研究成果の刊行に関する一覧表

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髙梨潤一	感染症に関連した小児の急性脳症	福井次矢、他	今日の治療指針 2024	医学書院	東京	2024	1524-1525
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髙梨潤一	急性脳症・脳炎	大場洋、高梨潤一、森墾	小児神経の画像診断	Gakken	東京	2024	412-431
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山本俊至	全ゲノム増幅と網羅的ゲノム解析の進歩		着床前遺伝学的検査（PGT）の最前線と遺伝カウンセリング	メディカル ドゥ	大阪	2024	33-37
小坂仁、久保田雅也、望月葉子、他	希少神経難病・知的障害の成人移行支援の手引き-遺伝性白質疾患も含めて	厚生労働科学研究費補助金難治性疾患政策研究事業　遺伝性白質疾患・知的障害をきたす疾患の診断・治療・研究システム構築班	希少神経難病・知的障害の成人移行支援の手引き-遺伝性白質疾患も含めて	診断と治療社	東京	2023	88
髙梨潤一	興奮毒性型脳症（AESD)とMERS	宮嵜治	小児画像診断の勘ドコロNEO	メディカルレビュー社	東京	2023	48-50

髙梨潤一	インフルエンザ脳症（ANE,HSES）	宮嵜治	小児画像診断の勘ドコロNEO	メディカルレビュー社	東京	2023	45-47
髙梨潤一	急性脳炎・脳症	高橋幸利	小児の治療指針	診断と治療社	東京	2023	759-768
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Sakuma H, Takanashi J, Muramatsu K, Kondo H, Shiihara T, Suzuki M, Okanari K, Kasai M, Mitani O, Nakazawa T, Omata T, Shimoda K, Abe Y, Maegaki Y, Murayama K, Hayashi Y, Nagase H, Okumura A, Sakai Y, Tada H, Mizuguchi M	Severe pediatric acute encephalopathy syndromes related to SARS-CoV-2	Front Neurosci	17	1085082	2023
Sano K, Miya F, Kato M, Omata T, Takanashi J.	Neurochemistry evaluated by magnetic resonance spectroscopy in a patient with *FBXO28*-related developmental and epileptic encephalopathy.	Brain Dev	45	583-587	2023

Shimojima Yamamoto K, Tamura T, Okamoto N, Nishi E, Noguchi A, Takahashi I, Sawaishi Y, Shimizu M, Kanno H, Minakuchi Y, Toyoda A, Yamamoto T	Identification of small-sized intrachromosomal segments at the ends of INV-DUP-DEL patterns.	J Hum Genet	68(11)	751-757	2023
Shimomura R, Yanagishita T, Ishiguro K, Shichiji M, Sato T, Shimojima Yamamoto K, Nagata M, Ishihara Y, Miyashita Y, Ishigaki K, Nagata S, Asano Y, Yamamoto T	Rare mosaic variant of GJA1 in a patient with a neurodevelopmental disorder.	Hum Genome Var	11(1)	2	2023
Shimojima Yamamoto K, Yoshimura A, Yamamoto T	Biallelic KCTD3 nonsense variant derived from paternal uniparental isodisomy of chromosome 1 in a patient with developmental epileptic encephalopathy and distinctive features	Hum Genome Var	10(1)	22	2023
Shirai R, Cho M, Isogai M, Fukatsu S, Okabe M, Okawa M, Miyamoto Y, Torii T, Yamauchi J	FTD/ALS Type 7-Associated Thr104Asn Mutation of CHMP2B Blunts Neuronal Process Elongation, and Is Recovered by Knockdown of Arf4, the Golgi Stress Regulator.	Neurol. Int.	15(3)	980-993	2023
Shirakawa Y, Li H, Inoue Y, Izumi H, Kaga Y, Goto YI, Inoue K, Inagaki M.	Abnormality in GABAergic postsynaptic transmission associated with anxiety in Bronxwaltzer mice with an Srrm4 mutation.	*IBRO Neurosci Rep*	16	67-77	2023
Takanashi J, Uetani H	Neuroimaging in acute infection-triggered encephalopathy syndromes.	Front Neurisci	17	1235364	2023
Tamura T, Shimojima Yamamoto K, Imaizumi T, Yamamoto H, Miyamoto Y, Yagasaki H, Morioka I, Kanno H, Yamamoto T	Breakpoint analysis for cytogenetically balanced translocation revealed unexpected complex structural abnormalities and suggested the position effect for MEF2C	Am J Med Genet A	191(6)	1632-1638	2023
