研究成果の刊行に関する一覧表

書　籍

著者氏名	タイトル	書籍全体の編集者名	書籍名	出版社名	出版地	出版年	ページ
森崎裕子	遺伝子診断	日本循環器学会/ 日本心臓血管外科学会/ 日本胸部外科学会/ 日本血管外科学会	2020 年改訂版　大動脈瘤・大動脈解離診療ガイドライン診療ガイドライン	日本循環器学会	東京	2020	138-141
仁科幸子	視覚器の異常	秋山千枝子、五十嵐隆、岡明、平岩幹男	小児保健ガイドブック	診断と治療社	東京	2021	96-98
仁科幸子	角膜の先天・周産期異常、網膜の周産期・発育異常	大鹿哲郎	眼科学 第３版	文光堂	東京	2020	111-113、349-351
渋谷絹子, 依田哲也, 田上順次, 若林則幸, 倉林亨, 森山啓司, 宮新美智世, 原田浩之, 紺野肖子, 月川和香奈	矯正歯科治療		系統看護学講座 専門分野 2-[15] 歯・口腔	医学書院	東京	2021	72-76
Kosho T, Miyake N	Coffin-Siris Syndrome	Carey JC, Cassidy SB, Battaglia A, Viskochil D	Cassidy and Allanson's Management of Genetic Syndromes, 4th Edition	Wiley-Blackwell	Hoboken, NJ, USA	2021	185-194
齋藤伸治、吉橋博史	Prader-Willi 症候群（PWS）と Angelman 症候群（AS）	関沢昭彦、佐村修、四元淳子	周産期遺伝カウンセリングマニュアル改定３版	中外医学社	東京	2020	126-130
水野誠司	Sotos 症候群、神経線維腫症１型		今日の小児治療指針　第 17 版	医学書院	東京	2020	172-173
岡本伸彦	性染色体異常症		今日の小児治療指針	医学書院	東京	2020	
岡本伸彦	先天性グリコシル化異常症		今日の小児治療指針	医学書院	東京	2020	
宮本達雄、藤田和将、松浦伸也	ゲノム編集技術を用いた培養細胞における疾患モデリング		医学のあゆみ	医歯薬出版株式会社	東京	2020	20977-20982

Soejima H, Ohba T.	Chapter 11 Genomic Imprinting Disorders (Including Mesenchymal Placental Dysplasia)	Masuzaki H (ed.)	Fetal Morph Functional Diagnosis	Springer	Singapore	2020	149-168
沼部 博直	22q11.2 欠失症候群	総編集 水口 雅, 市橋光, 伊藤秀一	今日の小児治療指針 第 17 版	医学書院	東京	2020	671-672
沼部 博直	先天異常，染色体異常 -総論	水口雅，山形崇倫編	クリニカルガイド 小児科専門医の診断・治療	南山堂	東京	2021	354-359
加藤光広	皮質形成異常	水口雅、市橋光、崎山弘、伊藤秀一編	今日の小児治療指針 第 17 版	医学書院	東京	2020	685-686
加藤光広	その他のてんかん発作を呈する神経疾患の遺伝子異常(脳形成異常・PME)	日本てんかん学会編	今日の小児治療指針 第 17 版	診断と治療社	東京	2020	21-23

雑　誌

発表者氏名	論文タイトル名	発表誌名	巻号	頁	出版年
Biesecker LG, Adam MP, Alkuraya FS, Amemiya AR, Bamshad MJ, Beck AE, Bennett JT, Bird LM, Carey JC, Chung B, Clark RD, Cox TC, Curry C, Dinulos MBP, Dobyns WB, Giampietro PF, Girisha KM, Glass IA, Graham JM Jr, Gripp KW, Haldeman-Englert CR, Hall BD, Innes AM, Kalish JM, Keppler-Noreuil KM, Kosaki K, Kozel BA, Mirzaa GM, Mulvihill JJ, Nowaczyk MJM, Pagon RA, Retterer K, Rope AF, Sanchez-Lara PA, Seaver LH, Shieh JT, Slavotinek AM,	A dyadic approach to the delineation of diagnostic entities in clinical genomics	American journal of human genetics	108(1)	8-15	2021

Sobering AK, Stevens CA, Stevenson DA, Tan TY, Tan WH, Tsai AC, Weaver DD, Williams MS, Zackai E, Zarate YA.					
Murakami H, Tsurusaki Y, Enomoto K, Kuroda Y, Yokoi T, Furuya N, Yoshihashi H, Minatogawa M, Abe-Hatano Chihiro, Ohashi I, Nishimura N, Kumaki T, Enomoto Y, Naruto T, Iwasaki F, Harada N, Ishikawa A, Kawame H, Sameshima K, Yamaguchi Y, Kobayashi M, Tominaga M, Ishikiriyama S, Tanaka T, Suzumura H, Ninomiya S, Kondo A, Kaname T, Kosaki K, Masuno M, Kuroki Y, Kurosawa K.	Update of the genotype and phenotype of KMT2D and KDM6A by genetic screening of 100 patients with clinically suspected Kabuki syndrome	American journal of medical genetics Part A	182(10)	2333-44	2020
Numabe H, Kosaki K.	Prevalence of Hallermann-Streiff syndrome in a Japanese pediatric population	Pediatrics international : official journal of the Japan Pediatric Society	63(4)	474-5	2021
Fujita H, Sasaki T, Miyamoto T, Akutsu SN, Sato S, Mori T, Nakabayashi K, Hata K, Suzuki H, Kosaki K, Matsuura S, Matsubara Y, Amagai M, Kubo A.	Premature aging syndrome showing random chromosome number instabilities with CDC20 mutation	Aging cell	19(11)	e13251	2020
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(®) (somatropin) in Noonan syndrome: a 4-year randomized, double-blind, multicenter trial in	Endocrine journal	67(8)	803-18	2020

