

**書籍**

著者氏名	論文タイトル	書籍全体の編集者、筆頭著者、監修など	書籍名	出版社名	出版地	出版年	頁
仁科 幸子	小児や障害児に適した眼鏡	日本近視学会・日本小児眼科学会・日本視能訓練士協会編	小児の近視診断と治療 第2版	三輪書店	東京	2023	144-147
仁科 幸子	疾患の早期発見の必要性	日本小児眼科学会編	やさしい小児の眼科	診断と治療社	東京	2023	90-96
仁科 幸子	簡便な機器を用いた乳幼児の眼科健診	五十嵐隆、中林正雄、竹田省編	母子保健マニュアル改定8版	南山堂	東京	2023	144
松永 達雄	盲ろうの生理・病理学的特徴	樫木暢子、金森克浩、船橋篤彦・編集.	特別支援教育免許シリーズ 重複障害教育領域① 複数の困難への対応	建帛社	東京	2023	14-23
松永 達雄	ミトコンドリア難聴とはどのような疾患ですか？	村山圭、小坂仁、三牧正和・編集.	ミトコンドリア病診療マニュアル 2023	診断と治療社	東京	2023	258-259
松永 達雄	ミトコンドリア難聴	小須賀基通、小林正久、野口篤子・編集.	先天代謝異常症クリニカルファイアル	診断と治療社	東京	2024	422-423
小崎 里華	18トリソミー	加藤 元博	小児科診療ガイドライン	総合医学社	東京	2023	763-766
小崎 里華	主な外表奇形	五十嵐 隆	母子保健マニュアル	南山堂	東京	2023	286
小崎 里華	主な染色体異常症	五十嵐 隆	母子保健マニュアル	南山堂	東京	2023	287-288,

雑誌

発表者氏名	論文タイトル名	発表誌名	巻号	頁	出版年
Akaba Y, Takahashi S, Suzuki K, <u>Kosaki K</u> , Tsujimura K	miR-514a promotes neuronal development in human iPSC-derived neurons	Frontiers in Cell and Developmental Biology	11		2023
Hama EY, Yamaguchi S, Uchiyama K, Kojima D, Nagasaka T, Yoshimoto N, Tajima T, Kanda T, Morimoto K, Yoshida T, <u>Kosaki K</u> , Itoh H, Hayashi K	Successful renal transplantation following hemodialysis as bridging therapy in a patient with Fechtner syndrome: a case report and literature review	Renal Replacement Therapy	9(1)		2023
Kaneko S, Sakura F, Tanita K, Shimbo A, Nambu R, Yoshida M, Umetsu S, Inui A, Okada C, Tsumura M, Yamada M, Suzuki H, <u>Kosaki K</u> , Ohara O, Shimizu M, Morio T, Okada S, Kanegae H	Janus kinase inhibitors ameliorate clinical symptoms in patients with STAT3 gain-of-function	Immunotherapy Advances	3(1)		2023
Kuroda A, Namkoong H, Iwami E, Tsutsumi A, Nakajima T, Shinoda H, Katada Y, Iimura J, Suzuki H, <u>Kosaki K</u> , Terashima T	X-linked inheritance of primary ciliary dyskinesia and retinitis pigmentosa due to RPGR variant: A case report and literature review	Respirology Case Reports	11(1 2)		2023
Nishio Y, Kato K, Tran Mau-Them F, Futagawa H, Quélin C, Masuda S, Vitobello A, Otsuji S, Shawki HH, Oishi H, Thauvin-Robinet C, Takenouchi T, <u>Kosaki K</u> , Takahashi Y, Saitoh S	Gain-of-function MYCN causes a megalencephaly-polydactyly syndrome manifesting mirror phenotypes of Feingold syndrome	Human Genetics and Genomics Advances	4(4)		2023
Nitta Y, Kawai H, Maki R, Osaka J, Hakeda-Suzuki S, Nagai Y, Doubková K, Uehara T, Watanabe K, <u>Kosaki K</u> , Suzuki T, Tavosanis G, Sugie A	Direct evaluation of neuroaxonal degeneration with the causative genes of neurodegenerative diseases in <i>Drosophila</i> using the automated axon quantification system, MeDUsA	Human Molecular Genetics	32(9)	1524-38	2023
Ogawa E, Hishiki T, Hayakawa N, Suzuki H, <u>Kosaki K</u> , Suematsu M, Takenouchi T	Ketogenic diet in action: Metabolic profiling of pyruvate dehydrogenase deficiency	Molecular Genetics and Metabolism Reports	35		2023

