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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
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 caused by dermatan sulfate epimerase deficiency (mcEDS-DSE): A detailed and comprehensive glycobiological and pathological investigation in a novel patient	Hum Mutat	43(12)	1829-36	2022
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
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	Frequency and clinical characteristics of distinct	J Hum Genet	67(10)	607-11	2022

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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
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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	Medicine (Baltimore)	101(50)	e32161	2022

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Negishi Y, Kurosawa K, Takano K, Matsubara K, Nishiyama T, Saitoh S	A nationwide survey of Schaaf-Yang syndrome in Japan	J Hum Genet	67(12)	735-8	2022
Saitoh S	Endosomal Recycling Defects and Neurodevelopmental Disorders	Cells	11(1)		2022
Minatogawa M, Tsuji T, Inaba M, Kawakami N, Mizuno S, Kosho T	Atypical Sotos syndrome caused by a novel splice site variant	Hum Genome Var	9(1)	41	2022
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Higashimoto K, Hara S, Soejima H	DNA Methylation Analysis Using Bisulfite Pyrosequencing	Methods Mol Biol	2577	3-20	2023
Soejima H, Hara S, Ohba T, Higashimoto K	Placental Mesenchymal Dysplasia and Beckwith-Wiedemann Syndrome	Cancers (Basel)	14(22)		2022
副島 英伸	【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】Beckwith-Wiedemann 症候群と Sotos 症候群	周産期医学	52(5)	755-9	2022
Hamaguchi Y, Mishima H, Kawai T, Saitoh S, Hata K, Kinoshita A, Yoshiura KI	Identification of unique DNA methylation sites in Kabuki syndrome using whole genome bisulfite	J Hum Genet	67(12)	711-20	2022

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Tamura S, Kosako H, Furuya Y, Yamashita Y, Mushino T, Mishima H, Kinoshita A, Nishikawa A, Yoshiura KI, Sonoki T	A Patient with Kabuki Syndrome Mutation Presenting with Very Severe Aplastic Anemia	Acta Haematol	145(1)	89-96	2022
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沼部 博直	先天異常の遺伝学的診断	東京医科大学雑誌	80(2)	81-7	2022
Akutsu K, Watanabe A, Yamada T, Sahara T, Hiraoka S, Shimizu W	Vascular Involvements Are Common in the Branch Arteries of the Abdominal Aorta Rather Than in the Aorta in Vascular Ehlers-Danlos Syndrome	CJC Open	5(1)	72-6	2023
Nagaya S, Maruyama K, Watanabe A, Meguro-Horike M, Imai Y, Hiroshima Y, Horike SI, Kokame K, Morishita E	First report of inherited protein S deficiency caused by paternal PROS1 mosaicism	Haematologica	107(1)	330-3	2022
渡邉 淳, 池田 和美, 関屋 智子	FOCUS　認定遺伝カウンセラー(CGC)	検査と技術	50(4)	418-21	2022
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Nakashima M, Argilli E, Nakano S, Sherr EH, Kato M, Saitsu H	De novo CLCN3 variants affecting Gly327 cause severe neurodevelopmental syndrome with brain structural abnormalities	J Hum Genet	68(4)	291-8	2023

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Yoshikawa Y, Koto T, Ishida T, Uehara T, Yamada M, Kosaki K, Inoue M	Rhegmatogenous Retinal Detachment in Musculocontractural Ehlers-Danlos Syndrome Caused by Biallelic Loss-of-Function Variants of Gene for Dermatan Sulfate Epimerase	J Clin Med	12(5)		2023