	Japanese patients				
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	Clinical dysmorphology	29(4)	186-8	2020
Oiso N, Kubo A, Shimizu A, Suzuki H, Kosaki K, Chikugo T, Nakabayashi K, Hata K, Yanagihara S, Ishikawa O, Matsubara Y, Amagai M, Kawada.	Epidermodysplasia verruciformis without progression to squamous cell carcinomas in an elderly man: α-human papillomavirus infection in the evolving verruca	International journal of dermatology	59(9)	e334-e6	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	Journal of human genetics			2021
Mori R, Matsumoto H, Muro S, Morisaki H, Otsuki R	Loeys-Dietz Syndrome Presenting with Giant Bullae and Asthma	The journal of allergy and clinical immunology In practice	8(6)	9-15	2020
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	The Journal of thoracic and cardiovascular surgery			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	The Journal of thoracic and cardiovascular surgery	159(4)	1214-20	2020
Tanaka H, Kamiya CA, Horiuchi C, Morisaki H, Tanaka K, Katsuragi S, Hayata E, Hasegawa J, Nakata M, Sekizawa A, Ishiwata I, Ikeda T.	Aortic dissection during pregnancy and puerperium: A Japanese nationwide survey	The journal of obstetrics and gynaecology research	47(4)	1265-71	2021
Nakao S, Nishina S, Tanaka S, Yoshida T, Yokoi T, Azuma N.	Early laser photocoagulation for extensive retinal avascularity in infants with incontinentia pigmenti	Japanese journal of ophthalmology	64(6)	613-20	2020
Tanaka S, Yokoi	Structure of the Retinal	Ophthalmology			2020

T, Katagiri S, Yoshida-Uemura T, Nishina S, Azuma N.	Margin and Presumed Mechanism of Retinal Detachment in Choroidal Coloboma	Retina			
仁科幸子	特集 遺伝情報と遺伝カウンセリング　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 male case with mosaicism	Journal of human genetics	66(2)	205-14	2021
Nishina S, Hosono K, Ishitani S, Kosaki K, Yokoi T, Yoshida T, Tomita K, Fukami M, Saitsu H, Ogata T, Ishitani T Hotta Y, Azuma N.	Biallelic CDK9 variants as a cause of a new multiple-malformation syndrome with retinal dystrophy mimicking the CHARGE syndrome	Journal of human genetics			2021
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	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 genetics	16(4)	e1008643	2020
Yamamoto N, Mutai H, Namba	Clinical Profiles of DFNA11 at Diverse Stages of	Otology & neurotology :	41(6)	e663-e73	2020

K, Goto F, Ogawa K, Matsunaga T.	Development and Aging in a Large Family Identified by Linkage Analysis	official publication of the American Otological Society, American Neurotology Society [and] European Academy of Otology and Neurotology			
松永達雄	小児の難聴	小児内科	52(8)	1090-4	2020
仲野敦子，有本友季子，務台英樹，松永達雄	GJB2 遺伝子変異が検出された小児難聴症例の臨床経過と遺伝学的検査	日本耳鼻咽喉科学会会報	123(10)	1225-30	2020
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
Yamashita Y, Ogawa T, Ogaki K, Kamo H,Sukigara T, Kitahara E, Izawa N, Iwamuro H, Oyama G, Kamagata K, Hatano T, Umemura A, Kosaki R, Kubota M, Shimo Y, Hattori N	Neuroimaging evaluation and successful treatment by using directional deep brain stimulation and levodopa in a patient with GNAO1-associated movement disorder: A case report	Journal of the neurological sciences	411	116710	2020
Kosaki R, Kubota M, Uehara T, Suzuki H, Takenouchi T, Kosaki K	Consecutive medical exome analysis at a tertiary center: Diagnostic and health-economic outcomes	American journal of medical genetics Part A	182(7)	1601-7	2020
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, Domínguez-Garrido E, Montin D, Vincent M, Milani D, Biondi A, Gervasini C, Badolato R		Journal of clinical immunology	40(6)	851-60	2020
Suzuki-Muromoto S, Kosaki R, Kosaki	Familial hemiplegic migraine with a PRRT2 mutation: Phenotypic	Brain & development	42(3)	293-7	2020

