

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
高橋幸利 東本和紀	てんかん	編集: 山崎麻美	小児脳神経外科学	金芳堂	東京		印刷中

雑誌 (和文・英文とも)

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
Ohba C, Okamoto N, Murakami Y, Suzuki Y, Tsurusaki Y, Nakahima M, Miyake N, Tanaka F, Kinoshita T, Matsumoto N, Saitsu	H. PIGN mutations cause congenital anomalies, developmental delay, hypotonia, epilepsy, and progressive cerebellar atrophy.	<i>Neurogenetics</i> .	15(2)	85-92	2014
Martin, H. C., G. Kim, A. T. Pagnamenta, Y. Murakami, G. Carvill, E. Meyer, R. Copley, A. Rimmer, G. Barcia, M. Fleming, J. Kronegold, M. R. Brown, K. A. Hudspith, J. Broxholme, A. Kanapin, J.-B. Cazier, T. Kinoshita, R. Nabbout, The WGS600 Consortium, D. Bentley, G. McVean, S. Heavin, Z. Zaiwalla, T. McShane, H. Mefford, D. Shears, H. Stewart, M. A. Kurian, I. E. Scheffer, E. Blair, P. Donnelly, L. K. Kaczmarek and J. Taylor.	whole-genome sequencing in severe early-onset epilepsy reveals new genes and improves molecular diagnosis.	<i>Hum. Mol. Genet.</i>	23	3200-3211	2014
	Clinical				

Kato, M <sup>1</sup> ., H. Saitsu <sup>1</sup> , Y. Murakami <sup>1</sup> , K. Kikuchi, S. Watanabe, M. Iai, K. Miya, R. Matsuyama, R. Takayama, C. Ohba, M. Nakashima, Y. Tsurusaki, N. Miyake, S. Hamano, H. Osaka, K. Hayasaka, T. Kinoshita and N. Matsumoto.	<i>PIGA</i> mutations cause early-onset epileptic encephalopathies and distinctive features.	<i>Neurology</i> ,	82	1587-1596	2014
Murakami, Y., H. Tawamie, Y. Maeda, C. Buttner, R. Buchert, F. Radwan, S. Schaffer, H. Sticht, M. Aigner, A. Reis, T. Kinoshita and R. A. Jamra.	Null mutation in <i>PGAP1</i> impairs GPI-anchor maturation and causes severe non-syndromic recessive intellectual disability.	<i>PLoS Genet.</i>	10(5)	e1004320.	2014
Nakashima, M., H. Kashii, Y. Murakami, M. Kato, Y. Tsurusaki, N. Miyake, M. Kubota, T. Kinoshita, H. Saitsu, N. Matsumoto.	Novel compound heterozygous <i>PIGT</i> mutations caused multiple congenital anomalies-hypotonia-seizures syndrome 3.	<i>Neurogenet.</i> ,	15	193-200	2014
Ueda, Y., J. Nishimura, Y. Murakami, S. Kajigaya, T. Kinoshita, Y. Kanakura and N. S. Young.	Paroxysmal nocturnal hemoglobinuria with copy number-neutral 6pL OH in GPI (+) but not in GPI (-) granulocytes.	<i>Eur. J. Haematol.</i>	92:	450-453 DOI: 10.1111/ejh.12253	2014.
Stokes, M., Y. Murakami, Y. Maeda, T. Kinoshita and Y. S. Morita.	New insights to the functions of PIGF, a protein involved in the ethanolamine phosphate transfer steps of glycosylphosphatidylinositol biosynthesis.	<i>Biochem. J.</i> ,	463(2)	249-256	2014
Fujiwara I, Murakami Y, Niihori T, Kanno J, Hakoda A, Sakamoto O, Okamoto N, Funayama R, Nagashima T, Nakayama K, Kinoshita T, Kure S, Matsubara Y, Aoki Y.	Mutations in <i>PIGL</i> in a patient with Mabry syndrome.	<i>Am J Med Genet A.</i>		doi: 10.1002/ajmg.a.36987	2015

