L., Hong, M., Liu, H., Andiappan, A. K. *et al.* (2016) Fine-mapping analysis revealed complex pleiotropic effect and tissue-specific regulatory mechanism of TNFSF15 in primary biliary cholangitis, Crohn's disease and leprosy Sci Rep **6**, 31429 10.1038/srep31429

1235. Hitomi, Y., Kojima, K., Kawashima, M., Kawai, Y., Nishida, N., Aiba, Y. *et al.* (2017) Identification of the functional variant driving ORMDL3 and GSDMB expression in human chromosome 17q12-21 in primary biliary cholangitis Sci Rep **7**, 2904 10.1038/s41598-017-03067-3

1236. Hitomi, Y., Nakatani, K., Kojima, K., Nishida, N., Kawai, Y., Kawashima, M. *et al.* (2019) NFKB1 and MANBA Confer Disease Susceptibility to Primary Biliary Cholangitis via Independent Putative Primary Functional Variants Cell Mol Gastroenterol Hepatol **7**, 515-532 10.1016/j.jcmgh.2018.11.006

1237. Hitomi, Y., Ueno, K., Kawai, Y., Nishida, N., Kojima, K., Kawashima, M. *et al.* (2019) POGLUT1, the putative effector gene driven by rs2293370 in primary biliary cholangitis susceptibility locus chromosome 3q13.33 Sci Rep **9**, 102 10.1038/s41598-018-36490-1

1238. Hitomi, Y., Aiba, Y., Kawai, Y., Kojima, K., Ueno, K., Nishida, N. *et al.* (2021) rs1944919 on chromosome 11q23.1 and its effector genes COLCA1/COLCA2 confer susceptibility to primary biliary cholangitis Sci Rep **11**, 4557 10.1038/s41598-021-84042-x

1239. Noguchi, E., Morii, W., Kitazawa, H., Hirota, T., Sonehara, K., Masuko, H. *et al.* (2025) A genome-wide meta-analysis reveals shared and population-specific variants for allergic sensitization J Allergy Clin Immunol **155**, 1321-1332 10.1016/j.jaci.2024.11.033

Japonica Array（1240-1332）

1240. Kuno, M., Okayama, Y., Yasui, Y., Shibata, W., Moriguchi, M., Nakaya, Y. *et al.* (2025) Effects of routes,

concentrations and genetic polymorphisms on posaconazole-tacrolimus interactions in allogeneic haematopoietic cell transplantation Br J Clin Pharmacol **91**, 3288-3296 10.1002/bcp.70264

1241. Mori, T., Ueno, K., Nagasaki, M., Matsuda, K., andBioBank Japan, P. (2025) A POLR1D-regulating single-nucleotide polymorphism as a predictive marker candidate for platinum-based chemotherapy in gastrointestinal cancers Ther Adv Med Oncol **17**, 17588359251355079 10.1177/17588359251355079
1242. Takahashi, N., Nishimura, T., Okumura, A., Iwabuchi, T., Harada, T., Rahman, M. S. *et al.* (2025) Maternal stress, cord blood zinc and attention deficit hyperactivity disorder Npj Ment Health Res **4**, 36 10.1038/s44184-025-00149-3
1243. Tanegashima, T., Shiota, M., Akamatsu, S., Miyake, H., Takahashi, M., Oya, M. *et al.* (2025) GWAS Identifies SNPs Associated With Severe Adverse Events and Efficacy in Advanced Renal Cell Carcinoma Treated With Nivolumab Cancer Sci 10.1111/cas.70204
1244. Liu, T. C., Naito, T., Liu, Z., VanDussen, K. L., Haritunians, T., Li, D. *et al.* (2017) LRRK2 but not ATG16L1 is associated with Paneth cell defect in Japanese Crohn's disease patients JCI Insight **2**, e91917 10.1172/jci.insight.91917
1245. Shiga, Y., Nishiguchi, K. M., Kawai, Y., Kojima, K., Sato, K., Fujita, K. *et al.* (2017) Genetic analysis of Japanese primary open-angle glaucoma patients and clinical characterization of risk alleles near CDKN2B-AS1, SIX6 and GAS7 PLoS One **12**, e0186678 10.1371/journal.pone.0186678
1246. Ueta, M., Sawai, H., Shingaki, R., Kawai, Y., Sotozono, C., Kojima, K. *et al.* (2017) Genome-wide association study using the ethnicity-specific Japonica array: identification of new susceptibility loci for cold medicine-related Stevens-Johnson syndrome with severe ocular complications Journal of human genetics **62**, 485-489 10.1038/jhg.2016.160
1247. Latt, K. Z., Honda, K., Thiri, M., Hitomi, Y., Omae, Y., Sawai, H. *et al.* (2018) Identification of a two-SNP PLA2R1 Haplotype and HLA-DRB1 Alleles as Primary Risk Associations in Idiopathic Membranous Nephropathy Sci Rep **8**, 15576 10.1038/s41598-018-33612-7
1248. Nakamura, T., Kawarabayashi, T., Seino, Y., Hirohata, M., Nakahata, N., Narita, S. *et al.* (2018) Aging and APOE-epsilon4 are determinative factors of plasma Abeta42 levels Ann Clin Transl Neurol **5**, 1184-1191 10.1002/acn3.635
1249. Okamoto-Uchida, Y., Nakamura, R., Matsunaga, K., Aihara, M., andSaito, Y. (2018) Specific association of the rs6500265 and rs9933632 single-nucleotide polymorphisms in Japanese patients with antipyretic analgesic-related Stevens-Johnson syndrome and toxic epidermal necrolysis with severe ocular involvements Pharmacogenet Genomics **28**, 95-98 10.1097/FPC.0000000000000324
1250. Ueta, M. (2018) Results of Detailed Investigations Into Stevens-Johnson Syndrome With Severe Ocular Complications Invest Ophthalmol Vis Sci **59**, DES183-DES191 10.1167/iovs.17-23537
1251. Choi, D., Tsuchiya, K. J., andTakei, N. (2019) Interaction effect of oxytocin receptor (OXTR) rs53576 genotype and maternal postpartum depression on child behavioural problems Sci Rep **9**, 7685 10.1038/s41598-019-44175-6
1252. Moon, S., Kim, Y. J., Han, S., Hwang, M. Y., Shin, D. M., Park, M. Y. *et al.* (2019) The Korea Biobank Array: Design and Identification of Coding Variants Associated with Blood Biochemical Traits Sci Rep **9**, 1382 10.1038/s41598-018-37832-9
1253. Shigemizu, D., Akiyama, S., Asanomi, Y., Boroevich, K. A., Sharma, A., Tsunoda, T. *et al.* (2019) A comparison of machine learning classifiers for dementia with Lewy bodies using miRNA expression data BMC Med Genomics

12, 150 10.1186/s12920-019-0607-3

1254. Kakuta, Y., Ichikawa, R., Fuyuno, Y., Hirano, A., Umeno, J., Torisu, T. *et al.* (2020) An Integrated Genomic and Transcriptomic Analysis Reveals Candidates of Susceptibility Genes for Crohn's Disease in Japanese Populations Sci Rep **10**, 10236 10.1038/s41598-020-66951-5
1255. Kim, Y. W., Kim, Y. J., Cheong, H. S., Shiga, Y., Hashimoto, K., Song, Y. J. *et al.* (2020) Exploring the Novel Susceptibility Gene Variants for Primary Open-Angle Glaucoma in East Asian Cohorts: The GLAU-GENDISK Study Sci Rep **10**, 221 10.1038/s41598-019-57066-7
1256. Nakayama, A., Nakatochi, M., Kawamura, Y., Yamamoto, K., Nakaoka, H., Shimizu, S. *et al.* (2020) Subtype-specific gout susceptibility loci and enrichment of selection pressure on ABCG2 and ALDH2 identified by subtype genome-wide meta-analyses of clinically defined gout patients Ann Rheum Dis **79**, 657-665 10.1136/annrheumdis-2019-216644
1257. Nishiya, Y., Daimon, M., Mizushiri, S., Murakami, H., Tanabe, J., Matsuhashi, Y. *et al.* (2020) Nutrient consumption-dependent association of a glucagon-like peptide-1 receptor gene polymorphism with insulin secretion Sci Rep **10**, 16382 10.1038/s41598-020-71853-7
1258. Okamoto, D., Kawai, Y., Kakuta, Y., Naito, T., Torisu, T., Hirano, A. *et al.* (2020) Genetic Analysis of Ulcerative Colitis in Japanese Individuals Using Population-specific SNP Array Inflamm Bowel Dis **26**, 1177-1187 10.1093/ibd/izaa033
1259. Shigemizu, D., Akiyama, S., Higaki, S., Sugimoto, T., Sakurai, T., Boroevich, K. A. *et al.* (2020) Prognosis prediction model for conversion from mild cognitive impairment to Alzheimer's disease created by integrative analysis of multi-omics data Alzheimers Res Ther **12**, 145 10.1186/s13195-020-00716-0
1260. Shiota, M., Fujimoto, N., Yamamoto, Y., Takeuchi, A., Tatsugami, K., Uchiumi, T. *et al.* (2020) Genome-wide association study of genetic variations associated with treatment failure after intravesical bacillus Calmette-Guerin therapy for non-muscle invasive bladder cancer Cancer Immunol Immunother **69**, 1155-1163 10.1007/s00262-020-02533-8
1261. Takahashi, N., Harada, T., Nishimura, T., Okumura, A., Choi, D., Iwabuchi, T. *et al.* (2020) Association of Genetic Risks With Autism Spectrum Disorder and Early Neurodevelopmental Delays Among Children Without Intellectual Disability JAMA Netw Open **3**, e1921644 10.1001/jamanetworkopen.2019.21644
1262. Takahashi, N., Nishimura, T., Harada, T., Okumura, A., Choi, D., Iwabuchi, T. *et al.* (2020) Polygenic risk score analysis revealed shared genetic background in attention deficit hyperactivity disorder and narcolepsy Transl Psychiatry **10**, 284 10.1038/s41398-020-00971-7
1263. Ueno, K., Aiba, Y., Hitomi, Y., Shimoda, S., Nakamura, H., Gervais, O. *et al.* (2020) Integrated GWAS and mRNA Microarray Analysis Identified IFNG and CD40L as the Central Upstream Regulators in Primary Biliary Cholangitis Hepatol Commun **4**, 724-738 10.1002/hep4.1497
1264. Yoshida, K., Yokota, K., Kutsuwada, Y., Nakayama, K., Watanabe, K., Matsumoto, A. *et al.* (2020) Genome-Wide Association Study of Lean Nonalcoholic Fatty Liver Disease Suggests Human Leukocyte Antigen as a Novel Candidate Locus Hepatol Commun **4**, 1124-1135 10.1002/hep4.1529
1265. Akiyama, S., Higaki, S., Ochiya, T., Ozaki, K., Niida, S., andShigemizu, D. (2021) JAMIR-eQTL: Japanese genome-wide identification of microRNA expression quantitative trait loci across dementia types Database (Oxford) **2021**, 10.1093/database/baab072
1266. Dang, J. T., Dang, T. T., Wine, E., Dicken, B., Madsen, K., andLaffin, M. (2021) The Genetics of Postoperative Recurrence in Crohn Disease: A Systematic Review, Meta-analysis, and Framework for Future Work Crohns

Colitis 360 **3**, otaa094 10.1093/crocol/otaa094

1267. Hishinuma, K., Moroi, R., Okamoto, D., Shimoyama, Y., Kuroha, M., Shiga, H. *et al.* (2021) Analysis of the Long-Term Prognosis in Japanese Patients with Ulcerative Colitis Treated with New Therapeutic Agents and the Correlation between Prognosis and Disease Susceptibility Loci Inflammatory Intestinal Diseases **6**, 154-164,
1268. Jinam, T., Kawai, Y., Kamatani, Y., Sonoda, S., Makisumi, K., Sameshima, H. *et al.* (2021) Genome-wide SNP data of Izumo and Makurazaki populations support inner-dual structure model for origin of Yamato people Journal of human genetics **66**, 681-687 10.1038/s10038-020-00898-3
1269. Nagakawa, S., Shiota, M., Fujimoto, N., Yamamoto, Y., Blas, L., Tsukahara, S. *et al.* (2021) The impact of single-nucleotide polymorphisms on intravesical recurrence after bacillus Calmette-Guerin therapy for non-muscle invasive bladder cancer in a genome-wide association study Urol Oncol **39**, 733 e717-733 e724 10.1016/j.urolonc.2021.05.034
1270. Nomura, A., Sato, T., Tada, H., Kannon, T., Hosomichi, K., Tsujiguchi, H. *et al.* (2021) Polygenic risk scores for low-density lipoprotein cholesterol and familial hypercholesterolemia Journal of human genetics **66**, 1079-1087 10.1038/s10038-021-00929-7
1271. Rahman, M. S., Takahashi, N., Iwabuchi, T., Nishimura, T., Harada, T., Okumura, A. *et al.* (2021) Elevated risk of attention deficit hyperactivity disorder (ADHD) in Japanese children with higher genetic susceptibility to ADHD with a birth weight under 2000 g BMC Med **19**, 229 10.1186/s12916-021-02093-3
1272. Shigemizu, D., Mitsumori, R., Akiyama, S., Miyashita, A., Morizono, T., Higaki, S. *et al.* (2021) Ethnic and trans-ethnic genome-wide association studies identify new loci influencing Japanese Alzheimer's disease risk Transl Psychiatry **11**, 151 10.1038/s41398-021-01272-3
1273. Takahashi, N., Nishimura, T., Harada, T., Okumura, A., Iwabuchi, T., Rahman, M. S. *et al.* (2021) Association Between Genetic Risks for Obesity and Working Memory in Children Front Neurosci **15**, 749230 10.3389/fnins.2021.749230
1274. Yamada, Y., Nakamura, H., Tsujiguchi, H., Hara, A., Miyagi, S., Kannon, T. *et al.* (2021) Relationships among the β3-adrenargic receptor gene Trp64Arg polymorphism, hypertension, and insulin resistance in a Japanese population PLoS One **16**, e0255444 10.1371/journal.pone.0255444
1275. Hara, A., Nguyen, P. M., Tsujiguchi, H., Nakamura, M., Yamada, Y., Suzuki, K. *et al.* (2022) Effect of beta3-adrenergic receptor gene polymorphism and lifestyle on overweight Japanese rural residents: A cross-sectional study Obes Sci Pract **8**, 199-207 10.1002/osp4.560
1276. Hasegawa, T., Kakuta, M., Yamaguchi, R., Sato, N., Mikami, T., Murashita, K. *et al.* (2022) Impact of salivary and pancreatic amylase gene copy numbers on diabetes, obesity, and functional profiles of microbiome in Northern Japanese population Sci Rep **12**, 7628 10.1038/s41598-022-11730-7
1277. Kakuta, Y., Iwaki, H., Umeno, J., Kawai, Y., Kawahara, M., Takagawa, T. *et al.* (2022) Crohn's Disease and Early Exposure to Thiopurines are Independent Risk Factors for Mosaic Chromosomal Alterations in Patients with Inflammatory Bowel Diseases J Crohns Colitis **16**, 643-655 10.1093/ecco-jcc/jjab199
1278. Kasai, M., Omae, Y., Kawai, Y., Shibata, A., Hoshino, A., Mizuguchi, M. *et al.* (2022) GWAS identifies candidate susceptibility loci and microRNA biomarkers for acute encephalopathy with biphasic seizures and late reduced diffusion Sci Rep **12**, 1332 10.1038/s41598-021-04576-y
1279. Kobayashi, M., Fujiyama, N., Tanegashima, T., Narita, S., Yamamoto, Y., Fujimoto, N. *et al.* (2022) Effect of HLA genotype on intravesical recurrence after bacillus Calmette-Guerin therapy for non-muscle-invasive bladder cancer Cancer Immunol Immunother **71**, 727-736 10.1007/s00262-021-03032-0

1280. Kress, S., Hara, A., Wigmann, C., Sato, T., Suzuki, K., Pham, K. O. *et al.* (2022) The Role of Polygenic Susceptibility on Air Pollution-Associated Asthma between German and Japanese Elderly Women Int J Environ Res Public Health **19**, 9869 10.3390/ijerph19169869
1281. Mishra, A., Malik, R., Hachiya, T., Jurgenson, T., Namba, S., Posner, D. C. *et al.* (2022) Stroke genetics informs drug discovery and risk prediction across ancestries Nature **611**, 115-123 10.1038/s41586-022-05165-3
1282. Mitamura, T., Zhai, T., Hatanaka, K. C., Hatanaka, Y., Amano, T., Wang, L. *et al.* (2022) Germline PRDM1 Variant rs2185379 in Long-Term Recurrence-Free Survivors of Advanced Ovarian Cancer Pharmgenomics Pers Med **15**, 977-984 10.2147/PGPM.S387120
1283. Nguyen, D. T., Tran, T. T. H., Tran, M. H., Tran, K., Pham, D., Duong, N. T. *et al.* (2022) A comprehensive evaluation of polygenic score and genotype imputation performances of human SNP arrays in diverse populations Sci Rep **12**, 17556 10.1038/s41598-022-22215-y
1284. Pham, K. O., Hara, A., Tsujiguchi, H., Suzuki, K., Suzuki, F., Miyagi, S. *et al.* (2022) Association between Vitamin Intake and Chronic Kidney Disease According to a Variant Located Upstream of the PTGS1 Gene: A Cross-Sectional Analysis of Shika Study Nutrients **14**, 2082 10.3390/nu14102082
1285. Semenova, E. A., Zempo, H., Miyamoto-Mikami, E., Kumagai, H., Larin, A. K., Sultanov, R. I. *et al.* (2022) Genome-Wide Association Study Identifies CDKN1A as a Novel Locus Associated with Muscle Fiber Composition Cells **11**, 10.3390/cells11233910
1286. Suzuki, K., Kakuta, Y., Naito, T., Takagawa, T., Hanai, H., Araki, H. *et al.* (2022) Genetic Background of Mesalamine-induced Fever and Diarrhea in Japanese Patients with Inflammatory Bowel Disease Inflamm Bowel Dis **28**, 21-31 10.1093/ibd/izab004
1287. Suzuki, K., Tsujiguchi, H., Hara, A., Pham, O. K., Miyagi, S., Nguyen, T. T. T. *et al.* (2022) Association Between Serum 25-Hydroxyvitamin D Concentrations, CDX2 Polymorphism in Promoter Region of Vitamin D Receptor Gene, and Chronic Pain in Rural Japanese Residents J Pain Res **15**, 1475-1485 10.2147/JPR.S356630
1288. Toyoda, Y., Nakayama, A., Nakatochi, M., Kawamura, Y., Nakaoka, H., Yamamoto, K. *et al.* (2022) Genome-wide meta-analysis between renal overload type and renal underexcretion type of clinically defined gout in Japanese populations Mol Genet Metab **136**, 186-189 10.1016/j.ymgme.2022.01.100
1289. Tsuboi, H., Matsunaga, M., Tsujiguchi, H., Kannon, T., Hosomichi, K., Sato, T. *et al.* (2022) Elevated ratio of serum anandamide to arachidonic acid intake in community-dwelling women with high depressive symptoms Neuro Endocrinol Lett **43**, 145-153, https://www.ncbi.nlm.nih.gov/pubmed/36179725
1290. Choi, J., Kim, S., Kim, J., Son, H. Y., Yoo, S. K., Kim, C. U. *et al.* (2023) A whole-genome reference panel of 14,393 individuals for East Asian populations accelerates discovery of rare functional variants Sci Adv **9**, eadg6319 10.1126/sciadv.adg6319
1291. Deguchi, M., Homma, H., de Almeida, K. Y., Kozuma, A., Saito, M., Tsuchiya, Y. *et al.* (2023) Association of MMP3 gene polymorphism and sex on recovery of muscle strength after eccentric exercise J Appl Physiol (1985) **135**, 527-533 10.1152/japplphysiol.00333.2023
1292. Khor, S. S., Ueno, K., Nishida, N., Kawashima, M., Kawai, Y., Aiba, Y. *et al.* (2023) Novel HLA allele associations with susceptibility, staging, symptomatic state, autoimmune hepatitis and hepatocellular carcinoma events for primary biliary cholangitis in the Japanese population Front Immunol **14**, 1151502 10.3389/fimmu.2023.1151502
1293. Liu, Z., Liu, R., Gao, H., Jung, S., Gao, X., Sun, R. *et al.* (2023) Genetic architecture of the inflammatory bowel diseases across East Asian and European ancestries Nature genetics **55**, 796-806 10.1038/s41588-023-01384-

0

1294. Matsunaga, M., Ohtsubo, Y., Masuda, T., Noguchi, Y., Yamasue, H., andIshii, K. (2023) Exploratory research on genetic polymorphisms associated with positive empathy and trait forgivingness among the Japanese Neuro Endocrinol Lett **44**, 506-516, https://www.ncbi.nlm.nih.gov/pubmed/38131174

1295. Mori, Y., Ueno, K., Chiba, D., Hashimoto, K., Kawai, Y., Baba, K. *et al.* (2023) Genome-Wide Association Study and Transcriptome of Japanese Patients with Developmental Dysplasia of the Hip Demonstrates an Association with the Ferroptosis Signaling Pathway Int J Mol Sci **24**, 5019 10.3390/ijms24055019

1296. Nakamura, T., Kawarabayashi, T., Nakahata, N., Itoh, K., Ihara, K., Nakaji, S. *et al.* (2023) Annual stability of the plasma Ass40/42 ratio and associated factors Ann Clin Transl Neurol **10**, 879-891 10.1002/acn3.51770

1297. Nayema, Z., Sato, T., Kannon, T., Tsujiguchi, H., Hosomichi, K., Nakamura, H. *et al.* (2023) Genetic factors associated with serum amylase in a Japanese population: combined analysis of copy-number and single-nucleotide variants Journal of human genetics **68**, 313-319 10.1038/s10038-022-01111-3

1298. Sakashita, T., Nakamura, Y., Sutoh, Y., Shimizu, A., Hachiya, T., Otsuka-Yamasaki, Y. *et al.* (2023) Comparison of the loci associated with HbA1c and blood glucose levels identified by a genome-wide association study in the Japanese population Diabetol Int **14**, 188-198 10.1007/s13340-023-00618-0

1299. Shigemizu, D., Akiyama, S., Suganuma, M., Furutani, M., Yamakawa, A., Nakano, Y. *et al.* (2023) Classification and deep-learning-based prediction of Alzheimer disease subtypes by using genomic data Transl Psychiatry **13**, 232 10.1038/s41398-023-02531-1

1300. Shimoda, F., Naito, T., Kakuta, Y., Kawai, Y., Tokunaga, K., Consortium, N. C. W. *et al.* (2023) HLA-DQA1*05 and upstream variants of PPARGC1B are associated with infliximab persistence in Japanese Crohn's disease patients Pharmacogenomics J **23**, 141-148 10.1038/s41397-023-00312-z

1301. Shiota, M., Miyake, H., Takahashi, M., Oya, M., Tsuchiya, N., Masumori, N. *et al.* (2023) Effect of genetic polymorphisms on outcomes following nivolumab for advanced renal cell carcinoma in the SNiP-RCC trial Cancer Immunol Immunother **72**, 1903-1915 10.1007/s00262-023-03367-w

1302. Shiota, M., Tatarano, S., Kamoto, T., Matsuyama, H., Sakai, H., Igawa, T. *et al.* (2023) Genome-wide association studies in advanced prostate cancer: KYUCOG-1401-A study Endocr Relat Cancer **30**, 10.1530/ERC-23-0044

1303. Takahashi, N., Nishimura, T., Harada, T., Okumura, A., Iwabuchi, T., Rahman, M. S. *et al.* (2023) Interaction of genetic liability for attention deficit hyperactivity disorder (ADHD) and perinatal inflammation contributes to ADHD symptoms in children Brain Behav Immun Health **30**, 100630 10.1016/j.bbih.2023.100630

1304. Yamagami, K., Tada, H., Sato, T., Nomura, A., Sakata, K., Usui, S. *et al.* (2023) Genetic Characteristics Contributing to Lipoprotein(a) Levels in the Japanese Population Journal of Coronary Artery Disease **29**, 104-108 10.7793/jcad.29.23-00015

1305. Yamamoto, R., Takeshita, Y., Tsujiguchi, H., Kannon, T., Sato, T., Hosomichi, K. *et al.* (2023) Nutrigenetic Interaction Between Apolipoprotein C3 Polymorphism and Fat Intake in People with Nonalcoholic Fatty Liver Disease Curr Dev Nutr **7**, 100051 10.1016/j.cdnut.2023.100051

1306. Yasukochi, Y., Sera, T., Kohno, T., Nakashima, Y., Uesugi, M., andKudo, S. (2023) Cold-induced vasodilation response in a Japanese cohort: insights from cold-water immersion and genome-wide association studies J Physiol Anthropol **42**, 2 10.1186/s40101-023-00319-2

1307. Yokoyama, S., Nakagawa, J., Kudo, M., Aiuchi, N., Seito, T., Isida, M. *et al.* (2023) Impact of solute carrier transporter gene polymorphisms on serum creatinine concentrations in healthy volunteers Pharmacol Res Perspect **11**, e01048 10.1002/prp2.1048

1308. Akiyama, M., Tamiya, G., Fujiwara, K., Shiga, Y., Yokoyama, Y., Hashimoto, K. *et al.* (2024) Genetic Risk Stratification of Primary Open-Angle Glaucoma in Japanese Individuals Ophthalmology **131**, 1271-1280 10.1016/j.ophtha.2024.05.026

1309. Deguchi, M., Homma, H., de Almeida, K. Y., Kozuma, A., Saito, M., Tsuchiya, Y. *et al.* (2024) Effect of the ACTN3 R577X Polymorphism on Serum Creatine Kinase and Interleukin-6 Levels after Maximal Eccentric Exercise Am J Phys Med Rehabil 10.1097/PHM.0000000000002623

1310. Goto, H., Yamamoto, Y., Tsujiguchi, H., Sato, T., Yamamoto, R., Takeshita, Y. *et al.* (2024) Oxytocin Receptor Polymorphism Is Associated With Sleep Apnea Symptoms J Endocr Soc **9**, bvae198 10.1210/jendso/bvae198

1311. Guilherme, J., Semenova, E. A., Kikuchi, N., Homma, H., Kozuma, A., Saito, M. *et al.* (2024) Identification of Genomic Predictors of Muscle Fiber Size Cells **13**, 10.3390/cells13141212

1312. Hanyuda, A., Raita, Y., Ninomiya, T., Hashimoto, K., Takada, N., Sato, K. *et al.* (2024) Metabolomic Profiling of Open-Angle Glaucoma Etiologic Endotypes: Tohoku Multi-Omics Glaucoma Study Invest Ophthalmol Vis Sci **65**, 44 10.1167/iovs.65.13.44

1313. Hishida, A., Nakatochi, M., Sutoh, Y., Nakano, S., Momozawa, Y., Narita, A. *et al.* (2024) GWAS Meta-analysis of Kidney Function Traits in Japanese Populations J Epidemiol **34**, 526-534 10.2188/jea.JE20230281

1314. Kimura, K., Suzuki, F., Tsujiguchi, H., Hara, A., Miyagi, S., Kannon, T. *et al.* (2024) Relationship between coronary artery disease with dyslipidaemia and trace mineral intake: a cross-sectional analysis of the Shika study J Nutr Sci **13**, e45 10.1017/jns.2024.26

1315. Koyanagi, Y. N., Nakatochi, M., Namba, S., Oze, I., Charvat, H., Narita, A. *et al.* (2024) Genetic architecture of alcohol consumption identified by a genotype-stratified GWAS and impact on esophageal cancer risk in Japanese people Sci Adv **10**, eade2780 10.1126/sciadv.ade2780

1316. Liu, X., Koyama, S., Tomizuka, K., Takata, S., Ishikawa, Y., Ito, S. *et al.* (2024) Decoding triancestral origins, archaic introgression, and natural selection in the Japanese population by whole-genome sequencing Sci Adv **10**, eadi8419 10.1126/sciadv.adi8419

