

.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
衛藤義勝 編集	ライソゾーム病 のすべて	衛藤義勝	別冊 医学のあゆみ	医歯薬出版	東京	2019年	1-151
衛藤義勝 編集	特集 ライソゾ ーム病—最新情 報と将来展望	衛藤義勝	日本臨床 特集	日本臨床社	東京	2019年	1-1384
衛藤義勝 (厚労省 研究班)	-	日本先天代 謝異常学会	ポンベ病診療 ガイドライン 2018	診断と治療 社	東京	2018年	1-80
衛藤義勝 (厚労省 研究班)	-	日本先天代 謝異常学会	ムコ多糖症 (MPS)Ⅱ型診 療ガイドライ ン2019	診断と治療 社	東京	2019年	1-40
衛藤義勝 (厚労省 研究班)	-	日本先天代 謝異常学会	副腎白質ジス トロフィー (ALD)診療ガ イドライン 2019	診断と治療 社	東京	2019年	1-64
衛藤義勝 (厚労省 研究班)	-	日本先天代 謝異常学会	シスチノーシ ス(シスチン蓄 積症)診療ガイ ドライン2019	診断と治療 社	東京	2019年	1-80

雑誌

衛藤義勝

1) Maekawa M, Jinnoh I, Matsumoto Y, Narita A, Mashima R, Takahashi H, Iwahori A, Saigusa D, Fujii K, Abe A, Higaki K, Yamauchi S, Ozeki Y, Shimoda K, Tomioka Y, Okuyama T, **Eto Y**, Ohno K, T Clayton P, Yamaguchi H, Mano N. Structural Determination of Lysosphingomyelin-509 and Discovery of Novel Class Lipids from Patients with Niemann-Pick Disease Type C. *Int J Mol Sci.* Oct 10;20(20), 2019.

2) Nojiri A, Anan I, Morimoto S, Kawai M, Sakuma T, Kobayashi M, Kobayashi H, Ida H, Ohashi T, **Eto Y**, Shibata T, Yoshimura M, Hongo K. Clinical findings of gadolinium-enhanced cardiac magnetic resonance in Fabry patients. *J Cardiol.* Oct 15. pii: S0914-5087(19)30292-8. 2019.

3) Hossain MA, Wu C, Yanagisawa H, Miyajima T, Akiyama K, **Eto Y**. Future clinical and biochemical predictions of Fabry disease in females by methylation studies of the GLA gene. *Mol Genet Metab Rep.* Jul 24;20:100497, 2019.

4) Akyol MU, Alden TD, Amartino H, Ashworth J, Belani K, Berger KI, Borgo A, Braunlin E, **Eto Y**, Gold JJ, Jester A, Jones SA, Karsli C, Mackenzie W, Marinho DR, McFadyen A, McGill J, Mitchell JJ, Muenzer J, Okuyama T, Orchard PJ, Stevens B, Thomas S, Walker R, Wynn R, Giugliani R, Harmatz P, Hendriksz C, Scarpa M; Recommendations for the management of MPS IVA: systematic evidence- and consensus-based guidance. MPS Consensus Programme Steering Committee; MPS Consensus Programme Co-Chairs. *Orphanet J Rare Dis.* Jun 13;14(1):137. doi: 10.1186/s13023-019-1074-9, 2019

5) Akyol MU, Alden TD, Amartino H, Ashworth J, Belani K, Berger KI, Borgo A, Braunlin E, Eto Y, Gold JI, Jester A, Jones SA, Karsli C, Mackenzie W, Marinho DR, McFadyen A, McGill J, Mitchell JJ, Muenzer J, Okuyama T, Orchard PJ, Stevens B, Thomas S, Walker R, Wynn R, Giugliani R, Harmatz P, Hendriksz C, Scarpa M Recommendations for the management of MPS VI: systematic evidence- and consensus-based guidance. MPS Consensus Programme Steering Committee; MPS Consensus Programme Co-Chairs. Orphanet J Rare Dis. May 29;14(1):118. doi: 10.1186/s13023-019-1080, 20

6) .Maekawa M, Narita A, Jinnoh I, Iida T, Marquardt T, Mengel E, Eto Y, Clayton PT, Yamaguchi H, Mano N. Diagnostic performance evaluation of sulfate-conjugated cholesterol metabolites as urinary biomarkers of Niemann-Pick disease type Clin Chim Acta. Mar 12;494:58-63, 2019.

