研究成果の刊行に関する一覧表

書　籍

著者氏名	論文タイトル名	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	Marfan 症候群およびその類縁疾患	日本小児循環器学会	小児・成育循環器学	診断と治療社	東京	2018	641-645
森崎裕子	先天性心疾患	櫻井晃洋	最新多因子遺伝性疾患研究と遺伝カウンセリング	メディカルドゥ	大阪	2018	89-94
仁科幸子	新生児・乳児の眼科的異常	五十嵐隆	小児科診療ガイドライン―最新の治療指針―第 4 版	総合医学社	東京	2019	741-744
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仁科幸子	未熟児網膜症―眼底検査法と写真撮影法．リハビリテーション・ロービジョンケア．類縁疾患	東範行	未熟児網膜症	三輪書店	東京	2018	
松永達雄	遺伝性難聴の診断の進歩	山岨達也	医学のあゆみ BOOKS 耳鼻咽喉科診療の進歩　40 のエッセンス	医歯薬出版	東京	2018	7-10
松永達雄	前庭水管拡大症	森山寛　監修．大森孝一、藤枝重治、小島博己、猪原秀典　編集	今日の耳鼻咽喉科・頭頸部外科治療指針 第 4 版	医学書院	東京	2018	257-258
松永達雄	耳介・外耳道の先天異常（耳瘻孔を含む）congenital anomalies of auricle and external ear canal	福井次矢、高木誠、小室一成	今日の治療指針 2019 年版（私はこう治療している）	医学書院	東京	2019	1545

黒澤健司	多発性翼状片症候群	柳瀬敏彦	内分泌症候（第 3 版）Ⅳ 領域別症候群シリーズ 4	日本臨床社	東京	2019	443-445
黒澤健司	CHARGE 症候群	柳瀬敏彦	内分泌症候（第 3 版）Ⅳ 領域別症候群シリーズ 4	日本臨床社	東京	2019	475-479
水野誠司	22q11.2 欠失症候群	五十嵐 隆	小児疾患の診断治療指針 第 5 版	東京医学社	東京	2018	144-145
岡本伸彦	最先端のゲノム医療と遺伝カウンセリング	大阪母子医療センター	こどもと妊婦の病気・治療がわかる本―大阪母子医療センターの今	バリューメディカル	東京	2018	
副島英伸	第 6 講義エピジェネティクス	日本人類遺伝学会編集	コアカリ準拠臨床遺伝学テキストノート	診断と治療社	東京	2018	72-83
渡邉淳		中込さと子（監修），西垣昌和（編集），渡邉淳（編集）	基礎から学ぶ遺伝看護学	羊土社	東京	2019	1-177
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小崎健次郎	【遺伝子解析研究の新時代】疾患ゲノム研究最前線 希少疾患　クリニカルシーケンスとデータ共有　ゲノム医療の実装に向けて	医学のあゆみ	266（5）	416-420	2018
小崎健次郎	【小児診療ガイドラインの読み解き方-ガイドラインの背景、使い方を中心に】 小児関連学会(分野)のガイドラインへの取り組み　先天異常症候群領域(日本小児遺伝学会)	小児内科	50（5）	852-855	2018
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占部良介, 早川格, 上田菜穂子, 武井剛, 鈴木智, 神岡哲治, 寺嶋宙, 久保田雅也, 小崎里華 and 加藤光広	新生児期発症の難治性てんかん性脳症をきたしたSCN2A変異の男児例	てんかん研究	36（2）	501	2018
三浦真理子, 村本美香, 益田博司, 小崎里華, 伊藤裕司, 鏡雅代 and 和田友香	Prader-Willi症候群との鑑別が困難であったTemple症候群の新生児例	小児科臨床	71（6）	1121-1126	2018
早川格, 寺嶋宙, 小崎里華 and 久保田雅也	大頭と発達遅滞を呈しPTEN遺伝子変異を認めた2例	脳と発達	50（5）	372	2018
A. Tamura, S. Uemura, K. Matsubara, E. Kozuki, T. Tanaka, N. Nino, T. Yokoi, A. Saito, T. Ishida, D. Hasegawa, I. Umeki, T. Niihori, Y. Nakazawa, K. Koike, Y.Aoki and Y. Kosaka	Co-occurrence of hypertrophic cardiomyopathy and juvenile myelomonocytic leukemia in a neonate with Noonan syndrome, leading to premature death	Clin Case Rep	6（7）	1202-1207	2018
S. Takahara, S. I. Inoue, S. Miyagawa-Tomita, K. Matsuura, Y. Nakashima, T. Niihori, Y.Matsubara, Y. Saiki and Y.Aoki	New Noonan syndrome model mice with RIT1 mutation exhibit cardiac hypertrophy and susceptibility to beta-adrenergic stimulation-induced cardiac fibrosis	EBioMedicine	42	43-53	2019

