研究成果の刊行に関する一覧表

辻　省次

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
Ishii A, Saito Y, Mitsui J, Ishiura H, Yoshimura J, Arai H, Yamashita S, Kimura S, Oguni H, Morishita S, Tsuji S, Sasaki M, Hirose S.	Identification of ATP1A3 mutations by exome sequencing as the cause of alternating hemiplegia of childhood in Japanese patients.	***PLOS One***	8	e56120	2013
Ichikawa Y, Ishiura H, Mitsui J, Takahashi Y, Kobayashi S, Takuma H, Kanazawa I, Doi K, Yoshimura J, Morishita S, Goto J, Tsuji S.	Exome analysis reveals a Japanese family with spinocerebellar ataxia, autosomal recessive 1.	***J. Neurol. Sci.***	331	158-60	2013
Mitsui J, Matsukawa T, Ishiura H, Fukuda Y, Ichikawa Y, Date H, Ahsan B, Nakahara Y, Momose Y, Takahashi Y, Iwata A, Goto J, Yamamoto Y, Komata M, Shirahige K, Hara K, Kakita A, Yamada M, Takahashi H, Onodera O, Nishizawa M, Takashima H, Kuwano R, Watanabe H, Ito M, Sobue G, Soma H, Yabe I, Sasaki H, Aoki M, Ishikawa K, Mizusawa H, Kanai K, Hattori T, Kuwabara S, Arai K, Koyano S, Kuroiwa Y, Hasegawa K, Yuasa T, Yasui K, Nakashima K, Ito H, Izumi Y, Kaji R, Kato T, Kusunoki S, Osaki Y, Horiuchi M, Kondo T, Murayama S, Hattori N, Yamamoto M, Murata M, Satake W, Toda T, Dürr A, Brice A, Filla A, Klockgether T, Wüllner U, Nicholson G, Gilman S, Shults CW, Tanner CM, Kukull WA, Lee V M-Y, Masliah E, Low PA, Sandroni P, Trojanowski JQ, Ozelius L, Foroud T, and Tsuji S.	Mutations of COQ2 in familial and sporadic multiple system atrophy.	***New Engl. J. Med.***	369	233-44	2013

Landouré G, Toro C, Zhu P-P, Johnson JO, Bricceno KV, Rinaldi C, Meilleur KG, Sangaré M, Diallo O, Pierson TM, Ishiura H, Tsuji S, Hein N, Fink JK, Stoll M, Nicholson G, Gonzalez M, Züchner S, Dürr A, Stevanin G, Biesecker LG, Accardi J, Landis D, Gahl WA, Traynor BJ, Blackstone C, Fischbeck KH, Burnett BG.	Hereditary spastic paraplegia type 43 (SPG43) is caused by mutation in C19ORF12.	***Human Mutation***	34	1357-60	2013
Isojima T, Doi K, Mitsui J, Oda Y, Tokuhiro E, Yasoda A, Yorifuji T, Horikawa R, Yoshimura J, Ishiura H, Morishita S, Tsuji S, and Kitanaka S.	A recurrent de novo FAM111A mutation causes Kenny–Caffey syndrome type 2.	***J Bone Mineral Res***			in press
Sasaki M, Ishii A, Saito Y, Morisada N, Iijima K, Takada S, Araki A, Tanabe Y, Arai H, Yamashita S, Ohashi T, Oda Y, Ichiseki H, Hirabayashi S, Yasuhara A, Kawawaki H, Kimura S, Shimono M, Narumiya M, Suzuki M, Yoshida T, Oyazato Y, Tsuneishi S, Ozasa S, Yokochi K, Dejima S, Akiyama T, Kishi N, Kira R, Ikeda T, Oguni H, Zhang B, Tsuji S and Hirose S.	Genotype–Phenotype Correlations in Alternating Hemiplegia of Childhood.	***Neurology***			in press

