

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	<i>Am J Med Genet A</i>	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, Takahashi 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, Saito H, Yoshiura K, Matsumoto N, Niikawa N	MLL2 and KDM6A mutations in patients with Kabuki syndrome.	<i>Am J Med Genet A</i>	161(9)	2234-43	2013
Aoki Y, Niihori T, Banjo T, Okamoto N, Mizuno S, Kurosawa K, Ogata T, Takada F, Yanagisawa M, Ando T, Hoshikawa 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

Aoki Y, Matsubara Y.	Ras/MAPK syndromes and childhood hemato-oncological diseases.	<i>Int J Hematol</i>	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.	<i>Acta Derm Venereol.</i>	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
<u>Okuyama T</u> , Yotsumoto J, Funato Y.	Survey of second-trimester maternal serum screening in Japan.	<i>J Obstet Gynaecol Res</i>	39	942-947	2013
Tajima G, Sakura N, Kosuga M, <u>Okuyama T</u> , Kobayashi M	Effects of idursulfase enzyme replacement therapy for Mucopolysaccharidosis type II when started in early infancy: comparison in two siblings.	<i>Mol Genet Metab</i>	108	172-177	2013
Niizeki H, Shiohama A, Sasaki T, Seki A, Kabashima K, Otsuka A, Takeshita M, Hirakiyama A, <u>Okuyama T</u> , 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.	<i>Br J Dermatol</i>	doi:10.1111/bjd.12790.		2013
<u>Okuyama T</u> , Yotsumoto J, Funato Y	Survey of second-trimester maternal serum screening in Japan.	<i>J Obstet Gynaecol Res</i>	39	942-947	2013
Tajima G, Sakura N, Kosuga M, <u>Okuyama T</u> , Kobayashi M.	Effects of idursulfase enzyme replacement therapy for Mucopolysaccharidosis type II when started in early infancy: comparison in two siblings.	<i>Mol Genet Metab</i>	108	172-177	2013
Niizeki H, Shiohama A, Sasaki T, Seki A, Kabashima K, Otsuka A, Takeshita M, Hirakiyama A, <u>Okuyama T</u> , 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.	<i>Br J Dermatol</i>		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、 <u>奥山虎之</u>	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	<i>日本マススクリーニング学会誌</i>	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, <u>Goto Y.</u>	MELAS phenotype associated with m.3302A>G mutation in mitochondrial tRNA(Leu(UUR)) gene.	<i>Brain Dev</i>	36	180-182	2014
後藤雄一	ミトコンドリア病の診断と治療	<i>内分泌・糖尿病・代謝内科</i>	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		<i>内科学、第10版</i>	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 <i>RYR1</i> Mutations in a Patient With Severe Congenital Nemaline Myopathy With Ophthalmoplegia Identified Through Massively Parallel Sequencing.	<i>Am J Med Genet A</i>	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, Kinoshita A, Takahashi H, Nakayama H, Koh K, Kigasawa H, Kosaka Y, <u>Miyachi H</u> , 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.	<i>Leukemia</i>	27	2413-16	2013
Tanaka Y, Matsushita H, Tanaka Y, Maruki Y, Hayashi F, Kondou T, Asai S, <u>Miyachi H</u> .	Elimination of interference by lipids in the low WBC mode in the automated hematology analyzer XN-2000.	<i>Intern J Lab Hematol</i>			in press

