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Kirino S, Suzuki M, Ogawa T, Takasawa K, Adachi E, Gau M, Takahashi K, Ikeno M, Yamada M, Suzuki H, Kosaki K, Moriyama K, Yoshida M, Morio T, Kashimada K	Clinical report: Chronic liver dysfunction in an individual with an AMOTL1 variant	Eur J Med Genet	65(11)	104623	2022
Maruwaka K, Nakajima Y, Yamada T, Tanaka T, Kosaki R, Inagaki H, Kosaki K, Kurahashi H	Two Japanese patients with Noonan syndrome-like disorder with loose anagen hair 2	Am J Med Genet A	188(7)	2246-50	2022

Matsukawa Y, Sakamoto K, Ikeda Y, Taga T, Kosaki K, Maruo Y	Familial hemophagocytic lymphohistiocytosis syndrome due to lysinuric protein intolerance: a patient with a novel compound heterozygous pathogenic variant in SLC7A7	Int J Hematol	116(4)	635-8	2022
Moriyama H, Endo J, Kataoka M, Shimanaka Y, Kono N, Sugiura Y, Goto S, Kitakata H, Hiraide T, Yoshida N, Isobe S, Yamamoto T, Shirakawa K, Anzai A, Katsumata Y, Suematsu M, Kosaki K, Fukuda K, Arai H, Sano M	Omega-3 fatty acid epoxides produced by PAF-AH2 in mast cells regulate pulmonary vascular remodeling	Nat Commun	13(1)	3013	2022
Murofushi Y, Hayakawa I, Abe Y, Ohto T, Murayama K, Suzuki H, Takenouchi T, Kosaki K, Kubota M	Ketogenic Diet for KARS-Related Mitochondrial Dysfunction and Progressive Leukodystrophy	Neuropediatrics	53(1)	23955	2022
Nakajima K, Suzuki H, Yamamoto M, Yamamoto T, Kawai T, Nakabayashi K, Hata K, Kosaki K, Nakajima H, Sano S, Kubo A	A familial case of periodontal Ehlers-Danlos syndrome lacking skin extensibility and joint hypermobility with a missense mutation in C1R	J Dermatol	49(7)	714-8	2022
Nishi E, Takenouchi T, Miya F, Uehara T, Yanagi K, Hasegawa Y, Ueda K, Mizuno S, Kaname T, Kosaki K, Okamoto N	The novel and recurrent variants in exon 31 of CREBBP in Japanese patients with Menke-Hennekam syndrome	Am J Med Genet A	188(2)	446-53	2022
Ogawa E, Sakaguchi Y, Enokizono M, Yoshihashi H, Yamada M, Suzuki H, Kosaki K, Miyama S, Takenouchi T	Vanishing basal ganglia in ATP1A3-related polymicrogyria	Am J Med Genet A	188(2)	665-7	2022
Ogura Y, Uehara T, Ujibe K, Yoshihashi H, Yamada M, Suzuki H, Takenouchi T, Kosaki K, Hirata H	The p.Thr395Met missense variant of NFIA found in a patient with intellectual disability is a defective variant	Am J Med Genet A	188(4)	1184-92	2022
Ohkawa T, Nishimura A, Kosaki K, Aoki-Nogami Y, Tomizawa D, Kashimada K, Morio T, Kato M, Mizutani S, Takagi M	PAX3/7-FOXO1 fusion-negative alveolar rhabdomyosarcoma in Schuurs-Hoeijmakers syndrome	J Hum Genet	67(1)	18719	2022
Okamoto N, Miya F, Tsunoda T, Kanemura Y, Saitoh S, Kato M, Yanagi K, Kaname T, Kosaki K	Four pedigrees with aminoacyl-tRNA synthetase abnormalities	Neurol Sci	43(4)	2765-74	2022
Saito S, Ono N, Sasaki T, Aoki S, Kosaki K, Kuze B, Nakabayashi K, Amagai M, Kubo A	Neurofibromatosis type 2 with mild Pierre-Robin sequence showing a heterozygous chromosome 22q12 microdeletion	J Hum Genet	67(11)	675-8	2022

	encompassing NF2 and MN1				
Shiraishi Y, Okada A, Chiba K, Kawachi A, Omori I, Mateos RN, Iida N, Yamauchi H, Kosaki K, Yoshimi A	Systematic identification of intron retention associated variants from massive publicly available transcriptome sequencing data	Nat Commun	13(1)	5357	2022
Suzuki H, Aoki K, Kurosawa K, Imagawa K, Ohto T, Yamada M, Takenouchi T, Kosaki K, Ishitani T	De novo non-synonymous CTR9 variants are associated with motor delay and macrocephaly: human genetic and zebrafish experimental evidence	Hum Mol Genet	31(22)	3846-54	2022
Suzuki H, Li S, Tokutomi T, Takeuchi C, Takahashi M, Yamada M, Okuno H, Miya F, Takenouchi T, Numabe H, Kosaki K, Ohshima T	De novo non-synonymous DPYSL2 (CRMP2) variants in two patients with intellectual disabilities and documentation of functional relevance through zebrafish rescue and cellular transfection experiments	Hum Mol Genet	31(24)	4173-82	2022
Suzuki H, Nozaki M, Yoshihashi H, Imagawa K, Kajikawa D, Yamada M, Yamaguchi Y, Morisada N, Eguchi M, Ohashi S, Ninomiya S, Seto T, Tokutomi T, Hida M, Toyoshima K, Kondo M, Inui A, Kurosawa K, Kosaki R, Ito Y, Okamoto N, Kosaki K, Takenouchi T	Genome Analysis in Sick Neonates and Infants: High-yield Phenotypes and Contribution of Small Copy Number Variations	J Pediatr	244	38-48 e1	2022
Suzuki N, Takai Y, Yonemura M, Negoro H, Motonaga S, Fujishiro N, Nakamura E, Takae S, Yoshida S, Uesugi K, Ohira T, Katsura A, Fujiwara M, Horiguchi I, Kosaki K, Onodera H, Nishiyama H	Guidance on the need for contraception related to use of pharmaceuticals: the Japan Agency for Medical Research and Development Study Group for providing information on the proper use of pharmaceuticals in patients with reproductive potential	Int J Clin Oncol	27(5)	829-39	2022
Takahashi Y, Date H, Oi H, Adachi T, Imanishi N, Kimura E, Takizawa H, Kosugi S, Matsumoto N, Kosaki K, Matsubara Y, Consortium I, Mizusawa H	Six years' accomplishment of the Initiative on Rare and Undiagnosed Diseases: nationwide project in Japan to discover causes, mechanisms, and cures	J Hum Genet	67(9)	505-13	2022
Tsuchida Y, Nagafuchi Y, Uehara T, Suzuki H, Yamada M, Kono M, Hatano H, Shoda H, Fujio K, Kosaki K	Rheumatoid arthritis in a patient with compound heterozygous variants in the COL11A2 gene and	Medicine (Baltimore)	101(7)	e28828	2022

