

別紙4

研究成果の刊行に関する一覧表レイアウト

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
黒田友紀子、大橋育子、黒澤健司	眼科疾患	福嶋義光監修、櫻井晃洋編集	遺伝カウンセリングマニュアル	南江堂	東京	2016	122-138

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
Inagaki H, Kato T, Tsutsumi M, Ouchi Y, Ohya T, Kurahashi H.	Palindrome-mediated translocations in humans: a new mechanistic model for gross chromosomal rearrangements.	Front Genet	7	125	2016
Tsutsumi M, Yokoi S, Miya F, Miyata M, Katoh 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(12)	1702-1706	2016
Boda H, Uchida H, Takaiso N, Ouchi Y, Fujita N, Kuno A, Hata T, Nagatani A, Funamoto Y, Miyata M, Yoshikawa T, Kurahashi H, Inagaki H.	A PDE3A mutation in familial hypertension and brachydactyly syndrome.	J Hum Genet	61(8)	701-3	2016
Taniguchi-Ikeda M, Takemoto Y, Lee T, Nishimura M, Awano H, Yamagishi M, Unzaki A, Nozu K, Nishio H, Matsuo M, Kurahashi H, Toda T, Morioka I, Iijima K.	Next-generation sequencing discloses a nonsense mutation in the dystrophin gene from long preserved dried umbilical cord and low-level somatic mosaicism in the proband mother.	J Hum Genet	61(4)	351-5	2016
Nagasaka M, Taniguchi-Ikeda M, Inagaki H, Ouchi Y, Kurokawa D, Yamana K, Harada R, Nozu K, Sakai Y, Mishra SK, Yamaguchi Y, Morikoka I, Toda T, Kurahashi H, Iijima K.	Novel missense mutation in DLL4 in a Japanese sporadic case of Adams-Oliver syndrome.	J Hum Genet	In press.	In press.	2017

Kato M, Kato T, Hosob a E, Ohashi M, Fujisaki M, Ozaki M, Yamaguc hi M, Sameshima H, K urahashi H.	PCS/MVA syndrome cau sed by an Alu insertion in the BUB1B gene.	Hum Genome Var	In press.	In press.	2017
Markoff A, Kurahashi H, Grandone E, Bogdan ova N.	Annexin A5 haplotype M2 is not a risk factor for recurrent miscarriages in Northern Europe, is there sufficient evidence?	Reprod Biomed Online	32(5)	469-73	2016
Markoff A, Kurahashi H, Grandone E, Bogdan ova N.	Authors' response to the letter of Nagirnaja et al., "Response to annexin A5 haplotype M2 is not a risk factor for recurrent miscarriages in Northern Europe, is there sufficient evidence?"	Reprod Biomed Online	33(1)	116-7	2016
Ohye T, Kawamura Y, Inagaki H, Yoshikawa A, Ihira M, Yoshikawa T, Kurahashi H.	A simple cytogenetic me thod to detect chromosom ally integrated human herpesvirus-6.	J Virol Method	228	74-8	2016
Kohmoto T, Okamoto N, Naruto T, Murata C, Ouchi Y, Fujita N, Ina gaki H, Satomura S, O kamoto N, Saito M, Ma suda K, Kurahashi H, I moto I.	A case with concurrent duplication, triplication, a nd uniparental isodisomy at 1q42.12-qter supporti ng microhomology- medi ated break-induced replic ation model for replicati ve rearrangements.	Mol Cytogenet	10	15	2017
Suzuki E, Shima H, To ki M, Hanew K, Matsul bara K, Kurahashi H, No rumi S, Ogata T, Kam imaki T, Fukami M.	Complex X-Chromosomal Rearrangements in Two Women with Ovarian Dysfunction: Implications of Chromothripsis/Chro moanasythesis-Dependen t and -Independent Origi ns of Complex Genomic Alterations.	Cytogenet Gen ome Res	150(2)	86-92	2016
Miyazaki J, Nishizawa H, Kambayashi A, Ito M, Noda Y, Terasawa S, Kato T, Miyamura H, Shiogama K, Sekiya T, Kurahashi H, Fujii T.	Increased levels of solub le corin in pre-eclampsia and fetal growth restrict ion.	Placenta	48	20-25	2016
Yasui T, Suzuki T, Har a F, Watanabe S, Uga N, Naoe A, Yoshikawa T, Ito T, Nakajima Y, Miura H, Sugioka A, Kato Y, Tokoro T, Tan ahashi Y, Kasahara M, Fukuda A, Kurahashi H.	Successful living donor liver transplantation for classical maple syrup urine disease.	Pediatr Transpl	20(5)	707-710	2016