酒井規夫

- 1) Yagita Y, Sakai N, Miwa K, Ohara N, Tanaka M, Sakaguchi M, Kitagawa K, Mochizuki H., Magnetic Resonance Imaging Findings Related to Stroke Risk in Japanese Patients With Fabry Disease., Stroke. Jul 24;STROKEAHA119025528. doi: 10.1161/STROKEAHA.119.025528, 2019
- 2) Shiraishi H, Yamada K, Oki E, Ishige M, Fukao T, Hamada Y, Sakai N, Ochi F, Watanabe A, Kawakami S, Kuzume K, Watanabe K, Sameshima K, Nakamagoe K, Tamaoka A, Asahina N, Yokoshiki S, Miyakoshi T, Oba K, Ise T, Hayashi H, Yamaguchi S, Sato N., Open-label clinical trial of bezafibrate treatment in patients with fatty acid oxidation disorders in Japan; 2nd report QOL survey., Mol Genet Metab Rep. 2019 Jul 25;20:100496. doi: 10.1016/j.yimgmr.2019.100496. eCollection Sep. PMID:3137234, 2019.
- 3) Sakurai M, Azuma J, Hamada Y, Yamamoto T, Sakai N., Early juvenile Tay-Sachs disease with atypical symptoms., Pediatr Int. Jun;61(6):611-613. doi:10.1111/ped.13848, 2019
- 4) Ichiei Narita, Toya Ohashi, Norio Sakai, Takashi Hamazaki, Nina Skuban, Jeffrey P. Castelli, Hjalmar Lagast & Jay A. Barth, Efficacy and safety of migalastat in a Japanese population: a subgroup analysis of the ATTRACT study, Clinical and Experimental Nephrology 24, 157–166, 2020

高橋 勉

- 1) Kawazoe T, Yamamoto T, Narita A, Ohno K, Adachi K, Nanba E, Noguchi A, Takahashi T, Maekawa M, Eto Y, Ogawa M, Murata M, Takahashi Y. Phenotypic variability of Niemann-Pick disease type C including a case with clinically pure schizophrenia: a case report. *MC Neurol.*, 18, 117, 2018.
- 2) Ota S, Noguchi A, Kondo D, Nakajima Y, Ito T, Arai H, Takahashi T. An early-onset neuronopathic form of acid sphingomyelinase deficiency: A *SMPD1* p.C133Y mutation in the saposin domain of acid sphingomyelinase. *Tohoku J Exp Med.*, 250: 5-11., 2020

高柳正樹

- 1) 高柳 正樹. 先天代謝異常症におけるトランジションの現状と問題点. 外来小児科 vol18:p304-308,2015.
- 2) 高柳 正樹.【小児慢性疾患の成人期移行の現状と問題点】先天性代謝異常 糖原病. 小児科臨床 vol69: p684-688, 2016.

辻 省次

- 1) T Matsukawa, et al. Clinical efficacy of haematopoietic stem cell transplantation for adult adrenoleukodystrophy. *Brain Communications*. 2:fcz048, 2020

鈴木康之

- 1) Orii KE, Lim A, Tomatsu S, Stapleton M, Suzuki Y, Simonaro CM, Schuchman EH, Fukao T, Matsumoto T. Safety Study of Sodium Pentosan Polysulfate for Adult Patients with Mucopolysaccharidosis Type II. *Diagnostics* 9:226doi:10.3390/diagnostics9040226,2019

奥山虎之

ムコ多糖症(MPS)I型診療ガイドライン 2019、監修：厚生労働省難治性疾患等政策研究事業ライソゾーム病(ファブリー病含む)に関する調査研究班、編集：ムコ多糖症(MPS)I型診療ガイドライン作成委員会、診断と治療社、2019

坪井一哉

- 1) 坪井一哉, 神崎保. ファブリー病の皮膚病変. *BRAIN and NERVE* 74(4): 354-359, 2019
- 2) 坪井一哉. *脳神経内科* 91(2): 233-240, 2019

下澤信行

1. 英文著書 (ALD & ペルオキシソーム病関連)

