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：2024 年 3 月 7 日

種類		新規件数 2023/11/1 -2024/1/31		これまでの件数 2015 – 2023/10/31		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	34 件	(1-34)	826 件	(35-860)	860 件
	1_2. 標準ゲノム	2 件	(2, 19)	8 件	(730, 828, 861-866)	10 件
	1_3. 全ゲノム	0 件		7 件	(614, 846, 867-871)	7 件
	1_4. SNP Array データ					
	1_4_1. Japonica Array	2 件	(872, 873)	79 件	(96, 113, 114, 126, 160, 205, 229, 372, 375, 492, 614, 620, 631, 812, 868-871, 874-934)	81 件
	1_4_2. OmniExpressExome	0 件		10 件	(614, 926, 935-942)	10 件
	1_5. iMETHYL	2 件	(943, 944)	31 件	(620, 945-974)	33 件
	1_6. その他のゲノムデータ	1 件	(975)	0 件		1 件
	2.　メタボローム情報	0 件		3 件	(492, 976, 977)	3 件
	3.　GWAS レポジトリ	0 件		4 件	(926, 978-980)	4 件
	4. 試料					
	4_1. 細胞	0 件		1 件	(163)	1 件
	4_2. 血清・血漿	0 件		2 件	(981, 982)	2 件
	4_3. DNA	1 件	(975)	3 件	(923, 983, 984)	4 件
	4_4. 母乳	1 件	(985)	0 件		1 件
	5. 健康調査情報					
	5_1. 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	5 件	(975, 985-988)	20 件	(614, 812, 846, 911, 923, 926, 983, 989-1001)	25 件
	5_2. MRI 検査情報	1 件	(987)	0 件		1 件
	6. 施設、技術・方法	0 件		19 件	(320, 1002-1019)	19 件
	小計（※重複を除いた件数）	**43** 件		**976** 件		**1019** 件
知財申請	1.　ゲノム情報	0 件		2 件	(1020, 1021)	2 件
	2.　健康調査情報	0 件		2 件	(1021, 1022)	2 件
	3.　血清	0 件		1 件	(1022)	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	8 件	(1023-1030)	231 件	(1031-1261)	239 件
	2.　レビュー	3 件	(1262-1264)	131 件	(1265-1395)	134 件
	3.　学会抄録	1 件	(1396)	43 件	(1397-1439)	44 件
	4.　学位論文	1 件	(1440)	20 件	(1441-1460)	21 件
	小計（※重複を除いた件数）	**13** 件		**425** 件		**438** 件
合計（※重複を除いた件数）		**56** 件	―	**1404** 件	―	**1460** 件

分譲利用者による成果

種類		新規件数 2023/11/1 -2024/1/31		これまでの件数 2015 – 2023/10/31		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					
	全ゲノムデータ	0 件		1 件	(614)	1 件
	SNP Array データ	0 件		2 件	(614, 923)	2 件
	その他のゲノムデータ　（ミトコンドリアゲノム）	1 件	(975)	0 件		1 件
	メタボローム情報	0 件		0 件		0 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	3 件	(975, 987, 988)	7 件	(614, 923, 983, 991-993, 1001)	10 件
	MRI 検査情報	1 件	(987)	0 件		1 件
	試料:DNA	1 件	(975)	2 件	(923, 983)	3 件
	小計（※重複を除いた件数）	**3** 件		**7** 件		**10** 件
知財申請	ゲノム情報	0 件		1 件	(1021)	1 件
	健康調査情報	0 件		2 件	(1021, 1022)	2 件
	試料：血清	0 件		1 件	(1022)	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）		**3** 件	―	**9** 件	―	**12** 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、iJGVD、Integrative Japanese Genome Variation、JG1、JG2、JRGA、JSV1、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（1-860）

1. 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
2. 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
3. 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
4. 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
5. 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
6. 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
7. 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
8. 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
9. 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
10. Takase, K., Susa, S., Sato, H., Hada, Y., Nagaoka, K., Takakubo, N. *et al.* (2023) Identification of causative gene variants for patients with known monogenic diabetes using a targeted next-generation sequencing panel in a single-center study Diabetology International 10.1007/s13340-023-00669-3
11. 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
12. 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
13. 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
14. 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

15. 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 Molecular Genetics and Metabolism **141**, 108098 10.1016/j.ymgme.2023.108098
16. 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
17. Honda, S., Yamaguchi, H., Aimono, E., Hara, S., Minamiguchi, S., Norose, T. *et al.* (2024) High-grade Solid Pseudopapillary Neoplasms of the Pancreas American Journal of Surgical Pathology **48**, 353-363 10.1097/pas.0000000000002177
18. 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
19. 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
20. 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 10.1038/s10038-023-01217-2
21. 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
22. 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
23. 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 10.1038/s10038-023-01210-9
24. 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 10.1111/cas.16058
25. 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
26. 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
27. 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
28. 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

