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
小坂　仁、井上　健	大脳白質変性症		「小児疾患診療のための病態生理3　改訂第6版」小児内科2022年54巻増刊号	東京医学社		2022	p348-352
髙梨潤一	序文、CQ1、画像診断、AESD、MERS	日本小児神経学会	小児急性脳症診療ガイドライン2023	診断と治療社	東京	2023	1-125
髙梨潤一	頭部画像検査	前垣義弘	小児急性脳炎・脳症のとらえ方と治療戦略	中山書店	東京	2022	43-49
髙梨潤一	けいれん重積型(二相性)急性脳症(AESD)	前垣義弘	小児急性脳炎・脳症のとらえ方と治療戦略	中山書店	東京	2022	124-133
髙梨潤一	画像によるてんかんの病因・鑑別診断	高橋幸利	ペランパネルによるてんかん治療のストラテジー　第2版	先端医学社	東京	2022	45-51
山本俊至	小児科領域におけるゲノム医療による診断率は40%程度が限界か？	金子一成(監修)	小児科診療Controversy	中外医学社	東京	2022	34-9
山本俊至	神経発達症における遺伝学的検査は必要か？	金子一成(監修)	小児科診療Controversy	中外医学社	東京	2022	76-80

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
Akahoshi K, Nakagawa E, Goto YI, Inoue K.	Duplication within two regions distal to MECP2: clinical similarity with MECP2 duplication syndrome.	BMC Med Genomics.	16(1)	43	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			In press
Fukahori K, Yamoto K, Saitsu H, Ogata T, Nagasaki K.	*PORCN*-related microphthalmia with limb anomalies: Case report and literature review.	Am J Med Genet A.	191(2)	636-639	2023
Hiraide T, Shimizu K, Okumura Y, Miyamoto S, Nakashima M, Ogata T, Saitsu H.	A deep intronic *TCTN2* variant activating a cryptic exon predicted by SpliceRover in a patient with Joubert syndrome.	J Hum Genet.	In press	In press	2023 Mar 10. Online publication
Iwayama H, Kawahara K, Takagi M, Numoto S, Azuma Y, Kurahashi H, Yasue Y, Kawajiri H, Yanase A, Ito T, Kimura S, Kumagai T, Okumura A.	Long-term efficacy of nusinersen and its evaluation in adolescent and adult patients with spinal muscular atrophy types 1 and 2.	Brain Dev.	45	110-116	2023
Kanbara Y, Takeuchi C, Mochizuki Y, Osako M, Sasaki M, Hidehiko M	Medical needs of adults with Down syndrome in a regional medical and rehabilitation center in Japan	J of Nippon Medical School	in press		2023
Kato K, Kuroda T, Yamadera-Egawa R, Ezoe K, Aoyama N, Usami A, Miki T, Yamamoto T, Takeshita T	Preimplantation Genetic Testing for Aneuploidy for Recurrent Pregnancy Loss and Recurrent Implantation Failure in Minimal Ovarian Stimulation Cycle for Women Aged 35-42 Years: Live Birth Rate, Developmental Follow-up of Children, and Embryo Ranking.	Reprod Sci	30	974-83	2023
Kodama Kazuo, Aoyama Hiromi, Murakami Yoshimi, Takanashi Jun-ichi, Koshimizu Eriko, Miyatake Satoko, Iwama Kazuhiro, Mizuguchi Takeshi, Matsumot	A case of early-infantile onset, rapidly progressive leukoencephalopathy with calcifications and cysts caused by biallelic SNORD118 variants	Radiol Case Rep	18(2023)	1217-1220	2023

