

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
後藤雄一	ミトコンドリア病	永田智	小児の治療指針	診断と治療社	東京	2018.4	857-859
三牧正和	ミトコンドリア遺伝	日本人類遺伝学会	臨床遺伝学ノート	診断と治療社	東京	2018.10	42-52
井川正道, 米田誠	ミトコンドリア病, 疾病別での診断・治療の現状と求める医薬品・医療機器・再生医療像.	株式会社技術情報協会	「希少疾患用医薬品の採算性ある事業化と適応拡大戦略」	株式会社技術情報協会	東京	2018	477-485
黒澤健司	多発性翼状片症候群	柳瀬敏彦	内分泌症候(第3版)IV 領域別症候群シリーズ4	日本臨床社	東京	2019.3	443-445
黒澤健司	CHARGE症候群	柳瀬敏彦	内分泌症候(第3版)IV 領域別症候群シリーズ4	日本臨床社	東京	2019.3	475-479

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Ishiyama A, Muramatsu K, Uchino S, Sakai C, Matsushima Y, Makioka N, Ogata T, Suzuki E, Komaki H, Sasaki M, <u>Mimaki M</u> , Goto Y I, Nishino I.	NDUFAF3 Variants that Disrupt Mitochondrial Complex I Assembly may associate with Cavitating Leukoencephalopathy.	Clin Gene t.	93(5)	1103-1106	2018
Ikeda T, <u>Osaka H</u> , Shimbo H, Tajika M, Yamazaki M, Ueda A, et al.	Mitochondrial DNA 3243A>T mutation in a patient with MELAS syndrome.	Hum Genome Var	5	25	2018
Kuwajima M, Goto M, Kurane K, Shimbo H, Omika N, Jimbo EF, et al.	MELAS syndrome with m.4450G>A mutation in mitochondrial tRNA(Met) gene.				
Matsuda Y, <u>Tanaka M</u> , Sawabe M, Mori S, Muramatsu M, Naka-Mirom M, Arai T	Relationship between pancreatic intraepithelial neoplasias, pancreatic ductal adenocarcinoma, and single nucleotide polymorphisms in autopsy aged elderly patients	Genes Chromosomes Cancer	57(1)	12-18	2018
Verechshagina N, Nikitichina N, Yamada Y, Harashima H, <u>Tanaka M</u> , Orishchenko K, Mazunin I	Future of human mitochondrial DNA editing technologies.	Mitochondrial DNA Part A	30(2)	214-221	2018
Ikawa M, Kimura H, Kitazaki Y, Sugimoto K, Matsunaga A, Hayashi K, Yamamura O, Tsubujikawa T, Hamano T, <u>Yoneda M</u> , Okazawa H, Nakamoto Y	Arterial spin labeling MRI imaging for the clinical detection of cerebellar hypoperfusion in patients with spinocerebellar degeneration.	J Neurol Sci	394	58-62	2018
井川正道, 米田誠	ミトコンドリア脳筋症のメカニズム	Medical Science Digest	44	559-562	2018
井川正道, 岡沢秀彦, 米田誠	酸化ストレスイメージング ミトコンドリア病, 神経変性疾患への応用.	Brain & Nerve	71	161-166	2019
Itoh M, Ide S, Iwasaki Y, Saito T, Narita K, Dai H, Yamakura S, Furue T, Kitayama H, Maeda K, Takahashi E, Matsui K, <u>Goto Y</u> , Takeda S, Arima M.	Arima Syndrome with specific variations of <i>CEP290</i> gene; clinical comparison with Joubert syndrome and Joubert syndrome-related diseases.	Brain Dev	40	259-267	2018