S. Suzuki-Muromoto, T. Miyabayashi, K. Nagai, S. Yamamura-Suzuki, M. Anzai, Y. Takezawa, R. Sato, Y. Okubo, W. Endo, T. Inui, N. Togashi, A. Kikuchi, T. Niihori, Y.Aoki, S. Kure and K. Haginoya	Leucine-485 deletion variant of BRAF may exhibit the severe end of the clinical spectrum of CFC syndrome	J Hum Genet	64（5）	499-504	2019
N. Ahiko, Y. Baba, M. Tsuji, R. Horikawa and K.Moriyama	Investigation of maxillofacial morphology and oral characteristics with Turner syndrome and early mixed dentition	Congenit Anom (Kyoto)	59（1）	43786	2019
S. Matsuno, M. Tsuji, R. Hikita, T. Matsumoto, Y. Baba and K.Moriyama	Clinical study of dentocraniofacial characteristics in patients with Williams syndrome	Congenit Anom (Kyoto)		2018	
K. Yamaji, J. Morita, T. Watanabe, K. Gunjigake, M. Nakatomi, M. Shiga, K. Ono, K.Moriyama and T. Kawamoto	Maldevelopment of the submandibular gland in a mouse model of apert syndrome	Dev Dyn	247（11）	1175-1185	2018
N. Higashihori, J. I. Takada, M. Katayanagi, Y. Takahashi and K.Moriyama	Frequency of missing teeth and reduction of mesiodistal tooth width in Japanese patients with tooth agenesis	Prog Orthod	19（1）	30	2018
Y. Takahashi, N. Higashihori, Y. Yasuda, J. I. Takada and K.Moriyama	Examination of craniofacial morphology in Japanese patients with congenitally missing teeth: a cross-sectional study	Prog Orthod	19（1）	38	2018
森山啓司	顎顔面先天異常に対する歯科矯正学的アプローチ　頭蓋縫合早期癒合症の臨床・研究を中心に	中・四国矯正歯科学会雑誌	30（1）	43471	2018
T. Yokoi, T. Saito, J. I. Nagai and K.Kurosawa	17q21.32-q22 Deletion in a girl with osteogenesis imperfecta, tricho-dento-osseous syndrome, and intellectual disability	Congenit Anom (Kyoto)	59（2）	51-52	2019

HayashiS., YokoiT., HatanoC., EnomotoY., TsurusakiY., NarutoT., KobayashiM., IdaH. and KurosawaK.	Biallelic mutations of EGFR in a compound heterozygous state cause ectodermal dysplasia with severe skin defects and gastrointestinal dysfunction	Hum Genome Var	5	11	2018
T. Yokoi, Y. Enomoto, Y. Tsurusaki, T. Naruto and K.Kurosawa	Nonsyndromic intellectual disability with novel heterozygous SCN2A mutation and epilepsy	Hum Genome Var	5	20	2018
黒澤健司	指定難病最前線(Volume72) ヤング・シンプソン症候群	新薬と臨牀	67（11）	1371-1374	2018
L. Guo, D. R. Bertola, A. Takanohashi, A. Saito, Y. Segawa, T. Yokota, S. Ishibashi, Y. Nishida, G. L. Yamamoto, J. Franco, R. S. Honjo, C. A. Kim, C. M. Musso, M. Timmons, A. Pizzino, R. J. Taft, B. Lajoie, M. A. Knight, K. H. Fischbeck, A. B. Singleton, C. R. Ferreira, Z. Wang, L. Yan, J. Y. Garbern, P. O. Simsek-Kiper, H.Ohashi, P. G. Robey, A. Boyde, N. Matsumoto, N. Miyake, J. Spranger, R. Schiffmann, A. Vanderver, G. Nishimura, M. Passos-Bueno, C. Simons, K. Ishikawa and S. Ikegawa	Bi-allelic CSF1R Mutations Cause Skeletal Dysplasia of Dysosteosclerosis-Pyle Disease Spectrum and Degenerative Encephalopathy with Brain Malformation	Am J Hum Genet	104（5）	925-935	2019
T. Motojima, K. Fujii, H.Ohashi and H. Arakawa	Catathrenia in Pitt-Hopkins syndrome associated with 18q interstitial deletion	Pediatr Int	60（5）	479-481	2018
R. Matsuura, S. I. Hamano, T. Iwamoto, K. Shimizu and H.Ohashi	First Patient With Salla Disease Confirmed by Genomic Analysis in Japan	Pediatr Neurol	81（52-53	2018	
E. Nishi, M. Takasugi, R. Kawamura, S. Shibuya, S. Takamizawa, T. Hiroma, T. Nakamura and T.Kosho	Clinical courses of children with trisomy 13 receiving intensive neonatal and pediatric treatment	Am J Med Genet A	176（9）	1941-1949	2018

S. Shibuya, Y. Miyake, S. Takamizawa, E. Nishi, K. Yoshizawa, T. Hatata, K. Yoshizawa, K. Fujita, M. Noguchi, J. Ohata, T. Hiroma, T. Nakamura and T. Kosho	Safety and efficacy of noncardiac surgical procedures in the management of patients with trisomy 13: A single institution-based detailed clinical observation	Am J Med Genet A	176（5）	1137-1144	2018
M. Uehara, T. Kosho, N. Yamamoto, H. E. Takahashi, T. Shimakura, J. Nakayama, H. Kato and J. Takahashi	Spinal manifestations in 12 patients with musculocontractural Ehlers-Danlos syndrome caused by CHST14/D4ST1 deficiency (mcEDS-CHST14)	Am J Med Genet A	176（11）	2331-2341	2018
T. Yamaguchi, K. Takano, Y. Inaba, M. Morikawa, M. Motobayashi, R. Kawamura, K. Wakui, E. Nishi, S. I. Hirabayashi, Y. Fukushima, H. Kato, J. Takahashi and T. Kosho	PIEZO2 deficiency is a recognizable arthrogryposis syndrome: A new case and literature review	Am J Med Genet A	179（6）	948-957	2019
T. Hirose, N. Takahashi, P. Tangkawattana, J. Minaguchi, S. Mizumoto, S. Yamada, N. Miyake, S. Hayashi, A. Hatamochi, J. Nakayama, T. Yamaguchi, A. Hashimoto, Y. Nomura, K. Takehana, T.Kosho and T. Watanabe	Structural alteration of glycosaminoglycan side chains and spatial disorganization of collagen networks in the skin of patients with mcEDS-CHST14	Biochim Biophys Acta Gen Subj	1863（3）	623-631	2019