- 1) Imanaka T, Shimozawa N (Eds.), *Peroxisomes: Biogenesis, Function, and Role in Human Disease*, Springer, pp1-279, 2020

2. 英文原著 (ALD & ペルオキシソーム病関連)

- 1) Takashima S, Saito H, Shimozawa N. Expanding the concept of peroxisomal diseases and efficient diagnostic system in Japan. *J Hum Genet* 64, 145-152 (2019)
- 2) Zaabi NA, Kendi A, Al-Jasmi F, Takashima S, Shimozawa N, Al-Dirbashi OY: Atypical PEX16 peroxisome biogenesis disorder with mild biochemical disruptions and long survival. *Brain Dev* 41, 57-65 (2019)
- 3) Sakurai K, Ohashi T, Shimozawa N, Seo JH, Okuyama T, Ida H. Characteristics of Japanese patients with X-linked adrenoleukodystrophy and concerns of their families from the 1st registry system. *Brain Dev* 41: 50-56 (2019)
- 4) 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)
- 4) H, Amano N, Tanaka K, Katsuki T, Adachi T, Shimozawa N, Kawai T. A 29-year patient with adrenoleukodystrophy presenting with Addison's disease. *Endocr J.* doi: 10.1507/endocrj.EJ19-0576 (2020)
- 5) Hama K, Fujiwara Y, Takashima S, Hayashi Y, Yamashita A, Shimozawa N, Yokoyama K. Hexacosenoyl-CoA is the most abundant very long-chain acyl-CoA in ATP-binding cassette transporter D1-deficient cells. *J Lipid Res pii: jlr.P119000325* (2020)

3. 和文著書

- 1) 下澤伸行：副腎白質ジストロフィー・ペルオキシソーム病と遺伝カウンセリング 遺伝子医学 MOOK 別冊シリーズ 4 最新小児・周産期遺伝医学研究と遺伝カウンセリング pp195-200.メディカルドウ.大阪. 2019年11月
- 2) 日本先天代謝異常学会編：副腎白質ジストロフィー(ALD)診療ガイドライン 2019 p1-46.診断と治療社.東京.2019年11月

今中常雄

1. 日本先天代謝異常学会：副腎白質ジストロフィー(ALD)診療ガイドライン 2019
2. ムコ多糖症(MPS)I型診療ガイドライン作成委員会：ムコ多糖症(MPS)I型診療ガイドライン 2019
3. Imanaka, T., Shimozawa, N. eds.: *Peroxisomes: Biogenesis, Function, and Role in Human Disease*. Springer Nature 2020

小林博司

副腎白質ジストロフィー(ALD)診療ガイドライン2019 日本先天代謝異常学会編集、第1版発行 診断と治療社、2019

加我牧子

- 1) Kaga M. Neurophysiology and Neuropsychology in ALD. In Imanaka T, Shimozawa N(ed.): *Peroxisomes: Biogenesis, Function, and Role in Human Disease*. Springer, Nature, 261-79, 2019.
- 2) Kato K et al. Allogeneic stem cell transplantation with reduced intensity conditioning for patients with ALD. *Molec Gen Metabol Rep* 18:1-6, 2019

横山和明

- 1) Hexacosenoyl-CoA is the most abundant very long-chain acyl-CoA in ATP-binding cassette transporter D1-deficient cells. Hama K., Yokoyama K. et al. J Lipid Res. 2020, doi: 10.1194/jlr.P119000325, PMID: 32075856
- 2) Mass spectrometry in combination with a chiral column and multichannel-MRM allows comprehensive analysis of glycosphingolipid molecular species from mouse brain. Fujiwara Y., Yokoyama K. et al. Carbohydr Res. 2020, doi: 10.1016/j.carres.2020.107959, PMID: 32120021
- 3) Lipidomics of Peroxisomal Disorders, Chapter 11 p249-260, in Peroxisomes: Biogenesis, Function, and Role in Human Disease, Hama K., Yokoyama K. et al. Springer, 2019

