衛藤義勝、井田博幸、大橋十也、小林博司

【論文発表】

1) Takamura A, Sakai N, Shinpoo M, Noguchi A, Takahashi T, Matsuda S, Yamamoto M, Narita A, Ohno K, Ohashi T, Ida H, Eto Y.：The useful preliminary diagnosis of Niemann-Pick disease type C by filipin test in blood smear. Mol Genet Metab. 2013 Nov;110(3):401-4. Epub 2013 Aug 17.
2) Takenori D, Takeo I, Minami M, Masahiro E, Toya O, Yoshikatu E.：A practical fluorometric assay method to measure lysosomal acid lipase activity in dried blood spots for the screening of cholesteryl ester storage disease and Wolman disease. Mol Genet Metab. Available online 16 November 2013
3) Sato Y, Fujiwara M, Kobayashi H, Ida H. Massive accumulation of glycosaminoglycans in the aortic valve of a patient with hunter syndrome during enzyme replacement therapy. Pediatr Cardiol. 2013 Dec;34(8):2077-9.
4) Akiyama K, Shimada Y, Higuchi T, Ohtsu M, Nakauchi H, Kobayashi H, Fukuda T, Ida H, Eto Y, Crawford BE, Brown JR, Ohashi T. Enzyme augmentation therapy enhances the therapeutic efficacy of bone marrow transplantation in mucopolysaccharidosis type II mice. Mol Genet Metab. 2013 Sep 21.
5) Kawagoe S, Higuchi T, Otaka M, Shimada Y, Kobayashi H, Ida H, Ohashi T, Okano HJ, Nakanishi M, Eto Y. Morphological features of iPS cells generated from Fabry disease skin fibroblasts using Sendai virus vector (SeVdp). Mol Genet Metab. 2013 Aug;109(4):386-9.
6) Sato T, Ikeda M, Yotsumoto S, Shimada Y, Higuchi T, Kobayashi H, Fukuda T, Ohashi T, Suda T, Ohteki T. Novel interferon-based pre-transplantation conditioning in the treatment of a congenital metabolic disorder. Blood. 2013 Apr 18;121(16):3267-73.
7) J. Ito, T. Saito, C. Numakura, A. Iwaba, S. Sugahara, R. Ishii, C. Sato, H. Haga, K. Okumoto, Y. Nishise, H. Watanabe, H. Ida, K. Hayasaka, H. Togashi, S. Kawata, Y. Ueno: A Case of Adult Type1 Gaucher Disease Complicated by Temporal Intestinal Hemorrhage. Case Rep Gastroenterol 2013;7:340-346

【学会発表】

1) M Fujisaki, J Matsumoto, A Takamura, T Higuchi, M Furujo, S Kawagoe, H Kobayashi, H Ida, Y Shimada, T Ohashi, T Dairaku, Y Eto Enzymatic Diagnosis of Maroteaux-Lamy disease (MPⅥ) in dried Blood Spots on Filter Paper, The 54th Annual Meeting of The Japanese Society for Inherited Metabolic Diseases (JSIMD), Gifu, Japan, 2012 November 15th – 17th
2) M Fujisaki, A Takamura, T Dairaku, T Ohashi, H Ida, Y Eto：Enzymatic screening in dried blood spots and gene analysis of Mucopolysaccharidosis IVA (MPS IⅣA) in Japanese, 12th International Congress of Inborn Errors of Metabolism, Barcelona, Spain, 2013 September 3rd – 6th
3) M Fujisaki, A Takamura, T Dairaku, T Ohashi, H Ida, Y Eto：Enzymatic screening using dried blood spots and gene analysis of Mucopolysaccharidosis ⅣA (MPS IⅣA) in Japanese The 3rd Asian Congress for Inherited Metabolic Diseases (ACIMD) The 55th Annual Meeting of The Japanese Society for Inherited Metabolic Diseases (JSIMD), Chiba, Japan, 2013 November 27th – 29th
4) Ayumi Takamura, Miwa Fujisaki, Hiroyuki Ida, Toya Ohashi, Yoshikatsu Eto Abnormal Intracellular Membrane Traffic in Juvenile Neural Ceroid Lipofuscinosis, Novel CLN1 Mutated Cases.3rd Asian Congress for Inherited Metabolic Diseases (ACIMD) and the 55th Annual Meeting of the Japanese Society for Inherited Metabolic Diseases (JSIMD), November 27-29, 2013, Chiba, Japan

5) Ayumi Takamura, Norio Sakai, Michiko Shinpo, Masanari Yamamoto, Aya Narita, Kosaku Ohno, Toya Ohashi, Hiroyuki Ida, Yoshikatsu Eto：The Useful Preliminary Diagnosis of Niemann-Pick Disease type C by Filipin Test in Blood Smear 12th International Congress of Inborn Errors of Metabolism (ICIEM), September 3-6, 2013, Barcelona, Spain

6) Eto Y：Immunity of inborn error of metabolism, Asian Society of Pediatric Research, Kuching, Malaysia, 2013.5.10