Shima Y, Sasagawa S, Ota N, Oyama R, Tanaka M, Kubota-Sakashita M, Kawakami H, Kobayashi M, Takubo N, Ozeki AN, Sun X, Kim YJ, Kamatani Y, Matsuda K, Maejima K, Fujita M, Noda K, Kamiyama H, Tanikawa R, Nagane M, Shibahara J, Tanaka T, Rikitake Y, Mataga N, Takahashi S, <u>Kosaki</u> K, Okano H, Furihata T, Nakaki R, Akimitsu N, Wada Y, Ohtsuka T, Kurihara H, Kamiguchi H, Okabe S, Nakafuku M, Kato T, Nakagawa H, Saito N, Nakatomi H	Increased PDGFRB and NF-κB signaling caused by highly prevalent somatic mutations in intracranial aneurysms	Science Translational Medicine	15(7 00)		2023
Takahashi I, Noguchi A, Kondo D, Sato Y, Suzuki H, Yamada M, <u>Kosaki</u> K, Takahashi T	A novel missense variant of FGD1 disrupts critical cysteine residues of the FYVE domain in Japanese siblings with Aarskog–Scott syndrome	Clinical Pediatric Endocrinology	33(1)	39-42	2024
Yamada M, Tanito K, Suzuki H, Nakato D, Miya F, Takenouchi T, <u>Kosaki</u> K	Café-au-lait Spots and Cleft Palate: Not a Chance Association	Cleft Palate Craniofacial Journal			2023
Yamashita T, Hotta J, Jogu Y, Sakai E, Ono C, Bamba H, Suzuki H, Yamada M, Takenouchi T, <u>Kosaki</u> K, Yorifuji T, Hamazaki T, Seto T	Oculofaciocardiodental syndrome caused by a novel BCOR variant	Human Genome Variation	10(1)		2023
Yamashita T, Tanaka Y, Miwatani T, <u>Okamoto</u> N, Takenouchi T, Uehara T, Suzuki H, <u>Kosaki</u> K, Nishigaki T	Case of ADNP syndrome with autism spectrum disorder, intellectual disability, and characteristic early eruption of primary teeth	No To Hattatsu	56(2)	142-4	2024
Yoshida Y, Uchida K, Kodo K, Ishizaki-Asami R, Maeda J, Katsumata Y, Yuasa S, Fukuda K, <u>Kosaki</u> K, Watanabe Y, Nakagawa O, Yamagishi H	A genetic and developmental biological approach for a family with complex congenital heart diseases—evidence of digenic inheritance	Frontiers in Cardiovascular Medicine	10		2023

Yoshikawa Y, Koto T, Ishida T, Uehara T, Yamada M, <u>Kosaki K</u> , Inoue M	Rhegmatogenous Retinal Detachment in Musculocontractural Ehlers–Danlos Syndrome Caused by Biallelic Loss-of-Function Variants of Gene for Dermatan Sulfate Epimerase	Journal of Clinical Medicine	12(5)		2023
Otsuji S, Nishio Y, Tsujita M, Rio M, Huber C, Anton-Plagaro C, <u>Mizuno S</u> , Kawano Y, Miyatake S, Simon M, van Binsbergen E, van Jaarsveld RH, Matsumoto N, Cormier-Daire V, P JC, Saitoh S, Kato K	Clinical diversity and molecular mechanism of VPS35L-associated Ritscher-Schinzel syndrome	J Med Genet	60(4)	359-67	2023
Fujimoto M, Nakamura Y, Iwaki T, Sato E, Ieda D, Hattori A, Shiraki A, <u>Mizuno S</u> , Saitoh S	Angelman syndrome with mosaic paternal uniparental disomy suggestive of mitotic nondisjunction	J Hum Genet	68(2)	87-90	2023
Fukahori K, Yamoto K, Saitsu H, <u>Ogata T</u> , Nagasaki K	PORCN-related microphthalmia with limb anomalies: Case report and literature review	Am J Med Genet A	191(2)	636-9	2023
Komatsu K, Sakaguchi K, Shimizu D, Yamoto K, Kato F, Miyairi I, <u>Ogata T</u> , Saitsu H	Characterization of KMT2A::MATR3 fusion in a patient with acute lymphoblastic leukemia and monitoring of minimal residual disease by nanoplate digital PCR	Pediatr Blood Cancer	70(4)	e301-20	2023
Sakamoto M, Shiiki T, Matsui S, <u>Okamoto N</u> , Koshimizu E, Tsuchida N, Uchiyama Y, Hamanaka K, Fujita A, Miyatake S, Misawa K, Mizuguchi T, Matsumoto N	A novel homozygous CHMP1A variant arising from segmental uniparental disomy causes pontocerebellar hypoplasia type 8	J Hum Genet	68(4)	247-53	2023
Kuroda Y, Kumaki T, Saito Y, Enomoto Y, Suzuki H, Takenouchi T, <u>Kosaki K</u> , <u>Kurosawa K</u>	A novel variant of ARPC4-related neurodevelopmental disorder	Am J Med Genet A	191(3)	893-5	2023
Imai Y, Nagaya S, Araiso Y, Meguro-Horike M, Togashi T, Ohmori K, Makita Y,	Identification and functional analysis of three novel genetic variants resulting in premature termination	Int J Hematol	117(4)	523-9	2023

