

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
長尾雅悦	ウィルソン病	猿田享男、他	専門家による私の治療（2019-20年度版）	日本医事新報社	東京	2018	114-115
倉信奈緒 美、村山圭	症候と鑑別診断：筋力低下・筋痛．	日本小児栄養消化器肝臓学会編集	小児臨床栄養学 改訂第2版	診断と治療社	東京	2018	51-53
市本景子、 村山 圭	症候と鑑別診断：筋力低下・筋痛、意識障害	日本小児栄養消化器肝臓学会編集 診断と治療社	小児臨床栄養学 改訂第2版	診断と治療社	東京	2018	151-153
羽田 明	疾患の遺伝要因と予防医学	森 千里	予防医学の未来	医歯薬出版	東京都	2019	81-86

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
Nakagama Y, Hamanaka K, Mimaki M, Shintaku H, Miyatake S, Matsumoto N, Hirohata K, Inuzuka R, Oka A.	Leaky splicing variant in sepiapterin reductase deficiency: Are milder cases escaping diagnosis?	Neurol Genet.	5(2):	e319	2019
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Yuasa M, Hata I, Sugihara K, Isozaki Y, Ohshima Y, Hara K, Tajima G, Shigematsu Y	Evaluation of metabolic defects in fatty acid oxidation using peripheral blood mononuclear cells loaded with deuterium-labeled fatty acids	Disease Markers		Doi:10.1155/2019/2984747	2019
Hamada H, Hata A et al.	Efficacy of primary treatment with immunoglobulin plus ciclosporin for prevention of coronary artery abnormalities in patients with Kawasaki disease predicted to be at increased risk of non-response to intravenous immunoglobulin (KAICA): a randomised controlled, open-label, blinded-endpoints, phase 3 trial	Lancet	393(10176)	1128-1137	2019

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Fukao T	Defects in ketone body metabolism and pregnancy.	J Korean Soc Inher Metab Dis			In press
Wada Y., Kikuchi A., Arai-Ichinoi N., Sakamoto O., Takezawa Y., Iwasawa S., Niihori T., Nyuzuki H., Nakajima Y., Ogawa E., Ishige M., Hirai H., Sasai H., Fujiki R., Shirota M., Funayama R., Yamamoto M., Ito T., Ohara O., Nakayama K., Aoki Y., Koshiba S., Fukao T., Kure S.	Biallelic GALM pathogenic variants cause a novel type of galactosemia.	Genet Med.			In press
Alijanpour M, Sasai H, Abdelkreem E, Ago Y, Soleimani S, Moslemi L, Yamaguchi S, Rezapour M, Hakimi MT, Matsumoto H, Fukao T	Beta-Ketothiolase deficiency: A case with unusual presentation of non-ketotic hypoglycemic episodes due to coexistent probable secondary carnitine deficiency.	JIMD report			In press

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, Nabatame S, Okazaki S, Kawawaki H, Ueno N, Goto YI, Kato Y.	KARS pathogenic variants cause an early-onset progressive leukodystrophy.	Brain				In press
新宅治夫、保科隆男、濱崎考史	メンケス病	Clinical Neuroscience	37(3)	324-328	2019	
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Hoshina T, Nozaki S, Hamazaki T, Kudo S, Nakatani Y, Kodama H, Shintaku H, Watanabe Y.	Disulfiram enhanced delivery of orally administered copper into the central nervous system in Menkes disease mouse model.	J Inherit Metab Dis.	41(6)	1285-1291	2018	
Sohn YB, Ko AR, Seong MR, Lee S, Kim MR, Cho SY, Kim JS, Sakaguchi M, Nakazawa T, Kosuga M, Seo	The efficacy of intracerebroventricular idursulfase-beta enzyme replacement therapy in mucopolysaccharidosis II murine model: heparan sulfate in cerebrospinal fluid as	J Inherit Metab Dis.		[Epub ahead of print] doi: 10.1007/s10545-018-0221-0.	2018	

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Kobayashi H, Ariga M, Sato Y, Fujiwara M, Fukasawa N, Fukuda T, Takahashi H, Ikegami M ⁵ , Kosuga M, Okuyama	P-Tau and Subunit c Mitochondrial ATP Synthase Accumulation in the Central Nervous System of a Woman with Hurler-Scheie Syndrome Treated with Enzyme	JIMD Rep	41	101-107	2018
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Furujo M, Kubo T, Kinoshita M, Nagao M.	Diagnostic value of the MAT1A gene mutations in methionine adenosyltransferase I/III deficiency: Possible relevance to various neurological manifestations.	Neuropsychiatry (London)	8	1564-1570	2018
Iijima H, Iwano R, Tanaka Y, Muroya K, Fukuda T, Sugie H, Kurosawa K, Adachi M	Analysis of GBE1 mutations via protein expression studies in glycogen storage disease type IV: A report on a non-progressive form with a literature review.	Mol Genet Metab Rep	13;17	31-37	2018
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清水教一	先天代謝異常 Wilson 病	周産期医学	48	1357-1361	2018
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村山 圭	ミトコンドリア病の診断と治療の最前線	日本マス・スクリーニング学会誌	28 巻 1 号	109-119	2018
村山 圭	新生児ライソゾーム病スクリーニングのパイロットスタディ	調査研究ジャーナル	7 巻 2 号	179-179	2018
李知子, 山本和宏, 起塚庸, 山田健治, 小林弘典, 湯浅光織, 重松陽介, 原圭一, 但馬剛, 竹島泰弘	新生児スクリーニングで異常を認めず、横紋筋融解症を機にカルニチンパルミトイルトランスフェラーゼ2(CPT2)欠損症と診断された幼児例	日本マススクリーニング学会誌	28 (3)	253-260	2018