

研究成果の刊行に関する一覧表

【令和2年度】

(ガイドライン)

ニーマンピック病C型(NPC)診療ガイドライン2021, 診療ガイドライン作成委員会, 診断と治療社
ムコ多糖症(MPS)IVA診療ガイドライン2021, 診療ガイドライン作成委員会, 診断と治療社

書籍・雑誌 (和文)

- 衛藤義勝, 特集 ライソゾーム病の早期発見早期治療を目指して—新生児スクリーニングの重要性予防医学, 9号, 4-7, 2020
- 松永綾子, 村山圭, 脾腫の急激な進行により診断に至ったゴーシェ病, ゴーシェ病症例集2020, クリエイトアール, 東京, 2020, 62-66
- 村山圭, パーキンソニズムを合併した小児期発症ゴーシェ病 ゴーシェ病症例集2020, クリエイトアール, 74-77
- 志村優, 村山圭, 新生児スクリーニングを契機に診断されたゴーシェ病I型の女児例 ゴーシェ病症例集2020, クリエイトアール, 2020, 42-44
- 坪井一哉, 酵素補充療法から基質合成抑制療法に切り替えたゴーシェ病の1例, p. 18-p. 22
- 今中常雄, ペルオキシソーム病, 第17版 今日の小児治療指針, 医学書院, 2020, 204-205
- 山川裕之, 胸痛を主訴に救急外来を受診した60歳代の男性が、心肥大を認めた症例, 特集 見逃していませんかその症状, 疾患!—日常診療で見逃さないためのTips—, Heart View 24巻12号, 2020年11月, pp. 99-110
- 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
- 小林正久, 松田純子, 笹井英雄, 石毛信之, 大橋十也, 井田博幸. 新生児マススクリーニングでC5-OH 持続高値例の遺伝子型についての検討. 日本マススクリーニング学会誌 30: 53-57, (2020).
- 春石和子, 三木淳司, 荒木俊介, 後藤克聡, 赤池洋人, 松田純子, 尾内一信, 桐生純一. 視覚障害を契機に副腎白質ジストロフィーと診断された1例. 神経眼科 37:165-170 (2020)
- 下澤伸行. 副腎白質ジストロフィー 小児科臨床 特集 診断・治療可能な遺伝性疾患を見逃さないために73 (5) 726-730 (2020)
- 下澤伸行. 副腎白質ジストロフィー (副腎白質ジストロフィー (ALD) 診療ガイドライン2019) 小児科61 臨時増刊号、小児診療ガイドラインのダイジェスト解説&プロGRESS 649-654 (2020)
- 高島茂雄, 下澤伸行. ペルオキシソーム病 生体の科学 増大特集「難病研究の進歩」71(5): 412-413 (2020)
- 松川 敬志. 副腎白質ジストロフィー. 遺伝医学32号Vol.10 67-70. 2020

書籍・雑誌 (英文)

Kaga M, Kaga K, Adrenoleukodystrophy, Landau-Kleffner syndrome and Central Auditory disorders in children, Springer, Singapore, 2021, 97-117

Satoshi Morimoto, Ayumi Nojiri, Eiko Fukuro, Ikuko Anan, Makoto Kawai, Ken Sakurai, Masahia Kobayashi, Hiroshi Kobayashi, Hiroyuki Ida, Toya Ohashi, Takahiro Shibata, Michihiro Yoshiura, Y oshikatsu Eto, Kenichi Hongo Characteristics of the electrocardiogram in Japanese Fary patie nts under long-term enzyme replacement therapy. *Frontiers in Cardiovascular Medicine*, section Gen eral Cardiovascular Medicine, 2020

Torayuki Okuyama,¹ Yoshikatsu Eto,² Norio Sakai,³ Kimitoshi Nakamura,⁴ Tatsuyoshi Yamamoto,⁵ Mariko Yamaoka,⁵ Toshiaki Ikeda,⁵ Sairei So,⁵ Kazunori Tanizawa,⁵ Hiroyuki Sonoda,⁵ and Yu ji Sato A Phase 2/3 Trial of Pabinafusp Alfa, IDS Fused with Anti-Human Transferrin Receptor Antibody, Targeting Neurodegeneration in MPS-II *Mol. Therapy* Vol. .29, No. 2 1-9, 2020

Wu C, Iwamoto T, Hossain MA, Akiyama K, Igarashi J, Miyajima T, Eto Y. *PLoS One*. 2020 Sep 8;15(9):e0238624. A combination of 7-ketocholesterol, lysosphingomyelin and bile acid-408 to diagnos e Niemann-Pick disease type C using LC-MS/MS. *PLoS One*. 2020 Sep 8;15(9):e0238624.

Iwahori A, Maekawa M, Narita A, Kato A, Sato T, Ogura J, Sato Y, Kikuchi M, Noguchi A, Higak i K, Okuyama T, Takahashi T, Eto Y, Mano N. Development of a Diagnostic Screening Strategy for Niemann-Pick Diseases Based on Simultaneous Liquid Chromatography-Tandem Mass Spectrometry Analyses of N-Palmitoyl-O-phosphocholine-serine and Sphingosylphosphorylcholine. *Biol Pharm Bull*. 2020 Sep 1;43(9):1398-1406. doi: 10.1248/bpb.b20-00400. Epub 2020 Jun 25.

K, Harada T, Fukuro E, Kobayashi M, Ohashi T, Eto Y. Massive accumulation of globotriaosylcera mide in various tissues from a Fabry patient with a high antibody titer against alpha-galactosidase A after 6 years of enzyme replacement therapy. *Hongo Mol Genet Metab Rep*. 2020 Jul 16;24:1006 23. doi: 10.1016/j.ymgmr.2020.10062 eCollection 2020 Sep.

Yuskiv N, Higaki K, Stockler-Ipsiroglu S, Morquio B disease, disease characteristics and treatment options of a distinct GLB1-related dystostosis multiplex. *Int J Mol Sci*, 21, 9121, 2020

Sturley SL, Rajakumar T, Hammond N, Higaki K, Marka Z, Marka S, Munkacsi AB, Potential CO VID-19 thearpeutics from a rare disease: weaponizing lipid dysregulation to combat viral infectivity. *J Lipid Res*, 61, 972-982, 2020

Fukaura M, Ishitsuka Y, Shirakawa S, Ushihama N, Yamada Y, Kondo Y, Takeo T, Nakagata N, Motoyama K, Higashi T, Arima H, Kurauchi Y, Seki T, Katsuki H, Higaki K, Matsuo M, Irie T, Intracerebroventricular treatment with 2-hydroxypropyl- β -cyclodextrin decreased cerebellar and hepatic glycoprotein nonmetastatic melanoma protein B (GPNMB) expression in Niemann-Pick disease type C model mice. *Int J Mol Sci*, 22, 452, 2021

Shibata Y, Matsushima M, Matsukawa T, Ishiura H, Tsuji S, Yabe I. Adrenoleukodystrophy sibling s with a novel ABCD1 missense variant presenting with phenotypic differences: a case report a nd literature review. *J Hum Genet*. 2020 Oct 30. doi: 10.1038/s10038-020-00866-x. Online ahead of print

Hiraide T, Yamoto K, Masunaga Y, Asahina M, Endoh Y, Ohkubo Y, Matsubayashi, T, Tsurui S, Y amada H, Yanagi, K, Nakashima M, Hirano K, Sugimura H, Fukuda T, Ogata T, Saitu H. Geneti c and phenotypic analysis of 101 patients with intellectual disability using 3 whole-exome sequencin g *Clin Genet*. doi: 10.1111/cge.13951 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.

Fuseya Y, Sakurai T, Miyahara JI, Sato K, Kaji S, Saito Y, Takahashi M, Nishino I, Fukuda T, Su gie H, Yamashita H. Adult-onset Repeat Rhabdomyolysis with a Very Long-chain Acyl-CoA Dehydro genase Deficiency Due to Compound Heterozygous ACADVL Mutations. *Intern Med*. 59:2729-2732. 2 020.

Ichimoto K, Fujisawa T, Shimura M, Fushimi T, Tajika M, Matsunaga A, Ogawa-Tominaga M, Akiy

ama 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.

Inamura N, Go S, Watanabe T, Takase H, Takakura N, Nakayama A, akebayashi H, Matsuda J and Enokido Y. Reduction in MIR-219 expression underlies cellular pathogenesis of oligodendrocytes in a mouse model of Krabbe disease. *Brain Pathology in press* (2021).