渡邊順子

1. 論文発表
個別に発表は行っていない

石垣景子

1. 論文発表
個別に発表は行っていない。

成田 綾

1. 論文発表なし

井田博幸

- 1) Kobayashi M, Ohashi T, Kaneshiro E, Higuchi T, Ida H. Mutation spectrum of α -galactosidase A gene in Japanese patients with Fabry disease. J Hum Genet. Jul ; 64(7) : 695-699, 2019
- 2) Nojiri A, Anan I, Morimoto S, Kawai M, Sakuma T, Kobayashi M, Kobayashi H, Ida H, Ohashi T, Eto Y, Shibata T, Yoshimura M, Hongo K. Clinical findings of gadolinium-enhanced cardiac magnetic resonance in Fabry patients. J Cardiol. Jan ; 75(1) : 27-33, 2020
- 3) Hiroyuki Ida : Gaucher Disease , Human Pathobiochemistry eds. Oohashi T et al , Springer , : p57-66, 2019
- 4) 井田博幸：酵素補充療法の有効性と限界，日本臨床，日本臨床社，77；1326-1331, 2019

大橋十也

当該論文 なし

小林正之

- 1) Kobayashi M, Ohashi T, Kaneshiro E, Higuchi T, Ida H. Mutation spectrum of α -galactosidase A gene in Japanese patients with Fabry disease. J Hum Genet. 2019; 64: 695-699
- 2) Hongo K, Ito K, Date T, Anan I, Inoue Y, Morimoto S, Ogawa K, Kawai M, Kobayashi H, Kobayashi M, Ida H, Ohashi T, Taniguchi I, Yoshimura M, Eto Y. The beneficial effects of long-term enzyme replacement therapy on cardiac involvement in Japanese Fabry patients. Mol Genet Metab. 2018; 142: 143-151
- 3) Nojiri A, Anan I, Morimoto S, Kawai M, Sakuma T, Kobayashi M, Kobayashi H, Ida H, Ohashi T, Eto Y, Shibata T, Yoshimura M, Hongo K. Clinical findings of gadolinium-enhanced cardiac magnetic resonance in Fabry patients. J Cardiol. 2020; 75: 27-33

福田冬季子

- 1) 平野 恵子, 遠藤 彰, 白井 眞美, 福田 冬季子, 松林 朋子 酵素補充療法中に腸間膜リンパ節の石灰化と難聴を呈した1型Gaucher病、日本小児科学会雑誌 123, 1673-1680, 2019.
- 2) 福田 冬季子 特集 小児の負荷試験2019 筋型糖原病鑑別のための負荷試験 小児内科 51, 506-508, 2019
- 3) 福田 冬季子 脂肪酸代謝異常症 治療可能な神経疾患診断治療の手引き 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 編集 診断と治療社 2020 . 38-41
- 4) 福田 冬季子 アミノ酸代謝異常症 治療可能な神経疾患診断治療の手引き 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 編集 診断と治療社 2020 . 46-50
- 5) 福田 冬季子 神経筋疾患新たな治療の時代へ 各疾患の治療の現代 筋型糖原病 小児科診療 83, 87-92, 2020.01
- 6) Ago Y, Sugie H, Fukuda T, Otsuka H, Sasai H, Nakama M, Abdelkreem E, Fukao T.A

rare PHKA2 variant (p.G991A) identified in a patient with ketotic hypoglycemia. JIMD Rep. 2019 28;48:15-18.

- 6) 平野 恵子, 遠藤 彰, 白井 眞美, 福田 冬季子, 松林 朋子 酵素補充療法中に腸間膜リンパ節の石灰化と難聴を呈した1型Gaucher病、日本小児科学会雑誌 123, 1673-1680, 2019.
- 7) 福田 冬季子 特集 小児の負荷試験2019 筋型糖原病鑑別のための負荷試験 小児内科 51, 506-508, 2019
- 8) 福田 冬季子 神経筋疾患新たな治療の時代へ 各疾患の治療の現代 筋型糖原病 小児科診療 83, 87-92, 2020.01

中村公俊

- 1) Momosaki K, Kido J, Yoshida S, Sugawara K, Miyamoto T, Inoue T, Okumiya T, Matsumoto S, Endo F, Hirose S, and Nakamura D Newborn screening for Pompe disease in Japan: report and literature review of mutations in the GAA gene in Japanese and Asian patients. J Hum Genet. Aug;64(8):741-755. doi: 10.1038/s10038-019-0603-7, 2019.
- 2) Newborn screening for Fabry disease in the western region of Japan. Sawada T, Kido J, Yoshida S, Sugawara K, Momosaki K, Inoue T, Tajima G, Sawada H, Mastumoto S, Endo F, Hirose S, Nakamura K. Mol Genet Metab Rep. 2020 Jan 11;22:100562. doi: 10.1016/j.ymgmr. 2019.100562. eCollection 2020