	progressive hearing loss: A case report				
Uwamino Y, Yokoyama T, Shimura T, Nishimura T, Sato Y, Wakui M, Kosaki K, Hasegawa N, Murata M	The effect of the E484K mutation of SARS-CoV-2 on the neutralizing activity of antibodies from BNT162b2 vaccinated individuals	Vaccine	40(13)	1928-31	2022
Yamada M, Arimitsu T, Suzuki H, Miwa T, Kosaki K	Early diagnosis of lateral meningocele syndrome in an infant without neurological symptoms based on genomic analysis	Childs Nerv Syst	38(3)	659-63	2022
Yamada M, Suzuki H, Adachi H, Noguchi A, Miya F, Takahashi T, Kosaki K	Diagnosis of SLC25A46-related pontocerebellar hypoplasia in two siblings with fulminant neonatal course: role of postmortem CT and whole genomic analysis: a case report	BMC Neurol	22(1)	20	2022
Yamada M, Suzuki H, Futagawa H, Takenouchi T, Miya F, Yoshihashi H, Kosaki K	Phenotypic overlap between cardioacrofacial dysplasia-2 and oral-facial-digital syndrome	Eur J Med Genet	65(6)	104512	2022
Yamada M, Suzuki H, Miya F, Takenouchi T, Kosaki K	Deciphering complex rearrangements at the breakpoint of an apparently balanced reciprocal translocation t(4:18)(q31;q11.2)dn and at a cryptic deletion: Further evidence of TLL1 as a causative gene for atrial septal defect	Am J Med Genet A	188(8)	209134	2022
Yamada M, Suzuki H, Shima T, Uehara T, Kosaki K	A patient with compound heterozygosity of SMPD4: Another example of utility of exome-based copy number analysis in autosomal recessive disorders	Am J Med Genet A	188(2)	613-7	2022
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, Kosaki K, Saito MK, Takita J, Yoshida T	Involvement of mTOR pathway in neurodegeneration in NSF-related developmental and epileptic encephalopathy	Hum Mol Genet	32(10)	1683-97	2023
Kuroda Y, Kumaki T, Saito Y, Enomoto Y, Suzuki H, Takenouchi T, Kosaki K, Kurosawa K	A novel variant of ARPC4-related neurodevelopmental	Am J Med Genet A	191(3)	893-5	2023

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Nitta Y, Kawai H, Maki R, Osaka J, Hakeda-Suzuki S, Nagai Y, Doubkova K, Uehara T, Watanabe K, Kosaki K, Suzuki T, Tavosanis G, Sugie A	Direct evaluation of neuroaxonal degeneration with the causative genes of neurodegenerative diseases in Drosophila using the automated axon quantification system, MeDUsA	Hum Mol Genet	32(9)	1524-38	2023
Uwamino Y, Yokoyama T, Sato Y, Shibata A, Kurafuji T, Tanabe A, Noguchi M, Arai T, Ohno A, Yokota H, Namkoong H, Nishimura T, Kosaki K, 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
Yamada M, Okuno H, Okamoto N, Suzuki H, Miya F, Takenouchi T, Kosaki K	Diagnosis of Prader-Willi syndrome and Angelman syndrome by targeted nanopore long-read sequencing	Eur J Med Genet	66(2)	104690	2023
Yoshihama K, Mutai H, Sekimizu M, Ito F, Saito S, Nakamura S, Mikoshiba T, Nagai R, Takebayashi A, Miya F, Kosaki K, Ozawa H, Matsunaga T	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
Horikawa R, Ogata T, Matsubara Y, Yokoya S, Ogawa Y, Nishijima K, Endo T, Ozono K	Long-term efficacy and safety of two doses of Norditropin((R)) (somatropin) in Noonan syndrome: a 4-year randomized, double-blind, multicenter trial in Japanese patients	Endocr J	67(8)	803-18	2020
Narumi-Kishimoto Y, Ozawa H, Yanagi K, Kawai T, Okamura K, Hata K, Kaname T, Matsubara Y	A novel EFTUD2 mutation identified an adult male with mandibulofacial dysostosis Guion-Almeida type	Clin Dysmorphol	29(4)	186-8	2020
Yanagi K, Morimoto N, Iso M, Abe Y, Okamura K, Nakamura T, Matsubara Y, Kaname T	A novel missense variant of the GNAI3 gene and recognisable morphological characteristics of the mandibula in ARCND1	J Hum Genet	66(10)	1029-34	2021
Kanno M, Suzuki M, Tanikawa K, Numakura C, Matsuzawa SI, Niihori T, Aoki Y, Matsubara Y, Makino S, Tamiya G, Nakano S, Funayama R, Shirota M, Nakayama K, Mitsui T, Hayasaka K	Heterozygous calcyclin-binding protein/Siah1-interacting protein (CACYBP/SIP) gene pathogenic variant linked to a dominant family with	J Hum Genet	67(7)	393-7	2022

