**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 |
|---|---|---|---|---|---|
| Miyatake S,*Matsumoto N (*: correspondence). | Clinical exome sequencing in neurology practice. | Nat Rev Neurol | 10(12) | 676-678, 2 | 2014 |
| Tsurusaki Y, et al., *Matsumoto N. | De novo SOX11 mutations cause Coffin-Siris syndrome. Nat Commun | Nat Commun | 5 | 4011 | 2014 |
| Miyatake S, et al., *Matsumoto N, *Saitsu H (*: co-correspondence) | Expanding the phenotypic spectrum of TUBB4A-associated hypomyelinating leukoencephalopathies, | Neurology | 82(24) | 2230-2237 | 2014 |
| *Kato M, et al., Matsumoto N. | PIGA mutations cause early-onset epileptic encephalopathies and distinctive features. | Neurology | 82(18) | 1587-1596, | 2014 |
| *Nakamura K, et al., Matsumoto N, Saitsu H. | AKT3 and PIK3R2 mutations in two patients with megalencephaly-related syndromes. | Clin Genet | 85(4): | 396-398 | 2014 |
| *Miyake N, et al., Matsumoto N. | Ehlers–Danlos syndrome associated with glycosaminoglycan abnormalities. J. Halper (ed.), Progress in heritable soft connective Tissue diseases, | Advances in Experimental Medicine and Biology | 802 | 145-159 | 2014 |

| | | | | | |
|---|---|---|---|---|---|
| Ohba C, et al., *Matsumoto N, *Saitsu H (*: co-corresponden ce). | PIGN mutations cause congenital anomalies, developmental delay, hypotonia, epilepsy, and progressive cerebellar atrophy. | Neurogenet | 59(5) | 292-295 | 2014 |
| Leventer RJ, et al., Matsumoto N, et al. | Is Focal Cortical Dysplasia sporadic? Family evidence for genetic susceptibility. | Epilepsia | 55(3) | e22-26 | 2014 |
| Ohba C, et al., *Matsumoto N, *Saitsu H (*: co-corresponden ce). | Early onset epileptic encephalopathy caused by de novo SCN8A mutations. | Epilepsia | 55(7) | 994-1000 | 2014 |
| #Miyatake S, #Koshimizu E (# denotes equal contribution), et al., *Matsumoto N. | Deep sequencing detects very low-grade somatic mosaicism in the unaffected mother of siblings with nemaline myopathy. | Neuromuscul Disord | 24(7) | 642-647 | 2014 |
| Nakashima M, et al., Matsumoto N*. | Novel compound heterozygous PIGT mutations caused multiple congenital anomalies-hypotonia-s eizures syndrome 3. | Neurogenet | 15(3) | 193-200 | 2014 |
| *Miyake N (*: corresponding author), et al., Matsumoto N. | Numerous BAF complex genes are mutated in Coffin-Siris syndrome. | Am J Med Genet Part C | 166(3) | 257-261 | 2014 |
| Katagiri S, et al., Matsumoto N, et al. | Whole exome analysis identifies frequent CNGA1 mutations in Japanese population with autosomal recessive retinitis pigmentosa. | Plos One | 9(9) | e108721 | 2014 |
| Tsurusaki Y, et al., *Matsumoto N | Whole exome sequencing revealed causative biallelic IFT122 mutations in a family with CED1 and recurrent pregnancy loss. | Clin Genet | 85(6) | 592-594 | 2014 |

| | | | | | |
|---|---|---|---|---|---|
| Nakamura K, et al., *Matsumoto N, *Saitsu H (*: co-corresponden ce). | PIGO mutations in epileptic encephalopathy with mild elevation of alkaline phosphatase levels. | Epilepsia | 55(2) | e13-7 | 2014 |
| Tsurusaki Y, et al., *Matsumoto N. | Coffin-Siris syndrome is a SWI/SNF complex disorder. | Clin Genet | 85(6) | 548-554 | 2014 |
| Ohashi T, et al., Matsumoto N, et al. | Infantile epileptic encephalopathy with a hyperkinetic movement disorder and hand stereotypies arising from a novel SCN1A mutation. | Epileptic Disord | 16(2) | 208-212 | 2014 |
| Imagawa E, et al., *Matsumoto N, *Miyake N | A hemizygous GYG2 mutation causes Leigh syndrome. | Hum Genet | 133 (2) | 225-234 | 2014 |
| Okabe T, Aida N, Niwa T, Noz awa K, Shibasak i J, Osaka H. | Early magnetic resonance detection of cortical necrosis and acute network injury associated with neonatal and infantile cerebral infarction. | Pediatr Radiol | 53 | 448-458 | 2014 |
| Akiyama T, Osaka H, Shimbo H, Nakajiri T, Kobayashi K, Oka M, Endoh F,Yoshinaga H. | A Japanese Adult Case of Guanidinoacetate Methyltransferase Deficiency. | JIMD Rep | 12 | 65-69 | 2014 |
| Wada T, Haddad MR, Yi L, Murakami T, Sasaki A, Shimbo H, Kodama H, Osaka H, Kaler SG. | A Novel Two-Nucleotide Deletion in the ATP7A Gene Associated With Delayed Infantile Onset of Menkes Disease. | Pediatr Neurol | 50 | 417-420 | 2014 |
| Shimbo H, Takagi M, Okuda M, Tsuyusaki Y, Takano K, Iai M, Yamashita S, Murayama K, Ohtake A, Goto Y, AidaN, Osaka | A rapid screening with direct sequencing from blood samples for the diagnosis of Leigh syndrome. | Mol Genet Metab Report | 1 | 133-138 | 2014 |

| | | | | | |
|---|---|---|---|---|---|
| Ohshiro-Sasaki A, Shimbo H, Takano K, Wada T, Osaka H. | A Three-Year-Old Boy With Glucose Transporter Type 1 Deficiency Syndrome Presenting With Episodic Ataxia. | Pediatr Neurol | 50(1) | 99-100 | 2014 |
| Numata Y, Gotoh L, Iwaki A, Kurosawa K, Takanashi J, Deguchi K, Yamamoto T,Osaka H, Inoue K. | Epidemiological, clinical, and genetic landscapes ofhypomyelinating leukodystrophies. | J Neurol | 261(4) | 752-758 | 2014 |
| Numasawa-Kuroiwa Y, Okada Y, Shibata S, Kishi N, Akamatsu W, Shoji M, Nakanishi A, Oyama M, Osaka H, Inoue K, Takahashi K, Yamanaka S, | Involvement of ER Stress in Dysmyelination of Pelizaeus-Merzbacher Disease with PLP1 Missense Mutations Shown by iPSC-Derived Oligodendrocytes. | Stem Cell Reports | 24;2(5) | 648-661 | 2014 |
| Yasuda Y, **Hashimoto R**, Ohi K, Yamamori H, Fujimoto M, Umeda-Yano S, Fujino H, Takeda M. | Cognitive inflexibility in Japanese adolescents and adults with autism spectrum disorders. | Workd J Psychiatry | 22;4(2) | 42-48 | 2014 |