Oji Y, Hatano T, Ueno SI, Funayama M, Ishikawa KI, Okuzumi A, Noda S, Sato S, Satake W, Toda T, Li Y, Hino-Takai T, Kakuta S, Tsunemi T, Yoshino H, Nishioka K, Hattori T, Mizutani Y, Mutoh T, Yokochi F, Ichinose Y, Koh K, Shindo K, Takiyama Y, Hamaguchi T, Yamada M, Farrer MJ, Uchiyama Y, Akamatsu W, Wu YR, Matsuda J, Hattori N. Variants in saposin D domain of prosaposin gene linked to Parkinson's disease. *Brain* 143 (4):1190-1205 (2020).

Tsuboi K, Yamamoto H. Efficacy and safety of enzyme-replacement-therapy with agalsidase alfa in 36 treatment-naïve Fabry disease patients. *BMC Pharmacol Toxicol*. 2017 Jun 7; 18(1):43.

Kazuya Tsuboi, Tamotsu Kanzaki. Skin Lesion in Fabry Disease. *Brain Nerve*. 2019 Apr;71(4):354-359. doi: 10.11477/mf.1416201275

Fei Wang, Hiroshi Yamamoto, Tadao Yoshida, Satofumi Sugimoto, Masaaki Teranishi, Kazuya Tsuboi, Michihiko Sone. Otolological aspects of Fabry disease in patients with normal hearing. *Nagoya J Med Sci*. 2019 Aug;81(3):469-475. doi: 10.18999/nagjms.81.3.469

Tomoaki Haga, Takahiro Okumura, Satoshi Isobe, Fuji Somura, Naoaki Kano, Tasuku Kuwayama, Tsuyoshi Yokoi, Hiroaki Hiraiwa, Toru Kondo, Akinori Sawamura, Ryota Morimoto, Hiroshi Yamamoto, Kazuya Tsuboi, Toyoaki Murohara. Potential prognostic implications of myocardial thallium-201 and iodine-123-beta-methylpentadecanoic acid dual scintigraphy in patients with Anderson-Fabry disease. *Ann Nucl Med*. 2019 Dec;33(12):930-936. doi: 10.1007/s12149-019-01406-0. Epub 2019 Oct 11.

Takaaki Sawada, Jun Kido, Keishin Sugawara, Shirou Matsumoto, Fumio Takada, Kazuya Tsuboi, Akira Ohtake, Fumio Endo, Kimitoshi Nakamura. Detection of novel Fabry disease-associated pathogenic variants in Japanese patients by newborn and high-risk screening. *Mol Genet Genomic Med*. 2020 Nov;8(11):e1502. doi: 10.1002/mgg3.1502. Epub 2020 Oct 5.

Sakai T, Honzawa S, Kaga M et.al. Osteoporosis pathology in people with severe motor and intellectual disability. *Brain Dev*. 2020;42:256-63.

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* 2021; 27

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* 2021; 29(2): 671-679

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* 2021; 15 (1)

Sawada T, Kido J, Sugawara K, Matsumoto S, Takada F, Tsuboi K, Ohtake A, Endo F, Nakamura K. Detection of novel Fabry disease-associated pathogenic variants in Japanese patients by newborn and high-risk screening. *Molecular Genetics and Genomic Medicine* 2020; 8(11) : e1502

Lee T, Yoshii K, Yoshida S, Suga T, Nakamura K, Sasai H, Murayama K, Kobayashi H, Hasegawa Y, Takeshima Y. Retrospective evaluations revealed pre-symptomatic citrulline concentrations measured by newborn screening were significantly low in late-onset ornithine transcarbamylase deficiency patients. *Clinica Chimica Acta* 2020; 510: 633-637

Naramura T, Tanaka K, Inoue T, Imamura H, Yoshimatsu H, Mitsubuchi H, Nakamura K, Iwai M. New reference ranges of procalditonin excluding respiratory failure in neonates. *Pediatrics International* 2020; 62(10): 1151-1157

Kido J, Inoue H, Shimotsu H, Yoshida Y, Suzuki Y, Nakamura K, Endo F, Matsumoto S. Effect of L-Carnitine on Amino Acid Metabolism in Elderly Patients Undergoing Regular Hemodialysis. *Blood Purification* 2020; 49(5): 614-621

Yoshida S, Kido J, Sawada T, Momosaki K, Sugawara K, Matsumoto S, Endo F, Nakamura K. Fabry disease screening in high-risk populations in Japan: A nationwide study. *Orphanet Journal of Rare Diseases* 2020; 15(1)

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. *Molecular Genetics and Metabolism* 2020; 130(3): 215-224

Liang K-H, Lu Y-H, Niu C-W, Chang S-K, Chen Y-R, Cheng C-Y, Hsu T-R, Yang C-F, Nakamura K, Niu D-M. The Fabry disease-causing mutation, GLA IVS4+919G>A, originated in Mainland China more than 800 years ago. *Journal of Human Genetics* 2020; 65 (7): 619-625

Sawada T, Kido J, Yoshida S, Sugawara K, Momosaki K, Inoue T, Tajima G, Sawada H, Mastumoto S, Endo F, Hirose S, Nakamura K. Newborn screening for Fabry disease in the western region of Japan. *Molecular Genetics and Metabolism Reports* 2020; 22: 100562

Tanaka K, Sakamoto R, Imamura H, Naramura T, Matsumoto S, Iwai M, Mitsubuchi H, Nakamura K. Reversal of blood flow in deep cerebral vein in preterm intraventricular hemorrhage: two case reports. *BMC Pediatrics* 2020; 20(1)

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* 2020; 8(11): 2314-2315

Sato S, Ohnishi T, Uejima Y, Furuichi M, Fujinaga S, Imai K, Nakamura K, Kawano Y, Suganuma E. Induction therapy with rituximab for lupus nephritis due to prolidase deficiency. *Rheumatology (United Kingdom)* 2020; 59(10): E57-E59

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* 2020; 40(5): 780-782

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* 2020; 12(3)

Kusunoki S, Kido J, Momosaki K, Sawada T, Kashiki T, Matsumoto S, Nakamura K. Effect of Flunarizine on Alternating Hemiplegia of Childhood in a Patient with the p.E815K Mutation in ATP1A3: A Case Report. *Case Reports in Neurology* 2020; 299-306

Kido J, Nakamura K, Era T. Role of induced pluripotent stem cells in lysosomal storage diseases. *Molecular and Cellular Neuroscience* 2020; 108

Sawada T, Kido J, Nakamura K. Newborn screening for pompe disease. *International Journal of Neonatal Screening* 2020; 6(2)

Matsuda Y, Morino H, Miyamoto R, Kurashige T, Kume K, Mizuno N, Kanaya Y, Tada Y, Ohsawa R, Yokota K, Shimozawa N, Maruyama H, Kawakami H. Biallelic mutation of HSD17B4 induces middle age-onset spinocerebellar ataxia. *Neurol Genet.* 16;6: e396 (2020)

Tanaka H, Amano N, Tanaka K, Katsuki T, Adachi T, Shimozawa N, Kawai T. A 29-year-old patient with adrenoleukodystrophy presenting with Addison's disease. *Endocr J.* 67(6): 655-658 (2020)

Hama K, Fujiwara Y, Takashima S, Hayashi Y, Yamashita A, Shimozawa N, Yokoyama K. Hexacos

enoyl-CoA is the most abundant very long-chain acyl-CoA in ATP-binding cassette transporter D1-deficient cells. *J Lipid Res* 61(4): 523-53 (2020)

Obara K, Abe E, Shimozawa N, Toyoshima I. A case of female adrenoleukodystrophy carrier with incontinent neurogenic bladder. *J Gen Fam Med*. 21: 146-147 (2020)

Takashima S, Toyoshi K, Yamamoto T, Shimozawa N. Positional determination of the carbon-carbon double bonds in unsaturated fatty acids mediated by solvent plasmation using LC-MS. *Sci Rep*. 10(1): 12988 (2020)

Morita A, Enokizono T, Ohto T, Tanaka M, Watanabe S, Takada Y, Iwama K, Mizuguchi T, Matsumoto N, Morita M, Takashima S, Shimozawa N, Takada H. Novel ACOX1 mutations in two siblings with peroxisomal acyl-CoA oxidase deficiency. *Brain Dev*. 43(3): 475-481 (2021)