Tanaka Y, Matsushita H, Tanaka Y, Maruki Y, Kondo T, Asai S, <u>Miyachi H.</u>	Evaluation of the body fluid mode of automated hematology analyzer XN-Series for extremely low peripheral White Blood Cell Counts.	<i>Intern J Lab Hematol</i>			in press
Tanaka Y, Tanaka Y, Gondo K, Maruki Y, Kondo T, Asai S, Matsushita H, <u>Miyachi H.</u>	Performance evaluation of platelet counting by novel fluorescent dye staining in the automated hematology analyzers XN-series.	<i>J Clin Lab Anal</i>			in press
Asai S, Okami K, Nakamura N, Shiraishi S, Sugimoto R, Damdinsuren A, Sato S, Matsushita H, Suzuki Y. <u>Miyachi H.</u>	Localized or diffuse lesions of the submandibular glands in IgG4-related disease in association with differential organ involvement.	<i>J Ultrasound Med</i>	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.	<i>Am J Med Genet A</i>	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.	<i>Am J Med Genet A</i>	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.	<i>J Hum Genet</i>	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, Koshio T..	Surgical intervention for esophageal atresia in patients with trisomy 18.	<i>Am J Med Genet A</i>	164A	324-330	2014
Shimizu K, Wakui K, Koshio T, Okamoto N, Mizuno S, Itomi K, Hattori S, Nishio K, Samura O, Kobayashi Y, Kako Y, Arai T, Ohishi 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(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.	<i>Leukemia & Lymphoma</i>	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.	<i>Surg Today</i>	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	<i>Yonago Acta Med</i>			in press

Chiba Y, Komori H, T akei S, Hasegawa-Ishii S, Kawamura N, Ada chi K, Nanba E, Hoso kawa M, Enokido Y, Kouchi Z, Yoshida F, Shimada A	Niemann-Pick disease type C1 predominantly involving the fro ntotemporal region, with cortic al and brainstem Lewy bodies: An autopsy case	<i>Neuropathology</i>	34(1)	49-57	2014
Fujimoto S, Manabe Y, Fujii D, Kozai Y, Matsuzono K, Takahas hi 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-on set Pompe disease lacking a disease-specific pathology	<i>Intern Med</i>	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 finding s of a sialidosis type I patient with a novel missense mutati on in the NEU1 gene	<i>Intern Med</i>	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 A, Yamaguchi M, Taka hashi H, Shoda Y, Mit suma T, Tsuda K, Nish ida E, Togawa Y, Nak ajima K, Sakakibara A, Kawachi S, Shimizu M, Ito Y, Takeichi T, Kono M, Ogawa Y, M uro Y, Ishida-Yamamot o A, Sano S, Matsue H, Morita A, Mizutani H, Iizuka H, Muto M, Akiyama M.	The majority of generalized pus ular psoriasis without psoriasis vulgaris is caused by deficiency of interleukin-36 receptor antag onist.	<i>J Invest Dermatol</i>	133(11)	2514-2521	2013
Kono M, Sugiura K, S uganuma M, Hayashi M, Takama H, Suzuki T, Matsunaga K, Tom ita Y, Akiyama M.	Whole-exome sequencing identif ies ADAM10 mutations as a ca use of reticulate acropigmentatio n of Kitamura, a clinical entity distinct from Dowling-Degos d isease.	<i>Hum Mol Genet</i>	22(17)	3524-3533	2013
Ogawa Y, Takeichi T, Kono M, Hamajima N, Yamamoto T, Sugiura K, Akiyama M.	Revertant mutation releases conf ined lethal mutation, opening Pa ndora's box: a novel genetic pat hogenesis.	<i>PLoS Genet ics</i>	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, <u>Noguchi Y</u> , Sato T, Naruse TK, Kimura A, Takagi A, Kitamura K	A <i>DFNA5</i> mutation identified in Japanese families with autosomal dominant hereditary hearing loss	<i>Annals of Human Genetics</i>	78	83-91	2014
<u>野口佳裕</u> , 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 <i>SLC26A4</i> , <i>ATP6V1B1</i> , <i>SIX1</i> 変異例の聴平衡覚所見の検討	<i>Equilibrium Research</i>	72	97-106	2013
川島慶之, <u>野口佳裕</u>	平衡覚と遺伝子	<i>JOHNS</i>			印刷中
本田圭司, <u>野口佳裕</u> , 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	<i>Otology Japan</i>	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
<u>野口佳裕</u>	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
<u>Morita H</u>	Human genomics in cardiovascular medicine-implications and perspectives-	<i>Circ J</i>	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.	<i>日内学誌</i>	102(5)	1233-1242	2013
<u>Morita H</u> , Komuro I	A novel channelopathy in pulmonary arterial hypertension.	<i>N Engl J Med</i>	369(22)	2161-2162	2013
<u>Morita H</u>	Genetic variants and dilated cardiomyopathy – To be or not to be causative: Is that the question? –	<i>Circ J</i>	77(12)	2879-2880	2013
<u>森田啓行</u>	遺伝子から心筋症をみる.	<i>日内会誌</i>	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, <u>Kosaki K.</u>	Concurrent deletion of BMP4 and OTX2 genes, two master genes in ophthalmogenesis.	<i>Eur J Med Genet</i>	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, <u>Kosaki K.</u> , 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.	<i>Jpn J Clin Oncol</i>	43(5)	515-519	2013
Yamazaki F, Osumi T, <u>Kosaki K.</u> , Mikami S, Hirato J, Shimada H.	Large Congenital Melanocytic Nevi With Atypical Teratoid/Rhabdoid Tumor.	<i>Pediatr Blood Cancer</i>	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.	<i>Am J Med Genet A</i>	161(5)	1154-1157	2013
Komoike Y, Matsuka M, <u>Kosaki K.</u>	Potential Teratogenicity of Methimazole: Exposure of zebrafish embryos to Methimazole causes similar developmental Anomalies to Human Methimazole Embryopathy.	<i>Birth Defects Res B Dev Reprod Toxicol</i>	98(3)	222-229	2013
Takenouchi T, Kosaki R, Torii C, <u>Kosaki K.</u>	Daytime somnolence in an adult with Smith-Magenis syndrome	<i>Am J Med Genet</i>	161(7)	1803-1805	2013

