

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
Ogi T, Nakazawa Y, Sasaki K, Guo C, Yoshiura K, Utani A, Nagayama Y.	Molecular cloning and characterisation of UVSSA, the responsible gene for UV-sensitive syndrome	公益社団 法人日本 生化学会	Journal of Biochemistry		Tokyo	2013	85: 133-144
吉浦孝一郎	遺伝性疾患におけ るエクソーム解析 の有用性と近将来	松本直通	医学の歩み	医師薬出 版株式会 社	東京	2013	245: 363-368
黒滝直弘, 小野慎二, 小澤寛樹, 吉浦孝一郎	発作性運動誘発性 舞蹈アテトーゼの 分子メカニズム		神経内科	化学評論 社	東京	2013	79: 718-725
齋藤伸治	Prader-Willi 症候群 と Angelman 症候群	佐々木裕 之	エピジェネテ ィクスと病気 ( 遺伝子医学 MOOK 25 )	メディカ ルドウ	東京	2013	189-94.
森崎裕子、 森崎隆幸	遺伝性大動脈疾患 の診断	高本眞一	大動脈外科の 要点と盲点	文光堂	東京	2013	53-58
森崎裕子、 森崎隆幸	大動脈疾患による 遺伝子異常	山口徹 他	Annual Review 循環器 2012	中外医学 社	東京	2012	240 - 246
森崎裕子	ロイス・ディーツ症 候群	遠藤文夫	先天代謝異常 症候群	日本臨床 社	大阪	2012	731-735
Kosho T	Discovery and delineation of dermatan 4-O-sulfotransferase- 1 (D4ST1)-deficient Ehlers-Danlos syndrome	Oiso N, Kawada A	Current Genetics in Dermatology	InTech	Croatia	2013	73-86

古庄知己	その他の遺伝性大動脈瘤・大動脈解離—血管型エーラス・ダンロス症候群.	鈴木亨, 永井良三	最新医学別冊 新しい診断と治療のABC(42) 大動脈瘤・大動脈解離	最新医学社	大阪	2013	85-92
古庄知己	エーラス・ダンロス症候群		先天代謝異常症候群	日本臨牀社	大阪	2012	721-726
古庄知己	Marfan症候群, Ehlers-Danlos症候群	五十嵐隆	小児疾患の診断治療基準 第4版	東京医学社	東京	2012	850-853
増井徹	第10章バイオバンク	玉井真理子, 松田純	シリーズ生命倫理学 11巻 遺伝子と医療	丸善出版	東京	2013	188 - 203
増井徹	バイオバンクプロジェクトの開始と終了に向けて検討すべき ELSI	個人の遺伝情報に応じた医療の実現プロジェクトELSI委員会	平成16年度～平成24年度個人の遺伝情報に応じた医療の実現プロジェクト	個人の遺伝情報に応じた医療の実現プロジェクトELSI委員会	東京	2013	51 - 67

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
Kosaki R, Takenouchi T, Takeda N, Kagami M, Nakabayashi K, Hata K, Kosaki K.	Somatic CTNNB1 mutation in hepatoblastoma from a patient with Simpson-Golabi-Behmel syndrome and germline GPC3 mutation.	<i>Am J Med Genet A</i>	164(4)	993-7	2014
Takenouchi T, Hashida N, Torii C, Kosaki R, Takahashi T, Kosaki K.	1p34.3 deletion involving GRIK3: Further clinical implication of GRIK family glutamate receptors in the pathogenesis of developmental delay.	<i>Am J Med Genet A.</i>	164(2)	456-60	2014
Nomura T, Takenouchi T, Fukushima H, Shimozato S, Kosaki K, Takahashi T.	Catastrophic Autonomic Crisis With Cardiovascular Collapse in Spinal Muscular Atrophy With Respiratory Distress Type 1.	<i>J Child Neurol.</i>	28(7)	949-951.	2013
Takenouchi T, Nishina S, Kosaki R, Torii C, Furukawa R, Takahashi T, Kosaki K.	Concurrent deletion of BMP4 and OTX2 genes, two master genes in ophthalmogenesis.	<i>Eur J Med Genet.</i>	56(1)	50-53.	2013
Ueda K, Awazu M, Konishi Y, Takenouchi T, Shimozato S, Kosaki K, Takahashi T.	Persistent hypertension despite successful dilation of a stenotic renal artery in a boy with neurofibromatosis type 1.	<i>Am J Med Genet A.</i>	161(5)	1154-1157.	2013
Takenouchi T, Saito H, Maruoka R, Oishi N, Torii C, Maeda J, Takahashi T, Kosaki K.	Severe obstructive sleep apnea in Loeys-Dietz syndrome successfully treated using continuous positive airway pressure.	<i>Am J Med Genet A.</i>	161(7)	1733-1736.	2013
Hirasawa A, Zama T, Akahane T, Nomura H, Kataoka F, Saito K, Okubo K, Tominaga E, Makita K, Susumu N, Kosaki K, Tanigawara Y, Aoki D.	Polymorphisms in the UGT1A1 gene predict adverse effects of irinotecan in the treatment of gynecologic cancer in Japanese patients.	<i>J Hum Genet.</i>	58(12)	794-798.	2013
Takenouchi T, Hida M, Sakamoto Y, Torii C, Kosaki R, Takahashi T, Kosaki K.	Severe congenital lipodystrophy and a progeroid appearance: Mutation in the penultimate exon of FBN1 causing a recognizable phenotype.	<i>Am J Med Genet A</i>	161(12)	3057-3062.	2013