P. J. van der Sluijs, S. Jansen, S. A. Vergano, M. Adachi-Fukuda, Y. Alanay, A. AlKindy, A. Baban, A. Bayat, S. Beck-Wodl, K. Berry, E. K. Bijlsma, L. A. Bok, A. F. J. Brouwer, I. van der Burgt, P. M. Campeau, N. Canham, K. Chrzanowska, Y. W. Y. Chu, B. H. Y. Chung, K. Dahan, M. De Rademaeker, A. Destree, T. Dudding-Byth, R. Earl, N. Elcioglu, E. R. Elias, C. Fagerberg, A. Gardham, B. Gener, E. H. Gerkes, U. Grasshoff, A. van Haeringen, K. R. Heitink, J. C. Herkert, N. S. den Hollander, D. Horn, D. Hunt, S. G. Kant, M. Kato, H. Kayserili, R. Kersseboom, E. Kilic, M. Krajewska-Walasek, K. Lammers, L. W. Laulund, D. Lederer, M. Lees, V. Lopez-Gonzalez, S. Maas, G. M. S. Mancini, C. Marcelis, F. Martinez, I. Maystadt, M. McGuire, S. McKee, S. Mehta, K. Metcalfe, J. Milunsky, S. Mizuno, J. B. Moeschler, C. Netzer, C. W. Ockeloen, B. Oehl-Jaschkowitz, N. Okamoto, S. N. M. Olminkhof, C. Orellana, L. Pasquier, C. Pottinger, V. Riehmer, S. P. Robertson, M. Roifman, C. Rooryck, F. G. Ropers, M. Rosello, C. A. L. Ruivenkamp, M. S. Sagiroglu, S. Sallevelt, A. S. Calvo, P. O. Simsek-Kiper, G. Soares, L. Solaeche, F. M. Sonmez, M. Splitt, D. Steenbeek, A. P. A. Stegmann, C. Stumpel, S. Tanabe, E. Uctepe, G. E. Utine, H. E. Veenstra-Knol, S. Venkateswaran, C. Vilain, C. Vincent-Delorme, A. T. Vulto-van Silfhout, P. Wheeler, G. N. Wilson, L. C. Wilson, B. Wollnik, T.Kosho, D. Wieczorek, E. Eichler, R. Pfundt, B. B. A.	Correction: The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffin-Siris syndrome	Genet Med		2019	

de Vries, J. Clayton-Smith and G. W. E. Santen					
H. Morokawa, M. Kamiya, K. Wakui, M. Kobayashi, T. Kurata, K. Matsuda, R. Kawamura, H. Kanno, Y. Fukushima, Y. Nakazawa and T.Kosho	Myelodysplastic syndrome in an infant with constitutional pure duplication 1q41-qter	Hum Genome Var	5（6	2018	
N. Koitabashi, T. Yamaguchi, D. Fukui, T. Nakano, A. Umeyama, K. Toda, R. Funada, M. Ishikawa, R. Kawamura, K. Okada, A. Hatamochi, T.Kosho and M. Kurabayashi	Peripartum Iliac Arterial Aneurysm and Rupture in a Patient with Vascular Ehlers-Danlos Syndrome Diagnosed by Next-Generation Sequencing	Int Heart J	59（5）	1180-1185	2018
Y. Ogawa, K. Nakamura, N. Ezawa, T. Yamaguchi, T. Yoshinaga, D. Miyazaki, T.Kosho and Y. Sekijima	A novel CACNA1A nonsense variant in a patient presenting with paroxysmal exertion-induced dyskinesia	J Neurol Sci	399（214-216	2019	
D. Kumaki, Y. Nakamura, N. Sakai, T.Kosho, A. Nakamura, S. Hirabayashi, T. Suzuki, M. Kamimura and H. Kato T. Kawakami, H. Nakazawa, F. Kawakami, S. Matsuzawa, Y. Sudo, H. Sakai, S. Nishina, N. Senoo, Y. Senoo, M. Komatsu, T. Umemura, T. Yamaguchi, T.Kosho, T. Fujiwara, H. Harigae and F. Ishida	Efficacy of Denosumab for Glucocorticoid-Induced Osteoporosis in an Adolescent Patient with Duchenne Muscular Dystrophy: A Case Report	JBJS Case Connect	8（2）	e22	2018
T. Kawakami, H. Nakazawa, F. Kawakami, S. Matsuzawa, Y. Sudo, H. Sakai, S. Nishina, N. Senoo, Y. Senoo, M. Komatsu, T. Umemura, T. Yamaguchi, T.Kosho, T. Fujiwara, H. Harigae and F. Ishida	[Successful treatment of X-linked sideroblastic anemia with ALAS2 R452H mutation using vitamin B6]	Rinsho Ketsueki	59（4）	401-406	2018