K, Kubota M	variations and carbamazepine efficacy				
小﨑里華	先天性疾患染色体検査　検査値を読む　2020　内科		125((4))	p588	2020
Ishikawa T, Tamura E, Kasahara M, Uchida H, Higuchi M, Kobayashi H, Shimizu H, Ogawa E, Yotani N, Irie R, Kosaki R, Kosaki K, Uchiyama T, Onodera M, Kawai T		Journal of clinical immunology			2021
Takahashi Y, Kubota M, Kosaki R, Kosaki K, Ishiguro A	A severe form of autosomal recessive spinocerebellar ataxia associated with novel PMPCA variants	Brain & development	43(3)	464-9	2021
小﨑里華	本人に伝える遺伝カウンセリング	臨床遺伝専門医テキストシリーズ③ 小児領域			2021
Yamamoto M, Takashio S, Nakashima N, Hanatani S, Arima Y, Sakamoto K, Kaikita K, Aoki Y.	Double-chambered right ventricle complicated by hypertrophic obstructive cardiomyopathy diagnosed as Noonan syndrome	ESC heart failure	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 reports in neurology	13(1)	108-18	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	日本小児科学会雑誌	HRAS G12V 変異による最重症の Costello 症候群	125(3)	461-6	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	Congenital anomalies	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	Journal of oral biosciences			2021
Ogawa T, Cheng ES, Muramoto K, Moriyama K.	Long-Term Management and Maxillofacial Growth in a Klippel-Trenaunay Syndrome Patient	The Cleft palate-craniofacial journal : official	57(6)	782-90	2020

		publication of the American Cleft Palate-Craniofacial Association			
Inagaki Y, Ogawa T, Tabata MJ, Nagata Y, Watanabe R, Kawamoto T, Moriyama K, Ono K, Kawamoto T.	Identification of OPN3 as associated with non-syndromic oligodontia in a Japanese population	Journal of human genetics			2021
Min Swe NM, Kobayashi Y, Kamimoto H, Moriyama K.	Aberrantly activated Wnt/ß-catenin pathway co-receptors LRP5 and LRP6 regulate osteoblast differentiation in the developing coronal sutures of an Apert syndrome (Fgfr2(S252W) (/+)) mouse model	Developmental dynamics : an official publication of the American Association of Anatomists	250(3)	465-76	2021
Murakami H, Tsurusaki Y, Enomoto K, Kuroda Y, Yokoi T, Furuya N, Yoshihashi H,Minatogawa M,Abe-Hatano C, Ohashi I,Nishimura N, Kumaki T, Enomoto Y,Naruto T, Iwasaki F, Harada N, Ishikawa A, Kawame H, Sameshima K,Yamaguchi Y, Kobayashi M, Tominaga M,Ishikiriyama S, Tanaka T,Suzumura H, Ninomiya S,Kondo A, Kaname T, Kosaki K, Masuno M, Kuroki Y, Kurosawa K.	Update of the genotype and phenotype of KMT2D and KDM6A by genetic screening of 100 patients with clinically suspected Kabuki syndrome	American journal of medical genetics Part A	182(10)	2333-44	2020
Nishimura N, Kumaki T, Murakami H, Enomoto Y, Tsurusaki Y, Tsuji M, Tsuyusaki Y, Goto T, Aida N, Kurosawa K.	Expanding the phenotype of COL4A1-related disorders-Four novel variants	Brain & development	42(9)	639-45	2020
Nishimura N, Murakami H, Hayashi T, Sato	Multiple craniosynostosis and facial dysmorphisms with homozygous IL11RA	Congenital anomalies	60(5)	153-5	2020

