

研究成果の刊行に関する一覧表

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
Yamamoto T, Shimojima K.	Pelizaeus-Merzbacher disease as a chromosomal disorder.	Congenit Anom (Kyoto)	53	3-8	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	411-419	2013
Okumura A, Shimojima K, Kubota T, Abe S, Yamashita S, Imai K, Okanishi T, Enoki H, Fukasawa T, Tanabe T, Dibbens LM, Shimizu T, Yamamoto T.	PRRT2 mutation in Japanese children with benign infantile epilepsy.	Brain Dev (Early On-line View)			
Shimada S, Maegaki Y, Osawa M, Yamamoto T.	Mild developmental delay and obesity in two patients with mosaic 1p36 deletion syndrome.	Am J Med Genet	in press		
Yamamoto T, Togawa M, Shimada S, Sangu N, Shimojima K, Okamoto N.	Narrowing of the responsible region for severe developmental delay and autistic behaviors in WAGR syndrome down to 1.6 Mb including PAX6, WT1, and PRRG4.	Am J Med Genet	in press		

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, Shioda M, Okumura A, Kidokoro H, Sakauchi M, Shimada S, Shimizu T, Osawa M, Hirose S, Yamamoto T.	A recurrent KCNT1 mutation in two sporadic cases with malignant migrating partial seizures in infancy.	Gene	531	467-471	2013
Sangu N, Shimojima K, Shimada S, Ando T, Yamamoto T.	Growth patterns of patients with 1p36 deletion syndrome.	Congenit Anom (Kyoto)	in press		
Eto K, Sakai N, Shimada S, Shioda M, Ishigaki K, Hamada Y, Shinpo M, Azuma J, Tominaga K, Shimojima K, Ozono K, Osawa M, Yamamoto T.	Microdeletions of 3p21.31 characterized by developmental delay, distinctive features, elevated serum creatine kinase levels, and white matter involvement.	Am J Med Genet	161A	3049-3056	2013
Okamoto N, Ohmachi K, Shimada S, Shimojima K, Yamamoto T.	109 kb deletion of chromosome 4p16.3 limited to WHSCR2 in a patient with mild phenotype of Wolf-Hirschhorn syndrome.	Am J Med Genet	161A	1465-1469	2013
Yamamoto T, Shimada S, Shimojima K.	Fiber-FISH analyses as a diagnostic application for orientation of the microduplications.	World J Med Genet	3	5-8	2013
Okumura A, Hayashi M, Tsurui H, Yamakawa Y, Abe S, Kudo T, Suzuki R, Shimizu T, Yamamoto T.	Lissencephaly with marked ventricular dilation, agenesis of corpus callosum, and cerebellar hypoplasia caused by TUBA1A mutation.	Brain Dev	35	274-279	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
Shichiji M, Ito Y, Shimojima K, Nakamu H, Oguni H, Osawa M, Yamamoto T.	A cryptic microdeletion including MBD5 occurring within the breakpoint of a reciprocal translocation between chromosomes 2 and 5 in a patient with developmental delay and obesity.	Am J Med genet A	161A	850-855	2013
Kobayashi S, Inui T, Wakusawa K, Tanaka S, Nakayama T, Uematsu M, Takayanagi M, Yamamoto T, Haginoya K.	A case of atypical benign partial epilepsy with action myoclonus.	Seizure	22	242-245	2013
Yamamoto T, Matsuo M, Shimada S, Sangu N, Shimojima K, Aso S, Saito K.	De novo triplication of 11q12.3 in a patient with developmental delay and distinctive facial features.	Mol Cytogenet	6	15	2013
Shimojima K, Shimada S, Sugawara M, Yoshikawa N, Nijima S, Urao M, Yamamoto T.	Challenges in genetic counseling because of intra-familial phenotypic variation of oral-facial-digital syndrome type 1.	Congenit Anom (Kyoto)	53	155-159	2013
Usui D, Shimada S, Shimojima K, Sugawara M, Kawasaki H, Shigematu H, Takahashi Y, Inoue Y, Imai K, Yamamoto T.	Interstitial duplication of 2q32.1-q33.3 in a patient with epilepsy, developmental delay, and autistic behavior.	Am J Med Genet A	161A	1078-1084	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
Sangu N, Shimosato T, Inoda H, Shimada S, Shimojima K, Ando T, Yamamoto T.	A novel nucleotide mutation leading to a recurrent amino acid alteration in SH3BP2 in a patient with cherubism.	Congenit Anom (Kyoto)	53	166-169	2013
Okumura A, Hayashi M, Shimojima K, Ikeno M, Uchida T, Takanashi J, Okamoto N, Hisata K, Shoji H, Saito A, Furukawa T, Kishida T, Shimizu T, Yamamoto T.	Whole-exome sequence for a unique brain malformation with periventricular heterotopia, cingulate polymicrogyria, and midbrain tectal hyperplasia.	Neuropathology	33	553-560	2013
Okamoto N, Ohmachi K, Shimada S, Shimojima K, Yamamoto T.	101 kb deletion of chromosome 4p16.3 limited to WHSCR2 in a patient with mild phenotype of Wolf-Hirschhorn syndrome.	Am J Med Genet A	161A	1465-1469	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	161A	2078-2083	2013
Shimojima K, Tanaka R, Shimada S, Sangu N, Nakayama J, Iwasaki N, Yamamoto T.	A novel homozygous mutation of GJC2 derived from maternal uniparental disomy in a female patient with Pelizaeus-Merzbacher-like disease.	J Neurol Sci	330	123-126	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
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	108-110	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
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	173-180	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
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	161A	2291-2293	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, Saito H, Yoshiura K, Matsumoto N, Niikawa N.	MLL2 and KDM6A mutations in patients with Kabuki syndrome.	Am J Med Genet A	161A	2234-2243	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, Saito H.	De novo mutations in SLC35A2 encoding a UDP-galactose transporter cause early-onset epileptic encephalopathy.	Hum Mutat	34	1708-1714	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
Koshimizu E, Miyatake S, Okamoto N, Nakashima M, Tsurusaki Y, Miyake N, Saito 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, Saito H, Kawashima H, Matsumoto N, Miyake N.	Novel FIG4 mutations in Yunis-Varon syndrome.	J Hum Genet	in press		
Ichikawa K, Kadota M, Wada Y, Okamoto N.	Congenital disorder of glycosylation type Ic: report of a Japanese case.	Brain Dev	35	586-589	2013
Miyatake S, Murakami A, Okamoto N, Sakamoto M, Miyake N, Saito 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	1073-1077	2013
Iida A, Okamoto N, Miyake N, Nishimura G, Minami S, Sugimoto T, Nakashima M, Tsurusaki Y, Saito H, Shiina M, Ogata K, Watanabe S, Ohashi H, Matsumoto N, Ikegawa S.	Exome sequencing identifies a novel INPPL1 mutation in opsismodysplasia.	J Hum Genet	58	391-394	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
Sakai Y, Ohkubo K, Matsushita Y, Akamine S, Ishizaki Y, Torisu H, Ihara K, Sanefuji M, Kim MS, Lee KU, Shaw CA, Lim J, Nakabeppu Y, Hara T.	Neuroendocrine phenotypes in a boy with 5q14 deletion syndrome implicate the regulatory roles of myocyte-specific enhancer factor 2C in the postnatal hypothalamus.	Eur J Med Genet	56	475-483	2013
Torisu H, Yoshikawa Y, Yamaguchi-Takada Y, Yano T, Sanefuji M, Ishizaki Y, Sawaishi Y, Hara T.	Alexander disease with mild dorsal brainstem atrophy and infantile spasms.	Brain Dev	35	441-444	2013
Uike K, Matsushita Y, Sakai Y, Togao O, Nagao M, Ishizaki Y, Nagata H, Yamamura K, Torisu H, Hara T.	Systemic vascular phenotypes of Loey's-Dietz syndrome in a child carrying a de novo R381P mutation in TGFBR2: a case report.	BMC Research Notes	6	456	2013
Torisu H, Watanabe K, Shimojima K, Sugawara K, Sanefuji M, Ishizaki Y, Sakai Y, Yamashita H, Yamamoto T, Hara T.	Girl with a PRRT2 mutation and infantile focal epilepsy with bilateral spikes.	Brain Dev	in press		
Abe Y, Kobayashi S, Wakusawa K, Tanaka S, Inui T, Yamamoto T, Kunishima S, Haginoya K.	Bilateral periventricular nodular heterotopia with megalencephaly: a case report.	J Child Neurol	in press		

