III.　研究成果の刊行に関する一覧表

別紙４

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
Miyake N, Kosho T, Matsumoto N	Ehlers–Danlos syndrome associated with glycosaminoglycan abnormalities	Jaroslava Halper	Progress in heritable soft connective tissue disorders	Springer	Springer Netherlands	2013	145-159
Vergano SS, Santen G, Wieczorek D, Wollnik B, Matsumoto N, Deardorff MA	Coffin-Siris Syndrome	Roberta A Pagon	GeneReviews™ [Internet]	University of Washington	Seattle	2013	http://www.ncbi.nlm.nih.gov/books/NBK131811/

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
Fujita A, Ochi N, Fujimaki H, Muramatsu H, other, **Matsumoto N**, **Miyake N**	A novel *WTX* mutation in a female patient with osteopathia striata with cranial sclerosis and hepatoblastoma.	Am J Med Genet A.	(in press)	(in press)	2014
Fukai R, Hiraki Y, Nishimura G, others, **Matsumoto N**, **Miyake N**	A de novo 1.4-Mb deletion at 21q22.11 in a boy with developmental delay.	Am J Med Genet A.	(in press)	(in press)	2014
Nakajima J, Eminoglu TF, Vatansever G, others, **Matsumoto N**, **Miyake N**.	A novel homozygous *YARS2* mutation causes severe myopathy, lactic acidosis, and sideroblastic anemia 2.	J Hum Genet	(in press)	(in press)	2014
Nakamura K, Osaka H, others, **Miyake N**, Kinoshita T, **Matsumoto N**, Saitsu H	*PIGO* mutations in intractable epilepsy and severe developmental delay with mild elevation of alkaline phosphatase levels.	Epilepsia	(in press)	(in press)	2014
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Ohba C, **Okamoto N**, others, **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)	(in press)	2013
Kodera H, others, **Okamoto N**, others, **Miyake N**, others, **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
Imagawa E, others, **Matsumoto N**, **Miyake N**.	A hemizygous *GYG2* mutation and Leigh syndrome: a possible link?	Hum Genet.	133	225-34	2014
Ohba C, others, **Miyake N**, Eto Y, Tanaka F, **Matsumoto N**, Saitsu H.	Diagnostic utility of whole exome sequencing in patients showing cerebellar and/or vermis atrophy in childhood.	Neurogenetics.	14	225-32	2013
Nakajima J, **Okamoto N**, others, **Matsumoto N**, **Miyake N**.	Novel *FIG4* mutations in Yunis-Varon syndrome.	J Hum Genet.	58	822-4	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
Nishiguchi KM, Tearle RG, Liu YP, Oh EC, **Miyake N**, others, **Matsumoto N**, Terasaki H, Berson EL, Katsanis N, Rivolta C.	Whole genome sequencing in patients with retinitis pigmentosa reveals pathogenic DNA structural changes and NEK2 as a new disease gene.	Proc Natl Acad Sci U S A.	110	16139-44	2013
Nakamura K,others, **Miyake N**, Hayasaka K, Ogata K, Fukuda A, **Matsumoto N**, Saitsu H.	De Novo mutations in *GNAO1*, encoding a Gαo subunit of heterotrimeric G proteins, cause epileptic encephalopathy.	Am J Hum Genet.	93	496-505	2013
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Miyake N, Koshimizu E, **Okamoto N**, others, **Matsumoto N**, Niikawa N.	*MLL2* and *KDM6A* mutations in atients with Kabuki syndrome.	Am J Med Genet A.	161	2234-43	2013
Doi H, Ohba C, Tsurusaki Y, Miyatake S, **Miyake N**, others, **Matsumoto N**.	Identification of a novel homozygous *SPG7* mutation in a Japanese patient with spastic ataxia: making an efficient diagnosis using exome sequencing for autosomal recessive cerebellar ataxia and spastic paraplegia.	Intern Med.	52	1629-33	2013
Tsurusaki Y, Yonezawa R, Furuya M, Nishimura G, Pooh R, Nakashima M, Saitsu H, **Miyake N**, Saito S, **Matsumoto N**.	Whole exome sequencing revealed biallelic *IFT122* mutations in a family with CED1 and recurrent pregnancy loss.	Clin Genet.	(in press)	(in press)	2013
Fujita A, Suzumura H, Nakashima M, Tsurusaki Y, Saitsu H, Harada N, **Matsumoto N**, **Miyake N**.	A unique case of de novo 5q33.3-q34 triplication with uniparental isodisomy of 5q34-qter.	Am J Med Genet A.	161	1904-9	2013
Tsurusaki Y, **Okamoto N**, others, **Miyake N**, **Matsumoto N**.	Coffin-Siris syndrome is a SWI/SNF complex disorder	Clin Genet.	(in press)	(in press)	2013
Ravenscroft G, Miyatake S, Lehtokari VL, Todd EJ, Vornanen P, Yau KS, Hayashi YK, **Miyake N**, others, **Matsumoto N**, Laing NG.	Mutations in *KLHL40* are a frequent cause of severe autosomal-recessive nemaline myopathy.	Am J Hum Genet	93	6-18	2013