H, Kurosawa K.	variant caused by maternal uniparental isodisomy of chromosome 9				
Ohashi I, Kuroda Y, Enomoto Y, Murakami H, Masuno M, Kurosawa K.	6p21 33 Deletion encompassing CSNK2B is associated with relative macrocephaly, facial dysmorphism, and mild intellectual disability	Clinical dysmorphology			2021
Yokoi T, Enomoto Y, Tsurusaki Y, Kurosawa K.	Siblings with vascular Ehlers-Danlos syndrome inherited via maternal mosaicism	Congenital anomalies	61(3)	101-2	2021
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	Publish Ahead of Print		2020
Ayoub S, Ghali N, Angwin C, Baker D, Baffini S, Brady AF, Giovannucci Uzielli ML, Giunta C, Johnson DS, Kosho T, Neas K, Pope F M et al.	Clinical features, molecular results, and management of 12 individuals with the rare arthrochalasia Ehlers-Danlos syndrome	American journal of medical genetics Part A	182(5)	994-1007	2020
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	Molecular genetics & genomic medicine	8(5)	e1197	2020
Malfait F, Castori M, Francomano CA, Giunta C, Kosho T, Byers PH.	The Ehlers-Danlos syndromes	Nature reviews Disease 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 neurosurgery	143	454-61	2020
古庄知己	特集　難病研究の進歩　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	特集・研修医と指導医に贈る小児科学研究・論文のススメ　Ⅰ．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	特集・臨床研究のための指針・法令を知る　ヒトゲノム・遺伝子解析研究における倫理指針	Precision Medicine	3((7))	19-22	2020
Ishikawa S, Kosho T, Kaminaga T, Miyamoto M, Hamasaki Y, Yoshihara S, et al	Endoplasmic reticulum stress and collagenous formation anomalies in vascular-type Ehlers-Danlos syndrome via electron microscopy	The Journal of dermatology	48(4)	481-5	2021
Fujisawa Y, Kitaoka T, Ono H, Nakashima S, Ozono K, Ogata T.	Case Report: Efficacy of Reduced Doses of Asfotase Alfa Replacement Therapy in an Infant With Hypophosphatasia Who Lacked Severe Clinical Symptoms	Frontiers in endocrinology	11	590455	2020
Fukami M, Fujisawa Y, Ono H, Jinno T, Ogata T.	Human Spermatogenesis Tolerates Massive Size Reduction of the Pseudoautosomal Region	Genome biology and evolution	12(11)	4-61	2020
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	Clinical epigenetics	12(1)	159	2020
Hara-Isono K, Matsubara K, Mikami M, Arima T, Ogata T, Fukami M, et al	Assisted reproductive technology represents a possible risk factor for development of epimutation-mediated imprinting disorders for mothers aged ≥ 30 years	Clinical epigenetics	12(1)	111	2020
Hayashi T, Hosono K, Kubo A, Kurata K, Katagiri S, Mizobuchi K, Kurai M, Mamiya N, Kondo M, Tachibana T, Saitsu H, Ogata T, Nakano T, Hotta Y.	Long-term observation of a Japanese mucolipidosis IV patient with a novel homozygous p F313del variant of MCOLN1	American journal of medical genetics Part A	182(6)	1500-5	2020
Hiraide T, Kubota K, Kono Y, Watanabe S, Matsubayashi T, Nakashima M, Kaname T, Fukao T, Shimozawa N, Ogata T, Saitsu H.	POLR3A variants in striatal involvement without diffuse hypomyelination	Brain & development	42(4)	363-8	2020
Hiraide T, Watanabe S, Matsubayashi T,	A de novo TOP2B variant associated with global developmental delay and	Molecular genetics & genomic	8(3)	e1145	2020

Yanagi K, Nakashima M, Ogata T, Saitsu H.	autism spectrum disorder	medicine			
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(®) (somatropin) in Noonan syndrome: a 4-year randomized, double-blind, multicenter trial in Japanese patients	Endocrine journal	67(8)	803-18	2020
Igarashi M, Masunaga Y, Hasegawa Y, Kinjo K, Miyado M, Saitsu H, Kato-Fukui Y, Horikawa R, Okubo Y, Ogata T, Fukami M .	Nonsense-associated altered splicing of MAP3K1 in two siblings with 46,XY disorders of sex development	Scientific reports	10(1)	17375	2020
Imura K, Ikeya S, Ogata T, Tokura Y.	Erythrokeratodermia variabilis et progressiva with a rare GJB3 mutation	The Journal of dermatology	47(4)	e111-e3	2020
Inoue T, Nakamura A, Iwahashi-Odano M, Tanase-Nakao K, Matsubara K, Nishioka J, Maruo Y.Hasegawa Y,Suzumura H, Sato S, Kobayashi Y, Murakami N, Nakabayashi K,Yamazawa K, Fuke T,Narumi S, Oka A, Ogata T, Fukami M, Kagami, M.	Contribution of gene mutations to Silver-Russell syndrome phenotype: multigene sequencing analysis in 92 etiology-unknown patients	Clinical epigenetics	12(1)	86	2020
Iwahashi-Odano M, Fujisawa Y, Ogata T, Nakashima S, Muramatsu M, Narumi S.	Identification and functional characterization of a novel PAX8 mutation (p,His39Pro) causing familial thyroid hypoplasia	Clinical pediatric endocrinology : case reports and clinical investigations : official journal of the Japanese Society for Pediatric Endocrinology	29(4)	173-8	2020
Kinjo K, Nagasaki K, Muroya K, Suzuki E, Ishiwata K, Nakabayashi K, Hattori A, Nagao K, Nozawa RS, Obuse C, Miyado K, Ogata T,	Rare variant of the epigenetic regulator SMCHD1 in a patient with pituitary hormone deficiency	Scientific reports	10(1)	10985	2020

