

〔IV〕

研究成果の刊行に関する一覧表

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
小崎 里華	心疾患と染色体異常、単一遺伝病		新版 心臓病児者の幸せのために	一社)全国心臓病の子どもを守る会	東京	2016	70-78
森崎裕子	血管型エーラス・ダンロス症候群	桜井章洋	遺伝カウンセリングマニュアル	南江堂	東京	2016	167-168
森崎裕子	遺伝性出血性毛細血管拡張症	桜井章洋	遺伝カウンセリングマニュアル	南江堂	東京	2016	168-169
森崎裕子	遺伝性肺動脈性肺高血圧症	桜井章洋	遺伝カウンセリングマニュアル	南江堂	東京	2016	170
森崎裕子	マルファン症候群、およびその類縁疾患	桜井章洋	遺伝カウンセリングマニュアル	南江堂	東京	2016	222-224
仁科幸子	眼の発生	大鹿哲郎	眼科診療クオリファイ 23 眼科診療と関連法規	中山書店	東京	2016	2-20
仁科幸子	斜視		小児疾患診療のための病態生理 3, 改訂第5版	東京医学社	東京	2016	1035-1040
Yamamoto N, Kanno A, Matsunaga T*	Genetics of Inner Ear Malformation and Cochlear Nerve Deficiency	Kaga K	Cochlear Implantation in Children with Inner Ear Malformation and Cochlear Nerve Deficiency	Springer	Tokyo	2016	47-59
松永達雄	遺伝子診断・平衡障害	永井良三・シリーズ総監修・山岨達也、小川郁、丹生健一、久育男、森山寛、宇佐美真一	耳鼻咽喉科・頭頸部外科研修ノート改訂第2版	診断と治療社	東京	2016	622
水野誠司、中島好美	遺伝子疾患と遺伝カウンセリング	本城秀次、野邑健二、岡田 俊	臨床児童青年精神医学	西村書店	東京	2016	444
水野誠司	染色体異常・先天異常歌舞伎症候群		小児内科 特集【慢性疾患児の一生を診る】	東京医学社		2016	1394-1397
水野誠司	総論 先天異常の記述と分類		小児科診療 特集【先天異常症候群の新しい展開】	診断と治療社		2016	1711-1717
水野誠司	先天異常症候群に見られる行動発達の特徴 遺伝と行動とその理解 特集:発達障害と神経眼科	山田謙一	神経眼科 特集:発達障害と神経眼科	日本神経眼科学会		2016	222-228
岡本伸彦	岡本伸彦 遺伝カウンセリング	上野昌江、和泉京子	公衆衛生看護学 第2版	中央法規	東京	2016	399-405

小崎健次郎	先天異常症候群 成長に応じた診療のポイントと政策的支援の手引き	小崎健次郎 他	先天異常症候群 成長に応じた診療のポイントと政策的支援の手引き	鳥影社	東京	2017	1-339
Kobayashi Y, Duarte C, <u>Moriyama K.</u>	Hormone Relaxin as Biomarkers for Bone Health and Disease.	Victor R. Preedy	Biomarkers in Bone Disease, Biomarkers in Disease: Methods, Discoveries and Applications.	Springer	Dordrecht	2017	329-353
水野誠司	染色体異常症		医薬ジャーナル 特集【移行期医療～小児期から成人期への円滑な橋渡しを目指して～】	医薬ジャーナル社		2017	83-88

雑誌

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
Matsubara K, Murakami N, Fukami M, Kagami M, Nagai T, <u>Ogata T</u>	Risk assessment of medically assisted reproduction and advanced maternal ages in the development of Prader-Willi syndrome due to UPD(15)mat.	Clin Genet	89 (5)	614-619	2015
Asahina M, Endoh Y, Matsubayashi T, Fukuda T, <u>Ogata T</u>	Novel RAB3GAP1 compound heterozygous mutations in Japanese siblings with Warburg Micro syndrome.	Brain Dev	38 (3)	337-340	2016
Asahina M, Endoh Y, Matsubayashi T, Hirano K, Fukuda T, <u>Ogata T</u>	Genomewide array comparative genomic hybridization in 55 Japanese normokaryotypic patients with non-syndromic intellectual disability.	J Pediatr Neurol Disord	2(1)	108	2016
Ayabe T, Fukami M, <u>Ogata T</u> , Kawamura T, Urakami T, Kikuchi N, Yokota I, Ihara K, Takemoto K, Mukai T, Nishii A, Kikuchi T, Mori T, Shimura N, Sasaki G, Kizu R, Takubo N, Soneda S, Fujisawa T, Takaya R, Kizaki Z, Kanzaki S, Hanaki K, <u>Matsuura N</u> , Kasahara Y, <u>Kosaka K</u> , Takahashi T, Minamitani K, Matsuo S, Mochizuki H, Kobayashi K, Koike A, Horikawa R, Teno S, Tsubouchi K, Mochizuki T, Igarashi Y, Amemiya S, Sugihara S; Japanese Study Group of Insulin Therapy for Childhood and Adolescent Diabetes (JSGIT). The Japanese	Variants associated with autoimmune type 1 diabetes in Japanese children: implications for age-specific effects of cis-regulatory haplotypes at 17q12-q21.	Diabet Med	33 (12)	1717-1722	2016