29. 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
30. 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
31. 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
32. 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
33. 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
34. 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
35. 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
36. 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
37. 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
38. 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
39. 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
40. 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
41. 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

42. 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
43. 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
44. 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
45. 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
46. 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
47. 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
48. 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
49. 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
50. 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
51. 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
52. 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
53. 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
54. 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
55. 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

56. 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
57. 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
58. 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
59. 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
60. 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
61. 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
62. 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
63. 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
64. 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
65. 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
66. 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
67. 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
68. 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

69. 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

70. 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

71. 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

72. 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

73. 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

74. 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

75. 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

76. 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

77. 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

78. 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

79. 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

80. 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

81. 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

82. 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

83. 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

84. 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

85. 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

86. 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

87. 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

88. 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

89. 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

90. 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,

91. 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

92. 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

93. 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

94. 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

95. 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

96. 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

97. 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

98. 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

99. 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

100. 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

101. 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

102. 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

103. 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

104. 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

105. 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

106. 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

107. 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

108. 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

109. 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

110. 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

111. 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

112. 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

113. 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

114. 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

115. 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

116. 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

117. 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

118. 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

119. 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

120. 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

121. 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

122. 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

123. 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

124. 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

125. 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

126. 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

127. 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

128. 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

129. 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

130. 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

131. 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

132. 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

133. 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

134. 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

135. 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

136. 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

137. 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

138. 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

139. 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

140. 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

141. 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

142. 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

143. 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

144. 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

145. 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

146. 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

147. 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

148. 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

149. 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

150. 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

151. 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,

152. 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

153. 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

154. 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

155. 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
156. 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
157. 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
158. 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
159. 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
160. 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
161. 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
162. 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
163. 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
164. 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
165. 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
166. 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
167. 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
168. 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

169. 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

170. 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

171. 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

172. 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

173. 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

174. 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

175. 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

176. 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

177. 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

178. 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

179. 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

180. 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

181. 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

182. 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

183. 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

184. 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

185. 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

186. 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

187. 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

188. 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

189. 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

190. 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

191. 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

192. 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

193. 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

194. 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

195. 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

196. 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

197. 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

198. 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

199. 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

200. 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

201. 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

202. 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

203. 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

204. 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

205. 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

206. 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

207. 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

208. 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

209. 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

210. 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

211. 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
212. 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
213. 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
214. 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
215. 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
216. 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
217. 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
218. 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
219. 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
220. 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
221. 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
222. 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
223. 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
224. 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

225. 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
226. 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
227. 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
228. 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
229. 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
230. 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
231. 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
232. 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
233. 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
234. 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
235. 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
236. 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
237. 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
238. 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

239. 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
240. 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
241. 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
242. 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
243. 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
244. 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
245. 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
246. 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
247. 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
248. 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
249. 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
250. 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
251. 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
252. 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

253. 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
254. 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
255. 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
256. 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
257. 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
258. 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
259. 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
260. 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
261. 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
262. 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
263. 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
264. 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
265. 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
266. 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

267. 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

268. 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

269. 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

270. 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

271. 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

272. 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

273. 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

274. 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

275. 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

276. 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

277. 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

278. 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

279. 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

280. 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

281. 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

282. 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

283. 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

284. 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

285. 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

286. 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

287. 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

288. 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

289. 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

290. 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

291. 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

292. 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

293. 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

294. 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

295. 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
296. 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
297. 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
298. 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
299. 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
300. 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
301. 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
302. 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
303. 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
304. 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
305. 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
306. 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
307. 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
308. 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

309. 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

310. 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

311. 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

312. 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

313. 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

314. 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

315. 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

316. 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

317. 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

318. 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

319. 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

320. 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

321. 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

322. 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

323. 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

324. 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

325. 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

326. 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

327. 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

328. 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

329. 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

330. 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

331. 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

332. 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

333. 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

334. 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

335. 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

336. 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

337. 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

338. 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

339. 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

340. 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

341. 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

342. 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

343. 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

344. 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

345. 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

346. 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

347. 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

348. 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

349. 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

350. 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

351. 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
352. 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
353. 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
354. 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
355. 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
356. 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
357. 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
358. 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
359. 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
360. 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
361. 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
362. 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
363. 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
364. 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