Sato E, Yujiri T, Nagamori Y, Horike SI, <u>Watanabe A</u> , Morishita E	codons in three unrelated patients with hereditary antithrombin deficiency				
Inoue Y, Tsuchida N, <u>Okamoto N</u> , Shuichi S, Ohashi K, Saitoh S, Ogawa A, Hamada K, Sakamoto M, Miyake N, Hamanaka K, Fujita A, Koshimizu E, Miyatake S, Mizuguchi T, Ogata K, Uchiyama Y, Matsumoto N	Three KINSHIP syndrome patients with mosaic and germline AFF3 variants	Clin Genet	103(5)	590-5	2023
Yamada M, Okuno H, <u>Okamoto N</u> , Suzuki H, Miya F, Takenouchi T, <u>Kosaki K</u>	Diagnosis of Prader-Willi syndrome and Angelman syndrome by targeted nanopore long-read sequencing	Eur J Med Genet	66(2)	1046 90	2023
Yoshihamo K, Mutai H, Sekimizu M, Ito F, Saito S, Nakamura S, Mikoshiba T, Nagai R, Takebayashi A, Miya F, <u>Kosaki K</u> , Ozawa H, <u>Matsunaga T</u>	Molecular basis of carotid body tumor and associated clinical features in Japan identified by genomic, immunohistochemical, and clinical analyses	Clin Genet	103(4)	466-71	2023
Nitta Y, Kawai H, Maki R, Osaka J, Hakeda-Suzuki S, Nagai Y, Doubkova K, Uehara T, Watanabe K, <u>Kosaki K</u> , Suzuki T, Tavosanis G, Sugie A	Direct evaluation of neuroaxonal degeneration with the causative genes of neurodegenerative diseases in <i>Drosophila</i> using the automated axon quantification system, MeDUsA	Hum Mol Genet	32(9)	1524 -38	2023
Seyama R, Uchiyama Y, Kaneshi Y, Hamanaka K, Fujita A, Tsuchida N, Koshimizu E, Misawa K, Miyatake S, Mizuguchi T, Makino S, Itakura A, <u>Okamoto N</u> , Matsumoto N	Distal arthrogryposis in a girl arising from a novel TNNI2 variant inherited from paternal somatic mosaicism	J Hum Genet	68(5)	363-7	2023
Hayashi T, Yano N, Kora K, Yokoyama A, Maizuru K, Kayaki T, Nishikawa K, Osawa M, Niwa A, Takenouchi T, Hijikata A, Shirai T, Suzuki H, <u>Kosaki K</u> , Saito MK, Takita J,	Involvement of mTOR pathway in neurodegeneration in NSF-related developmental and epileptic encephalopathy	Hum Mol Genet	32(10)	1683 -97	2023

Yoshida T					
Itai T, Sugie A, Nitta Y, Maki R, Suzuki T, Shinkai Y, Watanabe Y, Nakano Y, Ichikawa K, <u>Okamoto N</u> , Utsuno Y, Koshimizu E, Fujita A, Hamanaka K, Uchiyama Y, Tsuchida N, Miyake N, Misawa K, Mizuguchi T, Miyatake S, Matsumoto N	A novel NONO variant that causes developmental delay and cardiac phenotypes	Sci Rep	13(1)	975	2023
Uwamino Y, Yokoyama T, Sato Y, Shibata A, Kurafuji T, Tanabe A, Noguchi M, Arai T, Ohno A, Yokota H, Namkoong H, <u>Nishimura T, Kosaki K</u> , Hasegawa N, Wakui M, Murata M, Matsushita H	Humoral and cellular immune response dynamics in Japanese healthcare workers up to six months after receiving a third dose of BNT162b2 monovalent vaccine	Vaccine	41(9)	1545 -9	2023
Hirano S, Suzuki Y, Ikeda T, <u>Okamoto N</u>	Time course of serum neuron-specific enolase levels from infancy to early adulthood in a female patient with beta-propeller protein-associated neurodegeneration	Am J Med Genet A	191(5)	1384 -7	2023
Kawai M, Muroya K, Murakami N, Ihara H, Takahashi Y, Horikawa R, <u>Ogata T</u>	A questionnaire-based survey of medical conditions in adults with Prader-Willi syndrome in Japan: implications for transitional care	Endocr J	70(5)	519-28	2023
Azuma N, Yokoi T, Tanaka T, Matsuzaka E, Saida Y, <u>Nishina S</u> , Terao M, Takada S, Fukami M, Okamura K, Maehara K, Yamasaki T, Hirayama J, Nishina H, Handa H, Yamaguchi Y	Integrator complex subunit 15 controls mRNA splicing and is critical for eye development	Hum Mol Genet	32(12)	2032 -45	2023
Shen XM, Nakata T, <u>Mizuno S</u> , Imoto I, Selcen D, Ohno K, Engel AG	Impaired gating of gamma- and epsilon-AChR respectively causes Escobar syndrome and fast-channel myasthenia	Ann Clin Transl Neurol	10(5)	732-43	2023