1317. Lyu, H., Sugita, N., Komatsu, S., Wakasugi, M., Yokoseki, A., Yoshihara, A. *et al.* (2024) UCP2 polymorphisms, daily step count, and number of teeth associated with all-cause mortality risk in Sado City: A hospital-based cohort study Heliyon **10**, e32512 10.1016/j.heliyon.2024.e32512

1318. Naito, T., Osaka, R., Kakuta, Y., Kawai, Y., Khor, S. S., Umeno, J. *et al.* (2024) Genetically Predicted Higher Levels of Caffeic Acid Are Protective Against Ulcerative Colitis: A Comprehensive Metabolome Analysis Inflamm Bowel Dis izae143 10.1093/ibd/izae143

1319. Nakayama, A., Kawamura, Y., Nakatochi, M., Toyoda, Y., Nakajima, M., Maehara, K. *et al.* (2024) Strong genetic effect on gout revealed by genetic risk score from meta-analysis of two genome-wide association studies Hum Cell **38**, 16 10.1007/s13577-024-01138-y

1320. Sato, S., Iino, C., Sasada, T., Soma, G., Furusawa, K., Yoshida, K. *et al.* (2024) An epidemiological study on the factors including genetic polymorphism influencing ALT >30 U/L and liver fibrosis progression in metabolic dysfunction-associated steatotic liver disease among the general population JGH Open **8**, e70043 10.1002/jgh3.70043

1321. Shiota, M., Nemoto, S., Ikegami, R., Tatarano, S., Kamoto, T., Kobayashi, K. *et al.* (2024) Predictive model of castration resistance in advanced prostate cancer by machine learning using genetic and clinical data: KYUCOG-1401-A study BJC Rep **2**, 69 10.1038/s44276-024-00093-3

1322. Takahashi, N., Nishimura, T., Okumura, A., Harada, T., Iwabuchi, T., Rahman, M. S. *et al.* (2024) Association

between genetic risk of melatonin secretion and attention deficit hyperactivity disorder Psychiatry Research Communications **4**, 100188 10.1016/j.psycom.2024.100188

1323. Tanegashima, T., Shiota, M., Fujiyama, N., Narita, S., Habuchi, T., Fukuchi, G. *et al.* (2024) Effect of HLA Genotype on Anti-PD-1 Antibody Treatment for Advanced Renal Cell Carcinoma in the SNiP-RCC Study J Immunol **213**, 23-28 10.4049/jimmunol.2300308
1324. Ishida, N., Harada, S., Toki, R., Hirata, A., Matsumoto, M., Miyagawa, N. *et al.* (2025) Causal relationship between body mass index and insulin resistance: Linear and nonlinear Mendelian randomization study in a Japanese population J Diabetes Investig **16**, 1305-1314 10.1111/jdi.14377
1325. Kawahara, T., Nawa, N., Murakami, K., Tanaka, T., Ohseto, H., Takahashi, I. *et al.* (2025) Genetic effects on gestational diabetes mellitus and their interactions with environmental factors among Japanese women Journal of human genetics **70**, 265-273 10.1038/s10038-025-01330-4
1326. Khor, S. S., Hirayasu, K., Kawai, Y., Kim, H. L., Nagasaki, M., andTokunaga, K. (2025) LILR genotype imputation with attribute bagging (LIBAG): leukocyte immunoglobulin-like receptor copy number imputation system Front Immunol **16**, 1559301 10.3389/fimmu.2025.1559301
1327. Mori, Y., van Dijk, E. H. C., Miyake, M., Hosoda, Y., den Hollander, A. I., Yzer, S. *et al.* (2025) Genome-wide association and multi-omics analyses provide insights into the disease mechanisms of central serous chorioretinopathy Sci Rep **15**, 9158 10.1038/s41598-025-92210-6
1328. Omae, Y., Khor, S. S., Shimada, M., Kawai, Y., Yamaguchi, T., Yagi, M. *et al.* (2025) Genome-wide association study of common side effects following COVID-19 booster vaccination in a cohort of corporate employees in Japan Sci Rep **15**, 12728 10.1038/s41598-025-90787-6
1329. Sato, S., Iino, C., Sasada, T., Furusawa, K., Yoshida, K., Sawada, K. *et al.* (2025) A 4-year cohort study of the effects of PNPLA3 rs738409 genotypes on liver fat and fibrosis and gut microbiota in a non-fatty liver population Environ Health Prev Med **30**, 17 10.1265/ehpm.24-00365
1330. Shiota, M., Nemoto, S., Ikegami, R., Tanegashima, T., Blas, L., Miyake, H. *et al.* (2025) Predictive Model of Objective Response to Nivolumab Monotherapy for Advanced Renal Cell Carcinoma by Machine Learning Using Genetic and Clinical Data: The SNiP-RCC Study JCO Clin Cancer Inform **9**, e2400167 10.1200/CCI-24-00167
1331. Toki, R., Fushiki, S., Kojima, S., Sutoh, Y., Otsuka-Yamasaki, Y., Harada, S. *et al.* (2025) Genome-wide association study of plasma amino acids and Mendelian randomization for cardiometabolic traits Sci Rep **15**, 14569 10.1038/s41598-025-98992-z
1332. Yamamoto, Y., Shirai, Y., Sonehara, K., Namba, S., Ojima, T., Yamamoto, K. *et al.* (2025) Dissecting cross-population polygenic heterogeneity across respiratory and cardiometabolic diseases Nat Commun **16**, 3765 10.1038/s41467-025-58149-y

OmniExpressExome 1 万人分の SNP（1333-1340）

1333. Low, S. K., Takahashi, A., Ebana, Y., Ozaki, K., Christophersen, I. E., Ellinor, P. T. *et al.* (2017) Identification of six new genetic loci associated with atrial fibrillation in the Japanese population Nature genetics **49**, 953-958 10.1038/ng.3842
1334. Kanai, M., Akiyama, M., Takahashi, A., Matoba, N., Momozawa, Y., Ikeda, M. *et al.* (2018) Genetic analysis of quantitative traits in the Japanese population links cell types to complex human diseases Nature genetics **50**, 390-400 10.1038/s41588-018-0047-6
1335. Shiga, Y., Akiyama, M., Nishiguchi, K. M., Sato, K., Shimozawa, N., Takahashi, A. *et al.* (2018) Genome-wide

association study identifies seven novel susceptibility loci for primary open-angle glaucoma Human molecular genetics **27**, 1486-1496 10.1093/hmg/ddy053

1336. Tanikawa, C., Kamatani, Y., Takahashi, A., Momozawa, Y., Leveque, K., Nagayama, S. *et al.* (2018) GWAS identifies two novel colorectal cancer loci at 16q24.1 and 20q13.12 Carcinogenesis **39**, 652-660 10.1093/carcin/bgy026
1337. Tanikawa, C., Kamatani, Y., Toyoshima, O., Sakamoto, H., Ito, H., Takahashi, A. *et al.* (2018) Genome-wide association study identifies gastric cancer susceptibility loci at 12q24.11-12 and 20q11.21 Cancer Sci **109**, 4015-4024 10.1111/cas.13815
1338. Yodsurang, V., Tang, Y., Takahashi, Y., Tanikawa, C., Kamatani, Y., Takahashi, A. *et al.* (2018) Genome-wide association study (GWAS) of ovarian cancer in Japanese predicted regulatory variants in 22q13.1 PLoS One **13**, e0209096 10.1371/journal.pone.0209096
1339. Tanikawa, C., Kamatani, Y., Terao, C., Usami, M., Takahashi, A., Momozawa, Y. *et al.* (2019) Novel Risk Loci Identified in a Genome-Wide Association Study of Urolithiasis in a Japanese Population J Am Soc Nephrol **30**, 855-864 10.1681/ASN.2018090942
1340. Lin, Y., Nakatochi, M., Hosono, Y., Ito, H., Kamatani, Y., Inoko, A. *et al.* (2020) Genome-wide association meta-analysis identifies GP2 gene risk variants for pancreatic cancer Nat Commun **11**, 3175 10.1038/s41467-020-16711-w

iMETHYL（1341-1378）

1341. Guiraud, V., Fauchois, A., Soulie, C., Leducq, V., Veyri, M., Assoumou, L. *et al.* (2025) Impacts of immune checkpoint inhibitors use on the HIV reservoir are linked to provirus sequences but not integration sites Sci Rep **15**, 31726 10.1038/s41598-025-15349-2
1342. Sugawara, H., Murata, Y., Ikegame, T., Sawamura, R., Shimanaga, S., Takeoka, Y. *et al.* (2018) DNA methylation analyses of the candidate genes identified by a methylome-wide association study revealed common epigenetic alterations in schizophrenia and bipolar disorder Psychiatry and clinical neurosciences **72**, 245-254 10.1111/pcn.12645
1343. Joo, J., Omae, Y., Hitomi, Y., Park, B., Shin, H. J., Yoon, K. A. *et al.* (2019) The association of integration patterns of human papilloma virus and single nucleotide polymorphisms on immune- or DNA repair-related genes in cervical cancer patients Sci Rep **9**, 13132 10.1038/s41598-019-49523-0
1344. Fujimoto, M., Arai, E., Tsumura, K., Yotani, T., Yamada, Y., Takahashi, Y. *et al.* (2020) Establishment of diagnostic criteria for upper urinary tract urothelial carcinoma based on genome-wide DNA methylation analysis Epigenetics **15**, 1289-1301 10.1080/15592294.2020.1767374
1345. Jiang, C., Lian, X., Gao, C., Sun, X., Einkauf, K. B., Chevalier, J. M. *et al.* (2020) Distinct viral reservoirs in individuals with spontaneous control of HIV-1 Nature **585**, 261-267 10.1038/s41586-020-2651-8
1346. Meijer, M., Klein, M., Hannon, E., van der Meer, D., Hartman, C., Oosterlaan, J. *et al.* (2020) Genome-Wide DNA Methylation Patterns in Persistent Attention-Deficit/Hyperactivity Disorder and in Association With Impulsive and Callous Traits Front Genet **11**, 16 10.3389/fgene.2020.00016
1347. Pinhel, M. A. S., Noronha, N. Y., Nicoletti, C. F., Pereira, V. A., de Oliveira, B. A., Cortes-Oliveira, C. *et al.* (2020) Changes in DNA Methylation and Gene Expression of Insulin and Obesity-Related Gene PIK3R1 after Roux-en-Y Gastric Bypass Int J Mol Sci **21**, 10.3390/ijms21124476
1348. Sluiter, F., Incollingo Rodriguez, A. C., Nephew, B. C., Cali, R., Murgatroyd, C., andSantos, H. P., Jr. (2020)

Pregnancy associated epigenetic markers of inflammation predict depression and anxiety symptoms in response to discrimination Neurobiol Stress **13**, 100273 10.1016/j.ynstr.2020.100273

1349. Smith, A. K., Ratanatharathorn, A., Maihofer, A. X., Naviaux, R. K., Aiello, A. E., Amstadter, A. B. *et al.* (2020) Epigenome-wide meta-analysis of PTSD across 10 military and civilian cohorts identifies methylation changes in AHRR Nat Commun **11**, 5965 10.1038/s41467-020-19615-x
1350. Teng, C. S., Wu, B. H., Yen, M. R., andChen, P. Y. (2020) MethGET: web-based bioinformatics software for correlating genome-wide DNA methylation and gene expression BMC Genomics **21**, 375 10.1186/s12864-020-6722-x
1351. Bravo-Gutierrez, O. A., Falfan-Valencia, R., Ramirez-Venegas, A., Sansores, R. H., Hernandez-Zenteno, R. J., Hernandez-Perez, A. *et al.* (2021) Hypomethylation of AHRR (cg05575921) Is Related to Smoking Status in the Mexican Mestizo Population Genes (Basel) **12**, 10.3390/genes12081276
1352. Chilunga, F. P., Henneman, P., Venema, A., Meeks, K. A. C., Requena-Mendez, A., Beune, E. *et al.* (2021) Genome-wide DNA methylation analysis on C-reactive protein among Ghanaians suggests molecular links to the emerging risk of cardiovascular diseases NPJ Genom Med **6**, 46 10.1038/s41525-021-00213-9
1353. Huang, A. S., Ramos, V., Oliveira, T. Y., Gaebler, C., Jankovic, M., Nussenzweig, M. C. *et al.* (2021) Integration features of intact latent HIV-1 in CD4+ T cell clones contribute to viral persistence J Exp Med **218**, e20211427 10.1084/jem.20211427
1354. Huang, S., Qin, P., Chen, Q., Zhang, D., Cheng, C., Guo, C. *et al.* (2021) Association of FTO gene methylation with incident type 2 diabetes mellitus: A nested case-control study Gene **786**, 145585 10.1016/j.gene.2021.145585
1355. Lei, S., Li, L., Yang, X., Yin, Q., Xu, T., Zhou, W. *et al.* (2021) The association between RAPSN methylation in peripheral blood and breast cancer in the Chinese population Journal of human genetics **66**, 1069-1078 10.1038/s10038-021-00933-x
1356. Marion, M. C., Ramos, P. S., Bachali, P., Labonte, A. C., Zimmerman, K. D., Ainsworth, H. C. *et al.* (2021) Nucleic Acid-Sensing and Interferon-Inducible Pathways Show Differential Methylation in MZ Twins Discordant for Lupus and Overexpression in Independent Lupus Samples: Implications for Pathogenic Mechanism and Drug Targeting Genes (Basel) **12**, 1898 10.3390/genes12121898
1357. Neri de Souza Reis, V., Tahira, A. C., Daguano Gastaldi, V., Mari, P., Portolese, J., Feio Dos Santos, A. C. *et al.* (2021) Environmental Influences Measured by Epigenetic Clock and Vulnerability Components at Birth Impact Clinical ASD Heterogeneity Genes (Basel) **12**, 10.3390/genes12091433
1358. Chaar, D. L., Nguyen, K., Wang, Y. Z., Ratliff, S. M., Mosley, T. H., Kardia, S. L. R. *et al.* (2022) SNP-by-CpG Site Interactions in ABCA7 Are Associated with Cognition in Older African Americans Genes (Basel) **13**, 10.3390/genes13112150
1359. Chen, L., Li, J., Zhang, M., Zhang, Q., Wu, L., Lu, Y. *et al.* (2022) Serum Atrial Natriuretic Peptide, NPPA Promoter Methylation, and Cardiovascular Disease: A 10-year Follow-Up Study in Chinese Adults Glob Heart **17**, 27 10.5334/gh.1116
1360. Chilunga, F. P., Meeks, K. A. C., Henneman, P., Agyemang, C., Doumatey, A. P., Rotimi, C. N. *et al.* (2022) An epigenome-wide association study of insulin resistance in African Americans Clin Epigenetics **14**, 88 10.1186/s13148-022-01309-4
1361. Cusenza, V. Y., Braglia, L., andFrazzi, R. (2022) Methylation Heterogeneity and Gene Expression of SPG20 in Solid Tumors Genes (Basel) **13**, 861 10.3390/genes13050861

1362. Einkauf, K. B., Osborn, M. R., Gao, C., Sun, W., Sun, X., Lian, X. *et al.* (2022) Parallel analysis of transcription, integration, and sequence of single HIV-1 proviruses Cell **185**, 266-282 e215 10.1016/j.cell.2021.12.011

1363. He, Y., Li, Y., Zhang, J., Chen, L., Li, J., Zhang, M. *et al.* (2022) FURIN Promoter Methylation Predicts the Risk of Incident Diabetes: A Prospective Analysis in the Gusu Cohort Front Endocrinol (Lausanne) **13**, 873012 10.3389/fendo.2022.873012

1364. Peng, H., Fan, Y., Li, J., Zheng, X., Zhong, C., Zhu, Z. *et al.* (2022) DNA Methylation of the Natriuretic Peptide System Genes and Ischemic Stroke: Gene-Based and Gene Set Analyses Neurol Genet **8**, e679 10.1212/NXG.0000000000000679

1365. Rubino, E., Boschi, S., Giorgio, E., Pozzi, E., Marcinno, A., Gallo, E. *et al.* (2022) Analysis of the DNA methylation pattern of the promoter region of calcitonin gene-related peptide 1 gene in patients with episodic migraine: An exploratory case-control study Neurobiol Pain **11**, 100089 10.1016/j.ynpai.2022.100089

1366. Shi, J., Wu, L., Chen, Y., Zhang, M., Yu, J., Ren, L. *et al.* (2022) Association between CORIN methylation and hypertension in Chinese adults Postgrad Med J **99**, 753-762 10.1136/pmj-2022-141802

1367. van der Linden, E. L., Halley, A., Meeks, K. A. C., Chilunga, F., Hayfron-Benjamin, C., Venema, A. *et al.* (2022) An explorative epigenome-wide association study of plasma renin and aldosterone concentration in a Ghanaian population: the RODAM study Clin Epigenetics **14**, 159 10.1186/s13148-022-01378-5

1368. Sumalde, A. A. M., Yang, I. V., Yarza, T. K. L., Tobias-Grasso, C. A. M., Tantoco, M. L. C., Davidson, E. *et al.* (2023) Lack of Methylation Changes in GJB2 and RB1 Non-coding Regions of Cochlear Implant Patients with Sensorineural Hearing Loss Acta Med Philipp **57**, 116-120 10.47895/amp.v57i9.5200

1369. Swart, G., Meeks, K., Chilunga, F., Venema, A., Agyemang, C., van der Linden, E. *et al.* (2023) Associations between epigenome-wide DNA methylation and height-related traits among Sub-Saharan Africans: the RODAM study J Dev Orig Health Dis **14**, 658-669 10.1017/S204017442300034X

1370. Taylor, J. Y., Huang, Y., Zhao, W., Wright, M. L., Wang, Z., Hui, Q. *et al.* (2023) Epigenome-wide association study of BMI in Black populations from InterGEN and GENOA Obesity (Silver Spring) **31**, 243-255 10.1002/oby.23589

1371. Ustiugova, A. S., Ekaterina, D. M., Nataliya, M. V., Alexey, D. A., Dmitry, K. V., andMarina, A. A. (2023) CRISPR/Cas9 genome editing demonstrates functionality of the autoimmunity-associated SNP rs12946510 Biochim Biophys Acta Mol Basis Dis **1869**, 166599 10.1016/j.bbadis.2022.166599

1372. van der Laan, L., Rooney, K., Haghshenas, S., Silva, A., McConkey, H., Relator, R. *et al.* (2023) Functional Insight into and Refinement of the Genomic Boundaries of the JARID2-Neurodevelopmental Disorder Episignature Int J Mol Sci **24**, 10.3390/ijms241814240

1373. van der Linden, E. L., Meeks, K. A. C., Chilunga, F., Hayfron-Benjamin, C., Bahendeka, S., Klipstein-Grobusch, K. *et al.* (2023) Epigenome-wide association study of plasma lipids in West Africans: the RODAM study Ebiomedicine **89**, 104469 10.1016/j.ebiom.2023.104469

1374. Karaś, K., Pastwińska, J., Sałkowska, A., Karwaciak, I., andRatajewski, M. (2024) Epigenetic regulation of the human GDAP1 gene Biochem Biophys Rep **40**, 101827 10.1016/j.bbrep.2024.101827

1375. Maiti, A. K. (2024) Bioinformatic analysis predicts the regulatory function of noncoding SNPs associated with Long COVID-19 syndrome Immunogenetics **76**, 279-290 10.1007/s00251-024-01348-6

1376. Guiraud, V., Denis, J. A., Benhafoun, G., Ablin, E., Sayon, S., Souchet, L. *et al.* (2025) Longitudinal analysis of lentiviral and retroviral chimeric antigen receptors' integration sites reveals distinct clonal evolutionary patterns British Journal of Haematology **206**, 1173-1177,

https://onlinelibrary.wiley.com/doi/pdfdirect/10.1111/bjh.20020?download=true

1377. Ivie, J. J., Dill-McFarland, K. A., Simmons, J. D., Peterson, G. J., Benchek, P. H., Mayanja-Kizza, H. *et al.* (2025) Myeloid cell genome-wide screen identifies variants associated with Mycobacterium tuberculosis-induced cytokine transcriptional responses J Clin Invest **135**, 10.1172/JCI179822

1378. Sadri, F., Alipour, M., andNejatizadeh, A. (2025) Significant Association Among the Epigenetic Alterations in and Genes and Non-Small Cell Lung Cancer: A Prospective Emerging Molecular Biomarker Annals of Human Genetics e12597 10.1111/ahg.12597

その他のゲノム情報（1379）

1379. Fukunaga, H., andIkeda, A. (2023) Mitochondrial DNA copy number variation across three generations: a possible biomarker for assessing perinatal outcomes Hum Genomics **17**, 113 10.1186/s40246-023-00567-4

メタゲノム（1380）

1380. Lin, W., Liang, Z., Fang, J., Liu, Y., Lei, L., Lin, J. *et al.* (2025) Association Between Gut Microbiota and Chronic Kidney Disease: A Two-Sample Mendelian Randomization Study in a Chinese Population Biomedicines **13**, 10.3390/biomedicines13061397

メタボローム情報（1381-1387）

1381. Hatori, M., Fukuoka, S., Kimura, S., Kawakami, K., Yamaguchi, K., andYamaguchi, M. (2025) Impact of Comorbidity on Antiemetic Efficacy in Patients with Esophageal Cancer Treated with Cisplatin-Based Chemotherapy: A Retrospective Study from Japan Oncology 1-11 10.1159/000547684

1382. Iwasaki, T., Shirota, H., Hishinuma, E., Kawaoka, S., Matsukawa, N., Kasahara, Y. *et al.* (2025) Plasma Lysophosphatidylcholine Levels Correlate with Prognosis and Immunotherapy Response in Squamous Cell Carcinoma Int J Mol Sci **26**, 10.3390/ijms26157528

1383. Rashad, S., Saigusa, D., Yamazaki, T., Matsumoto, Y., Tomioka, Y., Saito, R. *et al.* (2020) Metabolic basis of neuronal vulnerability to ischemia; an in vivo untargeted metabolomics approach Sci Rep **10**, 6507 10.1038/s41598-020-63483-w

1384. Adam, M. G., Limonciel, A., andUstaszewski, B. (2022) Validation and application of a unique reference database for metabolomics–The Quantitative Metabolomics Database (QMDB) biocrates,

1385. Mino, M., Kakazu, E., Sano, A., Tsuruoka, M., Matsubara, H., Kakisaka, K. *et al.* (2024) Comprehensive analysis of peripheral blood free amino acids in MASLD: the impact of glycine-serine-threonine metabolism Amino Acids **57**, 3 10.1007/s00726-024-03433-2

1386. Mino, M., Sano, A., Kakazu, E., Matsubara, H., Kakisaka, K., Kogure, T. *et al.* (2024) Differences in branched-chain amino acid to tyrosine ratio (BTR) among etiologies of chronic liver disease progression compared to healthy adults J Gastroenterol **59**, 483-493 10.1007/s00535-024-02092-0

1387. Yang, H., Wang, T., Qian, C., Wang, H., Yu, D., Shi, M. *et al.* (2025) Gut microbial-derived phenylacetylglutamine accelerates host cellular senescence Nat Aging **5**, 401-418 10.1038/s43587-024-00795-w

GWAS レポジトリ（1388-1391）

1388. Feng, Y., Fu, M., Guan, X., Wang, C., Yuan, F., Bai, Y. *et al.* (2021) Uric Acid Mediated the Association Between BMI and Postmenopausal Breast Cancer Incidence: A Bidirectional Mendelian Randomization Analysis and

Prospective Cohort Study Front Endocrinol (Lausanne) **12**, 742411 10.3389/fendo.2021.742411

1389. Hu, S., Lin, Z., Hu, M. J., Tan, J. S., Guo, T. T., Huang, X. *et al.* (2023) Causal relationships of circulating amino acids with cardiovascular disease: a trans-ancestry Mendelian randomization analysis J Transl Med **21**, 699 10.1186/s12967-023-04580-y

1390. Iwasaki, T., Kamatani, Y., Sonomura, K., Kawaguchi, S., Kawaguchi, T., Takahashi, M. *et al.* (2023) Genetic influences on human blood metabolites in the Japanese population iScience **26**, 105738 10.1016/j.isci.2022.105738

1391. Fujii, R., Nakatochi, M., andDel Greco, M. F. (2024) Coffee Intake, Plasma Caffeine Levels, and Kidney Function: Two-Sample Mendelian Randomization Among East Asian and European Ancestries Kidney Int Rep **9**, 1083-1092 10.1016/j.ekir.2024.01.024

試料：血清･血漿（1392-1395）

1392. Kino, S., Ishida, T., Niizuma, K., Kanamori, M., Sato, K., Shida, N. *et al.* (2026) Developing a predictive model for rapid stroke diagnosis via blood using mid-infrared spectroscopy Spectrochim Acta A Mol Biomol Spectrosc **346**, 126883 10.1016/j.saa.2025.126883

1393. Kabe, Y., Suematsu, M., Sakamoto, S., Hirai, M., Koike, I., Hishiki, T. *et al.* (2018) Development of a Highly Sensitive Device for Counting the Number of Disease-Specific Exosomes in Human Sera Clin Chem **64**, 1463-1473 10.1373/clinchem.2018.291963

1394. Yokose, T., Kabe, Y., Matsuda, A., Kitago, M., Matsuda, S., Hirai, M. *et al.* (2020) O-Glycan-Altered Extracellular Vesicles: A Specific Serum Marker Elevated in Pancreatic Cancer Cancers (Basel) **12**, 2469 10.3390/cancers12092469

1395. Hanada, K., Shimizu, A., Tsushima, K., and Kobayashi, M. (2024) Potential of Carbohydrate Antigen 19-9 and Serum Apolipoprotein A2-Isoforms in the Diagnosis of Stage 0 and IA Pancreatic Cancer Diagnostics (Basel) **14**, 10.3390/diagnostics14171920

試料：DNA（1396-1397）

1396. Ogiwara, Y., Miyado, M., Suzuki, E., Niida, S., Ozaki, K., andFukami, M. (2021) Structural and numerical Y chromosomal variations in elderly men identified through multiplex ligation-dependent probe amplification Journal of human genetics **66**, 1181-1184 10.1038/s10038-021-00943-9

1397. Shiraishi, K., Takahashi, A., Momozawa, Y., Daigo, Y., Kaneko, S., Kawaguchi, T. *et al.* (2024) Identification of telomere maintenance gene variations related to lung adenocarcinoma risk by genome-wide association and whole genome sequencing analyses Cancer Commun (Lond) **44**, 287-293 10.1002/cac2.12498

試料：母乳（1398）

1398. Saito, Y., Sato, K., Jinno, S., Nakamura, Y., Nobukuni, T., Ogishima, S. *et al.* (2023) Effect of Nicotinamide Mononucleotide Concentration in Human Milk on Neurodevelopmental Outcome: The Tohoku Medical Megabank Project Birth and Three-Generation Cohort Study Nutrients **16**, 10.3390/nu16010145

健康調査情報（1399-1421）

1399. Iwata, H., Tojo, M., Tsuchiya, K. J., Ishikuro, M., Chen, G., Suyama, S. *et al.* (2025) Small for gestational age as a predictor of developmental coordination disorders: Exploring early risk from Japan birth cohort consortium