7) Eto Y：Applications of iPS Cell Technology for the Pathogenesis and Possible Treatment of LSD、International Fabry disease symposium, Hong Kong,June 6, 2013

8) Eto Y：Novel Strategies of the Treatment for Lysosomal Storage disease, Korean Human Genetic Seminar, Seoul, 2013, 11,14-16

9) Eto Y：New Strategies of the Treatment of Lysosomal Storage disease, 13th Asia LSD symposium, Nov.26, 2013

10)Eto Y：Future of clinical and research prospects in inborn error of metabolism, 第3回アジア先天代謝学会、舞浜、東京 2013.11.27-29

田中あけみ

【学会発表】

1) Tanaka A, Maeda M. Kadono C, Morimoto H, Efficacy of oral administration of chloroquine on the neuronal cells of iduronate 2-sulfatatse gene knock-out mouse **第 55 回日本先天代謝異常学会** 2013 **年** 11 **月** 27－29 **日 千葉**

酒井 規夫

【論文発表】

1) Hara M, Inokuchi T, Taniwaki T, Otomo T, Sakai N, Matsuishi T, Yoshino M., An adult patient with mucolipidosis III alpha/beta presenting with parkinsonism., ***Brain Dev***. 35(5):462-5, 2013

2) Tokushige SI, Sonoo T, Maekawa R, Shirota Y, Hanajima R, Terao Y, Matsumoto H, Hossain MA, Sakai N, Shiio Y., Isolated pyramidal tract impairment in the central nervous system of adult-onset Krabbe disease with novel mutations in the GALC gene., ***Brain Dev***. 35(6):579-81, 2013

3) Chang B, Gorbea C, Lezin G, Li L, Shan L, Sakai N, Kogaki S, Otomo T, Okinaga T, Hamaoka A, Yu X, Hata Y, Nishida N, Yost HJ, Bowles NE, Brunelli L, Ichida F., 14-3-3ε Gene variants in a Japanese patient with left ventricular noncompaction and hypoplasia of the corpus callosum., ***Gene***. 515(1):173-80, 2013

4) Kardas F, Uzak AS, Hossain A, Sakai N, Canpolat M, Yıkılmaz A., A novel homozygous GALC mutation: Very early onset and rapidly progressive Krabbe disease., ***Gene***. 517(1):125-7, 2013

5) Eto K, **Sakai** N, Shimada S, Shioda M, Ishigaki K, Hamada Y, Shinpo M, Azuma J, Tominaga K, Shimojima K, Ozono K, Osawa M, Yamamoto T., Microdeletions of 3p21.31 characterized by developmental delay, distinctive features, elevated serum creatine kinase levels, and white matter involvement., ***Am J Med Genet A***. 2013 Dec;161(12):3049-56.

6) Hossain MA, Otomo T, Saito S, Ohno K, Sakuraba H, Hamada Y, Ozono K, **Sakai** N., Late-onset Krabbe disease is predominant in Japan and its mutant precursor protein undergoes more effective processing than the infantile-onset form., ***Gene***. 2013 Nov 16. doi:pii: S0378-1119(13)01515-1. 10.1016/j.gene.2013.11.003.