<p>Takenouchi T, Saito H, Maruoka R, Oishi N, Torii C, Maeda J, Takahashi T, <u>Kosaki K</u>.</p>	<p>Severe obstructive sleep apnea in Loeys-Dietz syndrome successfully treated using continuous positive airway pressure.</p>	<p><i>Am J Med Genet A</i></p>	<p>161(7)</p>	<p>1733-1736</p>	<p>2013</p>
<p>Hirasawa A, Zama T, Akahane T, Nomura H, Kataoka F, Saito K, Okubo K, Tominaga E, Makita K, Susumu N, <u>Kosaki K</u>, Tanigawara Y, Aoki D.</p>	<p>Polymorphisms in the UGT1A1 gene predict adverse effects of irinotecan in the treatment of gynecologic cancer in Japanese patients.</p>	<p><i>J Hum Genet</i></p>	<p>58(12)</p>	<p>794-798</p>	<p>2013</p>
<p>Takenouchi T, Hida M, Sakamoto Y, Torii C, Kosaki R, Takahashi T, <u>Kosaki K</u>.</p>	<p>Severe congenital lipodystrophy and a progeroid appearance: Mutation in the penultimate exon of FBN1 causing a recognizable phenotype.</p>	<p><i>Am J Med Genet A</i></p>	<p>161(12)</p>	<p>3057-3062</p>	<p>2013</p>
<p>Kosaki R, Takenouchi T, Takeda N, Kagami M, Nakabayashi K, Hata K, <u>Kosaki K</u>.</p>	<p>Somatic CTNNB1 mutation in hepatoblastoma from a patient with Simpson-Golabi-Behmel syndrome and germline GPC3 mutation.</p>				<p>In press</p>
<p>Takenouchi T, Shimizu A, Torii C, Kosaki R, Takahashi T, Saya H, Kosaki K.</p>	<p>Multiple café au lait spots in familial patients with MAP2K2 mutation.</p>	<p><i>Am J Med Genet A</i></p>	<p>164(2)</p>	<p>392-396</p>	<p>2014</p>
<p>Mutai H, Suzuki N, Shimizu A, Torii C, Namba K, Morimoto N, Kudoh J, Kagawa K, Kosaki K, Matsunaga T.</p>	<p>Diverse spectrum of rare deafness genes underlies early-childhood hearing loss in Japanese patients: a cross-sectional, multi-center next-generation sequencing study.</p>	<p><i>Orphanet J Rare Dis</i></p>	<p>8(1)</p>	<p>172</p>	<p>2014</p>
<p>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.</p>	<p>Family History and BRCA1/BRCA2 Status Among Japanese Ovarian Cancer Patients and Occult Cancer in a BRCA1 Mutant Case.</p>	<p><i>Jpn J Clin Oncol</i></p>	<p>44(1)</p>	<p>49-56</p>	<p>2014</p>

