

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
伊藤康	グルコーストランスポーター1欠損症. 代謝異常症が原因で発症する脳症 ～代謝性脳症～		JaSMIn通信特別記事 No.42		東京	2020.06	
長尾雅悦	メープルシロップ尿症	水口 雅、他	今日の小児治療指針 第17版	医学書院	東京	2020	192-193
村山 圭,小坂 仁,三牧将和	ミトコンドリアと病気	村山 圭,小坂 仁,三牧将和	遺伝医学MOOK	メディカルドゥ	東京	2020	172-176
村山 圭	ミトコンドリア異常症	水口 雅, 市橋 光, 崎山弘, 伊藤 秀一	今日の小児診療指針	医学書院	東京	2020	194-195
笹井英雄	ミトコンドリアB酸化異常症	水口雅、市橋光、崎山弘、伊藤秀一	今日の小児治療指針	医学書院	東京	2020	196-197
大浦敏博、伊藤哲哉、中村公俊、他	特殊ミルク治療ガイドブック	日本小児医療保健協議会（四者協）、治療用ミルク安定供給委員会	特殊ミルク治療ガイドブック	診断と治療社	東京	2020年	全143ページ
野口篤子、矢野道広、高橋 勉	SLC7A7欠損症		日本臨床増刊号 原発性免疫不全症候群	日本臨床社	東京	2020	233-235
野口篤子	リジン尿性蛋白不耐症		特殊ミルクガイドブック	診断と治療社	東京	2020	33-35
清水教一	ウィルソン病（肝レンズ核変性症）	福井次矢ほか	今日の治療指針	医学書院	東京	2021年	792

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
Okuyama, T., Eto, Y., Sakai, N., Nakamura, K., Yamamoto, T., Yamaoka, M., Ikeda, T., So, S., Tanizawa, K., Sonoda, H., Sato, Y.	A Phase 2/3 Trial of Pabinafusp Alfa, IDS Fused with Anti-Human Transferrin Receptor Antibody, Targeting Neurodegeneration in MPS-II	Molecular Therapy	29(2)	671-679	2021
Shintaku H, Ohura T, Takayanagi M, Kure S, Owada M, Matsubara Y, et al.	Guide for diagnosis and treatment of hyperphenylalaninemia	Pediatrics International	63(1)	8-12	2021
Odagiri S, Kabata D, Tomita S, Kudo S, Sakaguchi T, Nakano N, et al.	Clinical and Genetic Characteristics of Patients with Mild Hyperphenylalaninemia Identified by Newborn Screening Program in Japan.	International Journal of Neonatal Screening	7(1)	17	2021
Shiraishi H, Yamada K, Egawa K, Ishige M, Ochi F, Watanabe A, Kawakami S, Kuzume K, Watanabe K, Sameshima K, Nakamagoe K, Tamaoka A, Asahina	Efficacy of bezafibrate for preventing myopathic attacks in patients with very long-chain acyl-CoA dehydrogenase deficiency.	Brain Dev	43(2)	214-219	2021
Mizuguchi M, Ichiyama T, Imataka G, Okumura A, Goto T, Sakuma H, Takanashi JI, Murayama K, Yamagata T, Yamanouchi H, Fukuda T, Maegaki Y.	Guidelines for the diagnosis and treatment of acute encephalopathy in childhood.	Brain Dev.	43	2-31	2021
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Hiraide T, Yamoto K, Masunaga Y, Asahina M, Endoh Y, Ohkubo Y, Matsubayashi, T, Tsurui S, Yamada H, Yanagi, K, Nakashima M, Hirano K, Sugimura H, Fukuda	Genetic and phenotypic analysis of 101 patients with intellectual disability using 3 whole-exome sequencing	Clin Genet.		doi: 10.1111/cge.13951	2021
Seo JH, Kosuga M, Hamazaki T, Shintaku H, Okuyama T.	Impact of intracerebroventricular enzyme replacement therapy in patients with neuronopathic mucopolysaccharidosis type II.	Mol Ther Methods Clin Dev.	21	67-75	2021
Hirashio S, Kagawa R, Tajima G, Masaki T	A classic variant of Fabry disease in a family with the M296I late - onset variant	CEN Case Reports	10 (1)	106-110	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
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Fatima A, Hoeber J, Schuster J, Koshimizu E, Maya-Gonzalez C, Keren B, Mignot C, Akram T, Ali Z, Miyatake S, Tanigawa J, Koike T, Kato M, Murakami Y,	Mono-allelic and bi-allelic variants in NCDN cause neurodevelopmental delay, intellectual disability, and epilepsy	Am J Hum Genet.	doi: 10.1016/j.ajhg.2021.02.015		2021
Salian S, Benkerroum H, Nguyen TTM, Nampoothiri S, Kinoshita T, Félix TM, Stewart F, Sisodiya SM, Murakami Y, Campeau PM.	PIGF deficiency causes a phenotype overlapping with DOORS syndrome.	Hum Genet.	doi: 10.1007/s00439-020-02251-2		2021