Takahashi Y, Fukuda Y, Yoshimura J, Toyoda A, Kurppa K, Moritoyo H, Belzil VV, Dion PA, Higasa K, Doi K, Ishiura H, Mitsui J, Date H, Ahsan B, Matsukawa T, Ichikawa Y, Moritoyo T, Ikoma M, Hashimoto T, Kimura F, Murayama S, Onodera O, Nishizawa N, Yoshida M, Atsuta N, Sobue G, JaCALS, Fifita JA, Williams KL, Blair IP, Nicholson GA, Gonzalez-Perez P, Brown, Jr.RH, Nomoto M, Elenius K, Rouleau GA, Fujiyama A, Morishita S, Goto J and Tsuji S.	ERBB4 Mutations that Disrupt the Neuregulin-ErbB4 Pathway Cause Amyotrophic Lateral Sclerosis Type 19.	***Am J Hum Genet***	93	900-5	2013
Doi K, et al.	Rapid detection of expanded short tandem repeats in personal genomics using hybrid sequencing.	***Bioinformatics***			in press
Yamada M, Tanaka M, Takagi M, Kobayashi S, Taguchi Y, Takashima S, Tanaka K, Touge T, Hatsuta H, Murayama S, Hayashi Y, Kaneko M, Ishiura H, Mitsui J, Astuta N, Sobue G, Shimozawa N, Inuzuka T, Tsuji S, and Hozumi I.	Evaluation of SLC20A2 mutations that cause idiopathic basal ganglia calcification in Japan.	***Neurol***			in press
Ishiura H, Takahashi Y, Hayashi T, Saito K, Furuya H, Watanabe M, Murata M, Suzuki M, Sugiura A, Sawai S, Shibuya K, Ueda N, Ichikawa Y, Kanazawa I, Goto J, Tsuji S.	Molecular epidemiology and clinical spectrum of hereditary spastic paraplegia in the Japanese population based on comprehensive mutational analyses.	***J. Hum. Genet.***			in press

松原洋一

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
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	*Am J Med Genet A*	161(10)	2600-3	2013
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.	*Am J Med Genet A*	161(9)	2234-43	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.	*Am J Hum Genet*	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.	*J Hum Genet.*	58(5)	259-66	2013
Aoki Y, Matsubara Y.	Ras/MAPK syndromes and childhood hemato-oncological diseases.	*Int J Hematol.*	97(1)	30-6	2013
Asano M, Fujimura T, Wakusawa C, Aoki Y, Matsubara Y, Aiba S.	A case of almost unilateral focal dermal hypoplasia resulting from a novel mutation in the PORCN gene.	*Acta Derm Venereol.*	93(1)	120-1	2013

奥山虎之

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
Okuyama T, Yotsumoto J, Funato Y.	Survey of second-trimester maternal serum screening in Japan.	*J Obstet Gynaecol Res.*	39	942-947	2013
Tajima G, Sakura N, Kosuga M, Okuyama T, Kobayashi M	Effects of idursulfase enzyme replacement therapy for Mucopolysaccharidosis type II when started in early infancy: comparison in two siblings.	*Mol Genet Metab*	108	172-177	2013
Niizeki H, Shiohama A, Sasaki T, Seki A, Kabashima K, Otsuka A, Takeshita M, Hirakiyama A, Okuyama T, Tanese K, Ishiko A, Amagai M, Kudoh J	The novel SLCO2A1 heterozygous missense mutation p.E427K and nonsense mutation p.R603* in a female patient with pachydermoperiostosis with an atypical phenotype.	*Br J Dermatol*	doi:10.1111/bjd.12790.		2013
Okuyama T, Yotsumoto J, Funato Y	Survey of second-trimester maternal serum screening in Japan.	*J Obstet Gynaecol Res.*	39	942-947	2013
Tajima G, Sakura N, Kosuga M, Okuyama T, Kobayashi M.	Effects of idursulfase enzyme replacement therapy for Mucopolysaccharidosis type II when started in early infancy: comparison in two siblings.	*Mol Genet Metab.*	108	172-177	2013

