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 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 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
和田敬仁	SLCトランスポーター異常症	厚生労働省難治性疾患等政策研究事業 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	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	63-65
和田敬仁	脳クレアチン欠乏症候群	厚生労働省難治性疾患等政策研究事業 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	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	107-109
Inoue K.	Pelizaeus-Merzbacher Disease: Molecular and Cellular Pathologies and Associated Phenotypes.	Sango K., Yamauchi J., Ogata T., Suzuki K.	Myelin. Advances in Experimental Medicine and Biology, vol 1190.	Springer	Singapore	2019	201-216.

Kubota M	Cockayne Syndrome: Clinical Aspects	Nishigori C., Sugasawa K.	DNA Repair Disorders	Springer	Singapore	2019	115-132
小坂　仁	有機酸代謝異常症	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	治療可能な遺伝性神経疾患	診断と治療社	東京	2019	42-45
小坂　仁	ミトコンドリア病	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	治療可能な遺伝性神経疾患	診断と治療社	東京	2019	55-57
黒澤健司	大頭症	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	治療可能な遺伝性神経疾患	診断と治療社	東京	2019	14-15
佐々木征行	付随運動	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	治療可能な遺伝性神経疾患	診断と治療社	東京	2019	16-19
髙梨潤一	尿素サイクル異常症	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	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	2019	58-62

松井　大	リボフラビン反応(1)ミトコンドリア呼吸鎖複合体1欠乏症（ACAD9欠損症）	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	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	2019	79-80
村松一洋	ライゾーム病，ペルオキシソーム病	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	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	2019	51-54
村松一洋	リボフラビン反応(1)Brown-Vialetto-Van Lsere症候群，Fazio-Londe病	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	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	2019	77-78
柳下友映，山本俊至	チアミン(ビタミンB_1)代謝異常症候群(2)ビオチン反応性大脳基底核病	厚生労働省難治性疾患等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	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	2019	75-76
山本俊至	リボフラミン反応(3)モリブデン補助因子欠損症A型.	厚生労働省難治性疾患等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	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	2019	81-83
今泉太一，山本俊至	ウリジン反応性てんかん性脳症	厚生労働省難治性疾患等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	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	2019	114-115

山本俊至	染色体微細構造異常と小児神経疾患	中村公俊、佐村修	【遺伝子医学MOOK別冊　最新遺伝医学研究と遺伝カウンセリング（シリーズ４）】　最新小児・周産期遺伝医学研究と遺伝カウンセリング	メディカルドゥ	大阪	2019	
山本俊至〔監修〕		山本俊至	症例でわかる小児神経疾患の遺伝学的アプローチ	診断と治療社	東京	2019	
山本俊至	11p13欠失症候群（WAGR症候群）		内分泌症候群（3版）IV-その他の内分泌疾患を含めて-	㈱日本臨床社	大阪	2019	
吉田誠克	リボフラビン反応　コエンザイムQ10欠損症	厚生労働省難治性疾患等政策研究事業　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	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	2019	84-86
和田敬仁	遺伝学的検査の手続き	厚生労働省難治性疾患等政策研究事業　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	小児神経疾患の遺伝学的アプローチ	診断と治療社	東京	2019	45-53
和田敬仁	知的障害を呈した１５才男児	厚生労働省難治性疾患等政策研究事業　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	小児神経疾患の遺伝学的アプローチ	診断と治療社	東京	2019	130-132
和田敬仁	X連鎖知的障害症候群の兄弟例	厚生労働省難治性疾患等政策研究事業　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	小児神経疾患の遺伝学的アプローチ	診断と治療社	東京	2019	133-135

和田敬仁	頭痛、視力障害を大した１５才女児	厚生労働省難治性疾患等政策研究事業 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	小児神経疾患の遺伝学的アプローチ	診断と治療社	東京	2019	141-142
井上　健	「メンデルの法則」「遺伝率」「集団遺伝」	一般社団法人日本人類遺伝学会	「コアカリ準拠　臨床遺伝学テキストノートーゲノム医療に必要な考え方を身につけるー」	診断と治療社	東京	2018	20-24
髙梨潤一	主に小児にみられる拡散強調画像高信号の鑑別	青木茂樹、大場洋	頭部の鑑別診断のポイント	秀潤社	東京	2018	48-52