Azuma Y, Töpf A, Evangelista T, Lorenzoni Pn J, Roos A, Viana P, Inagaki H, Kurahashi H, Lochmüller H.	Intragenic DOK7 deletion detected by whole-genome sequencing in congenital myasthenic syndromes.	Neurol Genet	3(3)	e152	2017
Inoue Y, Sakamoto Y, Sugimoto M, Inagaki H, Boda H, Miyata M, Kato H, Kurahashi H, Okamoto T.	A family with craniofrontonasal syndrome: the first report of familial case of craniofrontonasal syndrome with bilateral cleft lip and palate.	Cleft Palate Cr Anofac J	In press.	In press.	2017
Takaiso N, Nishizawa H, Nishiyama S, Sawada T, Hosoba E, Ohye T, Sato T, Inagaki H, Kurahashi H.	Mutation analysis of the JUNO gene in female infertility of unknown etiology.	Fujita Med J	2(3)	59-61	2016
Kawamura Y, Ohye T, Miura H, Ihira M, Kato Y, Kurahashi H, Yoshikawa T.	Analysis of the origin of inherited chromosomally integrated human herpesvirus 6 in the Japanese population.	J Gen Virol	In press.	In press.	2017
Nakajima M, Kou I, Ohhashi H; Genetic Study Group of the Investigation Committee on the Ossification of Spinal Ligaments., Ikegawa S.	Identification and functional characterization of SPO2 as a susceptibility gene for ossification of the posterior longitudinal ligament of the spine.	Am J Hum Genet	99	202-7	2016
Watanabe S, Shimizu K, Ohashi H, Kosaki R, Okamoto N, Shimojima K, Yamamoto T, Chen Y, Mizuno S, Doiwa Y, Shiomi N, Toda Y, Tashiro K, Shichijo K, Minatozaki K, Aso S, Minagawa K, Hirakawa Y, Shimokawa O, Matsumoto T, Fukuda M, Moriuchi H, Yoshiura K, Kondoh T.	Detailed analysis of 26 cases of 1q partial duplication/triplication syndrome.	Am J Med Genetics Part A	170	908-17	2016
Yaoita M, Niihori T, Mizuno S, Okamoto N, Hayashi S, Watanabe A, Yokozawa M, Suzumura H, Nakahara A, Nakanishi Y, Hokosaki T, Ohmori A, Sawada H, Migitaña O, Mima A, Lapunzina P, Santos-Simarro F, García-Miñáur S, Ogata T, Kawame H, Kuroswa K, Ohashi H, Inoue S, Matsubara Y, Kure S, Aoki Y.	Spectrum of mutations and genotype-phenotype analysis in Noonan syndrome patients with RIT1 mutations.	Hum Genet	135	209-2	2016