Study Group of Insulin Therapy for Childhood and Adolescent Diabetes (JSGIT)					
Dateki S, Watanabe S, Nakatomi A, Kinoshita E, Matsumoto T, <u>Yoshiura K</u> , Moriuchi H.	Genetic background of hyperphenylalaninemia in Nagasaki, Japan.	Pediatr Int	58(5)	431-433	2016
Eggermann T, Brioude F, Russo S, Lombardi MP, Blied J, Maher ER, Larizza L, Prawitt D, Netchine I, Gonzales M, Grønskov K, Tümer Z, Monk D, Mannens M, Chrzanowska K, Walasek MK, Begemann M, Soellner L, Eggermann K, Tenorio J, Nevado J, Moore GE, Mackay DJ, Temple K, Gillessen-Kaesbach G, <u>Ogata T</u> , Weksberg R, Algar E, Lapunzina P	Prenatal molecular testing for Beckwith-Wiedemann and Silver-Russell syndromes: a challenge for molecular analysis and genetic counseling.	Eur J Hum Genet	24(6)	784-793	2016
Negishi Y, Miya F, Hattori A, Johmura Y, Nakagawa M, Ando N, Hori I, Togawa T, Aoyama K, Ohashi K, Fukumura S, <u>Mizuno S</u> , Umemura A, Kishimoto Y, <u>Okamoto N</u> , Kato M, Tsunoda T, Yamasaki M, Kanemura Y, Kosaki K, Nakanishi M, <u>Saitoh S</u> .	A combination of genetic and biochemical analyses for the diagnosis of PI3K-AKT-mTOR pathway-associated megalencephaly.	BMC Med Genet.	13	4	2016
Fujisawa Y, Fukami M, Hasegawa T, Uematsu A, Muroya M, <u>Ogata T</u>	Long-term clinical course in three patients with MAMLD1 mutations.	Endocr J	63(9)	835-839	2016
Fujisawa Y, Sakaguchi K, Ono H, Yamaguchi R, Kato F, Kagami M, Fukami M, <u>Ogata T</u>	Combined steroidogenic characters of fetal adrenal and Leydig cells in childhood adrenocortical carcinoma.	J Steroid Biochem Mol Biol	159	86-93	2016
Fujita A, Isidor B, Piloquet H, Corre P, <u>Okamoto N</u> , Nakashima M, Tsurusaki Y, Saitsu H, Miyake N, Matsumoto N.	De novo MEIS2 mutation causes syndromic developmental delay with persistent gastro-esophageal reflux.	J Hum Genet.	61	835-8.	2016
Fukami M, Shima H, Suzuki E, <u>Ogata T</u> , Matsubara K, Kamimaki T.	Catastrophic Cellular Events Leading to Complex Chromosomal Rearrangements in the Germline.	Clin Genet (accepted)	91(5)	653-660	2016
Fukami M, Suzuki E, Shima H, Toki M, Hanew K, Matsubara K, Kurahashi H, Narumi S, <u>Ogata T</u> , Kamimaki T	Complex X-chromosomal rearrangements in two women with ovarian dysfunction: implications for chromothripsis/chromoanaphase-dependent and -independent origins of complex genomic alterations.	Cytogenet Genome Res	150(2)	86-92	2016

Fukami M, Seki A, <u>Ogata T</u>	SHOX haploinsufficiency as a cause of syndromic and non-syndromic short stature.	Mol Syndromol	7(1)	3-11	2016
Hirai M, <u>Muramatsu Y</u> , <u>Mizuno S</u> , Kurahashi N, Kurahashi H, Nakamura M.	Typical visual search performance and atypical gaze behaviors in response to faces in Williams syndrome.	Journal of Neurodevelopmental Disorders	24	38	2016
Hori I, <u>N Okamoto</u> , <u>K Kosaki</u> , <u>Saitoh S</u> et al.	Novel splicing mutation in the ASXL3 gene causing Bainbridge-Ropers syndrome.	Am J Med Genet A	170	1863-7	2016
Ishiwata T, Tanabe N, Shigeta A, Yokota H, Tsushima K, Terada J, Sakao S, <u>Morisaki H</u> , <u>Morisaki T</u> , <u>Tatsumi K</u>	Moyamoya disease and artery tortuosity as rare phenotypes in a patient with an elastin mutation.	Am J Med Genet	A 170	1924-1927	2016
Isojima T, Sakazume S, Haegawa T, <u>Ogata T</u> , Nakanishi T, Nagai T, Yokoya S	Growth references for Japanese individuals with Noonan syndrome.	Pediatr Res	79(4)	543-548	2016
Ito Y, Maehara K, Kaneki E, Matsuoka K, Sugahara N, Miyata T, Kamura H, Yamaguchi Y, Kono A, Nakabayashi K, Migita O, Higashimoto K, <u>Soejima H</u> , <u>Okamoto A</u> , <u>Nakamura H</u> , <u>Kimura T</u> , <u>Wake N</u> , <u>Taniguchi T</u> , <u>Hata K</u> .	Novel Nonsense Mutation in the NLRP7 Gene Associated with Recurrent Hydatidiform Mole.	Gynecol Obstet Invest	81(4)	353-358	2016
Kagami M, Matsubara K, Nakabayashi K, Nakamura A, Sano S, Okamura K, Hata K, Fukami M, <u>Ogata T</u>	Genomewide multilocus imprinting disturbance analysis in Temple syndrome and Kagami-Ogata syndrome.	Genet Med	19(4)	476-482	2016
Kanno A, Mutai M, Namba K, Morita N, Nakano A, Ogahara N, Sugiuchi T, Ogawa K, <u>Matsunaga T*</u>	Frequency and Specific Characteristics of the Incomplete Partition Type III Anomaly in Children	Laryngoscope	doi: 10.1002/lary.26245.	Epub ahead of print	2016
Katagiri S, Yokoi T, Mikami M, <u>Nishina S</u> , Azuma N.	Outer retinal deformity detected by optical coherence tomography in eyes with foveal hypoplasia.	Graefes Arch Clin Exp Ophthalmol.	254(11)	2197-2201	2016
Katagiri S, Yokoi T, <u>Nishina S</u> , Azuma N.	Structure and morphology of radial retinal folds with familial exudative vitreoretinopathy.	Ophthalmology	123(3)	666-668	2016
Koga T, Migita K, Sato S, Umeda M, Nonaka F, Kawashiri SY, Iwamoto N, Ichinose K, Tamai M, Nakamura H, Origuchi T, Ueki Y, Masumoto J, Agematsu K, Yachie A, <u>Yoshiura K</u> , <u>Eguchi K</u> , <u>Kawakami A</u> .	Multiple Serum Cytokine Profiling to Identify Combinational Diagnostic Biomarkers in Attacks of Familial Mediterranean Fever.	Medicine (Baltimore)	95(16)	e3449	2016
Kon M, Saio K, Mitsui T, Miyado M, Igarashi M, Moriya K, Nonomura K,	Copy-number variations of the azoospermia factor region or SRY are not associated with the risk of	Sex Dev	10(1)	12-15	2016