Tamura T, Yamamoto Shimojima K, Okamoto N, Yagasaki H, Morioka I, Kanno H, Minakuchi Y, Toyoda A, Yamamoto T	Long-read sequence analysis for clustered genomic copy number aberrations revealed architectures of intricately intertwined rearrangements	Am J Med Genet A	191(1)	112-119	2023
Tamura T, Yamamoto Shimojima K, Shiihara T, Sakazume S, Okamoto N, Yagasaki H, Morioka I, Kanno H, Yamamoto T	Interstitial microdeletions of 3q26.2q26.31 in two patients with neurodevelopmental delay and distinctive features	Am J Med Genet A	191(2)	400-407	2023

Torii K, Nishina S, Morikawa H, Mizobuchi K, Takayama M, Tachibana N, Kurata K, Hikoya A, Sato M, Nakano T, Fukami M, Azuma N, Hayashi T, Saitsu H, Hotta Y.	The Structural Abnormalities Are Deeply Involved in the Cause of RPGRIP1-Related Retinal Dystrophy in Japanese Patients.	Int J Mol Sci.	24(18)	13678	2023
Torii T, Yamauchi J	Molecular Pathogenic Mechanisms of Hypomyelinating Leukodystrophies (HLDs)	Neurol. Int.	15(3)	1155-1173	2023
Watanabe K, Kubota K, Nakashima M*, Saitsu H*	A case of infantile spasms with three possibly pathogenic de novo missense variants in NF1 and GABBR1.	Hum Genome Var.	10(1)	30	2023
Yamashita K, Kikuchi K, Hatai E Fujii, F, Chong P F, Sakai Y, Saitsu H, Inoue K, Togao O, Ishigami K.	Diagnostic MR imaging features of hypomyelination of early myelinating structures: A case report.	Neuroradiol J	Dec 25	19714009231224419	2023
Yiu RS, Ling TK, Ko CH, Poon SW, Poon GW, Wong FC, Law CY, Iwayama H, Lam CW.	Allan-Herndon-Dudley syndrome in Hong Kong: Implication for newborn screening.	Clin Chim Acta.	551	117621	2023
Yoh Y, Shiohama T, Uchida T, Ebata R, Kobayashi H, Okunushi K, Kato M, Watanabe K, Nakashima M, Saitsu H, Hamada H.	Case report: Progressive pulmonary artery hypertension in a case of megalencephaly-capillary malformation syndrome.	Front Genet.	14	1221745	2023
Yukiko Kuroda, Mayumi Matsufuji, Yumi Enomoto, Hitoshi Osaka, Jun-ichi Takanashi, et al.	A de novo U2AF2 heterozygous variant associated with hypomyelinating leukodystrophy.	Am J Med Genet A	191	2245-2248	2023
河野岳生, 近藤孝之, 井上治久	iPS細胞とAIによる神経変性疾患早期診断の展望	NEURO LOGICA	7	6-9	2023
黒澤健司	先天異常症候群	小児科臨床	76	193-196	2023
鈴木英文, 今村恵子, 井上治久	iPS細胞技術を用いた神経変性疾患研究	実験医学増刊	41 (12)	144-149	2023
宗実悠佳, 行武洋, 今村恵子, 井上治久	iPS細胞データと人工知能を用いた神経変性疾患研究	実験医学増刊	41 (15)	191-196	2023
村田静風, 今村恵子, 井上治久	疾患特異的iPS細胞によるALS創薬	BIO Clinica	38 (11)	3-9	2023
髙梨潤一	小児の白質病変をみたらどのように考えたらよいですか？	画像診断	43(1)	77-79	2023
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Arisa Ochiai, Sui Sawaguchi, Shiori Memezawa, Yoichi Seki, Takako Morimoto, Hiroaki Oizumi, Katsuya Ohbuchi, Masahiro Yamamoto, Kazushige Mizoguchi, Yuki Miyamoto, and Junji Yamauchi	Knockdown of Golgi stress-responsive caspase-2 ameliorates HLD17-associated AIMP2 mutant-mediated inhibition of oligodendroglial cell morphological differentiation.	Neurochem. Res.	47	2617-2631	2022
Aso K, Soutome T, Satoh M, Aoki T, Ogura H, Yamamoto T, Kanno H, Takahashi H	Association of autosomal-recessive-type distal renal tubular acidosis and Glanzmann thrombasthenia as a consequence of runs of homozygosity.	Clinical Case Reports	10	e06070	2022