365. 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

366. 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

367. 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

368. 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

369. 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

370. 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

371. 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

372. 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

373. 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

374. 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

375. 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

376. 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

377. 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

378. 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

379. 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

380. 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

381. 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

382. 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

383. 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

384. 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

385. 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

386. 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

387. 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

388. 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

389. 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

390. 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

391. 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

392. 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

393. 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

394. 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
395. 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
396. 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
397. 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
398. 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
399. 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
400. 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
401. 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
402. 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,
403. 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
404. 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
405. 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
406. 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
407. 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

408. 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

409. 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

410. 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

411. 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

412. 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

413. 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

414. 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

415. 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

416. 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

417. 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

418. 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

419. 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

420. 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

421. 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

422. 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

423. 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

424. 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

425. 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

426. 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

427. 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

428. 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

429. 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

430. 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

431. 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

432. 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

433. 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

434. 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

435. 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

436. 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

437. 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

438. 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

439. 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

440. 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

441. 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

442. 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

443. 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

444. 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

445. 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

446. 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

447. 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

448. 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

449. 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

450. 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

451. Katayanagi, S., Setoguchi, Y., Kitagawa, S., Okamoto, T., andMiyazaki, Y. (2021) Alternative gene expression by TOLLIP variant is associated with lung function in chronic hypersensitivity pneumonitis Chest,

452. 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

453. 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

454. 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

455. Kikkawa, Y., Hashimoto, T., Takizawa, K., Urae, S., Masuda, H., Matsunuma, M. *et al.* (2021) Laminin β2 variants associated with isolated nephropathy that impact matrix regulation JCI insight **6**,

456. 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

457. 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

458. 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

459. 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

460. Kubota, T., Nabatame, S., Sato, R., Hama, M., Nishiike, U., Mochizuki, H. *et al.* (2021) Hemiplegic migraine type 2 caused by a novel variant within the P-type ATPase motif in ATP1A2 concomitant with a CACNA1A variant Brain Dev **43**, 952-957 10.1016/j.braindev.2021.05.011

461. 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

462. 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

463. 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

464. 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

465. 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

466. 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

467. 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

468. 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

469. 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 EMD Neurology and Clinical Neuroscience **9**, 490-493 10.1111/ncn3.12552

470. 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

471. 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

472. 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

473. 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

474. 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

475. 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

476. 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

477. 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

478. 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

479. 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

480. 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

481. 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

482. 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

483. 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

484. 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

485. 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

486. 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

487. 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

488. 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

489. 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

490. 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

491. 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

492. 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

493. 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

494. 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

495. 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

496. 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

497. 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

498. 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

499. 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

500. 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

501. 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

502. 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

503. 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

504. 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

505. 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

506. 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

507. 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

508. 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

509. 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

510. 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

511. 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

512. 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

513. 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

514. 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

515. 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

516. 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

517. 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

518. 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

519. 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

520. 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

521. 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

522. 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,

523. 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,

524. 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,

525. 中村, 憲., 松田, 貴., 花岡, 拓., 後藤, 勝., and 松原, 悦. (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

526. 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

527. 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

528. 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**,

529. 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

530. 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

531. 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

532. 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

533. 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

534. 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

535. 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

536. 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

537. 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

538. 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

539. 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

540. 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

541. 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

542. 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

543. 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

544. 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

545. 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

546. 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

547. 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

548. 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

549. 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

550. 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
551. 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
552. 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
553. 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
554. 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
555. 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
556. 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
557. 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
558. 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
559. 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
560. 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
561. 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
562. 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
563. 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

564. 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

565. 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

566. 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

567. 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

568. 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

569. 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

570. 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

571. 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

572. 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

573. 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

574. 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

575. 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

576. 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

577. 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

578. 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

579. 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

580. 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

581. 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

582. 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

583. 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

584. 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

585. 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

586. 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

587. 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

588. 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

589. 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

590. 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

591. 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

592. 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

593. 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

594. 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

595. 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

596. 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

597. 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

598. 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

599. 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

600. 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

601. 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

602. 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

603. 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

604. 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

605. 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

606. 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

607. 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

608. 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

609. 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

610. 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

611. 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

612. 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

613. 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

614. 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

615. 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

616. 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

617. 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

618. 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

619. 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

620. 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

621. 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

622. 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

623. 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

624. 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

625. 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

626. 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

627. 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

628. 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

629. 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

630. 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

631. 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

632. 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
633. 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
634. 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
635. 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
636. 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
637. 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
638. 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
639. 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
640. 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
641. 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
642. 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
643. 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
644. 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
645. 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
646. 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
647. 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
648. 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

649. 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: A Case Report Internal Medicine 8508-8521,

