

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
岡本伸彦	神経皮膚症候群診断マニュアル	大槻泰介他	稀少難治てんかん診療マニュアル	診断と治療社	東京	2013	117-121
岡本伸彦	遺伝性疾患の発達と予後	氏家 達夫	発達心理学事典	丸善	東京	2013	

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
Tachibana Y, Aida N, Enomoto K, Iai M, Kurosawa K.	A case of Sjögren-Larsson syndrome with minimal MR imaging findings facilitated by proton spectroscopy.	Pediatr Radiol	42	380-2.	2012
Kurosawa K, Enomoto K, Tominaga M, Furuya N, Sameshima K, Iai M, Take H, Shinkai M, Ishikawa H, Yamanaka M, Matsui M, Masuno M.	Spastic quadriplegia in Down syndrome with congenital duodenal stenosis/atresia.	Cong Anom	52	78-81.	2012
Enomoto K, Kishitani Y, Tominaga M, Ishikawa A, Furuya N, Aida N, Masun M, Yamada K, Kurosawa K.	Expression Analysis of a 17p Terminal Deletion, including YWHAE, but not PAFAH1B1, associated with normal brain structure on MRI in a young girl.	Am J Med Genet Part A	158A	2347-2352.	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.	Am J Med Genet A.	61(9)	2300-4	2013

Hayashi S, Okamoto N, Chinen Y, Takanashi JI, Makita Y, Hata A, Imoto I, Inazawa J.	Novel intragenic duplications and mutations of CASK in patients with mental retardation and microcephaly with pontine and cerebellar hypoplasia (MICPCH).	Hum Genet.	131	99-110	2012
Yukiko Kawazu, Noboru Inamura, Futoshi Kayatani, Nobuhiko Okamoto, Hiroko Morisaki	Prenatal complex congenital heart disease with Loeys–Dietz syndrome	Cardiology in the Young	22	116-119	2012
Miyatake S, Miyake N, Touho H, Nishimura-Tadaki A, Kondo Y, Okada I, Tsurusaki Y, Doi H, Sakai H, Saitsu H, Shimojima K, Yamamoto T, Higurashi M, Kawahara N, Kawauchi H, Nagasaka K, Okamoto N, Mori T, Koyano S, Kuroiwa Y, Taguri M, Morita S, Matsubara Y, Kure S, Matsumoto N.	Homozygous c.14576G>A variant of RNF213 predicts early-onset and severe form of moyamoya disease.	Neurology	78	803-810	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.	Am J Med Genet A.	158A	514-518	2012
Tsurusaki Y, Okamoto N, Ohashi H, Mizuno S,ほか	Mutations affecting components of the SWI/SNF complex cause Coffin-Siris syndrome.	Nat Genet. 2	44	376-378	2012
Honda S, Hayashi S, Nakane T, Imoto I, Kurosawa K, Mizuno S, Okamoto N, Kato M, Yoshihashi H, Kubota T, Nakagawa E, Goto Y, Inazawa J.	The incidence of hypoplasia of the corpus callosum in patients with dup (X)(q28) involving MECP2 is associated with the location of distal breakpoints.	Am J Med Genet A..	158A	1292-1303	2012
Abe Y, Aoki Y, Kuriyama S, Kawame H, Okamoto N, Kurosawa K, Ohashi H, Mizuno S, Ogata T, Kure S, Nihori T, Matsubara Y.	Costello and CFC syndrome study group in Japan. Prevalence and clinical features of Costello syndrome and cardio-facio-cutaneous syndrome in Japan: findings from a nationwide epidemiological survey.	Am J Med Genet A.	158A	1083-1094	2012