Eto K, Machida O, Yanagishita T, Yamamoto Shimojima K, Chiba K, Aihara Y, Nagata M, Ishihara Y, Miyashita Y, Asano Y, Nagata S, Yamamoto T	Novel BCL11B truncation variant in a patient with developmental delay, distinctive features, epilepsy, and early craniosynostosis.	Hum Genome Var	9	43	2022
Fujita Yuji, Imataka Gyoji, Sakuma Hiroshi, Takanashi Jun-ichi, Yoshihara Shigemi	Multiple encephalopathy syndrome: a case of a novel radiological subtype of acute encephalopathy in childhood	Eur Rev Med Pharmacol Sci	26	5729-5735	2022
Fukumura S, Hiraide T, Yamamoto A, Tsuchida K, Aoto K, Nakashima M, Saitsu H.	A novel de novo *TMEM63A* variant in a patient with severe hypomyelination and global developmental delay.	Brain Dev.	44(2)	178-183	2022
Furukawa S, Miyamoto S, Fukumura S, Kubota K, Taga T, Nakashima M, Saitsu H.	Two novel heterozygous variants in *ATP1A3* cause movement disorders.	Hum Genome Var.	9(1)	7	2022
Hashiguchi M, Monden Y, Nozaki Y, Watanabe K, Nakashima M, Saitsu H, Yamagata T, Osaka H.	A TUBB4A Met363Thr variant in pediatric hypomyelination without atrophy of the basal ganglia.	Hum Genome Var.	9(1	19	2022
Hayakawa M, Matsubara T, Mochizuki Y, Takeuchi C, Minamitani N, Imai M, Kosaki K, Arai T, Murayama S	An autopsied case report of spastic paraplegia with thin corpus callosum carrying a novel mutation in the SPG11 gene: widespread degeneration with eosinophilic inclusions.	BMC Neurology	22(2):2	1-9	2022

Hiraide T, Akita T, Uematsu K, Miyamoto S, Nakashima M, Sasaki M, Fukuda A, Kato M, Saitsu H.	A novel de novo *KCNB1* variant altering channel characteristics in a patient with periventricular heterotopia, abnormal corpus callosum, and mild seizure outcom	J Hum Genet.	68(1)	25-31	2022
Hiraide T, Masunaga Y, Honda A, Kato F, Fukuda T, Fukami M, Nakashima M, Saitsu H, Ogata T.	Retrotransposition disrupting *EBP* in a girl and her mother with X-linked dominant chondrodysplasia punctata.	J Hum Genet.	67(5)	303-306	2022
Hiraide T, Shimizu K, Miyamoto S, Aoto K, Nakashima M, Yamaguchi T, Koshio T, Ogata T, Saitsu H.	Genome sequencing and RNA sequencing of urinary cells reveal an intronic *FBN1* variant causing aberrant splicing.	J Hum Genet.	67(7)	387-392	2022
Hoshino Y, Kodaira M, Matsuno A, Kaneko T, Fukuyama T, Takano K, Yazaki M, Sekijima Y.	Reversible Leukoencephalopathy in a Man with Childhood-onset Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome	Intern Med.	61 (4)	553-557	2022
Hyodo Y, Akiyama T, Fukuyama T, Mimaki M, Watanabe K, Kumagai T, Kobayashi K	Simultaneous assay of urine sepiapterin and creatinine in patients with sepiapterin reductase deficiency	Clin Chim Acta	534	167-172	2022
Inoue Y, Machida O, Kita Y, Yamamoto T	Need for revision of the ACMG/AMP guidelines for interpretation of X-linked variants.	Intractable & Rare Diseases Research	11	120-4	2022
Iwayama H, Ishihara N, Kawahara K, Madokoro Y, Togawa Y, Muramatsu K, Murakami A, Kuru S, Kumagai T, Ohashi W, Nanya K, Hasegawa S, Katsuno M, Okumura A.	Early immunological responses to the mRNA SARS-CoV-2 vaccine in patients with neuromuscular disorders.	Front Immunol.	13	996134	2022