Fukami M, Miyado M.					
Kinjo K, Yoshida T, Kobori Y, Okada H, Suzuki E, Ogata T, Miyado M, Fukami M.	Random X chromosome inactivation in patients with Klinefelter syndrome	Molecular and cellular pediatrics	7(1)	1	2020
Mano H, Fujiwara S, Takamura K, Kitoh H, Takayama S, Ogata T, Haga N.	Treatment approaches for congenital transverse limb deficiency: Data analysis from an epidemiological national survey in Japan	Journal of orthopaedic science : official journal of the Japanese Orthopaedic Association			2020
Masunaga Y, Inoue T, Yamoto K, Fujisawa Y, Sato Y, Kawashima-Sonoyama Y, Morisada N, Iijima K, Ohata Y, Kagami M, Namba N, Suzumura H, Kuribayashi R, Yamaguchi Y, Yoshihashi H, Fukami M, Saitsu H, Kagami M, Kagami M, Ogata T.	IGF2 Mutations	The Journal of clinical endocrinology and metabolism	105(1)		2020
Uchiyama H, Masunaga Y, Ishikawa T, Fukuoka T, Fukami M, Saitsu H, Ogata T.	TSC1 intragenic deletion transmitted from a mosaic father to two siblings with cardiac rhabdomyomas: Identification of two aberrant transcripts	European journal of medical genetics	63(11)	104060	2020
Yamazawa K, Inoue T, Sakemi Y, Nakashima T, Yamashita H, Khono K, Fujita H, Fujita H, Nakabayashi K, Hata K, Nakashima M, Matsunaga T, Nakamura A, Matsubara K, Ogata T, Kagami M.	Loss of imprinting of the human-specific imprinted gene ZNF597 causes prenatal growth retardation and dysmorphic features: implications for phenotypic overlap with Silver-Russell syndrome	Journal of medical genetics			2020
Yamoto K, Saitsu H, Fujisawa Y, Kato F, Matsubara K, Fukami M, Kagami M, Ogata T.	Coffin-Lowry syndrome in a girl with 46,XX,t(X;11)(p22;p15)dn: Identification of RPS6KA3 disruption by whole genome sequencing	Clinical case reports	8(6)	1076-80	2020

Binder G, Nakamura A, Schweizer R, Ogata T, Fukami M, Nagasaki K	Long-term Effect of Aromatase Inhibition in Aromatase Excess Syndrome	The Journal of clinical endocrinology and metabolism	106(5)	1491-500	2021
Fuke T, Nakamura A, Inoue T, Kawashima S, Hara KI, Matsubara K, Sano S, Fukami M, Ogata T.	Role of Imprinting Disorders in Short Children Born SGA and Silver-Russell Syndrome Spectrum	The Journal of clinical endocrinology and metabolism	106(3)	802-13	2021
Hiraide T, Yamoto K, Masunaga Y, Asahina M, Endoh Y, Ohkubo Y, Matsubayashi T, Tsurui S, Yamada H, Yanagi K, Nakashima M, Hirano K, Sugimura H, Fukuda T, Ogata T, Saitsu H.	Genetic and phenotypic analysis of 101 patients with developmental delay or intellectual disability using whole-exome sequencing	Clinical genetics			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	Endocrine journal	68(1)	111-7	2021
Masunaga Y, Kagami M, Kato F, Usui T, Yonemoto T, Mishima K, Fukami M, Kagami M, Saitsu H, Ogata T.	Parthenogenetic mosaicism: generation via second polar body retention and unmasking of a likely causative PER2 variant for hypersomnia	Clinical epigenetics	13(1)	73	2021
Masunaga Y, Mochizuki M, Kadoya M, Wada Y, Okamoto N, Fukami M, Kato F, Saitsu H, Ogata T.	Primary ovarian insufficiency in a female with phosphomannomutase-2 gene (PMM2) mutations for congenital disorder of glycosylation	Endocrine journal			2021
Nishina S, Hosono K, Ishitani S, Kosaki K, Yokoi T, Yoshida T, Tomita K, Fukami M, Saitsu H, Ogata T, Ishitani T Hotta Y, Azuma N.	Biallelic CDK9 variants as a cause of a new multiple-malformation syndrome with retinal dystrophy mimicking the CHARGE syndrome	Journal of human genetics			2021
Omark J, Masunaga Y,	Kagami-Ogata syndrome in a patient with	Journal of human genetics	66(4)	439-43	2021

Hannibal M, Shaw B, Fukami M, Kato F, Saitsu H, Kagami M, Ogata T.	46,XX,t(2;14)(q11,2;q322)mat disrupting MEG3				
Saida K, Fukuda T, Scott DA, Sengoku T, Ogata K, Nicosia A, Hernandez-Garcia A, Lalani SR, Azamian MS, Streff H, Liu P, Dai H, Mizuguchi T, Miyatake S, Miyatake S, Ogata T, Miyake N, Matsumoto N.	OTUD5 Variants Associated With X-Linked Intellectual Disability and Congenital Malformation	Frontiers in cell and developmental biology	9	631428	2021
Shima H TE, Okamoto S, Nagamori M,_ Ogata T, Narumi S, Nakamura A, Izumi Y, Jinno T, Suzuki E, Fukami M	SOX10 Mutation Screening for 117 Patients with Kallmann Syndrome	J Endocr Soc			2021
Tamaoka S, Suzuki E, Hattori A, Ogata T, Fukami M, Katoh-Fukui Y.	NDNF variants are rare in patients with congenital hypogonadotropic hypogonadism	Human genome variation	8(1)	5	2021
Abe J, Takeda A, Saitoh S.	A case of tricuspid atresia with Prader-Willi syndrome	Pediatrics international : official journal of the Japan Pediatric Society	62(9)	1105-6	2020
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	Clinical epigenetics	12(1)	159	2020
Ieda D, Negishi Y, Miyamoto T, Johmura Y, Kumamoto N, Kato K, Miyoshi I, Nakanishi M, Ugawa S, Ugawa S, 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,	De novo 2q36,3q37,1 deletion encompassing TRIP12 and NPPC yields distinct phenotypes	Human genome variation	0.304861111		2020