Kubo A, Shiohama A, Sasaki T, Nakabayashi K, Kawasaki H, Atsugi T, Sato S, Shimizu A, Mikami S, Tanizaki H, Uchiyama M, Maeda T, Ito T, Sakabe J, Heike T, Okuyama T, Kosaki R, Kosaki K, Kudoh J, Hata K, Umezawa A, Tokura Y, Ishiko A, Niizeki H, Kabashima K, Mitsuhashi Y, Amagai M.	Mutations in SERPINB7, Encoding a Member of the Serine Protease Inhibitor Superfamily, Cause Nagashima-type Palmoplantar Keratosis.	<i>Am J Hum Genet.</i>	93(5)	945-956.	2013
Takagi M, Ishii T, Torii C, Kosaki K, Hasegawa T.	A novel mutation in SOX3 polyalanine tract: a case of kabuki syndrome with combined pituitary hormone deficiency harboring double mutations in MLL2 and SOX3.	<i>Pituitary</i>		[Epub ahead of print]	2013
Mutai H, Suzuki N, Shimizu A, Torii C, Namba K, Morimoto N, Kudoh J, Kaga K, Kosaki K, Matsunaga T.	Diverse spectrum of rare deafness genes underlies early-childhood hearing loss in Japanese patients: A cross-sectional, multi-center next-generation sequencing study.	<i>Orphanet J. Rare Dis.</i>	8(1)	172	2013
Takenouchi T, Kosaki R, Torii C, Kosaki K	Daytime Somnolence in an Adult with Smith-Magenis Syndrome	<i>Am J Med Genet A.</i>	161A (7)	1803-5	2013
Sasaki K, Mishima H, Miura K, Yoshiura KI.	Uniparental disomy analysis in trios using genome-wide SNP array and whole-genome sequencing data imply segmental uniparental isodisomy in general populations.	<i>Gene</i>	512(2)	267-274	2013
Yamada A, Ishikawa T, Ota I, Kimura M, Shimizu D, Tanabe M, Chishima T, Sasaki T, Ichikawa Y, Morita S, Yoshiura KI,	High expression of ATP-binding cassette transporter ABCC11 in breast tumors is associated with aggressive subtypes and low disease-free survival.	<i>Breast Cancer Res Treat</i>	137(3)	773-782	2013
Ishikawa T, Toyoda Y, Yoshiura K, Niikawa N.	Pharmacogenetics of human ABC transporter ABCC11: new insights into apocrine gland growth and metabolite secretion.	<i>Front. Genet.</i>	3	306	2013

Higashijima A, Miura K, Mishima H, Kinoshita A, Jo O, Abe S, Hasegawa Y, Miura S, Yamasaki K, Yoshida A, Yoshiura K, Masuzaki H.	Characterization of placenta-specific microRNAs in fetal growth restriction pregnancy.	<i>Prenat Diagn.</i>	33(3)	214-222	2013
Abe S, Miura K, Kinoshita A, Mishima H, Miura S, Yamasaki K, Hasegawa Y, Higashijima A, Jo O, Sasa K, Yoshida A, Yoshiura K, Masuzaki H.	Copy number variation of the antimicrobial-gene, defensin beta 4, is associated with susceptibility to cervical cancer.	<i>J Hum Genet</i>	58(5)	250-253	2013
Kashiyama K, Nakazawa Y, Pilz D, Guo C, Shimada M, Sasaki K, Fawcett H, Wing J, Lewin S, Carr L, Yoshiura K, Utani A, Hirano A, Yamashita S, Greenblatt D, Nardo T, Stefanini M, McGibbon D, Sarkany R, Fassihi H, Takahashi Y, Nagayama Y, Mitsutake N, Lehmann AR, and Ogi T.	Malfunction of the ERCC1/XPF endonuclease results in diverse clinical manifestations and causes three nucleotide excision-repair-deficient disorders, Cockayne Syndrome, xeroderma pigmentosum and Fanconi Anemia.	<i>Am J Hum Genet</i>	92(5)	807-819	2013
Nakao K, Oikawa M, Arai J, Mussazhanova Z, Kondo H, Shichijo K, Nakashima M, Hayashi T, Yoshiura K, Hatachi T, Nagayasu T.	A Predictive Factor of the Quality of Microarray Comparative Genomic Hybridization Analysis for Formalin-fixed Paraffin-embedded Archival Tissue.	<i>Diagn Mol Pathol</i>	22(3)	174-180	2013
Hasegawa Y, Miura K, Furuya K, Yoshiura K, Masuzaki H.	Identification of Complete Hydatidiform Mole Pregnancy-Associated MicroRNAs in Plasma.	<i>Clin Chem</i>	59(9)	1410-1412	2013
Higashimoto K, Maeda T, Okada J, Ohtsuka Y, Sasaki K, Hirose A, Nomiya M, Takayanagi T, Fukuzawa R, Yatsuki H, Koide K, Nishioka K, Joh K, Watanabe Y, Yoshiura KI, Soejima H.	Homozygous deletion of DIS3L2 exon 9 due to non-allelic homologous recombination between LINE-1s in a Japanese patient with Perlman syndrome.	<i>Eur J Hum Genet</i>	21(11)	1316-1319	2013

Hamaguchi D, Miura K, Abe S, Kinoshita A, Miura S, Yamasaki K, Yoshiura KI, Masuzaki H.	Initial viral load in cases of single human papillomavirus 16 or 52 persistent infection is associated with progression of later cytopathological findings in the uterine cervix.	<i>J Med Virol</i>	85(12)	2093-2100	2013
Sekiguchi K, Maeda T, Suenobu S, Kunisaki N, Shimizu M, Kiyota K, Handa YS, Akiyoshi K, Korematsu S, Aoki Y, Matsubara Y, Izumi T.	A transient myelodysplastic/myeloproliferative neoplasm in a patient with cardio-facio-cutaneous syndrome and a germline BRAF mutation	<i>Am J Med Genet A</i>	161(10)	2600-3	2013
Aoki Y, Niihori T, Banjo T, Okamoto N, Mizuno S, Kurosawa K, Ogata T, Takada F, Yano M, Ando T, Hoshika T, Barnett C, Ohashi H, Kawame H, Hasegawa T, Okutani T, Nagashima T, Hasegawa S, Funayama R, Nagashima T, Nakayama K, Inoue S, Watanabe Y, Ogura T, Matsubara Y.	Gain-of-function mutations in RIT1 cause Noonan syndrome, a RAS/MAPK pathway syndrome.	<i>Am J Hum Genet</i>	93(1)	173-80	2013
Izumi R, Niihori T, Aoki Y, Suzuki N, Kato M, Warita H, Takahashi T, Tateyama M, Nagashima T, Funayama R, Abe K, Nakayama K, Aoki M, Matsubara Y.	Exome sequencing identifies a novel TTN mutation in a family with hereditary myopathy with early respiratory failure.	<i>J Hum Genet.</i>	58(5)	259-66	2013
Narisawa A, Komatsuzaki S, Kikuchi A, Niihori T, Aoki Y, Fujiwara K, Tanemura M, Hata A, Suzuki Y, Relton CL, Grinham J, Leung KY, Partridge D, Robinson A, Stone V, Gustavsson P, Stanier P, Copp AJ, Greene ND, Tominaga T, Matsubara Y, Kure S	Mutations in genes encoding the glycine cleavage system predispose to neural tube defects in mice and humans.	<i>Hum Mol Genet.</i>	21	1496-503	2013
Kato F, Hamajima T, Hasegawa T, Ogata T, et al.	IMAGE syndrome: clinical and genetic implications based on investigations in three Japanese patients.	<i>Clin Endocrinol</i>	80(5)	706-13	2014
Tsuchiya T, Shibata M, Numabe H, Ogata T, Fukami M, et al.	Compound heterozygous deletions in pseudoautosomal region 1 in an infant with mild manifestations of Langer mesomelic dysplasia.	<i>Am J Med Genet A</i>	64 A(2):	505-10.	2014