	paucity of interlobular bile duct				
Takeuchi I, Yanagi K, Takada S, Uchiyama T, Igarashi A, Motomura K, Hayashi Y, Nagano N, Matsuoka R, Sugiyama H, Yoshioka T, Saito H, Kawai T, Miyaji Y, Inuzuka Y, Matsubara Y, Ohya Y, Shimizu T, Matsumoto K, Arai K, Nomura I, Kaname T, Morita H	STAT6 gain-of-function variant exacerbates multiple allergic symptoms	J Allergy Clin Immunol	151(5)	1402-9 e6	2023
Yanagi K, Coker J, Miyana K, Aso S, Kobayashi N, Satou K, Richman A, Indupuru S, Matsubara Y, Kaname T	Biallelic CC2D2A variants, SNV and LINE-1 insertion simultaneously identified in siblings using long-read whole-genome sequencing and haplotype phasing	J Hum Genet			2023
Mori R, Matsumoto H, Muro S, Morisaki H, Otsuki R	Loeys-Dietz Syndrome Presenting with Giant Bullae and Asthma	J Allergy Clin Immunol Pract	8(6)	2058-9	2020
Seike Y, Minatoya K, Matsuda H, Ishibashi-Ueda H, Morisaki H, Morisaki T, Kobayashi J	Histologic differences between the ascending and descending aortas in young adults with fibrillin-1 mutations	J Thorac Cardiovasc Surg	159(4)	1214-20 e1	2020
森崎　裕子	【遺伝情報と遺伝カウンセリング】小児科領域別のポイント　循環器疾患の遺伝学的検査	小児内科	52(8)	1071-4	2020
森崎　裕子	【診断・治療可能な遺伝性疾患を見逃さないために】循環器疾患　マルファン症候群	小児科臨床	73(5)	757-62	2020
森崎　裕子	難治性疾患(難病)を学ぶ　マルファン症候群	遺伝子医学	10(2)	109-14	2020
Kitayama K, Ishiguro T, Komiyama M, Morisaki T, Morisaki H, Minase G, Hamanaka K, Miyatake S, Matsumoto N, Kato M, Takahashi T, Yorifuji T	Mutational and clinical spectrum of Japanese patients with hereditary hemorrhagic telangiectasia	BMC Med Genomics	14(1)	288	2021
Seike Y, Matsuda H, Ishibashi-Ueda H, Morisaki H, Morisaki T, Minatoya K, Ogino H	Surgical Outcome and Histological Differences between Individuals with TGFBR1 and TGFBR2 Mutations in Loeys-Dietz Syndrome	Ann Thorac Cardiovasc Surg	27(1)	56-63	2021
森崎　裕子	【小児疾患診療のための病態生理2　改訂第6版】染色体異常、先天異常　Ehlers-Danlos 症候群、Marfan 症候群	小児内科	53(増刊)	304-8	2021
森崎　裕子	【先天代謝異常症】結	糖尿病・	53(4)	409-15	2021

	合組織異常症	内分泌代謝科			
森崎　裕子	【小児遺伝子疾患事典】循環器疾患 TGFBR1(関連疾患:Loeys-Dietz 症候群関連遺伝子:TGFBR2、SMAD3)	小児科診療	84(11)	1623-6	2021
森崎　裕子	いま知っておきたい最新の臨床検査　身近な疾患を先端技術で診断(Vol.15)　循環器関連疾患の遺伝学的検査	医学のあゆみ	278(9)	788-94	2021
森崎　裕子	臨床遺伝学・人類遺伝学誌上講義　遺伝的多様性と多因子疾患	遺伝子医学	11(1)	128-33	2021
森崎　隆幸, 森崎　裕子	各種難病の最新治療情報　マルファン症候群の最新情報	難病と在宅ケア	26(10)	55-8	2021
Imamura T, Omura T, Sasaki N, Arino S, Nohara H, Saito A, Ichinose M, Yamaguchi K, Kojima N, Inagawa H, Takahashi K, Unno T, Morisaki H, Ishikawa O, Yoshikawa G, Okada Y	Case Report: Spontaneous Postpartum Quadruple Cervicocephalic Arterial Dissection With a Heterozygous COL5A1 Variant of Unknown Significance	Front Neurol	13	928803	2022
Minatogawa M, Unzaki A, Morisaki H, Syx D, Sonoda T, Janecke AR, Slavotinek A, Voermans NC, Lacassie Y, Mendoza-Londono R, Wierenga KJ, Jayakar P, Gahl WA, Tifft CJ, Figuera LE, Hilhorst-Hofstee Y, Maugeri A, Ishikawa K, Kobayashi T, Aoki Y, Ohura T, Kawame H, Kono M, Mochida K, Tokorodani C, Kikkawa K, Morisaki T, Kobayashi T, Nakane T, Kubo A, Ranells JD, Migita O, Sobey G, Kaur A, Ishikawa M, Yamaguchi T, Matsumoto N, Malfait F, Miyake N, Kosho T	Clinical and molecular features of 66 patients with musculocontractural Ehlers-Danlos syndrome caused by pathogenic variants in CHST14 (mcEDS-CHST14)	J Med Genet	59(9)	865-77	2022
Muroi A, Shiono J, Ihara S, Morisaki H, Nakai Y	Nonsurgical treatment of cerebral ischemia associated with ACTA2 cerebral arteriopathy: a case report and literature review	Childs Nerv Syst	38(6)	1209-12	2022
Seike Y, Matsuda H, Inoue Y, Sasaki H, Morisaki H, Morisaki T, Kobayashi J	The differences in surgical long-term outcomes between Marfan syndrome and Loeys-Dietz syndrome	J Thorac Cardiovasc Surg	164(1)	16-25 e2	2022
Seike Y, Yokawa K, Koizumi S, Masada K,	Long-term durability of	Eur J	61(6)	1318-	2022

Inoue Y, Morisaki H, Morisaki T, Sasaki H, Matsuda H	a reimplantation valve-sparing aortic root replacement can be expected in both Marfan syndrome and Loeys-Dietz syndrome	Cardiothorac Surg		25	
Yokota T, Koiwa H, Matsushima S, Tsujinaga S, Naya M, Morisaki H, Morisaki T	Loeys-Dietz Cardiomyopathy? Long-term Follow-up After Onset of Acute Decompensated Heart Failure	Can J Cardiol	38(3)	389-91	2022
森崎　裕子	【心疾患のプレコンセプションケア】Marfan 症候群類縁疾患のプレコンセプションケア	心臓	54(12)	1325-9	2022
森崎　裕子	遺伝性大動脈疾患遺伝学的検査による早期診断と管理	脈管学	62(10)	105-10	2022
森崎　裕子	【知っておくべき周産期・新生児領域の遺伝学的検査を展望する】結合織疾患 Marfan 症候群、Loeys-Dietz 症候群、Ehlers-Danlos 症候群	周産期医学	52(5)	734-7	2022
Greene D, Genomics England Research C, Pirri D, Frudd K, Sackey E, Al-Owain M, Giese APJ, Ramzan K, Riaz S, Yamanaka I, Boeckx N, Thys C, Gelb BD, Brennan P, Hartill V, Harvengt J, Kosho T, Mansour S, Masuno M, Ohata T, Stewart H, Taibah K, Turner CLS, Imtiaz F, Riazuddin S, Morisaki T, Ostergaard P, Loeys BL, Morisaki H, Ahmed ZM, Birdsey GM, Freson K, Mumford A, Turro E	Genetic association analysis of 77,539 genomes reveals rare disease etiologies	Nat Med	29(3)	679-88	2023
Nakao S, Nishina S, Tanaka S, Yoshida T, Yokoi T, Azuma N	Early laser photocoagulation for extensive retinal avascularity in infants with incontinentia pigmenti	Jpn J Ophthalmol	64(6)	613-20	2020
仁科 幸子	【遺伝情報と遺伝カウンセリング】小児科領域別のポイント　眼疾患	小児内科	52(8)	1095-9	2020
Haque MN, Ohtsubo M, Nishina S, Nakao S, Yoshida K, Hosono K, Kurata K, Ohishi K, Fukami M, Sato M, Hotta Y, Azuma N, Minoshima S	Analysis of IKBKG/NEMO gene in five Japanese cases of incontinentia pigmenti with retinopathy: fine genomic assay of a rare	J Hum Genet	66(2)	205-14	2021

