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
伊藤雅之	MECP2 Rett症候群	山岸敬幸、三牧正和、古庄知己	小児遺伝子疾患辞典	診断と治療社	東京	2021	1484-1486
伊藤雅之	Rett症候群	中村公俊	遺伝子医学	メディカルドウ	東京	2021	112-119
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.
青天目信	FOXG1欠損症（先天型Rett症候群).	厚労省「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	診断と治療社	東京	2021	56-57
青天目信	グルタル酸尿症1型(GA1)	厚労省「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	診断と治療社	東京	2021	78-79

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
Takeguchi R, Takahashi S, Akaba Y, Tanaka R, Nabatame S, Kurosawa K, Matsuishi T, Itoh M.	Early diagnosis of MECP2 duplication syndrome: insights from a nationwide survey in Japan.	J Neurol Sci	422	117321	2021
Saikusa T, Kawaguchi M, Tanioka T, Nabatame S, Takahasi S, Yuge K, Nagamitsu S, Takahashi T, Yamashita Y, Kobayashi Y, Hirayama C, Kakuma T, Matsuishi T, Itoh M	Meaningful word acquisitionis associated with walking ability over 10 years in Rett syndrome.	Brain Dev	42	705-712	2020

Takeguchi R, Takahashi S, Kuroda M, Tanaka R, Suzuki N, Tomonoh Y, Ihara Y, Sugiyama N, Itoh M.	MeCP2_e2 partially compensates for lack of MeCP2_e1: a male case of Rett syndrome.	Mol Genet Genomic Med	8(2)	e1088	2020
Wakabayashi T, Fukumura S, Takahashi S, Kurosawa K, Miyamoto S, Tsuchida K, Kato S, Tsugawa T, Sakai Y, Kawasaki Y.	Stereotyped upper limb movement in *MECP2* duplication syndrome.	Neurology	97:92-94	92-94	2021
Akaba Y, Takahashi S, Takeguchi R, Tanaka R, Nabatame S, Saitsu H, Matsumoto N	Phenotypic overlap between FOXG1 syndrome and PDH deficiency.	Clin Case Rep	9	1711-1715	2021
Ikeda S, Akamatsu C, Ijuin A, Nagashima A, Sasaki M, Mochizuki A, Nagase H, Enomoto Y, Kuroda Y, Kurosawa K, Ishikawa H.	Prenatal diagnosis of Fraser syndrome caused by novel variants of FREM2.	Hum Genome Var.	7	32	2020
Nishimura N, Kumaki T, Murakami H, Enomoto Y, Katsumata K, Toyoshima K, Kurosawa K.	Arthrogryposis multiplex congenita with polymicrogyria and infantile encephalopathy caused by a novel GRIN1 variant.	Hum Genome Var.	7	29	2020
Natsume J, Ishihara N, Azuma Y, Nakata T, Takeuchi T, Tanaka M, Sakaguchi Y, Okai Y, Ito Y, Yamamoto H, Ohno A, Kidokoro H, Hattori A, Nabatame S, Kato K.	Lenticular nuclei to thalamic ratio on PET is useful for diagnosis of GLUT1 deficiency syndrome.	Brain Dev	43(1)	69-77	2021
Kagitani-Shimono K, Kato H, Kuwayama R, Tominaga K, Nabatame S, Kishima H, Hatazawa J, Taniike M.	Clinical evaluation of neuroinflammation in child-onset focal epilepsy: a translocator protein PET study.	J Neuroinflammation	18(1)	8	2021
Ohashi I, Kuroda Y, Enomoto Y, Murakami H, Masuno M, Kurosawa K.	6p21.33 Deletion encompassing CSNK2B is associated with relative macrocephaly, facial dysmorphism, and mild intellectual disability.	Clinical Dysmorphology	30(3)	139-141.	2021

Tanigawa J, Nabatame S, Tominaga K, Nishimura Y, Maegaki Y, Kinosita T, Murakami Y, Ozono K	High-dose pyridoxine treatment for inherited glycosylphosphatidylinositol deficiency.	Brain Dev	43(6)	680-7	2021
Kimizu T, Ida S, Okamoto K, Awano H, Niba ETE, Wijaya YOS, Okazaki S, Shimomura H, Lee T, Tominaga K, Nabatame S, Saito T, Hamazaki T, Sakai N, Saito K, Shintaku H, Nozu K, Takeshima Y, Iijima K, Nishio H, Shinohara M.	Spinal Muscular Atrophy: Diagnosis, Incidence, and Newborn Screening in Japan.	Int J Neonatal Screen	3(45)	45	2021
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Yoshitomi S, Hamano SI, Hayashi M, Sakuma H, Hirose S, Ishii A, Honda R, Ikeda A, Imai K, Jin K, Kada A, Kakita A, Kato M, Kawai K, Kawakami T, Kobayashi K, Matsuishi T, Matsuo T, Nabatame S, Okamoto N, Ito S, Okumura A, Saito A, Shiraishi H, Shirozu H, Saito T, Sugano H, Takahashi Y, Yamamoto H, Fukuyama T, Kuki I, Inoue Y.	Current medico-psycho-social conditions of patients with West syndrome in Japan.	Epileptic Disord	23(4)	579-89	2021
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Satomura Y, Bessho K, Nawa N, Kondo H, Ito S, Togawa T, Yano M, Yamano Y, Inoue T, Fukui M, Onuma S, Fukuoka T, Yasuda K, Kimura T, Tachibana M, Kitaoka T, Nabatame S, Ozono K.	Novel gene mutations in three Japanese patients with ARC syndrome associated mild phenotypes: a case series.	J Med Case Rep	16(1)	60	2022

Kishimoto K, Nabatame S, Kagitani-Shimono K, Kato M, Tohyama J, Nakashima M, Matsumoto N, Ozono K.	Ketogenic diet for focal epilepsy with SPTAN1 encephalopathy.	Epileptic Disord	in press		2022