Kaneko S, Shimbo A, Irabu H, Yamamoto T, Shimizu M	Inverted-duplication-deletion of chromosome 10q identified in a patient with systemic lupus erythematosus.	Pediatr Int	65(1)	e15396	2022
Kawashima S, Yuno A, Sano S, Nakamura A, Ishiwata K, Kawasaki T, Hosomichi K, Nakabayashi K, Akutsu H, Saitsu H, Fukami M, Usui T, Ogata T, Kagami M.	Familial pseudohypoparathyroidism type 1B caused by an SVA retrotransposon insertion on the GNAS locus.	J Bone Miner Res.	37(10)	1850-1859	2022
Kojima K, Wada T, Shimbo H, Ikeda T, Jimbo EF, Saitsu H, Matsumoto N, Yamagata T.	The ATRX splicing variant c.21-1G>A is asymptomatic.	Hum Genome Var	9	1-4	2022

Komatsu K, Fukumura S, Minagawa K, Nakashima M, Saitsu H.	A new case of concurrent existence of PRRT2-associated paroxysmal movement disorders with c.649dup variant and 16p11.2 microdeletion syndrome.	Brain Dev.	44(7)	474-479	2022
Kondo T, Yada Y, Ikeuchi T, Inoue H	CDiP technology for reverse engineering of sporadic Alzheimer's disease.	Journal of Human Genetics.	68(3)	231-235	2022
Kumaki, T., Enomoto, Y., Aida, N., Goto, T., & Kurosawa, K.	Progression of cerebral and cerebellar atrophy in congenital contractures of limbs and face, hypotonia, and developmental delay.	Pediatrics International	64(1)	e14734.	2022
Ludwig L, Lareau C, EBao E, Liu N, Utsugisawa T, Tseng A, Myers S, Verboon J, Ulirsch J, Luo W, Muus C, Fiorini C, Olive M, Vockley C, Munschauer M, Hunter A, Ogura H, Yamamoto T, Inada H, Nakagawa S, Ohzono S, Subramanian V, Chiarle R, Glader B, Carr S, Aryee M, Kundaje A, Orkin S, Regev A, McCavit T, Kanno H, Sankaran V	A Congenital Anemia Reveals Distinct Targeting Mechanisms for Master Transcription Factor GATA1.	Blood	139	2534-46	2022
Machida O, Yamamoto Shimojima K, Shiihara T, Akamine S, Kira R, Hasegawa Y, Nishi E, Okamoto N, Nagata S, Yamamoto T	Interstitial deletions in the proximal regions of 6q: 12 original cases and a literature review.	Intractable & Rare Diseases Research	11	143-8	2022
Masunaga Y, Nishimura G, Takahashi K, Hishiyama T, Imamura M, Kashimada K, Kadoya M, Wada Y, Okamoto N, Oba D, Ohashi H, Ikeno M, Sakamoto Y, Fukami M, Saitsu H, Ogata T.	Clinical and molecular findings in three Japanese patients with N-acetylneuraminic acid synthetase-congenital disorder of glycosylation (NANS-CDG).	Sci Rep.	12(1)	17079	2022
Masunaga Y, Ohkubo Y, Nishimura G, Ueno T, Fujisawa Y, Fukami M, Saitsu H, Ogata T.	ACAN biallelic variants in a girl with severe idiopathic short stature.	J Hum Genet.	67(8)	481-486	2022
Mitsuishi Tsuyoshi, Miyata Kazunori, Ando Akiko, Sano Kentaro, Takanashi Jun-ichi, Hamada Hiromichi	Author reply to"Onycholysis associated with Kawasaki disease:A comment on characteristic nail lesions in Kawasaki disease:Case series and literature review"	J Dermatol	49	e293- e294	2022

Miyamoto S, Nakashima M, Fukumura S, Kumada S, Saitsu H.	An intronic *GNAO1* variant leading to in-frame insertion cause movement disorder controlled by deep brain stimulation.	Neurogenetics.	23(2)	129-135	2022