Shinohara M, <u>Ogata T</u> , Fukami M	hypospadias.				
Kono M (corresponding author), Hasegawa-Murakami Y, Sugiura K, Ono M, Toriyama K, Miyake N, Hatamochi A, Kamei Y, <u>Kosho T</u> (corresponding author), Akiyama M	A 45-year-old Woman with Ehlers-Danlos Syndrome Caused by Dermatan 4-O-sulfotransferase-1 Deficiency: Implications for Early Ageing.	Acta Derm Venereol	96(6)	830-1	2016
Konomoto T, Imamura H, Orita M, Tanaka E, Moritake H, Sato Y, Fujimoto S, Harita Y, Hisano S, <u>Yoshiura KI</u> , Nunoi H.	Clinical and histological findings of autosomal dominant renal-limited disease with LMX1B mutation.	Nephrology (Carlton).	21(9)	765-773	2016
Koyama Y, Homma K, Fukami M, Miwa M, Ikeda K, <u>Ogata T</u> , Murata M, Hasegawa T	Classic and non-classic 21-hydroxylase deficiency can be discriminated from P450 oxidoreductase deficiency in Japanese infants by urinary steroid metabolites.	Clin Pediatr Endocrinol	25(2)	37-44	2016
Luk H-M, Lo F-M I, Sano S, Matsbara K, Nakamura A, <u>Ogata T</u> , Kagami M	Silver-Russell syndrome in a patient with somatic mosaicism for upd(11)mat identified by buccal cell analysis.	Am J Med Genet A	170(7)	1938-1941	2016
Marchini A, <u>Ogata T</u> , Rappold GA	A track record on SHOX: from basic research to complex models and therapy.	Endocr Rev	37(4)	417-448	2016
Migita K, Izumi Y, Jiuchi Y, Iwanaga N, Kawahara C, Agematsu K, Yachie A, Masumoto J, Fujikawa K, Yamasaki S, Nakamura T, Ubara Y, Koga T, Nakashima Y, Shimizu T, Umeda M, Nonaka F, Yasunami M, Eguchi K, <u>Yoshiura K</u> , Kawakami A.	Familial Mediterranean fever is no longer a rare disease in Japan.	Arthritis Res Ther	18	175	2016
Miura K, Mishima H, Yasunami M, Kaneuchi M, Kitajima M, Abe S, Higashijima A, Fuchi N, Miura S, <u>Yoshiura KI</u> , Masuzaki H.	A significant association between rs8067378 at 17q12 and invasive cervical cancer originally identified by a genome-wide association study in Han Chinese is replicated in a Japanese population. doi:10.1038/jhg.2016.50.	J Hum Genet	61(9)	793-796	2016
Watanabe S, Shimizu K, Ohashi H, <u>Kosaki R</u> , Okamoto N, Shimojima K, Yamamoto T, Chinen Y, Mizuno S, Dowa Y, Shiomi N, Toda Y, Tashiro K, Shichijo K, Minatozaki K, Aso S, Minagawa K, Hiraki Y, Shimokawa O, Matsumoto T, Fukuda M,	Detailed analysis of 26 cases of 1q partial duplication/triplication syndrome.	Am J Med Genet A.	170(4)	908-17	2016

Moriuchi H, Yoshiura K, Kondoh T.					
Ishimaru D, Gotoh M, Takayama S, <u>Kosaki R</u> , Matsumoto Y, Narimatsu H, Sato T, Kimata K, Akiyama H, Shimizu K, Matsumoto K.	Large-scale mutational analysis in the EXT1 and EXT2 genes for Japanese patients with multiple osteochondromas.	BMC Genet.	17(1)	52	2016
Miyado M, Inui M, Igarashi M, Katoh-Fukui Y, Takasawa K, Hakoda A, Kanno J, Kashimada K, Miyado K, Tamano M, <u>Ogata T</u> , Takada S, Fukami M	The p.R92W variant of NR5A1/Nr5a1 induces testicular development of 46,XX gonads in humans, but not in mice: Phenotypic comparison of human patients and mutation-induced mice.	Biol Sex Differ	56(7)	eCollection 10.1186/s13293-016-0114-6	2016
Miyake N, Abdel-Salam G, Yamagata T, Eid MM, Osaka H, <u>Okamoto N</u> , Mohamed AM, Ikeda T, Afifi HH, Piard J, van Maldergem L, Mizuguchi T, Miyatake S, Tsurusaki Y, Matsumoto N.	Clinical features of SMARCA2 duplication overlap with Coffin-Siris syndrome.	Am J Med Genet A.	170A	2662-70	2016
Miyake N, Fukai R, Ohba C, Chihara T, Miura M, Shimizu H, Kakita A, Imagawa E, Shiina M, Ogata K, Okuno-Yuguchi J, Fueki N, Ogiso Y, Suzumura H, Watabe Y, Imataka G, Leong HY, Fattal-Valevski A, Kramer U, Miyatake S, Kato M, <u>Okamoto N</u> , Sato Y, Mitsuhashi S, Nishino I, Kaneko N, Nishiyama A, Tamura T, Mizuguchi T, Nakashima M, Tanaka F, Saitsu H, Matsumoto N	Biallelic TBCD Mutations Cause Early-Onset Neurodegenerative Encephalopathy	Am J Hum Genet	99	950-961	2016
Miyake N, Tsurusaki Y, Koshimizu E, <u>Okamoto N</u> , <u>Kosho T</u> , Jane Brown N, Yang Tan T, Jia Jiunn Yap P, Suzumura H, Tanaka T, Nagai T, Nakashima M, Saitsu H, Niikawa N, Matsumoto N.	Delineation of clinical features in Wiedemann-Steiner syndrome caused by KMT2A mutations	Clin Genet	89	115-9	2016
Miyamichi D, Asahina M, Nakajima J, Sato M, Hosono K, Nomura T, Negishi T, Miyake N, Hotta Y, <u>Ogata T</u> , Matsumoto N	Novel HPS6 mutations identified by whole-exome sequencing in two Japanese sisters with suspected ocular albinism.	J Hum Genet	61(9)	839-842	2016
Miyoshi Y, Yorifuji T, Horikawa R, Takahashi I, Nagasaki K, Ishiguro H, Fujiwara I, Ito J, Oba M,	Gonadal function, fertility, and reproductive medicine in childhood and adolescent cancer patients: a national survey of Japanese pediatric	Clin Pediatr Endocrinol	25(2)	45-57	2016