Brain Dev **47**, 104435 10.1016/j.braindev.2025.104435

1400. Wojcicki, J. M., Tsuchiya, K. J., Murakami, K., Ishikuro, M., Obara, T., andMorisaki, N. (2021) Limited consumption of 100% fruit juices and sugar sweetened beverages in Japanese toddler and preschool children Prev Med Rep **23**, 101409 10.1016/j.pmedr.2021.101409
1401. Iwagami, M., Goto, A., Katagiri, R., Sutoh, Y., Koyanagi, Y. N., Nakatochi, M. *et al.* (2022) Blood Lipids and the Risk of Colorectal Cancer: Mendelian Randomization Analyses in the Japanese Consortium of Genetic Epidemiology Studies Cancer Prev Res (Phila) **15**, 827-836 10.1158/1940-6207.CAPR-22-0146
1402. Yokokawa, H., Suzuki, M., Aoki, N., Sato, Y., andNaito, T. (2022) Achievement of target blood pressure among community residents with hypertension and factors associated with therapeutic failure in the northern territory of Japan J Int Med Res **50**, 3000605221126878 10.1177/03000605221126878
1403. Egawa, M., Kanda, E., Ohtsu, H., Nakamura, T., andYoshida, M. (2023) Number of Children and Risk of Cardiovascular Disease in Japanese Women: Findings from the Tohoku Medical Megabank J Atheroscler Thromb **30**, 131-137 10.5551/jat.63527
1404. Egawa, M., Kanda, E., Ohtsu, H., Nakamura, T., andYoshida, M. (2023) Hypertensive Disorders of Pregnancy are Associated with Cardiovascular Disease in Middle- and Older-Aged Japanese Women J Atheroscler Thromb **30**, 1420-1426 10.5551/jat.63816
1405. Hanyuda, A., Goto, A., Katagiri, R., Koyanagi, Y. N., Nakatochi, M., Sutoh, Y. *et al.* (2023) Investigating the association between glycaemic traits and colorectal cancer in the Japanese population using Mendelian randomisation Sci Rep **13**, 7052 10.1038/s41598-023-33966-7
1406. Hanyuda, A., Goto, A., Nakatochi, M., Sutoh, Y., Narita, A., Nakano, S. *et al.* (2023) Association Between Glycemic Traits and Primary Open-Angle Glaucoma: A Mendelian Randomization Study in the Japanese Population Am J Ophthalmol **245**, 193-201 10.1016/j.ajo.2022.09.004
1407. Machida, T., Obara, T., Ishikuro, M., Murakami, K., Ueno, F., Noda, A. *et al.* (2023) Liver steatosis and fibrosis markers' association with cardiovascular and renal damage in Japanese adults: the TMM BirThree cohort study Ann Hepatol **28**, 100761 10.1016/j.aohep.2022.100761
1408. Morisaki, N., Obara, T., Piedvache, A., Kobayashi, S., Miyashita, C., Nishimura, T. *et al.* (2023) Association Between Smoking and Hypertension in Pregnancy Among Japanese Women: A Meta-analysis of Birth Cohort Studies in the Japan Birth Cohort Consortium (JBiCC) and JECS J Epidemiol **33**, 498-507 10.2188/jea.JE20220076
1409. Ohe, T., Yamada, M., Hozawa, A., Nakaya, N., Nakamura, T., Tsuchiya, N. *et al.* (2023) Associations between birth weight and lung function in a Japanese adult population: The tohoku medical megabank community-based cohort study Respir Investig **61**, 588-600 10.1016/j.resinv.2023.06.004
1410. Watarai, G., Suzuki, J., Motoike, I. N., Sakurai, M., Ikeda, R., Kawase, T. *et al.* (2023) Relationship between age-related hearing loss and consumption of coffee and tea Geriatr Gerontol Int **23**, 453-456 10.1111/ggi.14589
1411. Yokokawa, H., Suzuki, M., Aoki, N., Fukuda, H., Sato, Y., Hisaoka, T. *et al.* (2023) Association between serum uric acid levels and achievement of target blood pressure among Japanese community residents with hypertension J Clin Hypertens (Greenwich) **25**, 295-303 10.1111/jch.14644
1412. Benton, M., Ishikuro, M., Obara, T., Noda, A., Murakami, K., Kuriyama, S. *et al.* (2024) The effect of gestational diabetes mellitus on postnatal mother-infant bonding: Findings from the Tohoku Medical Megabank Project Birth and Three-Generation Cohort Study Prev Med **187**, 108101 10.1016/j.ypmed.2024.108101
1413. Ishitsuka, K., Piedvache, A., Kobayashi, S., Iwama, N., Nishimura, T., Watanabe, M. *et al.* (2024) The Population-

Attributable Fractions of Small-for-Gestational-Age Births: Results from the Japan Birth Cohort Consortium Nutrients **16**, 10.3390/nu16020186

1414. Minami, S., Takahashi, M., Shinden, S., Shirai, K., Oishi, N., Nishimura, H. *et al.* (2024) Prediction of Cochlear Implant Effectiveness With Surface-Based Morphometry Otol Neurotol **45**, 114-120 10.1097/MAO.0000000000004070
1415. Okabe, T., Kunikata, H., Yasuda, M., Kodama, S., Maeda, Y., Nakano, J. *et al.* (2024) Relationship between nailfold capillaroscopy parameters and the severity of diabetic retinopathy Graefes Arch Clin Exp Ophthalmol **262**, 759-768 10.1007/s00417-023-06220-z
1416. Sato, Y., Yokokawa, H., Suzuki, R., Goto, T., andNaito, T. (2024) The association between milk and dairy products intake and insomnia symptoms among Japanese adults in community-based cohort Nutr Health 2601060241283133 10.1177/02601060241283133
1417. Takano, R., Tominaga, Y., Fu, D. J., Moyer, J. A., Cheng, Y., andOkada, K. (2024) Self-administered generational surveys combine with genetic analysis to reveal foundations of depression in Japanese adults J Affect Disord **356**, 204-214 10.1016/j.jad.2024.04.021
1418. Wang, H., Iwama, N., Yuwaki, K., Nakamichi, Y., Hamada, H., Tomita, H. *et al.* (2024) Association of parity with the prevalence of hypertension in Japan: The Tohoku Medical Megabank Community-based cohort study J Clin Hypertens (Greenwich) **26**, 102-121 10.1111/jch.14756
1419. Wang, H., Iwama, N., Yuwaki, K., Nakamichi, Y., Hamada, H., Tomita, H. *et al.* (2024) Relationship between parity and the prevalence of chronic kidney disease in Japan considering hypertensive disorders of pregnancy and body mass index BMC Nephrol **25**, 166 10.1186/s12882-024-03604-z
1420. Aoki, N., Suzuki, M., Sato, Y., Yokokawa, H., andNaito, T. (2025) Relationship between tea intake and cedar pollen allergy: a population-based cross-sectional study J Nutr Sci **14**, e2 10.1017/jns.2024.96
1421. Kawaguchi, C., Ozawa, M., Hidaka, T., Murakami, K., Ishikuro, M., Ueno, F. *et al.* (2025) Skin health survey on atopic dermatitis among Japanese children: The Tohoku Medical Megabank Project Birth and Three-Generation Cohort Study Allergol Int **74**, 246-253 10.1016/j.alit.2024.09.008

施設、技術･方法、データベース（1422-1447）

1422. Tanizawa, Y., Murakami, A., Takahashi, R., Doi, K., Kujiraoka, M., Sato, H. *et al.* (2025) Quantum Key Distribution Network and Quantum Secure Cloud Technologies for Genome Medicine Use Cases Ieee Transactions on Quantum Engineering **6**, Artn 4101215 10.1109/Tqe.2025.3611335
1423. Yamauchi, T., andHiyama, S., Daisuke Ochi (2016) Initiative toward Prevention and Early Detection of Disease Using Healthcare Data and Genome Analysis NTT DOCOMO Technical Journal **17**,
1424. Zhang, M., Huang, F., Wang, G., Liu, X., Wen, J., Zhang, X. *et al.* (2017) Geographic distribution of cadmium and its interaction with the microbial community in the Longjiang River: risk evaluation after a shocking pollution accident Sci Rep **7**, 227 10.1038/s41598-017-00280-y
1425. Zhang, M., Liu, X., Li, Y., Wang, G., Wang, Z., andWen, J. (2017) Microbial community and metabolic pathway succession driven by changed nutrient inputs in tailings: effects of different nutrients on tailing remediation Sci Rep **7**, 474 10.1038/s41598-017-00580-3
1426. Dong, S., Zhang, S., Chen, Z., Zhang, R., Tian, L., Cheng, L. *et al.* (2018) Berberine Could Ameliorate Cardiac Dysfunction via Interfering Myocardial Lipidomic Profiles in the Rat Model of Diabetic Cardiomyopathy Front Physiol **9**, 1042 10.3389/fphys.2018.01042

1427. Malmberg, M. M., Shi, F., Spangenberg, G. C., Daetwyler, H. D., andCogan, N. O. I. (2018) Diversity and Genome Analysis of Australian and Global Oilseed Brassica napus L. Germplasm Using Transcriptomics and Whole Genome Re-sequencing Front Plant Sci **9**, 508 10.3389/fpls.2018.00508

1428. Sekine, H., Okazaki, K., Kato, K., Alam, M. M., Shima, H., Katsuoka, F. *et al.* (2018) O-GlcNAcylation Signal Mediates Proteasome Inhibitor Resistance in Cancer Cells by Stabilizing NRF1 Mol Cell Biol **38**, 10.1128/MCB.00252-18

1429. Ishii, H., Saitoh, M., Sakamoto, K., Sakamoto, K., Saigusa, D., Kasai, H. *et al.* (2020) Lipidome-based rapid diagnosis with machine learning for detection of TGF-beta signalling activated area in head and neck cancer Br J Cancer **122**, 995-1004 10.1038/s41416-020-0732-y

1430. Nishizawa, H., Matsumoto, M., Shindo, T., Saigusa, D., Kato, H., Suzuki, K. *et al.* (2020) Ferroptosis is controlled by the coordinated transcriptional regulation of glutathione and labile iron metabolism by the transcription factor BACH1 J Biol Chem **295**, 69-82 10.1074/jbc.RA119.009548

1431. Okazaki, K., Anzawa, H., Liu, Z., Ota, N., Kitamura, H., Onodera, Y. *et al.* (2020) Enhancer remodeling promotes tumor-initiating activity in NRF2-activated non-small cell lung cancers Nat Commun **11**, 5911 10.1038/s41467-020-19593-0

1432. Ando, T., Nakamura, R., Kuru, S., Yokoi, D., Atsuta, N., Koike, H. *et al.* (2021) The wide-ranging clinical and genetic features in Japanese families with valosin-containing protein proteinopathy Neurobiol Aging **100**, 120 e121-120 e126 10.1016/j.neurobiolaging.2020.10.028

1433. Onoki, T., Izumi, Y., Takahashi, M., Murakami, S., Matsumaru, D., Ohta, N. *et al.* (2021) Skeletal muscle-specific Keap1 disruption modulates fatty acid utilization and enhances exercise capacity in female mice Redox Biol **43**, 101966 10.1016/j.redox.2021.101966

1434. Tayama, H., Karasawa, H., Yamamura, A., Okamura, Y., Katsuoka, F., Suzuki, H. *et al.* (2021) The association between ERK inhibitor sensitivity and molecular characteristics in colorectal cancer Biochem Biophys Res Commun **560**, 59-65 10.1016/j.bbrc.2021.04.130

1435. Nishiumi, S., Izumi, Y., Hirayama, A., Takahashi, M., Nakao, M., Hata, K. *et al.* (2022) Comparative Evaluation of Plasma Metabolomic Data from Multiple Laboratories Metabolites **12**, 135 10.3390/metabo12020135

1436. Okazaki, K., Anzawa, H., Katsuoka, F., Kinoshita, K., Sekine, H., andMotohashi, H. (2022) CEBPB is required for NRF2-mediated drug resistance in NRF2-activated non-small cell lung cancer cells J Biochem **171**, 567-578 10.1093/jb/mvac013

1437. Akiyama, M., Akiyama, T., Saigusa, D., Hishinuma, E., Matsukawa, N., Shibata, T. *et al.* (2023) Comprehensive study of metabolic changes induced by a ketogenic diet therapy using GC/MS- and LC/MS-based metabolomics Seizure **107**, 52-59 10.1016/j.seizure.2023.03.014

1438. Chen, Z., Watanabe, S., Hashida, H., Inoue, M., Daigaku, Y., Kikkawa, M. *et al.* (2023) Cryo-EM structures of human SPCA1a reveal the mechanism of Ca(2+)/Mn(2+) transport into the Golgi apparatus Sci Adv **9**, eadd9742 10.1126/sciadv.add9742

1439. Fujino, M., Morito, N., Hayashi, T., Ojima, M., Ishibashi, S., Kuno, A. *et al.* (2023) Transcription factor c-Maf deletion improves streptozotocin-induced diabetic nephropathy by directly regulating Sglt2 and Glut2 JCI Insight **8**, 10.1172/jci.insight.163306

1440. Ishikawa, T., Ogawa, T., Shiihara, M., Usubuchi, H., Omori, Y., Hirose, K. *et al.* (2023) Salivary gland cancer organoids are valid for preclinical genotype-oriented medical precision trials iScience **26**, 106695 10.1016/j.isci.2023.106695

1441. Akiyama, T., Saigusa, D., Inoue, T., Tokorodani, C., Akiyama, M., Michiue, R. *et al.* (2024) Exploration of urine metabolic biomarkers for new-onset, untreated pediatric epilepsy: A gas and liquid chromatography mass spectrometry-based metabolomics study Brain Dev **46**, 180-186 10.1016/j.braindev.2023.12.004
1442. Gutierrez Rico, E., Joseph, P., Noutsos, C., andPoon, K. (2024) Hypothalamic and hippocampal transcriptome changes in App(NL-G-F) mice as a function of metabolic and inflammatory dysfunction Neuroscience **554**, 107-117 10.1016/j.neuroscience.2024.07.007
1443. Itoh, T., Omori, Y., Seino, M., Hirose, K., Date, F., Ono, Y. *et al.* (2024) Gene Rearrangement and Expression of PRKACA and PRKACB Govern Morphobiology of Pancreatobiliary Oncocytic Neoplasms Mod Pathol **37**, 100358 10.1016/j.modpat.2023.100358
1444. Kino, S., Kanamori, M., Shimoda, Y., Niizuma, K., Endo, H., andMatsuura, Y. (2024) Distinguishing IDH mutation status in gliomas using FTIR-ATR spectra of peripheral blood plasma indicating clear traces of protein amyloid aggregation BMC Cancer **24**, 222 10.1186/s12885-024-11970-y
1445. Bekker, G. J., Nagao, C., Shirota, M., Nakamura, T., Katayama, T., Kihara, D. *et al.* (2025) Protein Data Bank Japan: Improved tools for sequence-oriented analysis of protein structures Protein Sci **34**, e70052 10.1002/pro.70052
1446. Bekker, G. J., Nagao, C., Shirota, M., Nakamura, T., Katayama, T., Kihara, D. *et al.* (2025) Protein Data Bank Japan: Computational Resources for Analysis of Protein Structures J Mol Biol **437**, 169013 10.1016/j.jmb.2025.169013
1447. Narita, D., Hishinuma, E., Ebina-Shibuya, R., Miyauchi, E., Matsukawa, N., Motoike, I. N. *et al.* (2025) Histological and genetic features and therapeutic responses of lung cancers explored via the global analysis of their metabolome profile Lung Cancer **200**, 108082 10.1016/j.lungcan.2025.108082

知財申請（1448-1450）

1448. Yamaguchi, K., Kusuhara, M., Serizawa, M., Mochizuki, T., Ohshima, K., Hatakeyama, K. *et al.* inventors; Google Patents assignee. Method for determining presence or absence of risk of developing cancer
1449. 株式会社東芝 inventors; 特許庁 assignee. 形質予測モデル作成装置、形質予測装置及び形質予測モデル作成方法
1450. 東ソー株式会社 inventors; 特許庁 assignee. がん罹患者の全生存期間を予測する方法

論文･本での引用（1451-1752）

1451. Chen, Y. A., Kawashima, H., Park, J., Mohsen, A., Hosomi, K., Nakagata, T. *et al.* (2025) NIBN Japan Microbiome Database, a database for exploring the correlations between human microbiome and health Sci Rep **15**, 19640 10.1038/s41598-025-04339-z
1452. Upadhyay, A. K., andSethy, N. K. (2025) Multi-omics Approaches for Understanding High-Altitude Pathophysiology In Health and Sustenance in Extreme Environment: A Military Perspective, Springer, 115-135
1453. Zhang, G., Wang, J., Yang, J., Li, W., Deng, Y., Li, J. *et al.* (2015) Comparison and evaluation of two exome capture kits and sequencing platforms for variant calling BMC Genomics **16**, 581 10.1186/s12864-015-1796-6
1454. Chung, R. H., Tsai, W. Y., Kang, C. Y., Yao, P. J., Tsai, H. J., andChen, C. H. (2016) FamPipe: An Automatic Analysis Pipeline for Analyzing Sequencing Data in Families for Disease Studies PLoS Comput Biol **12**, e1004980 10.1371/journal.pcbi.1004980
1455. Higasa, K., Miyake, N., Yoshimura, J., Okamura, K., Niihori, T., Saitsu, H. *et al.* (2016) Human genetic variation database, a reference database of genetic variations in the Japanese population Journal of human genetics **61**,

547-553 10.1038/jhg.2016.12

1456. Huang, M. C., Chuang, T. P., Chen, C. H., Wu, J. Y., Chen, Y. T., Li, L. H. *et al.* (2016) An integrated analysis tool for analyzing hybridization intensities and genotypes using new-generation population-optimized human arrays BMC Genomics **17**, 266 10.1186/s12864-016-2478-8

1457. Lacoste, C., Desvignes, J. P., Salgado, D., Pecheux, C., Villard, L., Bartoli, M. *et al.* (2016) Coverage Analysis of Lists of Genes involved in Heterogeneous Genetic Diseases following Benchtop Exome Sequencing using the Ion Proton J Genet **95**, 203-208 10.1007/s12041-016-0619-0

1458. Shinozuka, H., andForster, J. W. (2016) Use of the melting curve assay as a means for high-throughput quantification of Illumina sequencing libraries PeerJ **4**, e2281 10.7717/peerj.2281

1459. Telenti, A., Pierce, L. C., Biggs, W. H., di Iulio, J., Wong, E. H., Fabani, M. M. *et al.* (2016) Deep sequencing of 10,000 human genomes Proc Natl Acad Sci U S A **113**, 11901-11906 10.1073/pnas.1613365113

1460. Adachi, T., Kawamura, K., Furusawa, Y., Nishizaki, Y., Imanishi, N., Umehara, S. *et al.* (2017) Japan's initiative on rare and undiagnosed diseases (IRUD): towards an end to the diagnostic odyssey Eur J Hum Genet **25**, 1025-1028 10.1038/ejhg.2017.106

1461. Ahmad, M., Sinha, A., Ghosh, S., Kumar, V., Davila, S., Yajnik, C. S. *et al.* (2017) Inclusion of Population-specific Reference Panel from India to the 1000 Genomes Phase 3 Panel Improves Imputation Accuracy Sci Rep **7**, 6733 10.1038/s41598-017-06905-6

1462. Ameur, A., Dahlberg, J., Olason, P., Vezzi, F., Karlsson, R., Martin, M. *et al.* (2017) SweGen: a whole-genome data resource of genetic variability in a cross-section of the Swedish population Eur J Hum Genet **25**, 1253-1260 10.1038/ejhg.2017.130

1463. Carbonell-Caballero, J., Amadoz, A., Alonso, R., Hidalgo, M. R., Cubuk, C., Conesa, D. *et al.* (2017) Reference genome assessment from a population scale perspective: an accurate profile of variability and noise Bioinformatics **33**, 3511-3517 10.1093/bioinformatics/btx482

1464. Einhorn, Y., Weissglas-Volkov, D., Carmi, S., Ostrer, H., Friedman, E., andShomron, N. (2017) Differential analysis of mutations in the Jewish population and their implications for diseases Genet Res (Camb) **99**, e3 10.1017/S0016672317000015

1465. Konagaya, R., Naitoh, K., Suzuki, K., andTakashima, H. (2017) Prognostic medication: for predicting premonition and recovery Artificial Life and Robotics **22**, 449-456 10.1007/s10015-017-0375-0

1466. Machida, J., Goto, H., Tatematsu, T., Shibata, A., Miyachi, H., Takahashi, K. *et al.* (2017) WNT10A variants isolated from Japanese patients with congenital tooth agenesis Hum Genome Var **4**, 17047 10.1038/hgv.2017.47

1467. Nagai, A., Hirata, M., Kamatani, Y., Muto, K., Matsuda, K., Kiyohara, Y. *et al.* (2017) Overview of the BioBank Japan Project: Study design and profile J Epidemiol **27**, S2-S8 10.1016/j.je.2016.12.005

1468. Narimatsu, H. (2017) Gene-Environment Interactions in Preventive Medicine: Current Status and Expectations for the Future Int J Mol Sci **18**, 302-308 10.3390/ijms18020302

1469. Pan, C., McInnes, G., Deflaux, N., Snyder, M., Bingham, J., Datta, S. *et al.* (2017) Cloud-based interactive analytics for terabytes of genomic variants data Bioinformatics **33**, 3709-3715 10.1093/bioinformatics/btx468

1470. Popitsch, N., Consortium, W. G. S., Schuh, A., andTaylor, J. C. (2017) ReliableGenome: annotation of genomic regions with high/low variant calling concordance Bioinformatics **33**, 155-160 10.1093/bioinformatics/btw587

1471. Reisetter, A. C., Muehlbauer, M. J., Bain, J. R., Nodzenski, M., Stevens, R. D., Ilkayeva, O. *et al.* (2017) Mixture model normalization for non-targeted gas chromatography/mass spectrometry metabolomics data BMC

Bioinformatics **18**, 84 10.1186/s12859-017-1501-7

1472. Sato, K., Koyasu, M., Nomura, S., Sato, Y., Kita, M., Ashihara, Y. *et al.* (2017) Mutation status of RAD51C, PALB2 and BRIP1 in 100 Japanese familial breast cancer cases without BRCA1 and BRCA2 mutations Cancer Sci **108**, 2287-2294 10.1111/cas.13350
1473. Tanisawa, K., Arai, Y., Hirose, N., Shimokata, H., Yamada, Y., Kawai, H. *et al.* (2017) Exome-wide Association Study Identifies CLEC3B Missense Variant p.S106G as Being Associated With Extreme Longevity in East Asian Populations J Gerontol A Biol Sci Med Sci **72**, 309-318 10.1093/gerona/glw074
1474. Weisenfeld, N. I., Kumar, V., Shah, P., Church, D. M., andJaffe, D. B. (2017) Direct determination of diploid genome sequences Genome Res **27**, 757-767 10.1101/gr.214874.116
1475. Yoshizawa, G., Sasongko, T. H., Ho, C. H., andKato, K. (2017) Social and Communicative Functions of Informed Consent Forms in East Asia and Beyond Front Genet **8**, 99 10.3389/fgene.2017.00099
1476. Zhang, X., Liang, B., Xu, X., Zhou, F., Kong, L., Shen, J. *et al.* (2017) The comparison of the performance of four whole genome amplification kits on ion proton platform in copy number variation detection Biosci Rep **37**, 10.1042/BSR20170252
1477. Ahn, Y. J., Markkandan, K., Baek, I. P., Mun, S., Lee, W., Kim, H. S. *et al.* (2018) An efficient and tunable parameter to improve variant calling for whole genome and exome sequencing data Genes Genomics **40**, 39-47 10.1007/s13258-017-0608-6
1478. Amemiya, K., Hirotsu, Y., Oyama, T., andOmata, M. (2018) Simple and Rapid Method to Obtain High-quality Tumor DNA from Clinical-pathological Specimens Using Touch Imprint Cytology J Vis Exp 10.3791/56943
1479. Corbett, S., Courtiol, A., Lummaa, V., Moorad, J., andStearns, S. (2018) The transition to modernity and chronic disease: mismatch and natural selection Nat Rev Genet **19**, 419-430 10.1038/s41576-018-0012-3
1480. Dolgova, O., andLao, O. (2018) Evolutionary and Medical Consequences of Archaic Introgression into Modern Human Genomes Genes (Basel) **9**, 358 10.3390/genes9070358
1481. Hussing, C., Kampmann, M. L., Mogensen, H. S., Borsting, C., andMorling, N. (2018) Quantification of massively parallel sequencing libraries - a comparative study of eight methods Sci Rep **8**, 1110 10.1038/s41598-018-19574-w
1482. Jain, A., Gandhi, S., Koshy, R., andScaria, V. (2018) Incidental and clinically actionable genetic variants in 1005 whole exomes and genomes from Qatar Mol Genet Genomics **293**, 919-929 10.1007/s00438-018-1431-8
1483. Kim, J., Weber, J. A., Jho, S., Jang, J., Jun, J., Cho, Y. S. *et al.* (2018) KoVariome: Korean National Standard Reference Variome database of whole genomes with comprehensive SNV, indel, CNV, and SV analyses Sci Rep **8**, 5677 10.1038/s41598-018-23837-x
1484. Kocka, K., Slusarska, B., Bartoszek, A., Rzaca, M., Zdybel-Faldyga, U., Nowicki, G. *et al.* (2018) Comparative analysis of health problems in students from elementary school and middle school in Lublin, 2010-2015 Family Medicine and Primary Care Review **20**, 7-12 10.5114/fmpcr.2018.73697
1485. Komar, P., andKural, D. (2018) geck: trio-based comparative benchmarking of variant calls Bioinformatics **34**, 3488-3495 10.1093/bioinformatics/bty415
1486. Kondratyev, N., Golov, A., Alfimova, M., Lezheiko, T., andGolimbet, V. (2018) Prediction of smoking by multiplex bisulfite PCR with long amplicons considering allele-specific effects on DNA methylation Clin Epigenetics **10**, 130 10.1186/s13148-018-0565-1
1487. La Frano, M. R., Carmichael, S. L., Ma, C., Hardley, M., Shen, T., Wong, R. *et al.* (2018) Impact of post-collection freezing delay on the reliability of serum metabolomics in samples reflecting the California mid-term pregnancy

biobank Metabolomics **14**, 151 10.1007/s11306-018-1450-9

1488. Lee, S. H., Nam, H. S., Kim, H. B., Kim, E. J., Noh, J. W., andChae, J. H. (2018) Factors Associated with Complicated Grief in Students Who Survived the Sewol Ferry Disaster in South Korea Psychiatry Investig **15**, 254-260 10.30773/pi.2017.05.04

1489. Lencz, T., Yu, J., Palmer, C., Carmi, S., Ben-Avraham, D., Barzilai, N. *et al.* (2018) High-depth whole genome sequencing of an Ashkenazi Jewish reference panel: enhancing sensitivity, accuracy, and imputation Hum Genet **137**, 343-355 10.1007/s00439-018-1886-z

1490. Minari, J., Brothers, K. B., andMorrison, M. (2018) Tensions in ethics and policy created by National Precision Medicine Programs Hum Genomics **12**, 22 10.1186/s40246-018-0151-9

1491. Okada, Y., Momozawa, Y., Sakaue, S., Kanai, M., Ishigaki, K., Akiyama, M. *et al.* (2018) Deep whole-genome sequencing reveals recent selection signatures linked to evolution and disease risk of Japanese Nat Commun **9**, 1631 10.1038/s41467-018-03274-0

1492. Tada, M., Hirata, M., Sasaki, M., Sakate, R., Kohara, A., Takahashi, I. *et al.* (2018) The Rare Disease Bank of Japan: establishment, current status and future challenges Hum Cell **31**, 183-188 10.1007/s13577-018-0204-3

1493. Takashima, Y., Sasaki, Y., Hayano, A., Homma, J., Fukai, J., Iwadate, Y. *et al.* (2018) Target amplicon exome-sequencing identifies promising diagnosis and prognostic markers involved in RTK-RAS and PI3K-AKT signaling as central oncopathways in primary central nervous system lymphoma Oncotarget **9**, 27471, https://www.oncotarget.com/article/25463/pdf/

1494. Teitz, L. S., Pyntikova, T., Skaletsky, H., andPage, D. C. (2018) Selection Has Countered High Mutability to Preserve the Ancestral Copy Number of Y Chromosome Amplicons in Diverse Human Lineages Am J Hum Genet **103**, 261-275 10.1016/j.ajhg.2018.07.007

1495. Wong, K. H. Y., Levy-Sakin, M., andKwok, P. Y. (2018) De novo human genome assemblies reveal spectrum of alternative haplotypes in diverse populations Nat Commun **9**, 3040 10.1038/s41467-018-05513-w

