

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. and Nishimura, J.	<i>Paroxysmal Nocturnal Hemoglobinuria-From bench to bedside</i>	Springer Japan	Tokyo	2017	229-251
<u>Yukitoshi Takahashi et al.,</u>	Autoimmune-mediated encephalitis with antibodies to NMDA-type GluRs: Early clinical diagnosis.	Yamanouchi H, et al.,	Acute Encephalopathy and Encephalitis in Infancy and Its Related Disorders.	Elsevier		2018	151-156
Kiyoshi Egawa, YukitoshiTakahashi	Epilepsy in Dentato-rubro-pallido-luysian atrophy (DRPLA).	Shorvon et al.,	The Causes of Epilepsy.	Cambridge University Press		in press	
高橋幸利、堀野朝子	Rasmussen脳炎(症候群)		稀少てんかん診療指標	診断と治療社	東京	2017年	142-145
高橋幸利、小池敬義	その他の内科的薬物治療		稀少てんかん診療指標	診断と治療社	東京	2017年	203-206
高橋幸利、大松泰生	免疫とてんかん		稀少てんかん診療指標	診断と治療社	東京	2017年	23-27
高橋幸利	免疫介在性てんかん/抗体介在性てんかん(てんかんの自己抗体を含む)	須貝研司	てんかん学用語事典	診断と治療社	東京	2017年	77

## 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
Pagnamenta AT, <u>Murakami Y</u> , Taylor JM, Anzilotti C, Howard MF, Miller V, Johnson DS, Tadros S, Mansour S, Temple IK, Firth R, Rosser E, Harrison RE, Kerr B, Popitsch N; DDD Study, Kinoshita T, Taylor JC, Kini U.	Analysis of exome data for 4293 trios suggests GPI-anchor biogenesis defects are a rare cause of developmental disorders.	Eur J Hum Genet.	25(6)	669-679	2017
Johnstone DL, Nguyen TT, <u>Murakami Y</u> , Kernohan KD, Tétreault M, Goldsmith C, Doja A, Wagner JD, Huang L, Hartley T, St-Denis A, le Deist F, Majewski J, Bulman DE; Wagner JD, Huang L, Hartley T, St-Denis A, le Deist F, Majewski J, Bulman DE;	Johnstone DL, Nguyen TT, <u>Murakami Y</u> , Kernohan KD, Tétreault M, Goldsmith C, Doja A, Wagner JD, Huang L, Hartley T, St-Denis A, le Deist F, Majewski J, Bulman DE;	Hum Mol Genet.	26(9)	1706-1715	2017
Tanigawa J, Mimatsu H, Mizuno S, Okamoto N, Fukushi D, Tominaga K, Kidokoro H, Muramatsu Y, Nishi E, Nakamura S, Motooka D, Nomura N, Hayasaka K, Niihori T, Aoki Y, Nabatame S, Hayakawa M, Natsume J, Ozono K, Kinoshita T, Wakamatsu N, <u>Murakami Y</u> .	Phenotype-genotype correlations of PIGO deficiency with variable phenotypes from infantile lethality to mild learning difficulties.	Hum Mutat..	38(7)	805-815	2017

Kohashi K, Ishiyama A, Yuasa S, Tanaka T, Miya K, Adachi Y, Sato N, Saitsu H, Ohba C, Matsumoto N, <u>Murakami Y</u> , Kinoshita T, Sugai K, Sasaki M	Epileptic apnea in a patient with inherited glycosylphosphatidylinositol anchor deficiency and PIGT mutations.	Brain Dev	40(1)	53-57.	2018
Nguyen TTM, <u>Murakami Y</u> , Sheridan E, Ehresmann S, Rousseau J, St-Denis A, Chai G, Ajeawung NF, Fairbrother L, Reimschisel T, Bateman A, Berry-Kravis E, Xia F, Tardif J, Parry DA, Logan CV, Diggle C, Bennett CP, Hattingh L, Rosenfeld JA, Perry MS, Parker MJ, Le Deist F, Zaki MS, Ignatius E, Isohanni P, Lönnqvist T, Carroll CJ, Johnson CA, Gleeson JG, Kinoshita T, Campeau PM.	Mutations in GPAA1, Encoding a GPI Transamidase Complex Protein, Cause Developmental Delay, Epilepsy, Cerebellar Atrophy, and Osteopenia.	Am J Hum Genet.	101(5)	856-865	2017
Liu YS, Guo XY, Hirata T, Rong Y, Motooka D, Kitajima T, <u>Murakami Y</u> , Gao XD, Nakamura S, Kinoshita T, Fujita M.	N-Glycan-dependent protein folding and endoplasmic reticulum retention regulate GPI-anchor processing.	J Cell Biol.	217(2)	585-599	2017