Muramatsu K, Muramatsu SI.	Adeno-associated virus vector-based gene therapies for pediatric diseases.	Pediatr Neonatol	64 Suppl 1	S3-S9	2022
Muramatsu M, Shimojima Yamamoto K, Pin Fee Chong P-F, Ryutaro Kira R, Nobuhiko Okamoto N, Yamamoto T	Genotype-phenotype correlation in six patients with interstitial deletions spanning 13q31.	No To Hattatsu	54	317-22	2022
Murase H, Zhu Y, Sakaida K, Mizuno H, Mori H, Iwayama H, Suzuki N, Nagai N, Okumura A.	Case report: Five patients with myocarditis after mRNA COVID-19 vaccination.	Front Pediatr.	10	977476	2022
Nemoto Koko, Sano Kentaro, Sato Satoko, Maeda Yasuhiro, Murayama Kei, Takanashi Jun-ichi	A child with mitochondrial DNA deletion presenting diabetes mellitus as an initial symptom	Radiol Case Rep	17	2915-2918	2022
Okamoto Go, Furuya Emari, Terada Kanae, Yasukawa Kumi, Takanashi Jun-ichi, Kobayashi Emiko	Fosphenytoin dosing regimen including optimal timing for the measurement of serum phenytoin concentration in pediatric patients	Brain Dev	44	725-731	2022
Okuda T, Moroto M, Yamamoto T	Non-invasive prenatal testing suggesting an abnormality in chromosome 15 confirmed to be a case of Prader-Willi syndrome caused by trisomy rescue in the neonatal period	J Obstet Gynaecol Res	48(8)	2214-2218	2022
Ozaki H, Suga H, Sakakibara M, Soen M, Miyake N, Miwata T, Taga S, Nagai T, Kano M, Mitsumoto K, Miyata T, Kobayashi T, Sugiyama M, Onoue T, Takagi H, Hagiwara D, Iwama S, Banno R, Iguchi G, Takahashi Y, Muguruma K, Inoue H & Arima H	Differentiation of human induced pluripotent stem cells into hypothalamic vasopressin neurons with minimal exogenous signals and partial conversion to the naive state	Scientific Reports	12	17381	2022

Sakamoto M, Iwama K, Sasaki M, Ishiyama A, Komaki H, Saito T, Takeshita E, Shimizu-Motohashi Y, Haginoya K, Kobayashi T, Goto T, Tsuyusaki Y, Iai M, Kurosawa K, Osaka H, Tohyama J, Kobayashi Y, Okamoto N, Suzuki Y, Kumada S, Inoue K, Mashimo H, Arisaka A, Kuki I, Saijo H, Yokochi K, Kato M, Inaba Y, Gomi Y, Saitoh S, Shirai K, Morimoto M, Izumi Y, Watanabe Y, Nagamitsu SI, Sakai Y, Fukumura S, Muramatsu K, Ogata T, Yamada K, Ishigaki K, Hirasawa K, Shimoda K, Akasaka M, Kohashi K, Sakakibara T, Ikuno M, Sugino N, Yonekawa T, Gürsoy S, Cinleti T, Kim CA, Teik KW, Yan CM, Haniffa M, Ohba C, Ito S, Saitsu H, Saida K, Tsuchida N, Uchiyama Y, Koshimizu E, Fujita A, Hamanaka K, Misawa K, Miyatake S, Mizuguchi T, Miyake N, Matsumoto N.	Genetic and clinical landscape of childhood cerebellar hypoplasia and atrophy.	Genet Med.	22	S1098-3600	2022
Sasaki Yusuke, Fujimori Makoto, Hirose Shoko, Hamada Hiromichi, Takanashi Jun-Ichi	A 11-Year-old Male with Fever,Abdominal Pain and Progressive Renal Dysfunction	Pediatr Infect Dis J	41(11)	938- 940	2022
Sakata Y, Sano K, Aoki S, Saitsu H, Takanashi J I.	Neurochemistry evaluated by MR spectroscopy in a patient with *SPTAN1*-related developmental and epileptic encephalopathy.	Brain Dev.	44(6)	415-420	2022