	male case with mosaicism				
Tanaka S, Yokoi T, Katagiri S, Yoshida-Uemura T, Nishina S, Azuma N	Structure of the Retinal Margin and Presumed Mechanism of Retinal Detachment in Choroidal Coloboma	Ophthalmol Retina	5(7)	702-10	2021
松岡　真未, 仁科　幸子, 三井田　千春, 他	6 ヵ月以下の乳児に対する Spot Vision Screener の使用経験	眼科臨床紀要	15(1)	Jun-42	2022
Morikawa H, Nishina S, 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
Fujioka M, Akiyama T, Hosoya M, Kikuchi K, Fujiki Y, Saito Y, Yoshihama K, Ozawa H, Tsukada K, Nishio SY, Usami SI, Matsunaga T, Hasegawa T, Sato Y, Ogawa K	A phase I/IIa double blind single institute trial of low dose sirolimus for Pendred syndrome/DFNB4	Medicine (Baltimore)	99(19)	e19763	2020
Fujioka M, Hosoya M, Nara K, Morimoto N, Sakamoto H, Otsu M, Nakano A, Arimoto Y, Masuda S, Sugiuchi T, Masuda S, Morita N, Ogawa K, Kaga K, Matsunaga T	Differences in hearing levels between siblings with hearing loss caused by GJB2 mutations	Auris Nasus Larynx	47(6)	938-42	2020
Mutai H, Wasano K, Momozawa Y, Kamatani Y, Miya F, Masuda S, Morimoto N, Nara K, Takahashi S, Tsunoda T, Homma K, Kubo M, Matsunaga T	Variants encoding a restricted carboxy-terminal domain of SLC12A2 cause hereditary hearing loss in humans	PLoS Genet	16(4)	e1008643	2020
Yamamoto N, Mutai H, Namba K, Goto F, Ogawa K, Matsunaga T	Clinical Profiles of DFNA11 at Diverse Stages of Development and Aging in a Large Family Identified by Linkage Analysis	Otol Neurotol	41(6)	e663-e73	2020
Hosoya M, Fujioka M, Nara K, Morimoto N, Masuda S, Sugiuchi T, Katsunuma S, Takagi A, Morita N, Ogawa K, Kaga K, Matsunaga T	Investigation of the hearing levels of siblings affected by a single GJB2 variant: Possibility of genetic modifiers	Int J Pediatr Otorhinolaryngol	149	110840	2021
Isobe A, Maeda N, Fujita H, Banno S, Kageyama T, Hatabu N, Sato R, Suzuki E, Miharu M, Komiyama O, Nakashima M, Matsunaga T, Nishimura G, Yamazawa K	Metacarpophalangeal pattern profile analysis for a 3-month-old infant with Feingold syndrome 2	Am J Med Genet A	185(3)	952-4	2021
Matsunaga T	Clinical genetics, practice, and research of deafblindness: From uncollected experiences to the national registry in Japan	Auris Nasus Larynx	48(2)	185-93	2021
Yamazawa K, Inoue T, Sakemi Y, Nakashima T, Yamashita H, Khono K, Fujita H, Enomoto K, Nakabayashi K, Hata K,	Loss of imprinting of the human-specific imprinted gene ZNF597 causes	J Med Genet	58(6)	427-32	2021

Nakashima M, Matsunaga T, Nakamura A, Matsubara K, Ogata T, Kagami M	prenatal growth retardation and dysmorphic features: implications for phenotypic overlap with Silver-Russell syndrome				
Yamazawa K, Shimizu K, Ohashi H, Haruna H, Inoue S, Murakami H, Matsunaga T, Iwata T, Tsunoda K, Fujinami K	A Japanese boy with double diagnoses of 2p15p16.1 microdeletion syndrome and RP2-associated retinal disorder	Hum Genome Var	8(1)	46	2021
Masuda M, Kanno A, Nara K, Mutai H, Morisada N, Iijima K, Morimoto N, Nakano A, Sugiuchi T, Okamoto Y, Masuda S, Katsunuma S, Ogawa K, Matsunaga T	Phenotype-genotype correlation in patients with typical and atypical branchio-oto-renal syndrome	Sci Rep	12(1)	969	2022
Mutai H, Momozawa Y, Kamatani Y, Nakano A, Sakamoto H, Takiguchi T, Nara K, Kubo M, Matsunaga T	Whole exome analysis of patients in Japan with hearing loss reveals high heterogeneity among responsible and novel candidate genes	Orphanet J Rare Dis	17(1)	114	2022
Nakano A, Arimoto Y, Mutai H, Nara K, Inoue S, Matsunaga T	Clinical and genetic analysis of children with hearing loss and bilateral enlarged vestibular aqueducts	Int J Pediatr Otorhinolaryngol	152	110975	2022
Saettini F, Herriot R, Prada E, Nizon M, Zama D, Marzollo A, Romaniouk I, Lougaris V, Cortesi M, Morreale A, Kosaki R, Cardinale F, Ricci S, Dominguez-Garrido E, Montin D, Vincent M, Milani D, Biondi A, Gervasini C, Badolato R	Prevalence of Immunological Defects in a Cohort of 97 Rubinstein-Taybi Syndrome Patients	J Clin Immunol	40(6)	851-60	2020
Kawashima S, Hattori A, Suzuki E, Matsubara K, Toki M, Kosaki R, Hasegawa Y, Nakabayashi K, Fukami M, Kagami M	Methylation status of genes escaping from X-chromosome inactivation in patients with X-chromosome rearrangements	Clin Epigenetics	13(1)	134	2021
Ohashi E, Hayakawa I, Murofushi Y, Kawai M, Suzuki-Muromoto S, Abe Y, Yoshida M, Kono N, Kosaki R, Hoshino A, Mizuguchi M, Kubota M	Recurrent acute necrotizing encephalopathy in a boy with RANBP2 mutation and thermolabile CPT2 variant: The first case of ANE1 in Japan	Brain Dev	43(8)	873-8	2021
Hatakeyama S, Goto M, Yamamoto A, Ogura J, Watanabe N, Tsutsumi S, Yakuwa N, Yamane R, Nagase S, Takahashi K, Kosaki R, Murashima A, Yamaguchi H	The safety of pranlukast and montelukast during the first trimester of pregnancy: A prospective, two-centered cohort study in Japan	Congenit Anom (Kyoto)	62(4)	161-8	2022
Higashiyama H, Ohsone Y, Takatani R, Futatani T, Kosaki R, Kagami M	Two infants with mild, atypical clinical features	Eur J Med Genet	65(10)	104580	2022