Kubota K, Kawai H, Takashima S, Shimohata T, Otsuki M, Ohnishi H, Shimozawa N. Clinical evaluation of childhood cerebral adrenoleukodystrophy with balint's symptoms. *Brain Dev*. 43(3): 396-401 (2021)

Morita M, Kaizawa T, Yoda T, Oyama T, Asakura R, Matsumoto S, Nagai Y, Watanabe Y, Watanabe S, Kobayashi H, Kawaguchi K, Yamamoto S, Shimozawa N, So T, Imanaka T. Bone marrow transplantation into *Abcd1*-deficient mice: Distribution of donor derived-cells and biological characterization of the brain of the recipient mice. *J Inherit Metab Dis*. 2020 Dec 17. doi: 10.1002/jimd.123

Imanaka T., and Kawaguchi K.: A novel dynein-type AAA+ protein with peroxisomal targeting signal type 2. *J Biochem*. (2020) 167, 429-432

Morita M., Kaizawa T., Yoda T., Oyama T., Asakura R., Matsumoto S., Nagai Y., Watanabe Y., Watanabe S., Kobayashi H., Kawaguchi K., Yamamoto S., Shimozawa N., So T., and Imanaka T.: Bone marrow transplantation into *Abcd1*-deficient mice: Distribution of donor derived-cells and biological characterization of the brain of the recipient mice. *J Inherit Metab Dis*. (2020) Dec 17. Online ahead of print.

Kawaguchi K., Mukai E., Watanabe S., Yamashita A., Morita M., So T., and Imanaka T.: Acyl-CoA thioesterase activity of peroxisomal ABC protein ABCD1 is required for the transport of very long-chain acyl-CoA into peroxisomes. *Sci Rep*. (2021) 26, 11: 2192.

Morita M., Toida A., Horiuchi Y., Watanabe S., Sasahara M., Kawaguchi K., So T., and Imanaka T.: Generation of an immortalized astrocytic cell line from *Abcd1*-deficient H-2K^btsA58 mice to facilitate the study of the role of astrocytes in X-linked adrenoleukodystrophy. *Heliyon*. (2021) 11, 7: e06228.

Kitai K., Kawaguchi K., Tomohiro T., Morita M., So T., and Imanaka T.: The lysosomal protein ABCD4 transports vitamin B₁₂ across liposomal membranes *in vitro*. *J. Biol. Chem*. (2021) in press.

Kurotsu S, Sadahiro T, Fujita R, Tani H, Yamakawa H, Tamura F, Isomi M, Kojima H, Yamada Y, Abe Y, Murakata Y, Akiyama T, Muraoka N, Harada I, Suzuki T, Fukuda K, Ieda M. Soft Matrix Promotes Cardiac Reprogramming via Inhibition of YAP/TAZ and Suppression of Fibroblast Signatures. *Stem Cell Reports*. 2020 Sep 8;15(3):612-628. doi: 10.1016/j.stemcr.2020.07.022. Epub 2020 Aug 27.

Yamakawa H, Kusumoto D, Hashimoto H, Yuasa S. Stem Cell Aging in Skeletal Muscle Regeneration and Disease. *Int J Mol Sci*. 2020 Mar 6;21(5):1830. doi: 10.3390/ijms21051830

(その他)

「新型コロナウイルス感染拡大に伴う 緊急事態下のライソゾーム病酵素補充療法についての提言」

第11回、国際協力遺伝病遺伝子治療フォーラム「今、我が国の遺伝子細胞治療の課題を整理する」

第7回市民公開フォーラム「ライソゾーム病・ペルオキシソーム病 (LSD/PD) 診療の現状と未来～コロナ禍をいかに乗り切るか」

【令和3年度】

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
NPC診療ガイドライン作成委員会	ニーマンピック病C型(NPC)診療ガイドライン2021	NPC診療ガイドライン作成委員会	ニーマンピック病C型(NPC)診療ガイドライン2021	診断と治療社	東京	2021
奥山 虎之		衛藤 義勝 大橋 十也	ファブリー病 Up Date 改訂第2版	診断と治療社	東京	2021
衛藤 義勝	ファブリー病の歴史と概要	衛藤 義勝 大橋 十也	ファブリー病 Up Date 改訂第2版	診断と治療社	東京	2021
志村 優 村山 圭	疾患 3. 先天代謝異常	水口 雅 山形 崇倫	クリニカルガイド小児科 専門医の診断・治療	南山堂	東京	2021
村山 圭	各論の治療 5ミトコンドリア病	日本小児集中治療研究会	小児救急・ICUピックアップ5内 内分泌・代謝救急疾患	メディカル・サイエンス・インターナショナル	東京	2021
松川 敬志	Column 副腎白質ジストロフィーにおける造血幹細胞移植療法	矢部 一郎	臨床遺伝専門医テキスト④ 各論III 臨床遺伝学成人領域	診断と治療社	東京	2021
坪井 一哉	QOL、疼痛に対する効果	衛藤 義勝 大橋 十也	ファブリー病 Up Date 改訂第2版	診断と治療社	東京	2021
坪井 一哉	皮膚、自立神経症状に対する効果	衛藤 義勝 大橋 十也	ファブリー病 Up Date 改訂第2版	診断と治療社	東京	2021
坪井 一哉	ファブリー病	鈴木 則宏 監修 永田 栄一郎 伊藤 義彰 編	脳神経内科学レビュー2022-2023	総合医学社	東京	2022
坪井 一哉	ライソゾーム病の治療とマネジメント	鈴木 則宏 荒木 信夫 宇川 義一 桑原 聡 塩川 芳昭	Annual Review 神経2022	中外医学社	東京	2022

下澤 伸行	遺伝生化学	臨床遺伝専門医制度委員会監修	遺伝生化学 臨床遺伝専門医テキスト 1 臨床遺伝学総論	診断と治療社	東京	2021
下澤 伸行	副腎白質ジストロフィー	下畑 享良(編著)	脳神経内科診断ハンドブック	中外医学社	東京	2022
櫻井 謙	新規治療の開発(基質合成抑制治療薬, 核酸治療薬)	衛藤義勝 大橋十也	ファブリー病 Up Date 改訂第2版	診断と治療社	日本	2021
右田 王介	ムコ多糖症(MPS)I型診療ガイドライン 2020	日本先天代謝異常学会	ムコ多糖症(MPS)I型診療ガイドライン 2020	診断と治療社	東京	2021
小須賀 基通	疾患 3. 先天代謝異常	水口 雅 山形 崇倫	クリニカルガイド 小児科 専門医の診断・治療	南山堂	東京	2021
小須賀 基通	VI章 二次性心筋症(アミロイドーシス, サルコイドーシス, ファブリーなど)	清水 渉(監)	最新主要文献とガイドラインでみる循環器内科学レビュー2022-'23	総合医学社	東京	2021

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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
福原 康之, 奥山 虎之	確定的な遺伝子解析法とその活用 単一遺伝子疾患の家族発症: 次子妊娠時の対応	周産期医学	51巻5号	719-722	2021
奥山 虎之	ファブリー病の早期診断をいかに進めるか	日本皮膚科学会雑誌	131巻5号	1320	2021
高橋 勉	ライソゾーム病における低分子療法	小児科診療	12	1803-1808	2021
杉山 洋平 村山 圭	糖尿病・内分泌代謝科	先天代謝異常症のガイドライン	53巻4号	459 - 464	2021
坪井 一哉	ファブリー病 子供たちの叫びが聴こえますか?	月刊母子保健	11月号		2021
川合 裕規, 久保田 一生, 下澤 伸行	高次脳機能障害で発症した小児大脳型副腎白質ジストロフィーの早期診断のための臨床的検討	小児の精神と神経	第61巻1号	35-41	2021