Kawamoto H, Fujisaki H, Kato M, Shimizu C, Kato T, Matsumoto K, Sago H, Takimoto T, Okada H, Suzuki N, Yokoya S, <u>Ogata T</u> , Ozono K	endocrinologists.				
Mochida K, Amano M (corresponding author), Miyake N, Matsumoto N, Hatamochi A, <u>Kosho T</u> (corresponding author)	Dermatan 4-O-sulfotransferase 1-deficient Ehlers-Danlos syndrome complicated by a large subcutaneous hematoma on the back.	J Dermatol	43(7)	832-3	2016
Montalbano A Juergensen A, Roeth R, Weiss B, Fukami M, Fricke-Otto S, Binder G, <u>Ogata T</u> , Decker E, Nuernberg G, Hassel 2, Rappold GA	Retinoic acid catabolizing enzyme CYP26C1 is a genetic modifier in SHOX deficiency.	EMBO Mol Med	8 (12)	1455-1469	2016
Morisaki T, <u>Morisaki H</u>	Genetics of hereditary large vessel diseases	J Hum Genet	61	21-26	2016
Moritani M, Yokota I, Horikawa R, Urakami T, Nishii A, Kawamura T, Kikuchi N, Kikuchi T, <u>Ogata T</u> , Sugihara S, Amemiya S; Japanese Study Group of Insulin Therapy for Childhood and Adolescent Diabetes (JSGIT)	Identification of monogenic gene mutations in Japanese subjects diagnosed with type 1B diabetes between >5 and 15.1 years of age.	J Pediatr Endocrinol Metab	229(9)	1047-1054	2016
Muramatsu Y, Tokita Y, <u>Mizuno S</u> , Nakamura M.	Disparities in visuo-spatial constructive abilities in Williams syndrome patients with typical deletion on chromosome 7q11.23.	Brain and Development	39	145	2016
Mussazhanova Z, Akazawa Y, Matsuda K, Shichijo K, Miura S, Otsubo R, Oikawa M, <u>Yoshiura KI</u> , Mitsutake N, Rogounovitch T, Saenko V, Kozykenova Z, Zhetpisbaev B, Shabdarbaeva D, Sayakenov N, Amantayev B, Kondo H, Ito M, Nakashima M.	Association between p53-binding protein 1 expression and genomic instability in oncocyctic follicular adenoma of the thyroid.	Endocr J	63(5)	457-467	2016
Naiki Y, Miyado M, Horikawa R, Katsumata N, Onodera M, Pang S, <u>Ogata T</u> , Fukami M	Extra-Adrenal Induction of Cyp21a1 Ameliorates Systemic Steroid Metabolism in a Mouse Model of Congenital Adrenal Hyperplasia.	Endocr J	63 (10)	897-904	2016
Nakamura A, Hamaguchi E, Horikawa R, Nishimura Y, Matsubara K, Sano S, Nagasaki K, <u>Matsubara Y</u> , Umezawa A, Tajima T, <u>Ogata T</u> , Kagami M, Okamura K, Fukami M	Complex genomic rearrangement within the GNAS region associated with familial pseudohypoparathyroidism Ttype 1b.	J Clin Endocrinol Metab	101 (7)	2623-2627	2016

Nakayama Y, Katagiri S, Yokoi T, Ui M, <u>Nishina S</u> , Azuma N.	Successful scleral buckling of late-onset visual decrease in eye with retinal folds.	Doc Ophthalmol.	133 (2)	145-149	2016
Namba K, Mutai H, Takiguchi Y, Yagi H, Okuyama T, Oba S, Yamagishi R, Kaneko H, Shintani T, Kaga K, <u>Matsunaga T*</u>	Molecular impairment mechanisms of novel OPA1 mutations predicted by molecular modeling in patients with autosomal dominant optic atrophy and auditory neuropathy spectrum disorder.	Otol Neurotol	37(4)	394-402	2016
Nikitski A, Rogounovitch T, Bychkov A, Takahashi M, <u>Yoshiura K</u> , Mitsutake N, Kawaguchi T, Matsuse M, Drozd VM, Demidchik YE, Nishihara E, Hirokawa M, Miyauchi A, Rubanovich AV, Matsuda F, Yamashita S, Saenko VA.	Genotype analyses in the Japanese and Belarusian populations reveal independent effects of rs965513 and rs1867277 but do not support the role of FOXE1 polyalanine tract length in conferring risk for papillary thyroid carcinoma.	Thyroid	27(2)	224-235.	2016
Oda H, Sato T, Kunishima S, Nakagawa K, Izawa K, Hiejima E, Kawai T, Yasumi T, Doi H, Katamura K, <u>Numabe H</u> , Okamoto S, Nakase H, Hijikata A, Ohara O, Suzuki H, <u>Morisaki H</u> , Morisaki T, Nuno H, Hattori S, Nishikomori R, Heike T	Exon skipping causes atypical phenotypes associated with a loss-of-function mutation in FLNA by restoring its protein function.	Eur J Hum Genet	24	408-414	2016
<u>Ogata T</u> , Kagami M	Kagami-Ogata syndrome: a clinically recognizable upd(14)pat and related disorder affecting the chromosome 14q32.2 imprinted region.	J Hum Genet	61 (2)	87-94	2016
Ogura K, Iimura T, Makino Y, Sugie-Oya A, Takakura A, Takao-Kawabata R, Ishizuya T, <u>Moriyama K</u> , Yamaguchi A.	Short-term intermittent administration of parathyroid hormone facilitates osteogenesis by different mechanisms in cancellous and cortical bone.	Bone Reports	16(5)	7-14	2016
Ohtsuka Y, Higashimoto K, Oka T, Yatsuki H, Jozaki K, Maeda T Kawahara K, Hamasaki Y, Matsuo M, Nishioka K, Joh K, Mukai T, <u>Soejima H</u> .	Identification of consensus motifs associated with mitotic recombination and clinical characteristics in patients with paternal uniparental isodisomy of chromosome 11.	Hum Mol Genet	25(7)	1406-1419	2016
<u>Okamoto N</u> , Toribe Y, Shimojima K, Yamamoto T.	Tatton-Brown-Rahman syndrome due to 2p23 microdeletion.	Am J Med Genet A.	170A	1339-42.	2016
Okuno M, Kasahara Y, Onodera M, Takubo N, Okajima M, Suga S, Watanabe N, Suzuki J, Ayabe T, Urakami T, Kawamura T, Kikuchi N,	Nucleotide substitutions in CD101, the human homolog of a diabetes susceptibility gene in non-obese diabetic mouse, in patients with type 1 diabetes.	J Diabetes Investig	8(3)	286-294	2016