Niizeki H, Shiohama A, Sasaki T, Seki A, Kabashima K, Otsuka A, Takeshita M, Hirakiyama A, Okuyama T, Tanese K, Ishiko A, Amagai M, Kudoh J.	The novel SLCO2A1 heterozygous missense mutation p.E427K and nonsense mutation p.R603* in a female patient with pachydermoperiostosis with an atypical phenotype.	*Br J Dermatol.*		Dec16.doi:10.1111/bjd.12790.	2013
後藤由紀、柿島裕樹、藤直子、渡辺靖、小関満、松林守、木田和宏、小須賀基通、奥山虎之	ポンペ病を対象とした新生児マススクリーニングの運用	*日本マス.スクリーニング学会誌*	23	251-55	2013

後藤雄一

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
Goto M, Komaki H, Saito T, Saito Y, Nakagawa E, Sugai K, Sasaki M, Nishino I, Goto Y.	MELAS phenotype associated with m.3302A>G mutation in mitochondrial tRNA(Leu(UUR)) gene.	*Brain Dev.*	36	180-182	2014
後藤雄一	ミトコンドリア病の診断と治療	*内分泌・糖尿病・代謝内科*	37	481-486	2013

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
後藤雄一	ミトコンドリア病		*内科学、第10版*	朝倉書店	東京	2013	2339-2342

斎藤加代子

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
Kondo E, Nishimura T, Kosho T, Inaba Y, Matsuhashi S, Ishida T, Baba A, Koike K, Nishino I, Nonaka I, Furukawa T, Saito K.	Recessive *RYR1* Mutations in a Patient With Severe Congenital Nemaline Myopathy With Ophthalomoplegia Identified Through Massively Parallel Sequencing.	*Am J Med Genet A*	158(A)	772-778	2012

宮地勇人

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
Tomizawa D, Tawa A, Watanabe T, Saito AM, Kudo K, Taga T, Iwamoto S, Shimada A, Terui K, Moritake H, Kinoshital A, Takahashi H, Nakayama H, Koh K, Kigasawa H, Kosaka Y, Miyachi H, Horibe K, Nakahata T, Adachi S.	Excess treatment reduction including anthracyclines results in, higher incidence of relapse in core binding factor acute myeloid, leukemia in children.	*Leukemia*	27	2413-16	2013
Tanaka Y, Matsushita H, Tanaka Y, Maruki Y, Hayashi F, Kondo T, Asai S, Miyachi H.	Elimination of interference by lipids in the low WBC mode in the automated hematology analyzer XN-2000.	*Intern J Lab Hematol*		in press	2014
Tanaka Y, Matsushita H, Tanaka Y, Maruki Y, Kondo T, Asai S, Miyachi H.	Evaluation of the body fluid mode of automated hematology analyzer XN-Series for extremely low peripheral White Blood Cell Counts.	*Intern J Lab Hematol*		in press	2014
Tanaka Y, Tanaka Y, Gondo K, Maruki Y, Kondo T,Asai S, Matsushita H, Miyachi H.	Performance evaluation of platelet counting by novel fluorescent dye staining in the automated hematology analyzers XN-series.	*J Clin Lab Analysis*		in press	2014
Asai S, Okami K,Nakamura N, Shiraishi S, Sugimoto R, Damdinsuren A, Sato S, Matsushita H, Suzuki Y. Miyachi H.	Localized or diffuse lesions of the submandibular glands in IgG4-related disease in association with differential organ involvement.	*J Ultrasound Med*	32	731-736	2013

福嶋義光

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年

Narumi Y, Min BJ, Shimizu K, Kazukawa I, Sameshima K, Nakamura K, Kosho T, Rhee Y, Chung YS, Kim OH, Fukushima Y, Park WY, Nishimura G.	Clinical consequences in truncating mutations in exon 34 of NOTCH2: report of six patients with Hajdu-Cheney syndrome and a patient with serpentine fibula polycystic kidney syndrome.	*Am J Med Genet A*	161A	518-526	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.	*Am J Med Genet A.*	161A	1221-1237	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.	*J Hum Genet*	58	560-563	2013
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.	*Am J Med Genet A*	164A	324-330	2014
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.	*Am J Med Genet A*	164 (3)	597-609	2014