【学会発表】

1) **酒井規夫、神経遺伝病治療戦略セミナー、ニーマン・ピック病 C 型の診断と治療、第５５回日本小児神経学会学術集会、**2013.5
2) Norio Sakai, Potential effects and obstacles for NBS of metabolic leukodystrophy, including adrenoleukodystrophy, Krabbe and metachromatic leukodystrophy, Asian Congress for Lysosomal Storage Disease Screening, 2013.5
3) **新寶 理子、青天目 信、近藤 秀仁、**Mohammad Arif Hossain**、濱田 悠介、酒井 規夫、大薗 恵一、当科で診断した** GM2 **ガングリオシドーシス症例の検討、第９回近畿先天代謝異常研究会、**2013.6
4) Mohammad Arif Hossain, Michiko Shinpo, Keiichi Ozono, Norio Sakai, Molecular and biochemical diagnosis for three Japanese patients of galactosialidosis, **第９回近畿先天代謝異常研究会、**2013.6
5) **佐藤友紀、金川武司、酒井規夫、望月秀樹、大阪大学医学部附属病院における羊水染色体検査の現状報告、第 37 回日本遺伝カウンセリング学会、**2013.6
6) **酒井規夫、ファブリー病の診療；今患者がもとめるもの、神奈川酵素補充療法研究会、**2013.7
7) **酒井規夫、ムコ多糖症と遺伝カウンセリング、第１回ムコ多糖症フォーラム、**2013.7
8) Hossain MA[1], Higaki K[2], Nanba E[2], Suzuki Y[3], Ozono K[1], Sakai N、CHAPERONE THERAPY FOR KRABBE DISEASE; JAPANESE LATE-ONSET MUTATIONS CAN BE TREATED EFFECTIVELY BY NOEV、ICIEM2013.9
9) Hidehito Kondo[1], Michiko Shinpo[1], Mohammad Arif Hossain[1], Yusuke Hamada[1], Norio Sakai[1], Yoshihiro Asano[2], Takeshi Masaki[2], Tadayasu Togawa[3], Keichi Ozono, A case report of Fabry disease with chronic heart failure treated with amiodarone, ACIMD2013.11
10) Mohammad Arif Hossain[1], Katsumi Higaki[2], Eiji Nanba[2], Yoshiyuki Suzuki[3], Keiichi Ozono[1], Norio Sakai[1] , NOEV treatment option for Japanese Krabbe disease, ACIMD2013.11
11) Mohammad Arif Hossain[1], Michiko Shinpo[1] Katsumi Higaki[2], Eiji Nanba[2], Yoshiyuki Suzuki[3], Keiichi Ozono[1], Norio Sakai[1] , NOEV can stabilize β-galactosidase in galactosialidosis patients' skin fibroblasts effectively and cause normalization of its activity, ACIMD2013.11
12) Michiko Shinpo, Sayaka Nakano, Yusuke Hamada, Kouji Tominaga, Shin Nabatame, Takeshi Okinaga, Yoshiko Hashii, Norio Sakai, Keichi Ozono, Outcomes of hematopoietic stem cell transplantation for three patients with metachromatic leukodystrophy , ACIMD2013.11
13) Yusuke Hamada1, Hidehito Kondo1, Michiko Shinpo 1, Yoshiro Wada2 , Norio Sakai1, Yutaka Sumida2, Kiichi Ozono1, Different clinical course of propionic acidemia in two siblings, ACIMD2013.11
14) Motohiro Akagi1, 2, Mohammad Arif Hossain1, Keiichi Ozono1, Norio Sakai1, Yoshinori Okumura3, Clinicogenetical features of a Japanese patient with static encephalopathy of childhood with neurodegeneration in adulthood (SENDA), ACIMD2013.11
15) N. Sakai1, X.F. Gu2, H. Ida3 on behalf of the EDGE investigators, O. Kawaguchi4, Y. Xue5, A phase 3 study evaluating once versus twice daily dosing of eliglustat in patients with Gaucher disease type 1 (GD1): Interim

results from the lead in period, ACIMD2013.11

16)Akemi Tanaka1, Takashi Hamazaki1, Motomichi Kosuga2. Torayuki Okuyama2, Yasuyuki Suzuki3, Norio Sakai4, Tomo Sawada1, Hiromasa Yabe5, Mika Ishige6, Hideo Mugishima6, Shunichi Kato5, Efficacy of hematopoietic stem cell transplantation versus enzyme replacement therapy on intelligence in the patients with Mucopolysaccharidosis type II, ACIMD2013.11

高橋 勉

【論文発表】

1) Takamura A, Sakai N, Shinpoo M, Noguchi A, Takahashi T**,** Matsuda S, Yamamoto M, Narita A, Ohno K, Ohashi T, Ida H, Eto Y: The useful preliminary diagnosis of Niemann-Pick disease type C by filipin test in blood smear. Mol Genet metab **111:** 401-404, 2013

【著書】

1) **高橋 勉：**Niemann-Pick **病、新領域別症候群シリーズ** No.23**、血液症候群III（第** 2 **版）―その他の血液疾患を含めて―、日本臨床、日本臨床社、**491-75**、**2013.

2) **小山千嘉子、高橋 勉：ニーマンピック病** A, B **型、新領域別症候群シリーズ** No.20**、先天代謝異常症候群（第** 2 **版）下―病因・病態研究、診断治療の進歩―、日本臨床、日本臨床社、**472-75**、**2012.

【学会発表】

1) Hirayama, M., Oyama, C., Noguchi, A., Takahashi, T. Histone deacetylase inhibitors need acid sphingomyelinase to reduce the abnormal storage of cholesterol in Niemann-Pick C1 cells. The 3rd Asian congress for inherited metabolic diseases, Chiba, Japan, Nov. 2013.

高柳 正樹

【論文発表】

1) **井田 博幸**, **衞藤 義勝**, **田中 あけみ**, **高柳 正樹**, **酒井 規夫**, **川合 基司**, **田畑 恭裕。薬剤の臨床日本人** Gaucher **病(**I **型、**II **型および** III **型)患者に対するセレザイムの** 8 **年間の製造販売後調査結果による有効性と安全性の検討。小児科診療** 76**：** 1325-1334**、**2013

難波 栄ニ

【論文発表】

1) X Luan Z, Li L, Higaki K, Nanba E, Suzuki Y, Ohno K, The chaperone activity and toxicity of ambroxol on GD cells and normal mice. Brain Dev, 2013 35: 317-322.

2) Takai T, Higaki K, Aguilar-Moncayo M, Mena-Barragán T, Hirano Y, Yura K, Yu L, Ninomiya H, García-Moreno I, Sakakibara Y, Ohno K, Nanba E, Ortiz Mellet C, García Fernández JM, Suzuki **1** Y, A bicyclic 1-deoxygalactonojirimycin derivative as a novel pharmacological chaperone for GM_1 gangliosidosis. Mol Ther, 2013 21: 526-532.