	of Kagami-Ogata syndrome caused by epimutation				
Saito T, Okamura K, Kosaki R, Wakamatsu K, Ito S, Nakajima O, Yamashita H, Hozumi Y, Suzuki T	Impact of a SLC24A5 variant on the retinal pigment epithelium of a Japanese patient with oculocutaneous albinism type 6	Pigment Cell Melanoma Res	35(2)	212-9	2022
Shiohama T, Fujii K, Kosaki R, Watanabe Y, Uchida T, Hagiwara S, Kinoshita K, Sugita K, Aoki Y, Shimojo N	Severe neuroglycopenic symptoms due to nonketotic hypoglycemia in children with cardio-facio-cutaneous syndrome	Am J Med Genet A	188(12)	3505-9	2022
Urabe R, Abe Y, Kosaki R, Koshimizu E, Miyatake S, Matsumoto N, Kato M, Kubota M	A case of epilepsy of infancy with migrating focal seizures caused by mosaic SCN2A mutation	Epilepsy & Seizure	14(1)	17-24	2022
Fukui K, Amari S, Yotani N, Kosaki R, 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
Murofushi Y, Hayakawa I, Kawai M, Abe Y, Kosaki R, 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
Yaga T, Iguchi A, Nakayama R, Kosaki R, Ishiguro A	Potocki-Shaffer syndrome revealed in a WAGR syndrome case with multiple exostoses	Pediatr Int	65(1)	e15405	2023
Yamamoto M, Takashio S, Nakashima N, Hanatani S, Arima Y, Sakamoto K, Yamamoto E, Kaikita K, Aoki Y, Tsujita K	Double-chambered right ventricle complicated by hypertrophic obstructive cardiomyopathy diagnosed as Noonan syndrome	ESC Heart Fail	7(2)	721-6	2020
Ando Y, Sawada M, Kawakami T, Morita M, Aoki Y	A Patient with Noonan Syndrome with a KRAS Mutation Who Presented Severe Nerve Root Hypertrophy	Case Rep Neurol	13(1)	108-18	2021
Gualtieri A, Kyprianou N, Gregory LC, Vignola ML, Nicholson JG, Tan R, Inoue SI, Scagliotti V, Casado P, Blackburn J, Abollo-Jimenez F, Marinelli E, Besser REJ, Hogler W, Karen Temple I, Davies JH, Gagunashvili A, Robinson I, Camper SA, Davis SW, Cutillas PR, Gevers EF, Aoki Y, Dattani MT, Gaston-Massuet C	Activating mutations in BRAF disrupt the hypothalamo-pituitary axis leading to hypopituitarism in mice and humans	Nat Commun	12(1)	2028	2021
藤井 隆, 須藤 陽介, 佐々木 綾子, 永井 康貴, 青木 洋子, 三井 哲夫	HRAS G12V 変異による最重症の Costello 症候群	日本小児科学会雑誌	125(3)	461-6	2021

Leoni C, Viscogliosi G, Tartaglia M, Aoki Y, Zampino G	Multidisciplinary Management of Costello Syndrome: Current Perspectives	J Multidiscip Healthc	15	1277-96	2022
Nagai K, Niihori T, Okamoto N, Kondo A, Suga K, Ohhira T, Hayabuchi Y, Homma Y, Nakagawa R, Ifuku T, Abe T, Mizuguchi T, Matsumoto N, Aoki Y	Duplications in the G3 domain or switch II region in HRAS identified in patients with Costello syndrome	Hum Mutat	43(1)	3 月 15 日	2022
Igarashi N, Miyata K, Loo TM, Chiba M, Hanyu A, Nishio M, Kawasaki H, Zheng H, Toyokuni S, Kon S, Moriyama K, Fujita Y, Takahashi A	Hepatocyte growth factor derived from senescent cells attenuates cell competition-induced apical elimination of oncogenic cells	Nat Commun	13(1)	4157	2022
Inagaki Y, Ogawa T, Tabata MJ, Nagata Y, Watanabe R, Kawamoto T, Moriyama K, Tanaka T	Identification of OPN3 as associated with non-syndromic oligodontia in a Japanese population	J Hum Genet	66(8)	769-75	2021
Min Soe K, Ogawa T, Moriyama K	Molecular mechanism of hyperactive tooth root formation in oculo-facio-cardio-dental syndrome	Front Physiol	13	946282	2022
Min Swe NM, Kobayashi Y, Kamimoto H, Moriyama K	Aberrantly activated Wnt/beta-catenin pathway co-receptors LRP5 and LRP6 regulate osteoblast differentiation in the developing coronal sutures of an Apert syndrome (Fgfr2(S252W)(/+)) mouse model	Dev Dyn	250(3)	465-76	2021
Ogawa T, Cheng ES, Muramoto K, Moriyama K	Long-Term Management and Maxillofacial Growth in a Klippel-Trenaunay Syndrome Patient	Cleft Palate Craniofac J	57(6)	782-90	2020
Ogura K, Kobayashi Y, Hikita R, Tsuji M, Moriyama K	Three-dimensional analysis of the palatal morphology in growing patients with Apert syndrome and Crouzon syndrome	Congenit Anom (Kyoto)	62(4)	153-60	2022
Sagawa Y, Ogawa T, Matsuyama Y, Nakagawa Kang J, Yoshizawa Araki M, Unnai Yasuda Y, Tumurkhuu T, Ganburged G, Bazar A, Tanaka T, Fujiwara T, Moriyama K	Association between Smoking during Pregnancy and Short Root Anomaly in Offspring	Int J Environ Res Public Health	18(21)		2021
Shih-Wei Cheng E, Tsuji M, Suzuki S, Moriyama K	An overview of the intraoral features and craniofacial morphology of growing and adult Japanese cleidocranial dysplasia subjects	Eur J Orthod	44(6)	711-22	2022