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
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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
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Hiraide T, Fukumura S, Yamamoto A, Nakashima M, Saitsu H.	Familial periodic paralysis associated with a rare KCNJ5 variant that supposed to have incomplete penetrance.	Brain Dev.	43(3)	470-474	2021
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Nakashima M, Kato M, Matsukura M, Kira R, Ngu LH, Lichtenbelt KD, van Gassen KLI, Mitsuhashi S, Saitsu H, Matsumoto N.	De novo variants in CUL3 are associated with global developmental delays with or without infantile spasms.	J Hum Genet.	65(9)	727-734	2020
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Wakabayashi K, Osaka H, Kojima K, Imaizumi T, Yamamoto T, Yamagata T	MCT8 deficiency in a patient with a novel frameshift variant in the SLC16A2 gene.	Hum Genome Var	8	10	2020
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松山友美, 吉田誠克, 他	繰り返す意識消失と転倒を契機に急激に増悪する歩行障害を認めたAlexander病の1例.	臨床神経	60	137-141	2020
村松みゆき, 白井謙太朗, 今泉太一, 柳下友映, 山本圭子, 山本俊至.	Duchenne型筋ジストロフィー患者の母親で認められたモザイク変異と遺伝カウンセリング	脳と発達	52	41-44	2020
吉田誠克	アレキサンダー病の臨床的特徴と診断基準.	臨床神経	60	581-588	2020
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山本俊至	進行性白質脳症	日本医師会雑誌 【指定難病ペディア2019】	148		2019
山本俊至	Williams症候群の遺伝学	小児科診療	82	895-900	2019
山本俊至, 山本圭子	マイクロアレイ染色体検査の実際	遺伝子医学	9(1)	122-127	2019
吉田誠克.	アレキサンダー病の臨床と病態.	京都府立医科大学雑誌	128	1-8	2019
和田敬仁	個別の指定難病　染色体・遺伝子関連　ATR-X症候群[指定難病180]	日本医師会雑誌 【指定難病ペディア2019】	148	S307-S308	2019
和田敬仁	指定難病最前線(Volume 84)　ATR-X(X連鎖αサラセミア・知的障がい)症候群	新薬と臨牀	68	672-676	2019
藤田瑞穂, 下山恭平, 大塚直哉, 前田泰宏, 林北見、才津浩智, 松本直通, 髙梨潤一.	先天性片麻痺を呈したCOL4A1関連症候群の父子例	脳と発達	50	424-428	2018
黒田友紀子, 黒澤健司	序論：シンポジウム2　遺伝学的検査に振り回されない小児神経診療：適応から結果解釈・説明まで	脳と発達	50	181-182.	2018
黒澤健司	希少難病における診断・治療の進歩	こども医療センター医学誌	47	76-78.	2018

前田憲多郎, 吉田誠克, 他	首下がりを主訴としたアレキサンダー病の1例.	臨床神経	58	198-201	2018
山本俊至	遺伝性腫瘍症候群とその対応	小児科診療 Up-to-Date	33	9-12	2018
Akaboshi K, Yamamoto T	Interstitial deletion within 7q31.1q31.3 in a woman with mild intellectual disability and schizophrenia	Neuropsychiatric Disease and Treatment	14	1773-1778	2018
Akizawa Y, Yamamoto T, Tamura K, Kanno T, Takahashi N, Ohki T, Omori T, Tokushige K, Yamamoto M, Saito K.	A novel MLH1 mutation in a Japanese family with Lynch syndrome associated with small bowel cancer	Hum Genome Var	5	13	2018
Akutsu Y, Shirai K, Takei A, Goto Y, Aoyama T, Watanabe A, Imamura M, Enokizono T, Oto T, Hori T, Suzuki K, Hayashi M, Masumoto K, Inoue K.	A patient with peripheral demyelinating neuropathy, central dysmyelinating leukodystrophy, Waardenburg syndrome, and severe hy	Am J Med Genet Part A	176(5)	1195-1199	2018
Babaya N, Noso S, Hiromine Y, Ito H, Taketomo Y, Yamamoto T, Kawabata Y, Ikegami H	Early-Onset Diabetes Mellitus in a Patient With a Chromosome 13q34qter Microdeletion Including IRS2	J Endocr Soc	2(10)	1207-1213	2018
Belal H, Nakashima M, Matsumoto H, Yokochi K, Taniguchi-Ikeda M, Aoto K, Amin MB, Maruyama A, Nagase H, Mizuguchi T, Miyatake S, Miyake N, Iijima K, Nonoyama S, Matsumoto N, Saitsu H.	De novo variants in RHOBTB2, an atypical Rho GTPase gene, cause epileptic encephalopathy.	Hum Mutat	39(8)	1070-1075.	2018
Calmels N, Botta E, Jia N, Fawcett H, Nardo T, Nakazawa Y, Lanzafame M, Moriwaki S, Sugita K, Kubota M, Obringer C, Spitz MA, Stefanini M, Laugel V, Orioli D, Ogi T, Lehmann AR.	Functional and clinical relevance of novel mutations in a large cohort of patients with Cockayne syndrome.	J Med Genet	55	329- 343	2018
Chong PF, Saitsu H, Sakai Y, Imagi T, Nakamura R, Matsukura M, Matsumoto N, Kira R.	Deletions of SCN2A and SCN3A genes in a patient with West syndrome and autistic spectrum disorder.	Seizure.	60	91-93	2018