村上良子 木下タロウ	知的障害とてんかんを主症状とする新しい疾患—先天性 GPI 欠損症	脳と発達誌	第 47 巻 1 号	5-13	2015
Inoue N, Akazawa T.	IL17A (interleukin 17A).	Atlas of Genetics and Cytogenetics in Oncology and Haematology	19	18-27	2014
Akazawa T, Ohashi T, Nakajima H, Nishizawa Y, Kodama K, Sugiyama K, Inaba T, <u>Inoue N.</u>	Development of a dendritic cell-targeting lipopeptide as an immunoadjuvant that inhibits tumor growth without inducing local inflammation.	<i>Int. J. Cancer</i>	135	2847-2856	2014
Nobusuke Kimura, <u>Yukitoshi Takahashi,</u> Hideo Shigematsu, Katsumi Imai, Hiroko Ikeda, Hideyuki Ootani, Rumiko Takayama, Yukiko Mogami, Noriko Kimura, Koichi Baba, Kazumi Matsuda, Takayasu Tottori, Naotaka Usui, Yushi Inoue	Developmental outcome after surgery in focal cortical dysplasia patients with early-onset epilepsy.	Epilepsy Research	108(10)	1845-1852	2014
Hiroyuki Fujita Hiroyuki, Matsukura S, Watanabe T, Komitsu N, Watanabe Y, <u>Takahashi Y,</u> Kambara T, Ikezawa Z, Aihara M	The serum level of HMGB1 (high mobility group box 1 protein) is preferentially high in drug-induced hypersensitivity syndrome/drug reaction with eosinophilia and systemic symptoms.	British Journal of Dermatology	171(6)	1585-1588	2014

Chiba Yuhei, Katsuse Omi, Fujishiro Hiroshige, Kamada Ayuko, Saito Tomoyuki, Ikura Takahiro, <u>Takahashi Yukitoshi</u> , Kunii Misako, Takeno Mitsuhiro Hirayasu, Yoshio	Lymphopenia Helps Early Diagnosis of Systemic Lupus Erythematosus for Patients with Psychosis as an Initial Symptom	Psycho-somatics.		2013 Sep 23. doi-pii: S0033-3182(13)00136-9. 10.1016/j.psym..	in press
Takahiro Furukawa, Naoko Matsui, Koji Fujita, Ai Miyashiro, Yuishin Izumi, Fumitaka Shimizu, Katsuichi Miyamoto, <u>Yukitoshi Takahashi</u> , Takashi Kanda, Susumu Kusunoki, Ryuji Kaj	Increased proinflammatory cytokines in sera of patients with multifocal motor neuropathy.	J Neurol Sci.		2014 Aug 4. pii: S0022-510X(14)00505-X. doi: 10.1016/j.jns.2014.07.059.	in press
Hiroshi Sakuma, Naoyuki Tanuma, Ichiro Kuki, <u>Yukitoshi Takahashi</u> , Masashi Shiomi, Masaharu Hayashi	Intrathecal overproduction of pro-inflammatory cytokines and chemokines in febrile infection related refractory status epilepticus.	Journal of Neurology, Neurosurgery, and Psychiatry			in press
Masaki Yoshimura, Zhang Shouwen, Yuki Ueda, Kazumi Matsuda, Katsumi Imai, <u>Yukitoshi Takahashi</u> , Yushi Inoue	An analysis of epileptic negative myoclonus by magnetoencephalography.	Epilepsy Research			in press
Norimichi Higurashi, <u>Yukitoshi Takahashi</u> , Ayako Kashimada, Yuji Sugawara, Hiroshi Sakuma, Yuko Tomono, Takahito Inoue, Megumi Hoshina, Ruri Satomi, Masaharu Ohfu, Kazuya Itomi, Kyoko Takano, Tomoko Kirino, Shinichi Hirose	Immediate suppression of seizure clusters by corticosteroids in PCDH19 female epilepsy.	Seizure			in press

高橋幸利、森達夫、 大星大観、東本和紀、	神経疾患と NMDA 型 グルタミン酸受容	日本小児科学会誌	118(12)	1695-1707	2014
高橋幸利、木水友一、 小池敬義、堀野朝子、	免疫性神経疾患-基 礎・臨床研究の最新	日本臨床 増刊号			印刷中