3) Higaki K, Ninomiya H, Suzuki Y, Nanba E. Two candidate molecules for chemical chaperone therapy for GM1-gangliosidosis. Future Med Chem. 2013 5(13): 1551-1558.

【学会発表】

1) **難波栄二. ライソゾーム病に対するシャペロン療法. 第２回先天代謝異常症患者会フォーラム. 東**

京, 2013. 8

2) **難波栄二**, **檜垣克美**, **髙井知子**, **由良敬**, **榊原康文**, Carmen Ortiz Mellet, Jose M. Garcia Fernandez, **鈴木義之**. □-**ガラクトシダーゼ欠損症に対するシャペロン治療薬の開発**. **第** 58 **回日本人類遺伝学会**. **仙台**, 2013. 11

3) Nanba E. Chaperone therapy for lysosomal storage diseases. The 3rd Asian Congress for Inherited Metabolic Diseases / The 55th Annual Meeting for The Japanese Society for Inherited Metabolic Diseases. Chiba, Nov. 2013

4) Takai T, Higaki K, Suzuki Y, Nanba E. Comparison of two Chaperone candidates for treatment of GM1-gangliosidosis. The 3rd Asian Congress for Inherited Metabolic Diseases / The 55th Annual Meeting for The Japanese Society for Inherited Metabolic Diseases. Chiba, Nov. 2013

5) Hossain MA, Higaki K, Nanba E, Suzuki Y, Ozono K, Sakai N. NOEV treatment option for Japansese late-onset Krabbe disease. The 3rd Asian Congress for Inherited Metabolic Diseases / The 55th Annual Meeting for The Japanese Society for Inherited Metabolic Diseases. Chiba, Nov. 2013

鈴木 康之

【論文発表】

1) Pravin Patel, Yasuyuki Suzuki2, Miho Maeda, Eriko Yasuda, Tsutomu Shimada, Kenji E. Orii , Tadao Orii, Shunji Tomatsu. Growth charts for patients with Hunter Syndrome. Molecular Genetics and Metabolism (in press)

【学会発表】

1) Yasuyuki Suzuki, Kenji Orii, Tadao Orii, Shunji Tomatsu. Overgrowth in infants with Hunter disease: Implication for the early clinical detection. 3rd Asian Congress for Inherited Metabolic Diseases (ACIMD) / 55th Annual Meeting of the Japanese Society for Interited Metabolic Diseases (JSIMD). Nov 27-29, 2013, Maihama, Chiba, Japan

櫻庭 均

【論文発表】

1) Nakano S, Morizane Y, Makisaka N, et al：Development of a highly sensitive immuno-PCR assay for the measurement of α-galactosidase A protein levels in serum and plasma. PLoS ONE 8: e78588, 2013

2) Saito S, Ohno K, Maita N, et al：Structural and clinical implications of amino acid substitutions in α-L-iduronidase：Insight into the basis of mucopolysaccharidosis type I. Mol Genet Metab, in press.

3) Saito S, Ohno K, Sakuraba H：Comparative study of structural changes caused by different substitutions at the same residue on α-galactosidase A. PLoS ONE,in press.

【学会発表】

1) Sakuraba H：High risk screening for Fabry disease. Asian Congress for Lysosomal Storage Disease Screening, Kumamoto, Japan, May. 2013

2) Sakuraba H：E66Q: Biochemical, pathological and structural studies. 3rd Update on Fabry Nephropathy: Biomarkers, Progression and Treatment Opportunities, Hong Kong, China, Jun. 2013

3) Sakuraba H：Genotype/Phenotype correlation in Fabry disease. The 15th Annual Asia LSD Symposium, Chiba, Japan, Nov. 2013

4) Kawashima I, Mitobe S, Kodama T, Tsukimura T, Togawa T, Sakuraba H: Development of enzyme replacement therapy with a modified enzyme and an activator for Fabry disease. The 3rd Asian Congress for Inherited Metabolic Diseases / The 55th Annual Meeting of The Japanese Society for Inherited Metabolic Diseases. Chiba, Japan, Nov. 2013

5) Shibasaki F, Nakano S, Togawa T, Tsukimura T, Kawashima I, Sakuraba H.: Development of a highly sensitive immuno-PCR measurement of α-galactosidase A protein levels in serum and plasma. The 3rd Asian Congress for Inherited Metabolic Diseases / The 55th Annual Meeting of The Japanese Society for Inherited Metabolic Diseases. Chiba, Japan, Nov. 2013

6) Nakano S, Togawa T, Tsukimura T, Kawashima I, Sakuraba H, Futoshi S.: Rapid Immunochromatographic measurement of anti-α-galactosidase A antibodies in Fabry patients Treated with enzyme replacement therapy. The 3rd Asian Congress for Inherited Metabolic Diseases / The 55th Annual Meeting of The Japanese Society for Inherited Metabolic Diseases. Chiba, Japan, Nov. 2013

7) Togawa T, Tsukimura T, Katayama M, Mitobe S, Sakuraba H.: Fabry patients exhibiting no elevation in plasma globotriaosylsphingosine level. The 3rd Asian Congress for Inherited Metabolic Diseases / The 55th Annual Meeting of The Japanese Society for Inherited Metabolic Diseases. Chiba, Japan, Nov. 2013