Shimojima K, Okamoto N, Suzuki Y, Saito M, Mori M, Yamagata T, Momoi MY, Hattori H, Okano Y, Hisata K, Okumura A, Yamamoto T.	Subtelomeric deletions of 1q43q44 and severe brain impairment associated with delayed myelination.	J Hum Genet.	57	593-600	2012
Shimojima K, Mano T, Kashiwagi M, Tanabe T, Sugawara M, Okamoto N, Arai H, Yamamoto T.	Pelizaeus-Merzbacher disease caused by a duplication-inverted triplication-duplication in chromosomal segments including the PLP1 region.	Eur J Med Genet.	55	400-403	2012
Wada Y, Kadoya M, Okamoto N.	Mass spectrometry of apolipoprotein C-III, a simple analytical method for mucin-type O-glycosylation and its application to an autosomal recessive cutis laxa type-2 (ARCL2) patient.	Glycobiology.	22	1140-1144	2012
Naiki M, Mizuno S, Yamada K, Yamada Y, Kimura R, Oshiro M, Okamoto N, Makita Y, Seishima M, Wakamatsu N.	MBTPS2 mutation causes BRESEK/BRESHECK syndrome	Am J Med Genet Part A	158	97	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	Congenit Anom (Kyoto)	52	82	2012
Takanashi J, Okamoto N, Yamamoto Y, Hayashi S, Arai H, Takahashi Y, Maruyama K, Mizuno S, Shimakawa S, Ono H, Oyanagi R, Kubo S, Barkovich AJ, Inazawa J	Clinical and radiological features of Japanese patients with a severe phenotype due to CASK mutations	Am J Med Genet A	158	3112	2012
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.	Congenit Anom (Kyoto)	53(1)	54-7	2013
Takenouchi T, Hashida N, Torii C, Kosaki R, Takahashi T, <u>Kosaki K.</u>	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

<p>Takenouchi T, Nishina S, Kosaki R, Torii C, Furukawa R, Takahashi T, <u>Kosaki K.</u></p>	<p>Concurrent deletion of BMP4 and OTX2 genes, two master genes in ophthalmogenesis.</p>	<p>Eur J Med Genet.</p>	<p>56(1)</p>	<p>50-53</p>	<p>2013</p>
<p>Takenouchi T, Enomoto K, Nishida T, Torii C, Okazaki T, Takahashi T, <u>Kosaki K.</u></p>	<p>12q14 microdeletion syndrome and short stature with or without relative macrocephaly.</p>	<p>Am J Med Genet A.</p>	<p>158(10)</p>	<p>2542-2544</p>	<p>2012</p>
<p>Takenouchi T, Okuno H, Kosaki R, Ariyasu D, Torii C, Momoshima S, Harada N, Yoshinashi H, Takahashi T, Awazu M, <u>Kosaki K.</u></p>	<p>Microduplication of Xq24 and Hartsfield syndrome with holoprosencephaly, ectrodactyly, and clefting.</p>	<p>Am J Med Genet A.</p>	<p>158(10)</p>	<p>2537-2541</p>	<p>2012</p>
<p>Takenouchi T, Yagihashi T, Tsuchiya H, Torii C, Hayashi K, Kosaki R, Saitoh S, Takahashi T, <u>Kosaki K.</u></p>	<p>Tissue-limited ring chromosome 18 mosaicism as a cause of Pitt-Hopkins syndrome.</p>	<p>Am J Med Genet A.</p>	<p>158(10)</p>	<p>2621-2623</p>	<p>2012</p>
<p>Kosaki R, Takenouchi T, Takeda N, Kagami M, Nakabayashi K, Hata K, Kosaki K.</p>	<p>Somatic CTNNB1 mutation in hepatoblastoma from a patient with Simpson-Golabi-Behmel syndrome and germline GPC3 mutation.</p>	<p>Am J Med Genet A.</p>		<p>Epub ahead of print</p>	<p>2014</p>
<p>Takeuchi T, Hayashida N, Torii C, Kosaki R, Takahashi T, Saya H, Kosaki K.</p>	<p>1p34.3 deletion involving GRIK3: Further clinical implication of GRIK family glutamate receptors in the pathogenesis of developmental delay.</p>	<p>Am J Med Genet A.</p>	<p>164(2)</p>	<p>456-60</p>	<p>2014</p>
<p>Takeuchi 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>Am J Med Genet A.</p>	<p>164(2)</p>	<p>392-6</p>	<p>2014</p>
<p>Sasaki A, Sumie M, Wada S, Kosaki R, Kuroswa K, Fukami M, Sago H, Ogata T, Kagami M.</p>	<p>Prenatal genetic testing for a microdeletion at chromosome 14q32.2 imprinted region leading to UPD(14)pat-like phenotype.</p>	<p>Am J Med Genet A.</p>	<p>164A(1)</p>	<p>264-6</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, Kosak 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 Hum Genet.	7:93(5)	945-56	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)	3057-62.	2013
Nakajima M, Mizumoto S, Miyake N, Kogawa R, Iida A, Ito H, Kitoh H, Hirayama A, Mitsubuchi H, Miyazaki O, Kosaki R, ほか	Mutations in B3GALT6, which Encodes a Glycosaminoglycan Linker Region Enzyme, Cause a Spectrum of Skeletal and Connective Tissue Disorders.	J Hum Genet.	6:92(6)	927-34	2013
小崎 里華	遺伝の基礎知識・考え方	月刊母子保健	12月号		2013
小崎 里華	遺伝性内分泌疾患に関する遺伝カウンセリング	内分泌・糖尿病・代謝内科	Vo137 No.4		2013
小崎 里華	先天異常の分類	小児科臨床	66巻増刊号		2013
Miyake N, Koshimizu E, Okamoto N, Mizuno S, Ogata T, Nagai T, Kosho T, Ohashi H, ほか	MLL2 and KDM6A mutations in patients with Kabuki syndrome.	Am J Med Genet	A 161	2234-43	2013
Aoki Y, Niihori T, Banjo T, Okamoto N, Mizuno S, Kurosawa K, Ohashi H, ほか	Gain-of-function mutations in RIT1 cause Noonan syndrome, a RAS/MAPK pathway syndrome.	Am J Hum Genet	93	173-80	2013
Takahashi M, Ohashi H.	Craniofacial and dental malformations in Costello syndrome: A detailed evaluation using multi-detector row computed tomography.	Congenit Anom	53	67-72	2013

