

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
Inoue, N., Kinoshita, T.	Pathogenesis of Clonal Dominance in PNH; Growth Advantage in PNH.	Kanakura, Y., Kinoshita, T., Nishimura, J.	Paroxysmal Nocturnal Hemoglobinuria-From bench to bedside	Springer-Verlag	Tokyo	2016	In press
井上徳光	PNH型細胞のクローン性拡大の機序	金倉謙、西村純一	発作性夜間ヘモグロビン尿症(PNH)～遺伝子異常の解析から新規治療の臨床・開発まで～	医薬ジャーナル社	大阪	2016	69-82
高橋幸利、西田拓司、山口解冬	自己免疫性脳炎	編集：辻省次、吉良潤一	アキュアル脳・神経疾患の臨床、免疫性神経疾患病態と治療のすべて	中山書店	東京	2016	270-279
高橋幸利	診断(免疫・その他の診断)		「てんかん白書」～てんかん医療・研究のアクションプラン				印刷中
高橋幸利	臨床研究(小児期)		「てんかん白書」～てんかん医療・研究のアクションプラン				印刷中

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
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 PIGL in a patient with Mabry syndrome.	Am J Med Genet A.	167A(4)	777-85	2015
Hirata T, Fujita M, Nakamura S, Gotoh K, Motooka D, Murakami Y, Maeda Y, Kinoshita T.	Post-Golgi anterograde transport requires GARP-dependent endosome-to-TGN retrograde transport.	Mol Biol Cell.	26(17)	3071-84	2015

Ilkovski B, Pagnamenta AT, O'Grady GL, Kinoshita T, Howard MF, Lek M, Thomas B, Turner A, Christodoulou J, Sillence D, Knight SJ, Popitsch N, Keays DA, Anzilotti C, Goriely A, Waddell LB, Brilot F, North KN, Kanzawa N, Macarthur DG, Taylor JC, Kini U, <u>Murakami Y</u> , Clarke NF.	Mutations in PIGY: expanding the phenotype of inherited glycosylphosphatidylinositol deficiencies.	Hum Mol Genet.	24(21)	6146-59	2015
Bosch DG, Boonstra FN, Kinoshita T, Jhangiani S, de Ligt J, Cremers FP, Lupski JR, <u>Murakami Y</u> , de Vries BB.	Cerebral visual impairment and intellectual disability caused by PGAP1 variants.	Eur J Hum Genet.	23(12)	1689-93	2015
Nakagawa T, Taniguchi-Ikeda M ¹ , <u>Murakami Y</u> ¹ , Nakamura S, Motooka D, Emoto T, Satake W, Nishiyama M, Toyoshima D, Morisada N, Takada S, Tairaku S, Okamoto N, Morioka I, Kurahashi H, Toda T, Kinoshita T, Iijima K. (¹ Correspondence)	A novel <i>PIGN</i> mutation and prenatal diagnosis of inherited glycosylphosphatidylinositol deficiency.	Am J Med Genet A	170(1)	183-8	2016
Makrythanasis P, Kato M, Zaki MS, Saitu H, Nakamura K, Santoni FA, Miyatake S, Nakashima M, Issa MY, Guipponi M, Letourneau A, Logan CV, Roberts N, Parry DA, Johnson CA, Matsumoto N, Hamamy H, Sheridan E, Kinoshita T, Antonarakis SE, <u>Murakami Y</u> .	Pathogenic Variants in PIGG Cause Intellectual Disability with Seizures and Hypotonia.	Am J Hum Genet.	98(4)	615-26	2016
Roy, N., Ohtani, K., Matsuda, Y., Mori, K., Hwang, I., Suzuki, Y., <u>Inoue, N.</u> , Wakamiya, N.	Collectin CL-P1 utilizes C-reactive protein for complement activation.	<i>Biochim Biophys Acta</i>	1860	1118-1128	2016
Nakanishi, K., Kukita, Y., Segawa, H., <u>Inoue, N.</u> , Ohue, M., and Kato, K.	Characterization of the T cell receptor beta chain repertoire in tumor infiltrating lymphocytes	<i>Cancer Medicine</i>			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	27	1-5	2015
Tetsuhiro Fukuyama, <u>Yukitoshi Takahashi</u> , Yuko Kubota, Yukiko Mogami, Katsumi Imai, Yoshiyuki Kondo, Hiroshi Sakuma, Koji Tominaga, Hirokazu Oguni, Shigeko Nishimura	Semi-quantitative analyses of antibodies to N-methyl-D-aspartate type glutamate receptor subunits (GluN2B & GluN1) in the clinical course of Rasmussen syndrome.	Epilepsy Research	113	34-43	2015
Shinsaku Yoshitomi, <u>Yukitoshi Takahashi</u> , Mamiko Hotate Ishizuka, Tokito Yamaguchi, Akito Watanabe, Hirosato Nasu, Yuki Ueda, Hideyuki Ohtani, Hiroko Ikeda, Katsumi Imai, Hideo Shigematsu, Yushi Inoue, Yoshihiro Tanahashi, Kaori Aiba, Hodaka Ohta, Shino Shimada, Toshiyuki Yamamoto	Three patients manifesting early infantile epileptic spasms associated with 2q24.3 microduplications.	Brain & Development	37(9)	874-879	2015
Mori T, Imai K, Oboshi T, Fujiwara Y, Takeshita S, Saitsu H, Matsumoto N, <u>Takahashi Y</u> , Inoue Y.	Usefulness of ketogenic diet in a girl with migrating partial seizures in infancy.	Brain & Development	Jan 11		2016
Hiroko Ikeda, Katsumi Imai, Hitoshi Ikeda, Hideo Shigematsu, <u>Yukitoshi Takahashi</u> , Yushi Inoue, Norimichi Higurashi, Shinichi Hirose	Characteristic phasic evolution of convulsive seizure in PCDH19-related epilepsy.	Epileptic Disord.	18	26-33	2016
<u>高橋幸利</u>	特別企画シリーズ：てんかんを分かり 易く理解するための神経科学、6：免 疫	てんかん研 究	33	683-687	2016