8) Tsukimura T, Takada M, Aizawa Y, Suzuki T, Katayama M, Sakuraba H, Togawa T.: Comparative study on the content of mannose 6-phosphate residues of recombinant lysosomal enzymes. The 3rd Asian Congress for Inherited Metabolic Diseases / The 55th Annual Meeting of The Japanese Society for Inherited Metabolic Diseases. Chiba, Japan, Nov. 2013

9) Itoh K, Tsuji D, Namba K, Yamaguchi S, Imataki I, Ishimaru N, Sakuraba H.: Establishment of human neural cell culture systems induced from ips cells derived from Tay-Sachs disease patient for drug discovery. The 3rd Asian Congress for Inherited Metabolic Diseases / The 55th Annual Meeting of The Japanese Society for Inherited Metabolic Diseases. Chiba, Japan, Nov. 2013

10)Kitakaze K, Kawano K, Tsuji D, Asanuma D, Kamiya M, Urano Y, Sakuraba H, Itoh K.: Imaging of enzyme replacement with a novel fluorescent probe and purified lysosomal β-hexosaminidase carrying M6P-type glycans. The 3rd Asian Congress for Inherited Metabolic Diseases / The 55th Annual Meeting of The Japanese Society for Inherited Metabolic Diseases. Chiba, Japan, Nov. 2013

11)**櫻庭　均**：**ファブリー病を疑うとき 診断・治療について**, **ファブリー病セミナー** in **新潟**, **新潟**, 2013. 5

12)**櫻庭　均**：**ファブリー病の治療戦略**. **熊本ファブリー病フォーラム**, **熊本**, 2013. 5

13)**櫻庭　均：ファブリー病の新しい治療薬開発に向かって. ふくろうの会　東京シンポジウム** 2013, **東京**, 2013. 6

14)**櫻庭　均：よくわかるシリーズ ファブリー病：ファブリー病の診断法, 第** 58 **回日本透析医学会学術集会・総会, 福岡**,2013. 6

15)**櫻庭　均：ファブリー病の最近の話題. ファブリー病セミナー　腎臓** Special Lecture, **福岡**, 2013. 6

16)**櫻庭　均：心疾患の中に潜在するファブリー病　ファブリー病の病態・診断について. 第** 23 **回** Educational Seminar in Cardiology, **東京**, 2013. 7

17)**櫻庭　均：日常診療に潜在するファブリー病：病態・診断・治療. 福井ファブリー病セミナー, 福井**, 2013. 7

18)**櫻庭　均：ファブリー病の最近の話題. 川口ファブリー病セミナー, 川口** 2013. 7

19)**櫻庭　均：ファブリー病の診断方法と最近の話題. 秋田ファブリー病セミナー, 秋田**, 2013. 8

20)**櫻庭　均：ファブリー病へのアプローチ　診断・治療可能な希少疾患を見逃さないために. 西湘ファブリー病セミナー, 神奈川**, 2013. 10

21)**櫻庭　均：ファブリー病を知ろう　病態・診断・治療. 函館ファブリー病セミナー, 函館**, 2013. 10

22)**櫻庭　均：　ファブリー病　その診断, 治療の核心に迫る. 弘前ファブリー病セミナー, 青森**, 2013. 10

23)**櫻庭　均：治療可能な希少疾病ファブリー病～酵素補充療法の実際～. 第** 40 **回 日本小児臨床薬理学会 学術集会, 横浜**, 2013. 11

24)**北風圭介, 辻　大輔, 浅沼大祐, 神谷真子, 浦野泰照, 櫻庭　均, 伊藤孝司：酵素の分子構造改変に基づく** Tay-Sachs **病治療薬の開発. 日本薬学会第** 133 **年会. 横浜**, 2013. 3

25)**水戸部さゆり, 兎川忠靖, 月村考宏, 齋藤静司, 鈴木俊宏, 櫻庭　均：血漿中** Lyso-Gb3 **濃度の増加を伴わない特異なファブリー病患者群に関する研究. 日本薬学会第** 133 **年会. 横浜**, 2013. 3

26)**月村考宏, 高澤かおり, 山下翔悟, 水戸部さゆり, 鈴木俊宏, 片山昌勅, 兎川忠靖, 櫻庭　均：グロボトリアオシルセラミドの新規測定法の開発: ファブリー病バイオマーカーへの応用. 日本薬学会第** 133 **年会. 横浜**, 2013. 3

27)**高田　大, 相澤良明, 月村考宏, 兎川忠靖, 鈴木俊宏, 櫻庭　均：リソソーム酵素中のマンノース-6-リン酸残基の測定. 日本薬学会第** 133 **年会. 横浜**, 2013. 3

28)**月村考宏, 高田　大, 相澤良明, 鈴木俊宏, 片山昌勅, 櫻庭　均, 兎川忠靖：マンノース-6-リン酸残基の新規定量法の開発：組換えヒトリソソーム酵素解析への応用. 第** 86 **回日本生化学会大会. 横浜**, 2013. 9