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Kido, J., Matsumoto, S., Ito, T., Hirose, S., Fukui, K., Kojima-Ishii, K., Mushimoto, Y., Yoshida, S., Ishige, M., Sakai, N., Nakamura, K.	Physical, cognitive, and social status of patients with urea cycle disorders in Japan	Molecular Genetics and Metabolism Reports	27	100724	2021
Katayama, D., Baba, H., Kuwabara, T., Kido, J., Mitsubuchi, H., Matsumoto, S., Nakamura, K.	SGLT2 inhibition alleviated hyperglycemia, glucose intolerance, and dumping syndrome-like symptoms in a patient with glycogen storage disease type Ia: a case report	Journal of Medical Case Reports	15(1)	10.1186/s13256-020-02658-5	2021
Nakamura K, Kawashima S, Tozawa H, Yamaoka M, Yamamoto T, Tanaka N, Yamamoto R, Okuyama T, Eto Y.	Pharmacokinetics and pharmacodynamics of JR-051, a biosimilar of agalsidase beta, in healthy adults and patients with Fabry disease: Phase I and II/III clinical studies.	Mol Genet Metab.	130(3)	215-224	2020
Naramura T, Tanaka K, Inoue T, Imamura H, Yoshimatsu H, Mitsubuchi H, Nakamura K, Iwai M.	New reference ranges of procalcitonin excluding respiratory failure in neonates.	Pediatrics International	62(10)	1151-1157	2020
Momosaki K, Kido J, Matsumoto S, Ozasa S, Nakamura K.	Adrenocorticotrophic hormone therapy improved spasms and sleep disturbance in smith-magenis syndrome: A case report.	Pediatric Reports	12(3)	72-76	2020
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Kohroggi K, Migita M, Anan T, Sugahara T, Yoshimoto K, Kanegane H, Nakamura K.	Successful Artery Embolization in a Patient with Autoimmune Lymphoproliferative Syndrome Associated with Splenic Rupture.	Journal of Clinical Immunology	40(5)	780-782	2020
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Ichimoto K, Fujisawa T, Shimura M, Fushimi T, Tajika M, Matsunaga A, Ogawa-Tominaga M, Akiyama N, Naruke Y, Horie H, Fukuda T, Sugie H, Inui A, Murayama K.	Two cases of a non-progressive hepatic form of glycogen storage disease type IV with atypical liver pathology.	Mol Genet Metab Rep.		doi: 10.1016/j.ymgmr.2020.100601.	2020
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Kido J, Nakamura K, Era T.	Role of induced pluripotent stem cells in lysosomal storage diseases.	Molecular and Cellular Neuroscience	108	103540	2020
Nakama M., Sasai H., Kubota M., Hasegawa Y., Fujiki R., Okuyama T., Ohara O., Fukao T.	Novel HADHB mutations in a patient with mitochondrial trifunctional protein deficiency.	Hum Genome.	7(10)	doi: 10.1038/s41439-020-0097-z	2020
Fuseya Y, Sakurai T, Miyahara JI, Sato K, Kaji S, Saito Y, Takahashi M, Nishino I, Fukuda T, Sugie H, Yamashita H.	Adult-onset Repeat Rhabdomyolysis with a Very Long-chain Acyl-CoA Dehydrogenase Deficiency Due to Compound Heterozygous ACADVL Mutations.	Intern Med.	59	2729-2732	2020.
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Takaki Y, Yamashita T, Kataoka N, Yokoyama S, Anan T, Nakamura K, Yoshimoto K, Hayashida S, Yamamoto H, Hibi T, Migita M.	Hepatoblastoma with multiple tumors in a school-aged child.	Clinical Case Reports	8(11)	2314-2315	2020
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濱崎考史	アミノ酸代謝異常症 (特集 新ガイドラインの理解を深める 新生児マススクリーニング)	小児科診療	84(2)	157-62	2021
濱崎考史	対象疾患の診療アミノ酸代謝異常症 (特集 みんなで役立てよう 新生児スクリーニング検査)	周産期医学	51(2)	181-4	2021
笹井英雄	ケトン体代謝異常症	小児科診療	84(2)	187-192	2021
笹井英雄	確定検査：遺伝子検査を中心に	小児科診療	84(2)	199-204	2021
小野田幸男、稲岡一考、竹内真、笹井英雄、深尾敏幸、藤木亮次、小原収、吉長正博	MRI検査により早期の拡張型心筋症と考えられた遅発型極長鎖アシル-CoA脱水素酵素欠損症の兄妹例	月刊心臓	53(2)	209-215	2021
児玉浩子、岡山和代、道堯浩二郎	患者アンケートから見た移行期医療の課題	肝胆膵	82 (3)	405-412	2021
但馬剛, 佐倉文祥, 原圭一	脂肪酸代謝異常症	小児科診療	84 (2)	181-186	2021
但馬剛, 香川礼子	先天性門脈-体循環シャントによる高ガラクトース血症	小児科診療	84 (2)	279-283	2021