家里明日美, 黄瀬恵美子, 石川真澄, 山本佳那, 大場崇旦, 伊藤勅子, 金井敏晴, 前野一真, 山下浩美, 高野亨子, 鹿島大靖, 菊地範彦, 宮本強, 塩沢丹里, 福嶋義光, 伊藤研一 and 古庄知己	遺伝性乳がん卵巣がん症候群における遺伝カウンセリング受診者の臨床的特徴　信州大学医学部附属病院における20年間の取り組みから	日本遺伝カウンセリング学会誌	39（1）	53-59	2018
K. Yamoto, S. Okamoto, Y. Fujisawa, M. Fukami, H. Saitsu and T.Ogata	FGFR1 disruption identified by whole genome sequencing in a male with a complex chromosomal rearrangement and hypogonadotropic hypogonadism	Am J Med Genet A	176（1）	139-143	2018
S. Ohsako, T. Aiba, M. Miyado, M. Fukami, T.Ogata, Y. Hayashi, K. Mizuno and Y. Kojima	Expression of Xenobiotic Biomarkers CYP1 Family in Preputial Tissue of Patients with Hypospadias and Phimosis and Its Association with DNA Methylation Level of SRD5A2 Minimal Promoter	Arch Environ Contam Toxicol	74（2）	240-247	2018
M. Igarashi, K. Mizuno, M. Kon, S. Narumi, Y. Kojima, Y. Hayashi, T.Ogata and M. Fukami	GATA4 mutations are uncommon in patients with 46,XY disorders of sex development without heart anomaly	Asian J Androl	20（6）	629-631	2018
H. Mano, S. Fujiwara, K. Takamura, H. Kitoh, S. Takayama, T.Ogata, S. Hashimoto and N. Haga	Congenital limb deficiency in Japan: a cross-sectional nationwide survey on its epidemiology	BMC Musculoskelet Disord	19（1）	262	2018
T. Hiraide, T.Ogata, S. Watanabe, M. Nakashima, T. Fukuda and H. Saitsu	Coexistence of a CAV3 mutation and a DMD deletion in a family with complex muscular diseases	Brain Dev	41（5）	474-479	2019
D. Shimizu, S. Iwashima, K. Sato, S. Hayano, M. Fukami, H. Saitsu and T.Ogata	GATA4 variant identified by whole-exome sequencing in a Japanese family with atrial septal defect: Implications for male sex development	Clin Case Rep	6（11）	2229-2233	2018

H. Hattori, H. Hiura, A. Kitamura, N. Miyauchi, N. Kobayashi, S. Takahashi, H. Okae, K. Kyono, M. Kagami, T.Ogata and T. Arima	Association of four imprinting disorders and ART	Clin Epigenetics	11（1）	21	2019
M. Kagami, A. Yanagisawa, M. Ota, K. Matsuoka, A. Nakamura, K. Matsubara, K. Nakabayashi, S. Takada, M. Fukami and T.Ogata	Temple syndrome in a patient with variably methylated CpGs at the primary MEG3/DLK1:IG-DMR and severely hypomethylated CpGs at the secondary MEG3:TSS-DMR	Clin Epigenetics	11（1）	42	2019
K. Matsubara, M. Itoh, K. Shimizu, S.Saito, K. Enomoto, K. Nakabayashi, K. Hata, K.Kurosawa, T.Ogata, M. Fukami and M. Kagami	Exploring the unique function of imprinting control centers in the PWS/AS-responsible region: finding from array-based methylation analysis in cases with variously sized microdeletions	Clin Epigenetics	11（1）	36	2019
M. Okuno, T. Ayabe, I. Yokota, I. Musha, K. Shiga, T. Kikuchi, N. Kikuchi, A. Ohtake, A. Nakamura, K. Nakabayashi, K. Okamura, Y. Momozawa, M. Kubo, J. Suzuki, T. Urakami, T. Kawamura, S. Amemiya, T.Ogata, S. Sugihara, M. Fukami, C. Japanese Study Group of Insulin Therapy for and D. Adolescent	Protein-altering variants of PTPN2 in childhood-onset Type 1A diabetes	Diabet Med	35（3）	376-380	2018
T. Yoshida, T. Matsuzaki, M. Miyado, K. Saito, T. Iwasa, Y.Matsubara, T.Ogata, M. Irahara and M. Fukami	11-oxygenated C19 steroids as circulating androgens in women with polycystic ovary syndrome	Endocr J	65（10）	979-990	2018

J. R. Hernandez Mora, C. Tayama, M. Sanchez-Delgado, A. Monteagudo-Sanchez, K. Hata, T.Ogata, J. Medrano, M. E. Poo-Llanillo, C. Simon, S. Moran, M. Esteller, J. Tenorio, P. Lapunzina, M. Kagami, D. Monk and K. Nakabayashi	Characterization of parent-of-origin methylation using the Illumina Infinium MethylationEPIC array platform	Epigenomics	10（7）	941-954	2018
A. Montalbano, L. Juergensen, M. Fukami, C. T. Thiel, N. H. Hauer, R. Roeth, B. Weiss, Y. Naiki, T.Ogata, D. Hassel and G. A. Rappold	Functional missense and splicing variants in the retinoic acid catabolizing enzyme CYP26C1 in idiopathic short stature	Eur J Hum Genet	26（8）	1113-1120	2018
T. Hiraide, M. Nakashima, K. Yamoto, T. Fukuda, M.Kato, H. Ikeda, Y. Sugie, K. Aoto, T. Kaname, K. Nakabayashi, T.Ogata, N. Matsumoto and H. Saitsu	De novo variants in SETD1B are associated with intellectual disability, epilepsy and autism	Hum Genet	137（1）	95-104	2018
E. Suzuki, H. Shima, M. Kagami, S. Soneda, T. Tanaka, S. Yatsuga, J. Nishioka, Y. Oto, T. Kamiya, Y. Naiki, T.Ogata, Y. Fujisawa, A. Nakamura, S. Kawashima, S. Morikawa, R. Horikawa, S. Sano and M. Fukami	(Epi)genetic defects of MKRN3 are rare in Asian patients with central precocious puberty	Hum Genome Var	6	7	2019
K. Hamanaka, A. Takata, Y. Uchiyama, S. Miyatake, N. Miyake, S. Mitsuhashi, K. Iwama, A. Fujita, E. Imagawa, A. N. Alkanaq, E. Koshimizu, Y. Azuma, M. Nakashima, T. Mizuguchi, H. Saitsu, Y. Wada, S. Minami, Y. Katoh-Fukui, Y. Masunaga, M. Fukami, T. Hasegawa, T.Ogata and N. Matsumoto	MYRF haploinsufficiency causes 46,XY and 46,XX disorders of sex development: bioinformatics consideration	Hum Mol Genet			2019