650. 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

651. 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

652. 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

653. 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

654. 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

655. 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

656. 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

657. 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

658. 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

659. 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

660. 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

661. 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

662. 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

663. 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

664. 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

665. 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

666. 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

667. 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

668. 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

669. 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

670. 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

671. 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

672. 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

673. 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

674. 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

675. 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

676. 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

677. 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

678. 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

679. 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

680. 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

681. 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

682. 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

683. 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

684. 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

685. 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

686. 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

687. 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

688. 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

689. 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

690. 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

691. 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

692. 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

693. 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

694. 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

695. 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

696. 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

697. 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

698. 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

699. 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

700. 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,

701. 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,

702. 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

703. 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

704. 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

705. 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

706. Akbari, M., Tapeh, Z. E., 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 investigations Egyptian Journal of Medical Human Genetics **24**, 1-8 ARTN 12 10.1186/s43042-023-00393-2

707. 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

708. 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

709. 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

710. 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

711. Azam, F. K., Sohrabi, B., Rahimi, H., andGanji, M. (2023) Trio whole-exome sequencing reveals a novel mutation in gene in an Iranian patient with hypochondroplasia Gene Reports **31**, 101754 ARTN 101754 10.1016/j.genrep.2023.101754

712. 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

713. 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

714. 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

715. 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

716. 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

717. 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

718. 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 10.1002/ajmg.a.63328
719. 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
720. Furukawa, S., Kushima, I., Aleksic, B., andOzaki, N. (2023) Case reports of two siblings with autism spectrum disorder and 15q13.3 deletions Neuropsychopharmacol Rep 10.1002/npr2.12340
721. 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
722. 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
723. 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
724. 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
725. 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
726. 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
727. 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
728. 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
729. 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
730. 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
731. 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

732. 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
733. 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
734. 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
735. 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
736. 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
737. 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
738. 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
739. 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
740. 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
741. Ito, M., Morimoto, K., Ohfuji, T., Miyabayashi, A., Wakabayashi, K., Yamada, H. *et al.* (2023) FOXJ1 Variants Causing Primary Ciliary Dyskinesia with Hydrocephalus: A Case Report from Japan Intern Med 10.2169/internalmedicine.2565-23
742. 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
743. 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
744. 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
745. 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

746. Kanamori, H., Yamada, Y., Ito, Y., Shirosaki, K., Yamagishi, S., Maeda, Y. *et al.* (2023) Noninvasive graft monitoring using donor-derived cell-free DNA in Japanese liver transplantation Hepatol Res 10.1111/hepr.13978

747. 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

748. 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

749. 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

750. 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

751. 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

752. 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

753. 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

754. 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

755. Kishida, D., Nakamura, A., Yazaki, M., Tsuchiya-Suzuki, A., Ichikawa, T., Shimojima, Y. *et al.* (2023) Variables for differential diagnosis of familial Mediterranean fever: multiple correspondence analysis of a large Japanese cohort Mod Rheumatol 10.1093/mr/road084

756. 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

757. 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

758. 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

759. 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

760. 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

761. Kuroda, Y., Saito, Y., Enomoto, Y., Naruto, T., andKurosawa, K. (2023) A Japanese patient with Teebi hypertelorism syndrome and a novel CDH11 EC1 domain variant Am J Med Genet A 10.1002/ajmg.a.63386
762. 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
763. 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
764. 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
765. 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
766. 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
767. Matsubayashi, H., Kiyozumi, Y., Harada, R., Mukaigawa, T., Sugiura, T., Ishiwatari, H. *et al.* (2023) A Japanese Family Meeting the Clinical Diagnostic Criteria for MEN1 with a MEN1 Variant of Uncertain Significance Intern Med 10.2169/internalmedicine.2300-23
768. 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
769. 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
770. 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
771. Miyamoto, H., Kawakami, F., Abe, S., Sugita, H., andMatsui, H. (2023) Comprehensive Cancer Genomic Profiling of Liver Metastasis Led to the Unexpected Identification of Colorectal Cancer Intern Med 1845-1823 10.2169/internalmedicine.1845-23
772. 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
773. 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
774. 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

775. 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

776. 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 10.1016/j.joen.2023.09.008

777. 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

778. 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

779. Nakano, Y., Taira, Y., Sasaki, R., Tadokoro, K., Yunoki, T., Nomura, E. *et al.* (2023) Novel ABCD1 mutation detected in a symptomatic female carrier of adrenoleukodystrophy Neurology and Clinical Neuroscience **11**, 58-60 10.1111/ncn3.12667

780. 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

781. 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

782. 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

783. 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

784. 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

785. 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

786. 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

787. 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

788. Odate, T., Satomi, K., Kubo, T., Matsushita, Y., Ueno, T., Kurose, A. *et al.* (2023) Inflammatory Rhabdomyoblastic Tumor: Clinicopathologic and Molecular Analysis of 13 Cases Mod Pathol **37**, 100359 10.1016/j.modpat.2023.100359