1496. Yokozeki, H., andIzuhara, K. (2018) Two facets of sweat: A defensive factor in skin tissues and an accelerating factor for allergic skin diseases Allergol Int **67**, 433-434 10.1016/j.alit.2018.09.001

1497. Zamzaireen, Z., Natalia, C., andJuni, M. H. (2018) Post disaster governance and disease surveillance following earthquake International Journal of Public Health and Clinical Sciences **5**, 49-55,

1498. Ballardini, N., Kramer, M. S., Oken, E., Henderson, A. J., Bogdanovich, N., Dahhou, M. *et al.* (2019) Associations of atopic dermatitis and asthma with child behaviour: Results from the PROBIT cohort Clin Exp Allergy **49**, 1235-1244 10.1111/cea.13417

1499. Chen, Z., Chu, S., Xu, X., Jiang, J., Wang, W., Shen, H. *et al.* (2019) Analysis of longissimus muscle quality characteristics and associations with DNA methylation status in cattle Genes Genomics **41**, 1147-1163 10.1007/s13258-019-00844-4

1500. Dahary, D., Golan, Y., Mazor, Y., Zelig, O., Barshir, R., Twik, M. *et al.* (2019) Genome analysis and knowledge-driven variant interpretation with TGex BMC Med Genomics **12**, 200 10.1186/s12920-019-0647-8

1501. Du, Z., Ma, L., Qu, H., Chen, W., Zhang, B., Lu, X. *et al.* (2019) Whole Genome Analyses of Chinese Population and De Novo Assembly of A Northern Han Genome Genomics Proteomics Bioinformatics **17**, 229-247 10.1016/j.gpb.2019.07.002

1502. Fattahi, Z., Beheshtian, M., Mohseni, M., Poustchi, H., Sellars, E., Nezhadi, S. H. *et al.* (2019) Iranome: A catalog of genomic variations in the Iranian population Hum Mutat **40**, 1968-1984 10.1002/humu.23880

1503. GenomeAsia, K. C. (2019) The GenomeAsia 100K Project enables genetic discoveries across Asia Nature **576**, 106-111 10.1038/s41586-019-1793-z

1504. Ido, K., Nakamura, N., andNakayama, M. (2019) Miyagi Medical and Welfare Information Network: A Backup System for Patient Clinical Information after the Great East Japan Earthquake and Tsunami Tohoku J Exp Med **248**, 19-25 10.1620/tjem.248.19

1505. Ju, C., Fiori, L. M., Belzeaux, R., Theroux, J. F., Chen, G. G., Aouabed, Z. *et al.* (2019) Integrated genome-wide methylation and expression analyses reveal functional predictors of response to antidepressants Transl Psychiatry **9**, 254 10.1038/s41398-019-0589-0

1506. Kanemitsu, Y., Mishima, E., Maekawa, M., Matsumoto, Y., Saigusa, D., Yamaguchi, H. *et al.* (2019) Comprehensive and semi-quantitative analysis of carboxyl-containing metabolites related to gut microbiota on chronic kidney disease using 2-picolylamine isotopic labeling LC-MS/MS Sci Rep **9**, 19075 10.1038/s41598-019-55600-1

1507. Kishikawa, T., Momozawa, Y., Ozeki, T., Mushiroda, T., Inohara, H., Kamatani, Y. *et al.* (2019) Empirical evaluation of variant calling accuracy using ultra-deep whole-genome sequencing data Sci Rep **9**, 1784 10.1038/s41598-018-38346-0

1508. Kosugi, S., Momozawa, Y., Liu, X., Terao, C., Kubo, M., andKamatani, Y. (2019) Comprehensive evaluation of structural variation detection algorithms for whole genome sequencing Genome Biol **20**, 117 10.1186/s13059-019-1720-5

1509. Lacaze, P., Pinese, M., Kaplan, W., Stone, A., Brion, M. J., Woods, R. L. *et al.* (2019) The Medical Genome Reference Bank: a whole-genome data resource of 4000 healthy elderly individuals. Rationale and cohort design Eur J Hum Genet **27**, 308-316 10.1038/s41431-018-0279-z

1510. Lee, C. Y., Chattopadhyay, A., Chiang, L. M., Juang, J. J., Lai, L. C., Tsai, M. H. *et al.* (2019) VariED: the first integrated database of gene annotation and expression profiles for variants related to human diseases Database (Oxford) **2019**, 1-11 10.1093/database/baz075

1511. Liang, Y., He, L., Zhao, Y., Hao, Y., Zhou, Y., Li, M. *et al.* (2019) Comparative Analysis for the Performance of Variant Calling Pipelines on Detecting the de novo Mutations in Humans Front Pharmacol **10**, 358 10.3389/fphar.2019.00358

1512. Liu, S., Fang, L., Zhou, Y., Santos, D. J. A., Xiang, R., Daetwyler, H. D. *et al.* (2019) Analyses of inter-individual variations of sperm DNA methylation and their potential implications in cattle BMC Genomics **20**, 888 10.1186/s12864-019-6228-6

1513. Murata, Y., Fujii, A., Kanata, S., Fujikawa, S., Ikegame, T., Nakachi, Y. *et al.* (2019) Evaluation of the usefulness of saliva for DNA methylation analysis in cohort studies Neuropsychopharmacol Rep **39**, 301-305 10.1002/npr2.12075

1514. Nagura, H., Hokugo, J., andUeda, K. (2019) Long-Term Observation of the Safety and Effectiveness of Enzyme Replacement Therapy in Japanese Patients with Pompe Disease: Results From the Post-marketing Surveillance Neurol Ther **8**, 397-409 10.1007/s40120-019-00157-4

1515. Panagiotara, A., Skoufas, E., Chalikiopoulou, C., Tsermpini, E.-E., Bartsakoulia, M., Katsila, T. *et al.* (2019) Implementation of Genomic Medicine: An international perspective In Emery and Rimoin's Principles and Practice of Medical Genetics and Genomics, Elsevier, 369-380

1516. Sakurai, K., andMori, C. (2019) Current Findings in a Birth Cohort Study with Omics Analysis: Chiba Study of Mother and Child Health (C-MACH) In Pre-emptive Medicine: Public Health Aspects of Developmental Origins of

Health and Disease, Springer, 165-174

1517. Sata, F. F., H; Hanson, M (2019) Appendix B: Birth Cohorts and Consortia in the World In Pre-emptive Medicine: Public Health Aspects of Developmental Origins of Health and Disease, 211

1518. Someya, Y., Tamura, Y., Kaga, H., Nojiri, S., Shimada, K., Daida, H. *et al.* (2019) Skeletal muscle function and need for long-term care of urban elderly people in Japan (the Bunkyo Health Study): a prospective cohort study BMJ Open **9**, e031584 10.1136/bmjopen-2019-031584

1519. Toepfer, P., O'Donnell, K. J., Entringer, S., Garg, E., Heim, C. M., Lin, D. T. S. *et al.* (2019) Dynamic DNA methylation changes in the maternal oxytocin gene locus (OXT) during pregnancy predict postpartum maternal intrusiveness Psychoneuroendocrinology **103**, 156-162 10.1016/j.psyneuen.2019.01.013

1520. Ueta, M., Sotozono, C., Nishigaki, H., Ohsako, S., Yokoi, N., Mizushima, K. *et al.* (2019) Gene expression analysis of conjunctival epithelium of patients with Stevens-Johnson syndrome in the chronic stage BMJ Open Ophthalmol **4**, e000254 10.1136/bmjophth-2018-000254

1521. Xu, B., Zhang, Y. M., Yang, Y. W., Liu, Y. S., andFeng, J. F. (2019) Diagnostic performance of serum cystatin C and complement component 1q in lupus nephritis Arthritis Res Ther **21**, 267 10.1186/s13075-019-2065-x

1522. Yoo, S. K., Kim, C. U., Kim, H. L., Kim, S., Shin, J. Y., Kim, N. *et al.* (2019) NARD: whole-genome reference panel of 1779 Northeast Asians improves imputation accuracy of rare and low-frequency variants Genome Med **11**, 64 10.1186/s13073-019-0677-z

1523. Zhou, L., Zhang, P., Zhang, Z., Fan, L., Tang, S., Hu, K. *et al.* (2019) A Bibliometric Profile of Disaster Medicine Research from 2008 to 2017: A Scientometric Analysis Disaster Med Public Health Prep **13**, 165-172 10.1017/dmp.2018.11

1524. Zuo, W., Wen, L. P., Li, J., Mei, D., Fu, Q., andZhang, B. (2019) Oseltamivir induced Stevens-Johnson syndrome/toxic epidermal necrolysis-case report Medicine (Baltimore) **98**, e15553 10.1097/MD.0000000000015553

1525. 増元浩, 松木大造, 田中則之, 上杉幸嗣, 別府学, 岡本光弘 *et al.* (2019) 日米における疾患レジストリの臨床試験および新薬承認申請への利用比較 レギュラトリーサイエンス学会誌 **9**, 51-67,

1526. Adachi, T., Kainuma, K., Asano, K., Amagai, M., Arai, H., Ishii, K. J. *et al.* (2020) Strategic Outlook toward 2030: Japan's research for allergy and immunology - Secondary publication Allergol Int **69**, 561-570 10.1016/j.alit.2020.04.006

1527. Boulygina, E. A., Borisov, O. V., Valeeva, E. V., Semenova, E. A., Kostryukova, E. S., Kulemin, N. A. *et al.* (2020) Whole genome sequencing of elite athletes Biol Sport **37**, 295-304 10.5114/biolsport.2020.96272

1528. Dolzhenko, E., Bennett, M. F., Richmond, P. A., Trost, B., Chen, S., van Vugt, J. *et al.* (2020) ExpansionHunter Denovo: a computational method for locating known and novel repeat expansions in short-read sequencing data Genome Biol **21**, 102 10.1186/s13059-020-02017-z

1529. Fleming, M., McLay, J. S., Clark, D., King, A., Mackay, D. F., andPell, J. P. (2020) Health, educational and employment outcomes among children treated for a skin disorder: Scotland-wide retrospective record linkage cohort study of 766,244 children PLoS One **15**, e0243383 10.1371/journal.pone.0243383

1530. Gao, Y., Zhang, C., Yuan, L., Ling, Y., Wang, X., Liu, C. *et al.* (2020) PGG.Han: the Han Chinese genome database and analysis platform Nucleic Acids Res **48**, D971-D976 10.1093/nar/gkz829

1531. Harville, E. W., Breckner, D., Shu, T., Cooper, M., andBazzano, L. A. (2020) Establishing a three-generation prospective study: Bogalusa daughters J Dev Orig Health Dis **11**, 188-195 10.1017/S2040174419000357

1532. Hosoda, Y., Miyake, M., Meguro, A., Tabara, Y., Iwai, S., Ueda-Arakawa, N. *et al.* (2020) Keratoconus-

susceptibility gene identification by corneal thickness genome-wide association study and artificial intelligence IBM Watson Commun Biol **3**, 410 10.1038/s42003-020-01137-3

1533. Hosoi, A., Takizawa, T., Konagaya, R., andNaitoh, K. (2020) Prognostic medication: prediction by a macroscopic equation model for actual medical histories of illness with various recovery speeds Artificial Life and Robotics **25**, 189-198 10.1007/s10015-020-00596-5
1534. Jehan, F., Sazawal, S., Baqui, A. H., Nisar, M. I., Dhingra, U., Khanam, R. *et al.* (2020) Multiomics Characterization of Preterm Birth in Low- and Middle-Income Countries JAMA Netw Open **3**, e2029655 10.1001/jamanetworkopen.2020.29655
1535. Jeon, S., Bhak, Y., Choi, Y., Jeon, Y., Kim, S., Jang, J. *et al.* (2020) Korean Genome Project: 1094 Korean personal genomes with clinical information Sci Adv **6**, eaaz7835 10.1126/sciadv.aaz7835
1536. Kimble, L. P., Leslie, S., andCarlson, N. (2020) Metabolomics Research Conducted by Nurse Scientists: A Systematic Scoping Review Biol Res Nurs **22**, 436-448 10.1177/1099800420940041
1537. Konagaya, R., Takizawa, T., andNaitoh, K. (2020) A macroscopic theory for predicting catastrophic phenomena in both biological and mechanical chemical reactions Artificial Life and Robotics **25**, 178-188 10.1007/s10015-020-00595-6
1538. Krause, C., Geissler, C., Tackenberg, H., El Gammal, A. T., Wolter, S., Spranger, J. *et al.* (2020) Multi-layered epigenetic regulation of IRS2 expression in the liver of obese individuals with type 2 diabetes Diabetologia **63**, 2182-2193 10.1007/s00125-020-05212-6
1539. Kuo, C. W., Hwu, W. L., Chien, Y. H., Hsu, C., Hung, M. Z., Lin, I. L. *et al.* (2020) Frequency and spectrum of actionable pathogenic secondary findings in Taiwanese exomes Mol Genet Genomic Med **8**, e1455 10.1002/mgg3.1455
1540. Lancaster, S. M., Sanghi, A., Wu, S., andSnyder, M. P. (2020) A Customizable Analysis Flow in Integrative Multi-Omics Biomolecules **10**, 10.3390/biom10121606
1541. Liang, K. H., Lu, Y. H., Niu, C. W., Chang, S. K., Chen, Y. R., Cheng, C. Y. *et al.* (2020) The Fabry disease-causing mutation, GLA IVS4+919G>A, originated in Mainland China more than 800 years ago Journal of human genetics **65**, 619-625 10.1038/s10038-020-0745-7
1542. Liang, Y., Ke, X., Xiao, Z., Zhang, Y., Chen, Y., Li, Y. *et al.* (2020) Untargeted Metabolomic Profiling Using UHPLC-QTOF/MS Reveals Metabolic Alterations Associated with Autism Biomed Res Int **2020**, 6105608 10.1155/2020/6105608
1543. Logue, M. W., Miller, M. W., Wolf, E. J., Huber, B. R., Morrison, F. G., Zhou, Z. *et al.* (2020) An epigenome-wide association study of posttraumatic stress disorder in US veterans implicates several new DNA methylation loci Clin Epigenetics **12**, 46 10.1186/s13148-020-0820-0
1544. Manolio, T. A., Goodhand, P., andGinsburg, G. (2020) The International Hundred Thousand Plus Cohort Consortium: integrating large-scale cohorts to address global scientific challenges Lancet Digit Health **2**, e567-e568 10.1016/S2589-7500(20)30242-9
1545. Melis, E., Gallo, E., di Martino, S., Gallina, F. T., Laquintana, V., Casini, B. *et al.* (2020) Thymic Epithelial Tumors as a Model of Networking: Development of a Synergistic Strategy for Clinical and Translational Research Purposes Front Oncol **10**, 922 10.3389/fonc.2020.00922
1546. Merah-Mourah, F., Cohen, S. O., Charron, D., Mooney, N., andHaziot, A. (2020) Identification of Novel Human Monocyte Subsets and Evidence for Phenotypic Groups Defined by Interindividual Variations of Expression of Adhesion Molecules Sci Rep **10**, 4397 10.1038/s41598-020-61022-1

1547. Mishima, E., Mori, T., Nakajima, Y., Toyohara, T., Kikuchi, K., Oikawa, Y. *et al.* (2020) HPRT-related hyperuricemia with a novel p.V35M mutation in HPRT1 presenting familial juvenile gout CEN Case Rep **9**, 210-214 10.1007/s13730-020-00459-9

1548. Morrison, M., Teare, H., Bertier, G., Buchanan, J., Bylstra, Y., Gaff, C. *et al.* (2020) Implications of secondary findings for clinical contexts In Secondary Findings in Genomic Research, Chapter 8, 155-201

1549. Mudla, A., Jiang, Y., Arimoto, K. I., Xu, B., Rajesh, A., Ryan, A. P. *et al.* (2020) Cell-cycle-gated feedback control mediates desensitization to interferon stimulation Elife **9**, 10.7554/eLife.58825

1550. Nagai, K., Uranbileg, B., Chen, Z., Fujioka, A., Yamazaki, T., Matsumoto, Y. *et al.* (2020) Identification of novel biomarkers of hepatocellular carcinoma by high-definition mass spectrometry: Ultrahigh-performance liquid chromatography quadrupole time-of-flight mass spectrometry and desorption electrospray ionization mass spectrometry imaging Rapid Commun Mass Spectrom **34 Suppl 1**, e8551 10.1002/rcm.8551

1551. Oshikata, C., Watanabe, M., Ishida, M., Kobayashi, S., Kubosaki, A., Yamazaki, A. *et al.* (2020) Increase in asthma prevalence in adults in temporary housing after the Great East Japan earthquake International Journal of Disaster Risk Reduction **50**, 101696 ARTN 101696
10.1016/j.ijdrr.2020.101696

1552. Rocha, C. S., Secolin, R., Rodrigues, M. R., Carvalho, B. S., andLopes-Cendes, I. (2020) The Brazilian Initiative on Precision Medicine (BIPMed): fostering genomic data-sharing of underrepresented populations NPJ Genom Med **5**, 42 10.1038/s41525-020-00149-6

1553. Sawada, N., Iwasaki, M., Yamaji, T., Goto, A., Shimazu, T., Inoue, M. *et al.* (2020) The Japan Public Health Center-based Prospective Study for the Next Generation (JPHC-NEXT): Study Design and Participants J Epidemiol **30**, 46-54 10.2188/jea.JE20180182

1554. Seidualy, M., Blazyte, A., Jeon, S., Bhak, Y., Jeon, Y., Kim, J. *et al.* (2020) Decoding a highly mixed Kazakh genome Hum Genet **139**, 557-568 10.1007/s00439-020-02132-8

1555. Singh, K., Bijarnia-Mahay, S., Ramprasad, V. L., Puri, R. D., Nair, S., Sharda, S. *et al.* (2020) NGS-based expanded carrier screening for genetic disorders in North Indian population reveals unexpected results - a pilot study BMC Med Genet **21**, 216 10.1186/s12881-020-01153-4

1556. Svensson, D., Rentoft, M., Dahlin, A. M., Lundholm, E., Olason, P. I., Sjodin, A. *et al.* (2020) A whole-genome sequenced control population in northern Sweden reveals subregional genetic differences PLoS One **15**, e0237721 10.1371/journal.pone.0237721

1557. Tomiyoshi, K., Sato, H., Tominaga, K., Kawata, Y., Okamoto, D., Kakuta, Y. *et al.* (2020) Rare Genotype of His/His in NUDT15 Codon 139 and Thiopurine-associated Adverse Events in a Case of Ulcerative Colitis Intern Med **59**, 1611-1613 10.2169/internalmedicine.4261-19

1558. Yamaguchi, C., Ebara, T., Futamura, M., Ohya, Y., andAsano, M. (2020) Childhood allergy symptoms increase the risk of behavioral problems: A cross-sectional study Authorea Preprints,

1559. Yamasaki, M., Makino, T., Khor, S. S., Toyoda, H., Miyagawa, T., Liu, X. *et al.* (2020) Sensitivity to gene dosage and gene expression affects genes with copy number variants observed among neuropsychiatric diseases BMC Med Genomics **13**, 55 10.1186/s12920-020-0699-9

1560. Yoshihara, H., Sugiura-Ogasawara, M., Ozawa, F., Kitaori, T., Ozaki, Y., Aoki, K. *et al.* (2020) Polo-like kinase 4 and Stromal antigen 3 are not associated with recurrent pregnancy loss caused by embryonic aneuploidy Hum Genome Var **7**, 18 10.1038/s41439-020-0106-2

1561. Zhao, S., Zhang, C., Mu, J., Zhang, H., Yao, W., Ding, X. *et al.* (2020) All-in-one sequencing: an improved library

preparation method for cost-effective and high-throughput next-generation sequencing Plant Methods **16**, 74 10.1186/s13007-020-00615-3

1562. 尾崎るりこ, 中村篤大, 田中桂輔, 宮部泉, 若月智和, 菊地茉莉 *et al.* (2020) BRCA1: c. 852G> C p.(Gln284His) 遺伝性腫瘍 **20**, 41-42,

1563. 堀田喜裕, and 細野克博 (2020) 難治性小児眼疾患症例の遺伝学的診断に関する研究 In 厚生労働科学研究費補助金 分担研究報告書, 20210420 Ed.

1564. Akdel, M., van de Geest, H., Schijlen, E., van Rijswijck, I. M. H., Smid, E. J., Sanchez-Perez, G. *et al.* (2021) Signal-based optical map alignment PLoS One **16**, e0253102 10.1371/journal.pone.0253102

1565. Asteggiano, A., Franceschi, P., Zorzi, M., Aigotti, R., Dal Bello, F., Baldassarre, F. *et al.* (2021) HPLC-HRMS Global Metabolomics Approach for the Diagnosis of "Olive Quick Decline Syndrome" Markers in Olive Trees Leaves Metabolites **11**, 10.3390/metabo11010040

1566. Breton, G., Johansson, A. C. V., Sjodin, P., Schlebusch, C. M., andJakobsson, M. (2021) Comparison of sequencing data processing pipelines and application to underrepresented African human populations BMC Bioinformatics **22**, 488 10.1186/s12859-021-04407-x

1567. Chen, Y., Wang, J., Zhou, H., Huang, Z., Qian, L., andShi, W. (2021) Identification of Prognostic Risk Model Based on DNA Methylation-Driven Genes in Esophageal Adenocarcinoma Biomed Res Int **2021**, 6628391 10.1155/2021/6628391

1568. da Rocha, J. E. B., Othman, H., Botha, G., Cottino, L., Twesigomwe, D., Ahmed, S. *et al.* (2021) The Extent and Impact of Variation in ADME Genes in Sub-Saharan African Populations Front Pharmacol **12**, 634016 10.3389/fphar.2021.634016

1569. Drljaca, T., Zukic, B., Kovacevic, V., Gemovic, B., Klaassen-Ljubicic, K., Perovic, V. *et al.* (2021) The first insight into the genetic structure of the population of modern Serbia Sci Rep **11**, 13995 10.1038/s41598-021-93129-4

1570. Emwas, A.-H., Szczepski, K., McKay, R. T., Asfour, H., Chang, C.-k., Lachowicz, J. *et al.* (2021) Pharmacometabolomics: A New Horizon in Personalized Medicine In Metabolomics-Methodology and Applications in Medical Sciences and Life Sciences, IntechOpen, Chapter 3, 31-66

1571. Fujimoto, A., Wong, J. H., Yoshii, Y., Akiyama, S., Tanaka, A., Yagi, H. *et al.* (2021) Whole-genome sequencing with long reads reveals complex structure and origin of structural variation in human genetic variations and somatic mutations in cancer Genome Med **13**, 65 10.1186/s13073-021-00883-1

1572. Huang, C., Fan, Z., Han, D., Johnston, L. J., Ma, X., andWang, F. (2021) Pyrroloquinoline quinone regulates the redox status in vitro and in vivo of weaned pigs via the Nrf2/HO-1 pathway J Anim Sci Biotechnol **12**, 77 10.1186/s40104-021-00595-x

1573. Huang, H. Y., Li, J., Tang, Y., Huang, Y. X., Chen, Y. G., Xie, Y. Y. *et al.* (2021) MethHC 2.0: information repository of DNA methylation and gene expression in human cancer Nucleic Acids Res **49**, D1268-D1275 10.1093/nar/gkaa1104

1574. Inoue, Y., Hasebe, Y., Igarashi, T., Kawagishi-Hotta, M., Okuno, R., Yamada, T. *et al.* (2021) Search for genetic loci involved in the constitution and skin type of a Japanese women using a genome-wide association study Exp Dermatol **30**, 1787-1793 10.1111/exd.14430

1575. Jain, A., Bhoyar, R. C., Pandhare, K., Mishra, A., Sharma, D., Imran, M. *et al.* (2021) IndiGenomes: a comprehensive resource of genetic variants from over 1000 Indian genomes Nucleic Acids Res **49**, D1225-D1232 10.1093/nar/gkaa923

1576. Jain, A., Sharma, D., Bajaj, A., Gupta, V., andScaria, V. (2021) Founder variants and population genomes-Toward precision medicine In Advances in Genetics, 2021/03/02 Ed. Chapter 4, 121-152

1577. Jain, N., Virmani, D., andAbraham, A. (2021) Tsunami in the last 15 years: a bibliometric analysis with a detailed overview and future directions Natural Hazards **106**, 139-172 10.1007/s11069-020-04454-2

1578. Jeon, Y., Jeon, S., Blazyte, A., Kim, Y. J., Lee, J. J., Bhak, Y. *et al.* (2021) Welfare Genome Project: A Participatory Korean Personal Genome Project With Free Health Check-Up and Genetic Report Followed by Counseling Front Genet **12**, 633731 10.3389/fgene.2021.633731

1579. Jia, L., andFan, W. (2021) Medical Sports Data Privacy Protection Method Based on Legal Risk Control J Healthc Eng **2021**, 6630429 10.1155/2021/6630429

1580. Kalogeropoulos, A., andPikouli, A. (2021) Natural Disasters: Medical Management In Emergency Medicine, Trauma and Disaster Management, Springer, 433-449

1581. Kang, H. J., Lee, H. Y., Kim, K. T., Kim, J. W., Lee, J. Y., Kim, S. W. *et al.* (2021) Genetic Differences between Physical Injury Patients With and Without Post-traumatic Syndrome: Focus on Secondary Findings and Potential Variants Revealed by Whole Exome Sequencing Clin Psychopharmacol Neurosci **19**, 683-694 10.9758/cpn.2021.19.4.683

1582. Katsuki, M., Kakizawa, Y., Nishikawa, A., Yamamoto, Y., andUchiyama, T. (2021) Postsurgical functional outcome prediction model using deep learning framework (Prediction One, Sony Network Communications Inc.) for hypertensive intracerebral hemorrhage Surg Neurol Int **12**, 203 10.25259/SNI_222_2021

1583. Kausthubham, N., Shukla, A., Gupta, N., Bhavani, G. S., Kulshrestha, S., Das Bhowmik, A. *et al.* (2021) A data set of variants derived from 1455 clinical and research exomes is efficient in variant prioritization for early-onset monogenic disorders in Indians Hum Mutat **42**, e15-e61 10.1002/humu.24172

1584. Keller, W., Vogel, M., Prenzel, F., Genuneit, J., Jurkutat, A., Hilbert, C. *et al.* (2021) Atopic diseases in children and adolescents are associated with behavioural difficulties BMC Pediatr **21**, 197 10.1186/s12887-021-02663-7

1585. Kim, M., Xi, H., andPark, J. (2021) Genome-wide comparative analyses of GATA transcription factors among 19 Arabidopsis ecotype genomes: Intraspecific characteristics of GATA transcription factors PLoS One **16**, e0252181 10.1371/journal.pone.0252181

1586. Liu, D. X., Rajaby, R., Wei, L. L., Zhang, L., Yang, Z. Q., Yang, Q. Y. *et al.* (2021) Calling large indels in 1047 Arabidopsis with IndelEnsembler Nucleic Acids Res **49**, 10879-10894 10.1093/nar/gkab904

1587. Luger, T., Adaskevich, U., Anfilova, M., Dou, X., Murashkin, N. N., Namazova-Baranova, L. *et al.* (2021) Practical algorithm to inform clinical decision-making in the topical treatment of atopic dermatitis J Dermatol **48**, 1139-1148 10.1111/1346-8138.15921

1588. Mao, Y., Huang, P., Wang, Y., Wang, M., Li, M. D., andYang, Z. (2021) Genome-wide methylation and expression analyses reveal the epigenetic landscape of immune-related diseases for tobacco smoking Clin Epigenetics **13**, 215 10.1186/s13148-021-01208-0

1589. Meister, I., Zhang, P., Sinha, A., Skold, C. M., Wheelock, A. M., Izumi, T. *et al.* (2021) High-Precision Automated Workflow for Urinary Untargeted Metabolomic Epidemiology Anal Chem **93**, 5248-5258 10.1021/acs.analchem.1c00203