Kosho T, Okamoto N, Ohashi H,ほか	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	161	1221-37	2013
Fuke T, Mizuno S, Nagai T, Hasegawa T, Horikawa R, Miyoshi Y, Muroya K, Kondoh T, Numakura C, Sato S, Nakabayashi K, Tayama C, Hata K, Sano S, Matsubara K, Kagami M, Yamazawa K, Ogata T	Molecular and Clinical Studies in 138 Japanese Patients with Silver-Russell Syndrome.	PLoS ONE	8(3)	e60105	2013
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	Front Hum Neurosci	11	856	2013
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.	2013	Dec 19.[Epub ahead of print]	[Epub ahead of print]
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.	2013	Dec 5.	[Epub ahead of print]
Suzumori N, Kaname T, Muramatsu Y, Yanagi K, Kumagai K, Mizuno S, Naritomi K, Saitoh S, Sugiura Ogasawara M.	Prenatal diagnosis of X-linked recessive Lenz microphthalmia syndrome.	J Obstet Gynaecol Res.	39(11)	1545-7	2013
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
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.	164	213-9	2014

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.	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, Saitsu H, Miyake N, Matsumoto N.	Coffin-Siris syndrome is a SWI/SNF complex disorder.	Clin Genet.	In press		
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, Hibi-Ko Y, Kawame H,ほか	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.	161	1221-37.	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.	Am J Med Genet A.	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.	Am J Med Genet A.	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.	Am J Med Genet A.	161	2078-83.	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
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
Kodera H, Nakamura K, Osaka H, Maegaki Y, Haginoya K, Mizumoto S, Kato M, Okamoto N, ほか	De Novo Mutations in SLC35A2 Encoding a UDP-Galactose Transporter Cause Early-Onset Epileptic Encephalopathy.	Hum Mutat.	34	1708-14	2013
Nakajima J, Okamoto N, Shiraishi J, ほか	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
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
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	161A:	1779-85	2013
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	467-71	2013
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

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	164A	634-638	2013
Sangu N, Shimojima K, Shimada S, Ando T, Yamamoto T	Growth patterns of patients with 1p36 deletion syndrome.	Congenit Anom (Kyoto)			(in press)
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
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, 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	36:	315-321	2014
Yamamoto T, Wilsdon A, Joss S, Isidor B, Erlandsson A, Suri M, Sangu N, Shimada S, Shimojima K, LeCaignec C, Samuelsson L, Stefanova M.	An emerging phenotype of Xq22 microdeletions in females with severe intellectual disability, hypotonia and behavioral abnormalities.	J Hum Genet			(early on-line view)