789. 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

790. 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,

791. 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

792. 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

793. 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

794. 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

795. 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

796. 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

797. 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

798. 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

799. 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

800. 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

801. 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

802. 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

803. Setoguchi, A., Kawano, H., Okano, S., Honda, T., Kato, T., Dateki, S. *et al.* (2023) An Iron-chelating Agent Improved the Cardiac Function in a Patient with Severe Heart Failure Due to Hereditary Hemochromatosis Intern Med 10.2169/internalmedicine.1809-23

804. 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

805. 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

806. 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

807. 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 10.1038/s41416-023-02427-3

808. 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

809. 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

810. 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

811. 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

812. 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

813. 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

814. 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

815. 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

816. 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

817. 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

818. Takegami, N., Matsukawa, T., Hamada, M., Tanifuji, S., Tamura, T., Yamaguchi-Takegami, N. *et al.* (2023) A Case of Adrenomyeloneuropathy with Later Development of Cerebral Form Caused by a Hemizygous Splice-site Variant in ABCD1 Intern Med 10.2169/internalmedicine.2240-23

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

820. 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

821. 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

822. 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 10.1111/hepr.13946

823. 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

824. 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

825. 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

826. 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

827. Torazawa, S., Miyawaki, S., Imai, H., Hongo, H., Ishigami, D., Shimizu, M. *et al.* (2023) RNF213 p.Arg4810Lys Wild Type is Associated with De Novo Hemorrhage in Asymptomatic Hemispheres with Moyamoya Disease Transl Stroke Res 1-10 10.1007/s12975-023-01159-z

828. 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

829. 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

830. Tsuchida, M., Goto, S., Watanabe, H., Goto, S., Yamaguchi, H., andNarita, I. (2023) Defective C3d caused by C3 p.W1034R in inherited atypical hemolytic uremic syndrome Mol Genet Genomic Med e2288 10.1002/mgg3.2288

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

832. Turan, B., Goktas, E., Sonmez, F. M., Aydin, H. I., Aydogdu, D., Zamani, A. G. *et al.* (2023) Cerebral creatine deficiency syndrome with a novel missense variant in SLC6A8 gene Neurology and Clinical Neuroscience **n/a**, 10.1111/ncn3.12726

833. 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

834. 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

835. 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

836. 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: A Case Report Intern Med 10.2169/internalmedicine.1612-23

837. 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

838. 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

839. 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

840. 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

841. 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

842. 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

843. 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

844. 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

845. 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

846. 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

847. 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

848. 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

849. 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

850. 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

851. 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

852. 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

853. 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

854. 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 10.1111/jns.12590

855. 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

856. 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

857. 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,

858. 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,

859. 田村友紀, 落合恵理子, 山崎一樹, and 大澤資樹 (2023) 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 856,

860. 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,

標準ゲノム情報（861-866）

861. 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

862. 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

863. 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

864. 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

865. 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

866. 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

全ゲノム情報（867-871）

867. 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

868. 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

869. 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

870. 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

871. 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

Japonica Array（872-934）

872. 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

873. 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

874. 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

875. 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

876. 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

877. 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

878. 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

879. 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

880. 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

881. 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

882. 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

883. 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

884. 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

885. 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

886. 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

887. 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

888. 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

889. 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

890. 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

891. 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

892. 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

893. 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

894. 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

895. 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

896. 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

897. 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,

898. 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

899. 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

900. 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

901. 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

902. 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

903. 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

904. 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

905. 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

906. 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

907. 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

908. 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

909. 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

910. 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

911. 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

912. 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

913. 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

914. 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

915. 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

916. 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

917. 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

918. 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

919. 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

920. 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

921. 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

922. 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

923. 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

924. 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

925. 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

926. 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

927. 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
928. 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
929. 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
930. 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
931. 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
932. 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
933. 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
934. 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

OmniExpressExome 1 万人分の SNP（935-942）

935. 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
936. 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
937. 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
938. 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
939. 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

940. 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

941. 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

942. 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 (943-974)

943. 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

944. 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

945. 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

946. 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

947. 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

948. 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

949. 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

950. 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

951. 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

952. 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

953. 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
954. 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
955. 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
956. 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
957. 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
958. 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
959. 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
960. 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
961. 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
962. 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
963. 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
964. 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
965. 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
966. 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

967. 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
968. 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
969. 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 10.1136/pmj-2022-141802
970. 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
971. 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
972. 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
973. 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
974. 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