1590. Miyaguti, N., Chiocchetti, G. M. E., Salgado, C. M., Lopes-Aguiar, L., Viana, L. R., Blanchard, L. *et al.* (2021) Walker-256 Tumour-Induced Cachexia Altered Liver Metabolomic Profile and Function in Weanling and Adult Rats Metabolites **11**, 10.3390/metabo11120831

1591. Nakamura, H., Kikkawa, M., andMurata, T. (2021) Technical development and sharing of high-resolution cryo-electron microscopes Biophys Physicobiol **18**, 265-266 10.2142/biophysico.bppb-v18.030
1592. Nielsen, J. E., Maltesen, R. G., Havelund, J. F., Faergeman, N. J., Gotfredsen, C. H., Vestergard, K. *et al.* (2021) Characterising Alzheimer's disease through integrative NMR- and LC-MS-based metabolomics Metabol Open **12**, 100125 10.1016/j.metop.2021.100125
1593. Oshikata, C., Watanabe, M., Ishida, M., Kobayashi, S., Hashimoto, K., Kobayashi, N. *et al.* (2021) Association between Temporary Housing Habitation after the 2011 Japan Earthquake and Mite Allergen Sensitization and Asthma Development Int Arch Allergy Imm **182**, 949-961 10.1159/000515870
1594. Oshikata, C., Watanabe, M., Ishida, M., Kobayashi, S., Hashimoto, K., Kobayashi, N. *et al.* (2021) Mite avoidance decreased mite-specific IgE levels and ameliorated asthma symptoms in subjects who lived in temporary housing after natural disasters Allergologia Et Immunopathologia **49**, 171-179 10.15586/aei.v49i4.240
1595. Pathak, G. A., Wendt, F. R., De Lillo, A., Nunez, Y. Z., Goswami, A., De Angelis, F. *et al.* (2021) Epigenomic Profiles of African-American Transthyretin Val122Ile Carriers Reveals Putatively Dysregulated Amyloid Mechanisms Circ Genom Precis Med **14**, e003011 10.1161/CIRCGEN.120.003011
1596. Pena-Chilet, M., Roldan, G., Perez-Florido, J., Ortuno, F. M., Carmona, R., Aquino, V. *et al.* (2021) CSVS, a crowdsourcing database of the Spanish population genetic variability Nucleic Acids Res **49**, D1130-D1137 10.1093/nar/gkaa794
1597. Roux, J., Zeghidi, M., Villar, S., andKozlakidis, Z. (2021) Biosafety and biobanking: Current understanding and knowledge gaps Biosaf Health **3**, 244-248 10.1016/j.bsheal.2021.06.003
1598. Saito, R., Sugimoto, M., Hirayama, A., Soga, T., Tomita, M., andTakebayashi, T. (2021) Quality Assessment of Untargeted Analytical Data in a Large-Scale Metabolomic Study J Clin Med **10**, 10.3390/jcm10091826
1599. Sasaki, T., Abe, Y., Takayama, M., Adachi, T., Okano, H., Hirose, N. *et al.* (2021) Association among extracellular superoxide dismutase genotype, plasma concentration, and comorbidity in the very old and centenarians Sci Rep **11**, 8539 10.1038/s41598-021-87982-6
1600. Shi, S., Qian, Q., Yu, S., Wang, Q., Wang, J., Zeng, J. *et al.* (2021) RefRGim: an intelligent reference panel reconstruction method for genotype imputation with convolutional neural networks Brief Bioinform **22**, 10.1093/bib/bbab326
1601. Shimomura, H., Tokunaga, S., Taniguchi, N., Inoue, K., Okuda, M., Kato, T. *et al.* (2021) Emotional and behavioral problems in pediatric patients with migraine and tension-type headache Brain Dev **43**, 826-832 10.1016/j.braindev.2021.04.004
1602. Stranneheim, H., Lagerstedt-Robinson, K., Magnusson, M., Kvarnung, M., Nilsson, D., Lesko, N. *et al.* (2021) Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients Genome Med **13**, 40 10.1186/s13073-021-00855-5
1603. Suvakov, M., Panda, A., Diesh, C., Holmes, I., andAbyzov, A. (2021) CNVpytor: a tool for copy number variation detection and analysis from read depth and allele imbalance in whole-genome sequencing Gigascience **10**, 10.1093/gigascience/giab074
1604. Takeuchi, K., Naito, M., Kawai, S., Tsukamoto, M., Kadomatsu, Y., Kubo, Y. *et al.* (2021) Study Profile of the Japan Multi-institutional Collaborative Cohort (J-MICC) Study J Epidemiol **31**, 660-668 10.2188/jea.JE20200147
1605. Watanabe, O., Narita, N., Katsuki, M., Ishida, N., Cai, S., Otomo, H. *et al.* (2021) Prediction Model of Deep Learning for Ambulance Transports in Kesennuma City by Meteorological Data Open Access Emerg Med **13**, 23-32 10.2147/OAEM.S293551

1606. Wei, C. Y., Yang, J. H., Yeh, E. C., Tsai, M. F., Kao, H. J., Lo, C. Z. *et al.* (2021) Genetic profiles of 103,106 individuals in the Taiwan Biobank provide insights into the health and history of Han Chinese NPJ Genom Med **6**, 10 10.1038/s41525-021-00178-9

1607. Wiegand, A., Blickle, A., Bruckmann, C., Weller, S., Nieratschker, V., andPlewnia, C. (2021) Dynamic DNA Methylation Changes in the COMT Gene Promoter Region in Response to Mental Stress and Its Modulation by Transcranial Direct Current Stimulation Biomolecules **11**, 10.3390/biom11111726

1608. Yadav, A., Kumari, R., Gautam, B., andKatara, P. (2021) PHARMACOGENOMICS: CURRENT TRENDS AND FUTURE POSSIBILITIES In Recent Advances in Computer Aided Drug Designing, Nova Science Publishers Inc,, United States Chapter 11, 249-275

1609. Yamaguchi, C., Ebara, T., Futamura, M., Ohya, Y., andAsano, M. (2021) Do allergic clinical manifestations increase the risk of behavioral problems in children? A cross-sectional study Pediatric Allergy and Immunology,

1610. Yang, T., andPei, D. (2021) Association of cystatin C levels with metabolic syndrome incidence: a nested case-control study with propensity score matching J Int Med Res **49**, 300060520986311 10.1177/0300060520986311

1611. You, M. P., Akhatar, J., Mittal, M., Barbetti, M. J., Maina, S., andBanga, S. S. (2021) Comparative analysis of draft genome assemblies developed from whole genome sequences of two Hyaloperonospora brassicae isolate samples differing in field virulence on Brassica napus Biotechnol Rep (Amst) **31**, e00653 10.1016/j.btre.2021.e00653

1612. Zhang, J. Y., Roberts, H., Flores, D. S. C., Cutler, A. J., Brown, A. C., Whalley, J. P. *et al.* (2021) Using de novo assembly to identify structural variation of eight complex immune system gene regions PLoS Comput Biol **17**, e1009254 10.1371/journal.pcbi.1009254

1613. 弓気田美香 (2021) 日本における小児アレルギー性疾患をもつ子どもの QOL や精神健康状態に関する文献検討 東京有明医療大学雑誌 **13**, 43-46,

1614. 高井淳 (2021) 東北メディカル・メガバンクの宮城県地域住民コホート情報と統合データベースを活用した転写因子 GATA2 の遺伝子多型と感染症既往歴に関する調査研究 In 公衆衛生情報みやぎ, 宮城県公衆衛生協会, 仙台

1615. Cao, Y., Zhao, Y. L., Wang, Q., Zhang, J., Ng, S. X., andHanzo, L. (2022) The Evolution of Quantum Key Distribution Networks: On the Road to the Qinternet Ieee Commun Surv Tut **24**, 839-894 10.1109/Comst.2022.3144219

1616. Chi, J. H., andLee, B. J. (2022) Risk factors for hypertension and diabetes comorbidity in a Korean population: A cross-sectional study PLoS One **17**, e0262757 10.1371/journal.pone.0262757

1617. Desai, S., Mishra, R., Ahmad, S., Hait, S., Joshi, A., andDutt, A. (2022) TMC-SNPdb 2.0: an ethnic-specific database of Indian germline variants Database (Oxford) **2022**, 10.1093/database/baac029

1618. Diaz-de Usera, A., Rubio-Rodriguez, L. A., Munoz-Barrera, A., Lorenzo-Salazar, J. M., Guillen-Guio, B., Jaspez, D. *et al.* (2022) Developing CIRdb as a catalog of natural genetic variation in the Canary Islanders Sci Rep **12**, 16132 10.1038/s41598-022-20442-x

1619. Duong, H. T. T., Suzuki, H., Katagiri, S., Shibata, M., Arai, M., andYura, K. (2022) Computational study of the impact of nucleotide variations on highly conserved proteins: In the case of actin Biophys Physicobiol **19**, e190025 10.2142/biophysico.bppb-v19.0025

1620. Fujiwara, M., Hashimoto, H., Doi, K., Kujiraoka, M., Tanizawa, Y., Ishida, Y. *et al.* (2022) Secure secondary utilization system of genomic data using quantum secure cloud Sci Rep **12**, 18530 10.1038/s41598-022-22804-x

1621. Giri, K., Bisht, V. S., Maity, S., andAmbatipudi, K. (2022) Integrated Omics Technology for Basic and Clinical Research In Biotechnological Advances for Microbiology, Molecular Biology, and Nanotechnology: An Interdisciplinary Approach to the Life Sciences,

1622. Hattori, K., andHattori, R. (2022) Predicted Number of Pregnant Women in Aichi Prefecture, Japan: Estimation by Machine Learning Database Construction for Disaster Preparation Disaster Med Public Health Prep **16**, 940-948 10.1017/dmp.2020.417

1623. He, Z., Lv, Y., Zheng, S., Pu, Y., Lin, Q., Zhou, H. *et al.* (2022) Association of COVID-19 Lockdown With Gestational Diabetes Mellitus Front Endocrinol (Lausanne) **13**, 824245 10.3389/fendo.2022.824245

1624. Jacobsen, J. O. B., Baudis, M., Baynam, G. S., Beckmann, J. S., Beltran, S., Buske, O. J. *et al.* (2022) The GA4GH Phenopacket schema defines a computable representation of clinical data Nat Biotechnol **40**, 817-820 10.1038/s41587-022-01357-4

1625. Lee, S., Kim, J., Kwon, Y., Kim, T., andCho, S. (2022) Privacy Preservation in Patient Information Exchange Systems Based on Blockchain: System Design Study J Med Internet Res **24**, e29108 10.2196/29108

1626. McHugh, M. C., andDiercks, D. B. (2022) Interpreting High-Sensitive Troponins in Patients with Hypertension Curr Hypertens Rep **24**, 349-352 10.1007/s11906-022-01197-2

1627. Muroya, T., Satoh, M., Murakami, T., Nakayama, S., Asayama, K., Hirose, T. *et al.* (2022) Association between urinary sodium-to-potassium ratio and home blood pressure and ambulatory blood pressure: the Ohasama study J Hypertens **40**, 862-869 10.1097/HJH.0000000000003087

1628. Nagai, M., Hikichi, H., Shiba, K., Kondo, K., Kawachi, I., andAida, J. (2022) Long-Term Trend in the Association Between Disaster Damage and Happiness Before and After the Great East Japan Earthquake Int J Public Health **67**, 1604901 10.3389/ijph.2022.1604901

1629. Nishida, N., Sugiyama, M., Kawai, Y., Naka, I., Iwamoto, N., Suzuki, T. *et al.* (2022) Genetic association of IL17 and the importance of ABO blood group antigens in saliva to COVID-19 Sci Rep **12**, 3854 10.1038/s41598-022-07856-3

1630. Okuno, R., Inoue, Y., Hasebe, Y., Igarashi, T., Kawagishi-Hotta, M., Yamada, T. *et al.* (2022) Genome-wide association studies in Japanese women identified genetic loci associated with wrinkles and sagging Exp Dermatol **31**, 1411-1420 10.1111/exd.14612

1631. Oshikata, C., Watanabe, M., Hashimoto, K., Yamazaki, A., Kobayashi, N., Konuma, R. *et al.* (2022) Mite allergen avoidance decreases allergic symptoms in children in Ishinomaki city of Japan after natural disasters Allergol Immunopathol (Madr) **50**, 23-32 10.15586/aei.v50i2.483

1632. Rodosthenous, R. S., Niemi, M. E. K., Kallio, L., Perala, M., Terho, P., Knopp, T. *et al.* (2022) Recontacting biobank participants to collect lifestyle, behavioural and cognitive information via online questionnaires: lessons from a pilot study within FinnGen BMJ Open **12**, e064695 10.1136/bmjopen-2022-064695

1633. Seo, J. Y., You, S. W., Shin, J. G., Kim, Y., Park, S. G., Won, H. H. *et al.* (2022) GWAS Identifies Multiple Genetic Loci for Skin Color in Korean Women J Invest Dermatol **142**, 1077-1084 10.1016/j.jid.2021.08.440

1634. Shibamoto, J., Kubota, T., Ohashi, T., Konishi, H., Shiozaki, A., Fujiwara, H. *et al.* (2022) Glucose variability and predicted cardiovascular risk after gastrectomy Surg Today **52**, 1634-1644 10.1007/s00595-022-02496-6

1635. Suhre, K., Stephan, N., Zaghlool, S., Triggle, C. R., Robinson, R. J., Evans, A. M. *et al.* (2022) Matching Drug Metabolites from Non-Targeted Metabolomics to Self-Reported Medication in the Qatar Biobank Study Metabolites **12**, 249 10.3390/metabo12030249

1636. Syafiq, A., Fikawati, S., andGemily, S. C. (2022) Household food security during the COVID-19 pandemic in

urban and semi-urban areas in Indonesia J Health Popul Nutr **41**, 4 10.1186/s41043-022-00285-y

1637. Takada, T., Fukuta, K., Usuda, D., Kushida, T., Kondo, S., Kawamoto, S. *et al.* (2022) MoG+: a database of genomic variations across three mouse subspecies for biomedical research Mamm Genome **33**, 31-43 10.1007/s00335-021-09933-w

1638. Tang, R., Han, C., Yin, R., Zhu, P., Zhu, L., Lu, Y. *et al.* (2022) Quality Control of DNA Extracted from All-Cell Pellets After Cryopreservation for More Than 10 Years Biopreservation and Biobanking **20**, 211-216, https://www.liebertpub.com/doi/pdf/10.1089/bio.2021.0052?download=true

1639. Timms, K., Holder, B., Day, A., McLaughlin, J., Forbes, K. A., andWestwood, M. (2022) Watermelon-Derived Extracellular Vesicles Influence Human Ex Vivo Placental Cell Behavior by Altering Intestinal Secretions Mol Nutr Food Res **66**, e2200013 10.1002/mnfr.202200013

1640. Toyama, N., Ekuni, D., Yokoi, A., Fukuhara, D., Islam, M. M., Sawada, N. *et al.* (2022) Features of the oral microbiome in Japanese elderly people with 20 or more teeth and a non-severe periodontal condition during periodontal maintenance treatment: A cross-sectional study Front Cell Infect Microbiol **12**, 957890 10.3389/fcimb.2022.957890

1641. Wallace, K., Bowles, T., Griffin, A., Robinson, R., Solis, L., Railey, T. *et al.* (2022) Evidence of Anxiety, Depression and Learning Impairments following Prenatal Hypertension Behav Sci (Basel) **12**, 53 10.3390/bs12020053

1642. Wang, P., Sun, X., Miao, Q., Mi, H., Cao, M., Zhao, S. *et al.* (2022) Novel genetic associations with five aesthetic facial traits: A genome-wide association study in the Chinese population Front Genet **13**, 967684 10.3389/fgene.2022.967684

1643. Woltamo, D. D., Meskele, M., Workie, S. B., andBadacho, A. S. (2022) Determinants of fetal macrosomia among live births in southern Ethiopia: a matched case-control study BMC Pregnancy Childbirth **22**, 465 10.1186/s12884-022-04734-8

1644. Xu, Z. M., Rueger, S., Zwyer, M., Brites, D., Hiza, H., Reinhard, M. *et al.* (2022) Using population-specific add-on polymorphisms to improve genotype imputation in underrepresented populations PLoS Comput Biol **18**, e1009628 10.1371/journal.pcbi.1009628

1645. Yagasaki, K., Mabuchi, A., Higashino, T., Hao Wong, J., Nishida, N., Fujimoto, A. *et al.* (2022) Practical forensic use of kinship determination using high-density SNP profiling based on a microarray platform, focusing on low-quantity DNA Forensic Sci Int Genet **61**, 102752 10.1016/j.fsigen.2022.102752

1646. Yang, W., Han, N., Jiao, M., Chang, X., Liu, J., Zhou, Q. *et al.* (2022) Maternal diet quality during pregnancy and its influence on low birth weight and small for gestational age: a birth cohort in Beijing, China Br J Nutr 1-10 10.1017/S0007114522000708

1647. Yang, X. F., Zhao, X. X., Qu, S. F., Jia, P., Wang, B., Gao, S. H. *et al.* (2022) Haplotype-resolved Chinese male genome assembly based on high-fidelity sequencing Fundamental Research **2**, 946-953 10.1016/j.fmre.2022.02.005

1648. Zhang, K., Siziba, L. P., Suo, N. J., Rothenbacher, D., andGenuneit, J. (2022) Breastfeeding duration is positively associated with decreased smoking relapse in the postpartum period Midwifery **108**, 103289 10.1016/j.midw.2022.103289

1649. Abdullah, A. H., Nathan, A. M., Jayanath, S., Kwan, Z., Azanan, M. S., Hng, S. Y. *et al.* (2023) Poor sleep quality in children with atopic dermatitis and its effects on behavior: A multicenter cross-sectional study from a low-middle-income country Pediatrics international : official journal of the Japan Pediatric Society **65**, e15473 10.1111/ped.15473

1650. Abo Asy, A. S., Ahmed, M. E., Attia, S. M., andYoussef, M. (2023) High Sensitivity Cardiac Troponin T in Patients with Type 2 Diabetes Mellitus, Relation to Cardiac Metabolic Risk Factors: Hypertension and Truncal Obesity The Egyptian Journal of Hospital Medicine **90**, 2149-2153,

1651. Akaishi, T., Tarasawa, K., Fushimi, K., Hamada, H., Saito, M., Kobayashi, N. *et al.* (2023) Risk Factors Associated With Peripartum Suicide Attempts in Japan JAMA Netw Open **6**, e2250661 10.1001/jamanetworkopen.2022.50661

1652. AlOmari, H., Alkhateeb, A., andHammo, B. (2023) Multi-Omics Databases In Machine Learning Methods for Multi-Omics Data Integration, Springer, 151-166

1653. Bizkarguenaga, M., Gil-Redondo, R., Bruzzone, C., Bernardo-Seisdedos, G., Laín, A., González-Valle, B. *et al.* (2023) Prospective Metabolomic Studies in Precision Medicine: The AKRIBEA Project In Handb Exp Pharmacol, 275-297

1654. Dekeyser, T., Genin, E., andHerzig, A. F. (2023) Opening the Black Box of Imputation Software to Study the Impact of Reference Panel Composition on Performance Genes (Basel) **14**, 410 10.3390/genes14020410

1655. Duran-Pinedo, A. E., Solbiati, J., Teles, F., andFrias-Lopez, J. (2023) Subgingival host-microbiome metatranscriptomic changes following scaling and root planing in grade II/III periodontitis J Clin Periodontol **50**, 316-330 10.1111/jcpe.13737

1656. Enomoto, T., Okamoto, A., Kim, J. H., Lai, C. H., Wu, X., andKim, Y. M. (2023) East Asian Gynecologic Oncology Trial Group (EAGOT): founding history and future perspective J Gynecol Oncol **34**, e86 10.3802/jgo.2023.34.e86

1657. Fabre-Estremera, B., Buno-Soto, A., Garcia-Esquinas, E., Cabanas-Sanchez, V., Martinez-Gomez, D., Rodriguez-Artalejo, F. *et al.* (2023) Device-measured movement behaviors and cardiac biomarkers in older adults without major cardiovascular disease: the Seniors-ENRICA-2 study Eur Rev Aging Phys A **20**, 5 10.1186/s11556-023-00313-8

1658. Ferreira, A. L. L., Freitas-Costa, N., da Silva Rosa Freire, S., Figueiredo, A. C. C., Padilha, M., Alves-Santos, N. H. *et al.* (2023) Association of pre-pregnancy maternal overweight/obesity and dietary intake during pregnancy with the concentrations of persistent organic pollutants in the human milk of women from Rio de Janeiro, Brazil Environ Sci Pollut Res Int **30**, 44999-45014 10.1007/s11356-023-25308-x

1659. Forray, A., Gunter-Riley, R. G., Maltz, C., andWaters, A. J. (2023) Pilot study of attentional retraining for postpartum smoking relapse Front Psychiatry **14**, 1231702 10.3389/fpsyt.2023.1231702

1660. Grammatikopoulou, M. G., Nigdelis, M. P., Haidich, A. B., Kyrezi, M., Ntine, H., Papaioannou, M. *et al.* (2023) Diet Quality and Nutritional Risk Based on the FIGO Nutrition Checklist among Greek Pregnant Women: A Cross-Sectional Routine Antenatal Care Study Nutrients **15**, 2019 10.3390/nu15092019

1661. Han, X., Pan, S., Liu, J., Ding, X., Lin, X., Wang, D. *et al.* (2023) Novel loci for ocular axial length identified through extreme-phenotype genome-wide association study in Chinese populations Br J Ophthalmol 10.1136/bjo-2023-323596

1662. Harville, E. W., andNorthland, B. (2023) Natural Disaster Epidemiology and Reproductive Health Current Epidemiology Reports 10.1007/s40471-023-00329-9

1663. Hiromoto, K., Yamada, T., Tsuchiya, M., Kawame, H., Nanba, E., Goto, Y. *et al.* (2023) Challenges of secondary finding disclosure in genomic medicine in rare diseases: A nation-wide survey of Japanese facilities outsourcing comprehensive genetic testing Journal of human genetics **68**, 1-9 10.1038/s10038-022-01084-3

1664. Jiang, C., Melles, R. B., Yin, J., Fan, Q., Guo, X., Cheng, C. Y. *et al.* (2023) A multiethnic genome-wide analysis of 19,420 individuals identifies novel loci associated with axial length and shared genetic influences with

refractive error and myopia Front Genet **14**, 1113058 10.3389/fgene.2023.1113058

1665. Li, Z., Jiang, X., Fang, M., Bai, Y., Liu, S., Huang, S. *et al.* (2023) CMDB: the comprehensive population genome variation database of China Nucleic Acids Res **51**, D890-D895 10.1093/nar/gkac638

1666. Liu, L., Cheng, Y. T., Xu, A., andCheung, B. M. Y. (2023) Association between high sensitivity cardiac troponin and mortality risk in the non-diabetic population: findings from the National Health and Nutrition Examination Survey Cardiovasc Diabetol **22**, 296 10.1186/s12933-023-02003-2

1667. Liu, Q. Y., Liao, Y., Wu, Y. X., Diao, H., Du, Y., Chen, Y. W. *et al.* (2023) The Oral Microbiome as Mediator between Oral Hygiene and Its Impact on Nasopharyngeal Carcinoma Microorganisms **11**, 10.3390/microorganisms11030719

1668. Liu, X., Matsunami, M., Horikoshi, M., Ito, S., Ishikawa, Y., Suzuki, K. *et al.* (2023) Natural Selection Signatures in the Hondo and Ryukyu Japanese Subpopulations Mol Biol Evol **40**, 10.1093/molbev/msad231

1669. Mahmud AZ, Nazni p, Darki HS, Lami Idris B, Aliyu HI, Umar AN *et al.* (2023) Basic Food Groups' Consumption Pattern Among Pregnant Women Journal of Food and Dietetics Research **3**, 24-28,

1670. Markman, K. M., Weicker, N. P., Klein, A. K., andSege, R. (2023) Community-engaged training in informed consent J Clin Transl Sci **7**, e108 10.1017/cts.2023.534

1671. Matsudaira, I., Yamaguchi, R., andTaki, Y. (2023) Transmit Radiant Individuality to Offspring (TRIO) study: investigating intergenerational transmission effects on brain development Front Psychiatry **14**, 1150973 10.3389/fpsyt.2023.1150973

1672. Okuno, R., Inoue, Y., Hasebe, Y., Igarashi, T., Kawagishi-Hotta, M., Yamada, T. *et al.* (2023) Genome-wide association studies in the Japanese population identified genetic loci and target gene associated with epidermal turnover Exp Dermatol **32**, 1856-1863 10.1111/exd.14908

1673. Oladayo, A., Gowans, L. J. J., Awotoye, W., Alade, A., Busch, T., Naicker, T. *et al.* (2023) Clinically actionable secondary findings in 130 triads from sub-Saharan African families with non-syndromic orofacial clefts Mol Genet Genomic Med 10.1002/mgg3.2237

1674. Rakhimova, M., andNobo'toyev, O. (2023) The Role of Cystatin C in the Diagnosis of Renal Dysfunction in Patients with Coronary Heart Disease Genius Repository **24**, 31-36,

1675. Shi, M., Tanikawa, C., Munter, H. M., Akiyama, M., Koyama, S., Tomizuka, K. *et al.* (2023) Genotype imputation accuracy and the quality metrics of the minor ancestry in multi-ancestry reference panels Brief Bioinform **25**, 10.1093/bib/bbad509

1676. Suzuki, S., Uchiyama, K., Motoi, Y., Yoshii, Y., Inoue, Y., Kubota, T. *et al.* (2023) Analysis of the NUDT15 gene and metabolites of azathioprine in Japanese patients with inflammatory bowel disease BMC Gastroenterol **23**, 239 10.1186/s12876-023-02881-6

1677. Suzuki, T., Nishigori, T., Obara, T., Mori, M., Sakurai, K., Ishikuro, M. *et al.* (2023) Factors associated with new onset of father-to-infant bonding failure from 1 to 6 months postpartum: an adjunct study of the Japan environment and children's study Soc Psychiatry Psychiatr Epidemiol 1-22 10.1007/s00127-023-02505-0

1678. Taguchi, R., Shigemi, D., andYasunaga, H. (2023) Re: Use of antihypertensive drugs during pregnancy in Japan Hypertens Res **46**, 550 10.1038/s41440-022-01120-x

1679. Thébault, P., Cailhier, J. F., andLapointe, R. (2023) Blood Sample Processing and Banking for Functional and Molecular Analyses In Methods Mol Biol, 37-46

1680. Tian, Z., Chen, F., Wang, J., Wu, B., Shao, J., Liu, Z. *et al.* (2023) CAS Array: design and assessment of a genotyping array for Chinese biobanking Precis Clin Med **6**, pbad002 10.1093/pcmedi/pbad002

1681. Weil, C. J. (2023) Ethical, Legal, and Policy Issues Surrounding Biospecimen Research Conducted or Supported in the USA Biopreserv Biobank **21**, 14-22 10.1089/bio.2021.0094

1682. Wong, E., Bertin, N., Hebrard, M., Tirado-Magallanes, R., Bellis, C., Lim, W. K. *et al.* (2023) The Singapore National Precision Medicine Strategy Nature genetics **55**, 178-186 10.1038/s41588-022-01274-x

1683. Yamakawa, Y., Maruta, M., Higuchi, Y., Tokunaga, A., Iwanaga, R., Honda, S. *et al.* (2023) Factors influencing postpartum depression among Japanese parents: A prospective longitudinal study Neuropsychopharmacol Rep **43**, 213-221 10.1002/npr2.12326

1684. Yang, G., Hisada, A., Yamamoto, M., Kawanami, A., Mori, C., Sakurai, K. *et al.* (2023) Effect of nausea and vomiting during pregnancy on mother-to-infant bonding and the mediation effect of postpartum depression: the Japan Environment and Children's Study BMC Pregnancy Childbirth **23**, 704 10.1186/s12884-023-06014-5

1685. Yoshihara, K., Lee, S. W., Kim, Y. M., andEnomoto, T. (2023) The 1st annual meeting of the East Asian Gynecologic Oncology Trial Group (EAGOT) Journal of Gynecologic Oncology **34**, ARTN e87 10.3802/jgo.2023.34.e87