古川洋一

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年

Yamamoto S, Ebihara Y, Mochizuki S, Kawakita T,Kato S, Ooi J, Tojo A, Yusa N, Furukawa Y, Oyaizu N, Watanabe J, Sato K, Kimura F, and Tsuji K.	Quantitative PCR detection of CEP110-FGFR1 fusion gene in a patient with 8p11 syndrome.	*Leukemia & Lymphoma,*	54(9)	2068-9	2013
Shigeyasu K, Tanakaya K, Nagasaka T, Aoki H,Fujiwara T, Sugano K, Ishikawa H, Yoshida T, Moriya Y, Furukawa Y, Goel A, Takeuchi H.	Early detection of metachronous bile duct cancer in Lynch syndrome: report of a case.	*Surg Today*	Jul 31	Epub	2013

難波栄二

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
Adachi K	Expansion of Genetic Testing in the Division of Functional Genomics, Research Center for Bioscience and Technology, Tottori University from 2000 to 2013	*Yonago Acta Med*			in press
Chiba Y, Komori H, Takei S, Hasegawa-Ishii S, Kawamura N, Adachi K, Nanba E, Hosokawa M, Enokido Y, Kouchi Z, Yoshida F, Shimada A	Niemann-Pick disease type C1 predominantly involving the frontotemporal region, with cortical and brainstem Lewy bodies: An autopsy case	*Neuropathology*	34(1)	49-57	2014
Fujimoto S, Manabe Y, Fujii D, Kozai Y, Matsuzono K, Takahashi Y, Narai H, Omori N, Adachi K, Nanba E, Nishino I, Abe K	A novel mutation of the GAA gene in a patient with adult-onset Pompe disease lacking a disease-specific pathology	*Intern Med*	52(21)	2461-4	2013
Sekijima Y, Nakamura K, Kishida D, Narita A, Adachi K, Ohno K, Nanba E, Ikeda S	Clinical and serial MRI findings of a sialidosis type I patient with a novel missense mutation in the NEU1 gene	*Intern Med*	52(1)	119-24	2013

秋山真志

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
Sugiura K, Takemoto	The majority of generalize	*J Invest D*	133巻11	2514-2521	2013

A, Yamaguchi M, Takahashi H, Shoda Y, Mitsuma T, Tsuda K, Nishida E, Togawa Y, Nakajima K, Sakakibara A, Kawachi S, Shimizu M, Ito Y, Takeichi T, Kono M, Ogawa Y, Muro Y, Ishida-Yamamoto A, Sano S, Matsue H, Morita A, Mizutani H, Iizuka H, Muto M, Akiyama M.	d pusular psoriasis without psoriasis vulgaris is caused by deficiency of interleukin-36 receptor antagonist.	*ermatol*	号		
Kono M, Sugiura K, Suganuma M, Hayashi M, Takama H, Suzuki T, Matsunaga K, Tomita Y, Akiyama M.	Whole-exome sequencing identifies ADAM10 mutations as a cause of reticulate acropigmentation of Kitamura, a clinical entity distinct from Dowling-Degos disease.	*Hum Mol Genet*	22巻17号	3524-3533	2013
Ogawa Y, Takeichi T, Kono M, Hamajima N, Yamamoto T, Sugiura K, Akiyama M.	Revertant mutation releases confined lethal mutation, opening Pandora's box: a novel genetic pathogenesis.	*PLoS Genetics*	In press		2014

野口佳裕

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
Nishio A, Noguchi Y, Sato T, Naruse TK, Kimura A, Takagi A, Kitamura K	A *DFNA5* mutation identified in Japanese families with autosomal dominant hereditary hearing loss	*Annals of Human Genetics*	78	83-91	2014
野口佳裕、伊藤　卓、川島慶之、西尾綾子、本田圭司、喜多村健	前庭水管拡大症を伴う*SLC26A4*, *ATP6V1B1*, *SIX1*変異例の聴平衡覚所見の検討	*Equilibrium Research*	72	97-106	2013
川島慶之,野口佳裕	平衡覚と遺伝子	*JOHNS*			印刷中
本田圭司,野口佳裕,加藤智史,奥野秀次,喜多村　健	網羅的解析により診断された耳小骨奇形を合併したミトコンドリア3243変異例	*Otology Japan*	23	227-232	2013