Takada K, Chiba T, Miyazaki T, Yagasaki L, Nakamichi R, Iwata T, Moriyama K, Harada H, Asahara H	Single Cell RNA Sequencing Reveals Critical Functions of Mkx in Periodontal Ligament Homeostasis	Front Cell Dev Biol	10	795441	2022
Thiha P, Higashihori N, Kano S, Moriyama K	Histone methyltransferase SET domain bifurcated 1 negatively regulates parathyroid hormone/parathyroid hormone-related peptide receptor to control chondrocyte proliferation in Meckel's cartilage	Arch Oral Biol	131	105251	2021
Tsuji M, Suzuki H, Suzuki S, Moriyama K	Three-dimensional evaluation of morphology and position of impacted supernumerary teeth in cases of cleidocranial dysplasia	Congenit Anom (Kyoto)	60(4)	106-14	2020
Watanabe T, Kometani-Gunjigake K, Nakao-Kuroishi K, Ito-Sago M, Mizuhara M, Iwata D, Moriyama K, Ono K, Kawamoto T	A Ser252Trp substitution in mouse FGFR2 results in hyperplasia of embryonic salivary gland parenchyma	J Oral Biosci	63(2)	184-91	2021
阿南 康太, 辻 美千子, 大河原 愛奈, 清水 美里, 稲垣 有美, 門田 千穂, 小笠原 毅, 森山 啓司	Turner 症候群を伴う患者における歯の特徴	Clinical and Investigative Orthodontics (Japanese Edition)	81(2)	79-86	2022
Kumaki T, Enomoto Y, Aida N, Goto T, Kurosawa K	Progression of cerebral and cerebellar atrophy in congenital contractures of limbs and face, hypotonia, and developmental delay	Pediatr Int	64(1)	e14734	2022
Murakami H, Uehara T, Enomoto Y, Nishimura N, Kumaki T, Kuroda Y, Asano M, Aida N, Kosaki K, Kurosawa K	Persistent Hyperplastic Primary Vitreous with Microphthalmia and Coloboma in a Patient with Okur-Chung Neurodevelopmental Syndrome	Mol Syndromol	13(1)	2333-44	2022
Nishimura N, Murakami H, Hayashi T, Sato H, Kurosawa K	Multiple craniosynostosis and facial dysmorphisms with homozygous IL11RA variant caused by maternal uniparental isodisomy of	Congenit Anom (Kyoto)	60(5)	153-5	2020

	chromosome 9				
Yokoi T, Enomoto Y, Tsurusaki Y, Kurosawa K	Siblings with vascular Ehlers-Danlos syndrome inherited via maternal mosaicism	Congenit Anom (Kyoto)	61(3)	101-2	2021
黒澤 健司	臨床検査アップデート　アレイ CGH 法	Modern Media	68(11)	470-3	2022
黒澤 健司	難治性疾患(難病)を学ぶ　ヤング・シンプソン症候群	遺伝子医学	12(4)	85-9	2022
大橋 博文	【症候・疾患からみる小児の検査】疾患からみる臨床検査の進めかた　染色体異常が疑われるとき	小児科診療	83(増刊)	204-9	2020
Machida M, Katoh H, Machida M, Miyake A, Taira K, Ohashi H	The Association of Scoliosis and NSD1 Gene Deletion in Sotos Syndrome Patients	Spine (Phila Pa 1976)	46(13)	E726-E33	2021
大橋 博文	【小児疾患診療のための病態生理 2　改訂第 6 版】染色体異常、先天異常　Angelman 症候群	小児内科	53(増刊)	223-5	2021
大橋 博文	【成人移行支援の二面性】成人医療との連携の場　成人移行が難しい患者における成人診療科との連携の構築 成人診療科に専門医が少ない領域　Down 症候群を中心に	小児内科	53(8)	1296-9	2021
Endo Y, Funakoshi Y, Koga T, Ohashi H, Takao M, Miura K, Yoshiura KI, Matsumoto T, Moriuchi H, Kawakami A	Large deletion in 6q containing the TNFAIP3 gene associated with autoimmune lymphoproliferative syndrome	Clin Immunol	235	108853	2022
Kaneko M, Oba D, Ohashi H	Survey on experiences and attitudes of parents toward disclosing information to children with genetic syndromes and their siblings in Japan	Sci Rep	12(1)	15234	2022
Lima AR, Ferreira BM, Zhang C, Jolly A, Du H, White JJ, Dawood M, Lins TC, Chiabai MA, van Beusekom E, Cordoba MS, Caldas Rosa ECC, Kayserili H, Kimonis V, Wu E, Mellado C, Aggarwal V, Richieri-Costa A, Brunoni D, Cano TM, Jorge AAL, Kim CA, Honjo R, Bertola DR, Dandalo-Girardi RM, Bayram Y, Gezdirici A, Yilmaz-Gulec E,	Phenotypic and mutational spectrum of ROR2-related Robinow syndrome	Hum Mutat	43(7)	900-18	2022

Gumus E, Yilmaz GC, Okamoto N, Ohashi H, Coban-Akdemir Z, Mitani T, Jhangiani SN, Muzny DM, Regattieri NAP, Pogue R, Pereira RW, Otto PA, Gibbs RA, Ali BR, van Bokhoven H, Brunner HG, Sutton VR, Lupski JR, Vianna-Morgante AM, Carvalho CMB, Mazzeu JF					
Machida M, Rocos B, Taira K, Nemoto N, Oikawa N, Ohashi H, Machida M, Kinoshita T, Kamata Y, Nakanishi K	Costello syndrome-associated orthopaedic manifestations focussed on kyphoscoliosis: a case series describing the natural course	J Pediatr Orthop B			2022
Masunaga Y, Nishimura G, Takahashi K, Hishiyama T, Imamura M, Kashimada K, Kadoya M, Wada Y, Okamoto N, Oba D, Ohashi H, Ikeno M, Sakamoto Y, Fukami M, Saitsu H, Ogata T	Clinical and molecular findings in three Japanese patients with N-acetylneuraminic acid synthetase-congenital disorder of glycosylation (NANS-CDG)	Sci Rep	12(1)	17079	2022
Mizukami T, Sonck J, Sakai K, Ko B, Maeng M, Otake H, Koo BK, Nagumo S, Norgaard BL, Leipsic J, Shinke T, Munhoz D, Mileva N, Belmonte M, Ohashi H, Barbato E, Johnson NP, De Bruyne B, Collet C	Procedural Outcomes After Percutaneous Coronary Interventions in Focal and Diffuse Coronary Artery Disease	J Am Heart Assoc	11(23)	e026960	2022
大橋 博文	【近未来の小児科のあり方・これからの展望】新生児医療　出生前診断の進歩	小児科	63(13)	1581-90	2022
大橋 博文	【見て,聞いて,触って,五感で診る新生児の異常とその対応】頭髪の異常　色・分布など	周産期医学	52(10)	1331-3	2022
大橋 博文	【知っておくべき周産期・新生児領域の遺伝学的検査を展望する】新生児における遺伝学的検査の現状と課題	周産期医学	52(5)	659-63	2022
Horiguchi A, Koichihara R, Kikuchi K, Nonoyama H, Daida A, Oba D, Hirata Y, Matsuura R, Ohashi H, Hamano SI	Efficacy of antiseizure medications in Wolf-Hirschhorn syndrome	Neuropediatrics			2023
Saito K, Nakagawa R, Narumi S, Ohashi H, Ishiguro A, Kabe K	A Small-for-Gestational-Age Infant with MIRAGE Syndrome Who Developed Heat Stroke and Rhabdomyolysis due to Severe Temperature Instability	Neonatology		1-5	2023
Ayoub S, Ghali N, Angwin C, Baker D, Baffini S, Brady AF, Giovannucci Uzielli ML, Giunta C, Johnson DS, Kosho T, Neas K, Pope FM, Rutsch F, Scarselli G, Sobey G,	Clinical features, molecular results, and management of 12 individuals with the rare	Am J Med Genet A	182(5)	994-1007	2020