Nakashima S, Watanabe Y, Okada J, Ogata T, et al.	Critical role of Yp inversion in <i>PRKX/PRKY</i> -mediated Xp;Yp translocation in a patient with 45,X testicular disorder of sex development.	<i>Endocr J</i>	Oct 3	Epub ahead of print	2013
Shihara D, Miyado M, Nakabayashi K, Ogata T, Fukami M, et al.	Aromatase excess syndrome in a family with upstream deletion of <i>CYP19A1</i> .	<i>Clin Endocrinol</i>	Sep 18	Epub ahead of print	2013
Fujisawa Y, Yamaguchi R, Satake E, Ogata T, et al.	Identification of <i>AP2S1</i> Mutation and Effects of Low Calcium Formula in an Infant with Hypercalcemia and Hypercalciuria.	<i>J Clin Endocrinol Metab</i>	98 (12)	E2022–2027	2013
Fukami M, Suzuki J, Ogata T, Shozu M, Noguchi S, et al.	Lack of genomic rearrangements involving the aromatase gene <i>CYP19A1</i> in breast cancer.	<i>Breast Cancer</i>	Apr 30	Epub ahead of print	2013
Fukami M, Tsuchiya T, Vollbach H, Ogata T, et al.	Genomic basis of aromatase excess syndrome: recombination- and replication-mediated rearrangements leading to <i>CYP19A1</i> overexpression.	<i>J Clin Endocrinol Metab</i>	98 (12)	E2013–2021	2013
Igarashi M, Dung VC, Suzuki E, Ogata T, Fukami M, et al.	Cryptic genomic rearrangements in three patients with 46,XY disorders of sex development.	<i>PLoS One</i>	8 (7)	e68194	2013
Fukami M, Iso M, Sato N, Ogata T, et al.	Submicroscopic deletion involving the fibroblast growth factor receptor 1 gene in a patient with combined pituitary hormone deficiency.	<i>Endocr J</i>	60 (8)	1013–1020	2013
Yamaguchi R, Kato F, Hasegawa T, Ogata T, et al.	A novel homozygous mutation of the nicotinamide nucleotide transhydrogenase gene in a Japanese patient with familial glucocorticoid deficiency.	<i>Endocr J</i>	60 (7)	855–859	2013
Moritani M, Yokota I, Tsubouchi K, Ogata T, et al.	Identification of <i>INS</i> and <i>KCNJ11</i> gene mutations in type 1B diabetes in Japanese children with onset of diabetes before 5 yr of age.	<i>Pediatr Diabetes</i>	14(2)	112-20	2013
Suzuki-Suwanai A, Ishii T, Haruna H, Ogata T, et al.	A report of two novel <i>NR5A1</i> mutation families: possible clinical phenotype of psychiatric symptoms of anxiety and/or depression.	<i>Clin Endocrinol</i>	78(6)	957-65	2013
Sekii K*, Itoh H, Ogata T, Iwashima S*	Possible contribution of fetal size and gestational age to myocardial tissue Doppler velocities in preterm fetuses.	<i>Eur J Obstet Gynecol Reprod Biol</i>	167(1)	121	2013

Nagasaki K*, Tsuchiya S, Saitoh A, Ogata T, Fukami M.	Neuromuscular symptoms in a patient with familial pseudohypoparathyroidism type Ib diagnosed by methylation-specific multiplex ligation-dependent probe amplification.	<i>Endocr J</i>	60(2)	231-6	2013
Ohishi A*, Ueno D, Matsuoka, H, Kawamoto, F Ogata T	Glucose-6-phosphate dehydrogenase deficiency and adrenal hemorrhage in a Filipino neonate with hyperbilirubinemia.	<i>Am J Perinatol Reports</i>	3(1)	5-8.	2013
Fuke T, Mizuno S, Nagai T, Hasegawa T, Ogata T*et al.	Molecular and clinical studies in 138 Japanese patients with Silver-Russell syndrome.	<i>PLoS ONE</i>	8(3)	e60105	2013
Ayabe T, Matsubara K, Ogata T, Ayabe A, Murakami N, Nagai T, Fukami M*	Birth seasonality in Prader-Willi syndrome resulting from chromosome 15 microdeletion.	<i>Am J Med Genet A</i>	161A (6)	1495-7	2013
Fukami M, Homma K, Hasegawa T, Ogata T*	Backdoor pathway for dihydrotestosterone biosynthesis: implications for normal and abnormal human sex development.	<i>Dev Dyn</i>	242(4)	320-9	2013
Ueda H, <i>et al.</i>	Combination of Miller-Dieker syndrome and VACTERL association causes extremely severe clinical presentation.	<i>Eur J Pediatr</i>		in press	2013
Suzumori N, <i>et al.</i>	Prenatal diagnosis of X-linked recessive Lenz microphthalmia syndrome.	<i>J Obstet Gynaecol Res</i>	39	1545-7	2013
Hamajima N, <i>et al.</i>	Increased protein stability of CDKN1C causes a gain-of-function phenotype in patients with IMAGE syndrome.	<i>PLoS One</i>	8	e75137.	2013
Yoneda Y, <i>et al.</i>	Phenotypic spectrum of COL4A1 mutations: porencephaly to schizencephaly.	<i>Ann Neurol</i>	73	48-57.	2013
Hosoki <i>et al.</i>	Hand-foot-genital syndrome with a 7p15 deletion demonstrates a clinically recognizable syndrome.	<i>Pediatr Int</i>	54	e22-25.	2012