下澤 伸行	副腎白質ジストロフィー新生児マススクリーニング国内導入に向けての現状と課題. 特集 新生児マススクリーニングと治療の最前線	遺伝子医学	Vol.11/No.3 [7月号]	80-87	2021
高島 茂雄、 <u>下澤 伸行</u>	ペルオキシソーム病における脂質代謝と治療	The Lipid (リピッド)	32 (2)	76-84	2021
下澤 伸行	ABCD1 (関連疾患: 副腎白質ジストロフィー)	小児科診療	84巻 11号	1511-1513	2021
加我 牧子	Landau-Kleffner症候群	小児内科	53(10)	1705-9	2021
加我 牧子	発達性構音障害	脳神経内科	95(1)	80-5	2021
右田 王介	遺伝子機能制御を担うエピゲノムの解析による疾患発現機序の理解	日本医師会雑誌	149(11)	1947-1951	2021
杉下 陽堂, <u>右田 王介</u> , 鈴木 由妃, 本吉 愛, 岩端 秀之, 高江 正道, 洞下 由記, 菅沼 真樹, 津川 浩一郎, 鈴木 直	当院で早発卵巣不全の経過をたどったトリプルX症候群5症例における心理カウンセリングの意義の検討	日本生殖心理学会誌	7(1)	48-52.	2021
小須賀 基通	【小児遺伝子疾患事典】代謝疾患 IDS(関連疾患: ムコ多糖症 II 型)	小児科診療	84巻11号	1535-1536	2021
蘇 哲民, 小須賀 基通	疑う臨床症状 外観の特徴から疑うポイント	小児科診療	84巻12号	1729-1734	2021
小須賀基通	臨床 ライソゾーム病	The Lipid	2021年10月号 (Vol. 32 No. 2)	156-162	2021
小須賀 基通	ライソゾーム病	糖尿病・内分泌代謝科	53巻4号	387-393	2021
小須賀 基通	ムコ多糖症	遺伝子医学	11巻3号	73-79	2021
小須賀基通	(VI章)二次性心筋症(アミロイドーシス、サルコイドーシス、ファブリーなど) ファブリー病の診断と治療	循環器内科学レビュー	2022-'23巻	259-267	2021
<u>Okuyama, T, Eto, Y, Sakai, N, Nakamura, K, Yamamoto, T, Yamaoka, M, Ikeda, T, So, S, Tanizawa, K, Sonoda, H, Sato, Y.</u>	A Phase 2/3 Trial of Pabinafusp Alfa, IDS Fused with Anti-Human Transferrin Receptor Antibody, Targeting Neurodegeneration in MPS-II.	Molecular Therapy	2021;29(2)	671-679	2021

Giugliani R, Martins A, <u>Okuyama T</u> , <u>Eto Y</u> , <u>Sakai N</u> , <u>Nakamura K</u> , Morimoto H, Minami K, Yamamoto T, Yamaoka M, Ikeda T, So S, Tanizawa K, Sonoda H, Schmidt M, Sato Y	Enzyme Replacement Therapy with Pabinafusp Alfa for Neuronopathic Mucopolysaccharidosis II: An Integrated Analysis of Preclinical and Clinical Data.	Int J Mol Sci.	2021 Oct 10;22(20)	10938	2021
Koto Y, <u>Sakai N</u> , Lee Y, Kakee N, <u>Matsuda J</u> , <u>Tsuboi K</u> , <u>Shimozawa N</u> , <u>Okuyama T</u> , <u>Nakamura K</u> , <u>Narita A</u> , <u>Kobayashi H</u> , Uehara R, Nakamura Y, Kato K, <u>Eto Y</u> .	Prevalence of patients with lysosomal storage disorders and peroxisomal disorders: A nationwide survey in Japan.	Mol Genet Metab	133(3)	277-288	2021
Saito R., <u>EtoY</u> . et al	A neuropathological cell model derived from Niemann-Pick disease type C patient-specific iPSCs shows disruption of the p62/SQSTM1- KEAP1- NRF2 Axis and impaired formation of neuronal networks	Mol. Genet. Metab. Rep	28	1-12	2021
Yamada Y, Ishitsuka Y, Kondo Y et al,	Differential mode of cholesterol inclusion with 2-hydroxypropyl-cyclodextrins increases safety margin in treatment of Niemann-Pick disease type C.	Br J Pharmacol	178(13)	2727-2746	2021
Chen H, Khan S, Celik B, <u>Suzuki Y</u> , Ago Y, Tomatsu S	Activity of daily living in mucopolysaccharidosis IVA patients: evaluation of therapeutic efficacy	Mol Genet Genom Med	9	e1806	2021
<u>Tsuboi K</u> , Tai T, Yamashita R, Ali H, Watanabe T, Uyama T, Okamoto Y, Kitakaze K, Takenouchi Y, Go S, Rahman IAS, Houchi H, Tanaka T, Okamoto Y, Tokumura A, <u>Matsuda J</u> , Ueda N.	Involvement of acid ceramidase in the degradation of bioactive N-acyl ethanolamines.	Biochim Biophys Acta Mol Cell Biol Lipids.	1866 (9)	158972	2021

Inamura N, Go S, Watanabe T, Takase H, Takakura N, Nakayama A, Takebayashi H, <u>Matsuda J</u> and Enokido Y.	Reduction in MIR-219 expression underlies cellular pathogenesis of oligodendrocytes in a mouse model of Krabbe disease.	Brain Pathology	31 (5)	e12951	2021
Morita M, Kaizawa T, Yoda T, Oyama T, Asakura R, Matsumoto S, Nagai Y, <u>Watanabe Y</u> , Watanabe S, <u>Kobayashi H</u> , Kawaguchi K, Yamamoto S, <u>Shimozawa N</u> , So T, <u>Imanaka T</u> .	Bone marrow transplantation into Abcd1-deficient mice: Distribution of donor derived-cells and biological characterization of the brain of the recipient mice.	J Inher Metab Dis.	44 (3)	718-727	2021
Kubota K, Kawai H, Takashima S, Shimohata T, Otsuki M, Ohnishi H, <u>Shimozawa N</u> . Clinical evaluation of childhood cerebral adrenoleukodystrophy with balint's symptoms.	Clinical evaluation of childhood cerebral adrenoleukodystrophy with balint's symptoms.	Brain Dev.	43(3)	396-401	2021
Takashima S, Takemoto S, Toyoshiki K, Ohba A, <u>Shimozawa N</u> .	Zebrafish model of human Zellweger syndrome reveals organ-specific accumulation of distinct fatty acid species and widespread gene expression changes.	Mol Genet Metab.	2021 May 8:S1096-7192(21)	00703-4	2021
Fujiwara Y, Hama K, <u>Shimozawa N</u> , <u>Yokoyama K</u> .	Glycosphingolipids with Very Long-Chain Fatty Acids Accumulate in Fibroblasts from Adrenoleukodystrophy Patients.	Int J Mol Sci.	2021 Aug 11;22(16)	8645	2021
<u>Shimozawa N</u> , Takashima S, Kawai H, Kubota K, Sasaki H, Orii K, Ogawa M, Ohnishi H.	Advanced Diagnostic System and Introduction of Newborn Screening of Adrenoleukodystrophy and Peroxisomal Disorders in Japan.	Int J Neonatal Screen.	2021 Aug 25;7(3)	58	2021

Koji Kato, <u>Hiro</u> <u>sa Yabe</u> , <u>Nobuyuki</u> <u>Shimozawa</u> , Souic hi Adachi, Mineo Kurokawa, Yoshiko Hashii, Atsushi S ato, Nao Yoshida, <u>Makiko Kaga</u> , Osa mu Onodera, Shun ichi Kato, Yoshiko Atsuta, and Tomoh iro Morio.	Stem Cell Transplantation for Pediatric Patients with Adrenoleukodystrophy: A nationwide retrospective a nalysis in Japan.	Pediatric Tra nsplantation.	2021 Oct 18;	e14125	2021
Kawaguchi K., and <u>Imanaka T.</u>	Substrate specificity and t he direction of transport i n the ABC transporters A BCD1-3 and ABCD4.	Chem Phar Bull	<i>in press</i>		2022
Kitai K., Kawaguc hi K., Tomohiro T., Morita M., So T., and <u>Imanaka T.</u>	The lysosomal protein AB CD4 transports vitamin B ₁ 2 across liposomal membra nes <i>in vitro</i> .	J. Biol. Che m	296	100654	2021
Sera Y., Sadoya M., Ichinose T., M atsuya S., <u>Imanaka</u> <u>T.</u> , and Yamaguchi, M.	SBDS interacts with RNF 2 and isdegraded through RNF2-dependent ubiquitin ation	Biochem Bio phys Res Co mmun	598	119-123	2022
Morimoto S, Nojiri A, Fukuro E, An an I, Kawai M, <u>Sa</u> <u>kurai K</u> , <u>Kobayash</u> <u>i M</u> , <u>Kobayashi H</u> , Ida H, Ohashi T, Shibata T, Yoshim ura M, <u>Eto Y</u> , Ho ngo K.	Characteristics of the Electrocardiogram in Japanese Fabry Patients Under Long-Term Enzyme Replacement Therapy	Front Cardio vasc Med.	7	614129	2021
Anan I, Sakuma T, Fukuro E, Mori moto S, Nojiri A, Kawai M, <u>Sakurai</u> <u>K</u> , <u>Kobayashi M</u> , <u>Kobayashi H</u> , Ida H, Ohashi T, Yosh imura M, <u>Eto Y</u> , Hongo K	The role of native T1 valu es on the evaluation of ca rdiac manifestation in Jap anese Fabry disease patie nts.	Mol Genet Metab rep	<i>in press</i>		2022
Sugihara K, Yuasa M, Isozaki Y, Hat a I, Ohshima Y, H amazaki T, Kakiuc hi T, Arao M, Igar ashi N, Kotani Y, <u>Fukuda T</u> , Kagaw a R, Tajima G, Sh igematsu Y.	Severity estimation of very -long-chain acyl-CoA dehyd rogenase deficiency via 13 C-fatty acid loading test.	Pediatr Res.	Online ah ead of pri nt	PMID: 3513 6200	2022