Hirata T, Mishra SK, Nakamura S, Saito K, Motooka D, Takada Y, Kanzawa N, <u>Murakami Y</u> , Maeda Y, Fujita M, Yamaguchi Y, Kinoshita T.	Identification of a Golgi GPI-N-acetylgalactosamine transferase with tandem transmembrane regions in the catalytic domain.	Nat Commun.	9(1)	405	2018
Mogami Y, Suzuki Y, <u>Murakami Y</u> , Ikeda T, Kimura S, Yanagihara K, Okamoto N, Kinoshita T.	Early infancy-onset stimulation-induced myoclonic seizures in three siblings with inherited glycosylphosphatidylinositol (GPI) anchor deficiency.	Epileptic Disord.	20(1)	42-50	2018
Pagnamenta AT, <u>Murakami Y</u> , Anzilotti C, Titheradge H, Oates AJ, Morton J; DDD Study, Kinoshita T, Kini U, Taylor JC.	A homozygous variant disrupting the PIGH start-codon is associated with developmental delay, epilepsy, and microcephaly.	Hum Mutat.	doi: 1 0.1002 /humu. 23420.		2018
Kojima-Kita, K., Kuramochi-Miyagawa, S., Ogonuki, N., Ogura, A., Hasuwa, H., Akazawa, T., <u>Inoue</u> , N., Nakano, T.	MIWI2 as an effector of DNA methylation and gene silencing in embryonic male germ cells	MIWI2 as an effector of DNA methylation and gene silencing in embryonic male germ cells			
Nakanishi, K., Kukit a, Y., Segawa, H., <u>Inob</u> ue, N., Ohue, M., Kat o, K.	Characterization of the T cell receptor beta chain repertoire in tumor infiltrati ng lymphocytes	Cancer Med.	5(9)	2513-25 21	2016
Roy, N., Ohtani, K., Matsuda, Y., Mori, K., Hwang, I., Suzuk i, Y., <u>Inoue</u> , N., Waka miya, N.	Collectin CL-P1 utilizes C-reactive prote in for complement activation.	Biochim Biophys A cta - General Subj ects	1860 (6)	1118-11 28	2016
Roy, N., Ohtani, K., Hidaka, Y., Amano, Y., Matsuda, Y., Mori, K., Hwang, I., <u>Inoue</u> , N., Wakamiya, N.	Three pentraxins C-reactive protein, ser um amyloid p component and pentraxin 3 mediate complement activation using Collectin CL-P1	Biochim Biophys A cta - General Subj ects	861(2)	1-14	2017
Ohashi, T., Aoki, M., Tomita, H., Akazawa, T., Sato, K., Kuze, B., Mizuta, K., Hara, A., Nagaoka, H., <u>Inou</u> e, N., and Ito, Y.	M2-like macrophage polarization in hig h lactic acid-producing head and neck c ancer	Cancer Science	108(6)	1128-11 34	2017

