

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

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
Tsurusaki Y, et al., Matsumoto N.	Exome sequencing identifies an <i>OFD1</i> mutation in a family of X-linked lethal congenital malformation syndrome: delineation of male Oral-facial-digital syndrome type 1.	Clin Genet	83 (2)	135-144	2013
Tsurusaki Y, et al., Matsumoto N.	The diagnostic utility of exome sequencing in Joubert syndrome related disorders.	J Hum Genet	58(2)	113-115	2013
Kondo Y, et al., Matsumoto N.	Whole-exome sequencing identified a homozygous <i>FBNP4</i> mutation in a family with a condition microphthalmia with limb anomalies-like	Am J Med Genet Part A	161A	1543-1546	2013
Miyake N, et al., Matsumoto N.	Mitochondrial complex III deficiency caused by a homozygous <i>UQCRC2</i> mutation presenting with neonatal-onset recurrent metabolic decompensation.	Hum Mut	34(3)	446-452	2013
Saitu H, et al., Matsumoto N.	<i>De novo</i> mutations in the autophagy gene <i>WDR45</i> cause static encephalopathy of childhood with neurodegeneration in adulthood.	Nat Genet	45(4)	445-449	2013

Kondo Y, et al., Matsumoto N.	Pathogenic mutations in two families with congenital cataract identified by whole-exome sequencing.	Mol Vis	19	384-389	2013
Nakamura K, et al., Matsumoto N, Saitsu H.	Clinical spectrum of <i>SCN2A</i> mutations expanding to Ohtahara syndrome.	Neurology	81(11)	992-998	2013
Koshimizu E, et al., Matsumoto N.	Exome sequencing identifies an <i>OFD1</i> mutation in a family of X-linked lethal congenital malformation syndrome: delineation of male Oral-facial-digital syndrome type 1.	Plos One	8(9)	e74167	2013
Nakajima M, et al., Matsumoto N, et al..	Mutations in <i>B3GALT6</i> , which encodes a glycosaminoglycan linker region enzyme, cause a spectrum of skeletal and connective tissue disorders.	Am J Hum Genet	92(6)	927-934	2013
Iida A, et al., Matsumoto N, Ikegawa S.	Exome sequencing identifies a novel <i>INPPL1</i> mutation in opsismodysplasia.	J Hum Genet	58(6)	391-394	2013
Nishiguchi KM, et al., Matsumoto N, et al.	Whole genome sequencing in patients with retinitis pigmentosa reveals pathogenic DNA structural changes and <i>NEK2</i> as a new disease gene.	Proc Natl Acad Sci USA	110(40)	16139-16144	2013
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Kodera H, et al., Matsumoto N, Saitu H.	De novo mutations in SLC35A2 encoding a UDP-galactose transporter cause early-onset epileptic encephalopathy.	Hum Mut	34(12)	1708-1714	2013
Ohba C, et al., Matsumoto N, Saitu H.	Diagnostic utility of whole exome sequencing in cerebellar atrophy in childhood.	Neurogenet	14 (3-4)	225-232	2013
Gupta VA, et al., Matsumoto N, et al.	Identification of KLHL41 mutations implicates BTB-Kelch-mediated ubiquitination as an alternate pathway to myofibrillar disruption in nemaline myopathy.	Am J Hum Genet	93(6)	1108-1117	2013
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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
Wada T, Ban H, Matsufuji M, Okamoto N, Enomoto K, Kurosawa K, Aida N.	Neuroradiologic Features in X-linked $\alpha$ -Thalassemia/Mental Retardation Syndrome.	Am J Neuroradiology	34	2034-8.	2013
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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
Mitani T, Aida N, Tomiyasu M, Wada T, Osaka H.	Transient ischemic attack-like episodes without stroke-like lesions in MELAS.	Pediatr Radiol	43	1400-1403	2013;
Anselm I, Azzouz H,(28名) Osaka H, (22名) Mancini GM, Salomons GS.	Phenotype and genotype in 101 males with X-linked creatine transporter deficiency.	J Med Genet	50	463-472	2013
Kato H, Miyake F, Shimbo H, Ohya M, Sugawara H, Aida N, Anzai R, Takagi M, Okuda M, Takano K, Wada T, Iai M, Yamashita S, Osaka H.	Urine screening for patients with developmental disabilities detected a patient with creatine transporter deficiency due to a novel missense mutation in SLC6A8.	Brain Dev	13	S0387-7604	2013
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<p>Abe J, Nakamura K, (14名) Osaka H, (9名) Ichinose H, Heike T.</p>	<p>A nationwide survey of Aicardi-Goutieres syndrome patients identifies a strong association between dominant TREX1 mutations and chilblain lesions: Japanese cohort study.</p>	<p>Rheumatology (Oxford)</p>			<p>印刷中</p>
<p>Okabe T, Aida N, Niwa T, Nozawa K, Shibasaki J, Osaka H.</p>	<p>Early magnetic resonance detection of cortical necrosis and acute network injury associated with neonatal and infantile cerebral infarction.</p>	<p>Pediatr Radiol</p>			<p>印刷中</p>