浜崎孝史

- 1) Narita I, Ohashi T, Sakai N, Hamazaki T, et.al. Efficacy and safety of migalastat in a Japanese population: a subgroup analysis of the ATTRACT study. CLINICAL AND EXPERIMENTAL NEPHROLOGY 24 巻 2 号 157-166 2020 年 2 月 (査読有り, 招待無し)
- 2) Okuyama T, Seo Joo-Hyun, Kosuga M, Shintaku H, Hamazaki T. Successful prevention and stabilization of cognitive decline in Japanese patients with neuronopathic mucopolysaccharidosis type II treated by intracerebroventricular enzyme replacement therapy: Results of the Phase clinical trial for two years. MOLECULAR GENETICS AND METABOLISM 129 巻 2 号 S120 2020 年 2 月 (査読有り, 招待無し)
- 3) Maeda M, Seto T, Kadono C, Morimoto H, Kida S, Suga M, Nakamura M, Kataoka Y, Hamazaki T, Shintaku H. Autophagy in the Central Nervous System and Effects of Chloroquine in Mucopolysaccharidosis Type II Mice. INTERNATIONAL JOURNAL OF MOLECULAR SCIENCES 20 巻 23 号 2019 年 12 月 (査読有り, 招待無し)
- 4) 濱崎孝史. ライソゾーム病-最新情報と将来展望-治療の最新情報 クリニカルクエスチョン(CQ)低分子治療薬 シャペロン治療の治療対象患者と有効性. 日本臨床 77 巻 8 号 1338-1343 2019 年 8 月

秋山けい子

- 1) Hossain MA, Wu C, Yanagisawa H, Miyajima T, Akiyama K, Eto Y. Future clinical and biochemical predictions of Fabry disease in females by methylation studies of the *GLA* gene. Mol Genet Metab Rep. 2019 Jul;20:100497.

矢部普正

1. Yabe H, Tabuchi K, Uchida N, Takahashi S, Onishi Y, Aotsuka N, Sugio Y, Ikegame K, Ichinohe T, Takanashi M, Kato K, Atsuta Y, Kanda Y. Could the minimum number of hematopoietic stem cells to obtain engraftment exist in unrelated, single cord blood transplantation? Br J Haematol. 2020 Feb 28. doi: 10.1111/bjh.16465. [Epub ahead of print] No abstract available. PMID: 32108331
2. Yamazaki N, Kosuga M, Kida K, Takei G, Fukuhara Y, Matsumoto H, Senda M, Honda A, Ishiguro A, Koike T, Yabe H, Okuyama T. Early enzyme replacement therapy enables a successful hematopoietic stem cell transplantation in mucopolysaccharidosis type IH: Divergent clinical outcomes in two Japanese siblings. Brain and development 2019 (in press)
3. Donovan FX, Solanki A, Mori M, Chavan N, George M, Kumar C S, Okuno Y, Muramatsu H, Yoshida K, Shimamoto A, Takaori-Kondo A, Yabe H, Ogawa S, Kojima S, Yabe M, Ramanagoudr-Bhojappa R, Smogorzewska A, Mohan S, Rajendran A, Auerbach AD, Takata M, Chandrasekharappa SC, Vundinti BR. A founder variant in the South Asian population leads to a high prevalence of FANCL Fanconi anemia cases in India. Hum Mutat. 2019 Sep