S. Nakamura, Y. Kobori, Y. Ueda, Y. Tanaka, H. Ishikawa, A. Yoshida, M. Katsumi, K. Saito, A. Nakamura, T.Ogata, H. Okada, H. Nakai, M. Miyado and M. Fukami	STX2 is a causative gene for nonobstructive azoospermia	Hum Mutat	39（6）	830-833	2018
M. Miyado, M. Fukami, S. Takada, M. Terao, K. Nakabayashi, K. Hata, Y.Matsubara, Y. Tanaka, G. Sasaki, K. Nagasaki, M. Shiina, K. Ogata, Y. Masunaga, H. Saitsu and T.Ogata	Germline-Derived Gain-of-Function Variants of Gsalpha-Coding GNAS Gene Identified in Nephrogenic Syndrome of Inappropriate Antidiuresis	J Am Soc Nephrol	30（5）	877-889	2019
M. Nakashima, J. Tohyama, E. Nakagawa, Y. Watanabe, C. G. Siew, C. S. Kwong, K. Yamoto, T. Hiraide, T. Fukuda, T. Kaname, K. Nakabayashi, K. Hata, T.Ogata, H. Saitsu and N. Matsumoto	Identification of de novo CSNK2A1 and CSNK2B variants in cases of global developmental delay with seizures	J Hum Genet	64（4）	313-322	2019
T. Inoue, H. Yagasaki, J. Nishioka, A. Nakamura, K. Matsubara, S. Narumi, K. Nakabayashi, K. Yamazawa, T. Fuke, A. Oka, T.Ogata, M. Fukami and M. Kagami	Molecular and clinical analyses of two patients with UPD(16)mat detected by screening 94 patients with Silver-Russell syndrome phenotype of unknown aetiology	J Med Genet			2018
A. Nakamura, K. Muroya, H. Ogata-Kawata, K. Nakabayashi, K. Matsubara, T.Ogata, K.Kurosawa, M. Fukami and M. Kagami	A case of paternal uniparental isodisomy for chromosome 7 associated with overgrowth	J Med Genet	55（8）	567-570	2018
H. Shima, K. Koehler, Y. Nomura, K. Sugimoto, A. Satoh, T.Ogata, M. Fukami, R. Juhlen, M. Schuelke, K. Mohnike, A. Huebner and S. Narumi	Two patients with MIRAGE syndrome lacking haematological features: role of somatic second-site reversion SAMD9 mutations	J Med Genet	55（2）	81-85	2018

N. K. Matsushita R*, Ayabe T, Kinjo S, Haruna H, Ihara K, Hasegawa T.Ogata T, Ozono K, Minamitani K, Thyroid Committee of the Japanese Society for Pediatric Endocrinology	Early Calcitonin Level-Based Thyroidectomy May Reduce Postoperative Complications and Improve Prognosis in MEN2	J Pediatr Endocrinol Metab (accepted)			
H. Ono, C. Numakura, K. Homma, T. Hasegawa, S. Tsutsumi, F. Kato, Y. Fujisawa, M. Fukami and T.Ogata	Longitudinal serum and urine steroid metabolite profiling in a 46,XY infant with prenatally identified POR deficiency	J Steroid Biochem Mol Biol	178	177-184	2018
K. Kurata, K. Hosono, A. Hikoya, A. Kato, H. Saitsu, S. Minoshima, T.Ogata and Y. Hotta	Clinical characteristics of a Japanese patient with Bardet-Biedl syndrome caused by BBS10 mutations	Jpn J Ophthalmol	62（4）	458-466	2018
H. Ono, H. Saitsu, R. Horikawa, S. Nakashima, Y. Ohkubo, K. Yanagi, K. Nakabayashi, M. Fukami, Y. Fujisawa and T.Ogata	Partial androgen insensitivity syndrome caused by a deep intronic mutation creating an alternative splice acceptor site of the AR gene	Sci Rep	8（1）	2287	2018
A. Takata, N. Miyake, Y. Tsurusaki, R. Fukai, S. Miyatake, E. Koshimizu, I. Kushima, T. Okada, M. Morikawa, Y. Uno, K. Ishizuka, K. Nakamura, M. Tsujii, T. Yoshikawa, T. Toyota, N.Okamoto, Y. Hiraki, R. Hashimoto, Y. Yasuda, S.Saitoh, K. Ohashi, Y. Sakai, S. Ohga, T. Hara, M.Kato, K. Nakamura, A. Ito, C. Seiwa, E. Shirahata, H. Osaka, A. Matsumoto, S. Takeshita, J. Tohyama, T. Saikusa, T. Matsuishi, T. Nakamura, T. Tsuboi, T. Kato, T. Suzuki, H. Saitsu, M. Nakashima, T. Mizuguchi, F. Tanaka, N. Mori, N. Ozaki and N. Matsumoto	Integrative Analyses of De Novo Mutations Provide Deeper Biological Insights into Autism Spectrum Disorder	Cell Rep	22（3）	734-747	2018