Hosoki <i>et al.</i>	Clinical Phenotype and Candidate Genes for the 5q31.3 Microdeletion Syndrome.	<i>Am J Med Genet A</i>	158A	1891-1896.	2012
Kawamura <i>et al.</i>	Visualization of the spatial positioning of the <i>SNRPN</i> , <i>UBE3A</i> , and <i>GABRB3</i> genes in the normal human nucleus by three-color 3D-fluorescence in situ hybridization.	<i>Chromosome Res</i>	20	659-672.	2012
Tsurusaki <i>et al.</i>	A <i>DYNC1H1</i> mutation causes a dominant spinal muscular atrophy with lower extremity predominance.	<i>Neurogenetics</i>	13	327-332.	2012
Egawa <i>et al.</i>	Decreased tonic inhibition in cerebellar granule cells causes motor dysfunction in a mouse model of Angelman syndrome.	<i>Sci Transl Med</i>	4	163ra157.	2012
Yatsuki H, Higashimoto K, Jozaki K, Koide K, Okada J, Watanabe Y, Okamoto N, Tsuno Y, Yoshida Y, Ueda K, Shimizu K, Ohashi H, Mukai T, Soejima H.	Novel Mutations of <i>CDKN1C</i> in Japanese Patients with Beckwith-Wiedemann Syndrome.	<i>Genes Genom</i>	35(2)	141-147	2013
Adachi H, Takahashi I, Higashimoto K, Tsuchida S, Noguchi A, Tamura H, Arai H, Ito T, Masue M, Nishibori H, Takahashi T, Soejima H.	Congenital hyperinsulinism in an infant with paternal uniparental disomy on chromosome 11p15: Few clinical features suggestive of Beckwith-Wiedemann syndrome.	<i>Endocr J</i>	60(4)	403-408	2013
Misago N, Joh K, Soejima H, Narisawa Y.	Multiple mucocutaneous (palisaded encapsulated) neuromas may be a distinct entity.	<i>JAMA Dermatol</i>	149(4)	498-500	2013
Soejima H, Higashimoto K	Epigenetic and genetic alterations of the imprinting disorder Beckwith-Wiedemann syndrome and related disorders.	<i>J Hum Genet</i>	58(7)	402-409	2013
Fukuda K, Ichiyanagi K, Yamada Y, Go Y, Udono T, Wada S, Maeda T, Soejima H, Saitou N, Ito T, Sasaki H.	Regional DNA methylation differences between humans and chimpanzees are associated with genetic changes, transcriptional divergence and disease genes.	<i>J Hum Genet</i>	58(7)	446-454	2013

Miyazaki H <sup>†</sup> , Higashimoto K <sup>†</sup> , Yada Y <sup>†</sup> , A. Endo T <sup>¶</sup> , Sharif J <sup>¶</sup> , Komori T, Matsuda M, Koseki Y, Nakayama M, Soejima H, Handa H, Koseki H, Hirose S, Nishioka K.( <sup>†</sup> , <sup>¶</sup> equal contribution)	Ash11 methylates Lys36 of histone H3 independently of transcriptional elongation to counteract Polycomb silencing.	<i>PLoS Genet</i>	9(11)	e1003897	2013
Rumbajan JM, Maeda T, Souzaki R, Mitsui K, Higashimoto K, Nakabayashi K, Yatsuki H, Nishioka K, Harada R, Aoki S, Kohashi K, Oda Y, Hata K, Saji T, Taguchi T, Tajiri T, Soejima H, Joh K.	Comprehensive analyses of imprinted differentially methylated regions reveal epigenetic and genetic characteristics in hepatoblastoma.	<i>BMC Cancer</i>	13	608	2013
Higashimoto K, Jozaki K, Kosho T, Matsubara K, Fuke T, Yamada D, Yatsuki H, Maeda T, Ohtsuka Y, Nishioka K, Joh K, Koseki H, Ogata T, Soejima H.	A novel de novo point mutation of the OCT-binding site in the IGF2/H19-imprinting control region in a Beckwith-Wiedemann syndrome patient.	<i>Clin Genet</i>			2013
Takahashi Y, Fujii K, Yoshida A, Morisaki H, Kohno Y, and Morisaki T,	Artery tortuosity syndrome exhibiting early-onset emphysema with novel compound heterozygous SLC2A10 mutations	<i>Am J Med Genet A</i>	12(10)	35776	2013
JCS Joint Working Group	Guidelines for Diagnosis and Treatment of Aortic Aneurysm and Aortic Dissection (JCS 2011)	<i>Circ J</i>	77(3)	789-828	2013
Katsuragi S, Neki N, Yoshimatsu J, Ikeda T, Morisaki H, Morisaki T	Acute aortic dissection (Stanford type B) during pregnancy	<i>J Perinatol</i>	33	484-485	2013
Kono AK, Higashi M, Morisaki H, Morisaki T, Naito H, Sugimura K	Prevalence of dural ectasia in loeys-dietz syndrome: comparison with marfan syndrome and normal controls	<i>PLoS One</i>	8	e75264	2013
Hayashi S, Utani A, Iwanaga A, Yagi Y, Morisaki H, Morisaki T, Hamasaki Y, Hatamochi A	Co-existence of mutations in the FBN1 gene and the ABCC6 gene in a patient with Marfan syndrome associated with pseudoxanthoma elasticum.	<i>J Dermatol Sci</i>	72	325-327	2013