29)**北風圭介, 河野加菜子, 田島陽一,　櫻庭　均, 伊藤孝司：テイ‐サックス病の新規治療薬開発を目指した機能改変型ヒトβ-ヘキソサミニダーゼ** B **の精製および評価. 第** 86 **回日本生化学会大会. 横浜**, 2013. 9

30)**田島陽一, 芝崎 太, 櫻庭　均：ポンペマウス骨格筋における** p62 **と** Parkin **の蓄積.　第** 36 **回日本分子生物学会年会. 神戸**, 2013. 12

北川 照男

【論文発表】

1) 北川照男、大和田操、他．新生児マス･スクリーニングの今後の飛躍を期待して．日本マス・スクリーニング学会誌　第 23 巻，2 号，P175,2013．
2) 鈴木　健、北川照男、他．ライソゾーム病（ファブリー病、糖原病 II 型、ムコ多糖症 I 型・II 型）スクリーニング法の基礎的検討―酵素活性測定の反応時間について．日本マス・スクリーニング学会誌　第 23 巻，2 号，P213,2013．

奥山　虎之

【論文発表】

1) T Tajima G, Sakura N, Kosuga M et al. Effects of idursulfase enzyme replacement therapy for Mucopolysaccharidosis type II when started in early infancy: comparison in two siblings. Mol Genet Metab. 2013;108:172-7.

【学会発表】

1) Tanaka A, Okuyama T, Suzuki Y, Sakai N, Hamazaki T, Kosuga M. Sawada T, Yabe H, Ishige M, Mugishima H, Kato S：EFFICACY OF ENZYME REPLACEMENT THERAPY VERSUS HEMATOPOIETIC STEM CELL TRANSPLANTATION ON BRAIN INVOLVEMENT IN MPS II, 12th International Congress of Inborn Errors of Metabolism,Barcelona,Spain, September.4.2013.
2) 小須賀基通、木田和宏、藤直子、奥山虎之:5 つのライソゾーム酵素同時測定によるライソゾーム病の新たなスクリーニング法. 第 116 回 日本小児科学会学術集会学会、広島、2013.4.19.
3) 奥山虎之：ライソゾーム病に対する新生児マス・スクリーニングの現状と今後の課題（シンポジウム）.第 40 回日本マス・スクリーニング学会学術集会、大阪、2013.8.24.

坪井　一哉

【学会発表】

1) 内富一仁, 村上敬之, 坪井一哉, 山本浩志. ファブリー病の腎障害における agalsidase aifa の有効性.　第 67 回日交通医学会総会; 2013 June 8-9; 広島
2) 内富一仁, 村上敬之, 坪井一哉, 山本浩志. ファブリー病の腎障害における agalsidase aifa の有効性.　第 67 回日交通医学会総会; 2013 June 8-9; 広島
3) 西山裕乃, 坪井一哉, 山本浩志. ファブリー病における Lyso-Gb3 を用いた治療有効性の検討.　第 67 回日交通医学会総会; 2013 June 8-9; 広島
4) Yamamoto H, Tsuboi K, Togawa T. Componential analysis of the cerumen in patients with Fabry disease. The 3rd Asian Congress for Inherited Metabolic Disease and The 55th Annual Meeting of The Japanese Society for Inherited Metabolic Disease; 2013 Nov 27-29; Chiba, Japan
5) Yamamoto H, Goto H, Tsuboi K. Histopathological findings of the nasal mucosa in 2 cosanguineous patients with Fabry disease. 12th International Congress of Inborn Errors of Metabolism; 2013 September 3rd - 6th Barcelona, Spain.
6) Tsuboi K, Yamamoto H, Goto H. Clinical course and safety in 13 Fabry Disease patients who switched from agalsidase-beta to agalsidase-aifa. 12th International Congress of Inborn

Errors of Metabolism; 2013 September 3rd - 6th Barcelona, Spain

7) Tsuboi K, Yamamoto H, Goto H. Switch from agalsidase beta to agalsidase alfa in 13 Fabry disease (FD) patients: Clinical course and safety. The 3rd Asian Congress for Inherited Metabolic Disease and The 55th Annual Meeting of The Japanese Society for Inherited Metabolic Disease; 2013 Nov 27-29; Chiba, Japan
8) Tsuboi K. Wish with a silver wing. 15th Asia LSD Symposium; 2013 November 26th Chiba, Japan
9) Goto H, Tsuboi K, Yamamoto H. Abnormal heart rate variability and left ventricular hypertrophy in patients with Fabry disease. 12th International Congress of Inborn Errors of Metabolism; 2013 September 3rd - 6th Barcelona, Spain
10) Goto H, Tsuboi K, Yamamoto H. Cardiac manifestations and enzyme replacement therapy of Fabry disease. 12th International Congress of Inborn Errors of Metabolism; 2013 September 3rd - 6th Barcelona, Spain

松田 純子

【論文発表】

1) Murakami, I, Mitsutake, S, Kobayashi, N, Matsuda, J, Suzuki, A, Shigyo, T, Igarashi, Y.: Improved high-fat diet-induced glucose intolerance by an oral administration of phytosphingosine. Biosci. Biotechnol. Biochem. 77, 194-197. 2013.