Akazawa, T., Ohashi, T., Wijewardana, V., Sugiura, K., Inoue, N.	Development of a vaccine based on bacteria-mimicking tumor cells coated with novel engineered TLR2 ligands	<i>Cancer Science</i>			2018 <i>In press</i>
John C Kingswood, Yukitoshi Takahashi et al.,	Tuberous Sclerosis registry to increase disease Awareness (TOSCA) – baseline data on 2093 patients.	<i>Orphanet Journal of Rare Diseases</i>	12(1)		2017
Yoshiaki Yamamoto, Yukitoshi Takahashi et al.,	Effect of CYP inducers/inhibitors on the topiramate concentration: Clinical value of therapeutic drug monitoring.	<i>Therapeutic Drug Monitoring</i>	39(1)		2017
Toshihiro Jogamoto, Yukitoshi Takahashi et al.,	Add-on stiripentol elevates serum valproate levels in patients with or without concomitant topiramate therapy.	<i>Epilepsy Research</i>	130	7-12	2017
Kimizu T, <u>Takahashi Y</u> et al.,	A case of early onset epileptic encephalopathy with de novo mutation in <i>SLC35A2</i> : Clinical features and treatment for epilepsy.	<i>Brain &amp; Development</i>	39	256-260	2017
Taku Omata, Yukitoshi Takahashi et al.,	Ovarian Teratoma Development after Anti-NMDA Receptor Encephalitis Treatment.	<i>Brain &amp; Development</i>	39	448-451	2017
Ryuki Matsuura, Yukitoshi Takahashi et al.,	Epilepsy with myoclonic atonic seizures and chronic cerebellar symptoms associated with antibodies against glutamate receptors N2B and D2 in serum and cerebrospinal fluid.	<i>Epileptic disorders</i>	19	94-98	2017
Shin-Seok Lee, D. Park, <u>Y. Takahashi</u> , J. Kang, Y. Yim, J. Kim, J. Lee, K. Lee, J. Lee, S. Lee	Anti-N-methyl-D-aspartate receptor antibodies are associated with fibromyalgia in patients with systemic lupus erythematosus: a case-control study	<i>Clinical and Experimental Rheumatology</i>	35	S54-S60	2017
Oikawa Y, Okubo Y, Numata-Uematsu Y, Aihara Y, Kitamura T, Takayanagi M, <u>Takahashi Y</u> , Kure S, Uematsu M,	Initial vasodilatation in a child with reversible cerebral vasoconstriction syndrome	<i>J Clin Neurosci.</i>	39	108-110	2017
高橋幸利	先生の知りたい最新医学がここにある：「小児てんかん」	健	45(10)	48-50	2017

四家達彦、 <u>高橋幸利</u> 、木治治療戦略の変更により ADL を改善し得た 村暢佑、今井克美、山下CDKL5 異常症による難治性てんかんの女 行雄、山本俊至、高橋孝児例. 雄	脳と発達	49	28-31	2017	
西口奈菜子、里龍晴、原 口康平、井上大嗣、渡邊 聖子、渡邊嘉章、 <u>高橋幸 利</u> 、森内浩幸	非ヘルペス性急性辺縁系脳炎の補助診断法としての脳血流シンチグラフィの有用性.	脳と発達	49	46-50	2017
束本和紀、 <u>高橋幸利</u> 、高 山留美子	Rufinamideが長期に奏功しているLennox-Gastaut症候群の3小児例.	脳と発達	49	54-56	2017
月田和人、下竹昭寛、中 谷光良、 <u>高橋幸利</u> 、池田 昭夫、高橋良輔	辺縁系脳炎で発症した神経梅毒の1例.	臨床神経学	57	37-40	2017
千葉悠平、勝瀬大海、斎 藤知之、須田顕、鎌田鮎 子、伊倉崇浩、阿部紀絵、 戸代原奈央、山口博行、 佐藤由佳、 <u>高橋幸利</u> 、平 安良雄	慢性自己免疫性脳炎を疑った際の検査、治療についての取り組みの紹介.	精神科治療学			印刷中
高橋幸利、西村成子、 高尾恵美子、笠井理沙、 榎田かある	非ヘルペス性急性辺縁系脳炎の分子病態.	Neuroinfection	22	56-61	2017
高橋幸利、松平敬史	ヒトパピローマウィルス（子宮頸がん）ワクチン後にみられる中枢神経関連症状.	日本内科学会雑誌	106 97	1591-15 97	2017