Saitoh, S.					
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 between Prader-Willi syndrome and Angelman syndrome	Brain & development	43(4)	521-7	2021
Ito Y, Ito T, Kurahashi N, Ochi N, Noritake K, Sugiura H, Mizuno S, Kidokoro H Natsume J, Nakamura M.	Gait characteristics of children with Williams syndrome with impaired visuospatial recognition: a three-dimensional gait analysis study	Experimental brain research	238(12)	2887-95	2020
Ivanovski I, Djuric O, Broccoli S, Caraffi SG, Accorsi P, Adam MP, Avela K, Badura-Stronka M, Bayat A, Clayton-Smith J, Cocco I, Cordelli DM, Cuturilo G, Di Pisa V, Dupont Garcia J, Dupont Garcia J, Gastaldi R, Giordano L, Guala Hoei-Hansen CA, Inaba M, Iodice A, Nielsen JEK, Kuburovic V, Lazalde-Medina B, Malbora B, Mizuno S, Moldovan O, Møller RS, et al.	Mowat-Wilson syndrome: growth charts	Orphanet journal of rare diseases	15(1)	151	2020
Kato K, Mizuno S, Morton J, Toyama M, Hara Y, Wasmer E, et al	Expanding the phenotype of biallelic loss-of-function variants in the NSUN2 gene: Description of four individuals with juvenile cataract, chronic nephritis, or brain anomaly as novel complications	American journal of medical genetics Part A	185(1)	282-5	2021
Suzuki H, Inaba M, Yamada M, Uehara T, Takenouchi T, Mizuno S, Kosaki	Biallelic loss of OTUD7A causes severe muscular hypotonia, intellectual disability, and seizures	American journal of medical genetics Part A	185(4)	1182-6	2021

K, Doi M.					
Yamada M, Suzuki H, Watanabe A, Uehara T, Takenouchi T, Mizuno S, Kosaki K.	Role of chimeric transcript formation in the pathogenesis of birth defects	Congenital anomalies	61(3)	76-81	2021
Zarate YA, Uehara T, Abe K, Oginuma M, Harako S, Ishitani S, , Lehesjoki AE, Bierhals T, Kloth K, Ehmke N, Horn D, Holtgrewe M, Anderson K, Viskochil D, Edgar-Zarate C, L, Sacoto MJG, Schnur RE, Morrow MM, Sanchez-Valle A, Pappas J, Rabin R, Muona M, Anttonen AK, Platzer K, Luppe J, Kaname T, Gburek-Augustat J, Okamoto N, Mizuno S, Kaido Y, Ohkuma Y, Hirose Y, Ishitani T, Kosaki K.	CDK19-related disorder results from both loss-of-function and gain-of-function de novo missense variants	Genetics in medicine : official journal of the American College of Medical Genetics			2021
Hirano M, Satake W, Moriyama N, Saida K, Okamoto N, Cha PC, et al	Bardet-Biedl syndrome and related disorders in Japan	Journal of human genetics	65(10)	847-53	2020
Yanagishita T, Imaizumi T, Yamamoto-Shimojima K, Yano T, Okamoto N, Nagata S, et al	Breakpoint junction analysis for complex genomic rearrangements with the caldera volcano-like pattern	Human mutation	41(12)	2119-27	2020
Faundes V, Goh S, Akilapa R, Bezuidenhout H, Bjornsson HT, Bradley L, Brady AF、Brischoux-Boucher E、Brunner H.Bulk S.Canham N, Cody D.Dentici ML, Digilio MC, Elmslie F, Fry AE, Gill H.Hurst	Clinical delineation, sex differences, and genotype-phenotype correlation in pathogenic KDM6A variants causing X-linked Kabuki syndrome type 2	Genetics in medicine : official journal of the American College of Medical Genetics			2021