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Kondo Y, Koshimizu E, Megarbane A, others, Miyatake S, Tsurusaki Y, Nakashima M, Doi H, **Miyake N**, Saitsu H, **Matsumoto N**.	Whole-exome sequencing identified a homozygous *FNBP4* mutation in a family with a condition similar to microphthalmia with limb anomalies.	Am J Med Genet A.	161	1543-6	2013
Nakajima M, Mizumoto S, **Miyake N**, others, **Matsumoto N**, Sugahara K, Nishimura G, Ikegawa S.	Mutations in *B3GALT6*, which Encodes a Glycosaminoglycan Linker Region Enzyme, Cause a Spectrum of Skeletal and Connective Tissue Disorders.	Am J Hum Genet.	92	927-34	2013
Kodera H, Kato M, others, **Miyake N**, Hayasaka K, King MC, **Matsumoto N**, Saitsu H.	Targeted capture and sequencing for detection of mutations causing early onset epileptic encephalopathy.	Epilepsia.	54	1262-9	2013
Kosho T, **Okamoto N**, Ohashi H, Tsurusaki Y, Imai Y, Hibi-Ko Y, Kawame H,others, **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.	161	1221-37	2013
Nakamura K, Nishiyama K, Kodera H, Nakashima M, Tsurusaki Y, **Miyake N**, **Matsumoto N**, others, Enoki H.	A *de novo CASK* mutation in pontocerebellar hypoplasia type 3 with early myoclonic epilepsy and tetralogy of Fallot.	Brain Dev.	(in press)	(in press)	2013
Kato M, Yamagata T, others, **Miyake N**, Saito K, Hayasaka K, **Matsumoto N**, Saitsu H.	Clinical spectrum of early onset epileptic encephalopathies caused by *KCNQ2* mutation.	Epilepsia.	54	1282-7	2013

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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
Kondo Y, Saitsu H, Miyamoto T, Lee BJ, Nishiyama K, Nakashima M, Tsurusaki Y, Doi H, **Miyake N**, Kim JH, Yu YS, **Matsumoto N**.	Pathogenic mutations in two families with congenital cataract identified with whole-exome sequencing.	Mol Vis.	19	384-9	2013
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Higashiyama Y, Doi H, Wakabayashi M, Tsurusaki Y, **Miyake N**, others, **Matsumoto N**.	A novel *SCARB2* mutation causing late-onset progressive myoclonus epilepsy.	Mov Disord.	28	552-3	2013
Takanashi JI, Osaka H, Saitsu H, others, Inoue K, **Matsumoto N**, James Barkovich A.	Different patterns of cerebellar abnormality and hypomyelination between *POLR3A* and *POLR3B* mutations.	Brain Dev.	(in press)	(in press)	2013
Nakayama T, Saitsu H, Endo W, Kikuchi A, others, Kure S, **Matsumoto N**.	RBPJ is disrupted in a case of proximal 4p deletion syndrome with epilepsy.	Brain Dev.	(in press)	(in press)	2013
Okamoto N, Yamagata T, Yada Y, Ichihashi K, **Matsumoto N**, Momoi MY, Miyuguchi T.	Williams-Beuren syndrome with brain malformation and hypertrophic cardiomyopathy.	Brain Dev.	(in press)	(in press)	2013

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Nonoda Y, Saito Y, Nagai S, Sasaki M, Iwasaki T, **Matsumoto N**, Ishii M, Saitsu H	Progressive diffuse brain atrophy in West syndrome with marked hypomyelination due to *SPTAN1* gene mutation.	Brain Dev.	35	280-3	2013
Yokoo N, Marumo C, Nishida Y, Iio J, Maeda S, Nonaka M,others, **Matsumoto N**, **Okamoto N**	A case of Toriello-Carey syndrome with severe congenital tracheal stenosis.	Am J Med Genet A.	161	2291-3	2013
Sun SL, Horino S, Itoh-Nakadai A, Kawabe T, Asao A, Takahashi T, So T, Funayama R, Kondo M, Saitsu H, **Matsumoto N**, Nakayama K, Ishii N.	Y chromosome-linked B and NK cell deficiency in mice.	J Immunol.	190	6209-20	2013