Akizu N, Cantagrel V, Schroth J, Cai N, Vaux V, McCloskey D, Naviaux RK, Van Vleet J, Fenstermaker AG, Silhavy JL, Scheliga JS, Toyama K, Morisaki H, Sonmez FM, Celep F, Oraby A, Zaki MS, Al-Baradie R, Faqeih EA, Saleh MAM, Spencer E, Rosti RO, Scott E, Nickerson E, Gabriel S, Morisaki T, Holmes EW, Gleeson JG	AMPD2 regulates GTP synthesis and is mutated in a potentially treatable neurodegenerative brainstem disorder	<i>Cell</i>	154	505-517	2013
Li P, Ogino K, Hoshikawa Y, Morisaki H, Toyama K, Morisaki T, Morikawa K, Ninomiya H, Yoshida A, Hashimoto K, Shirayoshi Y, Hisatome I	AMP deaminase 3 plays a critical role in remote reperfusion lung injury	<i>Biochem Biophys Res Commun</i>	434	131-136	2013
森崎裕子	大動脈瘤と遺伝子	<i>Heart View</i>	17	60-65	2013
Katsuragi S, Neki N, Yoshimatsu J, Ikeda T, Morisaki H, Morisaki T	Acute aortic dissection (Stanford type B) during pregnancy	<i>J Perinatol</i>	33(6)	484-5	2013
Fujita D, Takahashi M, Doi K, Abe M, Tazaki J, Kiyosue A, Myojo M, Ando J, Fujita H, Noiri E, Sugaya T, Hirata Y, Komuro I.	Response of urinary liver-type fatty acid-binding protein to contrast media administration has a potential to predict one-year renal outcome in patients with Ischemic heart disease.	<i>Heart and Vessel</i>		In press	2014
藤田大司、今井靖、平田恭信	循環器遺伝子診療の新展開- 遺伝子型から臨床へ- マルファン症候群	<i>心臓</i>	46(1)	21-26	2014
藤田大司、今井靖、平田恭信	大動脈瘤・大動脈解離 改訂第2版第4章 管理・治療 マルファン症候群の経過・治療・予後 1.内科治療	<i>最新医学・別冊 新しい診断と治療のABC</i> 42		244-252	2013
Ogawa N, Imai Y, Nishimura H, Kato M, Takeda N, Nawata K, Taketani T, Morota T, Takamoto S, Nagai R, Hirata Y	Circulating transforming growth factor $\beta$ -1 level in Japanese patients with Marfan syndrome.	<i>Int Heart J</i>	54	23-6	2013

Ashigaki N, Suzuki J, Ogawa M, Watanabe R, Aoyama N, Kobayashi N, Hanatani T, Sekinishi A, Zempo H, Tada Y, Takamura C, Wakayama K, Hirata Y, Nagai R, Izumi Y, Isobe M.	Periodontal bacteria aggravate experimental autoimmune myocarditis in mice.	<i>Am J Physiol Heart Circ Phys</i>	304	H740-8	2013
Takeda N, Jain R, Li D, Li L, Lu MM, Epstein JA.	Lgr5 identifies progenitor cells capable of taste bud regeneration after injury.	<i>PLoS One.</i>	8	e66314.	2013
Takeda N, Jain R, Leboeuf MR, Padmanabhan A, Wang Q, Li L, Lu MM, Millar SE, Epstein JA.	Hopx expression defines a subset of multipotent hair follicle stem cells and a progenitor population primed to give rise to K6+ niche cells.	<i>Development</i>	140	1655-64	2013
Takata M, Amiya E, Watanabe M, Omori K, Imai Y, Fujita D, Nishimura H, Kato M, Morota T, Nawata K, Ozeki A, Watanabe A, Kawarasaki S, Hosoya Y, Nakao T, Maemura K, Nagai R, Hirata Y, Komuro I.	Impairment of flow-mediated dilation correlates with aortic dilation in patients with Marfan syndrome.	<i>Heart Vessel</i>		epub	2013
Miyairi T, Miyata H, Taketani T, Sawaki D, Suzuki T, Hirata Y, Shimizu H, Motomura N, Takamoto S.	Risk Model of Cardiovascular Surgery in 845 Marfan Patients Using the Japan Adult Cardiovascular Surgery Database	<i>Intern Heart J</i>	54	401-404	2013
藤田大司、今井靖、平田恭信	マルファン症候群の経過・治療・予後 1.内科治療	最新医学・別冊「新しい診断と治療のABC42」		244-252	2013
Ochiai H., <i>et al</i>	TALEN-mediated single-base-pair editing identification of an intergenic mutation upstream of BUB1B as causative of PCS (MVA) syndrome.	<i>Proc Natl Acad Sci USA</i>	111	1461-1466	2014
Miyamoto R., <i>et al</i>	Exome sequencing reveals a novel MRE11 mutation in a patient with progressive myoclonic ataxia.	<i>J Neurol Sci</i>	337	219-223	2013

Sakuma T., <i>et al</i>	Repeating pattern of non-RVD variations in DNA-binding modules enhances TALEN activity.	<i>Sci Rep</i>	3	3379	2013
Sakuma T., <i>et al</i>	Efficient TALEN construction and evaluation methods for human cell and animal applications.	<i>Genes Cells</i>	18(4)	315-26	2013
落合 博 他	TALE nuclease (TALEN)を用いた培養細胞におけるゲノム編集	実験医学	31	95-100	2013
Kobayashi J., <i>et al.</i>	Nucleolin participates in DNA double-strand break-induced damage response through MDC1-dependent pathway.	<i>PLoS One</i>	7(11)	e49245	2012
Ochiai H., <i>et al.</i>	Zinc-finger nuclease-mediated targeted insertion of reporter genes for quantitative imaging of gene expression in sea urchin embryos.	<i>Proc Natl Acad Sci USA</i>	109	10915-10920	2012
Nakajima M, Mizumoto S, Miyake N, Kogawa R, Iida A, Ito H, Kitoh H, Hirayama A, Mitsubuchi H, Miyazaki O, Kosaki R, Horikawa R, Lai A, Mendoza-Londono R, Dupuis L, Chitayat D, Howard A, Leal GF, Cavalcanti D, Tsurusaki Y, Saito H, Watanabe S, Lausch E, Unger S, Bonafe L, Ohashi H, Superti-Furga A, Matsumoto N, Sugahara K, Nishimura G, Ikegawa S.	Mutations in B3GALT6, which Encodes a Glycosaminoglycan Linker Region Enzyme, Cause a Spectrum of Skeletal and Connective Tissue Disorders.	<i>Am J Hum Genet.</i>	92(6)	927-34.	2013
Sasaki A, Sumie M, Wada S, Kosaki R, Kuroswa K, Fukami M, Sago H, Ogata T, Kagami M.	Prenatal genetic testing for a microdeletion at chromosome 14q32.2 imprinted region leading to UPD(14)pat-like phenotype.	<i>Am J Med Genet A.</i>	[Epub ahead of print]		2013
小崎里華	先天異常の分類	小児科臨床	66 巻増 刊号		2013