その他のゲノム情報（975）

975. 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

メタボローム情報（976-977）

976. 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
977. Adam, M. G., Limonciel, A., andUstaszewski, B. (2022) Validation and application of a unique reference database for metabolomics–The Quantitative Metabolomics Database (QMDB) biocrates,

GWAS レポジトリ（978-980）

978. 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

979. 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

980. 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

試料：血清･血漿（981-982）

981. 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

982. 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

試料：DNA（983-984）

983. 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

984. Shiraishi, K., Takahashi, A., Momozawa, Y., Daigo, Y., Kaneko, S., Kawaguchi, T. *et al.* (2023) Identification of telomere maintenance gene variations related to lung adenocarcinoma risk by genome-wide association and whole genome sequencing analyses Cancer Commun (Lond) 10.1002/cac2.12498

試料：母乳（985）

985. 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（986-1001）

986. 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

987. 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

988. 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

989. 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

990. 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

991. 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

992. 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

993. 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 10.5551/jat.63816

994. 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

995. 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

996. 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

997. 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

998. 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

999. Okabe, T., Kunikata, H., Yasuda, M., Kodama, S., Maeda, Y., Nakano, J. *et al.* (2023) Relationship between nailfold capillaroscopy parameters and the severity of diabetic retinopathy Graefes Arch Clin Exp Ophthalmol 10.1007/s00417-023-06220-z

1000. 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

1001. 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

施設、技術、方法（1002-1019）

1002. 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**,

1003. 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

1004. 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

1005. 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

1006. 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

1007. 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

1008. 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

1009. 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

1010. 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

1011. 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

1012. 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

1013. 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

1014. 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

1015. 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

1016. 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

1017. 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

1018. 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

1019. 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

知財申請（1020-1022）

1020. 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

1021. 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

1022. 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（1023-1261）

1023. AlOmari, H., Alkhateeb, A., andHammo, B. (2023) Multi-Omics Databases In Machine Learning Methods for Multi-Omics Data Integration, Springer, 151-166

1024. 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

1025. 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,

1026. 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

1027. 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

1028. 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

1029. 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

1030. 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

1031. 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

1032. 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

1033. 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

1034. 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

1035. 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

1036. 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

1037. 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

1038. 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

1039. 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

1040. 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

1041. 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

1042. 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

1043. 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

1044. 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

1045. 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

1046. 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

1047. 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

1048. 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

1049. 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

1050. 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

1051. 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

1052. 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

1053. 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

1054. 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

1055. 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

1056. 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

1057. 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

1058. 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

1059. Dolgova, O., andLao, O. (2018) Evolutionary and Medical Consequences of Archaic Introgression into Modern Human Genomes Genes (Basel) **9**, 358 10.3390/genes9070358

1060. 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

1061. 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

1062. 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

1063. 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

1064. Komar, P., andKural, D. (2018) geck: trio-based comparative benchmarking of variant calls Bioinformatics **34**, 3488-3495 10.1093/bioinformatics/bty415

1065. 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

1066. 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

1067. 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

1068. 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

1069. 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

1070. 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

1071. 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

1072. 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/

1073. 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

1074. 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

1075. 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

1076. 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,

1077. 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

1078. 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

1079. 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

1080. 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

1081. 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

1082. GenomeAsia, K. C. (2019) The GenomeAsia 100K Project enables genetic discoveries across Asia Nature **576**, 106-111 10.1038/s41586-019-1793-z

1083. 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

1084. 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

1085. 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

1086. 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

1087. 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

1088. 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

1089. 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

1090. 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

1091. 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

1092. 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

1093. 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

1094. 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

1095. 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

1096. 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

1097. 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

1098. 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

1099. 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

1100. 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

1101. 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

1102. 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

1103. 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

1104. 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,

1105. 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

1106. 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

1107. 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

1108. 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

1109. 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

1110. 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

1111. 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

1112. 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

1113. 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

1114. 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

1115. 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

1116. 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

1117. 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

1118. 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

1119. Lancaster, S. M., Sanghi, A., Wu, S., andSnyder, M. P. (2020) A Customizable Analysis Flow in Integrative Multi-Omics Biomolecules **10**, 10.3390/biom10121606

1120. 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

1121. 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

1122. 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

1123. 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

1124. 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

1125. 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

1126. 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

1127. 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

1128. 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

1129. 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

1130. 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

1131. 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

1132. 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

1133. 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

1134. 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

1135. 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

1136. 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

1137. 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,

1138. 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

1139. 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

1140. 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

1141. 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.