1686. Yumiya, Y., Chimed-Ochir, O., Kayano, R., Hitomi, Y., Akahoshi, K., Kondo, H. *et al.* (2023) Emergency Medical Team Response during the Hokkaido Eastern Iburi Earthquake 2018: J-SPEED Data Analysis Prehosp Disaster Med **38**, 332-337 10.1017/S1049023X23000432

1687. Zhang, Y., Zhao, Y., Duan, Y., Liu, C., Yang, Z., Duan, J. *et al.* (2023) Effects of prepregnancy dietary patterns on infant birth weight: a prospective cohort study J Matern Fetal Neonatal Med **36**, 2273216 10.1080/14767058.2023.2273216

1688. Zheng, H., Wang, Y., Yang, B., Wu, J., Qian, Y., Wang, W. *et al.* (2023) Interaction of Dietary Sodium-to-potassium Ratio and Dinner Energy Ratio on Prevalence of Hypertension in Inner Mongolia, China J Epidemiol **33**, 547-555 10.2188/jea.JE20220045

1689. 小林澄貴, 宮下ちひろ, 伊藤佐智子, and 岸玲子 (2023) DOHaD, ライフコースアプローチによる 20 年に及ぶ 前向き出生コホート研究の成果：環境と子どもの健康に関する北海道スタディ DOHaD 研究 **11**, 63-95 https://doi.org/10.51067/dohad.11.2_63

1690. Belay, J. K., Abebe, S. M., Baffa, L. D., andMengistu, B. (2024) Food consumption score and predictors among pregnant women attending antenatal care services in health centers of Addis Ababa, Ethiopia: Using ordinal logistic regression model PLoS One **19**, e0306169 10.1371/journal.pone.0306169

1691. Brown, A., Griffin, S., Weaver, G., andShenker, N. (2024) Receiving screened donor human milk as part of a community-based lactation support programme reduces parental symptoms of anxiety and depression Matern Child Nutr **20**, e13686 10.1111/mcn.13686

1692. Brunette, C. A., Yi, T., Danowski, M. E., Cardellino, M., Harrison, A., Assimes, T. L. *et al.* (2024) Development and utility of a clinical research informatics application for participant recruitment and workflow management for a return of results pilot trial in familial hypercholesterolemia in the Million Veteran Program JAMIA Open **7**, ooae020 10.1093/jamiaopen/ooae020

1693. Chen, C., Mochizuki, Y., Okawa, S., Okubo, R., Nakagawa, S., andTabuchi, T. (2024) Postpartum loneliness predicts future depressive symptoms: a nationwide Japanese longitudinal study Arch Womens Ment Health **27**, 447-457 10.1007/s00737-024-01424-6

1694. Dinh, B. L., Tang, E., Taparra, K., Nakatsuka, N., Chen, F., andChiang, C. W. K. (2024) Recombination map tailored to Native Hawaiians may improve robustness of genomic scans for positive selection Hum Genet **143**, 85-99 10.1007/s00439-023-02625-2

1695. Egawa, M., Ikeda, M., Miyasaka, N., Harada-Shiba, M., andYoshida, M. (2024) Physicians' Perspective Toward Hypertensive Disorders of Pregnancy and Future Cardiovascular Diseases in Japan J Atheroscler Thromb **31**, 953-963 10.5551/jat.64453

1696. Geng, M., Ding, P., Wang, S., Wang, B., Tong, J., Gao, H. *et al.* (2024) Prenatal antibiotics exposure and preschoolers' internalizing and externalizing problems: A biomonitoring-based prospective birth cohort study Sci Total Environ **919**, 170891 10.1016/j.scitotenv.2024.170891

1697. Gondo, T., Hiraishi, N., Takeuchi, A., Moyes, D., andShimada, Y. (2024) Comparative analysis of microbiome in coronal and root caries BMC Oral Health **24**, 869 10.1186/s12903-024-04670-3

1698. Hanai, A., Ishikawa, T., Sugao, S., Fujii, M., Hirai, K., Watanabe, H. *et al.* (2024) Explainable Machine Learning Classification to Identify Vulnerable Groups Among Parenting Mothers: Web-Based Cross-Sectional Questionnaire Study JMIR Form Res **8**, e47372 10.2196/47372

1699. Hartmann, A., Grabitz, S. D., Wagner, F. M., Wild, P. S., Muller-Nurasyid, M., Lackner, K. J. *et al.* (2024) Bi-Gaussian analysis reveals distinct education-related alterations in spherical equivalent and axial length-results from the Gutenberg Health Study Graefes Arch Clin Exp Ophthalmol **262**, 1819-1828 10.1007/s00417-024-06395-z

1700. Hashimoto, K., andShimizu, K. (2024) Enhancing Privacy Protection for Human Genome Synthesis Using Gradient Clipping In Proceedings of the 15th ACM International Conference on Bioinformatics, Computational Biology and Health Informatics, Shenzhen China 1-6

1701. Hashimoto, T., Nakamura, Y., Oki, E., Kobayashi, S., Yuda, J., Shibuki, T. *et al.* (2024) Bridging horizons beyond CIRCULATE-Japan: a new paradigm in molecular residual disease detection via whole genome sequencing-based circulating tumor DNA assay Int J Clin Oncol **29**, 495-511 10.1007/s10147-024-02493-4

1702. Hattori, K., Kobayashi, K., Azuma-Suzuki, R., Iwasa, K., Higashi, S., Hamaguchi, T. *et al.* (2024) Nicotinamide phosphoribosyl transferase in mammary gland epithelial cells is required for nicotinamide mononucleotide production in mouse milk Biochem Biophys Res Commun **728**, 150346 10.1016/j.bbrc.2024.150346

1703. Herzig, A. F., Velo-Suarez, L., FrEx, C., FranceGenRef, C., Dina, C., Redon, R. *et al.* (2024) How local reference panels improve imputation in French populations Sci Rep **14**, 370 10.1038/s41598-023-49931-3

1704. Hiremath, G. B., Omkarbabu, K., Kokate, M. H., Venkidasamy, B., Krishnan, M., andMurugaiyan, A. (2024) Development of Oral Bio-banks Past, Present and Future; Challenges and Opportunities Curr Gene Ther **24**, 2-3 10.2174/1566523223666230801090355

1705. Hisamatsu, T., Kogure, M., Tabara, Y., Hozawa, A., Sakima, A., Tsuchihashi, T. *et al.* (2024) Practical use and target value of urine sodium-to-potassium ratio in assessment of hypertension risk for Japanese: Consensus Statement by the Japanese Society of Hypertension Working Group on Urine Sodium-to-Potassium Ratio Hypertens Res **47**, 3288-3302 10.1038/s41440-024-01861-x

1706. Hsu, F. M., Chen, H. C., Wang, K. C., Ling, W. L., andChen, N. C. (2024) Application of time series analysis in predicting postpartum depression: integrating data from the hospitalization period and early postpartum weeks Arch Womens Ment Health 10.1007/s00737-024-01521-6

1707. Imai, Y., Kusano, K., Aiba, T., Ako, J., Asano, Y., Harada-Shiba, M. *et al.* (2024) JCS/JCC/JSPCCS 2024 Guideline on Genetic Testing and Counseling in Cardiovascular Disease Circ J **88**, 2022-2099 10.1253/circj.CJ-23-0926

1708. Ingeborgrud, C. B., Oerbeck, B., Friis, S., Pripp, A. H., Aase, H., Biele, G. *et al.* (2024) Low perceived social support in mothers during pregnancy and early childhood; associations with anxiety and ADHD symptoms in children at 3 and 8 years Soc Psychiatry Psychiatr Epidemiol 10.1007/s00127-024-02792-1

1709. Kamitaki, N., Hujoel, M. L. A., Mukamel, R. E., Gebara, E., McCarroll, S. A., andLoh, P. R. (2024) A sequence of SVA retrotransposon insertions in ASIP shaped human pigmentation Nature genetics **56**, 1583-1591 10.1038/s41588-024-01841-4

1710. Kasai, F., Fukushima, M., Miyagi, Y., andNakamura, Y. (2024) Genetic diversity among the present Japanese population: evidence from genotyping of human cell lines established in Japan Hum Cell **37**, 944-950 10.1007/s13577-024-01055-0

1711. Kim, B., Kim, D. S., Shin, J. G., Leem, S., Cho, M., Kim, H. *et al.* (2024) Mapping and annotating genomic loci to prioritize genes and implicate distinct polygenic adaptations for skin color Nat Commun **15**, 4874 10.1038/s41467-024-49031-4

1712. Kim, S. (2024) The Relationship Between MOTS-c K14Q Polymorphism and Sarcopenia, Blood Lipids, and Mental Health in Older Korean Adults Biomedicines **12**, 10.3390/biomedicines12102384

1713. Kober, K. M., Berger, L., Roy, R., andOlshen, A. (2024) Torch-eCpG: a fast and scalable eQTM mapper for thousands of molecular phenotypes with graphical processing units BMC Bioinformatics **25**, 71 10.1186/s12859-024-05670-4

1714. Koiwa, C., Negishi, T., Sakemi, F., Takesue, A., andYokoyama, T. (2024) A Case of Strabismus Fixus With Extremely Long Axial Length Results in Improvement of Limitation of Abduction by the Yokoyama Procedure Cureus **16**, e58194 10.7759/cureus.58194

1715. Kok, E. Y., Kaur, S., Mohd Shukri, N. H., Abdul Razak, N., andTakahashi, M. (2024) Development, validation, and reliability of the Chrononutrition Profile Questionnaire-Pregnancy (CPQ-P) BMC Pregnancy Childbirth **24**, 217 10.1186/s12884-024-06403-4

1716. Lam, P. H., Hon, K. L., Loo, S., Li, C. K., Ip, P., Koh, M. J. *et al.* (2024) Mental health among parents and their children with eczema in Hong Kong Hong Kong Med J **30**, 362-370 10.12809/hkmj219738

1717. Lukasiewicz-Smietanska, D., Godlewski, D., Nowakowska, E., Szpak, A., Chabros, E., Juszczyk, G. *et al.* (2024) Association of the bitter taste genes TAS2R38 and CA6 and breast cancer risk; a case-control study of Polish women in Poland and Polish immigrants in USA PLoS One **19**, e0300061 10.1371/journal.pone.0300061

1718. Matsunaga, M., Okajima, J., Furutani, K., Kusakabe, N., andNakamura-Taira, N. (2024) Associations of rumination, behavioral activation, and perceived reward with mothers' postpartum depression during the COVID-19 pandemic: a cross-sectional study Front Psychiatry **15**, 1295988 10.3389/fpsyt.2024.1295988

1719. Matsuo, T., Hamasaki, I., Kamatani, Y., Kawaguchi, T., Yamaguchi, I., Matsuda, F. *et al.* (2024) Genome-Wide Association Study with Three Control Cohorts of Japanese Patients with Esotropia and Exotropia of Comitant Strabismus and Idiopathic Superior Oblique Muscle Palsy Int J Mol Sci **25**, 10.3390/ijms25136986

1720. Min, Q., Hu, W., Lei, D., He, X., Liu, C., Li, Q. *et al.* (2024) The correlation of shift work and CLOCK, BMAL1, and PER1 gene polymorphisms with hypertension in healthcare workers: A cross-sectional study Medicine (Baltimore) **103**, e40148 10.1097/MD.0000000000040148

1721. Mizuta, M., andSuzuki, I. (2024) Streptococcus mutans Membrane Vesicles, Containing Insoluble Glucan Synthase and Extracellular DNA, Contribute to the Promotion of Initial Attachment and Colonization of Actinomyces oris International Journal of Oral-Medical Sciences **22**, 57-68,

1722. Neda-Stepan, O., Giurgi-Oncu, C., Salcudean, A., Bernad, E., Bernad, B. C., Boeriu, E. *et al.* (2024) Evaluating the Impact of Obsessive-Compulsive Symptoms and Personality Types on Perinatal Depressive Symptoms Behav Sci (Basel) **14**, 10.3390/bs14070589

1723. Ng, J. Y., Zhou, H., Li, T., andChew, F. T. (2024) Comparisons between wrinkles and photo-ageing detected and

self-reported by the participant or identified by trained assessors reveal insights from Chinese individuals in the Singapore/Malaysia Cross-sectional Genetics Epidemiology Study (SMCGES) cohort J Physiol Anthropol **43**, 14 10.1186/s40101-024-00361-8

1724. Oshikata, C., Watanabe, M., Hashimoto, K., Yamazaki, A., Kobayashi, N., Konuma, R. *et al.* (2024) Mite allergen levels and fungal counts in children's bedding in four widely separated towns in Japan Revue Francaise D Allergologie **64**, 104084 10.1016/j.reval.2024.104084
1725. Ozsahin, Z. (2024) Determinants of the desire to avoid pregnancy after the disaster of the century in Turkiye BMC Womens Health **24**, 496 10.1186/s12905-024-03330-6
1726. Peng, F., Shen, Q., Zou, L. P., Cheng, F., Xue, Y. P., andZheng, Y. G. (2024) Design of NAMPTs with Superior Activity by Dual-Channel Protein Engineering Strategy J Agric Food Chem 10.1021/acs.jafc.4c02698
1727. Qu, Q., Shi, Y., Guo, Q., Yue, X., Chen, L., Sun, J. *et al.* (2024) Association of high-sensitivity cardiac troponin T with all-cause and cardiovascular mortality in older adults with low lean mass: A 14.6-year longitudinal study Arch Gerontol Geriatr **116**, 105140 10.1016/j.archger.2023.105140
1728. Rodriguez-Fuentes, M. E., Perez-Sayans, M., Barbeito-Castineiras, G., Molares-Vila, A., Prado-Pena, I. B., Camolesi, G. C. V. *et al.* (2024) Oral specimens as a tool for accurate metagenomic analysis: A pilot study J Stomatol Oral Maxillofac Surg **125**, 101991 10.1016/j.jormas.2024.101991
1729. Ronald, A., andGui, A. (2024) The potential and translational application of infant genetic research Nature genetics **56**, 1346-1354 10.1038/s41588-024-01822-7
1730. Sotodate, G., Onodera, S., Okuyama, I., Akasaka, M., Matsumoto, A., Toya, Y. *et al.* (2024) Relationship Between Frequency of Remote Visitation and Postpartum Depression in Mothers of Hospitalized Neonates During the COVID-19 Pandemic J Obstet Gynecol Neonatal Nurs **53**, 554-561 10.1016/j.jogn.2024.05.136
1731. Taji, A., Yumiya, Y., Chimed-Ochir, O., Fukunaga, A., Tsurugi, Y., Kiwaki, K. *et al.* (2024) Medical needs during the Kumamoto heavy rain 2020: analysis from emergency medical teams' responses BMC Emerg Med **24**, 94 10.1186/s12873-024-01009-7
1732. Tomita, H., Iwama, N., Hamada, H., Kudo, R., Tagami, K., Kumagai, N. *et al.* (2024) The impact of maternal and paternal birth weights on infant birth weights: the Japan environment and children's study J Dev Orig Health Dis 1-12 10.1017/S2040174423000387
1733. Vadsaria, K., Mohammed, N., Aamir, S., andNuruddin, R. (2024) Are first-trimester pregnant women consuming adequate and diverse diet? A hospital-based cross-sectional study in Karachi, Pakistan BMC Nutr **10**, 104 10.1186/s40795-024-00912-3
1734. Wang, X., Gao, X., Chen, D., Chen, X., Li, Q., Ding, J. *et al.* (2024) The effect of active and passive smoking during pregnancy on birth outcomes: A cohort study in Shanghai Tob Induc Dis **22**, 10.18332/tid/188866
1735. Wei, M., Meng, D., He, S., Guo, H., Yang, G., andWang, Z. (2024) Causal effect of physical activity and sedentary behavior on the risk of alcohol dependence: A bidirectional two-sample Mendelian randomization study Alcohol **120**, 15-24 10.1016/j.alcohol.2024.05.002
1736. Yamamoto, Y., Yamamoto, T., Miyamoto, N., Kinoshita, K., Nishikawa, S., Adachi, T. *et al.* (2024) Oral Function and the Oral Microbiome in the Elderly in the Kyotango Area Dent J (Basel) **12**, 10.3390/dj12010016
1737. Yoshida, A., Asakura, K., Imamura, H., Mori, S., Sugimoto, M., Michikawa, T. *et al.* (2024) Relationship between working hours and sleep quality with consideration to effect modification by work style: a community-based cross-sectional study Environ Health Prev Med **29**, 19 10.1265/ehpm.23-00252
1738. Zaabaar, E., Shing, E., Zhang, X. J., Wang, Y., Kam, K. W., Zhang, Y. *et al.* (2024) Associations of genetic variants

for refractive error and axial length in adults with ocular endophenotypes in children: a cross-sectional and longitudinal study Br J Ophthalmol 10.1136/bjo-2024-325606

1739. Zheng, W., Chu, J., Bambrick, H., Wang, N., Mengersen, K., Guo, X. *et al.* (2024) Impacts of heatwaves on type 2 diabetes mortality in China: a comparative analysis between coastal and inland cities Int J Biometeorol **68**, 939-948 10.1007/s00484-024-02638-0
1740. Zhou, S., Zhou, N., Zhang, H., Yang, W., Liu, Q., Zheng, L. *et al.* (2024) A prospective multicenter birth cohort in China: pregnancy health atlas Eur J Epidemiol **39**, 1297-1310 10.1007/s10654-024-01157-x
1741. 安形清彦 (2024) 糖鎖関連遺伝子の発現と疾患との関連解析 創価大学糖鎖生命システム融合研究所 (GaLSIC) 所報 3-7,
1742. 吉田輝彦, and 飛松由紀子 (2024) 遺伝性腫瘍セミナー: 25 年の歩みと展望 遺伝性腫瘍 **24**, 38-44,
1743. 篠澤圭子 (2024) 血液凝固異常症と血友病の分子遺伝学 日本血栓止血学会誌 **35**, 497-511,
1744. 小椋たみ子, 増田珠巳, and 平井純子 (2024) 家庭で養育されている乳幼児の遊び環境と言語発達, 精神発達 大阪総合保育大学児童保育論集 25-38,
1745. 等々力花歩 (2024) 日本の育児をめぐる現状と課題解決に向けた展望--発達科学の観点から 京都大学大学院教育学研究科紀要 **70**, 219-232,
1746. Brill, J., Clarke, B., Hong, I., andHuganir, R. L. (2025) Dissociation of SYNGAP1 enzymatic and structural roles: Intrinsic excitability and seizure susceptibility Proc Natl Acad Sci U S A **122**, e2427288122 10.1073/pnas.2427288122
1747. Dung, L. T., Lam, L. T., Trang, N. H., Anh, N. V. H., Nam, N. N., Nhung, D. T. *et al.* (2025) Toward a Kinh Vietnamese Reference Genome: Constructing a De Novo Genome Assembly Using Long-Read Sequencing and Optical Mapping Genes (Basel) **16**, 10.3390/genes16050536
1748. EMES, M., AM, D., AH. Atta, andFN, M. (2025) Emotional and Behavioural Problems among Children with Allergic Diseases in Sharqia Governorate Zagazig Nursing Journal,
1749. Grigio, T. R., Furuya, T. K., Slullitel, A., Murillo Carrasco, A. G., Uno, M., Alves, M. J. F. *et al.* (2025) Clinical, ethnic and genetic risk factors associated with postoperative nausea and vomiting in patients undergoing cancer surgery: a case-control study Am J Transl Res **17**, 3235-3246 10.62347/DGRM3907
1750. Matsushita, K., Ishige, T., Watanabe, K., Akahane, T., Tanimoto, A., Yoshimoto, M. *et al.* (2025) Importance of EQA/PT for the detection of genetic variants in comprehensive cancer genome testing Sci Rep **15**, 1036 10.1038/s41598-024-84714-4
1751. Mukuda, K., Inoue, R., Takata, M., Takazawa, K., Noma, H., Morishima, S. *et al.* (2025) Temporal effects of lascufloxacin on human gut and salivary microbiota: Analysis using next-generation sequencing method J Infect Chemother **31**, 102483 10.1016/j.jiac.2024.07.023
1752. Seidizadeh, O., Cairo, A., Mancini, I., George, J. N., andPeyvandi, F. (2025) Clarifying the role of p.Gln723Lys in hTTP prevalence Blood Adv **9**, 3024-3025 10.1182/bloodadvances.2025016030

レビュー（1753-1919）

1753. Mitra, S., Pandey, S., Gill, A., Roy, A., Dutta, S., Kumar, A. *et al.* (2025) Molecular pathways and biomarkers associated with age-related cognitive decline Academia Biology **3**, 10.20935/AcadBiol7909
1754. Rizner, T. L., andRomano, A. (2025) The discovery of biomarkers for endometrial cancer: update over the last years Expert Rev Mol Diagn **25**, 425-452 10.1080/14737159.2025.2505546
1755. Shareef, A., Mulaw, G. F., Rathore, K., Irwin, C., Vincze, L., Rigby, R. *et al.* (2025) Association between Bitter Taste Sensitivity and Weight Status in Adults: A Systematic Review and Meta-Analysis Nutr Rev

10.1093/nutrit/nuaf160

1756. Precone, V., Del Monaco, V., Esposito, M. V., De Palma, F. D., Ruocco, A., Salvatore, F. *et al.* (2015) Cracking the Code of Human Diseases Using Next-Generation Sequencing: Applications, Challenges, and Perspectives Biomed Res Int **2015**, 161648 10.1155/2015/161648
1757. Chalmers, D., Nicol, D., Kaye, J., Bell, J., Campbell, A. V., Ho, C. W. *et al.* (2016) Has the biobank bubble burst? Withstanding the challenges for sustainable biobanking in the digital era BMC Med Ethics **17**, 39 10.1186/s12910-016-0124-2
1758. Fukunaga, H., Yokoya, A., andTaki, Y. (2016) Now Is the Time to Consider Personalized Effective Dose Int J Radiat Oncol Biol Phys **96**, 479-480 10.1016/j.ijrobp.2016.06.012
1759. Lee, S., Jung, P. E., andLee, Y. (2016) Publicly-funded biobanks and networks in East Asia Springerplus **5**, 1080 10.1186/s40064-016-2723-2
1760. Zappala, Z., andMontgomery, S. B. (2016) Non-Coding Loss-of-Function Variation in Human Genomes Hum Hered **81**, 78-87 10.1159/000447453
1761. An, J. Y. (2017) National human genome projects: an update and an agenda Epidemiol Health **39**, e2017045 10.4178/epih.e2017045
1762. Baux, D., Sasorith, S., Bergougnoux, A., andClaustres, M. (2017) Mutation Databases In eLS, 1-9
1763. Chiara, M., andPavesi, G. (2017) Evaluation of Quality Assessment Protocols for High Throughput Genome Resequencing Data Front Genet **8**, 94 10.3389/fgene.2017.00094
1764. Elger, B. S., andDe Clercq, E. (2017) Returning Results: Let's Be Honest! Genet Test Mol Biomarkers **21**, 134-139 10.1089/gtmb.2016.0395
1765. Friedman, C. P., Allee, N. J., Delaney, B. C., Flynn, A. J., Silverstein, J. C., Sullivan, K. *et al.* (2017) The science of Learning Health Systems: Foundations for a new journal Learn Health Syst **1**, e10020 10.1002/lrh2.10020
1766. Fukunaga, H., Yokoya, A., Taki, Y., andPrise, K. M. (2017) Radiobiological Implications of Fukushima Nuclear Accident for Personalized Medical Approach Tohoku J Exp Med **242**, 77-81 10.1620/tjem.242.77
1767. Hellwege, J. N., Keaton, J. M., Giri, A., Gao, X., Velez Edwards, D. R., andEdwards, T. L. (2017) Population Stratification in Genetic Association Studies Curr Protoc Hum Genet **95**, 1 22 21-21 22 23 10.1002/cphg.48
1768. Hitomi, Y., andTokunaga, K. (2017) Significance of functional disease-causal/susceptible variants identified by whole-genome analyses for the understanding of human diseases Proc Jpn Acad Ser B Phys Biol Sci **93**, 657-676 10.2183/pjab.93.042
1769. Matzaraki, V., Kumar, V., Wijmenga, C., andZhernakova, A. (2017) The MHC locus and genetic susceptibility to autoimmune and infectious diseases Genome Biol **18**, 76 10.1186/s13059-017-1207-1
1770. Rigden, D. J., andFernández, X. M. (2017) The 2018 Nucleic Acids Research database issue and the online molecular biology database collection Nucleic acids research **46**, D1-D7,
1771. Lerch, M., Mainetti, C., Terziroli Beretta-Piccoli, B., andHarr, T. (2018) Current Perspectives on Stevens-Johnson Syndrome and Toxic Epidermal Necrolysis Clin Rev Allergy Immunol **54**, 147-176 10.1007/s12016-017-8654-z
1772. Rajagopalan, R. M., andFujimura, J. H. (2018) Variations on a Chip: Technologies of Difference in Human Genetics Research J Hist Biol **51**, 841-873 10.1007/s10739-018-9543-x
1773. Satterfield, K., Rubin, J. C., andFriedman, C. P. (2018) Toward a Learning Ecosystem for Diagnostic Excellence
1774. Suneja, A., Gakh, M., andRutkow, L. (2018) Burden and Management of Noncommunicable Diseases After Earthquakes and Tsunamis Health Secur **16**, 30-47 10.1089/hs.2017.0059
1775. Suzuki, K. (2018) The developing world of DOHaD J Dev Orig Health Dis **9**, 266-269

10.1017/S2040174417000691

1776. 岩上将夫, 青木事成, 赤沢学, 石黒智恵子, 今井志乃ぶ, 大場延浩 *et al.* (2018) 「日本における傷病名を中心とするレセプト情報から得られる指標のバリデーションに関するタスクフォース」 報告書 薬剤疫学 **23**, 95-123,
1777. 秋葉澄伯 (2018) 環境政策における意思決定とレギュラトリーサイエンス 保健医療科学 **67**, 255-260,
1778. 神沼英里, 藤澤貴智, and 中村保一 (2018) ライフサイエンス研究におけるクラウドソーシングの利用と実践 デジタルプラクティス **9**, 886-899,
1779. 野口佳裕, 西尾信哉, and 宇佐美真一 (2018) 次世代シークエンサーを用いた遺伝子診断のピットフォール Audiology Japan **61**, 129-135,
1780. Bregenzer, M. E., Horst, E. N., Mehta, P., Novak, C. M., Raghavan, S., Snyder, C. S. *et al.* (2019) Integrated cancer tissue engineering models for precision medicine PLoS One **14**, e0216564 10.1371/journal.pone.0216564
1781. Coppola, L., Cianflone, A., Grimaldi, A. M., Incoronato, M., Bevilacqua, P., Messina, F. *et al.* (2019) Biobanking in health care: evolution and future directions J Transl Med **17**, 172 10.1186/s12967-019-1922-3
1782. Ding, C., Qin, Z., Li, Y., Shi, W., Li, L., Zhan, D. *et al.* (2019) Proteomics and Precision Medicine Small Methods **3**, 1900075 https://doi.org/10.1002/smtd.201900075
1783. Donovan, B. M., Bastarache, L., Turi, K. N., Zutter, M. M., andHartert, T. V. (2019) The current state of omics technologies in the clinical management of asthma and allergic diseases Ann Allergy Asthma Immunol **123**, 550-557 10.1016/j.anai.2019.08.460
1784. Ebi, H., andBando, H. (2019) Precision Oncology and the Universal Health Coverage System in Japan JCO Precis Oncol **3**, 1-12 10.1200/PO.19.00291
1785. Fukunaga, H., Yokoya, A., Taki, Y., Butterworth, K. T., andPrise, K. M. (2019) Precision Radiotherapy and Radiation Risk Assessment: How Do We Overcome Radiogenomic Diversity? Tohoku J Exp Med **247**, 223-235 10.1620/tjem.247.223
1786. Guo, L., andYe, K. (2019) Mapping Genome Variants Sheds Light on Genetic and Phenotypic Differentiation in Chinese Genomics Proteomics Bioinformatics **17**, 226-228 10.1016/j.gpb.2019.09.001
1787. Klasberg, S., Surendranath, V., Lange, V., andSchofl, G. (2019) Bioinformatics Strategies, Challenges, and Opportunities for Next Generation Sequencing-Based HLA Genotyping Transfus Med Hemother **46**, 312-325 10.1159/000502487
1788. Misra, B. B., andMohapatra, S. (2019) Tools and resources for metabolomics research community: A 2017-2018 update Electrophoresis **40**, 227-246 10.1002/elps.201800428
1789. Ohkubo, K. (2019) Cybersecurity Technologies Essential in the Digital Transformation Era International Journal of Informatics Society (IJIS) **11**, 13-21,
1790. Park, S. C., andJeen, Y. T. (2019) Genetic Studies of Inflammatory Bowel Disease-Focusing on Asian Patients Cells **8**, 404-425 10.3390/cells8050404
1791. Sivadas, A., andScaria, V. (2019) Population-scale genomics-Enabling precision public health Adv Genet **103**, 119-161 10.1016/bs.adgen.2018.09.001
1792. Stark, Z., Dolman, L., Manolio, T. A., Ozenberger, B., Hill, S. L., Caulfied, M. J. *et al.* (2019) Integrating Genomics into Healthcare: A Global Responsibility Am J Hum Genet **104**, 13-20 10.1016/j.ajhg.2018.11.014
1793. Stylianou, E. (2019) Epigenetics of chronic inflammatory diseases J Inflamm Res **12**, 1-14 10.2147/JIR.S129027
1794. Suzuki, H., Watanabe, T., Uehara, T., andKosaki, K. (2019) Medical genetics and genomic medicine in Japan Am J Med Genet C Semin Med Genet **181**, 166-169 10.1002/ajmg.c.31702