Kasahara M, Sakamoto S, Kanazawa H, Karaki C, Kakiuchi S, Shigeta T, Fukuda A, Kosaki R, Nakazawa A, Ishige M, Nagao M, Shigematsu Y, Yorifuji T, Naiki Y, Horikawa R.	Living-donor liver transplantation for propionic acidemia.	<i>Pediatr Transplant.</i>	16(3)	230-234.	2012
Okamoto N, Hayashi S, Masui A, Kosaki R, Oguri I, Hasegawa T, Imoto I, Makita Y, Hata A, Moriyama K, Inazawa J.	Deletion at chromosome 10p11.23-p12.1 defines characteristic phenotypes with marked midface retrusion.	<i>J Hum Genet.</i>	57(3)	191-196	2012
Miyazaki O, Nishimura G, Sago H, Horiuchi T, Hayashi S, Kosaki R.	Prenatal diagnosis of fetal skeletal dysplasia with 3D CT.	<i>Pediatr Radiol.</i>	42(7)	842-852.	2012
Yagihashi T, Kosaki K, Okamoto N, Mizuno S, Kurosawa K, Takahashi T, Sato Y, Kosaki R.	Age-dependent change in behavioral feature in Rubinstein-Taybi syndrome.	<i>Congenit Anom.</i>	52(2)	82-86.	2012
Kosaki R, Nagao K, Kameyama K, Suzuki M, Fujii K, Miyashita T.	Heterozygous tandem duplication within the PTCH1 gene results in nevoid basal cell carcinoma syndrome.	<i>Am J Med Genet A.</i>	158(7)	1724-1728.	2012
野崎誠 佐々木りか子 土井亜紀子 重松由紀子 久保田雅也 関敦仁 東範行 小崎里華 新関寛徳	小児期のレックリングハウゼン病患者は初診時に何割が確定診断できるか	日本レックリングハウゼン病学会誌	第2巻第1号		2012
境信哉、真木誠、境直子、須藤章、加藤光広、齋藤伸治	脊髄性筋萎縮症I型児(者)におけるスイッチ使用状況・言語発達・上肢機能・QOL-親に対するアンケート調査より-	脳と発達	44	465-471	2012
Yagihashi T, Kosaki K, Okamoto N, Mizuno S, Kurosawa K, Takahashi T, Sato Y, Kosaki R.	Age-dependent change in behavioral feature in Rubinstein-Taybi syndrome.	<i>Congenit Anom (Kyoto)</i>	52(2)	Jun-82	2012
Kosaki R, Kaneko T, Torii C, Kosaki K.	EEC syndrome-like phenotype in a patient with an IRF6 mutation .	<i>Am J Med Genet A.</i>	158A (5)	1219-20	2012
Nishina S, Kosaki R, Yagihashi T, Azuma N, Okamoto N, Hatsukawa Y, Kurosawa K, Yamane T, Mizuno S, Tsuzuki K, Kosaki K.	Ophthalmic features of CHARGE syndrome with CHD7 mutations.	<i>Am J Med Genet A.</i>	158A (3)	514-8	2012

Ishikawa A, Enomoto K, Tominaga M, Saito T, Nagai JI, Furuya N, Ueno K, Ueda H, Masuno M, Kurosawa K.	Pure duplication of 19p13.3.	<i>Am J Med Genet A.</i>	61(9)	2300-4	2013
Nagase H, Ishikawa H, Kurosawa K, Furuya N, Itani Y, Yamanaka M.	Familial severe congenital diaphragmatic hernia: left herniation in one sibling and bilateral herniation in another.	<i>Congenit Anom (Kyoto)</i>	53(1)	54-7	2013
石川亜貴、榎本啓典、古谷憲孝、室谷浩二、朝倉由美、安達昌功、黒澤健司	CHARGE 症候群 26 例の臨床的検討	日本小児科学会雑誌	116	1357-1364	2012
Miyake N, Koshimizu E, Okamoto N, Mizuno S, Ogata T, Nagai T, Kosho T, Ohashi H, Kato M, Sasaki G, Mabe H, Watanabe Y, Yoshino M, Matsuishi T, Takanashi J, Shotelersuk V, Tekin M, Ochi N, Kubota M, Ito N, Ihara K, Hara T, Tonoki H, Ohta T, Saito K, Matsuo M, Urano M, Enokizono T, Sato A, Tanaka H, Ogawa A, Fujita T, Hiraki Y, Kitanaka S, Matsubara Y, Makita T, Taguri M, Nakashima M, Tsurusaki Y, Saitsu H, Yoshiura K, Matsumoto N, Niikawa N.	MLL2 and KDM6A mutations in patients with Kabuki syndrome.	<i>Am J Med Genet</i>	A 161	2234-43	2013
Takahashi M, Ohashi H.	Craniofacial and dental malformations in Costello syndrome: A detailed evaluation using multi-detector row computed tomography.	<i>Congenit Anom</i>	53	67-72	2013
Fujii K, Ohashi H, Suzuki M, Hatsuse H, Shiohama T, Uchikawa H, Miyashita T.	Frameshift mutation in the PTCH2 gene can cause nevoid basal cell carcinoma syndrome.	<i>Fam Cancer</i>	12(4)	611-4	2013
Mitsui N, Shimizu K, Nishimoto H, Mochizuki H, Iida M, Ohashi H	Patient with terminal 9 Mb deletion of chromosome 9p: Refining the critical region for 9p monosomy syndrome with trigonocephaly.	<i>Congenit Anom</i>	53	49-53	2013