Yokota I, Kikuchi T, Amemiya S, Nakabayashi K, Hayashi KK, Hata KK, <u>Matsubara Y</u> , <u>Ogata T</u> , Fukami M, Sugihara S					
Okuno M, Yorifuji T, Kagami M, Ayabe T, Urakami T, Kawamura T, Kikuchi N, Yokota I, Toru Kikuchi, Amemiya S, Suzuki J, <u>Ogata T</u> , Sugihara S, Fukami M and The Japanese Study Group of Insulin Therapy for Childhood and Adolescent Diabetes (JSGIT)	Chromosome 6q24 methylation defects are uncommon in childhood-onset non-autoimmune diabetes mellitus patients born appropriate-for-gestational age.	Clin Pediatr Endocrinol	25 (3)	99-102	2016
Onda Y, Sugihara S, <u>Ogata T</u> , Yokoya S, Yokoyama T, Tajima N; Type 1 Diabetes (T1D) Study Group	Incidence and prevalence of childhood-onset type 1 diabetes in Japan: The T1D Study.	Diabet Med	doi: 10.1111/dme.13295.	Epub ahead of print	2016
Rumbajan JM, Yamaguchi Y, Nakabayashi K, Higashimoto K, Yastuki H, Nishioka K, Matsuoka K, Aoki S, Toda S, Takeda S, Seki H, Hatada I, Hata K, <u>Soejima H</u> , Joh K.	The HUS1B promoter is hypomethylated in the placentas of low-birth-weight infants.	Gene	583(2)	141-146	2016
Saito K, Matsuzaki T, Iwasa T, Miyado M, Saito H, Hasegawa T, Homma K, Inoue E, Kubota T, Irahara M, <u>Ogata T</u> , Fukami M	Steroidogenic pathways involved in androgen biosynthesis in eumenorrhic women and patients with polycystic ovary syndrome.	J Steroid Biochem Mol Biol	158	31-37	2016
Saito K, Matsuzaki T, Iwasa T, Miyado M, Saito H, Kubota T, Irahara M, <u>Ogata T</u> , Fukami M	Blood allopregnanolone levels in women with polycystic ovary syndrome.	Clin Endocrinol	85	151-152	2016
Saito H <u>Saitoh S</u> et al.	Impaired neuronal KCC2 function by biallelic SLC12A5 mutations in migrating focal seizures and severe developmental delay.	Sci Rep	6	30072	2016
Sangu N, <u>Okamoto N</u> , Shimojima K, Ondo Y, Nishikawa M, Yamamoto T.	A de novo microdeletion in a patient with inner ear abnormalities suggests that the 10q26.13 region contains the responsible gene.	Hum Genome Var	3	16008	2016
Sano S, Nagasaki K, Kikuchi T, Nakabayashi K, Hata K, Fukami M, Kagami M, <u>Ogata T</u>	Beckwith-Wiedemann syndrome and pseudohypoparathyroidism type Ib in a patient with multilocus methylation defects: a female-dominant phenomenon?	J Hum Genet	61 (8)	765-769	2016
Sarafino M, Trivedi RH, Levin AV, Wilson ME, Nucci P, Lambert SR, Nischal KK, Plager DA, Bremond-Gignac D, Kekunnaya R, <u>Nishina S</u> , Tehrani NN, Ventura MC.	Use of the Delphi process in paediatric cataract management.	Br J Ophthalmol	100(5)	611-615	2016

Sato C, Ogawa T, Tsuge R, Shiga M, Tsuji M, Baba Y, Kosaki K, <u>Moriyama K.</u>	Systemic and maxillofacial characteristics of 11 Japanese children with Russell-Silver syndrome	Congenit Anom (Kyoto)	56(5)	217-225	2016
Sato M, Baba Y, Haruyama N, Higashihori N, Tsuji M, Suzuki S, <u>Moriyama K.</u>	Clinicostatistical analysis of congenitally missing permanent teeth in Japanese patients with cleft lip and/or palate	Orthodontic Waves.	75(2)	41-45	2016
Shima H, Tanaka T, Kamimaki T, Dateki S, Muroya K, Horikawa R, Kanno J, Adachi M, Naiki Y, Tanaka H, Mabe H, Yagasaki H, Kure S, <u>Matsubara Y</u> , Tajima T, Kashimada K, Ishii T, Asakura Y, Fujiwara I, Soneda S, Nagasaki K, Hamajima T, Kanzaki S, Jinno T, <u>Ogata T</u> , Fukami M	Japanese SHOX study group: Systematic molecular analyses of SHOX in Japanese patients with idiopathic short stature and Leri-Weill dyschondrosteosis.	J Hum Genet	61 (2)	585-591	2016
Shima H, Yatsuga S, Nakamura A, Sano S, Sasaki T, Katsumata N, Suzuki E, Hata K, Nakabayashi K, Momozawa Y, Kubo M, Okamura K, Kure S, <u>Matsubara Y</u> , <u>Ogata T</u> , Narumi S, Fukami M	NR0B1 frameshift mutation in a boy with idiopathic central precocious puberty.	Sex Dev	10 (4)	205-209	2016
Shimada K, Yanagisawa R, Kubota N, Hidaka E, Sakashita K, Ishii E, <u>Matsuura S</u> , <u>Ogiso Y</u>	Wilms tumor accompanied by premature chromatid separation	Pediatr Blood Cancer	63	e26255	2016
Shimojima K, Ondo Y, Nishi E, <u>Mizuno S</u> , Ito M, Ioi A, Shimizu M, Sato M, Inoue M, <u>Okamoto N</u> , Yamamoto T	Loss-of-function mutations and global rearrangements in GPC3 in patients with Simpson-Golabi-Behmel syndrome	Hum Genome Var	3	16033	2016
Suzuki T, Miyake N, Tsurusaki Y, <u>Okamoto N</u> , Alkindy A, Inaba A, Sato M, Ito S, Muramatsu K, Kimura S, Ieda D, <u>Saitoh S</u> , Hiyane M, Suzumura H, Yagy K, Shiraishi H, Nakajima M, Fueki N, Habata Y, Ueda Y, Komatsu Y, Yan K, Shimoda K, Shitara Y, <u>Mizuno S</u> , Ichinomiya K, Sameshima K, Tsuyusaki Y, <u>Kurosawa K</u> , Sakai Y, Haginoya K, Kobayashi Y, Yoshizawa C, Hisano M, Nakashima M, Saitsu H, Takeda S, Matsumoto N.	Molecular genetic analysis of 30 families with Joubert syndrome.	Clin Genet.	90	526-535	2016