1142. 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

1143. 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

1144. 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

1145. 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

1146. 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

1147. 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

1148. 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

1149. 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

1150. 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

1151. 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

1152. 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

1153. 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

1154. 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

1155. 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

1156. 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

1157. 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

1158. Kalogeropoulos, A., andPikouli, A. (2021) Natural Disasters: Medical Management In Emergency Medicine, Trauma and Disaster Management, Springer, 433-449

1159. 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

1160. 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

1161. 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

1162. 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

1163. 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

1164. 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

1165. 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

1166. 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

1167. 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

1168. 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

1169. 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

1170. 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

1171. 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

1172. 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

1173. 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

1174. 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

1175. 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

1176. 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

1177. 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

1178. 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

1179. 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

1180. 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

1181. 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

1182. 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

1183. 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

1184. 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

1185. 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

1186. 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

1187. 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,

1188. 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

1189. 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

1190. 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

1191. 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,

1192. 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

1193. 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

1194. 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

1195. 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

1196. 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

1197. 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

1198. 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

1199. 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,

1200. 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

1201. 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

1202. 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

1203. 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

1204. 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

1205. 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

1206. 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

1207. 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

1208. 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

1209. 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

1210. 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

1211. 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

1212. 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

1213. 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

1214. 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

1215. 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

1216. 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

1217. 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

1218. 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

1219. 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

1220. 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

1221. 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

1222. 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

1223. 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

1224. 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

1225. 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

1226. 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

1227. 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

1228. 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,

1229. 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

1230. 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

1231. 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

1232. 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

1233. 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

1234. 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

1235. 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

1236. 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

1237. 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

1238. Harville, E. W., andNorthland, B. (2023) Natural Disaster Epidemiology and Reproductive Health Current Epidemiology Reports 10.1007/s40471-023-00329-9

1239. 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

1240. 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
1241. 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
1242. 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
1243. 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
1244. 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,
1245. 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
1246. 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
1247. 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
1248. 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
1249. Qu, Q., Shi, Y., Guo, Q., Yue, X., Chen, L., Sun, J. *et al.* (2023) 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
1250. 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
1251. 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
1252. 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
1253. 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
1254. 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

1255. 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

1256. 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

1257. 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

1258. 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

1259. 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

1260. 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

1261. 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

レビュー（1262-1395）

1262. 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

1263. 鎌田, 真., and 河合, 洋. (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

1264. 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

1265. 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

1266. 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

1267. 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

1268. 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

1269. Zappala, Z., andMontgomery, S. B. (2016) Non-Coding Loss-of-Function Variation in Human Genomes Hum

Hered **81**, 78-87 10.1159/000447453

1270. An, J. Y. (2017) National human genome projects: an update and an agenda Epidemiol Health **39**, e2017045 10.4178/epih.e2017045

1271. 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

1272. 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

1273. 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

1274. 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

1275. 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

1276. 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

1277. 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

1278. 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,

1279. 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

1280. 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

1281. Satterfield, K., Rubin, J. C., andFriedman, C. P. (2018) Toward a Learning Ecosystem for Diagnostic Excellence

1282. Suzuki, K. (2018) The developing world of DOHaD J Dev Orig Health Dis **9**, 266-269 10.1017/S2040174417000691

1283. 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,

1284. 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,

1285. 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,

1286. 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,

1287. 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

1288. 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

1289. 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

1290. 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

1291. 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

1292. 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

1293. 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

1294. 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

1295. 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

1296. Ohkubo, K. (2019) Cybersecurity Technologies Essential in the Digital Transformation Era International Journal of Informatics Society (IJIS) **11**, 13-21,

1297. Park, S. C., andJeen, Y. T. (2019) Genetic Studies of Inflammatory Bowel Disease-Focusing on Asian Patients Cells **8**, 404-425 10.3390/cells8050404

1298. Sivadas, A., andScaria, V. (2019) Population-scale genomics-Enabling precision public health Adv Genet **103**, 119-161 10.1016/bs.adgen.2018.09.001

1299. 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

1300. Stylianou, E. (2019) Epigenetics of chronic inflammatory diseases J Inflamm Res **12**, 1-14 10.2147/JIR.S129027

1301. 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

1302. 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

1303. Takeoka, M., Fujiwara, M., andSasaki, M. (2019) R&D Trends and Future Prospects of Quantum Cryptography New Breeze 4-8,

1304. 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

1305. 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,

1306. 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,

1307. 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

1308. 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
1309. 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
1310. Cechova, M. (2020) Probably Correct: Rescuing Repeats with Short and Long Reads Genes (Basel) **12**, 48 10.3390/genes12010048
1311. 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
1312. 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
1313. 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
1314. 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
1315. 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
1316. 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
1317. Pan, Z., andXu, S. (2020) Population genomics of East Asian ethnic groups Hereditas **157**, 49 10.1186/s41065-020-00162-w
1318. 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
1319. 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
1320. 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
1321. 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
1322. 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
1323. 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
1324. 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,
1325. 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,
1326. 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,
1327. 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,
1328. 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