但馬剛	海外の新生児マススクリーニングと日本における対応	周産期医学	51 (2)	275-279	2021
清水教一	Wilson病	小児科	61	1410-1414	2020
清水教一	肝胆疾患Wilson病	小児科臨床	73	767-771	2020
羽田 明	脊髄性筋萎縮症新生児スクリーニングの実施とその課題	第9回九州新生児スクリーニング研究会報告集		20-25	2020
山口清次, 但馬剛	新生児マススクリーニングの全国標準化	公衆衛生情報	50 (4)	10-12	2020
但馬剛, 原圭一, 宇都宮朱里, 香川礼子, 佐倉文祥	有機酸代謝異常症	小児科	61 (10)	1359-1364	2020
児玉浩子	セルロプラスミン	内科	125	815-816	2020
岡山和代, 児玉浩子, 青木継稔, 徐朱玟, 奥山虎之, 池田修一, 玉井浩, 藤澤智雄, 松浦晃洋, 清水教一, 林久男, 原田大, 道堯浩二郎	移行期医療に関するWilson病患者のアンケート調査結果	肝臓	61	700-714	2020
中務秀嗣, 伊藤康, 小國弘量, 衛藤薫, 平澤恭子, 中村幸恵, 小坂仁, 永田智	グルコーストランスポーター1 (GLUT1) 欠損症の遺伝子治療に対する意識調査	脳と発達	52	S324	2020
田中藤樹	日常診療で先天代謝異常症を見逃さないために 5. シトリン欠損症.	小児科	61	1387-1393	2020
田中藤樹, 長尾雅悦, 小杉山清隆	札幌市における先天代謝異常症のハイリスクスクリーニングによる診断と経過観察	札幌市医学会雑誌	45	115-116	2020
村山 圭	ミトコンドリア病	特殊ミルク治療ガイドブック	24	71-73	2020
村山 圭	先天代謝異常症	今日の診断指針	第8版総	1918-1920	2020
村山 圭	カルニチン回路異常症	新臨床内科学	第10版第6章	784-786	2020
杉山 洋平 村山 圭	新生児期に緊急対応が必要な先天代謝異常症	新生児内分泌ハンドブック	新版7代謝	213-225	2020

伊藤康	【診断・治療可能な遺伝性疾患を見逃さないために】 グルコーストランスポーター1 (GLUT1) 欠損症	小児科臨床	73	649-653	2020
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