Hiraide T, Shimizu K, Okumura Y, Miyamoto S, Nakashima M, <u>Ogata T</u> , Saitsu H	A deep intronic TCTN2 variant activating a cryptic exon predicted by SpliceRover in a patient with Joubert syndrome	J Hum Genet	68(7)	499-505	2023
Yamamoto Y, Higashimoto K, Ohkawa Y, <u>Soejima H</u> , Kaneko K, Ohmi Y, Furukawa K, Furukawa K	Possible regulation of ganglioside GD3 synthase gene expression with DNA methylation in human glioma cells	Glycoconj J	40(3)	323-32	2023
Tsuzuki N, Namba K, Saegusa C, Mutai H, Nishiyama T, Oishi N, <u>Matsunaga T</u> , Fujioka M, Ozawa H	Apoptosis of type I spiral ganglion neuron cells in Otof-mutant mice	Neurosci Lett	803	1371 78	2023
Fukui K, Amari S, Yotani N, <u>Kosaki R</u> , Hata K, Kosuga M, Sago H, Isayama T, Ito Y	A Neonate with Mucopolysaccharidosis Type VII with Intractable Ascites	AJP Rep	13(1)	e25-e8	2023
Morikawa H, <u>Nishina S</u> , Torii K, Hosono K, Yokoi T, Shigeyasu C, Yamada M, Kosuga M, Fukami M, Saitsu H, Azuma N, Hori Y, Hotta Y	A pediatric case of congenital stromal corneal dystrophy caused by the novel variant c.953del of the DCN gene	Hum Genome Var	10(1)	9	2023
Nabatame S, Tanigawa J, Tominaga K, Kagitani-Shimono K, Yanagihara K, Imai K, Ando T, Tsuyusaki Y, Araya N, Matsufuji M, Natsume J, Yuge K, Bratkovic D, Arai H, Okinaga T, Matsushige T, Azuma Y, Ishihara N, Miyatake S, Kato M, Matsumoto N, <u>Okamoto N</u> , Takahashi S, Hattori S, Ozono K	Association between cerebrospinal fluid parameters and developmental and neurological status in glucose transporter 1 deficiency syndrome	J Neurol Sci	447	1205 97	2023
Kimizu T, Ida S, Oki K, Shima M, Nishimoto S, Nakajima K, Ikeda T, Mogami Y, Yanagihara K, Matsuda K, Nishi E, Hasegawa Y, Nozaki M, Fujita H, Irie A, Katayama T, <u>Okamoto</u>	Newborn screening for spinal muscular atrophy in Osaka -challenges in a Japanese pilot study	Brain Dev	45(7)	363-71	2023

<u>N</u> , Imai K, Nishio H, Suzuki Y					
Miyake N, Tsurusaki Y, Fukai R, Kushima I, <u>Okamoto N</u> , Ohashi K, Nakamura K, Hashimoto R, Hiraki Y, Son S, Kato M, Sakai Y, Osaka H, Deguchi K, Matsuishi T, Takeshita S, Fattal-Valevski A, Ekhilevitch N, Tohyama J, Yap P, Keng WT, Kobayashi H, Takubo K, Okada T, Saitoh S, Yasuda Y, Murai T, Nakamura K, Ohga S, Matsumoto A, Inoue K, Saikusa T, Hershkovitz T, Kobayashi Y, Morikawa M, Ito A, Hara T, Uno Y, Seiwa C, Ishizuka K, Shirahata E, Fujita A, Koshimizu E, Miyatake S, Takata A, Mizuguchi T, Ozaki N, Matsumoto N	Molecular diagnosis of 405 individuals with autism spectrum disorder	Eur J Hum Genet			2023
Masunaga Y, Fujisawa Y, Massart F, Spinelli C, Kojima Y, Mizuno K, Hayashi Y, Sasagawa I, Yoshida R, Kato F, Fukami M, Kamatani N, Saitsu H, <u>Ogata T</u>	Microdeletion at ESR1 Intron 6 (DEL_6_75504) Is a Susceptibility Factor for Cryptorchidism and Hypospadias	J Clin Endocrinol Metab	108(10)	2550-60	2023
Murofushi Y, Hayakawa I, Kawai M, Abe Y, <u>Kosaki R</u> , Suzuki H, Takenouchi T, Kubota M	Oral Baclofen Therapy for Multifocal Spinal Myoclonus with TBC1D24 Variant	Mov Disord Clin Pract	10(4)	719-21	2023
Oishi N, Noguchi M, Fujioka M, Nara K, Wasano K, Mutai H, Kawakita R, Tamura R, Karatsu K, Morimoto Y, Toda M, Ozawa H, <u>Matsunaga T</u>	Correlation between genotype and phenotype with special attention to hearing in 14 Japanese cases of NF2-related schwannomatosis	Sci Rep	13(1)	6595	2023
Goma M, Hagiwara SI, Wada T, Maeyama T, <u>Okamoto N</u> , Ishii S,	A case of early-onset idiopathic chronic pancreatitis associated with a	Clin J Gastroenterol	16(4)	623-7	2023

Etani Y, Masamune A	loss-of-function TRPV6 p.R483Q variant successfully treated by pancreatic duct stenting				
Unuma K, Tomomasa D, Noma K, Yamamoto K, Matsuyama TA, Makino Y, Hijikata A, Wen S, <u>Ogata T</u> , <u>Okamoto N</u> , Okada S, Ohashi K, Uemura K, Kanegane H	Case Report: Molecular autopsy underlie COVID-19-associated sudden, unexplained child mortality	Front Immunol	14	##### #	2023
Hara-Isono K, Matsubara K, Nakamura A, Sano S, Inoue T, Kawashima S, Fuke T, Yamazawa K, Fukami M, <u>Ogata T</u> , Kagami M	Risk assessment of assisted reproductive technology and parental age at childbirth for the development of uniparental disomy-mediated imprinting disorders caused by aneuploid gametes	Clin Epigenetics	15(1)	78	2023
Pintus E, Chinn AF, Kadlec M, Garcia-Vazquez FA, Novy P, Matson JB, Ross-Santaella JL	Correction to: N-thiocarboxyanhydrides, amino acid-derived enzyme-activated H(2)S donors, enhance sperm mitochondrial activity in presence and absence of oxidative stress	BMC Vet Res	19(1)	71	2023
Yamaguchi T, Hayashi S, Nagai S, Uchiyama A, Motegi SI, Fujikawa T, Takiguchi Y, <u>Kosho T</u>	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	##### #	2023
Kobayashi T, Fujishima F, Tokodai K, Sato C, Kamei T, Miyake N, Matsumoto N, <u>Kosho T</u>	Detailed Courses and Pathological Findings of Colonic Perforation without Diverticula in Sisters with Musculocontractural Ehlers-Danlos Syndrome Caused by Pathogenic Variant in CHST14 (mcEDS-CHST14)	Genes (Basel)	14(5)		2023
Ouchi S, Ishii K, <u>Kosaki K</u> , Suzuki H, Yamada M, Takenouchi T, Tamaoka A	Parkinsonism in spinocerebellar ataxia with axonal neuropathy caused by adult-onset COA7 variants: a case report	BMC Neurol	23(1)	211	2023
Shima Y, Sasagawa S, Ota N, Oyama R, Tanaka M, Kubota-	Increased PDGFRB and NF-kappaB signaling caused by highly prevalent somatic	Sci Transl Med	15(700)	eabq 7721	2023

