別添５

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
久保田雅也	① 一般診察　新生児 ② 不随意運動 ③ 小脳系 ④ トリビア	藤井克則	動画でわかる小児神経の診かた	羊土社	東京	2020	① pp14-23 ② pp47-73 ③ pp164-191 ④ pp202-218
髙梨潤一	尿素サイクル異常症	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	治療可能な遺伝性神経疾患 診断・治療の手引き	診断と治療社	東京	2020	58-62
山本俊至	遺伝子検査.		お医者さんオンライン	プレシジョン		2020	h00391
山本俊至	脊髄性筋萎縮症.		周産期遺伝カウンセリングマニュアル	中外医学社	東京	2020	146-148

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
Itai T, Miyatake S, Taguri M, Takanashi J, Saitsu H, Matsumoto N, et al.	Prenatal clinical manifestations in individuals with COL4A1/2 variants.	J Med Genet.		doi: 10.1136/jmedgenet-2020-106896.	In press
Yamamoto-Shimojima K, Akagawa H, Yanagi K, Kaname T, Okamoto N, Yamamoto T	Deep intronic deletion in intron 3 of PLP1 associated with severe phenotype of Pelizaeus-Merzbacher disease.	Hum Genome Var			In press
Yanagishita T, Eto K, Yamamoto-Shimojima K, Imaizumi T, Nagata S, Yamamoto T	A novel PAFAH1B1 splicing variant identified in a patient with classical lissencephaly.	Tokyo Women's Medical University Journal			In press

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			In press
Anzai R, Tsuji M, Yamashita S, Wada Y, Okamoto N, Saitsu H, Matsumoto N, Goto T.	Congenital disorders of glycosylation type IIb with MOGS mutations cause early infantile epileptic encephalopathy, dysmorphic features, and hepatic dysfunction.	Brain Dev.	43(3)	402-410	2021
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