Shiohama T, Fujii K, Hino M, Shimizu K, Ohashi H, Kambe M, Nakatani Y, Mitsunaga T, Yoshida H, Ochiai H, Shimojo N.	Coexistence of neuroblastoma and ganglioneuroma in a girl with a hemizygous deletion of chromosome 11q14.1-23.3.	Am J Med Genet A	170	492-7	2016
Ono H, <u>Kurosawa K</u> , Wakamatsu N, Masuda S.	Hearing impairment in a female infant with interstitial deletion of 2q24.1q24.3.	Congenit Anom (Kyoto)		Epub ahead of print	2016
Hossain MA, Yanagisawa H, Miyajima T, Wu C, Takamura A, Akiyama K, Itagaki R, Eto K, Iwamoto T, Yokoi T, <u>Kurosawa K</u> , Numabe H, Eto Y.	The severe clinical phenotype for a heterozygous Fabry female patient correlates to the methylation of non-mutated allele associated with chromosome 10q26 deletion syndrome.	Mol Genet Metab.	120(3):	173-179. doi: 10.1016/j.ymgme.2017.01.002.	2017
Akiyama T, Osaka H, Shimbo H, Kuhara T, Shibata T, Kobayashi K, <u>Kurosawa K</u> , Yoshinaga H.	SSADH deficiency possibly associated with enzyme activity-reducing SNPs.	Brain Dev.	38(9)	871-4	2016
Shimojima K, Okamoto N, <u>Yamamoto T</u> .	A 10q21.3q22.2 microdeletion identified in a patient with severe developmental delay and multiple congenital anomalies including congenital heart defects.	Congenit Anom	In press	In press	2017
Okamoto N, Shimojima K, <u>Yamamoto T</u> .	Neurological Manifestations of 2q31 Microdeletion Syndrome.	Congenit Anom	In press	In press	2017
Shimojima K, Okamoto N, <u>Yamamoto T</u> .	Possible genes responsible for developmental delay observed in patients with rare 2q23q24 microdeletion syndrome: literature review and description of an additional patient.	Congenit Anom	In press	In press	2017
Alber M, Kalscheuer V, Marco E, Sherr EH, Lesca G, Till M, Gradel G, Wiesener A, Korneke CG, Mecier S, Becker F, <u>Yamamoto T</u> , Scherer SW, Marshall C, Walker S, Dutta U, Dalal A, Suckow V, Jamali P, Kahrizi K, Najmabadi H, Minassian BA.	The ARHGEF9 Disease: Phenotype Clarification and Genotype-Phenotype Correlation.	Neurol Genet	In press	In press	2017
Shirai K, Higashi Y, Shimojima K, <u>Yamamoto T</u> .	An Xq22.1q22.2 nullisomy in a male patient with severe neurological impairment.	Am J Med Genet A	173A	1124-1127	2017

Murakoshi M, Takasawa K, Nishioka M, Asaka wa M, Kashimada K, YOSHIMOTO T, Takekoshi K, Ogawa Y, Shimohira M.	Abdominal paraganglioma in a young woman with 1p36 deletion syndrome.	Am J Med Genet A	173A	495-500	2017
Matsuo M, Yamauchi A, Ito Y, Sakauchi M, Yamamoto T, Okamoto N, Tsurusaki Y, Miyake N, Matsumoto N, Saito K.	Mandibulofacial dysostosis with microcephaly: A case presenting with seizures.	Brain Dev	39	177-181	2017
Shimojima K, Ondo Y, Matsufuji M, Sano N, Tsuru H, Oyoshi T, Higashino N, Tokimura H, Arita K, Yamamoto T..	Concurrent occurrence of an inherited 16p13.11 microduplication and a de novo 19p13.3 microdeletion involving MAP2K2 in a patient with developmental delay, distinctive facial features, and lambdoid synostosis.	Eur J Med Genet	59	559-563	2016
Yamamoto T, Shimojima K, Matsufuji M, Mashima R, Sakai E, Okuyama T.	Aspartylglucosaminuria caused by a novel homozygous mutation in the <i>AGA</i> gene was identified by an exome-first approach in a patient from Japan.	Brain Dev	39	422–425	2017
Yamamoto T, Shimojima K, Yamazaki S, Ikeno K, Tohyama J.	A 16q12.2q21 deletion identified in a patient with developmental delay, epilepsy, short stature, and distinctive features.	Congenit Anom (Kyoto)	56	253-255	2016
Hamatani M, Jingami N, Tsurusaki Y, Shimada S, Shimojima K, Asada Utsugi M, Yoshinaga K, Uemura N, Yamashita H, Uemura K, Takashashi R, Matsumoto N, Yamamoto T.	The first Japanese case of leukodystrophy with ovarian failure arising from novel compound heterozygous <i>AARS2</i> mutation.	J Hum Genet	61	899-902	2016
Shimojima K, Narai S, Togawa M, Doumoto T, Sangu N, Vanakkere OM, De Paeppe A, Edwards M, Whitehall J, Brescianini S, Petit F, Andrieux J, Yamamoto T.	7p22.1 microdeletions involving <i>ACTB</i> associated with developmental delay, short stature, and microcephaly.	Eur J Med Genet	59	502-6	2016
Shimojima K, Maruyama K, Kikuchi M, Imai A, Inoue K, Yamamoto T.	Novel SLC16A2 mutations in patients with Allane-Herndon-Dudley syndrome.	Intractable Rare Dis Res	5	214-217	2016