J, Johnson D.Julia S, Lachlan K, Lebel RR, Byler M, Gershon E, Lemire E, Gnazzo M, Lepri FR, Marchese A, McEntagart M, McGaughran J, Mizuno S, Okamoto N, Rieubland C.Rodgers J, Sasaki E, Scalais E et al.					
Imaizumi T, Yamamoto-Shimojima K, Yanagishita T, Ondo Y, Nishi E, Okamoto N, et al	Complex chromosomal rearrangements of human chromosome 21 in a patient manifesting clinical features partially overlapped with that of Down syndrome	Human genetics	139(12)	1555-63	2020
Kennedy J, Goudie D, Blair E, Chandler K, Joss S, McKay V, Brady AF、Brischoux-Boucher E、Brunner H.Bulk S.Canham N, Cody D.Dentici ML, Digilio MC, Elmslie F, Fry AE, Gill H.Hurst J, Johnson D.Julia S, Lachlan K, Lebel RR, Byler M, Gershon E, Lemire E, Gnazzo M, Lepri FR, Marchese A, McEntagart M, McGaughran J, Mizuno S, Okamoto N, Rieubland C.Rodgers J, Sasaki E, Scalais E et al.	Correction: KAT6A Syndrome: genotype-phenotype correlation in 76 patients with pathogenic KAT6A variants	Genetics in medicine : official journal of the American College of Medical Genetics	22(11)	1920	2020
Cappuccio G, Sayou C, Tanno PL, Tisserant E, Bruel AL, Kennani SE,Sá J, Low KJ, Dias C, Havlovicová M, Eichler EE, Devillard F, Hančárová M, Devillard F,	De novo SMARCA2 variants clustered outside the helicase domain cause a new recognizable syndrome with intellectual disability and blepharophimosis distinct from Nicolaides-Baraitser syndrome	Genetics in medicine : official journal of the American College of Medical Genetics	22(11)	1838-50	2020

Moutton S, Van-Gils J, Dubourg C, Odent S, Gerard B, Piton A, Yamamoto T, Okamoto N, Firth H, Metcalfe K, et al.					
Yamamoto-Shimojima K, Akagawa H, Yanagi K, Kaname T, Okamoto N, Yamamoto T	Deep intronic deletion in intron 3 of PLP1 is associated with a severe phenotype of Pelizaeus-Merzbacher disease	Human genome variation	8(1)	14	2021
Okamoto N, Miya F, Kitai Y, Tsunoda T, Kato M, Saitoh S, et al	Homozygous ADCY5 mutation causes early-onset movement disorder with severe intellectual disability	Neurological sciences : official journal of the Italian Neurological Society and of the Italian Society of Clinical Neurophysiology			2021
Tajima D, Nakamura T, Ichinose F, Okamoto N, Tomonoh Y, Uda K, et al	Transient hypoglycorrhachia with paroxysmal abnormal eye movement in early infancy	Brain & development	43(3)	482-5	2021
Fujita H, Sasaki T, Miyamoto T, Akutsu SN, Sato S, Mori T, Nakabayashi K, Hata K, Suzuki H, Kosaki K, Matsuura S, Matsubara Y, Amagai M, Kubo A.	Premature aging syndrome showing random chromosome number instabilities with CDC20 mutation	Aging cell	19(11)	e13251	2020
Yukimoto H, Miyamoto T, Kiyono T, Wang S, Matsuura S, Mizoguchi A, et al	A novel CDK-independent function of p27(Kip1) in preciliary vesicle trafficking during ciliogenesis	Biochemical and biophysical research communications	527(3)	716-22	2020
冨岡啓太, 阿久津シルビア夏子, 柳原啓見, 田内広, 山本卓, 工藤美樹, 小林正夫, 宮本達雄, 松浦伸也.	放射線感受性の遺伝的個人差を規定する候補素因としてのNBS1 遺伝子 I171V 多型の定量的評価	広島医学	73((4))	224-7	2020
Higashimoto K, Watanabe H, Tanoue Y, Tonoki H, Tokutomi T, Hara S,Yatsuki H, Soejima H.	Hypomethylation of a centromeric block of ICR1 is sufficient to cause Silver-Russell syndrome	Journal of medical genetics	58(6)	422-5	2021
Kodera C, Aoki S, Ohba T, Higashimoto K,	Clinical manifestations of placental mesenchymal dysplasia in Japan: A	The journal of obstetrics and gynaecology	47(3)	1118-25	2021

Mikami Y, Fukunaga M, Yatsuki H, Soejima H.	multicenter case series	research			
沼部博直.	先天異常症候群と SIDS	日本 SIDS・乳幼児突然死予防学会雑誌	20(1)	16-22	2020
Numabe H, Kosaki K.	Prevalence of Hallermann-Streiff syndrome in a Japanese pediatric population	Pediatrics international : official journal of the Japan Pediatric Society	63(4)	474-5	2021
沼部博直.	先天性疾患と遺伝カウンセリング	泌尿器科	13 (2)	203-8	2021
Okazaki T, Adachi K, Matsuura K, Oyama Y, Nose M, Shirahata E, Abe T, Hasegawa T, Maihara T, Maegaki Y, Nanba E.	Clinical Characteristics of Fragile X Syndrome Patients in Japan	Yonago acta medica	64(1)	3-30	2021
Io S, Watanabe A, Yamada S, Mandai M, Yamada T.	Perinatal benign hypophosphatasia antenatally diagnosed through measurements of parental serum alkaline phosphatase and ultrasonography	Congenital anomalies	60(6)	199-200	2020
Ishijima Y, Iizuka T, Kagami K, Masumoto S, Nakade K, Mitani Y, Niida Y, Watanabe A, Ono M, Yamazaki R, Fujiwara H.	Prenatal diagnosis facilitated prompt enzyme replacement therapy for prenatal benign hypophosphatasia	Journal of obstetrics and gynaecology : the journal of the Institute of Obstetrics and Gynaecology	40(1)	132-4	2020
Nagata M, Setoh K, Takahashi M, Higasa K, Kawaguchi T, Kawasaki H, Wada T, Watanabe A, Sawai H, Tabara Y, Yamada T, Matsuda F, Kosugi S.	Association of ALPL variants with serum alkaline phosphatase and bone traits in the general Japanese population: The Nagahama Study	Journal of human genetics	65(3)	337-43	2020
Nakamura-Takahashi A, Tanase T, Matsunaga S, Shintani S, Abe S, Nitahara-Kasahara Y, Watanabe A, Hirai Y, Okada T, Yamaguchi A, Kasahara M.	High-Level Expression of Alkaline Phosphatase by Adeno-Associated Virus Vector Ameliorates Pathological Bone Structure in a Hypophosphatasia Mouse Model	Calcified tissue international	106(6)	665-77	2020