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
Shimojima K, Shimada S, Tamasaki A, Akaboshi S, Komoike Y, Saito A, Furukawa T, Yamamoto T.	Novel compound heterozygous mutations of POLR3A revealed by whole-exome sequencing in a patient with hypomyelination.	Brain Dev	in press		
Irahara K, Saito Y, Sugai K, Nakagawa E, Saito T, Komaki H, Nakata Y, Sato N, Baba K, Yamamoto T, Chan W, Andrews C, Engle EC, Sasaki M.	Pontine malformation, undecussated pyramidal tracts and regional polymicrogyria: a new syndrome.	Pediatr Neurol	in press		
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.	Am J Med Genet A	in press		
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.	Neurogenetics	in press		
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, Saito H, Miyake N, Matsumoto N.	Coffin-Siris syndrome is a SWI/SNF complex disorder.	Clin Genet	in press		

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
Wada T, Ban H, Matsufuji M, Okamoto N, Enomoto K, Kurosawa K, Aida N.	Neuroradiologic Features in X-linked -Thalassemia/ Mental Retardation Syndrome.	AJNR Am J Neuroradiol	in press		
Shimada S, Okamoto N, Hirasawa K, Yoshii K, Tani Y, Sugawara M, Shimajima 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.	161	1779-1785	2013