Takano K*, Ogasawara N, Matsunaga T, Mutai H, Sakurai A, Ishikawa A, Himi T	A novel nonsense mutation in the NOG gene causes familial NOG-related symphalangism spectrum disorder	Hum Genome Variation	3	16023	2016
Takenouchi T, Miura K, Uehara T, Mizuno S, Kosaki K.	Establishing SON in 21q22.11 as a cause a new syndromic form of intellectual disability: Possible contribution to Braddock-Carey syndrome phenotype.	American Journal of Medical Genetics	170	2587	2016
Takenouchi T, Okamoto N, Ida S, Uehara T, Kosaki K.	Further evidence of a mutation in CDC42 as a cause of a recognizable syndromic form of thrombocytopenia.	Am J Med Genet A.	170A	852-5	2016
Tsutsumi M, Yokoi S, Miya F, Miyata M, Kato M, Okamoto N, Tsunoda T, Yamasaki M, Kanemura Y, Kosaki K, Saitoh S, Kurahashi H.	Novel compound heterozygous variants in PLK4 identified in a patient with autosomal recessive microcephaly and chorioretinopathy.	Eur J Hum Genet	24	1702-1706	2016
Tumurkhuu T, Fujiwara T, Komazaki Y, Kawaguchi Y, Tanaka T, Inazawa J, Ganburged G, Bazar A, Ogawa T, Moriyama K.	Association between maternal education and malocclusion in Mongolian adolescents: a cross-sectional study.	BMJ Open	1;6(11)	e012283	2016
Uchiyama Y, Nakashima M, Watanabe S, Miyajima M, Taguri M, Miyatake S, Miyake N, Saito H, Mishima H, Kinoshita A, Arai H, Yoshiura K, Matsumoto N.	Ultra-sensitive droplet digital PCR for detecting a low-prevalence somatic GNAQ mutation in Sturge-Weber syndrome.	Sci Rep	6	22985	2016
Uehara DT, Okamoto N, Kosaki R, Kurosawa K, Saith S, et al.	SNP array screening of cryptic genomic imbalances in 450 Japanese subjects with intellectual disability and multiple congenital anomalies previously negative for large rearrangements.	J Hum Genet	61	335-43	2016
Uemura R, Tachibana D, Kurihara Y, Pooh RK, Aoki Y, Koyama M.	Prenatal findings of hypertrophic cardiomyopathy in a severe case of Costello syndrome.	Ultrasound Obstet Gynecol	48(6)	799-800	2016
Wada H, Matsuda K, Akazawa Y, Yamaguchi Y, Miura S, Ueki N, Kinoshita A, Yoshiura K, Kondo H, Ito M, Nagayasu T, Nakashima M.	Expression of Somatostatin Receptor Type 2A and PTEN in Neuroendocrine Neoplasms Is Associated with Tumor Grade but Not with Site of Origin.	Endocr Pathol	27(3)	179-187	2016
Watanabe S, Shimizu K, Ohashi H, Kosaki R, Okamoto N, Shimojima K, Yamamoto T, Chinen Y, Mizuno S, Dowa Y, Shiomi N, Toda Y, Tashiro K, Shichijo K, Minatozaki K, Aso S, Minagawa K, Hiraki Y, Shimokawa O, Matsumoto T, Fukuda M,	Detailed analysis of 26 cases of 1q partial duplication/triplication syndrome.	Am J Med Genet A	170(4)	908-917	2016

Moriuchi H, <u>Yoshiura K</u> , <u>Kondoh T</u> .					
Yaguchi Y, Katagiri S, Fukushima Y, Yokoi T, <u>Nishina S</u> , Kondo M, Azuma N.	Electroretinographic effects of retinal dragging and retinal folds in eyes with familial exudative vitreoretinopathy.	Sci Rep.	doi: 10.1038/ srep3052 3.		2016
Yaoita M, Niihori T, <u>Mizuno S</u> , Okamoto N, Hayashi S, Watanabe A, Yokozawa M, Suzumura H, Nakahara A, Nakano Y, Hokosaki T, Ohmori A, Sawada H, Migita O, Mima A, Lapunzina P, Santos F, Garcia S, <u>Ogata</u> <u>T</u> , Kawame H, <u>Kurosawa</u> <u>K</u> , <u>Ohashi H</u> , Inoue S, <u>Matsubara Y</u> , Kure S, <u>Aoki Y</u>	Spectrum of mutations and genotype-phenotype analysis in Noonan syndrome patients with RIT1 mutations.	Hum Genet	135 (2)	209-222	2016
Yokoi T, Nakayama Y, <u>Nishina S</u> , Azuma N.	Abnormal traction of the vitreous detected by swept-source optical coherence tomography is related to the maculopathy associated with optic disc pits.	Graefes Arch Clin Exp Ophthalmol.	254	675-682	2016
Yokoi T, <u>Nishina S</u> , Fukami M, <u>Ogata T</u> , Hosono K, Hotta Y, Azuma N.	Genotype-Phenotype Correlation of the PAX6 Gene Mutations in Aniridia.	Human Genome Variation	3	15052	2016
伊藤里美・仁科幸子	小児ロービジョンケア	眼科	58 (12)	1487-1492	2016
永井遼斗、松永達雄*	図説シリーズ「目で見る遺伝医学」 ー難聴の遺伝医学	国立医療学会 誌「医療」	88(3)	240-247	2016
小崎里華	Rubinstein-Taybi 症候群	小児内科	48	1386-89	2016
岡本伸彦	日本が貢献した先天異常症候群 Coffin-Siris 症候群	小児科診療	79	1807-1812	2016
岡本伸彦	結節性硬化症	小児内科	43	1520-1523	2016
岡本伸彦	Sener 症候群	小児科診療	79	128	2016
岡本伸彦	染色体異常・先天奇形症候群 Smith-Lemli-Opitz 症候群	小児科診療	79	6	2016
吉田 朋世、仁科 幸子、 萬東 恭子、赤池 祥子、 越後貫 滋子、横井 匡、 東 範行	乳児内斜視早期手術後の両眼視 機能.	眼臨紀	10 (1)	58-63	2016
古庄知己	エーラスダンロス症候群の特徴と治 療の現状.	新薬と臨牀「指 定難病最前 線」	65(11)	124-130	2016
古庄知己	Ehlers-Danlos 症候群 Kosho type.	小児科診療 「日本人が貢 献した先天異 常症候群」	79(12)	1761-1769	2016
森崎裕子	マルファン症候群、ロイス・ディーツ 症候群	日本小児科学 会雑誌	120	1579-1586	2016
仁科 幸子	内斜視に対する手術治療.	眼科	58 (3)	251-257	2016
仁科 幸子	乳児期の斜視と両眼視機能	東京小児科医	35 (1)	61-66	2016