1329. 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

1330. 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

1331. Garg, S. (2021) Computational methods for chromosome-scale haplotype reconstruction Genome Biol **22**, 101 10.1186/s13059-021-02328-9

1332. 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

1333. 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

1334. Kiseleva, O., Kurbatov, I., Ilgisonis, E., andPoverennaya, E. (2021) Defining Blood Plasma and Serum Metabolome by GC-MS Metabolites **12**, 15 10.3390/metabo12010015

1335. 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

1336. 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

1337. 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

1338. 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

1339. 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

1340. 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

1341. 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

1342. 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

1343. 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

1344. 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,

1345. 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,

1346. 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,

1347. 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,

1348. 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,

1349. 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

1350. 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

1351. 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

1352. 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

1353. 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

1354. 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

1355. 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

1356. 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

1357. 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

1358. 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

1359. 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

1360. 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

1361. 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

1362. 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

1363. 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

1364. 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

1365. Miller, C. L. (2022) The Epigenetics of Psychosis: A Structured Review with Representative Loci Biomedicines **10**, 561 10.3390/biomedicines10030561

1366. Murakami, Y. (2022) Cohorts and Biobanks as Essential Resources for Cancer Research Gan to Kagaku ryoho Cancer & Chemotherapy **49**, 129-132,

1367. Nasution, D. R. S., andYamamoto, Z. (2022) Salivary Microflora and Its Diagnostic Importance Buletin Farmatera **7**, 84-93,

1368. 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

1369. Ogasawara, O. (2022) Building cloud computing environments for genome analysis in Japan Hum Genome Var **9**, 46 10.1038/s41439-022-00223-8

1370. Omae, Y., Goto, Y. I., andTokunaga, K. (2022) National Center Biobank Network Hum Genome Var **9**, 38 10.1038/s41439-022-00217-6

1371. 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

1372. 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

1373. Swan, M., dos Santos, R. P., andWitte, F. (2022) Quantum Neurobiology Quantum Reports **4**, 107-126,

1374. 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

1375. Tsuchida, S., andNakayama, T. (2022) Metabolomics Research in Periodontal Disease by Mass Spectrometry Molecules **27**, 2864 10.3390/molecules27092864

1376. 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

1377. 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

1378. 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

1379. 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

1380. 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

1381. 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

1382. 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

1383. 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

1384. 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

1385. 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

1386. 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

1387. 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

1388. 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

1389. 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

1390. 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

1391. 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

1392. 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

1393. Yanai, H. (2023) Parity and Maternal Cardiovascular Disease J Atheroscler Thromb **30**, 107-109 10.5551/jat.ED203

1394. 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,

1395. 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,

学会抄録（1396-1439）

1396. 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

1397. 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

1398. 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 2016 DDW, San Diego S75

1399. 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

1400. 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

1401. 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 2017 DDW, Chicago Chapter S78, S78-S78

1402. 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,

1403. 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

1404. 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

1405. 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

1406. 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

1407. 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

1408. 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

1409. 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

1410. 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

1411. 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

1412. 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

1413. 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

1414. 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

1415. 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

1416. 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

1417. 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

1418. 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,

1419. 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

1420. 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,

1421. 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

1422. 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

1423. 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

1424. 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

1425. 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

1426. 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

1427. 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

1428. 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

1429. 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

1430. 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

1431. 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

1432. 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

1433. 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

1434. 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

1435. 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

1436. 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

1437. 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

1438. 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

1439. 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

学位論文（1440-1460）

1440. 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

1441. 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

1442. 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

1443. 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

1444. 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

1445. 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

1446. 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

1447. Cui, H. (2020) In Silico Edgetic Profiling and Network Analysis of Human Genetic Variants, with an Application to Disease Module Detection, Worcester Polytechnic Institute

1448. 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

1449. Lilleväli, H. (2020) Hyperphenylalaninaemias and neurophysiological disorders associated with the condition, University of Tartu

1450. 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

1451. 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

1452. 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

1453. Jeon, S. (2021) Korean Genome Analysis, Ulsan National Institute of Science and Tevchnology

1454. 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

1455. 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

1456. Ferreira, J. M. G. (2022) Clinical phenotypes in autossomal dominant Alzheimer's disease, Universidade do Porto (Portugal)

1457. Howell, B. (2022) The Contribution of Structural Variants to 2,095 Molecular Phenotypes in 12,354 European Ancestry Individuals, University of Cambridge

1458. Xie, T. (2022) Environmental and Genetic Origins of Hypertension, University of Groningen

1459. 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

1460. 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