Sakashita M, Kawakami H, Kobayashi M, Takubo N, Ozeki AN, Sun X, Kim YJ, Kamatani Y, Matsuda K, Maejima K, Fujita M, Noda K, Kamiyama H, Tanikawa R, Nagane M, Shibahara J, Tanaka T, Rikitake Y, Mataga N, Takahashi S, <u>Kosaki</u> <u>K.</u> , Okano H, Furihata T, Nakaki R, Akimitsu N, Wada Y, Ohtsuka T, Kurihara H, Kamiguchi H, Okabe S, Nakafuku M, Kato T, Nakagawa H, Saito N, Nakatomi H	mutations in intracranial aneurysms				
Kume K, Kurashige T, Muguruma K, Morino H, Tada Y, Kikumoto M, Miyamoto T, Akutsu SN, Matsuda Y, <u>Matsuura</u> <u>S.</u> , Nakamori M, Nishiyama A, Izumi R, Niihori T, Ogasawara M, Eura N, Kato T, Yokomura M, Nakayama Y, Ito H, Nakamura M, Saito K, Riku Y, Iwasaki Y, Maruyama H, <u>Aoki</u> <u>Y.</u> , Nishino I, Izumi Y, Aoki M, Kawakami H	CGG repeat expansion in LRP12 in amyotrophic lateral sclerosis	Am J Hum Genet	110(7)	1086-97	2023
Furuhataya-Yoshimura M, Yamaguchi T, Izu Y, <u>Kosho</u> <u>T</u>	Homozygous splice site variant affecting the first von Willebrand factor A domain of COL12A1 in a patient with myopathic Ehlers- Danlos syndrome	Am J Med Genet A	191(10)	2672-37	2023
Tanabe Y, Nomura N, Minami M, Takaya J, <u>Okamoto</u> <u>N.</u> , Yanagi K, Kaname T, Fujii Y, Kaneko K	HIST1H1E syndrome with deficiency in multiple pituitary hormones	Clin Pediatr Endocrinol	32(3)	195-8	2023
Yamada M, Nitta Y, Uehara T, Suzuki H, Miya F, Takenouchi T, Tamura M, Ayabe S, Yoshiki A, Maeno A, Saga Y, Furuse T,	Heterozygous loss-of- function DHX9 variants are associated with neurodevelopmental disorders: Human genetic and experimental evidences	Eur J Med Genet	66(8)	1048-04	2023

<u>Yamada I</u> , <u>Okamoto N</u> , <u>Kosaki K</u> , Sugie A					
Nagaya S, Togashi T, Akiyama M, Imai Y, Matsumoto H, Moriya H, Meguro-Horike M, Yasuda I, Kikuchi Y, Kuwajima Y, Horike SI, <u>Watanabe A</u> , Morishita E	Protein S deficiency caused by cryptic splicing due to the novel intron variant c.346+5G>C in PROS1	Thromb Res	229	26-30	2023
Shimojima Yamamoto K, Tamura T, <u>Okamoto N</u> , Nishi E, Noguchi A, Takahashi I, Sawaishi Y, Shimizu M, Kanno H, Minakuchi Y, Toyoda A, Yamamoto T	Identification of small-sized intrachromosomal segments at the ends of INV-DUP-DEL patterns	J Hum Genet	68(1)	751-7	2023
Okubo Y, Shibuya M, Nakamura H, Kawashima A, Kodama K, Endo W, Inui T, Togashi N, Aihara Y, Shirota M, Funayama R, Niihori T, Fujita A, Nakayama K, <u>Aoki Y</u> , Matsumoto N, Kure S, Kikuchi A, Haginoya K	Neonatal developmental and epileptic encephalopathy with movement disorders and arthrogryposis: A case report with a novel missense variant of SCN1A	Brain Dev	45(9)	505-11	2023
Yamada M, Tanito K, Suzuki H, Nakato D, Miya F, Takenouchi T, <u>Kosaki K</u>	Cafe-au-lait Spots and Cleft Palate: Not a Chance Association	Cleft Palate Craniofac J	##### #		2023
Tsuji M, Ikeda A, Tsuyusaki Y, Iai M, <u>Kurosawa K</u> , <u>Kosaki K</u> , Goto T	Atypical clinical course in two patients with GNB1 variants who developed acute encephalopathy	Brain Dev	45(8)	462-6	2023
Kawakita M, Iwasaki S, Moteki H, Nishio SY, <u>Kosho T</u> , Usami SI	Otological Features of Patients with Musculocontractural Ehlers-Danlos Syndrome Caused by Pathogenic Variants in CHST14 (mcEDS-CHST14)	Genes (Basel)	14(7)		2023
Futagawa H, Ito S, <u>Kosaki K</u> , <u>Yoshihashi H</u>	Long-term clinical course of Heyn-Sproul-Jackson syndrome	Congenit Anom (Kyoto)	63(5)	174-5	2023
Yogi A, Iemura R, Nakatani H, Takasawa K, Gau M, Yamauchi T, Yoshida M, Moriyama K, Ishii T, Hosokawa S,	BMP2 is a potential causative gene for isolated dextrocardia situs solitus	Eur J Med Genet	66(9)	1048-20	2023