12. doi: 10.1002/humu.23914. [Epub ahead of print]
4. Hyakuna N, Hashii Y, Ishida H, Umeda K, Takahashi Y, Nagasawa M, Yabe H, Nakazawa Y, Koh K, Goto H, Fujisaki H, Matsumoto K, Kakuda H, Yano M, Tawa A, Tomizawa D, Taga T, Adachi S, Kato K. Retrospective analysis of children with high-risk acute myeloid leukemia who underwent allogeneic hematopoietic stem cell transplantation following complete remission with initial induction chemotherapy in the AML-05 clinical trial. *Pediatr Blood Cancer*. 2019 Oct;66(10):e27875. doi: 10.1002/pbc.27875. Epub 2019 Jul 16.
 5. Iguchi A, Cho Y, Yabe H, Kato S, Kato K, Hara J, Koh K, Takita J, Ishihara T, Inoue M, Imai K, Nakayama H, Hashii Y, Morimoto A, Atsuta Y, Morio T; Hereditary disorder Working Group of the Japan Society for Hematopoietic Cell Transplantation. Long-term outcome and chimerism in patients with Wiskott-Aldrich syndrome treated by hematopoietic cell transplantation: a retrospective nationwide survey. *Int J Hematol*. 2019 Jun 11. doi: 10.1007/s12185-019-02686-y. [Epub ahead of print] PMID: 31187438
 6. Matsuda M, Ono R, Iyoda T, Endo T, Iwasaki M, Tomizawa-Murasawa M, Saito Y, Kaneko A, Shimizu K, Yamada D, Ogonuki N, Watanabe T, Nakayama M, Koseki Y, Kezuka-Shiotani F, Hasegawa T, Yabe H, Kato S, Ogura A, Shultz LD, Ohara O, Taniguchi M, Koseki H, Fujii SI, Ishikawa F. Human NK cell development in hIL-7 and hIL-15 knockin NOD/SCID/IL2rgKO mice. *Life Sci Alliance*. 2019 Apr 1;2(2). pii: e201800195. doi: 10.26508/lsa.201800195. Print 2019 Apr. PMID: 30936185
 7. Mori M, Hira A, Yoshida K, Muramatsu H, Okuno Y, Shiraishi Y, Anmae M, Yasuda J, Tadaka S, Kinoshita K, Osumi T, Noguchi Y, Adachi S, Kobayashi R, Kawabata H, Imai K, Morio T, Tamura K, Takaori-Kondo A, Yamamoto M, Miyano S, Kojima S, Ito E, Ogawa S, Matsuo K, Yabe H, Yabe M, Takata M. Pathogenic mutations identified by a multimodality approach in 117 Japanese Fanconi anemia patients. *Haematologica*. 2019; 104(10): 1962-1973.
 8. Taylor M, Khan S, Stapleton M, Wang J, Chen J, Wynn R, Yabe H, Chinen Y, Boelens JJ, Mason RW, Kubaski F, Horovitz DDG, Barth AL, Serafini M, Bernardo ME, Kobayashi H, Orii KE, Suzuki Y, Orii T, Tomatsu S. Hematopoietic Stem Cell Transplantation for Mucopolysaccharidoses: Past, Present, and Future. *Biol Blood Marrow Transplant*. 2019 Feb 14. pii: S1083-8791(19)30137-5. doi: 10.1016/j.bbmt.2019.02.012. [Epub ahead of print] Review.
 9. Ono R, Watanabe T, Kawakami E, Iwasaki M, Tomizawa-Murasawa M, Matsuda M, Najima Y, Takagi S, Fujiki S, Sato R, Mochizuki Y, Yoshida H, Sato K, Yabe H, Kato S, Saito Y, Taniguchi S, Shultz LD, Ohara O, Amagai M, Koseki H, Ishikawa F. Co-activation of macrophages and T cells contribute to chronic GVHD in human IL-6 transgenic humanised mouse model. *EBioMedicine*. 2019 Mar;41:584-596. doi: 10.1016/j.ebiom.2019.02.001. Epub 2019 Feb 13.
 10. Yamazaki N, Kosuga M, Kida K, Takei G, Fukuhara Y, Matsumoto H, Senda M, Honda A, Ishiguro A, Koike T, Yabe H, Okuyama T. Early enzyme replacement therapy enables a successful hematopoietic stem cell transplantation in mucopolysaccharidosis type IH: Divergent clinical outcomes in two Japanese siblings. *Brain Dev*. 2019 Feb 9. pii: S0387-7604(18)30541-2. doi: 10.1016/j.braindev.2019.01.008. [Epub ahead of print]

著書

1. 指定難病ペディア 2019 ファンコニ貧血 日本医師会 2019 190-191 (共著)