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, Nizeki H, Kabashima K, Mitsunashi Y, Amagai M.	Mutations in SERPINB7, Encoding a Member of the Serine Protease Inhibitor Superfamily, Cause Nagashima-type Palmoplantar Keratosis.	<i>Am J Med 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.Pituitary.				In press
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

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	<i>J Hum Genet</i>	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.	<i>J Neurol Neurosurg Psychiatry</i>	84	433-40	2013

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.	<i>Nat Genet</i>	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 <u>Matsuda, F.</u>	Two susceptibility loci to Takayasu arteritis reveal a synergistic role of the IL12B and HLA-B regions in a Japanese population.	<i>Am J Hum Genet</i>	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, H., Mimori, T. and <u>Matsuda, F.</u>	Association of Takayasu arteritis with HLA-B*67:01 and two amino acids in HLA-B protein.	<i>Rheumatol (Oxford)</i>	52	1769-1774	2013

<p>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., <u>Matsuda, F.</u> and Keavney, B. D.</p>	<p>Quantitative variation in plasma angiotensin-I converting enzyme activity shows allelic heterogeneity in the ABO blood group locus.</p>	<p><i>Ann Hum Genet</i></p>	<p>77</p>	<p>465-471</p>	<p>2013</p>
<p>Yamakawa, N., Fujimoto, M., Kawabata, D., Terao, C., Nishikori, M., Nakashima, R., Imura, Y., Yukawa, N., Yoshifuji, H., Ohmura, K., Fuji, T., Kitano, T., Kondo, T., Yurugi, K., Miura, Y., Maekawa, T., Saji, S., Takaori-Kondo, A., <u>Matsuda, F.</u>, Haga, H. and Mimori, T.</p>	<p>A clinical, pathological and genetic characterization of methotrexate-associated lymphoproliferative disorders.</p>	<p><i>J Rheumatol</i></p>	<p>41</p>	<p>293-299</p>	<p>2014</p>
<p>Tanaka, K., Terao, C., Ohmura, K., Takahashi, M., Nakashima, R., Imura, Y., Yoshifuji, H., Yukawa, N., Usui, T., Fuji, T., Mimori, T. and <u>Matsuda, F.</u></p>	<p>Significant association between <i>CYP3A5</i> polymorphism and blood concentration of tacrolimus in patients with connective tissue diseases.</p>	<p><i>J Hum Genet</i></p>	<p>59</p>	<p>107-109</p>	<p>2014</p>
<p>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.,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., <u>Matsuda, F.</u>, Yamamoto, K. and Plenge, R. M.</p>	<p>Genetics of rheumatoid arthritis contributes to biology and drug discovery.</p>	<p><i>Nature</i></p>	<p>506</p>	<p>376-381</p>	<p>2014</p>

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	<i>J Hum Genets</i>			In press