Yamada M, Suzuki H, <u>Kosaki K</u> , Kashimada K, Morio T					
Machida M, Rocos B, <u>Ohashi H</u> , Taira K, Nemoto N, Oikawa N, Kaguchi R, Nakanishi K	RASopathies and spinal deformities for screening of scoliosis	Pediatr Int	65(1)	e155 89	2023
Sugimoto T, Inagaki H, Mariya T, Kawamura R, Taniguchi-Ikeda M, <u>Mizuno S</u> , Muramatsu Y, Tsuge I, <u>Ohashi H</u> , Saito N, Hasegawa Y, Ochi N, Yamaguchi M, Murotsuki J, Kurahashi H	Breakpoints in complex chromosomal rearrangements correspond to transposase-accessible regions of DNA from mature sperm	Hum Genet	142(10)	1451 -60	2023
Iimori H, <u>Nishina S</u> , Hieda O, Goseki T, Nishikawa N, Suzuki S, Hikoya A, Komori M, Suzuki H, Yoshida T, Hayashi S, Mori T, Kimura A, Morimoto T, Shimizu Y, Negishi T, Shimizu T, Yokoyama Y, Sugiyama Y, Azuma N, Sato M	Clinical presentations of acquired comitant esotropia in 5-35 years old Japanese and digital device usage: a multicenter registry data analysis study	Jpn J Ophthalmol	67(6)	629-36	2023
Nishio Y, Kato K, Tran Mau-Them F, Futagawa H, Quelin C, Masuda S, Vitobello A, Otsuji S, Shawki HH, Oishi H, Thauvin-Robinet C, Takenouchi T, <u>Kosaki K</u> , Takahashi Y, Saitoh S	Gain-of-function MYCN causes a megalencephaly-polydactyly syndrome manifesting mirror phenotypes of Feingold syndrome	HGG Adv	4(4)	1002 38	2023
Yamaguchi T, Yamada K, Nagai S, Nishikubo T, Koitabashi N, Minami-Hori M, Matsushima M, Shibata Y, Ishiguro H, Sanai H, Fujikawa T, Takiguchi Y, Matsumoto KI, <u>Koshio T</u>	Clinical and molecular delineation of classical-like Ehlers-Danlos syndrome through a comprehensive next-generation sequencing-based screening system	Front Genet	14	##### #	2023
Fujisawa Y, Masunaga Y, Tanikawa W, Nakashima S, Ueda D, Sano S, Fukami M, Saitsu H, Yazawa T,	Serum steroid metabolite profiling by LC-MS/MS in two phenotypic male patients with HSD17B3 deficiency: Implications for hormonal	J Steroid Biochem Mol Biol	234	1064 03	2023

<u>Ogata T</u>	diagnosis				
Torii K, <u>Nishina S</u> , Morikawa H, Mizobuchi K, Takayama M, Tachibana N, Kurata K, Hikoya A, Sato M, Nakano T, Fukami M, Azuma N, Hayashi T, Saitsu H, Hotta Y	The Structural Abnormalities Are Deeply Involved in the Cause of RPGRIP1-Related Retinal Dystrophy in Japanese Patients	Int J Mol Sci	24(18)		2023
Masuda Y, Nagayasu Y, Murakami H, Nishie R, Morita N, Hashida S, Daimon A, Nunode M, Maruoka H, Yoo M, Sano T, Odanaka Y, Fujiwara S, Fujita D, <u>Okamoto N</u> , Ohmichi M	Triple repeated fetal congenital heart disease linked to PLD1 mutation: a case report	J Med Case Rep	17(1)	411	2023
Pinto EM, Fridman C, Figueiredo BC, Salvador H, Teixeira MR, Pinto C, Pinheiro M, Kratz CP, Lavarino C, Legal E, Le A, Kelly G, Koeppe E, Stoffel EM, Breen K, Hahner S, Heinze B, Techavichit P, Krause A, <u>Ogata T</u> , Fujisawa Y, Walsh MF, Rana HQ, Maxwell KN, Garber JE, Rodriguez-Galindo C, Ribeiro RC, Zambetti GP	Multiple TP53 p.R337H haplotypes and implications for tumor susceptibility	HGG Adv	5(1)	1002 44	2024
Arai H, Noguchi A, Shina K, Otaka S, Takahashi I, <u>Kosaki K</u> , Takahashi T	A child with branchio-oto-renal spectrum disorder carrying an SIX1 variant	Pediatr Int	65(1)	e156 38	2023
Kurosawa R, Iida K, Ajiro M, Awaya T, Yamada M, <u>Kosaki K</u> , Hagiwara M	PDIVAS: Pathogenicity predictor for Deep-Intronic Variants causing Aberrant Splicing	BMC Genomics	24(1)	601	2023
Thanasegaran S, Daimon E, Shibukawa Y, Yamazaki N, <u>Okamoto N</u>	Modelling Takenouchi-Kosaki syndrome using disease-specific iPSCs	Stem Cell Res	73	1032 21	2023