【著書】

1) **松田純子：シアリドーシス．先天代謝異常ハンドブック．中山書店．** p.212-213, 2013.

【学会発表】

1) Matsuda J, Ono K, Muto M, Yoneshige A, Yoshimura S.: Overexpression of prosaposin causes severe retinal degeneration in mouse. **第 55 回日本先天代謝異常学会** 2013 **年** 11 **月** 27-29 **日 舞浜.**
2) **久樹晴美、只野－有冨桂子、宮川誠、内田俊也、松田純子、戸田年総、岡崎具樹．：**Saposin D **欠損マウスの** 2D-DIGE **タンパク質発現解析 - 炭酸脱水酵素**(CA2)**との関連**. **第** 86 **回日本生化学会大会**. 2013 **年** 9 **月** 11-13 **日 横浜.**

遠藤 文夫

【論文発表】

1) Kido J, Nakamura K, Matsumoto S, Mitsubuchi H, Ohura T, Shigematsu Y,Yorifuji T, Kasahara M, Horikawa R and Endo F Current status of hepatic glycogen storage disease in Japan: clinical manifestations, treatments and long-term outcomes. J. Hum. Genet. 58, 285-292 (2013)
2) Lee D, Oka T, Hunter B, Robinson A, Papp S, Nakamura K, Srisakuldee W, Nickel BE, Light PE, Dyck JRB, Lopaschuk GD, Kardami E, Opas M, and Michalak M Calreticulin induces dilated cardiomyopathy. Plos One 8, e56387 (2013)
3) Yamamoto A, Nakamura K, Matsumoto S, Iwai M, Shigematsu Y, Tajima G, Tsumura M, Okada S, Mitsubuchi H, Endo F. VLCAD deficiency in a patient who recovered from VF,

but died suddenly of an RSV infection. Pediatr Int. 55, 775–778 (2013)

4) Nakamura K, Sekijima Y, NakamuraK, Hattori K, Nagamatsu K, Shimizu Y, Yazaki M, Sakurai A, Endo F, Fukushima Y, Ikeda S p.E66Q Mutation in the GLA Gene is Associated with a High Risk of Cerebral Small-Vessel Occlusion in Elderly Japanese Males. Eur J Neurol (2013 in press)

5) Inoue T, Hattori K, Ihara K, Ishii A, Nakamura K, Hirose S Newborn screening for Fabry disease in Japan: Prevalence and genotypes of Fabry disease in a pilot study. J. Hum. Genet. (2013 in press)

6) Tanaka T, Mochida T, Maki Y, Shiraki Y, Mori H, Matsumoto S, Shimbo K, Ando T, Nakamura K, Endo F, Okamoto M. Interactive network analysis of the plasma amino acids profile in a mouse model of hyperglycemia. Springerplus. (2013 in press)

7) Fujisawa D, Nakamura K, Mitsubuchi H, Ohura T, Shigematsu Y, Yorifuji T, Kasahara M, Horikawa R and Endo F Clinical features and management of organic acidemias in Japan. J. Hum. Genet. (2013 in press)

8) Kido J, Nakamura K, Mitsubuchi H, Ohura T, Takayanagi M, Matsuo M, Yoshino M, Shigematsu Y, Yorifuji T, Kasahara M, Horikawa R, Endo F. Long-term outcome and intervention of urea cycle disorders in Japan. J Inherit Metab Dis. 35, 777–785 (2012).

9) Nishino T, Obata Y, Furusu A, Hirose M, Shinzato K, Hattori K, Nakamura K, Matsumoto T, Endo F, Kohno S Identification of a novel mutation and prevalence study for fabry disease in Japanese dialysis patients. Ren Fail. 34, 566-570 (2012)

10)Katsuren K, Nakamura K, Ohta T Effect of body mass index-z score on adverse levels of cardiovascular disease risk factors. Pediatr Int. 54, 200-204 (2012)

下澤 伸行

【論文発表】

1) Vu Chi Dung, Nobuyuki Shimozawa, Nguyen Ngoc Khanh, et al. Mutations of ABCD1 gene and phenotype of Vietnamese patients with X-linked adrenoleukodystrophy (X-ALD). **International Journal of Pediatric Endocrinology** Suppl 1: 127, 2013.

2) Ohba C, Osaka H, Shimozawa N, et al. Diagnostic utility of whole exome sequencing in patients showing cerebellar and/or vermis atrophy in childhood. **Neurogenetics** 14: 225-32, 2013.

3) Hama K, Nagai T, Shimozawa N, et al. Molecular Species of Phospholipids with Very Long Chain Fatty Acids in Skin Fibroblasts of Zellweger Syndrome. **Lipids** 48: 1253-1267, 2013.

4) Shuji Matsuia, Masuko Funahashia, Nobuyuki Shimozawa, et al. Newly identified milder phenotype of peroxisome biogenesis disorder caused by mutated PEX3 gene. **Brain Dev**; 35: 842-8, 2013.