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 R K, Nakashima M, Saito 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.	<i>Clin Genet</i>			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	<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, Saito H.	PIGN mutations cause congenital anomalies, developmental delay, hypotonia, epilepsy, and progressive cerebellar atrophy.	<i>Neurogenetics</i>		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.	<i>Clin Genet.</i>		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.	<i>AJNR Am J Neuroradiol.</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
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 A.</i>	161	1221-37	2013
Hitomi Yatsuki, Ken Higashimoto, Kosuke Jozaki, Kayoko Koida, Junichiro Okada, Yoriko Watanabe, Nobuhiko Okamoto, Yoshinobu Tsuno, Yoko Yoshida, Kazutoshi Ueda, Kenji Shimizu, Hirofumi Ohashi, Tsunehiro Mukai, Hidenobu Saejima	Novel mutations of CDKN1C in Japanese patients with Beckwith-Wiedemann syndrome.	<i>Genes and Genetics</i>			
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>	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.	<i>Am J Med Genet A</i>	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.	<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 Gene</i>	58	391-4	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	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.	<i>Am J Med Genet A</i>	161	2291-3	2013

Okumura A, Hayashi M, Shimojima K, Ikeno M, Uchida T, Takanashi JI, Okamoto N, Hisata K, Shoji H, Saito , Furukawa T, Kishida T, Shimizu T, Yamamoto T.	Whole-exome sequencing of a unique brain malformation with periventricular heterotopia, cingulate polymicrogyria and midbrain tectal hyperplasia.	<i>Neuropathology. Neuropathology</i>	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, Ogasawa 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 A</i>	161	2234-43	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
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

Nakajima J, Okamoto N, Shiraishi J, Nishimura G, Nakashima M, Tsurusaki Y, Saitsu H, Kawahima 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

V 班 構 成 員 名 簿

平成25年度 遺伝学的検査の実施拠点の在り方に関する研究班

区分	氏名	所属等	職名
研究代表者	辻 省次	東京大学医学部附属病院神経内科	教授
研究分担者	松原 洋一	国立成育医療研究センター研究所	所長
	奥山 虎之	国立成育医療研究センター臨床検査部	診療部長
	後藤 雄一	国立精神・神経医療研究センター神経研究所	部長
	斎藤 加代子	東京女子医科大学附属遺伝子医療センター	教授
	宮地 勇人	東海大学医学部基盤診療学系臨床検査学	教授
	福嶋 義光	信州大学医学部遺伝医学	教授
	古川 洋一	東京大学医科学研究所臨床ゲノム腫瘍学分野	教授
	難波 栄二	鳥取大学生命機能研究支援センター遺伝子探索分野	教授・センター長
	秋山 真志	名古屋大学大学院医学系研究科皮膚科学	教授
	野口 佳裕	東京医科歯科大学医学部附属病院耳鼻咽喉科学	講師
	森田 啓行	東京大学大学院医学系研究科健康医科学創造講座	特任准教授
	小崎 健次郎	慶應義塾大学医学部小児科学教室	教授
	青木 正志	東北大学病院神経内科	教授
	小野寺 理	新潟大学脳研究所生命科学リソース研究センター	教授
	松田 文彦	京都大学大学院医学研究科附属ゲノム医学センター	教授
	梅澤 明弘	国立成育医療研究センター研究所再生医療センター	センター長
	松本 直通	横浜市立大学医学研究科環境分子医科学	教授
	岡本 伸彦	大阪府立病院機構大阪府立母子保健総合医療センター遺伝診療科	主任部長
山内 泰子	川崎医療福祉大学医療福祉学部医療福祉学科	准教授	
武藤 香織	東京大学医科学研究所公共政策研究分野	教授	
事務局	後藤 順	東京大学医学部附属病院神経内科 〒113-8655 東京都文京区本郷7-3-1 TEL 03-5800-6542 FAX 03-5800-6844 e-mail gotoj-tky@umin.ac.jp	准教授
経理事務担当者	大溝真由美	東京大学医学部附属病院管理課 TEL 03-5800-9753 FAX 03-5800-9145 e-mail kenkyukyoryokuAll@edm.h.u-tokyo.ac.jp	