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

野口佳裕	感覚器疾患　29. 難聴	泉孝英	*ガイドライン外来診療2014*	日経メディカル開発	東京		印刷中

森田啓行

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
Morita H	Human genomics in cardiovascular medicine-implications and perspectives-	*Circ J*	77(4)	876-885	2013
森田啓行、山田奈美恵、小室一成	肥大型心筋症の遺伝子診断：推進に向けての方策.	*日内学誌*	102(5)	1233-1242	2013
Morita H, Komuro I	A novel channelopathy in pulmonary arterial hypertension.	*N Engl J Med*	369(22)	2161-2162	2013
Morita H	Genetic variants and dilated cardiomyopathy - To be or not to be causative: Is that the question? -	*Circ J*	77(12)	2879-2880	2013
森田啓行	遺伝子から心筋症をみる.	*日内会誌*	103(2)	285-292	2014

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
森田啓行	心筋症	矢崎義雄総編集	内科学第10版	朝倉書店	東京	2013	422-424
森田啓行、永井良三	循環器疾患と遺伝子異常	小川聡総編集	内科学書改訂第8版	中山書店	東京	2013	293-296

小崎健次郎

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年

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.	*Eur J Med Genet*	56(1)	50-53	2013
Hirasawa A, Masuda K, Akahane T, Tsuruta T, Banno K, Makita K, Susumu N,Jinno H, Kitagawa Y, Sugano K, Kosaki K, Aoki D.	Experience of Risk-reducing Salpingo-oophorectomy for a BRCA1 Mutation Carrier and Establishment of a System Performing a Preventive Surgery for Hereditary Breast and Ovarian Cancer Syndrome in Japan: Our Challenges for the Future.	*Jpn J Clin Oncol*	43(5)	515-519	2013
Yamazaki F, Osumi T, Kosaki K, Mikami S, Hirato J, Shimada H.	Large Congenital Melanocytic Nevi With Atypical Teratoid/Rhabdoid Tumor.	*Pediatr Blood Cancer*	60(7)	1240-1241	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.	*Am J Med Genet A*	161(5)	1154-1157	2013
Komoike Y, Matsuoka M, Kosaki K.	Potential Teratogenicity of Methimazole: Exposure of zebrafish embryos to Methimazole causes similar developmental Anomalies to Human Methimazole Embryopathy.	*Birth Defects Res B Dev Reprod Toxicol*	98(3)	222-229	2013
Takenouchi T, Kosaki R, Torii C, Kosaki K.	Daytime somnolence in an adult with Smith-Magenis syndrome	*Am J Med Genet*	161(7)	1803-1805	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.	*Am J Med Genet A*	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.	*J Hum Genet*	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.	*Am J Med Genet A*	161(12)	3057-3062	2013
Kosaki R, Takenouchi T, Takeda N, Kagami M, Nakabayashi K, Hata K, Kosaki K.	Somatic CTNNB1 mutation in hepatoblastoma from a patient withSimpson-Golabi-Behmel syndrome and germline GPC3 mutation.			In press	
Takenouchi T, Shimizu A, Torii C, Kosaki R, Takahashi T, Saya H, Kosaki K.	Multiple café au lait spots in familial patients with MAP2K2 mutation.	*Am J Med Genet A*	164(2)	392-396	2014
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.	*Orphanet J Rare Dis*	8(1)	172	2014
Hirasawa A, Masuda K, Akahane T, Ueki A, Yokota M, Tsuruta T, Nomura H, Kataoka F, Tominaga E, Banno K, Makita K, Susumu N, Sugano K, Kosaki K, Kameyama K, Aoki D.	Family History and BRCA1/BRCA2 Status Among Japanese Ovarian Cancer Patients and Occult Cancer in a BRCA1 Mutant Case.	*Jpn J Clin Onco*	44(1)	49-56	2014
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.	*Am J Med Genet*	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.Pituitary.			in press	2013
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.	*Am J Med Genet A.*	164(2)	456-60	2014

青木正志

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
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	*J Hum Genet*	58(5)	259-66	2013
Takahashi T, Aoki M, et al.,	Clinical features and a mutation with late onset of limb girdle muscular dystrophy 2B.	*J Neurol Neurosurg Psychiatry*	84	433-40	2013