1795. Takahashi, K., andShibahara, S. (2019) 100 Years of the Tohoku Journal of Experimental Medicine, and the Tohoku Medical Megabank Project in the Japanese New Era, Reiwa Tohoku J Exp Med **248**, 1-2 10.1620/tjem.248.1

1796. Takeoka, M., Fujiwara, M., andSasaki, M. (2019) R&D Trends and Future Prospects of Quantum Cryptography New Breeze 4-8,

1797. Wong, C. S., Kogon, A. J., Warady, B. A., Furth, S. L., Lantos, J. D., andWilfond, B. S. (2019) Ethical and Policy Considerations for Genomic Testing in Pediatric Research: The Path Toward Disclosing Individual Research Results Am J Kidney Dis **73**, 837-845 10.1053/j.ajkd.2019.01.020

1798. 鳥羽研二 (2019) オレンジレジストリと認知症研究 日本老年医学会雑誌 **56**, 97-106,

1799. 箕輪真理 (2019) NBDC ヒトデータベースと日本人ゲノム多様性統合データベース “TogoVar” 家族性腫瘍 **19**, 45-49,

1800. Afshinnekoo, E., Scott, R. T., MacKay, M. J., Pariset, E., Cekanaviciute, E., Barker, R. *et al.* (2020) Fundamental Biological Features of Spaceflight: Advancing the Field to Enable Deep-Space Exploration Cell **183**, 1162-1184 10.1016/j.cell.2020.10.050

1801. Angelopoulou, E., Paudel, Y. N., Villa, C., andPiperi, C. (2020) Arylsulfatase A (ASA) in Parkinson's Disease: From Pathogenesis to Biomarker Potential Brain Sci **10**, 713 10.3390/brainsci10100713

1802. Berezin, A. E., andBerezin, A. A. (2020) Circulating Cardiac Biomarkers in Diabetes Mellitus: A New Dawn for Risk Stratification-A Narrative Review Diabetes Ther **11**, 1271-1291 10.1007/s13300-020-00835-9

1803. Cechova, M. (2020) Probably Correct: Rescuing Repeats with Short and Long Reads Genes (Basel) **12**, 48 10.3390/genes12010048

1804. Farmakis, D., Mueller, C., andApple, F. S. (2020) High-sensitivity cardiac troponin assays for cardiovascular risk stratification in the general population Eur Heart J **41**, 4050-4056 10.1093/eurheartj/ehaa083

1805. Ho, S. S., Urban, A. E., andMills, R. E. (2020) Structural variation in the sequencing era Nat Rev Genet **21**, 171-189 10.1038/s41576-019-0180-9

1806. Huang, Y., Guo, Q., Ding, X. P., andWang, X. (2020) Mechanism of long noncoding RNAs as transcriptional regulators in cancer RNA Biol **17**, 1680-1692 10.1080/15476286.2019.1710405

1807. Kim, K. D., Kang, Y., andKim, C. (2020) Application of Genomic Big Data in Plant Breeding:Past, Present, and Future Plants (Basel) **9**, 10.3390/plants9111454

1808. Malsagova, K., Kopylov, A., Stepanov, A., Butkova, T., Sinitsyna, A., Izotov, A. *et al.* (2020) Biobanks-A Platform for Scientific and Biomedical Research Diagnostics (Basel) **10**, 10.3390/diagnostics10070485

1809. Mochida, K., Nishii, R., andHirayama, T. (2020) Decoding Plant-Environment Interactions That Influence Crop Agronomic Traits Plant Cell Physiol **61**, 1408-1418 10.1093/pcp/pcaa064

1810. Pan, Z., andXu, S. (2020) Population genomics of East Asian ethnic groups Hereditas **157**, 49 10.1186/s41065-020-00162-w

1811. Pereira, J. A. M., Porto-Figueira, P., Taware, R., Sukul, P., Rapole, S., andCamara, J. S. (2020) Unravelling the Potential of Salivary Volatile Metabolites in Oral Diseases. A Review Molecules **25**, 10.3390/molecules25133098

1812. Poot, M. (2020) Fanconi Anemia: A Syndrome of Anemia and Skeletal Malformations Progressing to a Gene Network Involved in Genomic Stability and Malignant Disease Mol Syndromol **11**, 178-182 10.1159/000510878

1813. Shen, L., McCormick, E. M., Muraresku, C. C., Falk, M. J., andGai, X. (2020) Clinical Bioinformatics in Precise Diagnosis of Mitochondrial Disease Clin Lab Med **40**, 149-161 10.1016/j.cll.2020.02.002

1814. Sherman, R. M., andSalzberg, S. L. (2020) Pan-genomics in the human genome era Nat Rev Genet **21**, 243-254 10.1038/s41576-020-0210-7

1815. Xiao, T., andZhou, W. (2020) The third generation sequencing: the advanced approach to genetic diseases Transl Pediatr **9**, 163-173 10.21037/tp.2020.03.06
1816. Yurkovich, J. T., Tian, Q., Price, N. D., andHood, L. (2020) A systems approach to clinical oncology uses deep phenotyping to deliver personalized care Nat Rev Clin Oncol **17**, 183-194 10.1038/s41571-019-0273-6
1817. 吉田輝彦 (2020) がんゲノム医療における二次的所見の取り組みと考察 遺伝性腫瘍 **20**, 18-23,
1818. 宮田敏行, and 小亀浩市 (2020) TMA の遺伝子診断: TTP と aHUS 日本血栓止血学会誌 **31**, 17-27,
1819. 小峰啓吾, and 石岡千加史 (2020) 個別化がん医療の臨床開発と診療体制の整備 遺伝性腫瘍 **20**, 13-17,
1820. 西條泰明 (2020) 採血が唯一の侵襲である医学研究における採血合併症への賠償・補償について 保健医療科学 **69**, 290-295,
1821. Dato, S., Crocco, P., Rambaldi Migliore, N., andLescai, F. (2021) Omics in a Digital World: The Role of Bioinformatics in Providing New Insights Into Human Aging Front Genet **12**, 689824 10.3389/fgene.2021.689824
1822. Douillard, V., Castelli, E. C., Mack, S. J., Hollenbach, J. A., Gourraud, P. A., Vince, N. *et al.* (2021) Approaching Genetics Through the MHC Lens: Tools and Methods for HLA Research Front Genet **12**, 774916 10.3389/fgene.2021.774916
1823. Ezekian, J. E., Rehder, C., Kishnani, P. S., andLandstrom, A. P. (2021) Interpretation of Incidental Genetic Findings Localizing to Genes Associated With Cardiac Channelopathies and Cardiomyopathies Circ Genom Precis Med **14**, e003200 10.1161/CIRCGEN.120.003200
1824. Garg, S. (2021) Computational methods for chromosome-scale haplotype reconstruction Genome Biol **22**, 101 10.1186/s13059-021-02328-9
1825. Jackson, C. L., Zordok, M., andKullo, I. J. (2021) Familial hypercholesterolemia in Southeast and East Asia Am J Prev Cardiol **6**, 100157 10.1016/j.ajpc.2021.100157
1826. Katoh, H., andIshikawa, S. (2021) Lifestyles, genetics, and future perspectives on gastric cancer in east Asian populations Journal of human genetics **66**, 887-899 10.1038/s10038-021-00960-8
1827. Kiseleva, O., Kurbatov, I., Ilgisonis, E., andPoverennaya, E. (2021) Defining Blood Plasma and Serum Metabolome by GC-MS Metabolites **12**, 15 10.3390/metabo12010015
1828. Li, Y., Deshpande, P., Hertzman, R. J., Palubinsky, A. M., Gibson, A., andPhillips, E. J. (2021) Genomic Risk Factors Driving Immune-Mediated Delayed Drug Hypersensitivity Reactions Front Genet **12**, 641905 10.3389/fgene.2021.641905
1829. Momozawa, Y., andMizukami, K. (2021) Unique roles of rare variants in the genetics of complex diseases in humans Journal of human genetics **66**, 11-23 10.1038/s10038-020-00845-2
1830. Okazaki, A., Yamazaki, S., Inoue, I., andOtt, J. (2021) Population genetics: past, present, and future Hum Genet **140**, 231-240 10.1007/s00439-020-02208-5
1831. Oyenihi, O. R., Oyenihi, A. B., Erhabor, J. O., Matsabisa, M. G., andOguntibeju, O. O. (2021) Unravelling the Anticancer Mechanisms of Traditional Herbal Medicines with Metabolomics Molecules **26**, 10.3390/molecules26216541
1832. Perrone, F., andZilbauer, M. (2021) Biobanking of human gut organoids for translational research Exp Mol Med **53**, 1451-1458 10.1038/s12276-021-00606-x
1833. Tada, H., Yeo, K. K., Li, J. J., Tan, K., Ako, J., Krittayaphong, R. *et al.* (2021) Polygenic Risk Scores for Atherosclerotic Cardiovascular Disease in the Asia-Pacific Region JACC Asia **1**, 294-302 10.1016/j.jacasi.2021.08.008
1834. Tanjo, T., Kawai, Y., Tokunaga, K., Ogasawara, O., andNagasaki, M. (2021) Practical guide for managing large-

scale human genome data in research Journal of human genetics **66**, 39-52 10.1038/s10038-020-00862-1

1835. Trost, B., Loureiro, L. O., andScherer, S. W. (2021) Discovery of genomic variation across a generation Human molecular genetics **30**, R174-R186 10.1093/hmg/ddab209
1836. Zhu, D. D., Tan, X. M., Lu, L. Q., Yu, S. J., Jian, R. L., Liang, X. F. *et al.* (2021) Interplay between nuclear factor erythroid 2-related factor 2 and inflammatory mediators in COVID-19-related liver injury World J Gastroenterol **27**, 2944-2962 10.3748/wjg.v27.i22.2944
1837. 伊藤稔 (2021) 認知症における疾病との共生 医薬産業政策研究所政策研ニュース= OPIR views and actions 37-47,
1838. 永井礼子 (2021) 肺高血圧症における基礎研究の最近の話題: 分子遺伝学的背景を中心に Pediatric Cardiology and Cardiac Surgery **37**, 2-9,
1839. 谷川洋介 (2021) 複数の表現型を用いた人類遺伝統計学の大規模情報解析 JSBi Bioinformatics Review **1**, 47-59,
1840. 池田匡志 (2021) ゲノム情報を利用した精神疾患の個別化医療 日本生物学的精神医学会誌 **32**, 85-88,
1841. 中杤昌弘 (2021) エピゲノムワイド関連研究 (EWAS) による形質関連 DNA メチル化サイトの探索 JSBi Bioinformatics Review **2**, 58-75,
1842. Abouzid, M., Glowka, F., Kagan, L., andKarazniewicz-Lada, M. (2022) Vitamin D Metabolism Gene Polymorphisms and Their Associated Disorders: A Literature Review Curr Drug Metab **23**, 630-651 10.2174/1389200223666220627104139
1843. Ashok, G., andRamaiah, S. (2022) A critical review of datasets and computational suites for improving cancer theranostics and biomarker discovery Med Oncol **39**, 206 10.1007/s12032-022-01815-8
1844. Auwerx, C., Sadler, M. C., Reymond, A., andKutalik, Z. (2022) From pharmacogenetics to pharmaco-omics: Milestones and future directions HGG Adv **3**, 100100 10.1016/j.xhgg.2022.100100
1845. Chuang, Y. C., Wang, C. Y., Huang, W. L., Wang, L. J., Kuo, H. C., Chen, Y. C. *et al.* (2022) Two meta-analyses of the association between atopic diseases and core symptoms of attention deficit hyperactivity disorder Sci Rep **12**, 3377 10.1038/s41598-022-07232-1
1846. Comber, D. A., Davies, B., Roberts, J. D., Tadros, R., Green, M. S., Healey, J. S. *et al.* (2022) Return of Results Policies for Genomic Research: Current Practices and the Hearts in Rhythm Organization (HiRO) Approach Can J Cardiol **38**, 526-535 10.1016/j.cjca.2021.10.006
1847. Edamitsu, T., Taguchi, K., Okuyama, R., andYamamoto, M. (2022) AHR and NRF2 in Skin Homeostasis and Atopic Dermatitis Antioxidants (Basel) **11**, 227 10.3390/antiox11020227
1848. Goto, A., Suzuki, S., Katagiri, R., Yamaji, T., Sawada, N., Nakatochi, M. *et al.* (2022) Public Access to Summary Statistics for Genome-wide Association Studies of Body Mass Index, Weight, and Height Among Healthy Japanese Individuals: The Japanese Consortium of Genetic Epidemiology Studies J Epidemiol **32**, 115-116 10.2188/jea.JE20210459
1849. Guan, F., Ni, T., Zhu, W., Williams, L. K., Cui, L. B., Li, M. *et al.* (2022) Integrative omics of schizophrenia: from genetic determinants to clinical classification and risk prediction Mol Psychiatry **27**, 113-126 10.1038/s41380-021-01201-2
1850. Guo, H. L., Zhao, Y. T., Wang, W. J., Dong, N., Hu, Y. H., Zhang, Y. Y. *et al.* (2022) Optimizing thiopurine therapy in children with acute lymphoblastic leukemia: A promising "MINT" sequencing strategy and therapeutic "DNA-TG" monitoring Front Pharmacol **13**, 941182 10.3389/fphar.2022.941182
1851. Hannon, S. E., Daly, D., andHiggins, A. (2022) Resilience in the Perinatal Period and Early Motherhood: A Principle-Based Concept Analysis Int J Environ Res Public Health **19**, 4754 10.3390/ijerph19084754
1852. Hon, K. L., Chu, S., Leung, A. K. C., andWong, A. (2022) Atopic Dermatitis: Conventional and Integrative

Medicine Curr Pediatr Rev **18**, 84-96 10.2174/1573396317666210716152325

1853. Jain, N., Nagaich, U., Pandey, M., Chellappan, D. K., andDua, K. (2022) Predictive genomic tools in disease stratification and targeted prevention: a recent update in personalized therapy advancements EPMA J **13**, 561-580 10.1007/s13167-022-00304-2

1854. Jones, L. K., Brownson, R. C., andWilliams, M. S. (2022) Applying implementation science to improve care for familial hypercholesterolemia Curr Opin Endocrinol Diabetes Obes **29**, 141-151 10.1097/MED.0000000000000692

1855. Kanazawa, N., Tani, T., Imai, S., Horiguchi, H., Fushimi, K., andInoue, N. (2022) Existing Data Sources for Clinical Epidemiology: Database of the National Hospital Organization in Japan Clin Epidemiol **14**, 689-698 10.2147/CLEP.S359072

1856. Kobayashi, S., Sata, F., andKishi, R. (2022) Gene-environment interactions related to maternal exposure to environmental and lifestyle-related chemicals during pregnancy and the resulting adverse fetal growth: a review Environ Health Prev Med **27**, 24 10.1265/ehpm.21-00033

1857. Kunii, Y., Usukura, H., Otsuka, K., Maeda, M., Yabe, H., Takahashi, S. *et al.* (2022) Lessons learned from psychosocial support and mental health surveys during the 10 years since the Great East Japan Earthquake: Establishing evidence-based disaster psychiatry Psychiatry and clinical neurosciences **76**, 212-221 10.1111/pcn.13339

1858. Lazareva, T. E., Barbitoff, Y. A., Changalidis, A. I., Tkachenko, A. A., Maksiutenko, E. M., Nasykhova, Y. A. *et al.* (2022) Biobanking as a Tool for Genomic Research: From Allele Frequencies to Cross-Ancestry Association Studies J Pers Med **12**, 10.3390/jpm12122040

1859. Miller, C. L. (2022) The Epigenetics of Psychosis: A Structured Review with Representative Loci Biomedicines **10**, 561 10.3390/biomedicines10030561

1860. Murakami, Y. (2022) Cohorts and Biobanks as Essential Resources for Cancer Research Gan to Kagaku ryoho Cancer & Chemotherapy **49**, 129-132,

1861. Nasution, D. R. S., andYamamoto, Z. (2022) Salivary Microflora and Its Diagnostic Importance Buletin Farmatera **7**, 84-93,

1862. Ng, J. Y., andChew, F. T. (2022) A systematic review of skin ageing genes: gene pleiotropy and genes on the chromosomal band 16q24.3 may drive skin ageing Sci Rep **12**, 13099 10.1038/s41598-022-17443-1

1863. Ogasawara, O. (2022) Building cloud computing environments for genome analysis in Japan Hum Genome Var **9**, 46 10.1038/s41439-022-00223-8

1864. Omae, Y., Goto, Y. I., andTokunaga, K. (2022) National Center Biobank Network Hum Genome Var **9**, 38 10.1038/s41439-022-00217-6

1865. Serelli-Lee, V., Ito, K., Koibuchi, A., Tanigawa, T., Ueno, T., Matsushima, N. *et al.* (2022) A State-of-the-Art Roadmap for Biomarker-Driven Drug Development in the Era of Personalized Therapies J Pers Med **12**, 669 10.3390/jpm12050669

1866. Sivolella, S., Scanu, A., Xie, Z., Vianello, S., andStellini, E. (2022) Biobanking in dentistry: A review Jpn Dent Sci Rev **58**, 31-40 10.1016/j.jdsr.2021.12.002

1867. Swan, M., dos Santos, R. P., andWitte, F. (2022) Quantum Neurobiology Quantum Reports **4**, 107-126,

1868. Tada, H., Fujino, N., Hayashi, K., Kawashiri, M. A., andTakamura, M. (2022) Human genetics and its impact on cardiovascular disease J Cardiol **79**, 233-239 10.1016/j.jjcc.2021.09.005

1869. Tsuchida, S., andNakayama, T. (2022) Metabolomics Research in Periodontal Disease by Mass Spectrometry

Molecules **27**, 2864 10.3390/molecules27092864

1870. Wang, C., Wu, W., Yang, H., Ye, Z., Zhao, Y., Liu, J. *et al.* (2022) Mendelian randomization analyses for PCOS: evidence, opportunities, and challenges Trends Genet **38**, 468-482 10.1016/j.tig.2022.01.005
1871. Aktoz, F., Gunes, A. C., andYakin, K. (2023) The effect of earthquake experience on pregnancy outcomes: A systematic review and meta-analysis Eur J Obstet Gynecol Reprod Biol **291**, 70-75 10.1016/j.ejogrb.2023.10.002
1872. Athieniti, E., andSpyrou, G. M. (2023) A guide to multi-omics data collection and integration for translational medicine Comput Struct Biotechnol J **21**, 134-149 10.1016/j.csbj.2022.11.050
1873. Cui, Y., Liao, M., Xu, A., Chen, G., Liu, J., Yu, X. *et al.* (2023) Association of maternal pre-pregnancy dietary intake with adverse maternal and neonatal outcomes: A systematic review and meta-analysis of prospective studies Crit Rev Food Sci Nutr **63**, 3430-3451 10.1080/10408398.2021.1989658
1874. Davis, K., Yost, E., Brauneis, J., Krumme, A., Geldhof, A., Tuck, A. *et al.* (2023) Landscape review of global real-world data sources for studying medication use in pregnancy and lactation that support regulatory decision making Pharmacoepidemiol Drug Saf 10.1002/pds.5711
1875. De Oliveira, T. C., Secolin, R., andLopes-Cendes, I. (2023) A review of ancestrality and admixture in Latin America and the caribbean focusing on native American and African descendant populations Front Genet **14**, 1091269 10.3389/fgene.2023.1091269
1876. Dumancas, G. G., Smith, K. F. K., Bugayong-Janagap, A. M., Zamora, P. R. F. C., Ferriols, V. M. E. N., Liwag, A. G. *et al.* (2023) Challenges and opportunities in establishing a regional biobank center in a developing country: A case from the Philippines Health Policy and Technology 100822 10.1016/j.hlpt.2023.100822
1877. Ferreira, C. R., Carpenter, T. O., andBraddock, D. T. (2023) ENPP1 in Blood and Bone: Skeletal and Soft Tissue Diseases Induced by ENPP1 Deficiency Annu Rev Pathol 10.1146/annurev-pathmechdis-051222-121126
1878. Kakuta, Y., Kinouchi, Y., andMasamune, A. (2023) Genetics of inflammatory bowel disease in East Asia: From population to individual J Gastroenterol Hepatol **38**, 1116-1122 10.1111/jgh.16244
1879. Krol, Z. J., Dobosz, P., Slubowska, A., andMroczek, M. (2023) WGS Data Collections: How Do Genomic Databases Transform Medicine? Int J Mol Sci **24**, 10.3390/ijms24033031
1880. Matsuda, S., Yoshimura, H., andKawachi, I. (2023) Impact of natural disaster on oral health: A scoping review Medicine (Baltimore) **102**, e33076 10.1097/MD.0000000000033076
1881. Miyashita, A., Kikuchi, M., Hara, N., andIkeuchi, T. (2023) Genetics of Alzheimer's disease: an East Asian perspective Journal of human genetics **68**, 115-124 10.1038/s10038-022-01050-z
1882. Ni, S. L., He, W., Zhang, J. K., Yang, F., Bai, G. N., Li, D. *et al.* (2023) Multigenerational birth cohort study in China: importance, necessity and beyond World J Pediatr **19**, 411-416 10.1007/s12519-023-00707-9
1883. Patel, K. K., Venkatesan, C., Abdelhalim, H., Zeeshan, S., Arima, Y., Linna-Kuosmanen, S. *et al.* (2023) Genomic approaches to identify and investigate genes associated with atrial fibrillation and heart failure susceptibility Hum Genomics **17**, 47 10.1186/s40246-023-00498-0
1884. Raz, A., Minari, J., Takashima, K., Gaydarska, H., Hashiloni-Dolev, Y., andHorn, R. (2023) Old and new challenges regarding comparable and viable data sharing in population-scale genomic research Eur J Hum Genet **31**, 617-618 10.1038/s41431-023-01355-3
1885. Rodriguez Llorian, E., Kopac, N., Waliji, L. A., Borle, K., Dragojlovic, N., Elliott, A. M. *et al.* (2023) A Rapid Review on the Value of Biobanks Containing Genetic Information Value Health **26**, 1286-1295 10.1016/j.jval.2023.02.017

1886. Shojima, N., andYamauchi, T. (2023) Progress in genetics of type 2 diabetes and diabetic complications J Diabetes Investig **14**, 503-515 10.1111/jdi.13970
1887. Suzuki-Barrera, K., Teramoto, A., Saez-Chandia, J., Nakakuki, K., andBracchiglione, J. (2023) Oral Health Interventions in Natural Disasters: A Scoping Review Disaster Med Public Health Prep **17**, e388 10.1017/dmp.2023.62
1888. Tonegawa-Kuji, R., Kanaoka, K., andIwanaga, Y. (2023) Current status of real-world big data research in the cardiovascular field in Japan J Cardiol **81**, 307-315 10.1016/j.jjcc.2022.09.006
1889. Wogayehu, B., Demissie, T., Wolka, E., Alemayehu, M., andDaka, K. (2023) The epidemiology of khat (catha edulis) chewing and alcohol consumption among pregnant women in Ethiopia: A systematic review and meta-analysis PLOS Glob Public Health **3**, e0002248 10.1371/journal.pgph.0002248
1890. Yanai, H. (2023) Parity and Maternal Cardiovascular Disease J Atheroscler Thromb **30**, 107-109 10.5551/jat.ED203
1891. 鎌田真由美, and 河合洋介 (2023) ゲノム医療と日本における取り組みについて JSBi Bioinformatics Review **4**, 81-90 10.11234/jsbibr.2023.2
1892. 三嶋博之 (2023) 難病・希少疾患のためのゲノム医療とバイオインフォマティクス JSBi Bioinformatics Review **4**, 26-34,
1893. 川嶋実苗 (2023) 人を対象とする生命科学・医学系研究において実施した解析データの共有：NBDC ヒトデータベースと TogoVar 薬学図書館 **68**, 28-34,
1894. Ando, M., Fukushima, K., andNishizaki, K. (2024) The discovery of acatalasemia (lack of catalase in the blood) and its significance in human genetics Proc Jpn Acad Ser B Phys Biol Sci **100**, 353-367 10.2183/pjab.100.024
1895. Bouwman, H. B., andGuchelaar, H. J. (2024) The efficacy and safety of eculizumab in patients and the role of C5 polymorphisms Drug Discov Today **29**, 104134 10.1016/j.drudis.2024.104134
1896. Cheng, Y., Lu, J. W., Wang, J. H., Loh, C. H., andChen, T. L. (2024) Associations of Atopic Dermatitis with Attention Deficit/Hyperactivity Disorder and Autism Spectrum Disorder: A Systematic Review and Meta-Analysis Dermatology **240**, 13-25 10.1159/000533366
1897. Cheong, I. H., Garcia, D. L., Kozlakidis, Z., Shao, Y., andWang, H. (2024) Challenges, Rewards, and Digital Aspects in Establishing Macau's First Regional Biobank Innovations in Digital Health, Diagnostics, and Biomarkers **4**, 44-49 10.36401/iddb-23-12
1898. Ezaki, J., Takahashi, Y., Saijo, H., Miya, F., andKosaki, K. (2024) Japanese Public Health Insurance System's new genomic strategic action to shorten the "diagnostic odyssey" for patients with rare and intractable diseases Journal of human genetics **69**, 549-552 10.1038/s10038-024-01285-y
1899. Fang, J., Yang, X., andNi, J. (2024) RNF213 in moyamoya disease: Genotype-phenotype association and the underlying mechanism Chin Med J (Engl) 10.1097/CM9.0000000000002985
1900. Li, W., Ballard, J., Zhao, Y., andLong, Q. (2024) Knowledge-guided learning methods for integrative analysis of multi-omics data Comput Struct Biotechnol J **23**, 1945-1950 10.1016/j.csbj.2024.04.053
1901. Luo, M., Zhu, J., Jia, J., Zhang, H., andZhao, J. (2024) Progress on network modeling and analysis of gut microecology: a review Appl Environ Microbiol **90**, e0009224 10.1128/aem.00092-24
1902. Luo, Y., Meng, X., Cui, L., andWang, S. (2024) Circadian Regulation of Lipid Metabolism during Pregnancy Int J Mol Sci **25**, 10.3390/ijms252111491
1903. Martínez-Gálvez, G., Lee, S., Niwa, R., andWoltjen, K. (2024) On the edge of deletion: Using natural and engineered microhomology to edit the human genome Gene and Genome Editing **7**, 100033 10.1016/j.ggedit.2024.100033

