

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
黒澤健司	遺伝学的検査	秋山千枝子、五十嵐隆、岡明、平岩幹夫	小児保健ガイドブック	診断と治療社	東京	2021	197-200
高梨潤一	MRIによる遺伝性白質疾患診断アプローチ.	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	画像で診る遺伝性白質疾患の診断の手引き.	診断と治療社	東京	2021	2-34
高梨潤一	Pelizaeus-Merzbacher病(PMD)、他15疾患	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	画像で診る遺伝性白質疾患の診断の手引き.	診断と治療社	東京	2021	
山本俊至	Prof. 山本のマイクロアレイ染色体検査入門	山本俊至	Prof. 山本のマイクロアレイ染色体検査入門	診断と治療社	東京	2021	120
山本俊至	生殖医療における染色体解析法 (PGT-Aなど)	関沢明彦ら	生殖医療遺伝カウンセリングマニュアル	中外医学社	東京	2021	23-27
山本俊至	Jacobsen症候群(JBS)	画像で診る遺伝性白質疾患診断の手引き	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 編集	診断と治療社	東京	2021	62-63
山本圭子, 山本俊至	マイクロアレイ染色体検査	小児疾患診療のための病態生理2改訂6版小児内科 53 (増刊号)		東京医学社	東京	2021	174-181

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Furukawa S, Miyamoto S, Fukumura S, Kubota K, Taga T, Nakashima M, Saitsu H.	Two novel heterozygous variants in <i>ATPIA3</i> cause movement disorders.	Hum Genome Var.	Feb 18 online publication	Online journal	2022
Hayakawa M, Matsubara T, Mochizuki Y, Takeuchi C, Minami 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 <i>SPG11</i> gene: widespread degeneration with eosinophilic inclusions.	BMC Neurology	22(2):2	1-9	2022
Hiraide T, Masunaga Y, Honda A, Kato F, Fukuda T, Fukami M, Nakashima M, Saitsu H, Ogata T.	Retrotransposition disrupting <i>EBP</i> in a girl and her mother with X-linked dominant chondrodysplasia punctata.	J Hum Genet.	In press	In press	2022 Jan 24. online publication
Hiraide T, Shimizu K, Miyamoto S, Aoto K, Nakashima M, Yamaguchi T, Kosho T, Ogata T, Saitsu H.	Genome sequencing and RNA sequencing of urinary cells reveal an intronic <i>FBNI</i> variant causing aberrant splicing.	J Hum Genet.	In press	In press	2022 Jan 24. online publication
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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	In press		
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Hiraide T, Tanaka T, Masunaga Y, Ohkubo Y, Nakashima M, Fukuda T, Ogata T, Saitsu H.	Global developmental delay, systemic dysmorphism and epilepsy in a patient with a <i>de novo</i> <i>U2AF2</i> variant.	J Hum Genet.	66(12)	1185-1187	2021
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Watanabe K, Nakashima M, Kumada S, Mashimo H, Enokizono M, Yamada K, Kato M, Saitsu H.	Identification of two novel de novo <i>TUBB</i> variants in cases with brain malformations: case reports and literature review.	J Hum Genet.	66(12)	1193-1197	2021	
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Yamamoto T	Genomic Aberrations Associated with the Pathophysiological Mechanisms of Neurodevelopmental Disorders	Cells	10	2317	2021	
Yamamoto-Shimojima K, Akagawa H, Yanagi K, Kamekura T, Okamoto N, Yamamoto T	Deep intronic deletion in intron 3 of <i>PLP1</i> associated with severe phenotype of Pelizaeus-Merzbacher disease	Hum Genome Var	8	14	2021	
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大迫美穂、竹内千 仙、望月葉子	小児期発症の神経系疾 患を有する患者の成人 診療科への移行—知的・ 運動障害を有する患者 への取り組み—	神経治療学	38巻2号	112-122	2021
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山本俊至	神経発達症とゲノム医 療	小児科	62	1545-1554	2021
山本俊至	小児科医が考える出生 前検査へのサポート体 制とは	周産期医学	51	765-768	2021
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和田敬仁	指定難病最前線(Volume 129) 脳クレアチン欠 乏症候群	新薬と臨牀	71	272-275	2021
和田敬仁	【小児遺伝子疾患事 典】先天異常症候群 A TRX(関連疾患:ATR-X症 候群)	小児科診療	84	1423-1425	2021
和田敬仁	【エピゲノムで新たな 解明が進む「先天性疾 患」】(第3章)先天性疾 患 クロマチンリモデ リング因子異常症 ATR -X(X連鎖αサラセミ ア・知的障害)症候群	遺伝子医学MO OK	36	144-150	2021
柳下友映, 下島 圭子, 西 恵理 子, チョン ピン フィー, 山田 博 之, 岡本 伸彦, 永田智, 山本俊 至	日本人Potocki-Lupski 症候群7症例の臨床症状	脳と発達	53	456-461	2021