Nishizawa H, Sato Y, Ishikawa M, Arakawa Y, Iijima M, Akiyama T, Iijima M, Akiyama T, Takano K, Watanabe A, Kosho T.	Marked motor function improvement in a 32-year-old woman with childhood-onset hypophosphatasia by asfotase alfa therapy: Evaluation based on standardized testing batteries used in Duchenne muscular dystrophy clinical trials	Molecular genetics and metabolism reports	25	100643	2020
Togashi T, Meguro-Horike M, Nagaya S, Sugihara S, Ichinohe T, Araiso Y, Yamaguchi K, Mori K, Imai Y, Kuzasa K, Horike SI, Asakura H, Watanabe A, Morishita E.	Molecular genetic analysis of inherited protein C deficiency caused by the novel large deletion across two exons of PROC	Thrombosis research	188	115-8	2020
Tsuchiya M, Yamada T, Akaishi R, Hamanoue H, Hirasawa A, Hyodo M, Shimizu K, Tamai M, Umemura H, Watanabe A, Yoshida A, Yoshihashi H, Yotsumoto J, Kosugi S.	Attitudes toward and current status of disclosure of secondary findings from next-generation sequencing: a nation-wide survey of clinical genetics professionals in Japan	Journal of human genetics	65(12)	1045-53	2020
Hiromoto Y, Azuma Y, Suzuki Y, Hoshina M, Uchiyama Y, Mitsuhashi S, Miyatake S, Mizuguchi T, Takata A, Miyake N, Kato M, Matsumoto N.	Hemizygous FLNA variant in West syndrome without periventricular nodular heterotopia	Human genome variation	7(1)	43	2020
Itai T, Miyatake S, Taguri M, Nozaki F, Ohta M, Osaka H, Nabatame S, Smigiel R, Kato M, Tanda K, Saito Y, Ishiyama A, Noguchi Y et al.	Prenatal clinical manifestations in individuals with COL4A1/2 variants	Journal of medical genetics			2020
Miyake N, Takahashi H, Nakamura K, Isidor B, Hiraki Y, Koshimizu E, Mizuguchi T,	Gain-of-Function MN1 Truncation Variants Cause a Recognizable Syndrome with Craniofacial and Brain Abnormalities	American journal of human genetics	106(1)	13-25	2020

Takata A, Obo K, Kato M, Ogata K, Matsumoto N.					
Nakashima M, Kato M, Matsukura M, Kira R, Ngu LH, Lichtenbelt KD, Mitsuhashi S, Saitsu H, Matsumoto N.	De novo variants in CUL3 are associated with global developmental delays with or without infantile spasms	Journal of human genetics	65(9)	727-34	2020
Chen Y, Sakurai K, Maeda S, Masui T, Okano H, Dewender J, Seltmann S, Kurtz A Masuya H, Nakamura Y, Sheldon M, Schneider J, Stacey GN, Panina Y, Fujibuchi W.	Integrated Collection of Stem Cell Bank Data, a Data Portal for Standardized Stem Cell Information	Stem cell reports	16(4)	997-1005	2021
Kosaki R, Kubota M, Uehara T, Suzuki H, Takenouchi T, Kosaki K	Consecutive medical exome analysis at a tertiary center: Diagnostic and health-economic outcomes	American journal of medical genetics Part A	182(7)	1601-7	2020
Sakaguchi Y, Yoshihashi H, Uehara T, Miyama S, Kosaki K, Takenouchi T	Coloboma may be a shared feature in a spectrum of disorders caused by mutations in the WDR37-PACS1-PACS2 axis	American journal of medical genetics Part A	185(3)	884-8	2021
Suzuki H, Yamada M, Uehara T, Takenouchi T, Kosaki K	Parallel detection of single nucleotide variants and copy number variants with exome analysis: Validation in a cohort of 700 undiagnosed patients	American journal of medical genetics Part A	182(11)	2529-32	2020
Takeshita Y, Ohto T, Enokizono T, Tanaka M, Suzuki H, Fukushima H, Uehara T, Takenouchi T, Kosaki K, Takada H.	Novel ARX mutation identified in infantile spasm syndrome patient	Human genome variation	7	9	2020