Mori T, Ishikawa A, Shigetomi H, <u>Fukuda T</u> , Sugie H	A novel PHKA2 variant in a Japanese boy with glycogen storage diseases type IXa	Pediatr Int.	64(1)	e14839.	2022
Munekane A, Ohsawa Y, <u>Fukuda T</u> , Nishimura H, Nishimatsu SI, Sugie H, Saito Y, Nishino I, Sunada Y.	Maximal Multistage Shuttle Run Test-induced Myalgia in a Patient with Muscle Phosphorylase B Kinase Deficiency.	Intern Med.	Online ahead of print	PMID: 34615823	2021
Kido, J, Matsumoto, S, Ito, T, Hirose, S, Fukui, K, Kojima-Ishii, K, Mushimoto, Y, Yoshida, S, Ishige, M, Sakai, N, <u>Nakamura, K</u> .	Physical, cognitive, and social status of patients with urea cycle disorders in Japan.	Molecular Genetics and Metabolism Reports	2021; 27		2021
Kido J, Matsumoto S, Häberle J, Nakajima Y, Wada Y, Mochizuki N, Murayama K, Lee T, Mochizuki H, Watanabe Y, Horikawa R, Kasahara M, <u>Nakamura K</u>	Long-term outcome of urea cycle disorders: Report from a nationwide study in Japan	J Inherit Metab Dis.	2021 Jul;44(4)	826-837	2021
Sawada J, Nakagawa N, Kano K, Saito T, Katayama T, Sawada T, Momosaki K, <u>Nakamura K</u> , Hasebe N	Characteristics of Neurological Symptoms in Adult Japanese Patients with Fabry Disease.	Intern Med.	2021 Jun 15;60(12)	1819-1826	2021
Miyashita Y, Kouwaki T, Tsukamoto H, Okamoto M, <u>Nakamura K</u> , Oshiumi H.	TICAM-1/TRIF associates with Act1 and suppresses IL-17 receptor-mediated inflammatory responses.	Life Sci Alliance.	2021 Nov 24;5(2)	e202101181	2021
Naramura T, Imamura H, Yoshimatsu H, Hirashima K, Irie S, Inoue T, Tanaka K, Mitsubuchi H, <u>Nakamura K</u> , Iwai M	The Predictive Value of Procalcitonin and High-Sensitivity C-Reactive Protein for Early Bacterial Infections in Preterm Neonates.	Neonatology.	2021;118(1)	28-36	2021
Kohrogi K, Hino S, Sakamoto A, Anan K, Takase R, Araki H, Hino Y, Araki K, Sato T, <u>Nakamura K</u> , Nakao M	LSD1 defines erythroleukemia metabolism by controlling the lineage-specific transcription factors GATA1 and C/EBP α .	Blood Adv.	2021 May 11;5(9)	2305-2318	2021

Kido J, Matsumoto S, Häberle J, Inomata Y, Kasahara M, Sakamoto S, Horikawa R, Tanemura A, Okajima H, Suzuki T, <u>Nakamura K</u>	Role of liver transplantation in urea cycle disorders: Report from a nationwide study in Japan.	J Inherit Metab Dis.	2021 Nov; 44(6)	1311-1322	2021
Hama R, Kido J, Sugawara K, Nakamura T, <u>Nakamura K</u>	Hyperprolinemia type I caused by homozygous p.T466M mutation in PRODH.	Hum Genome Var.	2021 Jul 20;8(1)	28	2021
Kido J, Matsumoto S, Takeshita E, Hayasaka C, Yamada K, Kagawa J, Nakajima Y, Ito T, Iijima H, Endo F, <u>Nakamura K</u>	Current status of surviving patients with arginase 1 deficiency in Japan.	Mol Genet Metab Rep.	2021 Oct 1;29:	100805	2021
Sawada T, Kido J, Sugawara K, Momosaki K, Yoshida S, Kojima-Ishii K, Inoue T, Matsumoto S, Endo F, Ohga S, Hirose S, <u>Nakamura K</u>	Current status of newborn screening for Pompe disease in Japan.	Orphanet J Rare Dis.	2021 Dec 18;16(1)	516	2021
Kinoshita Y, Momosaki K, Matsumoto S, Murayama K, <u>Nakamura K</u>	Severe metabolic acidosis with cardiac involvement in DNMT1L-related mitochondrial encephalopathy.	Pediatr Int.	2022 Jan; 64(1)	e14879	2022
Iwai M, Yoshimatsu H, Naramura T, Imamura H, Nakamura T, Sakamoto R, Inoue T, Tanaka K, Matsumoto S, Nakamura K, Mitsubuchi H	Procalcitonin is associated with postnatal respiratory condition severity in preterm neonate.	Pediatr Pulmonol.	2022 Jan 22. doi:	10.1002/ppu.125846.	2022
Kido J, Häberle J, Sugawara T, Tanaka T, Nagao M, Sawada T, Wada Y, Numakura C, <u>Murayama K</u> , <u>Watanabe Y</u> , Kojima-Ishii K, Sasai H, Kosugiyama K, <u>Nakamura K</u>	Clinical manifestation and long-term outcome of citrin deficiency: Report from a nationwide study in Japan.	J Inherit Metab Dis.	2022 Feb 10.	doi: 10.1002/jimd.12483.	2022

Sawada T, Kido J, Sugawara K, Yoshida S, Matsumoto S, Shimazu T, Matsushita Y, Inoue T, Hirose S, Endo F, <u>Nakamura K</u>	Newborn screening for Gaucher disease in Japan.	Mol Genet Metab Rep.	2022 Feb 18;31:	100850.	2022
Miyashita Y, Yoshida T, Takagi Y, Tsukamoto H, Takashima K, Kouwaki T, Makino K, Fukushima S, <u>Nakamura K</u> , Oshiumi H.	Circulating extracellular vesicle microRNAs associated with adverse reactions, proinflammatory cytokine, and antibody production after COVID-19 vaccination.	NPJ Vaccines.	2022 Feb 8;7(1)	16	2022
Tomita K, Okamoto S, Seto T, <u>Hamazaki T</u> .	Real world long-term outcomes in patients with mucopolysaccharidosis type II: A retrospective cohort study.	Mol Genet Metab Rep.	29	100816	2021
Tomita K, Okamoto S, Seto T, <u>Hamazaki T</u> , So S, Yamamoto T, Tanizawa K, Sonoda H, Satou Y.	Divergent developmental trajectories in two siblings with neuropathic mucopolysaccharidosis type II (Hunter syndrome) receiving conventional and novel enzyme replacement therapies: A case report.	JIMD Rep.	62(1)	9-14	2021
Seo JH, <u>Kosuga M</u> , <u>Hamazaki T</u> , Shintaku H, <u>Okuyama T</u> .	Impact of intracerebroventricular enzyme replacement therapy in patients with neuronopathic mucopolysaccharidosis type II.	Mol Ther Methods Clin Dev.	21	67-75	2021
Katsuki Y, Abe M, Park SY, Wu W, <u>Yabe H</u> , Yabe M, van Attikum H, Nakada S, Ohta T, Seidman MM,	RNF168 E3 ligase participates in ubiquitin signaling and recruitment of SLX4 during DNA crosslink repair.	Cell Rep	Oct 26;37(4)	109879. doi: 10.1016/j.celrep.	2021
Hayakawa A, Sato I, Kamibeppu K, Ishida Y, Inoue M, Sato A, Shiohara M, <u>Yabe H</u> , Koike K, Adachi S, Atsuta Y, Yamashita T, Kanda Y, Okamoto S	Impact of chronic GVHD on QOL assessed by visual analogue scale in pediatric HSCT survivors and differences between raters: a cross-sectional observational study in Japan	Int J Hematol	Oct 3. doi: 10.1007	s12185-021-03227-2.	2021
Mikami K, Akama F, Kimoto K, Okazawa H, Orihashi Y, Onishi Y, Takahashi Y, <u>Yabe H</u> , Ya	Iron supplementation for hypoferritinemia-related psychological symptoms in children and adolescents.	Nippon Med Sc	Sep 14.	doi: 10.1272/jnms.	2021