伊藤雅之	レット症候群の概要	難病と在宅ケア	24	30-34	2018
岩崎裕治	高度医療と療育.	日本重症心身障害学会誌	44	3-6	2019
Kuroda Y, Ohashi I, Naruto T, Ida K, Enomoto Y, Saito T, Nagai	Familial total anomalous pulmonary venous return with 15q11.2 (BP1-BP2)	J Hum Genet	63(11)	1185-1188	2018
Yokoi T, Saito T, Nagai JI, <u>Kurosawa K.</u>	17q21.32-q22 Deletion in a girl with osteogenesis imperfecta, tricho-dento-osseous syndrome, and intellectual disability.	Congenit Anom (Kyoto).	59	51-52	2019
Saikusa T, Hara M, Iwama K, Yuge K, Ohba C, Okada J, Hisano T, Yamashita Y, Okamoto N, Saitsu H, Matsumoto N, <u>Matsuishi T</u>	De novo HDAC8 mutation causes Rett-related disorder with distinctive facial features and multiple congenital anomalies	Brain Dev	40	406-409	2018
Yuge K, Iwama K, Yonee C, Matsufuji M, Sanjo N, Saikusa T, Yae Y, Yamashita Y, Mizuguchi T, Matsumoto N, <u>Matsuishi T</u>	A novel STXBPI mutation causes typical Rett syndrome in a Japanese girl	Brain Dev	40	493-497	2018
Kumada T, Imai K, Takahashi Y, <u>Nabatame S</u> , Oguni H.	Ketogenic diet using a Japanese ketogenic milk for patients with epilepsy: A multi-institutional study.	Brain Dev	40 (3)	188-95	2018
Tanigawa J, Kagitani-Shimono K, Matsuzaki J, Ogawa R, Hanaie R, Yamamoto T, Tominaga K, <u>Nabatame S</u> , Mohri I, Taniike M, Ozono K.	Atypical auditory language processing in adolescents with autism spectrum disorder.	Clin Neurophysiol	129(9)	2029-37	2018

Itoh M, Dai H, Horike SI, Gonzalez J, Kitami Y, Meguro-Horike M, Kuki I, Shimakawa S, Yoshinaga H, Ota Y, Okazaki T, Maegaki Y, <u>Nabatame S</u> , Okazaki S, Kawawaki H, Ueno N, Goto YI, Kato Y.	Biallelic KARS pathogenic variants cause an early-onset progressive leukodystrophy.	Brain	142(3)	560-573	2019
Harada K, Yamamoto M, Konishi Y, Koyano K, <u>Takahashi S</u> , Namba M, Kusaka T.	Hypoplastic hippocampus in atypical Rett syndrome with a novel FOXP1 mutation.	Brain Dev	40	49-52	2018
Shioda T, <u>Takahashi S</u> , Kaname T, Yamauchi T, Fukuoka T.	MECP2 mutation in a boy with severe apnea and sick sinus syndrome.	Brain Dev	40	714-718	2018
Suganuma E, <u>Oka A</u> , Sakata H, Adachi N, Asanuma S, Oguma E, Yamaguchi A, Furuichi M, Uejima Y, Sato S, Takano T, Kawano Y, Tanaka R, Arai T, Oh-Ishi T.	10-year follow-up of congenital cytomegalovirus infection complicated with severe neurological findings in infancy: a case report.	BMC Pediatr	18(1)	369	2018
Koyano S, Morioka I, <u>Oka A</u> , Moriuchi H, Asano K, Ito Y, Yoshikawa T, Yamada H, Suzutani T, Inoue N, Japanese Congenital Cytomegalovirus Study Group.	More than two years follow-up of infants with congenital cytomegalovirus infection in Japan.	Pediatr Int	60(1)	57-62	2018
Nakamura M, Kita S, Kikuchi R, Hirata Y, Shindo T, Shimizu N, Inuzuka R, <u>Oka A</u> , Kamibeppu K.	A Qualitative Assessment of Adolescent Girls' Perception of Living with Congenital Heart Disease: Focusing on Future Pregnancies and Childbirth.	J Pediatr Nurs	38	e12-e18	2018

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