Sakaue T, Obata Y, Fujishima Y, Kozawa J, Otsuki M, Yamamoto T, Maeda N, Nishizawa H, Shimomura I	A Japanese patient with a 2p25.3 terminal deletion presented with early-onset obesity, intellectual disability, and diabetes mellitus; a case report	J Diabetes Investig	13	391-396	2022
Satoshi Nishino, Yoko Fujiki, Takanari Sato, Yukino Kato, Remina Shirai, Hiroaki Oizumi, Masahiro Yamamoto, Katsuya Ohbuchi, Yuki Miyamoto, Kazushige Mizoguchi, and Junji Yamauchi	Hesperetin, a citrus flavonoid, ameliorates inflammatory cytokine-mediated inhibition of oligodendroglial cell morphological differentiation.	Neurol. Int.	14	471-487	2022

Shiori Memezawa, Takanari Sato, Arisa Ochiai, Miku Fukawa, Sui Sawaguchi, Kazunori Sango, Yuki Miyamoto, and Junji Yamauchi	The antiepileptic valproic acid ameliorates Charcot-Marie-Tooth 2W (CMT2W) disease-associated HARS1 mutation-induced inhibition of neuronal cell morphological differentiation through c-Jun N-terminal kinase.	Neurochem. Res.	47	2684-2702	2022
So Hayato, Ohashi Takashi, Yamagishi Sae, Mori Harushi, Takanashi Jun-ichi	Case of autoimmune glial fibrillary acidic protein astrocytopathy associated with Ebstein-Barr virus reactivation.	Clin Exp Neuroimmunol	13	106-110	2022
Sugiyama Y, Watanabe T, Tajika M, Matsuhashi T, Shimura M, Fushimi T, Ichimoto K, Matsunaga A, Ebihara T, Tsuruoka T, Akiyama T, Murayama K.	A Japanese single-center experience of the efficacy and safety of asfotase alfa in pediatric-onset hypophosphatasia.	Orphanet J Rare Dis	17(1)	78	2022
Sugitate R, Muramatsu K, Ogata T, Goto M, Hayashi S, Sawaura N, Kawada-Nagashima M, Matsui A, Yamagata T.	Recurrent pneumonia in three patients with MECP2 duplication syndrome with aspiration as the possible cause.	Brain Dev.	44(7)	486-491	2022
Sui Sawaguchi, Kenji Tago, Hiroaki Oizumi, Katsuya Ohbuchi, Masahiro Yamamoto, Kazushige Mizoguchi, Yuki Miyamoto, and Junji Yamauchi	Hypomyelinating leukodystrophy 7 (HLD7)-associated mutation of POLR3A is related to defective oligodendroglial cell differentiation, which is ameliorated by ibuprofen.	Neurol. Int.	14	11-33	2022
Sui Sawaguchi, Rimi Suzuki, Hiroaki Oizumi, Katsuya Ohbuchi, Masahiro Yamamoto, Kazushige Mizoguchi, Yuki Miyamoto, and Junji Yamauchi	Hypomyelinating leukodystrophy 8 (HLD8)-associated mutation of POLR3B leads to defective oligodendroglial morphological differentiation whose effect Is reversed by ibuprofen.	Neurol. Int.	14	212-244	2022
Tabata K, Ishiyama A, Nakamura Y, Sasaki M, Inoue K, Goto YI.	A familial 2p14 microdeletion disrupting actin-related protein 2 and Ras-related protein Rab-1A genes with intellectual disability and language impairment.	Eur J Med Genet.	65(3)	104446	2022
Takanari Sato, Remina Shirai, Mikinori Isogai, Masahiro Yamamoto, Yuki Miyamoto, and Junji Yamauchi	Hyaluronic acid and its receptor CD44, acting through TMEM2, inhibit morphological differentiation in oligodendroglial cells.	Biochem. Biophys. Res. Commun.	624	102-111	2022