松田文彦

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年

Plenge, R. M., Greenberg, J. D., Mangravite, L. M., Derry, J. M., Stahl, E. A., Coenen, M. J., Barton, A., Padyukov, L., Klareskog, L., Gregersen, P. K., Mariette, X., Moreland, L. W., Bridges, S. L. Jr, de Vries, N. Huizinga, T. W. Guchelaar, H. J., International Rheumatoid Arthritis Consortium (INTERACT), Friend, S. H. and Stolovitzky, G.	Crowdsourcing genetic prediction of clinical utility in the Rheumatoid Arthritis Responder Challenge.	*Nat. Genet.*	45	468-469	2013
Terao, C., Yoshifuji, H., Kimura, A., Matsumura, T., Ohmura, K., Takahashi, M., Shimizu, M., Kawaguchi, T., Chen, Z., Naruse, T. K., Sato-Otsubo, A., Ebana, Y., Maejima, Y., Kinoshita, H., Murakami, K., Kawabata, D., Wada, Y., Narita, I., Tazaki, J., Kawaguchi, Y., Yamanaka, H., Yurugi, K., Miura, Y., Maekawa, T., Ogawa, S., Komuro, K., Nagai, R., Yamada, R., Tabara, Y., Isobe, M., Mimori, T. and Matsuda, F.	Two susceptibility loci to Takayasu arteritis reveal a synergistic role of the IL12B and HLA-B regions in a Japanese population.	*Am. J. Hum. Genet*	93	289-297	2013
Terao, C., Yoshifuji, H., Ohmura, K., Murakami, K., Kawabata, D., Yurugi, K., Tazaki, J., Kinoshita, H., Kimura, A., Akizuki, M., Kawaguchi, Y., Yamanaka, H., Miura, Y., Maekawa, T., Saji,	Association of Takayasu arteritis with HLA-B*67:01 and two amino acids in HLA-B protein.	*Rheumatol.* (Oxford)	52	1769-1774	2013

H., Mimori, T. and Matsuda, F.					
Terao, C., Bayoumi, N., McKenzie, C. A., Zelenika, D., Muro, S., Mishima, M.; The Nagahama Cohort Research Group, Connell, J. M., Vickers, M. A., Lathrop, G. M., Farrall, M., Matsuda, F. and Keavney, B. D.	Quantitative variation in plasma angiotensin-I converting enzyme activity shows allelic heterogeneity in the ABO blood group locus.	*Ann. Hum. Genet*	77	465-471	2013
Yamakawa, N., Fujimoto, M., Kawabata, D., Terao, C., Nishikori, M., Nakashima, R., Imura, Y., Yukawa, N., Yoshifuji, H., Ohmura, K., Fujii, T., Kitano, T., Kondo, T., Yurugi, K., Miura, Y., Maekawa, T., Saji, S., Takaori-Kondo, A., Matsuda, F., Haga, H. and Mimori, T.	A clinical, pathological and genetic characterization of methotrexate- associated lymphoproliferative disorders.	*J. Rheumatol.*	41	293-299	2014
Tanaka, K., Terao, C., Ohmura, K., Takahashi, M., Nakashima, R., Imura, Y., Yoshifuji, H., Yukawa, N., Usui, T., Fujii, T., Mimori, T. and Matsuda, F.	Significant association between *CYP3A5* polymorphism and blood concentration of tacrolimus in patients with connective tissue diseases.	*J. Hum. Genet.*	59	107-109	2014

Okada, Y., Wu, D., Trynka, G., Raj, T., Terao, C., Ikari, K., Kochi, Y., Ohmura, K., Suzuki, A., Yoshida, S., Graham, R. R., Manoharan, A., Ortmann, W., ……Kawaguchi, T., ……Kamatani, Y., ……Taniguchi, A., Yamada, R., Kubo, M., ……De Jager, P. L., Franke, L., Visscher, P. M., Brown, M. A., Yamanaka, H., Mimori, T., Takahashi, A., Xu, H., Behrens, T. W., Siminovitch, K. A., Momohara, S., Matsuda, F., Yamamoto, K. and Plenge, R. M.	Genetics of rheumatoid arthritis contributes to biology and drug discovery.	*Nature*	506	376-381	2014