Suzuki Y, Nomura N, Yamada K, Yamada Y, Fukuda A, Hoshino K, Abe S, <u>Kurosawa</u> K, Inaba M, <u>Mizuno</u> S, Wakamatsu N, Hayashi S	Pathogenicity evaluation of variants of uncertain significance at exon-intron junction by splicing assay in patients with Mowat-Wilson syndrome	Eur J Med Genet	66(1 2)	1048 82	2023
Masunaga Y, Ono H, Fujisawa Y, Taniguchi K, Saitsu H, <u>Ogata</u> T	Sotos syndrome with marked overgrowth in three Japanese patients with heterozygous likely pathogenic NSD1 variants: case reports with review of literature	Endocr J	71(1)	75- 81	2024
Matsubara K, Kuki I, Ishioka R, Yamada N, Fukuoka M, Inoue T, Nukui M, <u>Okamoto</u> N, Mizuguchi T, Matsumoto N, Okazaki S	Abnormal axonal development and severe epileptic phenotype in Dynamin-1 (DNM1) encephalopathy	Epileptic Disord	26(1)	139- 43	2024
Sakamoto M, <u>Kurosawa</u> K, Tanoue K, Iwama K, Ishida F, Watanabe Y, <u>Okamoto</u> N, Tsuchida N, Uchiyama Y, Koshimizu E, Fujita A, Misawa K, Miyatake S, Mizuguchi T, Matsumoto N	A heterozygous germline deletion within USP8 causes severe neurodevelopmental delay with multiorgan abnormalities	J Hum Genet	69(2)	85- 90	2024
Urakawa T, Sano S, Kawashima S, Nakamura A, Shima H, Ohta M, Yamada Y, Nishida A, Narusawa H, Ohtsu Y, Matsubara K, Dateki S, Maruo Y, Fukami M, <u>Ogata</u> T, Kagami M	(Epi)genetic and clinical characteristics in 84 patients with pseudohypoparathyroidism type 1B	Eur J Endocrinol	189(6)	590- 600	2023
Oshika T, Endo T, Kurosaka D, Matsuki N, Miyagi M, Mori T, Nagamoto T, Negishi K, <u>Nishina</u> S, Nomura K, Unoki N, Yoshida S	Long-term surgical outcomes of pediatric cataract-multivariate analysis of prognostic factors	Sci Rep	13(1)	2164 5	2023
Yamoto K, Kato F, Yamoto M, Fukumoto K, Shimizu K, Saitsu H, <u>Ogata</u> T	TBX5 pathogenic variant in a patient with congenital heart defect and tracheal stenosis	Congenit Anom (Kyoto)	64(1)	4549 6	2024

Sonoda Y, Fujita A, Torio M, Mukaino T, Sakata A, Matsukura M, Yonemoto K, Hatae K, Ichimiya Y, Chong PF, Ochiai M, Wada Y, Kadoya M, <u>Okamoto N</u> , Murakami Y, Suzuki T, Isobe N, Shigeto H, Matsumoto N, Sakai Y, Ohga S	Progressive myoclonic epilepsy as an expanding phenotype of NGLY1-associated congenital deglycosylation disorder: A case report and review of the literature	Eur J Med Genet	67	1048 95	2024
Tokunaga S, Shimomura H, Taniguchi N, Yanagi K, Kaname T, <u>Okamoto N</u> , Takeshima Y	A novel DLG4 variant causes DLG4-related synaptopathy with intellectual regression	Hum Genome Var	11(1)	1	2024
Higashimoto K, Sun F, Imagawa E, Saida K, Miyake N, Hara S, Yatsuki H, Kubiura-Ichimaru M, Fujita A, Mizuguchi T, Matsumoto N, <u>Soejima H</u>	Whole-exome sequencing reveals causative genetic variants for several overgrowth syndromes in molecularly negative Beckwith-Wiedemann spectrum	J Med Genet			2024
Ichikawa Y, Kuroda H, Ikegawa T, Kawai S, Ono S, Kim KS, Yanagi S, <u>Kurosawa K</u> , Aoki Y, Iwamoto M, Ueda H	Electrocardiographic Changes with Age in Japanese Patients with Noonan Syndrome	J Cardiovasc Dev Dis	11(1)		2023
Udagawa T, Takahashi E, Tatsumi N, Mutai H, Saijo H, Kondo Y, Atkinson PJ, <u>Matsunaga T</u> , Yoshikawa M, Kojima H, Okabe M, Cheng AG	Loss of Pax3 causes reduction of melanocytes in the developing mouse cochlea	Sci Rep	14(1)	2210	2024
Takahashi I, Noguchi A, Kondo D, Sato Y, Suzuki H, Yamada M, <u>Kosaki K</u> , Takahashi T	A novel missense variant of FGD1 disrupts critical cysteine residues of the FYVE domain in Japanese siblings with Aarskog-Scott syndrome	Clin Pediatr Endocrinol	33(1)	39-42	2024
Watanabe D, Nakato D, Yamada M, Suzuki H, Takenouchi T, Miya F, <u>Kosaki K</u>	SALL4 deletion and kidney and cardiac defects associated with VACTERL association	Pediatr Nephrol			2024
Nakamura- <u>Utsunomiya A</u> , Yamaguchi K, Goshima N	Anti-ZSCAN1 Autoantibodies Are a Feasible Diagnostic Marker for ROHHAD Syndrome Not	Int J Mol Sci	25(3)		2024