M. Nakashima, Y. Negishi, I. Hori, A. Hattori, S.Saitoh and H. Saitsu	A case of early-onset epileptic encephalopathy with a homozygous TBC1D24 variant caused by uniparental isodisomy	Am J Med Genet A	179（4）	645-649	2019
I. Hori, F. Miya, Y. Negishi, A. Hattori, N. Ando, K. A. Boroevich, N.Okamoto, M.Kato, T. Tsunoda, M. Yamasaki, Y. Kanemura, K.Kosaki and S.Saitoh	A novel homozygous missense mutation in the SH3-binding motif of STAMBP causing microcephaly-capillary malformation syndrome	J Hum Genet	63（9）	957-963	2018
T. Takenouchi, M. Inaba, T. Uehara, T. Takahashi, K.Kosaki and S.Mizuno	Biallelic mutations in NALCN: Expanding the genotypic and phenotypic spectra of IHPRF1	Am J Med Genet A	176（2）	431-437	2018
T. Takenouchi, T. Uehara, K.Kosaki and S.Mizuno	Growth pattern of Rahman syndrome	Am J Med Genet A	176（3）	712-714	2018
M. Yamada, T. Uehara, H. Suzuki, T. Takenouchi, H. Yoshihashi, H. Suzumura, S.Mizuno and K.Kosaki	SATB2-associated syndrome in patients from Japan: Linguistic profiles	Am J Med Genet A	179（6）	896-899	2019
K. Kato, S.Mizuno, M. Inaba, S. Fukumura, N. Kurahashi, K. Maruyama, D. Ieda, K. Ohashi, I. Hori, Y. Negishi, A. Hattori and S.Saitoh	Distinctive facies, macrocephaly, and developmental delay are signs of a PTEN mutation in childhood	Brain Dev	40（8）	678-684	2018
T. Kato, Y. Ouchi, H. Inagaki, Y. Makita, S.Mizuno, M. Kajita, T. Ikeda, K. Takeuchi and H. Kurahashi	Genomic Characterization of Chromosomal Insertions: Insights into the Mechanisms Underlying Chromothripsis	Cytogenet Genome Res	153（1）	1-9	2017
D. Fukushi, K. Yamada, K. Suzuki, M. Inaba, N. Nomura, Y. Suzuki, K. Katoh, S.Mizuno and N. Wakamatsu	Clinical and genetic characterization of a patient with SOX5 haploinsufficiency caused by a de novo balanced reciprocal translocation	Gene	655	65-70	2018

H. Imura, S. Suzuki, S.Mizuno, C. Sakuma and N. Natsume	A case of Tetrasomy 15q with left cleft lip and alveolus	Journal of Oral and Maxillofacial Surgery Medicine and Pathology	29（5）	427-429	2017
Y. Suzuki, Y. Enokido, K. Yamada, M. Inaba, K. Kuwata, N. Hanada, T. Morishita, S.Mizuno and N. Wakamatsu	The effect of rapamycin, NVP-BEZ235, aspirin, and metformin on PI3K/AKT/mTOR signaling pathway of PIK3CA-related overgrowth spectrum (PROS)	Oncotarget	8（28）	45470-45483	2017
N.Okamoto	Okamoto syndrome has features overlapping with Au-Kline syndrome and is caused by HNRNPK mutation	Am J Med Genet A	179（5）	822-826	2019
A. Fassio, A. Esposito, M.Kato, H. Saitsu, D. Mei, C. Marini, V. Conti, M. Nakashima, N.Okamoto, A. Olmez Turker, B. Albuz, C. N. Semerci Gunduz, K. Yanagihara, E. Belmonte, L. Maragliano, K. Ramsey, C. Balak, A. Siniard, V. Narayanan, C. R. R. Group, C. Ohba, M. Shiina, K. Ogata, N. Matsumoto, F. Benfenati and R. Guerrini	De novo mutations of the ATP6V1A gene cause developmental encephalopathy with epilepsy	Brain	141（6）	1703-1718	2018
T. Saikusa, M. Hara, K. Iwama, K. Yuge, C. Ohba, J. I. Okada, T. Hisano, Y. Yamashita, N.Okamoto, H. Saitsu, N. Matsumoto and T. Matsuishi	De novo HDAC8 mutation causes Rett-related disorder with distinctive facial features and multiple congenital anomalies	Brain Dev	40（5）	406-409	2018
K. Ueda, K. Yanagi, T. Kaname and N.Okamoto	A novel mutation in the GATAD2B gene associated with severe intellectual disability	Brain Dev	41（3）	276-279	2019
K. Yamamoto-Shimojima, N.Okamoto, W. Matsumura, T. Okazaki and T. Yamamoto	Three Japanese patients with 3p13 microdeletions involving FOXP1	Brain Dev	41（3）	257-262	2019

K. Yamamoto-Shimojima, M. Kouwaki, Y. Kawashima, K. Itomi, K. Momosaki, S. Ozasa, N.Okamoto, K. Yokochi and T. Yamamoto	Natural histories of patients with Wolf-Hirschhorn syndrome derived from variable chromosomal abnormalities	Congenit Anom (Kyoto)			2018
M. Taniguchi-Ikeda, N. Morisada, H. Inagaki, Y. Ouchi, Y. Takami, M. Tachikawa, W. Satake, K. Kobayashi, S. Tsuneishi, S. Takada, H. Yamaguchi, H. Nagase, K. Nozu, N.Okamoto, H. Nishio, T. Toda, I. Morioka, H. Wada, H. Kurahashi and K. Iijima	Two patients with PNKP mutations presenting with microcephaly, seizure, and oculomotor apraxia	Clin Genet	93（4）	931-933	2018
T. Imaizumi, Y. Mogami, N.Okamoto, K. Yamamoto-Shimojima and T. Yamamoto	De novo 1p35.2 microdeletion including PUM1 identified in a patient with sporadic west syndrome	Congenit Anom (Kyoto)		2019	