梅澤明弘

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
Migita O, Maehara K, Kamura H, Miyakoshi K, Tanaka M, Morokuma S, Fukushima K, Shimamoto T, Saito S, Sago H, Nishihama K, Abe K, Nakabayashi K, Umezawa A, Okamura K, Hata K	Compilation of copy number variants identified in phenotypically normal and parous Japanese women	*J Hum Genets*		in press	2014

松本直道

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年

Tsurusaki Y, Yonezawa R, Furuya M, Nishimura G, Pooh RK, Nakashima M, Saitsu H, Miyake N, Saito S, and Matsumoto N.	Whole exome sequencing revealed causative biallelic IFT122 mutations in a family with CED1 and recurrent pregnancy loss.	*Clin Genet*		in press	

岡本伸彦

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
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	*Am J Med Genet A*	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.	*Neurogenetics*		Nov 20. [Epub ahead of print]	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.	*Clin Genet.*		Jul 1. doi: 10.1111/cge.12225. [Epub ahead of print]	2013
Wada T, Ban H, Matsufuji M, Okamoto N, Enomoto K, Kurosawa K, Aida N.	Neuroradiologic Features in X-linked α-Thalassemia/Mental Retardation Syndrome.	*AJNR Am J Neuroradiol.*	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	*Hum Mutat*	34	108-10	2013
Kosho T, Okamoto N, Ohashi H, Tsurusaki Y, Imai Y, Hi	Clinical correlations of mutations affecting six components of the SWI/S	*Am J Med Genet A.*	161	1221-37	2013

bi-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.	NF complex: Detailed description of 21 patients and a review of the literature.				
Hitomi Yatsuki, Ken Higashimoto, Kosuke Jozaki, Kayoko Koide, Junichiro Okada, Yoriko Watanabe, Nobuhiko Okamoto, Yoshinobu Tsuno, Yoko Yoshida, Kazutoshi Ueda, Kenji Shimizu , Hirofumi Ohashi, Tsunehiro Mukai, Hidenobu Soejima	Novel mutations of CDKN1C in Japanese patientswith Beckwith-Wiedemann syndrome.	*Genes and Genetics*			
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.	*Am J Med Genet A*	161A	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.	*Am J Med Genet A*	161A	1179-85	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.	*Am J Med Genet A*	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.	*Brain Dev*	35	411-9	2013
Iida A, Okamoto N, Miyake N, Nishimura G, Minami S, Sugimoto T, Nakashima M, Tsurusaki Y,	Exome sequencing identifies a novel INPPL1 mutation in opsismodysplasia.	*J Hum Gene*	58	391-4	2013

Saitsu H, Shiina M, Ogata K, Watanabe S, Ohashi H, Matsumoto N, Ikegawa S.					
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.	*Am J Hum Genet.*	93	173-80	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.	*Am J Med Genet A*	161	2291-3	2013
Okumura A, Hayashi M, Shimojima K, Ikeno M, Uchida T, Takanashi JI, Okamoto N, Hisata K, Shoji H, Saito A, Furukawa T, Kishida T, Shimizu T, Yamamoto T.	Whole-exome sequencing of a unique brain malformation with periventricular heterotopia, cingulate polymicrogyria and mid brain tectal hyperplasia.	*Neuropathology. Neuropathology*	33	533-60	2013
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, Mat	MLL2 and KDM6A mutations in patients with Kabuki syndrome.	*Am J Med Genet A*	161	2234-43	2013

subara Y, Makita T, Taguri M, Nakashima M, Tsurusaki Y, Saitsu H, Yoshiura K, Matsumoto N, Niikawa N.					
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.	*Hum Mutat*	34	1708-14	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.	*PLoS One*	8	e74167	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.	*J Hum Genet*	58	822-4	2013
Ichikawa K, Kadoya M, Wada Y, Okamoto N.	Congenital disorder of glycosylation type Ic: report of a Japanese case.	*Brain Dev*	35	586-9	2013