Miyamoto S, Umeda K, Kurata M, Nishimura A, Yanagimachi M, Ishimura M, Sato M, Shigemura T, Kato M, Sasahara Y, Iguchi A, Koike T, Takahashi Y, Kajiwara M, Inoue M, Hashii Y, <u>Yabe H</u> , Kato K, Atsuta Y, Imai K, Morio T.	Hematopoietic Cell Transplantation for Severe Combined Immunodeficiency Patients: a Japanese Retrospective Study	J Clin Immunol	Nov;41(8)	1865-1877	2021
Kada A, Kikuta A, Saito AM, Kato K, Iguchi A, <u>Yabe H</u> , Ishida H, Hyakuna N, Takahashi	Single-Arm Non-Blinded Multicenter Clinical Trial on T-Cell-Replete Haploidentical Stem Cell Transplantation Using Low-Dose Antit	Kurume Med J	Oct 6;66(3)	161-168	2021
Yabe T, Satake M, Odajima T, Watanabe-Okochi N, Azuma F, Kashiwase K, Matsumoto K, Orihara T, <u>Yabe H</u> , Kato S, Kato K, Kai S, Mori T, Morishima S, Takanaishi M, Nakajima K, Murata M, Morishima Y	Combined impact of HLA-allele matching and the CD34-positive cell dose on optimal unit selection for single-unit cord blood transplantation in adults	Leuk Lymphoma	Nov;62(11)	2737-2746	2021
Kanda Y, Inoue M, Uchida N, Onishi Y, Kamata R, Kotaki M, Kobayashi R, Tanaka J, Fukuda T, Fujii N, Miyamura K, Mori SI, Mori Y, Morishima Y, <u>Yabe H</u> , Koderu Y	Cryopreservation of Unrelated Hematopoietic Stem Cells from a Blood and Marrow Donor Bank During the COVID-19 Pandemic: A Nationwide Survey by the Japan Marrow Donor Program.	Transplant Cell Ther	Aug;27(8)	664.e1-664	2021
<u>Yamakawa H</u> , Kato TS, Noh JY, Yuasa S, Kawamura A, Fukuda K, Aizawa Y.	Thyroid Hormone Plays an Important Role in Cardiac Function: From Bench to Bedside.	Front Physiol.	2021 Oct 18;12	606931	2021
Adachi K, Tokuyama H, Oshima Y, Itoh T, Hashiguchi A, <u>Yamakawa H</u> , Togawa T, Sakuraba H, Wakino S, Itoh H.	Fabry disease associated with multiple myeloma: a case report.	CEN Case Rep.	2022 Feb; 11(1)	146-153	2021
<u>Yamakawa H</u> , Ieda M.	Cardiac regeneration by direct reprogramming in this decade and beyond.	Inflamm Regen.	2021 Jul 1;41(1)	20	2021

Isomi M, Sadahiro T, <u>Yamakawa H</u> , Fujita R, Yamada Y, Abe Y, Murakata Y, Akiyama T, Shu T, Mizukami H, Fukuda K, Ieda M.	Overexpression of Gata4, Mef2c, and Tbx5 Generate s Induced Cardiomyocytes Via Direct Reprogramming and Rare Fusion in the Heart.	Circulation	2021 May 25;143(21)	2123-2125	2021
Minatogawa M, Unzaki A, Morisaki H, Syx D, Sonoda T, Janecke AR, Slavotinek A, Voermans NC, Lacassie Y, Mendoza-Londono R, Wierenga KJ, Jayakar P, Gahl WA, Tift CJ, Figueroa LE, Hilhorst-Hofstee Y, Maugeri A, Ishikawa K, Kobayashi T, Aoki Y, Ohura T, Kawame H, Kono M, Mochida K, Tokoro dani C, Kikkawa K, Morisaki T, Kobayashi T, Nakane T, Kubo A, Ranelis JD, <u>Migita O</u> , Sobey G, Kaur A, Ishikawa M, Yamaguchi T, Matsumoto N, Malfait F, Miyake N, Kosho T	Clinical and molecular features of 66 patients with musculocontractural Ehlers-Danlos syndrome caused by pathogenic variants in CHST14 (mcEDS-CHST14)	J Med Genet	November 23, 2021	jmedgenet-2020-107623	2021
Hori A, <u>Migita O</u> , Kawaguchi-Kawata R, Narumi-Kishimoto Y, Takada F, Hata K	A novel TAB2 mutation detected in a putative case of frontometaphyseal dysplasia	Hum Genome Var.	8(1)	40	2021
Taniguchi K, Inoue M, Arai K, Uchida K, <u>Migita O</u> , Akemoto Y, Hirayama J, Takeuchi I, Shimizu H, Hata K	Novel TNFAIP3 microdeletion in a girl with infantile-onset inflammatory bowel disease complicated by a severe perianal lesion	Hum Genome Var.	8(1)	1	2021
Kuji S, Kondo H, Ohara T, Deura I, Tozawa-Ono A, <u>Migita O</u> , Kawamoto H, Tsugawa K, Chosokabe M, Koike J, Maeda I, Suzuki N	Value of adjuvant chemotherapy and informed microscopic examination for occult gynecologic cancer detected upon risk-reducing salpingo-oophorectomy after chemotherapy for BRCA1/2-associated breast cancer	Jpn J Clin Oncol	51(3)	492-497	2021

Morita M, Takeuchi I, Kato M, <u>Migita O</u> , Jimbo K, Shimizu H, Yoshimura S, Tomizawa D, Shimizu T, Hata K, Ishiguro A, Arai K	Intestinal outcome of bone marrow transplantation for monogenic inflammatory bowel disease	Pediatr Int.		e14750	2022
Hori A, Ogata-Kawata H, Sasaki A, Takahashi K, Taniguchi K, <u>Migita O</u> , Kawashima A, Okamoto A, Sekizawa A, Sago H, Takada F, Nakabayashi K, Hata K	Improved library preparation protocols for amplicon sequencing-based noninvasive fetal genotyping for RHD-positive D antigen-negative alleles	BMC Res Notes.	2021;14(1):380	380	2021
Yamauchi M, Nakayama H, Shiota S, Ohshima Y, Terada J, Nishijima T, <u>Kosuga M</u> , Kitamura T, Tachibana N, Oguri T, Shirahama R, Aoki Y, <u>Ishigaki K</u> , Sugie K, Yagi T, Muraki H, Fujita Y, Takatani T, Muro S.	Potential patient screening for late-onset Pompe disease in suspected sleep apnea: a rationale and study design for a Prospective Multicenter Observational Cohort Study in Japan (PSAP-J Study).	Sleep Breath.	2021 Jun; 25(2)	695-704	2021

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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
衛藤義勝	ファブリ病	鈴木則宏	最新ガイドラインに基づく 神経疾患診療 指針	総合医学社	東京	2021-22	238
高橋 勉	Niemann-Pick病 酸性スフィンゴリエリナーゼ欠損症とNiemann-Pick病C型	「小児内科」 「小児外科」 編集委員会	小児疾患診療のための病態生理 3 改訂第6版	東京医学社	東京	2022	
村山 圭 伏見 拓矢 杉山 洋平	II. 先天代謝異常 10. ミトコンドリア病	「小児内科」 「小児外科」 編集委員会 共編	小児疾患診療のための病態生理 3	東京医学社	東京	2022	84-91