Shimojima K, Ondo Y, Nishi E, Mizuno S, Itsuo M, Ioi A, Shimizu M, Sato M, Inoue M, Okamoto N, Yamamoto T.	Loss-of-function mutation in <i>GPC3</i> in patients with Simpson-Golabi-Behmel syndrome.	Human Genome Var	3	16033	2016
Morisada N, Ioroi T, Taniguchi-Ikeda M, Ye MJ, Okamoto N, Yamamoto T, Iijima K.	A 12p13 <i>GRIN2B</i> deletion is associated with developmental delay and macrocephaly.	Human Genome Var	3	16029	2016
Yamamoto T, Shimojima K, Ondo Y, Imai K, Chong P-F, Kira R, Amemiya M, Saito A, Okamoto N.	Challenges in detecting genomic copy number aberrations using next-generation sequencing data and the eXome Hidden Markov Model: a clinical exome-first diagnostic approach.	Human Genome Var	3	16025	2016
Iwasaki N, Tsurumi M, Asai K, Shimuzu W, Watanabe A, Ogata M, Takizawa M, Ide R, Yamamoto T, Saito K.	Pancreatic developmental defect evaluated byeliac artery angiography in a patient with MODY5.	Human Genome Var	3	16022	2016
Banno K, Omori S, Hirata K, Nawa N, Nakagae wa N, Nishimura K, Ohtaka M, Nakanishi M, Sakuma T, Yamamoto T, Toki T, Ito E, Yamamoto T, Kokubu C, Takeda J, Taniguchi H, Arahorri H, Wada K, Kitabatake Y, Ozono K.	Systematic cellular disease models reveal synergistic interaction of trisomy 21 and <i>GATA1</i> mutation in hematopoietic abnormalities.	Cell Reports	15	1228-41	2016
Itakura A, Saito Y, Nishimura Y, Okazaki T, Ohno K, Sejima H, Yamamoto T, Maegaki Y.	Successful treatment of migrating partial seizures in Wolf-Hirschhorn syndrome with bromide.	Brain Dev	38	658-62	2016
Oka M, Shimojima K, Yamamoto T, Hanaoka Y, Sato S, Yasuhara T, Yoshinaga H, Kobayashi K.	A novel <i>HYLS1</i> homozygous mutation in living siblings with Joubert syndrome.	Clin Genet	89	739-43	2016
Sangu N, Okamoto N, Shimojima K, Ondo Y, Nishikawa M, Yamamoto T.	A de novo microdeletion in a patient with inner ear abnormalities suggests the existence of the responsible gene in 10q26.	Human Genome Var	3	16008	2016
Shimojima K, Okamoto N, Yamamoto T..	A novel <i>TUBB3</i> mutation in a sporadic patient with asymmetric cortical dysplasia.	Am J Med Genetics Part A	170A	1076-9	2016

Watanabe S, Shimizu K, Ohashi H, Kosaki R, Okamoto N, Shimojima K, Yamamoto T, Cmehin Y, Mizuno S, Dowa Y, Shiomi N, Toda Y, Tashiro K, Shichijo K, Minatozaki K, Aso	Detailed analysis of 26 cases of 1q partial duplication/triplication 1 syndrome. Yamamoto T, Cmehin Y, Mizuno S, Dowa Y, Shiomi N, Toda Y, Tashiro K, Shichijo K, Minatozaki K, Aso	Am J Med Genet A	170A	908-17	2016
Yamamoto T, Igarashi N, Shimojima K, Sangue N, Sakamoto Y, Shimoji K, Niijima S..	Use of targeted next-generation sequencing for molecular diagnosis of craniosynostosis: identification of a novel de novo mutation of <i>EFNB1</i> .	Congenit Anom	56	91-3	2016
Yamamoto T, Shimojima K, Yano T, Ueda Y, Takayama R, Ikeda H, Imai K.	Loss-of-function mutations of <i>STXBPI</i> in patients with epileptic encephalopathy.	Brain Dev	38	4-7	2016
Moteki H, Azaiez H, Sloan-Heggen CM, Booth K, Nishio SY, Wakui K, Yamaguchi T, Kolbe DL, Iwasa YI, Shearer AE, Fukushima Y, Smith RJ, Usami SI.	Detection and Confirmation of Deafness-Causing Copy Number Variations in the STRC Gene by Massively Parallel Sequencing and Comparative Genomic Hybridization.	Ann Otol Rhinol Laryngol.	125(11)	918-923	2016