1904. Nakic Rados, S., Hairston, I., andHandelzalts, J. E. (2024) The concept analysis of parent-infant bonding during pregnancy and infancy: a systematic review and meta-synthesis J Reprod Infant Psychol **42**, 142-165 10.1080/02646838.2022.2162487
1905. Nam, Y., Kim, J., Jung, S. H., Woerner, J., Suh, E. H., Lee, D. G. *et al.* (2024) Harnessing Artificial Intelligence in Multimodal Omics Data Integration: Paving the Path for the Next Frontier in Precision Medicine Annu Rev Biomed Data Sci **7**, 225-250 10.1146/annurev-biodatasci-102523-103801
1906. Ovchinnikova, E. V., Garbuz, M. M., Ovchinnikova, A. A., andKumeiko, V. V. (2024) Epidemiology of Wilson's Disease and Pathogenic Variants of the ATP7B Gene Leading to Diversified Protein Disfunctions Int J Mol Sci **25**, 10.3390/ijms25042402
1907. Pashaei Asl, Y., Ghanbari-Homaie, S., Partash, N., Pakzad, A., andFaridaalaee, G. (2024) Consequences of Natural and Man-made Disasters on Pregnancy Outcomes and Complications: A Systematic Review Arch Acad Emerg Med **12**, e61 10.22037/aaem.v12i1.2268
1908. Rivera, N. V. (2024) Big data in sarcoidosis Curr Opin Pulm Med **30**, 561-569 10.1097/mcp.0000000000001102
1909. Sakai, T., Tsuboi, K., Takarada, S., Okabe, M., Nakaoka, H., Ibuki, K. *et al.* (2024) Tachycardia-Induced Cardiomyopathy in an Infant with Atrial Flutter and Prolonged Recovery of Cardiac Function Journal of Clinical Medicine **13**, 3313, https://www.mdpi.com/2077-0383/13/11/3313
1910. Sharma, A., Kumar, A., Saha, P. K., andSaha, L. (2024) The role of TMPRSS6 gene polymorphism in iron resistance iron deficiency anaemia (IRIDA): a systematic review Ann Hematol **103**, 1085-1102 10.1007/s00277-023-05576-w
1911. UEHARA, R. (2024) Epidemiological topics in Japan in the last decade Journal of the National Institute of Public Health **73**, 55-61,
1912. Ward, C., Beharry, A., Tennakoon, R., Rozik, P., Wilhelm, S. D. P., Heinemann, I. U. *et al.* (2024) Mechanisms and Delivery of tRNA Therapeutics Chem Rev **124**, 7976-8008 10.1021/acs.chemrev.4c00142
1913. Yang, C., Fu, R., Wang, H., Jiang, Y., Zhang, S., andJi, X. (2024) Evaluating the global prevalence of insomnia during pregnancy through standardized questionnaires and diagnostic criteria: a systematic review and meta-analysis Front Psychiatry **15**, 1427255 10.3389/fpsyt.2024.1427255
1914. Yeo, N. K., Lim, C. K., Yaung, K. N., Khoo, N. K. H., Arkachaisri, T., Albani, S. *et al.* (2024) Genetic interrogation for sequence and copy number variants in systemic lupus erythematosus Front Genet **15**, 1341272 10.3389/fgene.2024.1341272
1915. 行正信康 (2024) 遺伝子検査のはなし−polymerase chain reaction （PCR） 詳解− 順天堂医療科学雑誌 **2**, 2-12 https://doi.org/10.69235/junmedscij.2.1_2
1916. Aquina, N., Cimoli, B., Das, S., Hovelmanns, K., Weber, F. J., Okonkwo, C. *et al.* (2025) A critical analysis of deployed use cases for quantum key distribution and comparison with post-quantum cryptography EPJ Quantum Technol **12**, 51 10.1140/epjqt/s40507-025-00350-5
1917. Wang, H. F., Liu, S., Cao, Y., andLi, Q. S. (2025) Bidirectional association between atopic dermatitis and attention deficit hyperactivity disorder: a systematic review and meta-analysis Ann Med **57**, 2483370 10.1080/07853890.2025.2483370
1918. Yang, Q., Duan, S., Huang, Y., Liu, C., Wang, M., andHe, G. (2025) The large-scale whole-genome sequencing era expedited medical discovery and clinical translation EngMedicine **2**, 100055,
1919. Zoccali, C., Mallamaci, F., Rosenberg, K., Unwin, R., Silva, P. I., Simeoni, M. A. *et al.* (2025) Big databases and biobanks for studying the links between CKD, cognitive impairment, and dementia Nephrol Dial Transplant **40**,

ii37-ii45 10.1093/ndt/gfae255

学会抄録（1920-1977）

1920. Kume, K., Masamune, A., Nakano, E., Niihori, T., Aoki, Y., Funayama, R. *et al.* (2016) Next generation sequencing might become the new strategy to identify the candidate genes for pancreatitis In Pancreatology, S159-S160
1921. Naito, T., Liu, T.-C., Kakuta, Y., Head, R., Liu, Z., Haritunians, T. *et al.* (2016) Paneth Cell Phenotype is Associated With Novel Genetic Determinants and Clinical Outcome in Japanese Crohn's Disease Patients In DDW 2016, Gastroenterology, San Diego S75
1922. Nakano, E., Masamune, A., Niihori, T., Kume, K., Hamada, S., Aoki, Y. *et al.* (2016) Mutational analysis of the autophagy-related 16 like 1 (ATG16L1) gene in Japanese patients with chronic pancreatitis In 20th Meeting of the International Association of Pancreatology, Sendai, Japan S160
1923. Chiang, L. M., Lee, C. Y., Lai, L. C., Tsai, M. H., Lu, T. P., andChuang, E. Y. (2017) VariED: an integrated database of variants and gene expression profiles for genetic diseases In AACR Annual Meeting 2017, Washington, DC Chapter 3573, 3573-3573
1924. Kakuta, Y., Kawai, Y., Naito, T., Hirano, A., Umeno, J., Hiramoto, K. *et al.* (2017) Population-Optimized Snp Array Reveals Rap1a as a Novel Candidate Susceptibility Gene for Crohn's Disease in Japanese Individuals In DDW 2017, Gastroenterology, Chicago Chapter S78, S78-S78
1925. Hosoe, J., Kadowaki, H., Miya, F., Takakura, M., Tsunoda, T., Shojima, N. *et al.* (2018) Genotype–Phenotype Correlations and Structural Basis of INSR and IGF1R Mutations Causing Severe Insulin/IGF-1 Resistance In American Diabetes Association’s 78th Scientific Sessions, Am Diabetes Assoc, Orlando,
1926. Miyamoto, S., Naruto, T., Morio, T., Urayama, K., Manabe, A., andTakagi, M. (2018) TCF3 Inherited Variants in Children with B-cell Precursor Acute Lymphoblastic Leukemia In 第 60 回日本小児血液・がん学会学術集会 WILEY 111 RIVER ST, HOBOKEN 07030-5774, NJ USA, 京都 S21-S21
1927. Mori, M., Hira, A., Yoshida, K., Muramatsu, H., Okuno, Y., Anmae, M. *et al.* (2018) Characterization of Pathogenic Variants and Clinical Phenotypes in 117 Japanese Fanconi Anemia Patients In 60th ASH Annual Meeting, San Diego, CA Chapter 3860, 3860-3860
1928. Tanaka, M., Ishiura, H., Mitsui, J., Koh, K., Ichinose, Y., Takiyama, Y. *et al.* (2018) A New Method to Detect Potential Causative Genes for Spastic Paraplegia Utilizing Aggregate Data of Whole Exome Sequencing (P2. 456) In AAN 70th Annual Meeting, AAN Enterprises, Los Angeles
1929. 森亮樹 (2018) バイオデータベース:「作る」から「使う」へ In トーゴ―の日シンポジウム 2017, 828-831
1930. 人見祐基 (2018) 日本人原発性胆汁性胆管炎（PBC）感受 性遺伝子領域 NFKB1/MANBA における機能的遺伝 子多型（causal variant）の同定 In 第 54 回日本肝臓学会総会, 大阪
1931. 増永陽平, 福岡哲哉, 大久保由美子, 才津浩智, and 緒方勤 (2018) 低身長・黒色表皮腫の精査により INSR 変異が同定された 1 家系 In 第 28 回臨床内分泌代謝 Update, 135-137
1932. 白月遼 (2018) 小児外科疾患に対するゲノム解析の試み In 第 55 回日本小児外科学会, 新潟
1933. Hirano, A., Shibata, H., Kakuta, Y., Nagasaki, M., Tokunaga, K., Khor, S.-S. *et al.* (2019) The association study between hla genotype and mucosal microbial composition in patients with inflammatory bowel diseases In the 2019 Crohn's and Colitis Congress, Las Vegas S105
1934. Hirotsu, Y., Schmidt-Edelkraut, U., Wang, X., Bohnert, R., Hartenfeller, M., Koch, P. *et al.* (2019) BRCA variant classification is improved by Japanese SNP data and correlate with response to PARP inhibition in MH GUIDE In

2019 the Japanese Spcoety of Medical Oncology Annual Meeting, Kyoto vi106

1935. Ishizuka, K., Aleksic, B., andOzaki, N. (2019) Investigation of Novel Rare Variants in Nrxn1 Contributes to the Increased Risk of Autism Spectrum Disorders and Schizophrenia In European Neuropsychopharmacology, S829-S829

1936. Miyamoto, S., Naruto, T., Morio, T., Urayama, K. Y., Manabe, A., andTakagi, M. (2019) Association of Germline Variants of TCF3 and PAX5 with Pediatric Acute Lymphoblastic Leukemia Development In 61st ASH Annual Meeting, Orland, FL Chapter 1466, 1466-1466

1937. 森田正実 (2019) 未来ヘルスケアへの準備: データ駆動型研究を進めていくための方策 In 産官学少人数懇談会, 248-266

1938. 赤塚浩之 (2019) 研究開発ステージでの AI 活用 In 産官学シンポジウム, 403-409

1939. Okamoto, D., Kakuta, Y., Takeo, N., Moroi, R., Kuroha, M., Kanazawa, Y. *et al.* (2020) Genetic analysis of ulcerative colitis in Japanese individuals using population-specific SNP array In 15th Congress of the European Crohn's and Colitis Organisation (ECCO) 2020, S638-S639

1940. Shinya, Y., Hiraide, T., Kataoka, M., Momoi, M., Goto, S., Katsumata, Y. *et al.* (2020) A novel causative gene variant, TNFRSF13B p.Gly76Ser, in patients with pulmonary arterial hypertension In ESC Congress 2020-The Digital Experience 2310-2310

1941. Tsuchihara, K., Yamashita, R., Yoshino, T., Shitara, K., Watanabe, J., Yasui, H. *et al.* (2020) Clonal and subclonal mutational landscapes in circulating tumor DNA in metastatic colorectal cancer: An exploratory analysis from the phase III PARADIGM study In AACR Annual Meeting 2020, American Association for Cancer Research,

1942. 市川弥生子, 宮崎泰, 炭谷由計, 茂呂直紀, 渋谷裕彦, 永井健太郎 *et al.* (2020) 2 型糖尿病発祥の高リスク疾患関連遺伝子バリアントを有する日本人の臨床像 In 日本人類遺伝学会第 65 回大会, 名古屋

1943. Hirasawa, A., Futagawa, M., Ogawa, C., Aoki1, D., Imoto, I., Matsuo, K. *et al.* (2021) Prospective cohort study and biobanking with Japanese BRCA1/2 pathogenic variant carriers by the Japanese Gynecologic Oncology Group (JGOG) In Eighth International Symposium on hereditary breast and ovarian cancer,

1944. Nguyen, D. T., Dinh, H. Q., Vu, G. M., Nguyen, D. T., andVo, N. S. (2021) A comprehensive imputation-based evaluation of tag SNP selection strategies In 2021 13th International Conference on Knowledge and Systems Engineering (KSE), IEEE, Bangkok, Thailand 1-6

1945. Suharti, S., andWonorahardjo, S. (2021) Utilization of online data resources by final year student in completing final year project on biochemistry and biotechnology In AIP Conference Proceedings 2021, AIP Publishing LLC, 020052

1946. 高井淳 (2021) 東北メディカル・メガバンクのデータベースを用いた転写因子 GATA"の遺伝子多型に関する調査研究 In 第 57 回宮城県公衆衛生学会, web

1947. 西尾信哉, and 宇佐美真一 (2021) 次世代シークエンサーによる遺伝子解析の進歩 In 第 30 回日本耳科学会総会・学術講演会, 北九州市 116-124

1948. 萩原正敏 (2021) 人工知能と化学的介入によるスプライシングコード解読から深部イントロンの VUS 解明へ In 第 48 回日本毒性学会学術年会, 日本毒性学会, S9-2

1949. Barak, T., Yalcin, K., Miyagishima, D., andGunel, M. (2022) ZYX is a Novel Candidate Gene in Moyamoya Pathogenesis In 2022 Annual Meeting of the Congress of Neurological Surgeons, San Francisco, CA, 56

1950. Chiba, T., Miwa, K., Takahashi, A., Mizoguchi, T., Yi, K., Hosoki, S. *et al.* (2022) Genome Wide Association Study Of Intracranial Artery Dissection Reveals Potential Novel Loci In International Stroke Conference 2022, New Orleans, LA ATP199-ATP199

1951. Homma, H., Mika, S., Zempo, H., Kozuma, A., Saito, A., de Almeida, K. Y. *et al.* (2022) Genome-wide association

studies of Japanese weightliftes In The 77th Japanese Society of Physical Fitness and Sports Medicine 2022, Tochigi

1952. Oyanagi, J., Koh, Y., Akiyama, Y., Morimoto, A., Sato, K., Teraoka, S. *et al.* (2022) N-glycan moiety of IgG-Fc region is a prognostic biomarker for advancednon-small cell lung cancertreated with immune checkpoint inhibitors In AACR Annual Meeting 2022, New Orleans, LA 516-516

1953. SUGAWARA, Y., Hirakawa, Y., Nagasu, H., Narita, A., Wada, J., Wada, T. *et al.* (2022) GENOME-WIDE ASSOCIATION STUDY FOR MULTIPLE KIDNEY-RELATED TRAITS IN JAPANESE POPULATION In ISN World Congress of Nephrology (WCN) 2022, Kuala Lumpur, Malaysia S179-S180

1954. Tomioka, Y., Sugimoto, S., Kawana, S., Kubo, Y., Shimizu, D., Matsubara, K. *et al.* (2022) Identification of Single-Nucleotide Polymorphisms Associated with Renal Dysfunction After Lung Transplantation Using Ethnic-Specific SNP Array In ISHLT2022 42nd Annual Meeting & Scientific Sessions, Boston, Massachusetts S254-S255

1955. Yoshihara, H., Otani, T., Nishiyama, T., Omae, Y., Tokunaga, K., Fumiko, O. *et al.* (2022) Genome-wide association study identified meiotic variant associated with aneuploid pregnancy loss In 38th Hybrid Annual Meeting of the ESHRE European Societt of Human Reproduction and Embryology, Milan, Italy deac106. 094

1956. Zempo, H., Miyamoto-Mikami, E., Kikuchi, N., Kamiya, N., Kumagai, H., Takaragawa, M. *et al.* (2022) Genome-wide association study for stress fracture In The 77th Japanese Society of Physical Fitness and Sports Medicine, Tochigi

1957. 吉田誠克 (2022) Alexander 病疑い症例に対する GFAP-ε アイソフォームの遺伝子解析 In 第 63 回日本神経学会学術大会, 東京 S247-S393

1958. 渡耒剛右, 鈴木淳, 池田怜吉, 川瀬哲明, and 香取幸夫 (2022) 東北メディカル・メガバンクの大規模コホートデータを用いた加齢性難聴の疫学研究 In 第 67 回日本聴覚医学会総会・学術講演会, 山形 412-412

1959. 南修司郎, 大石直樹, 高橋優宏, 樫尾明憲, 吉田晴郎, 白井杏湖 *et al.* (2022) ToMMo 健聴データバンクと比較した両側高度重度難聴者の全脳 Surface―based Morphometry In 第 23 回日本耳鼻咽喉科頭頸部外科学会総会・学術講演会, 神戸 769-770

1960. 南修司郎, 大石直樹, 高橋優宏, 樫尾明憲, 吉田晴郎, 白井杏湖 *et al.* (2022) ToMMo の選別聴力検査に基づいた健聴群と難聴群, 及び NHO ネットワーク高度重度難聴群の脳皮質厚解析 In 第 67 回日本聴覚医学会総会・学術講演会, 山形 338-338

1961. Akasaki, Y., Fuse, N., Ogishima, S., Uruno, A., Shimizu, A., Nakamura, T. *et al.* (2023) Integration of Mobile Health and Biobank to Establish the Dry Eye Data Platform In 2023 ARVO Annual Meeting, Investigative Ophthalmology & Visual Science, New Orleans, LA 3953-3953

1962. Wei, X., Enatsu, N., Enatsu, K., Yamada, K., Yamada, A., Nao, H. *et al.* (2023) Genetic mutations in women with recurrent assisted reproductive technology failure In 39th Hybrid Annual Meeting of the ESHRE, Copenhagen, Denmark dead093. 1037

1963. 秋元孝文, 稲葉浩, 篠澤圭子, 三橋彩乃, and 天野景裕 (2023) Investigation of the pathogenic gene variants of von Willebrand disease (VWD) using the Japanese Multi-Omics Reference Panel (jMorp) in the Tohoku Medical Megabank Organization (ToMMo) In 第 192 回東京医科大学医学会総会, 東京医科大学 245-246

1964. 渡井健太郎, 関谷潔史, and 谷口正実 (2023) 喘息およびアレルギー疾患と鑑別を要する化学物質過敏症 In 第 72 回日本アレルギー学会学術大会, 日本アレルギー学会, 東京 729

1965. 務台英樹, 奈良清光, 南修司郎, 仲野敦子, 有本友季子, 守本倫子 *et al.* (2023) 難聴原因遺伝子の全エクソーム解析による効率的な同定 In 第 33 回日本耳科学会総会・学術講演会, 群馬

1966. Deguchi, M., Zempo, H., Homma, H., Saito, M., Kozuma, A., Tsuchiya, Y. *et al.* (2024) SLCO3A1 gene

polymrphism is associatd with recovery of muscle strength after maximaleccentric exercise In The 78th Japanese Society of Physical Fitness and Sports Medicine, The Journal of Physical Fitness and Sports Medicine, Saga

1967. Kozuma, A., Zempo, H., Homma, H., Saito, M., Deguchi, M., Matsumoto, R. *et al.* (2024) Genoome-wide association study and replication study in Japanese wrestlers In The 78th Japanese Society of Physical Fitness and Sports Medicine, The Journal of Physical Fitness and Sports Medicine, Saga
1968. Matsumoto, T., Tsukahara, h., Shiota, M., Goto, S., Mutaguchi, J., Monji, K. *et al.* (2024) ctDNA DETECTION OF NON-CORDING MUTATION IN PLEKHS1 BY DIGITAL PCR ENABLES EARLY PREDICTION OF RECURRENCE IN MUSCLE-INVASIVE BLADDER CANCER In American Urological Association (AUA) 2024 Annual Meeting, The Journal of Urology, San Antonio, Texas e73
1969. Miyashita, A., Hara, N., Obinata, A., Tsukie, T., Hasegawa, M., Kikuchi, M. *et al.* (2024) APOE rare missense variants in Japan In Alzheimer's Association International Confetence 2024, Alzheimer's & Dementia, Philadelphia, PA e088676
1970. Naito, T., Osaka, R., Kakuta, Y., Nagai, H., Shimoyama, Y., Moroi, R. *et al.* (2024) METABOLOME-WIDE MENDELIAN RANDOMIZATION IMPLICATES CAFFEIC ACID AS A PROTECTIVE FACTOR OF ULCERATIVE COLITIS IN JAPANESE POPULATION In DDW 2024, Gastroenterology, Washington, D.C. S-1111-S-1112
1971. Nakamura, S., Sato, Y., Maeda, Y., Sumitani, R., Oura, M., Sogabe, K. *et al.* (2024) Identification of Light Chain Amyloidosis-Associated Single Nucleotide Variants in Japanese People In The 66th ASH Annual Meeting, Blood, San Diego, CA 6867-6867
1972. Soma, R., Hashimoto, K., Sato, K., andNakazawa, T. (2024) Two cases of primary open-angle glaucoma with a rare variant of the MYOC gene In ARVO 2024 Annual Meeting, Investigative Ophthalmology & Visual Science, Seattle, WA, USA 3557-3557
1973. 下川和郎, 石川俊平, 田中博, and 於保祐子 (2024) Influence of heterozygosity in genomic variations of metabolic disorders In 日本人類遺伝学会 第 69 回大会, 札幌
1974. 橋本逸美, 岩澤聡子, 佐藤博基, 伊藤健太, 鈴木聡子, 吉岡範幸 *et al.* (2024) 嗜好飲料からの砂糖摂取量と腎機能（eGFRcys）の関係および嗜好飲料の摂取行動の分析 In 第 35 回日本疫学会学術総会, 高知
1975. 佐藤博基, 岩澤聡子, 橋本逸美, 伊藤健太, 竹内文乃, 角田正史 *et al.* (2024) 身体活動量と腎機能の関連：東北メディカル・メガバンク地域住民コホート In 第 35 回日本疫学会学術総会, 高知
1976. Mariya, T., Idogawa, M., Nakase, H., Tokino, T., Saito, T., andSakurai, A. (2025) Frequency and pathogenicity of hereditary tumor-associated pathogenic variants in the Japanese general population using ToMMo 54KJPN data In 2025 ACMG Annual Clinical Genetics Meeting, Genetics in Medicine Open, Los Angeles, CA 102947
1977. 津川裕司, 三枝大輔, and 有田誠 (2025) 分析の高度化とビッグデータ In Journal of the Mass Spectrometry Society of Japan, Chapter 34, 34-36

学位論文（1978-2014）

1978. Machado, A. J. (2024) Assessment of dihydropyrimidine dehydrogenase dimer formation in human liver samples, The University of Auckland
1979. 出口実 (2024) 伸張性運動後の筋損傷と遺伝子多型との関連性: ゲノムワイド関連解析を用いた検討, 日本体育大学大学院
1980. Abbasian, M. (2025) A computational approach to investigate the structural and functional consequences of residue mutations in human disease and protein design, UCL (University College London)
1981. Edwards, K. (2025) Epigenetic underpinnings of pediatric immune system development, University of British

Columbia

1982. Park, J. (2018) Korean Variant Archive (KOVA): a reference database of genetic variations in the Korean population, ソウル大学大学院
1983. 山本勝利 (2018) 日本人クローン病に対する抗 TNFα 抗体療法の長期治療成績および遺伝子多型と長期成績との関連について, 東北大学大学院
1984. 渋谷祐介 (2018) 次世代シークエンサーを用いた卵巣明細胞癌のゲノム異常の探索, 東北大学大学院
1985. 河野通大 (2019) 全身性エリテマトーデス患者 T 細胞における SRSF1 発現低下は RasGRP1 スプライシング異常を引き起こす, 北海道大学
1986. 岩澤伸哉 (2019) ガラクトース血症の原因遺伝子 galactose mutarotase (GALM) のバリアントの機能解析と GALM 欠損症の保因者頻度および発症頻度の推定, 東北大学大学院
1987. 三浦絵美里 (2019) 東北地方の地域住民コホート研究における強制オシレーション法測定データの分布, 東北大学大学院
1988. Cui, H. (2020) In Silico Edgetic Profiling and Network Analysis of Human Genetic Variants, with an Application to Disease Module Detection, Worcester Polytechnic Institute
1989. Kim, C. (2020) 1,779 whole-genome datasets unveil population-specific genetic architecture and pharmacogenomics profile in Northeast Asian reference panel, ソウル大学大学院
1990. Lilleväli, H. (2020) Hyperphenylalaninaemias and neurophysiological disorders associated with the condition, University of Tartu
1991. 戸澤雄介 (2020) 日本人における遺伝性葉酸吸収不全症の研究, 北海道大学
1992. 高橋雄太 (2020) 機械学習を用いたゲノムワイド遺伝子多型情報に基づくうつ状態のリスク予測, 東北大学大学院
1993. 松平泉 (2020) 視床と大脳基底核の灰白質体積に対する FK506 結合タンパク質 51 (FKBP5) 遺伝子の一塩基多型と母親の受容性の交互作用効果, 東北大学大学院
1994. Jeon, S. (2021) Korean Genome Analysis, Ulsan National Institute of Science and Tevchnology
1995. 吉原紘行 (2021) Polo-like kinase 4 and stromal antigen 3 are not associated with recurrent pregnancy loss caused by embryonic aneuploidy (Polo-like kinase 4・Stromal antigen 3 (STAG3) と絨毛染色体異常による不育症との関連性について)< 内容の要旨及び審査結果の要旨, 名古屋市立大学大学院
1996. 松本昌大 (2021) 日本人侵襲性歯周炎の疾患関連遺伝子 Lipase-A のヒト歯根膜細胞における機能解析, 大阪大学大学院
1997. Ferreira, J. M. G. (2022) Clinical phenotypes in autossomal dominant Alzheimer's disease, Universidade do Porto (Portugal)
1998. Howell, B. (2022) The Contribution of Structural Variants to 2,095 Molecular Phenotypes in 12,354 European Ancestry Individuals, University of Cambridge
1999. Xie, T. (2022) Environmental and Genetic Origins of Hypertension, University of Groningen
2000. 本間洋樹 (2022) 候補遺伝子アプローチおよびゲノムワイド関連解析を用いた筋力・パワー系アスリートの遺伝的特性の検討, 日本体育大学大学院
2001. Qamhia, D. S. (2023) Investigation of the Molecular Basis of Familial Mediterranean Fever-Like Disease in Palestinian population, Al-Quds University
2002. WEI, X. (2023) 体外受精における妊娠率向上に寄与する要因および不妊原因に関する研究, 岡山大学
2003. Zhu, B. (2023) CSF1R 関連白質脳症の分子病態メカニズムの解明, 新潟大学大学院
2004. 下田楓美子 (2023) 日本人クローン病に対する抗 TNFα 抗体療法の長期治療成績に影響を与える臨床的因子と遺伝子多型に関する研究, Tohoku University
2005. 宮部真悟 (2023) 家族発症肺癌の臨床および遺伝学的特徴, Tohoku University
2006. Choi, Y. (2024) Multi-omics analysis of pediatric diseases with Korean genome project ER, Ulsan National

Institute of Science and Technology

2007. Grigio, T. R. (2024) Enhancing the management of postoperative nausea and vomiting: from genetics and pharmacology to daily practice, University of Groningen

2008. Zheng, D. (2024) Investigating evolutionary conservation and population specificity of DNA methylation, University of British Columbia

2009. 大内司 (2024) DNA による個人識別法の強化を図る-超並列シーケンシングによる STR 型検査の実務導入に向けた検討と大規模災害に対するミトコンドリア DNA 検査の重要性の喚起, Tohoku University

2010. 張潔 (2024) 胆嚢癌及び十二指腸乳頭部癌における患者由来腫瘍同所移植 (PDOX) マウスモデルの樹立, 東北大学大学院

2011. 渡来剛右 (2024) 東北メディカル・メガバンク機構のコホート調査で収集したデータを用いた加齢性難聴に関連する生活習慣の疫学研究, Tohoku University

2012. 畠山哲八 (2024) 呼吸機能と関連があるヒト RAGE 一塩基多型ノックインマウスの作製とその機能解析, Tohoku University

2013. 矢尾板久雄 (2024) 日本人集団における総動脈幹症の遺伝学的解析と高頻度病的バリアント TMEM260 遺伝子 c. 1617del の同定, Tohoku University

2014. 鈴木智尚 (2024) 次世代シークエンシング技術を基盤としたヒト希少疾患へのアプローチ, 東北大学大学院