Vandersteen A, van Dijk FS	arthrochalasia Ehlers-Danlos syndrome				
Lautrup CK, Teik KW, Unzaki A, Mizumoto S, Syx D, Sin HH, Nielsen IK, Markholt S, Yamada S, Malfait F, Matsumoto N, Miyake N, Kosho T	Delineation of musculocontractural Ehlers-Danlos Syndrome caused by dermatan sulfate epimerase deficiency	Mol Genet Genomic Med	8(5)	e1197	2020
Malfait F, Castori M, Francomano CA, Giunta C, Kosho T, Byers PH	The Ehlers-Danlos syndromes	Nat Rev Dis Primers	6(1)	64	2020
Uehara M, Oba H, Hatakenaka T, Ikegami S, Kuraishi S, Takizawa T, Munakata R, Mimura T, Yamaguchi T, Kosho T, Takahashi J	Posterior Spinal Fusion for Severe Spinal Deformities in Musculocontractural Ehlers-Danlos Syndrome: Detailed Observation of a Novel Case and Review of 2 Reported Cases	World Neurosurg	143	454-61	2020
古庄 知己	【難病研究の進歩】代謝・免疫　エーラス・ダンロス症候群	生体の科学	71(5)	488-9	2020
古庄 知己	【研修医と指導医に贈る　小児科学研究・論文のススメ】研修医に贈る　研究の仕方・論文の書き方　症例報告の書き方	小児科診療	83(7)	861-8	2020
古庄 知己	【臨床研究のための指針・法令を知る】[3]ヒトゲノム・遺伝子解析研究における倫理指針	Precision Medicine	3(7)	615-8	2020
Ishikawa S, Kosho T, Kaminaga T, Miyamoto M, Hamasaki Y, Yoshihara S, Hayashi S, Igawa K	Endoplasmic reticulum stress and collagenous formation anomalies in vascular-type Ehlers-Danlos syndrome via electron microscopy	J Dermatol	48(4)	481-5	2021
Miyake N, Kosho T, Matsumoto N	Ehlers Danlos Syndrome with Glycosaminoglycan Abnormalities	Adv Exp Med Biol	1348	235-49	2021
古庄 知己	【小児遺伝子疾患事典】先天異常症候群 CHST14(関連疾患 :Ehlers-Danlos 症候群)	小児科診療	84(11)	1431-5	2021
Minatogawa M, Hirose T, Mizumoto S, Yamaguchi T, Nagae C, Taki M, Yamada S, Watanabe T, Kosho T	Clinical and pathophysiological delineation of musculocontractural Ehlers-Danlos syndrome	Hum Mutat	43(12)	1829-36	2022

	caused by dermatan sulfate epimerase deficiency (mcEDS-DSE): A detailed and comprehensive glycobiological and pathological investigation in a novel patient				
Minatogawa M, Miyake N, Tsukahara Y, Tanabe Y, Uchiyama T, Matsumoto N, Kosho T	Expanding the phenotypic spectrum of cardiospondylocarpofacial syndrome: From a detailed clinical and radiological observation of a boy with a novel missense variant in MAP3K7	Am J Med Genet A	188(1)	350-6	2022
Takeda R, Yamaguchi T, Hayashi S, Sano S, Kawame H, Kanki S, Taketani T, Yoshimura H, Nakamura Y, Kosho T	Clinical and molecular features of patients with COL1-related disorders: Implications for the wider spectrum and the risk of vascular complications	Am J Med Genet A	188(9)	2560-75	2022
Yamaguchi T, Hayashi S, Hayashi D, Matsuyama T, Koitabashi N, Ogiwara K, Noda M, Nakada C, Fujiki S, Furutachi A, Tanabe Y, Yamanaka M, Ishikawa A, Mizukami M, Mizuguchi A, Sugiura K, Sumi M, Yamazawa H, Izawa A, Wada Y, Fujikawa T, Takiguchi Y, Wakui K, Takano K, Nishio SY, Kosho T	Comprehensive genetic screening for vascular Ehlers-Danlos syndrome through an amplification-based next-generation sequencing system	Am J Med Genet A	191(1)	37-51	2023
Hara-Isono K, Matsubara K, Fuke T, Yamazawa K, Satou K, Murakami N, Saitoh S, Nakabayashi K, Hata K, Ogata T, Fukami M, Kagami M	Genome-wide methylation analysis in Silver-Russell syndrome, Temple syndrome, and Prader-Willi syndrome	Clin Epigenetics	12(1)	159	2020
Hara-Isono K, Matsubara K, Mikami M, Arima T, Ogata T, Fukami M, Kagami M	Assisted reproductive technology represents a possible risk factor for development of epimutation-mediated imprinting disorders for mothers aged >/= 30 years	Clin Epigenetics	12(1)	111	2020
Masunaga Y, Inoue T, Yamoto K, Fujisawa Y, Sato Y, Kawashima-Sonoyama Y, Morisada N, Iijima K, Ohata Y, Namba N, Suzumura H, Kuribayashi R, Yamaguchi Y, Yoshihashi H, Fukami M, Saitsu H, Kagami M, Ogata T	IGF2 Mutations	J Clin Endocrinol Metab	105(1)		2020
緒方 勤	【FGR/SGA をめぐるトピックス】産科 FGR/SGA の疫学と発症要因　主な発症要因:SGA を招く遺伝学的	周産期医学	50(11)	1816-21	2020