		会報			
Aoki R, Srivatanakul K, Osada T, Hotta K, Sorimachi T, Matsumae M, <u>Morisaki H</u>	Endovascular treatment of a dural arteriovenous fistula in a patient with Loeys-Dietz syndrome: A case report.	Interv Neuroradiol	23	206-210	2017
Brady AF, Demirdas S, Fournel-Gigleux S, Ghali N, Giunta C, Kapferer-Seebacher I, <u>Kosho T</u> , Mendoza-Londono R, Pope MF, Rohrbach M, Van Damme T, Vandersteen A, van Mourik C, Voermans N, Zschocke J, Malfait F	The Ehlers-Danlos syndromes, rare types.	Am J Med Genet C Semin Med Genet	175(1)	70-115	2017
Duarte C, Kobayashi Y, Morita J, Kawamoto T, <u>Moriyama K</u> .	A preliminary investigation of the effect of relaxin on bone remodelling in suture expansion.	The European Journal of Orthodontics	pii: cjuw037	Epub ahead of print	2017
Fukami M, Suzuki E, Izumi Y, Torii T, Narumi S, Igarashi M, Miyado M, Katsumi M, Fujisawa Y, Nakabayashi K, Hata K, Umezawa A, <u>Matsubara Y</u> , <u>Yamauchi J</u> , <u>Ogata T</u>	Paradoxical gain-of-function mutant of the G-protein coupled receptor PROKR2 promotes early puberty.	J Cell Mol Med	doi: 10.1111/jcmm.13146	Epub ahead of print	2017
Fukuoka M, Kuki I, Kawawaki H, Okazaki S, Kim K, Hattori Y, Tsuji H, Nukui M, Inoue T, Yoshida Y, Uda T, Kimura S, Mogami Y, Suzuki Y, <u>Okamoto N</u> , <u>Saito H</u> , <u>Matsumoto N</u> .	Quinidine therapy for West syndrome with KCNT1 mutation: A case report.	Brain Dev	39	80-83	2017
Hamada N, Negishi Y, Mizuno M, Miya F, Hattori A, <u>Okamoto N</u> , Kato M, Tsunoda T, Yamasaki M, Kanemura Y, <u>Kosaki K</u> , <u>Tabata H</u> , <u>Saitoh S</u> , <u>Nagata KI</u> .	Role of a heterotrimeric G-protein, Gi2, in the corticogenesis: Possible involvement in periventricular nodular heterotopia and intellectual disability	J Neurochem	140	92-95	2017
Hirai M, Muramatsu Y, <u>Mizuno S</u> , <u>Kurahashi N</u> , <u>Kurahashi H</u> , <u>Nakamura M</u> .	Preserved search asymmetry in the detection of fearful faces among neutral faces in individuals with Williams syndrome revealed by measurement of both manual responses and eye tracking.	J Neurodev Disord.	9	8	2017
Hosoya M, Fujioka M, Sone T, Okamoto S, Akamatsu W, Ukai H, Ueda HR, Ogawa K, <u>Matsunaga T</u> , <u>Okano H*</u>	Cochlear cell modeling using disease-specific iPSCs unveils a degenerative phenotype and suggests treatments for congenital progressive hearing loss	Cell Rep	18(1)	68-81	2017
Igarashi M, Takasawa K, Hakoda A, Kanno J, Takada S, Miyado M, Baba T, Morohashi KI, Tajima T, Hata K, Nakabayashi K,	Identical NR5A1 missense mutations in two unrelated 46,XX individuals with testicular tissues.	Hum Mutat	38(1)	39-42	2017

Matsubara Y, Sekido R, Ogata T, Kashimada K, Fukami M					
Ihara K, Fukano C, Ayabe T, Fukami M, Ogata T, Kawamura T, Urakami T, Kikuchi N, Yokota I, Takemoto K, Mukai T, Nishii A, Kikuchi T, Mori T, Shimura N, Sasaki G, Kizu R, Takubo N, Soneda S, Fujisawa T, Takaya R, Kizaki Z, Kanzaki S, Hanaki K, Matsuura N, Kasahara Y, Kosaka K, Takahashi T, Minamitani K, Matsuo S, Mochizuki H, Kobayashi K, Koike A, Horikawa R, Teno S, Tsubouchi K, Mochizuki T, Igarashi Y, Amemiya S, Sugihara S; Japanese Study Group of Insulin Therapy for Childhood and Adolescent Diabetes (JSGIT)	FUT2 nonsecretor status links type 1 diabetes susceptibility in Japanese children.	Diabet Med	34(4)	586-589	2017
Ikeda M, Miyamoto JJ, Takada JI, Moriyama K.	Association between 3-dimensional mandibular morphology and condylar movement in subjects with mandibular asymmetry.	Am J Orthod Dentofacial Orthop	151(2)	324-334	2017
Kasakura-Kimura N, Masuda M, Mutai H, Masuda S, Morimoto N, Ogahara N, Misawa H, Sakamoto H, Saito K, Matsunaga T*	WFS1 and GJB2 mutations in patients with bilateral low-frequency sensorineural hearing loss	Laryngoscope	doi: 10.1002/lary.26528.	Epub ahead of print	2017
Katagiri S, Nishina S, Yokoi T, Mikami M, Nakayama Y, Tanaka M, Azuma N	Retinal structure and function in eyes with optic nerve hypoplasia.	Sci Rep.	doi: 10.1038/srep42480	Epub ahead of print	2017
Katagiri S, Tanaka S, Yokoi T, Hayashi T, Matsuzaka E, Ueda K, Yoshida-Uemura T, Arakawa A, Nishina S, Kadonosono K, Azuma N.	Clinical features of a toddler with bilateral bullous retinoschisis with a novel RS1 mutation.	Am J Ophthalmol Case Rep.	5	76-80	2017
Koda N, Sato T, Shinohara M, Ichinose S, Ito Y, Nakamichi R, Kayama T, Suzuki H, Moriyama K, Asahara H.	The transcription factor mohawk homeobox regulates homeostasis of the periodontal ligament.	Development	144(2)	313-320	2017
Lin W, Izu Y, Smriti A, Kawasaki M, Pawaputanon C, Böttcher RT, Costell M, Moriyama K, Noda M, Ezura Y.	Profillin1 is expressed in osteocytes and regulate cell shape and migration.	J Cell Physiol	doi: 10.1002/jcp.25872.	Epub ahead of print	2017
Malfait F, Francomano C,	The 2017 international classification	Am J Med	175(1)	8-26	2017