Iijima K, Someya T, Ito S, Nozu K, Nakanishi K, Matsuoka K, Ohashi H, Nagata M, Kamei K, Sasaki S	Focal segmental glomerulosclerosis in patients with complete deletion of one WT1 allele.	<i>Pediatrics</i>	129	e1621-5	2012
Shimizu K, Wakui K, Kosho T, Okamoto N, Mizuno S, Itomi K, Hattori S, Nishio K, Samura O, Kobayashi Y, Kako Y, Arai T, Oh-Ishi T, Kawame H, Narumi Y, Ohashi H, Fukushima Y.	Microarray and FISH-based genotype-phenotype analysis of 22 Japanese patients with Wolf-Hirschhorn syndrome.	<i>Am J Med Genet A.</i>	164 A(3)	597-609	2014
Nishi E, Takamizawa S, Iio K, Yamada Y, Yoshizawa K, Hatata T, Hiroma T, Mizuno S, Kawame H, Fukushima Y, Nakamura T, Kosho T.	Surgical intervention for esophageal atresia in patients with trisomy 18.	<i>Am J Med Genet A.</i>	164 A(2)	324-30	2014
Hirai M, Muramatsu Y, Mizuno S, Kurahashi N, Kurahashi H, Nakamura M.	Developmental changes in mental rotation ability and visual perspective-taking in children and adults with Williams syndrome.	<i>Front Hum Neurosci</i>	11	856	2013
Okamoto N, Fujii T, Tanaka J, Saito K, Matsui T, Harada N.	A clinical study of patients with pericentromeric deletion and duplication within 16p12.2-p11.2.	<i>Am J Med Genet A.</i>	164	213-9	2014
Okamoto N, Ohmachi K, Shimada S, Shimojima K, Yamamoto T.	109 kb deletion of chromosome 4p16.3 in a patient with mild phenotype of Wolf-Hirschhorn syndrome	<i>Am J Med Genet A.</i>	161	1465-9	2013
Ohba C, Okamoto N, Murakami Y, Suzuki Y, Tsurusaki Y, Nakashima M, Miyake N, Tanaka F, Kinoshita T, Matsumoto N, Saitsu H.	PIGN mutations cause congenital anomalies, developmental delay, hypotonia, epilepsy, and progressive cerebellar atrophy.	<i>Neurogenetics.</i>	In press		2013
Tsurusaki Y, Okamoto N, Ohashi H, Mizuno S, Matsumoto N, Makita Y, Fukuda M, Isidor B, Perrier J, Aggarwal S, Dalal A, Al-Kindy A, Liebelt J, Mowat D, Nakashima M, Saitsu H, Miyake N, Matsumoto N.	Coffin-Siris syndrome is a SWI/SNF complex disorder.	<i>Clin Genet.</i>	In press		2013



Wada T, Ban H, Matsufuji M, Okamoto N, Enomoto K, Kurosawa K, Aida N.	Neuroradiologic Features in X-linked $\alpha$ -Thalassemia/Mental Retardation Syndrome.	<i>Am J Neuroradiology</i>	34	2034-8.	2013
Miyake N, Mizuno S, Okamoto N, Ohashi H, Shiina M, Ogata K, Tsurusaki Y, Nakashima M, Saitsu H, Niikawa N, Matsumoto N.	KDM6A Point Mutations Cause Kabuki Syndrome.	<i>Hum Mutat</i>	34	108-10	2013
Miyatake S, Murakami A, Okamoto N, Sakamoto M, Miyake N, Saitsu H, Matsumoto N.	A de novo deletion at 16q24.3 involving ANKRD11 in a Japanese patient with KBG syndrome.	<i>Am J Med Genet A.</i>	161	1073-7	2013
Shimada S, Okamoto N, Hirasawa K, Yoshii K, Tani Y, Sugawara M, Shimojima K, Osawa M, Yamamoto T.	Clinical manifestations of Xq28 functional disomy involving MECP2 in one female and two male patients.	<i>Am J Med Genet A.</i>	161	1779-85.	2013
Shimada S, Okamoto N, Nomura S, Fukui M, Shimakawa S, Sangu N, Shimojima K, Osawa M, Yamamoto T.	Microdeletions of 5.5 Mb (4q13.2-q13.3) and 4.1 Mb (7p15.3-p21.1) associated with a saethre-chotzen-like phenotype, severe intellectual disability, and autism.	<i>Am J Med Genet A.</i>	161	2078-83.	2013
Shimada S, Okamoto N, Ito M, Arai Y, Momosaki K, Togawa M, Maegaki Y, Sugawara M, Shimojima K, Osawa M, Yamamoto T.	MECP2 duplication syndrome in both genders.	<i>Brain Dev.</i>	35	411-9	2013
Iida A, Okamoto N, Miyake N, Nishimura G, Minami S, Sugimoto T, Nakashima M, Tsurusaki Y, Saitsu H, Shiina M, Ogata K, Watanabe S, Ohashi H, Matsumoto N, Ikegawa S.	Exome sequencing identifies a novel INPPL1 mutation in opsismodysplasia.	<i>J Hum Genet</i>	58	391-4	2013
Yokoo N, Marumo C, Nishida Y, Iio J, Maeda S, Nonaka M, Maihara T, Chujoh S, Katayama T, Sakazaki H, Matsumoto N, Okamoto N.	A case of Toriello-Carey syndrome with severe congenital tracheal stenosis.	<i>Am J Med Genet A.</i>	161	2291-3	2013

Koshimizu E, Miyatake S, Okamoto N, Nakashima M, Tsurusaki Y, Miyake N, Saitsu H, Matsumoto N.	Performance Comparison of Bench-Top Next Generation Sequencers Using Microdroplet PCR-Based Enrichment for Targeted Sequencing in Patients with Autism Spectrum Disorder.	<i>PLoS One.</i>	8	E74167	2013
Kodera H, Nakamura K, Osaka H, Maegaki Y, Haginoya K, Mizumoto S, Kato M, Okamoto N, Iai M, Kondo Y, Nishiyama K, Tsurusaki Y, Nakashima M, Miyake N, Hayasaka K, Sugahara K, Yuasa I, Wada Y, Matsumoto N, Saitsu H.	De Novo Mutations in SLC35A2 Encoding a UDP-Galactose Transporter Cause Early-Onset Epileptic Encephalopathy.	<i>Hum Mutat.</i>	34	1708-14	2013
Nakajima J, Okamoto N, Shiraishi J, Nishimura G, Nakashima M, Tsurusaki Y, Saitsu H, Kawashima H, Matsumoto N, Miyake N.	Novel FIG4 mutations in Yunis-Varon syndrome.	<i>J Hum Genet.</i>	58	822-4	2013
Ichikawa K, Kadoya M, Wada Y, Okamoto N.	Congenital disorder of glycosylation type Ic: report of a Japanese case.	<i>Brain Dev.</i>	35	586-9	2013
Miyake N, Kosho T, Matsumoto N	Ehlers-danlos syndrome associated with glycosaminoglycan abnormalities.	<i>Adv Exp Med Biol</i>	802	145-159	2014
Kosho T, Kuniba H, Tanikawa Y, Hashimoto Y, Sakurai H	Natural history and parental experience of children with trisomy 18 based on a questionnaire given to a Japanese trisomy 18 parental support group.	<i>Am J Med Genet Part A</i>	161A(7)	1531-1542	2013
Kosho T, Okamoto N, Ohashi H, Tsurusaki Y, Imai Y, Hibi-Ko Y, Kawame H, Homma T, Tanabe S, Kato M, Hiraki Y, Yamagata T, Yano S, Sakazume S, Ishii T, Nagai T, Ohta T, Niikawa N, Mizuno S, Kaname T, Naritomi K, Narumi Y, Wakui K, Fukushima Y, Miyatake S, Mizuguchi T, Saitsu H, Miyake N, Matsumoto N	Clinical correlations of mutations affecting six components of the SWI/SNF complex: detailed description of 21 patients and a review of the literature.	<i>Am J Med Genet Part A</i>	161A(6)	1221-1237	2013