	異常-Silver-Russell 症候群を主に				
Fuke T, Nakamura A, Inoue T, Kawashima S, Hara KI, Matsubara K, Sano S, Yamazawa K, Fukami M, Ogata T, Kagami M	Role of Imprinting Disorders in Short Children Born SGA and Silver-Russell Syndrome Spectrum	J Clin Endocrinol Metab	106(3)	802-13	2021
Hara-Isono K, Matsubara K, Hamada R, Shimada S, Yamaguchi T, Wakui K, Miyazaki O, Muroya K, Kurosawa K, Fukami M, Ogata T, Kosho T, Kagami M	A patient with Silver-Russell syndrome with multilocus imprinting disturbance, and Schimke immuno-osseous dysplasia unmasked by uniparental isodisomy of chromosome 2	J Hum Genet	66(11)	1121-6	2021
Kagami M, Hara-Isono K, Matsubara K, Nakabayashi K, Narumi S, Fukami M, Ohkubo Y, Saitsu H, Takada S, Ogata T	ZNF445: a homozygous truncating variant in a patient with Temple syndrome and multilocus imprinting disturbance	Clin Epigenetics	13(1)	119	2021
Masunaga Y, Fujisawa Y, Muramatsu M, Ono H, Inoue T, Fukami M, Kagami M, Saitsu H, Ogata T	Insulin resistant diabetes mellitus in SHORT syndrome: case report and literature review	Endocr J	68(1)	111-7	2021
Masunaga Y, Kagami M, Kato F, Usui T, Yonemoto T, Mishima K, Fukami M, Aoto K, Saitsu H, Ogata T	Parthenogenetic mosaicism: generation via second polar body retention and unmasking of a likely causative PER2 variant for hypersomnia	Clin Epigenetics	13(1)	73	2021
Omark J, Masunaga Y, Hannibal M, Shaw B, Fukami M, Kato F, Saitsu H, Kagami M, Ogata T	Kagami-Ogata syndrome in a patient with 46,XX,t(2;14)(q11.2;q32.2)mat disrupting MEG3	J Hum Genet	66(4)	439-43	2021
緒方 勤, 鏡 雅代	新規ヒトインプリンティング疾患「Kagami-Ogata 症候群」の樹立	浜松医科大学小児科学雑誌	1(1)	4-19	2021
緒方 勤, 村上 信行, 永井 敏郎	【成長ホルモン(GH)治療を考える】プラダーウイリ症候群と GH 治療	糖尿病・内分泌代謝科	53(2)	178-86	2021
Eggermann T, Yapici E, Bliek J, Pereda A, Begemann M, Russo S, Tannorella P, Calzari L, de Nanclares GP, Lombardi P, Temple IK, Mackay D, Riccio A, Kagami M, Ogata T, Lapunzina P, Monk D, Maher ER, Tumer Z	Trans-acting genetic variants causing multilocus imprinting disturbance (MLID): common mechanisms and consequences	Clin Epigenetics	14(1)	41	2022
Fuke T, Nakamura A, Inoue T, Kawashima S, Hara-Isono K, Matsubara K, Sano S, Yamazawa K, Fukami M, Ogata T, Kagami M	Frequency and clinical characteristics of distinct etiologies in patients with Silver-Russell syndrome diagnosed based on the	J Hum Genet	67(10)	607-11	2022

	Netchine-Harbison clinical scoring system				
Hara-Isono K, Nakamura A, Fuke T, Inoue T, Kawashima S, Matsubara K, Sano S, Yamazawa K, Fukami M, Ogata T, Kagami M	Pathogenic Copy Number and Sequence Variants in Children Born SGA With Short Stature Without Imprinting Disorders	J Clin Endocrinol Metab	107(8)	e3121-e33	2022
Kawashima S, Yuno A, Sano S, Nakamura A, Ishiwata K, Kawasaki T, Hosomichi K, Nakabayashi K, Akutsu H, Saitsu H, Fukami M, Usui T, Ogata T, Kagami M	Familial Pseudohypoparathyroidism Type IB Associated with an SVA Retrotransposon Insertion in the GNAS Locus	J Bone Miner Res	37(10)	1850-9	2022
Mackay D, Bliek J, Kagami M, Tenorio-Castano J, Pereda A, Brioude F, Netchine I, Papingi D, de Franco E, Lever M, Sillibourne J, Lombardi P, Gaston V, Tauber M, Diene G, Bieth E, Fernandez L, Nevado J, Tumer Z, Riccio A, Maher ER, Beygo J, Tannorella P, Russo S, de Nanclares GP, Temple IK, Ogata T, Lapunzina P, Eggermann T	First step towards a consensus strategy for multi-locus diagnostic testing of imprinting disorders	Clin Epigenetics	14(1)	143	2022
Tachibana N, Hosono K, Nomura S, Arai S, Torii K, Kurata K, Sato M, Shimakawa S, Azuma N, Ogata T, Wada Y, Okamoto N, Saitsu H, Nishina S, Hotta Y	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(2)		2022
緒方 勤	【知っておくべき周産期・新生児領域の遺伝学的検査を展望する】Silver-Russell 症候群とその周辺	周産期医学	52(5)	761-4	2022
緒方 勤	Genetics in CKD, インプリンティング	腎と透析	94(3)	339-45	2023
Abe J, Takeda A, Saitoh S	A case of tricuspid atresia with Prader-Willi syndrome	Pediatr Int	62(9)	1105-6	2020
Egawa K, Saitoh S, Asahina N, Shiraishi H	Short-latency somatosensory-evoked potentials demonstrate cortical dysfunction in patients with Angelman syndrome	eNeurologicalSci	22	100298	2021
Egawa K, Saitoh S, Asahina N, Shiraishi H	Variance in the pathophysiological impact of the hemizygosity of gamma-aminobutyric acid type A receptor subunit genes	Brain Dev	43(4)	521-7	2021

	between Prader-Willi syndrome and Angelman syndrome				
Fujimoto M, Nakamura Y, Iwaki T, Sato E, Ieda D, Hattori A, Shiraki A, Mizuno S, Saitoh S	Angelman syndrome with mosaic paternal uniparental disomy suggestive of mitotic nondisjunction	J Hum Genet	68(2)	87-90	2023
Ieda D, Negishi Y, Miyamoto T, Johmura Y, Kumamoto N, Kato K, Miyoshi I, Nakanishi M, Ugawa S, Oishi H, Saitoh S	Two mouse models carrying truncating mutations in Magel2 show distinct phenotypes	PLoS One	15(8)	e0237814	2020
Kondo Y, Aoyama K, Suzuki H, Hattori A, Hori I, Ito K, Yoshida A, Koroki M, Ueda K, Kosaki K, Saitoh S	De novo 2q36.3q37.1 deletion encompassing TRIP12 and NPPC yields distinct phenotypes	Hum Genome Var	7	19	2020
Mizumoto K, Kato K, Fujinami K, Sugita T, Sugita I, Hattori A, Saitoh S, Ueno S, Tsunoda K, Iwata T, Kondo M	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(50)	e32161	2022
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