

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
Tamura T, Imajima T, Shimajima Y, Yamamoto K, Yamamoto T	Genomic Copy Number Analysis Using Droplet Digital PCR: A Simple Method with EvaGreen Single-Color Fluorescent Design		Cerebral Cortex Development	Springer Nature	London	2024	In press
小坂 仁、井上 健	先天性大脳白質形成不全症	加藤元博	最新ガイドライン準拠 小児科診断・治療指針 改訂第三版	中山書店	東京	2024	812-814
高梨潤一	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
高梨潤一	小児急性脳症	門脇孝、他	診療ガイドライン UP-TO-DATE.	メディカルレビュー社	東京	2024	1001-1004
高梨潤一	MRSの臨床応用	大場洋、高梨潤一、森壘	小児神経の画像診断	Gakken	東京	2024	146-159
高梨潤一	急性脳症・脳炎	大場洋、高梨潤一、森壘	小児神経の画像診断	Gakken	東京	2024	412-431
高梨潤一	髄鞘形成不全性白質ジストロフィー	大場洋、高梨潤一、森壘	小児神経の画像診断	Gakken	東京	2024	432-443
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
高梨潤一	興奮毒性型脳症 (AE) と MERS	宮寄治	小児画像診断の勘道コロNEO	メディカルレビュー社	東京	2023	48-50
高梨潤一	インフルエンザ脳症 (ANE, HSES)	宮寄治	小児画像診断の勘道コロNEO	メディカルレビュー社	東京	2023	45-47
高梨潤一	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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Aoki S, Watanabe K, Kato M, Konishi Y, Kubota K, Kobayashi E, Nakashima M*, Saitsu H*.	Two novel cases of biallelic <i>SMPD4</i> variants with brain structural abnormalities.	Neurogenetics.	25(1)	3-11	2024
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Hayashi Y, Sehara Y, Watanabe R, Ohba K, Takayanagi Y, Sakiyama Y, Muramatsu K, Mizukami H.	Therapeutic Strategy for Fabry Disease by Intravenous Administration of Adeno-Associated Virus 9 in a Symptomatic Mouse Model.	Hum Gene Ther	35	192-201	2024
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Kawakami R, Hiraide T, Watanabe K, Miyamoto S, Hirai A K, Komatsu K, Ishigaki H, Sakaguchi K, Maekawa M, Yamashita K, Fukuda T, Miyairi I, Ogata T, Saitsu H*.	RNA sequencing and target long-read sequencing reveal an intronic transposon insertion causing aberrant splicing.	J Hum Genet.	69(2)	91-99	2024
Masunaga Y, Ono H, Fujisawa Y, Taniguchi K, Saitsu H, Ogata T.	Sotos syndrome with marked overgrowth in three Japanese patients with heterozygous likely pathogenic <i>NSD1</i> variants: case reports with review of literature.	Endocr J.	71(1)	75-81	2024
Matsumoto, A., Kano, S., Kobayashi, N., Matsuki, M., Furukawa, R., Yamagishi, H., Yoshinari, H., Nakakata, W., Wakabayashi, H., Tsuda, H., et al.	Unfavorable switching of skewed X chromosome inactivation leads to Menkes disease in a female infant.	Sci Rep.	14(1)	440	2024

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Yamoto K, Kato F, Yamoto M, Fukumoto K, Shimizu K, Saitsu H, Ogata T.	<i>TBX5</i> pathogenic variant in a patient with congenital heart defect and tracheal stenosis.	Congenit Anom (Kyoto).	64(1)	23-27	2024
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黒澤健司	マイクロアレイ染色体検査の原理と臨床応用	新生児成育医学 会雑誌	36	2-4	2024
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Fukawa M, Shirai R, Torii T, Nakata K, Fukatsu S, Sato T, Homma K, Miyamoto Y, Yamauchi J	Extracellular HSPA5 is autocritically involved in the regulation of neuronal process elongation	Biochem. Biophys. Res. Commun.	664	50-58	2023
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Kato Y, Shirai R, Ohbuchi K, Oizumi H, Yamamoto M, Miyata W, Iguchi T, Mimaki Y, Miyamoto Y, Yamauchi J	Hesperetin Ameliorates Inhibition of Neuronal and Oligodendroglial Cell Differentiation Phenotypes Induced by Knockdown of Rab2b, an Autism Spectrum Disorder-Associated Gene Product	Neurol. Int.	15(1)	371-391	2023
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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)	179 (6)	85-598	2023
Saito R, Murofushi Y, Kimura S, Yasukawa K, Murayama K, Takanashi J.	Multimodal MR imaging in acute exacerbation of methylmalonic acidemia	Radiol Case Reports	18	1010-1014	2023
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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 Bronx waltzer mice with an Srrm4 mutation.	IBRO Neurosci Rep	16	67-77	2023
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Tamura T, Shimajima 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 Shimajima 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
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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
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Yukiko Kuroda, Mayumi Matsufuji, Yumi Enomoto, Hitoshi Osaka, Jun-ichi Takayashi, 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