J. Kennedy, D. Goudie, E. Blair, K. Chandler, S. Joss, V. McKay, A. Green, R. Armstrong, M. Lees, B. Kamien, B. Hopper, T. Y. Tan, P. Yap, Z. Stark, N.Okamoto, N. Miyake, N. Matsumoto, E. Macnamara, J. L. Murphy, E. McCormick, H. Hakonarson, M. J. Falk, D. Li, P. Blackburn, E. Klee, D. Babovic-Vuksanovic, S. Schelley, L. Hudgins, S. Kant, B. Isidor, B. Cogne, K. Bradbury, M. Williams, C. Patel, H. Heussler, C. Duff-Farrier, P. Lakeman, I. Scurr, U. Kini, M. Elting, M. Reijnders, J. Schuurs-Hoeijmakers, M. Wafik, A. Blomhoff, C. A. L. Ruivenkamp, E. Nibbeling, A. J. M. Dingemans, E. D. Douine, S. F. Nelson, D. D. D. Study, V. A. Arboleda and R. Newbury-Ecob	KAT6A Syndrome: genotype-phenotype correlation in 76 patients with pathogenic KAT6A variants	Genet Med	21（4）	850-860	2019
Y. Mogami, Y. Suzuki, Y. Murakami, T. Ikeda, S. Kimura, K. Yanagihara, N.Okamoto and T. Kinoshita	Early infancy-onset stimulation-induced myoclonic seizures in three siblings with inherited glycosylphosphatidylinositol (GPI) anchor deficiency	Epileptic Disord	20（1）	42-50	2018
N.Okamoto, T. Kohmoto, T. Naruto, K. Masuda and I. Imoto	Primary microcephaly caused by novel compound heterozygous mutations in ASPM	Hum Genome Var	5	18015	2018
K. Shimojima, N.Okamoto, K. Ohmura, H. Nagase and T. Yamamoto	Infantile spasms related to a 5q31.2-q31.3 microdeletion including PURA	Hum Genome Var	5	18007	2018

I. Hori, F. Miya, Y. Negishi, A. Hattori, N. Ando, K. A. Boroevich, N.Okamoto, M.Kato, T. Tsunoda, M. Yamasaki, Y. Kanemura, K.Kosaki and S.Saitoh	A novel homozygous missense mutation in the SH3-binding motif of STAMBP causing microcephaly-capillary malformation syndrome	J Hum Genet	63（9）	957-963	2018
M. P. Adam, S. Banka, H. T. Bjornsson, O. Bodamer, A. E. Chudley, J. Harris, H. Kawame, B. C. Lanpher, A. W. Lindsley, G. Merla, N. Miyake, N.Okamoto, C. T. Stumpel, N. Niikawa and B. Kabuki Syndrome Medical Advisory	Kabuki syndrome: international consensus diagnostic criteria	J Med Genet	56（2）	89-95	2019
A. Inoko, T. Yano, T. Miyamoto, S.Matsuura, T. Kiyono, N. Goshima, M. Inagaki and Y. Hayashi	Albatross/FBF1 contributes to both centriole duplication and centrosome separation	Genes Cells	23（12）	1023-1042	2018
T. Miyamoto, S. N. Akutsu, H. Tauchi, Y. Kudo, S. Tashiro, T. Yamamoto and S.Matsuura	Exploration of genetic basis underlying individual differences in radiosensitivity within human populations using genome editing technology	J Radiat Res	59（suppl_2）	ii75-ii82	2018
H. Hidaka, K. Higashimoto, S. Aoki, H. Mishima, C. Hayashida, T. Maeda, Y. Koga, H. Yatsuki, K. Joh, H. Noshiro, R. Iwakiri, A. Kawaguchi, K. I. Yoshiura, K. Fujimoto and H.Soejima	Comprehensive methylation analysis of imprinting-associated differentially methylated regions in colorectal cancer	Clin Epigenetics	10（1）	150	2018
K. Joh, F. Matsuhisa, S. Kitajima, K. Nishioka, K. Higashimoto, H. Yatsuki, T. Kono, H. Koseki and H.Soejima	Growing oocyte-specific transcription-dependent de novo DNA methylation at the imprinted Zrsr1-DMR	Epigenetics Chromatin	11（1）	28	2018
T. Yamada, G. Sugiyama, K. Higashimoto, A. Nakashima, H. Nakano, T. Sumida, H.Soejima and Y. Mori	Beckwith-Wiedemann syndrome with asymmetric mosaic of paternal disomy causing hemihyperplasia	Oral Surg Oral Med Oral Pathol Oral Radiol	127（3）	e84-e88	2019