Tomohiro Torii, Remina Shirai, Risa Kiminami., Satoshi Nishino, Takanari Sato, Sui Sawaguchi, Nana Fukushima, Yoichi Seki, Yuki Miyamoto, and Junji Yamauchi	Hypomyelinating leukodystrophy 10 (HLD10)-associated mutations of PYCR2 form large size mitochondria, inhibiting oligodendroglial cell morphological differentiation.	Neurol. Int.	14	1062-1080	2022

Tsuchiya Y, Kobayashi H, Kanno H, Yamamoto T	Beta-tricalcium phosphate as a possible adjuvant in γδ Tcell-based immune therapy for human disorders.	Tokyo Women's Medical University Journal	6	101-7	2022
Tsukida K, Muramatsu SI, Osaka H, Yamagata T and Muramatsu K.	WDR45 variants cause ferrous iron loss due to impaired ferritinophagy associated with NCOA4 and WIPI4 reduction.	Brain Commun.	4(6)	fcac304	2022
Ueda R, Okada T, Kita Y, Ukezono M, Takada M, Ozawa Y, Inoue H, Shioda M, Kono Y, Kono C, Nakamura Y, Amemiya K, Ito A, Sugiura N, Matsuoka Y, Kaiga C, Shiraki Y, Kubota M, Ozawa H.	Quality of life of children with neurodevelopmental disorders and their parents during the COVID-19 pandemic: A one-year follow-up study	Sci Rep	12(1)	4298	2022
Yamamoto N, Okazaki S, Kuki I, Yamada N, Nagase S, Nukui M, Inoue T, Kawakita R, Yorifuji T, Hoshina T, Seto T, Yamamoto T, Kawawaki H	Possible critical region associated with late-onset spasms in 17p13.1-p13.2 microdeletion syndrome: a report of two new cases and review of the literature.	Epileptic Disorders	24	567-71	2022
Yamamoto Shimojima K, Utsugisawa T, Ogura H, Aoki T, Kawakami T, Ohga S, Ohara A, Ito E, Yamamoto T, Kanno H	Clinical and genetic diagnosis of thirteen Japanese patients with hereditary spherocytosis	Hum Genome Vari	9	1	2022
Yamane H, Seki M, Ikeda T, Matsumoto A, Furui S, Sato T, Muramatsu K, Tajima T, Yamagata T.	An Adolescent Patient with Sick Sinus Syndrome Complicated by Hypothyroidism Carrying an SCN5A Variant.	Int Heart J.	63(3)	627-632	2022
Yuki Miyamoto, Tomohiro Torii, Keiichi Homma, Hiroaki Oizumi, Katsuya Ohbuchi, Kazushige Mizoguchi, Shou Takashima, and Junji Yamauchi	The adaptor SH2B1 and the phosphatase PTP4A1 regulate the phosphorylation of cytohesin-2 in myelinating Schwann cells in mice.	Sci. Signal.	15	eabi5276	2022
Yukino Kato, Kenji Tago, Shoya Fukatsu, Miyu Okabe, Remina Shirai, Hiroaki Oizumi, Katsuya Ohbuchi, masahiro Yamamoto, Kazushige Mizoguchi, Yuki Miyamoto, and Junji Yamauchi	CRISPR/CasRx-mediated RNA knockdown reveals that ACE2 is involved in the regulation of oligodendroglial cell morphological differentiation.	Non-coding RNA	8	42	2022
Watanabe K, Nakashima M, Wakatsuki R, Bunai T, Ouchi Y, Nakamura T, Miyajima H, Saitsu H.	Cognitive Impairment in a Complex Family With AAGGG and ACAGG Repeat Expansions in RFC1 Detected by ExpansionHunter Denovo.	Neurol Genet.	8(3)	e682	2022

河野岳生，近藤孝之，井上治久	iPS細胞とAIによる神経変性疾患早期診断の展望	NEURO LOGICA	7	6 - 9	2022
望月葉子	特集　脳神経内科医に求められる移行期医療　移行医療の現状と課題－脳神経内科の立場から	Brain Nerve	74(6)	741-746	2022
望月葉子	特集　神経治療における小児ー成人移行医療　重症心身障害児（者）の移行医療	神経治療学	39(2)	78-83	2022
望月葉子、尾方克久、熊田聡子、鈴木保宏、一ノ瀬英史、崎山快夫、齊藤利雄、望月秀樹、日本神経学会小児－成人移行医療対策特別委員会	小児期発症神経系疾患を対象とする小児－成人移行医療への取り組み：小児診療科と成人診療科との連携推進	臨床神経	63(2)	67-72	2022
西下直希，近藤孝之，井上治久	自動細胞製造技術委とiPS細胞の産業化	生物工学	100(5)	252	2022
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