坪井一哉	QOL、疼痛に対する効果	衛藤義勝、大橋十也	ファブリー病 Update改訂版2	診断と治療社	東京	2021	210-212
坪井一哉	皮膚、自立神経症状に対する効果	衛藤義勝、大橋十也	ファブリー病 Update改訂版2	診断と治療社	東京	2021	213-215
坪井一哉	ファブリー病	鈴木則宏 永田栄一郎、 伊藤義彰	脳神経内科学レビュー2022-2023	総合医学社	東京	2022	354-360
坪井一哉	ライソゾーム病の治療とマネジメント	鈴木則宏、荒木信夫、宇川義一、桑原聡、塩川芳昭	Annual Review of Neurology 2022	中外医学社	東京	2022	233-239
下澤伸行	副腎白質ジストロフィー	下畑亨良	脳神経内科診断ハンドブック	中外医学社	東京	2022	
加我牧子	知的障害	松原康雄	社会福祉学習双書第14巻	全国社会福祉協議会	東京	2023	182-185
Kaga M	Normalization and deterioration of auditory brainstem response (ABR) in child neurology	Kaga K	ABRs and Electrically Evoked ABRs in Children	Springer,	Japan	2022	81-168
研究班診療ガイドライン作成委員会		日本先天代謝異常学会	ニーマンピック病診療ガイドライン2023	診断と治療社	東京	2023	1-71

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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
Tsurumi M, Eto Y. Et al	A survey on the patient journey in Fabry disease in Japan.	Mol. Genet. Metab. Rep	17	100909	2022
Hosseini AM Eto Y.et al.	Generation and characterization of motor neuron progenitors and motor neurons using metachromatic leukodystrophy-induced pluripotent stem cells.	Mol. Genet. Metab. Rep.	31	100852	2022
高橋 勉	ライソゾーム病における低分子療法	小児科診療	84	1803-1808	2021

村山 圭	序—ミトコンドリア病の概念と変遷	小児内科	54巻4号	539 - 543	2022
村山 圭	臨床編 新生児期発症のミトコンドリア病	医学のあゆみ	282巻5号	391-398	2022
松川敬志	【代謝性疾患と神経内科】副腎白質ジストロフィー	脳神経内科	97巻3号	320-323	2022
González-Cuesta M, Herrera-González I, García-Moreno MI, Ashmus RA, Vocadlo DJ, García Fernández JM, Nanba E, Higaki K*, Ortiz Mellet C	sp2-Iminosugars targeting human lysosomal β -hexosaminidase as pharmacological chaperone candidates for late-onset Tay-Sachs disease.	J Enzyme Inhib Med Chem	37(1)	1364-1374	2022
Yamada Y, Miwa T, Nakashima M, Shirakawa A, Ishii A, Namiba N, Kondo Y, Takeo T, Nakagata N, Motoyama K, Higashi T, Arima H, Kurauchi Y, Seki T, Katsuki H, Okada Y, Ichikawa A, Higaki K, Hayashikawa N, Ikeda R, Ishikawa Y, Kajii T, Tachii K, Takeda H, Orita Y, Matsuo M, Irie T, Ishitsuka Y	Fine-tuned cholesterol solubilizer, mono-6-O- α -D-maltosyl- γ -cyclodextrin, ameliorates experimental Niemann-Pick disease type C without hearing loss.	Biomed Pharmacother	155	113698	2022
Okada BY, Kuroiwa S, Noi A, Tanaka A, Nishikawa J, Kondo Y, Ishitsuka Y, Irie T, Higaki K, Matsuo M, Ichikawa A.	Effects of 6-O- α -maltosyl- β cyclodextrin on lipid metabolism in Npc1-deficient Chinese hamster ovary cells.	Mol Genet Metab	137(3)	239-248	2022
坪井一哉	ファブリー病 子供たちの叫びが聞こえますか？	月刊母子保健	11月号	10-11	2021
坪井一哉, 神崎保	ファブリー病の皮膚病変	BRAIN and NERVE	71(4)	354-359	2019
Watanabe T, Suzuki A, Ohira S, Go S, Ishizuka Y, Moriya T, Miyaji Y, Nakatsuka T, Hirata K, Nagai A, Matsuda J	The Urinary Bladder is Rich in Glycosphingolipids Composed of Phytoceramides.	J Lipid Res.	63 (12)	100303	2022
松田純子	代謝性疾患と神経内科 シアリドーシス・ガラクトシアリドーシス	脳神経内科	97 (3)	304-311	2022

Suzuki A, Silsirivanit A, Watanabe T, Matsuda J , Inamori K, Inokuchi J	Mass Spectrometry of Neutral Glycosphingolipids	Methods Mol Biol	2613	127-144	2023
Ota A, Morita H, Naganuma T, Miyamoto K, Matsuda J , Kohara A	Bifunctional DEGS2 has higher hydroxylase activity toward substrates with very-long-chain fatty acids in the production of phytosphingosine-ceramides	J Biol Chem.	doi: 10.1016/j.jbc.2023.104603. Online ahead of print.	104603	2023
Takashima S., Fujita H., Toyoshi K., Ohba A., Hirata Y., Shimozawa N., Oh-hashii K	Hypomorphic mutation of PEX3 with peroxisomal mosaicism reveals the oscillating nature of peroxisome biogenesis coupled with differential metabolic activities	Molecular Genetics and Metabolism	137	68-80	2022
Kato K., Yabe H., Shimozawa N., Adachi S., Kurokawa M., Hashii Y., Sato A., Yoshida N., Kaga M., Onodera O., Kato S., Atsuta Y., Morio T	Stem cell transplantation for pediatric patients with adrenoleukodystrophy:	A nationwide retrospective analysis in Japan Pediatric Transplantation	26	e14125	2022
Ikeda T., Kawahara Y., Miyauchi A., Niijima H., Furukawa R., Shimozawa N., Morimoto A., Osaka H., Yamagata T	Low donor chimerism may be sufficient to prevent demyelination in adrenoleukodystrophy	JIMD Reports	63	19-24	2022
Kawai H, Takashima S, Ohba A, Toyoshi K, Kubota K, Ohnishi H, Shimozawa N	Development of a system adapted for the diagnosis and evaluation of peroxisomal disorders by measuring bile acid intermediates	Brain Dev	45(1)	58-69	2023
下澤伸行	ペルオキシソーム病	小児内科	54、2022増刊号	143-153	2022
Sera Y., Sadoya M., Ichinose T., Matsuyama S., Imanaka T., and Yamaguchi M.	SBDS interacts with RNF2 and is degraded through RNF2-dependent ubiquitination.	Biochem Biophys Res Commun	598	119-123	2022
Kawaguchi K., and Imanaka T.	Substrate specificity and the direction of transport in the ABC transporters ABCD1-3 and ABCD4.	Chem Pharm Bull	70	533-539	2022

Anan I, Sakuma T, Fukuro E, Morimoto S, Nojiri A, Kawai M, Sakurai K, Kobayashi M, Kobayashi H, Ida H, Ohashi T, Yoshimura M, Eto Y, Hong K.	The role of native T1 values on the evaluation of cardiac manifestation in Japanese Fabry disease patients	Mol Genet Metab Rep.	16;31	100858	2022
小林博司.	特集 今知っておきたいゲノム医療と遺伝子治療～基礎から臨床まで	小児内科	54 (2)	343-353	2022
角皆季樹、小林博司.	特集I 代謝疾患と神経内科 GM1/GM2ガングリオシドーシス	脳神経内科	97 (3)	312-319	2022
小林博司	特集 遺伝性神経筋疾患—診断と研究の最前線 ポンペ病の病態, 診断, 治療法の開発	医学の歩み	283 (10)	1006-1015	2022
加我牧子	医療と福祉のあいだ	Support	70(1)	17-20	2023
加我牧子	学童思春期の子どもの自殺をめぐって	東京小児科医会報	41(2)	21-7	2022
Mori-Yoshimura M, Aizawa K, Oya Y, Saito Y, Fukuda T, Sugie H, Nishino I, Takahashi Y.	A 78-year-old Japanese male with late-onset PHKA1-associated distal myopathy: Case report and literature review	Neuromuscul Disord.	32	769-773	2022
Baba K, Fukuda T, Furuta M, Tada S, Imai A, Asano Y, Sugie H, P Takahashi M, Mochizuki H.	A Mild Clinical Phenotype with Myopathic and Hemolytic Forms of Phosphoglycerate Kinase Deficiency (PGK Osaka): A Case Report and Literature Review	Intern Med.	61	3589-3594.	2022
Saito Y, Nakamura K, Fukuda T, Sugie H, Hayashi S, Noguchi S, Nishino I.	Muscle biochemical and pathological diagnosis in Pompe disease.	J Neurol Neurosurg Psychiatry.	Online ahead of print.		2022