K. Nishioka, H. Miyazaki and H.Soejima	Unbiased shRNA screening, using a combination of FACS and high-throughput sequencing, enables identification of novel modifiers of Polycomb silencing	Sci Rep	8（1）	12128	2018
副島英伸	【婦人科医が注意すべき悪性腫瘍関連疾患の新知識-他科エキスパートに聞く】 Beckwith-Wiedemann症候群	産科と婦人科	85（6）	667-672	2018
T. Kikuiri, H. Mishima, H. Imura, S. Suzuki, Y. Matsuzawa, T. Nakamura, S. Fukumoto, Y. Yoshimura, S. Watanabe, A. Kinoshita, T. Yamada, M. Shindoh, Y. Sugita, H. Maeda, Y. Yawaka, T. Mikoya, N. Natsume and K. I. Yoshiura	Patients with SATB2-associated syndrome exhibiting multiple odontomas	Am J Med Genet A	176（12）	2614-2622	2018
T. Shirakawa, Y. Nakashima, S. Watanabe, S. Harada, M. Kinoshita, T. Kihara, Y. Hamasaki, S. Shishido, K. I. Yoshiura, H. Moriuchi and S. Dateki	A novel heterozygous GLI2 mutation in a patient with congenital urethral stricture and renal hypoplasia/dysplasia leading to end-stage renal failure	CEN Case Rep	7（1）	94-97	2018
K. Kiyota, K. I. Yoshiura, R. Houbara, H. Miyahara, S. Korematsu and K. Ihara	Auto-immune disorders in a child with PIK3CD variant and 22q13 deletion	Eur J Med Genet	61（10）	631-633	2018
Y. S. Hori, A. Yamada, N. Matsuda, Y. Ono, D. Starenki, N. Sosonkina, K. I. Yoshiura, N. Niikawa and T. Ohta	A Novel Association between the 27-bp Deletion and 538G>A Mutation in the ABCC11 Gene	Hum Biol	89（4）	305-307	2017
S. Sato, H. Itonaga, M. Taguchi, Y. Sawayama, D. Imanishi, H. Tsushima, T. Hata, Y. Moriuchi, H. Mishima, A. Kinoshita, K. I. Yoshiura and Y. Miyazaki	Clonal dynamics in a case of acute monoblastic leukemia that later developed myeloproliferative neoplasm	Int J Hematol	108（2）	213-217	2018

Y. Murakami, K. Miura, S. Sato, A. Higashijima, Y. Hasegawa, S. Miura, K. I. Yoshiura and H. Masuzaki	Reference values for circulating pregnancy-associated microRNAs in maternal plasma and their clinical usefulness in uncomplicated pregnancy and hypertensive disorder of pregnancy	J Obstet Gynaecol Res	44（5）	840-851	2018
T. Koga, K. Migita, T. Sato, S. Sato, M. Umeda, F. Nonaka, S. Fukui, S. Y. Kawashiri, N. Iwamoto, K. Ichinose, M. Tamai, H. Nakamura, T. Origuchi, Y. Ueki, J. Masumoto, K. Agematsu, A. Yachie, K. I. Yoshiura, K. Eguchi and A. Kawakami	MicroRNA-204-3p inhibits lipopolysaccharide-induced cytokines in familial Mediterranean fever via the phosphoinositide 3-kinase gamma pathway	Rheumatology (Oxford)	57（4）	718-726	2018
吉浦孝一郎	全ゲノム関連解析研究(GWAS)の原理と考え方	遺伝子医学	9（1）	114-121	2019
I. T. Ishijima Y, Kagami K, Masumoto S, Nakade K, Mitani Y, Niida Y, Watanabe A, Yamazaki R, Ono M, Fujiwara H	Prenatal diagnosis facilitated prompt enzyme replacement therapy for prenatal benign hypophosphatasia.	J Obstet Gynaecol	75	842-849	in press
T. Mushiroda, Y. Takahashi, T. Onuma, Y. Yamamoto, T. Kamei, T. Hoshida, K. Takeuchi, K. Otsuka, M. Okazaki, M. Watanabe, K. Kanemoto, T. Oshima, A.Watanabe, S. Minami, K. Saito, H. Tanii, Y. Shimo, M. Hara, S.Saitoh, T. Kinoshita, M.Kato, N. Yamada, N. Akamatsu, T. Fukuchi, S. Ishida, S. Yasumoto, A. Takahashi, T. Ozeki, T. Furuta, Y. Saito, N. Izumida, Y. Kano, T. Shiohara, M. Kubo and G. S. Group	Association of HLA-A*31:01 Screening With the Incidence of Carbamazepine-Induced Cutaneous Adverse Reactions in a Japanese Population	JAMA Neurol	75（7）	842-849	2018

R. Ikeue, A. Nakamura-Takahashi, Y. Nitahara-Kasahara, A.Watanabe, T. Muramatsu, T. Sato and T. Okada	Bone-Targeted Alkaline Phosphatase Treatment of Mandibular Bone and Teeth in Lethal Hypophosphatasia via an scAAV8 Vector	Mol Ther Methods Clin Dev	10	360-370	2018
渡邉淳	【最新遺伝医学研究と遺伝カウンセリング(シリーズ3) 最新 多因子遺伝性疾患研究と遺伝カウンセリング】 (第5章)多因子疾患の遺伝情報と社会　社会における遺伝リテラシー向上	遺伝子医学MOOK	別冊（最新多因子遺伝性疾患研究と遺伝カウンセリング）	283-288	2018
T. Sato, M.Kato, K.Moriyama, K. Haraguchi, H. Saitsu, N. Matsumoto and H. Moriuchi	A case of tubulinopathy presenting with porencephaly caused by a novel missense mutation in the TUBA1A gene	Brain Dev	40（9）	819-823	2018
加藤光広	新生児科医が知っておきたい脳の発生と脳形成異常	日本周産期・新生児医学会雑誌	53（2）	411	2017