Fassio A, Esposito A, Kato M, Saitsu H, Mei D, Marini C, Conti V, Nakashima M, Okamoto N, Olmez Turker A, Albuz B, Semerci Gündüz CN, Yanagihara K, Belmonte E, Maragliano L, Ramsey K, Balak C, Siniard A, Narayanan V; C4RCD Research Group, Ohba C, Shiina M, Ogata K, Matsumoto N, Benfenati F, Guerrini R.	De novo mutations of the ATP6V1A gene cause developmental encephalopathy with epilepsy.	Brain.	1;141(6)	1703-1718.	2018
Hamada N, Ogaya S, Nakashima M, Nishijo T, Sugawara Y, Iwamoto I, Ito H, Maki Y, Shirai K, Baba S, Maruyama K, Saitsu H, Kato M, Matsumoto N, Momiyama T, Nagata KI.	De novo PHACTR1 mutations in West syndrome and their pathophysiological effects.	Brain	1;141(11)	3098-3114.	2018
Hamanaka K, Miyatake S, Zerem A, Lev D, Blumkin L, Yokochi K, Fujita A, Imagawa E, Iwama K, Nakashima M, Mitsuhashi S, Mizuguchi T, Takata A, Miyake N, Saitsu H, van der Knaap MS, Lerman-Sagie T, Matsumoto N.	Expanding the phenotype of IBA57 mutations: related leukodystrophy can remain asymptomatic.	J Hum Genet	63(12)	1223-1229	2018
Hamilton EMC, van der Lei HDW, Vermeulen G, Gerver JAM, Lourenço CM, Naidu S, Mierzewska H, Gemke RJBJ, de Vet HCW, Uitdehaag BMJ, Lissenberg-Witte BI, VWM Research Group (Matsui M et al.), van der Knaap MS.	Natural History of Vanishing White Matter.	Ann Neurol	84	274-288	2018
Hayakawa I, Kubota M.	Ictal pouting: kabuki visage or chapeau de gendarme?	Pract Neurol	18(5)	410-412	2018
Imaizumi T, Kumakura A, Yamamoto-Shimojima K, Ondo Y, Yamamoto T	Identification of a rare homozygous SZT2 variant due to uniparental disomy in a patient with a neurodevelopmental disorder	Intractable & Rare Diseases Research	7(4)	245-250	2018
Ishiyama A, Muramatsu K, Uchino S, Sakai C, Matsushima Y, Makioka N, Ogata T, Suzuki E, Komaki H, Sasaki M, Mimaki M, Goto YI, Nishino I.	NDUFAF3 Variants that Disrupt Mitochondrial Complex I Assembly may associate with Cavitating Leukoencephalopathy.	Clin Genet	93	1103-6	2018