伊藤あかね, 平出 拓也, 古澤有花子, 松本由里香, 河崎 知子, 鶴井 聡, 才津 浩智, 緒方 勤, 福田冬季子	c.116G>A, p. (Arg39His) ホモ接合性バリエントが同定された一過性眼振を伴う遊離シアル酸蓄積症の姉弟例	浜松医科大学小児科学雑誌	3	36-44	2023
Sawada T, Kido J, Sugawara K, Yoshida S, Matsumoto S, Shimazu T, Matsushita Y, Inoue T, Hirose S, Endo F, Nakamura K	Newborn screening for Gaucher disease in Japan	Mol Genet Metab Rep	31	100850	2022
Saito Y., Nakamura K., Fukuda T., Sugie H., Hayashi S., Noguchi S., Nishino I	Muscle biochemical and pathological diagnosis in Pompe disease	Journal of Neurology, Neurosurgery and Psychiatry	93	1142-1145	2022
Shimizu M., Fujii H., Kono K., Watanabe K., Goto S., Nozu K., Nakamura K., Nishi S.	Screening for Fabry disease among male patients on hemodialysis in Awaji Island	Therapeutic Apheresis and Dialysis	10.1111/2022 Mar 8	1744-9987.13834	2022
Imasawa, T., Murayama, K., Sawada, T., Takayanagi, M., Nakamura, K	High-risk screening for Fabry disease in hemodialysis patients in Chiba Prefecture	Japan Clinical and Experimental Nephrology	27(3)	288-294	2023
Yanagisawa R, Hirakawa T, Doki N, Ikegame K, Matsuoka KI, Fukuda T, Nakamae H, Ota S, Hiramoto N, Ishikawa J, Ara T, Tanaka M, Koga Y, Kawakita T, Maruyama Y, Kanda Y, Hino M, Atsuta Y, Yabe H, Tsukada N.	Severe short-term adverse events in related bone marrow or peripheral blood stem cell donors.	Int J Hematol.	117(3)	421-427	2023
Toyama D, Koganesawa M, Akiyama K, Yabe H, Yamamoto S	Invasive Pulmonary Aspergillosis Successfully Treated with Granulocyte Transfusions Followed by Hematopoietic Stem Cell Transplantation in a Patient with Severe Childhood Aplastic Anemia	Tokai J Exp Clin Med	47(3)	136-138	2022

Ishida Y, Kamibeppu K, Sato A, Inoue M, Hayakawa A, Shiobara M, <u>Yabe H</u> , Koike K, Adachi S, Yamashita T, Kanda Y, Okamoto S, Atsuta Y	Karnofsky performance status and visual analogue scale scores are simple indicators for quality of life in long-term AYA survivors who received allogeneic hematopoietic stem cell transplantation in childhood	Int J Hematol	116(5)	787-797	2022
Kanda Y, Doki N, Kojima M, Kako S, Inoue M, Uchida N, Onishi Y, Kamata R, Kotaki M, Kobayashi R, Tanaka J, Fukuda T, Fujii N, Miyamura K, Mori SI, Mori Y, Morishima Y, <u>Yabe H</u> , Atsuta Y, Kodera Y	Cryopreservation in Unrelated Bone Marrow and Peripheral Blood Stem Cell Transplantation in the Era of the COVID-19 Pandemic: An Update from the Japan Marrow Donor Program	Transplant Cell Ther	28(10)	677.e1-677.e6.	2022
Murakami T, Hamada M, Odagiri K, Koike T, <u>Yabe H</u>	A Case of Intratemporal Rhabdomyosarcoma in a Child Presenting with VII th , IX th , and X th Cranial Nerve Paralysis	Tokai J Exp Clin Med	47(2)	85-89	2022
<u>Yabe H</u>	Allogeneic hematopoietic stem cell transplantation for inherited metabolic disorders	Int J Hematol	116(1)	28-40	2022
Tsumanuma R, Omoto E, Kumagai H, Katayama Y, Iwato K, Aoki G, Sato Y, Tsutsumi Y, Tsukada N, Iino M, Atsuta Y, Kodera Y, Okamoto S, <u>Yabe H</u> .	The safety and efficacy of hematopoietic stem cell mobilization using biosimilar filgrastim in related donors	Int J Hematol	115(6)	882-889	2022
Miyamoto S, Umeda K, Kurata M, Yanagimachi M, Iguchi A, Sasahara Y, Okada K, Koike T, Tanoshima R, Ishimura M, Yamada M, Sato M, Takahashi Y, Kajiwara M, Kawaguchi H, Inoue M, Hashii Y, <u>Yabe H</u> , Kato K, Atsuta Y, Imai K, Morio T	Hematopoietic Cell Transplantation for Inborn Errors of Immunity Other than Severe Combined Immunity deficiency in Japan: Retrospective Analysis for 1985-2016	J Clin Immunol	42(3)	529-545	2022

Tani H, Sadahiro T, Yamada Y, Isomi M, Yamakawa H, Fujita R, Abe Y, Akiyama T, Nakano K, Kuze Y, Seki M, Suzuki Y, Fujisawa M, Sakata-Yanagimoto M, Chiba S, Fukuda K, Ieda M.	Direct Reprogramming Improves Cardiac Function and Reverses Fibrinolytic Dysfunction in Chronic Myocardial Infarction.	Circulation	2023 Jan 17;147(3)	223-238	2022
Arai T, Kanazawa H, Kimura K, Munakata M, Yamakawa H, Shinmura K, Yuasa S, Sano M, Fukuda K.	Upregulation of neuropeptide Y in cardiac sympathetic nerves induces stress (Takotsubo) cardiomyopathy.	Front Neurosci	2022 Nov 3;16:1013712		2022
Ito S, Hashimoto H, Yamakawa H, Kusumoto D, Akiba Y, Nakamura T, Momoi M, Komuro J, Katsuki T, Kimura M, Kishino Y, Kashimura S, Kunitomi A, Lachmann M, Shimojima M, Yozu G, Motoda C, Seki T, Yamamoto T, Shinya Y, Hiraide T, Kataoka M, Kawakami T, Suzuki K, Ito K, Yada H, Abe M, Osaka M, Tsuru H, Yoshida M, Sakimura K, Fukumoto Y, Yuzaki M, Fukuda K, Yuasa S.	The complement C3-coplement factor D-C3a receptor signalling axis regulates cardiac remodeling in right ventricular failure.	Nat Commun	2022 Sep 15;13(1)		2022
山川 裕之	【心筋症診療のフロントライン-概念から最新の治療まで】心肥大と出会ったら 肥大を呈する二次性心筋症 心Fabry病	循環器ジャーナル	70巻1号	126-136	2022
Uryu H, Migita O, Ozawa M, Kamiyo C, Aoto S, Okamura K, Hasegawa F, Okuyama T, Kosuga M, Hata K.	Automated urinary sediment detection for Fabry disease using deep-learning algorithms.	Mol Genet Metab Rep.	33	100921	2022

Watanabe S, Lei M, Nakagawa E, Takeshita E, Inamori KI, Shishido F, Sasaki M, Mitsuhashi S, Matsudamoto N, Kimura Y, Iwasaki M, Takahashi Y, Mizusawa H, Migita O, Ohno I, Inokuchi JI.	Neurological insights on two siblings with GM3 synthase deficiency due to novel compound heterozygous ST3GAL5 variants.	Brain Dev.	45(5)	270-277	2023
Hori A, Migita O, Isogawa N, Takada F, Hata K.	A novel TP63 variant in a patient with ankyloblepharon-ectodermal defect-cleft lip/palate syndrome and Rapp-Hodgkin syndrome-like ectodermal dysplasia. Hum	Genome Var.	9(1)	17	2022
本吉 愛, 久我 亜沙美, 鈴木 由妃, 杉下 陽堂, 右田 王介, 津川 浩一郎	包括的がんゲノムプロファイリング検査で抽出される二次的所見に関する実態調査 単施設の遡及的観察研究	遺伝性腫瘍	22(2)	36-40	2022
Oshina K, Kamei Y, Hori A, Hasegawa F, Taniguchi K, Migita O, Itakura A, Hata K	A novel FLNA variant in a fetus with skeletal dysplasia	Human Genome Var.	9(1)	45	2022
Hattori A, Okuyama T, So T, Kosuga M, Ichimoto K, Murayama K, Kagami M, Fukami M, Fukuhara Y.	Maternal uniparental disomy of chromosome 7r. underlying argininosuccinic aciduria and Silver-Russell syndrome.	Hum Genome Var.	9(1)	32	2022