	Associated with a Tumor				
Nishi E, Yanagi K, Kaname T, <u>Okamoto</u> <u>N</u>	Clinical details of individuals with Rauch-Steindl syndrome due to NSD2 truncating variants	Mol Genet Genomic Med	12(2)	e239 6	2024
Machida O, Sakamoto H, Yamamoto KS, Hasegawa Y, Nii S, Okada H, Nishikawa K, Sumimoto SI, Nishi E, <u>Okamoto</u> <u>N</u> , Yamamoto T	Haploinsufficiency of NKX2-1 is likely to contribute to developmental delay involving 14q13 microdeletions	Intractable Rare Dis Res	13(1)	36-41	2024
Nishino M, Tanaka M, Imagawa K, Yaita K, Enokizono T, Ohto T, Suzuki H, Yamada M, Takenouchi T, <u>Kosaki</u> <u>K</u> , Takada H	Identification of a novel splice-site WWOX variant with paternal uniparental isodisomy in a patient with infantile epileptic encephalopathy	Am J Med Genet A			2024
Tocan V, Nakamura- <u>Utsunomiya</u> <u>A</u> , Sonoda Y, Matsuoka W, Mizuguchi S, Muto Y, Hijioka T, Nogami M, Sasaoka D, Nagamatsu F, Oba U, Kawakubo N, Hamada H, Mushimoto Y, Chong PF, Kaku N, Koga Y, Sakai Y, Oda Y, Tajiri T, Ohga S	High-Titer Anti-ZSCAN1 Antibodies in a Toddler Clinically Diagnosed with Apparent Rapid-Onset Obesity with Hypothalamic Dysfunction, Hypoventilation, and Autonomic Dysregulation Syndrome	Int J Mol Sci	25(5)		2024
Yamada M, Maeta K, Suzuki H, Kurosawa R, Takenouchi T, Awaya T, Ajiro M, Takeuchi A, Nishio H, Hagiwara M, Miya F, Matsuo M, <u>Kosaki</u> <u>K</u>	Successful skipping of abnormal pseudoexon by antisense oligonucleotides in vitro for a patient with beta-propeller protein-associated neurodegeneration	Sci Rep	14(1)	6506	2024
Tada H, Kawashiri MA, Nohara A, Sekiya T, <u>Watanabe</u> <u>A</u> , Takamura M	Genetic Counseling and Genetic Testing for Familial Hypercholesterolemia	Genes (Basel)	15(3)		2024
Kimizu T, Nozaki M, Okada Y, Sawada A, Morisaki M, Fujita H, Irie A, Matsuda K, Hasegawa Y, Nishi E, <u>Okamoto</u> <u>N</u> , Kawai M, Imai K, Suzuki Y, Wada K, Mitsuda N, Ida S	Multiplex Real-Time PCR-Based Newborn Screening for Severe Primary Immunodeficiency and Spinal Muscular Atrophy in Osaka, Japan: Our Results after 3 Years	Genes (Basel)	15(3)		2024

Hashimoto K, Miwa T, Ono C, Nara K, Mutai H, Seto T, Sakamoto H, <u>Matsunaga T</u>	Gap Junction Beta-2 p.Val84Met Can Cause Autosomal Dominant Syndromic Hearing Loss With Keratoderma	Cureus	16(2)	e549 92	2024
Yamada M, <u>Mizuno S</u> , Inaba M, Uehara T, Inagaki H, Suzuki H, Miya F, Takenouchi T, Kurahashi H, <u>Kosaki K</u>	Truncating variants of the sterol recognition region of SHH cause hypertelorism phenotype rather than hypotelorism-holoprosencephaly	Am J Med Genet A		e636 14	2024
Shoji Y, Hata A, Maeyama T, Wada T, Hasegawa Y, Nishi E, Ida S, Etani Y, Niihori T, <u>Aoki Y, Okamoto N</u> , Kawai M	Genetic backgrounds and genotype-phenotype relationships in anthropometric parameters of 116 Japanese individuals with Noonan syndrome	Clin Pediatr Endocrinol	33(2)	1847 6	2024
Saito S, Saito Y, Sato S, Aoki S, Fujita H, Ito Y, Ono N, Funakoshi T, Kawai T, Suzuki H, Sasaki T, Tanaka T, Inoie M, Hata K, Kataoka K, <u>Kosaki K</u> , Amagai M, Nakabayashi K, Kubo A	Gene-specific somatic epigenetic mosaicism of FDFT1 underlies a non-hereditary localized form of porokeratosis	Am J Hum Genet	111(5)	896-912	2024
黒澤 健司	【小児科学レビュー-最新主要文献とガイドライン-】先天異常症候群	小児科臨床	76(2)	193-6	2023
岡崎 哲也, 足立 香織, 難波 栄二	【重篤な遺伝性疾患の着床前診断-患者ニーズと診断・治療の現状】疾患各論 脆弱X症候群(fragile X syndrome)	臨床婦人科産科	78(2)	223-7	2024