Tsurusaki Y, Kosho T, Hatasaki K, Narumi Y, Wakui K, Fukushima Y, Doi H, Saitsu H, Miyake N, Matsumoto N	Exome sequencing in a family with an X-linked lethal malformation syndrome: clinical consequences of hemizygous truncating OFD1 mutations in male patients.	<i>Clin Genet</i>	83(2)	135-144	2013
Nitta H, Unoki M, Ichiyangi K, Kosho T, Shigemura T, Takahashi H, Velasco G, Francastel C, Picard C, Kubota T, Sasaki H	Three novel ZBTB24 mutations identified in Japanese and Cape Verdean type 2 ICF syndrome patients.	<i>J Hum Genet</i>	58(7)	455-460	2013
Tanaka K, Sekijima Y, Yoshida K, Tamai M, Kosho T, Sakurai A, Wakui K, Ikeda S, Fukushima Y	Follow-up nationwide survey on predictive genetic testing for late-onset hereditary neurological diseases in Japan.	<i>J Hum Genet</i>	58(8)	560-563	2013
古庄知己	遺伝カウンセリングロールプレイ実習～全人的医学教育としての取り組み～.	日本遺伝カウンセリング学会誌	34(1)	17-20	2013
古庄知己	デルマタン4-O-硫酸基転移酵素-1欠損に基づく新型エーラス・ダンロス症候群の発見と疾患概念の確立.	日本遺伝カウンセリング学会誌	34(1)	21-29	2013
古庄知己	18トリソミー児の調査を通じて	ネオネイタルケア	26(5)		2013
古庄知己	グリコサミノグリカンの異常と新型Ehlers-Danlos症候群(古庄型).	病理と臨床	31(8)	852-860	2013
古庄知己	18トリソミー症候群.	小児科臨床増刊号『臨床医が知っておきたい先天異常』	66	55-60	2013
古庄知己	4p-症候群, 5p-症候群.	周産期医学特集『染色体異常と先天異常症候群の診療ガイド』	43(3)	363-367	2013
Sugiura K, Takeichi T, Tanahashi K, Ito Y, Kosho T, Saida K, Uhara H, Okuyama R, Akiyama M	Lamellar ichthyosis in a collodion baby caused by CYP4F22 mutations in a non-consanguineous family outside the Mediterranean.	<i>J Dermatol Sci</i>	Epub ahead of print		

古庄知己	信州大学医学部附属病院遺伝子診療部の取り組み～小児科出身の臨床遺伝科医として思うこと .	日本遺伝カウンセリング学会誌	In press		
Takenouchi T, Shimizu A, Torii C, Kosaki R, Takahashi T, Saya H and Kosaki K	Multiple cafe' au lait spots in familial patients with MAP2K2 mutation.	<i>Am J Med Genet</i>	164	392-396	2014
Oshima H, Ishikawa T, Yoshida GJ, Naoi K, Maeda Y, Naka K, Ju X, Yamada Y, Minamoto T, Mukaida N, Saya H and Oshima M	TNF- /TNFR1 signaling promotes gastric tumorigenesis through induction of Nox1 and Gna14 in tumor cells.	<i>Oncogene</i>	印刷中		2013
DeBoer E, Azevedo R, Vega T, Brodtkin J, Akamatsu W, Okano H, Wagner G, Rasin MR.	Prenatal deletion of the RNA binding protein HuD disrupts postnatal cortical circuit maturation and behavior.	<i>The Journal of Neuroscience</i>		in press	2014
Bundo M, Toyoshima M, Ueda J, Nemoto-Miyake T, Sunaga F, Toritsuka M, Ikawa D, Kakita A, Okada Y, Akamatsu W, Kato M, Okano H, Kasai K, Kishimoto T, Nawa H, Yoshikawa T, Kato T, Iwamoto K:	Increased L1 Retrotransposition in the neuronal genome in Schizophrenia.	<i>Neuron</i>	81	306-313	2014
Kim C, Kim W, Lee H, Ji E, Choe YJ, Martindale JL, Akamatsu W, Okano H, Kim HS, Nam SW, Gorospe M, Lee EK	The RNA binding protein, HuD regulates autophagosome formation in pancreatic $\beta$ cells by promoting autophagy-related gene 5 expression.	<i>J Biol Chem.</i>	289	112-121	2013
Higurashi N, Uchida T, Christoph L, Misumi Y, Okada Y, Akamatsu W, Imaizumi Y, Zhang B, Nabeshima K, Mori M, Katsurabayashi S, Shirasaka S, Okano H and Hirose S	A human Dravet syndrome model from patient induced pluripotent stem cells.	<i>Molecular Brain</i>	6	19	2013

Veraitch O, Kobayashi T, Imaizumi Y, Akamatsu W, Sasaki T, Yamanaka , Amagai M, Okano H and Ohyama	Human induced pluripotent stem cell-derived ectodermal precursor cells contribute to hair follicle morphogenesis <i>in vivo</i> .	<i>J Invest Dermatol.</i>	133(6)	1479-88.	2013
増井徹	ヒトゲノム研究の規制について	<i>Organ Biology</i>	21	16 - 23	2014