Kuroda Y, Matsufuji M, Enomoto Y, Osaka Y, Takanashi J, Yamamoto T, Numata-Uematsu Y, Tabata K, Kurosawa K, Inoue K.	A *de novo U2AF2* heterozygous variant associated with hypomyelinating leukodystrophy.	American Journal of Medical Genetics Part A			2023［論文受理後］
Miyamoto S, Nakamura K, Kato M, Nakashima M, Saitsu H.	Identification of pathogenic deep intronic variant and exonic LINE-1 insertion in a patient with Meckel syndrome.	Ann Hum Genet.	In press	In press	2023 Mar 27. Online publication
Morikawa H, Nishina S, Torii K, Hosono K, Yokoi T, Shigeyasu C, Yamada M, Kosuga M, Fukami M, Saitsu H, Azu	A pediatric case of congenital stromal corneal dystrophy caused by the novel variant c.953del of the DCN gene.	Hum Genome Var.	24;10(1):9.	Online journal	2023
Murofushi Yuka, Sakuma Hiroshi, Tada Hiroko, Mizuguchi Masashi, Takanashi Jun-ichi	Changes in the treatment of pediatric acute encephalopathy in Japan between 2015 and 2021:A national questionnaire-based survey	Brain Dev	45	153-160	2023
Myojin shota, Michihata Nobuaki, Shoji Kensuke, Takanashi Jun-ichi, Matsui Hiroki, Fushimi Kiyohide, Miyairi Isao, Yasunaga Hideo	Prognostic factors among patients with Shiga toxin-producing Escherichia coli hemolytic uremic syndrome: A retrospective cohort study using a nationwide inpatient database in Japan	J Infect Chemother	in press		2023
Nakahara E, Shimojima Yamamoto K, OguraH, Aoki T, Utsugisawa T, Azuma K, Akagawa H, Watanabe K, Muraoka M, Nakamura F, Kamei M, Tatebayashi K, Shinozuka J, Yamane T, Hibino M, Katsura Y, Nakano-Akamatsu S, Kadowaki N, Maru Y, Ito E, Ohga S, Yagasaki H, Morioka I, Yamamoto T, Kanno H	Variant spectrum of PIEZO1 and KCNN4 in Japanese patients with dehydrated hereditary stomatocytosis.	Hum Genome Var	10	8	2023
Osako M, Yamaoka Y, Takeuchi C, Mochizuki Y, Fujiwara T	Health care transition for cerebral palsy with intellectual disabilities: A systematic review	Rev Neurol (Paris)	S0035-3787(23)00820-2.2023 Online ahead of prin		2023

Osako M, Yamaoka Y, Takeuchi C, Mochizuki Y, Fujiwara T	Benefits and challenges of pediatric-to-adult health care transition in childhood-onset neurological conditions	Neurology Clinical Practice	13(2)	e200139	2023
Saito Riho, Hayashi Yuka, Kimura Sho, Yasukawa Kumi, Murayama Kei, Takanashi Jun-ichi	Multimodal MR imaging in acute exacerbation of methylmalonic acidemia	Radiol Case Rep	18(3)	1010-1014	2023
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
Tamura T, Yamamoto Shimojima K, Shiihara T, Sakazume S, Okamoto N, Yagasaki H, Morioka I, Kanno H, Yamamoto T	nterstitial microdeletions of 3q26.2q26.31 in two patients with neurodevelopmental delay and distinctive features.	Am J Med Genet A	191	400-7	2023
Abe K, Ando K, Kato M, Saitsu H, Nakashima M, Aoki S, Kimura T. A	New Case With Cortical Malformation Caused by Biallelic Variants in *LAMC3*.	Neurol Genet.	9;8(3):e680	Online journal	2022
Akiyama T, Kuki I, Kim K, Yamamoto N, Yamada Y, Igarashi K, Ishihara T, Hatano Y, Kobayashi K	Folic acid inhibits 5-methyltetrahydrofolate transport across the blood-cerebrospinal fluid barrier: clinical biochemical data from two cases	JIMD Rep	63(6)	529-535	2022
Arisa Ochiai, Sui Sawaguchi, Shiori Memezawa, Yoichi Saeki, 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
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.	3;9(1):19	Online journal	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 outcome.	J Hum Genet.	68(1)	25-31	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		e15396	2022
Kawashima S, Yuno A, Sano S, Nakamura A, Ishiwata K, Kawasaki T, Hosomichi K, Nakabayashi K, Akustu 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
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, Shihara 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;12(1):17079.	Online journal	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
Muramatsu K, Muramatsu SI.	Adeno-associated virus vector-based gene therapies for pediatric diseases.	Pediatr Neonatol	S1875-9572(22)		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 Obster Gynecol Res	48	2214-8	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
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. Inflt.	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
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
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

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
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.	16;8(3):e682	Online journal	2022
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
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
菅三佳, 井上治久	iPS細胞研究の新知見からの発展：脳オルガノイドによるてんかんの病態研究	日本臨牀	80(12)	1905	2022
杉原 進, 竹内千仙, 沼部博直, 山本俊至, 今井祐之	日光過敏症から骨髄性プロトポルフィリン症と診断されたモザイク型18q21.2-q22.1欠失の1例.	脳と発達	54	352-5	2022
髙梨潤一	小児急性脳症の臨床・画像最新情報	日本小児放射線学会雑誌	38(1)	35-43	2022
和田敬仁	指定難病最前線　脳クレアチン欠乏症候群.	新薬と臨牀	71	272-275	2022
山本俊至	ゲノム医療.	小児科	63　(増刊号)	1499-1505	2022
山本俊至	出生前診断・着床前診断の現状と課題.	日本小児科学会雑誌	126	1459-64	2022
吉田　誠克	アレキサンダー病にみられる異常GFAP.	京府医大誌	131	131-139	2022