Byers P, Belmont J, Berglund B, Black J, Bloom L, Bowen JM, Brady AF, Burrows NP, Castori M, Cohen H, Colombi M, Demirdas S, De Backer J, De Paepe A, Fournel-Gigleux S, Frank M, Ghali N, Giunta C, Grahame R, Hakim A, Jeunemaitre X, Johnson D, Juul-Kristensen B, Kapferer-Seebacher I, Kazkaz H, <u>Kosho T</u> , Lavalley ME, Levy H, Mendoza-Londono R, Pepin M, Pope FM, Reinstein E, Robert L, Rohrbach M, Sanders L, Sobey GJ, Van Damme T, Vandersteen A, van Mourik C, Voermans N, Wheeldon N, Zschocke J, Tinkle B	of the Ehlers-Danlos syndromes.	Genet C Semin Med Genet			
Mizumoto S, <u>Kosho T</u> , Hatamochi A, Honda T, Yamaguchi T, Okamoto N, Miyake N, Yamada S, Sugahara K	Defect in dermatan sulfate in urine of patients with Ehlers-Danlos syndrome caused by a CHST14/D4ST1 deficiency.	Clin Biochem	doi: 10.1016/j.clinbiochem.2017.02.018.	Epub ahead of print	2017
Mizumoto S, <u>Kosho T</u> , Yamada S, Sugahara K	Pathophysiological Significance of Dermatan Sulfate Proteoglycans Revealed by Human Genetic Disorders.	Pharmaceuticals (Basel)	10(2)	E34	2017
Mutai H, Watabe T, <u>Kosaki K</u> , Ogawa K, <u>Matsunaga T*</u>	Mitochondrial mutations in maternally inherited hearing loss	BMC Medical Genetics	18(1)	32	2017
<u>Nishina S</u> , Katagiri S, Nakazawa A, Kiyotani C, Yokoi T, Azuma N	Atypical intravitreal growth of retinoblastoma with a multi-branching configuration.	Am J Ophthalmol Case Rep	7	4-8	2017
Ohishi A, Nishimura G, Kato F, Ono H, Maruwaka K, Ago M, Suzumura H, Hirose E, Uchida Y, Fukami M, <u>Ogata T</u>	Mutation analysis of FGFR1-3 in 11 Japanese patients with syndromic craniosynostoses.	Am J Med Genet A	173(1)	157-162	2017
Ohtaka K, Fujisawa Y, Takada F, Hasegawa Y, Miyoshi T, Hasegawa T, Miyoshi H, Kameda H, Kurokawa-Seo M, Fukami M, <u>Ogata T</u>	FGFR1 Analyses in Four Patients with Hypogonadotropic Hypogonadism with Split-Hand/Foot Malformation: Implications for the Promoter Region.	Hum Mutat	38(5)	503-506	2017
<u>Okamoto N</u> , Kimura S, Shimojima K, Yamamoto T.	Neurological Manifestations of 2q31 Microdeletion Syndrome.	Congenit Anom (Kyoto)	doi: 10.1111/cga.12212.	Epub ahead of print	2017
<u>Okamoto N</u> , Nakao H, Niihori T, <u>Aoki Y</u> .	Patient with a novel purine-rich element binding protein A mutation.	Congenit Anom (Kyoto).	doi: 10.1111/	Epub ahead of print	2017

			cga.1221 4		
Ozawa H, Yamane M, Inoue E, Yoshida-Uemura T, Katagiri S, Yokoi T, <u>Nishina S</u> , Azuma N.	Long-term surgical outcome of conventional trabeculotomy for childhood glaucoma.	Jpn J Ophthalmol	61(3)	237-244	2017
Sawada H, Ogawa T, Kataoka K, Baba Y, <u>Moriyama K.</u>	Measurement of distraction force in maxillary distraction osteogenesis for cleft lip and palate.	Journal of Craniofacial Surgery	28(2)	406-412	2017
Shiga M, Ogawa T, Ekprachayakoon I, <u>Moriyama K.</u>	Orthodontic treatment and long-term management of a patient with Marfan syndrome.	Cleft Palate-Craniofacial J	54(3)	358-367	2017
Tanigawa J, Mimatsu H, <u>Mizuno S</u> , <u>Okamoto N</u> , Fukushi D, Tominaga K, Kidokoro H, Muramatsu Y, Nishi E, Nakamura S, Motooka D, Nomura N, Hayasaka K, Niihori T, <u>Aoki Y</u> , Nabatame S, Hayakawa M, Natsume J, Ozono K, Kinoshita T, Wakamatsu N, Murakami Y.	Phenotype-genotype correlations of PIGO deficiency with variable phenotypes from infantile lethality to mild learning difficulties.	Human Mutation	doi: 10.1002	Epub ahead of print	2017
Wakeling EL, Brioude F, Lokulo-Sodipe O, O'Connell SM, Salem J, Blied J, Canton AP, Chrzanowska KH, Davies JH, Dias RP, Dubern B, Elbracht M, Giabicani E, Grimberg A, Grønskov K, Hokken-Koelega AC, Jorge AA, Kagami M, Linglart A, Maghnie M, Mohnike K, Monk D, Moore GE, Murray PG, <u>Ogata T</u> , Petit IO, Russo S, Said E, Toumba M, Tümer Z, Binder G, Eggermann T, Harbison MD, Temple IK, Mackay DJ, Netchine I	Diagnosis and management of Silver-Russell syndrome: first international consensus statement.	Nat Rev Endocrinol	13(2)	105-124	2017
Yahiro K, Higashihori N, <u>Moriyama K.</u>	Histone methyltransferase Setdb1 is indispensable for Meckel's cartilage development.	Biochem Biophys Res Commun	482(4)	883-888	2017
Yoshida-Uemura T, Katagiri S, Yokoi T, <u>Nishina S</u> , Azuma N.	Different foveal schisis patterns in each retinal layer in eyes with hereditary juvenile retinoschisis evaluated by en-face optical coherence tomography.	Graefes Arch Clin Exp Ophthalmol.	255 (4)	719-723	2017
<u>松永達雄*</u>	遺伝性難聴と内耳再生医療	日本医事新報	4846	29	2017
<u>森崎裕子</u>	遺伝カウンセリングが必要な循環器疾患と実際	HeartView	21	433-439	2017
<u>森崎裕子</u> , <u>森崎隆幸</u>	遺伝性大動脈疾患: NGS 時代の遺伝子診断	日本血栓止血学会誌	28	41-49	2017

大塚泰史、副島英伸.	モザイク病、インプリンティング.	腎と透析	82(3)	356-362	2017
樋野村亜希子、倉田真由美、小原有弘、松山晃文	指定難病はどのように選考されたのかー難病対策の検討委員会における検討過程の要点整理ー [前篇]	難病と在宅ケア	Vol.22 No.11	pp36-39	2017
樋野村亜希子、倉田真由美、小原有弘、松山晃文	指定難病はどのように選考されたのかー難病対策の検討委員会における検討過程の要点整理ー [後篇]	難病と在宅ケア	Vol.22 No.12	pp.44-47	2017
Morimoto N, Mutai H, Namba K, Kaneko H, Kosaki R, Matsunaga T*	Homozygous EDNRB Mutation in a Patient with Waardenburg Syndrome Type 1	Acta Oto-Laryngologica		in press	
Kagami M, Nagasaki K, Kosaki R, Horikawa R, Naiki Y, Saito S, Tajima T, Yorifuji T, Numakura C, Mizuno S, Nakamura A, Matsubara K, Fukami M, Ogata T	Temple syndrome: comprehensive molecular and clinical findings in 32 Japanese patients.	Genet Med (accepted).			
Kitaoka T, Tajima T, Nagasaki K, Kikuchi T, Yamamoto K, Michigami T, Okada S, Fujiwara I, Kokaji M, Mochizuki Hi, Ogata T, Tatebayashi K, Watanabe A, Yatsuga S, Kubota T, Ozono K: Safety and Efficacy of Treatment with Asfotase Alfa in Patients with Hypophosphatasia (HPP)	Results from Japanese Physician-Initiated Clinical Trial.	Clin Endocrinol (accepted)			