Kunii M, Doi H, Ishii Y, Ohba C, Tanaka K, Tada M, Fukai R, Hashiguchi S, Kishida H, Ueda N, Kudo Y, Kugimoto C, Nakano T, Udaka N, Miyatake S, Miyake N, Saitsu H, Ito Y, Takahashi K, Nakamura H, Tomita-Katsumoto A, Takeuchi H, Koyano S, Matsumoto N, Tanaka F.	Genetic analysis of adult leukoencephalopathy patients using a custom-designed gene panel.	Clin Genet.	94(2)	232-238	2018
Matsumoto, A., Tulyeu, J., Furukawa, R., Watanabe, C., Monden, Y., Nozaki, Y., Mori, M., Namekawa, M., Jimbo, E.F., Aihara, T., Yamagata, T., ○Osaka, H.	A case of severe Alexander disease with de novo c. 239T>C, p.(F80S), in GFAP.	Brain Dev.	40(7)	587-591	2018
Miyatake S, Schneeberger S, Koyama N, Yokochi K, Ohmura K, Shiina M, Mori H, Koshimizu E, Imagawa E, Uchiyama Y, Mitsuhashi S, Frith MC, Fujita A, Satoh M, Taguri M, Tomono Y, Takahashi K, Doi H, Takeuchi H, Nakashima M, Mizuguchi T, Takata A, Miyake N, Saitsu H, Tanaka F, Ogata K, Hennet T, Matsumoto N.	Biallelic COLGALT1 variants are associated with cerebral small vessel disease.	Ann Neurol.	84(6)	843-853	2018
Mizuguchi T, Nakashima M, Kato M, Okamoto N, Kurahashi H, Ekhilevitch N, Shiina M, Nishimura G, Shibata T, Matsuo M, Ikeda T, Ogata K, Tsuchida N, Mitsuhashi S, Miyatake S, Takata A, Miyake N, Hata K, Kaname T, Matsubara Y, Saitsu H, Matsumoto N.	Loss-of-function and gain-of-function mutations in PPP3CA cause two distinct disorders.	Hum Mol Genet.	15:27(8)	1421-1433.	2018
Nagai K, Maekawa T, Terashima H, Kubota M, Ishiguro A.	Severe anti-GAD antibody-associated encephalitis after stem cell transplantation.	Brain Dev.	41(3)	301-304	2018
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Nabbout R, Belousova E, Benedik MP, Carter T, Cottin V et al. ; TOSCA Consortium and TOSCA Investigators (Kubota M et al.)	Epilepsy in tuberous sclerosis complex: Findings from the TOSCA Study	Epilepsia Open	4	73-84.	2018

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Sato T, Kato M, Moriyama K, Haraguchi K, Saitsu H, Matsumoto N, Moriuchi H.	A case of tubulinopathy presenting with porencephaly caused by a novel missense mutation in the TUBA1A gene.	Brain Dev.	40(9)	819-823	2018
Sato, Y Shibasaki J, Aida N, Hiiragi K, Kimura Y, Akahira-Azuma M, Yumi Enomoto Y, Tsurusaki Y, Kuronsawa K.	Novel COL4A1 mutation in a fetus with early prenatal onset of schizencephaly.	Human Genome Variation	5	4	2018
Shimada S, Hirasawa K, Takeshita A, Nakatsukasa H, Yamamoto-Shimojima K, Imaizumi T, Nagata S, Yamamoto T.	Novel compound heterozygous EPG5 mutations consisted with a missense mutation and a microduplication in the exon 1 region identified in a Japanese patient with Vici syndrome.	Am J Med Genet	176(12)	2803-2807	2018
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Shioda N*, Yabuki Y, Yamaguchi K, Onozato M, Li Y, Kurosawa K, Tanabe H, Okamoto N, Era T, Sugiyama H, Wada T* and Fukunaga K.	Targeting G-quadruplex DNA as cognitive function therapy for ATR-X syndrome.	Nature Medicine	24	802-813	2018
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Tsuchida N, Hamada K, Shiina M, Kato M, Kobayashi Y, Tohyama J, Kimura K, Hoshino K, Ganesan V, Teik KW, Nakashima M, Mitsuhashi S, Mizuguchi T, Takata A, Miyake N, Saitsu H, Ogata K, Miyatake S, Matsumoto N.	GRIN2D variants in three cases of developmental and epileptic encephalopathy.	Clin Genet.	94(6)	538-547	2018
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Yamamoto T, Yamamoto-Shimojima K, Ueda Y, Imai K, Takahashi Y, Imagawa E, Miyake N, Matsumoto N	Independent occurrence of de novo HSPD1 and HIP1 variants in brothers with different neurological disorders - leukodystrophy and autism	Hum Genome Var	5	18	2018
Yamashita K, Seto T, Fukushima S, Fujita K, Hikita N, Yamamoto T, Shintaku H.	Evaluation of the relationship between the serum immunoglobulin G2 level and repeated infectious diseases in children.	Osaka City Med J	64	19-30	2018
Yanagishita T, Yamamoto-Shimojima K, Nakano S, Sasaki T, Shigematsu H, Imai K, Yamamoto T	Phenotypic features of 1q41q42 microdeletion including WDR26 and FBXO28 are clinically recognizable: The first case from Japan	Brain Dev	41(5)	452-455	2018
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