5) Yumi Mizuno, Yuichi Ninomiya, Nobuyuki Shimozawa, et al. Tysnd1 deficiency in mice interferes with the peroxisomal localization of PTS2 enzymes, causing lipid metabolic abnormalities and male infertility. **PLOS Genetics** 9 :e1003286, 2013.

6) Masashi Morita, Junpei Kobayashi, Nobuyuki Shimozawa, et al. A novel double mutation in the ABCD1 gene in a patient with X-linked adrenoleukodystrophy: Analysis of the stability and function of the mutant ABCD1 protein. **J Inher Metab Dis**, Rep 10: 95-102, 2013.

7) Iwasa M, Yamagata T, Shimozawa N et al. Contiguous ABCD1 DXS1357E deletion syndrome: Report of an autopsy case. **Neuropathology** 33: 292-8, 2013.

診療ハンドブック

1) **下澤伸行 ペルオキシソーム病ハンドブック 2013 −全てのペルオキシソーム病患者の診断治療を目指して− 日本臨床社 大阪 2013 年 6 月**

2) **下澤伸行：監修、副腎白質ジストロフィー診療ハンドブック 2013 作成委員会：編集 副腎白質ジストロフィー診療ハンドブック 2013 −ALD 患者を支えている関係者の皆様へ− 協力：日本先天代謝異常学会 厚生労働省難治性疾患克服事業「ライソゾーム病(ファブリ病を含む)に関する調査研究」 西濃印刷 岐阜 2013 年 9 月**

その他の論文

1) 下澤伸行 Zellweger spectrum 先天代謝異常ハンドブック pp248-249. 中山書店. 東京. 2013 年

2) 下澤伸行 rhizomelic chondrodysplasia punctate (RCDP) type 1 先天代謝異常ハンドブック pp250-251. 中山書店. 東京. 2013 年

3) 下澤伸行 副腎白質ジストロフィー 先天代謝異常ハンドブック pp252-253. 中山書店. 東京. 2013 年

4) 下澤伸行 ペルオキシソーム β 酸化酵素欠損症 先天代謝異常ハンドブック pp254-256. 中山書店. 東京. 2013 年

5) 下澤伸行 Refsum 病、rhizomelic chondrodysplasia punctate (RCDP) type 2･3 先天代謝異常ハンドブック pp257-259. 中山書店. 東京. 2013 年

6) 塩田睦記、舟塚 真、下澤伸行、他 極長鎖脂肪酸の反復検査で診断し得た D-bifunctional protein 欠損症の 1 例 東京女子医科大学雑誌 83: E103-106, 2013 年

7) 下澤伸行 ペルオキシソーム病 小児科診療 76(1) 35-43. 2013 年

【学会発表】

1) **下澤伸行：「これだけは伝えたい診断法 ペルオキシソーム病」第 9 回先天代謝異常学会セミナー、品川、7 月** 2013

2) Shimozawa N: Peroxisomal disorder 12th Asian and Oceanian Congress on Child Neurology. Riyadh. September 2013.

3) Shimozawa N: Diagnosis and treatment of Peroxisomal diseases 3rd ACIMD & 55th JSIMD. Maihama. Nobember 2013.

今中 常雄

【論文発表】

1) Morita M, Kobayashi J, Yamazaki K, *et al.*: A novel double mutation in the *ABCD1* gene in a patient with X-linked adrenoleukodystrophy: Analysis of the stability and function of the mutant ABCD1 protein. **J Inherit Metab Dis Rep** 10: 95-102, 2013

2) Hama K, Nagai T, Nishizawa C, *et al.*: Molecular species of phospholipids with very long chain fatty acids in skin fibroblasts of Zellweger syndrome. **Lipids** 48: 1253-1267, 2013

【学会発表】

有村洸平，守田雅志，Kostsin DG**，山崎こず枝，下澤伸行，今中常雄：副腎白質ジストロフィーの治療薬開発：**ABCD1 **タンパク質の安定化を指標としたスクリーニング系の構築患．第** 14 **回**
Pharmaco-Hematology **シンポジウム．東京，**2013. 6

2) **池島俊季，川口甲介，守田雅志，今中常雄：ペルオキシソーム膜** ABC **タンパク質** ABCD1 **の機能解析．第** 86 **回日本生化学会大会．横浜，**2013. 9

1) Kostsin DG, Morita M, Yamazaki K, Arimura K, Shimozawa N, Imanaka T: Establishment and application of fluorescence-based assay for screening of chemical compounds that stabilize mutant ABCD1 protein responsible for adrenoleukodystrophy. **第** 86 **回日本生化学会大会. 横浜,** 2013. 9

2) **池島俊季*，川口甲介，守田雅志，今中常雄：メタノール資化性酵母を用いたペルオキシソーム膜** ABC **タンパク質** ABCD1 **の 発現と機能解析．第** 12 **回次世代を担う若手ファーマ・バイオフォーラム．東京，**2013. 9

3) **岡元拓海，川口甲介，金林峰，守田雅志，今中常雄：**ABC **トランスポーター**ABCD4 **のリソソームへの局在化機構の解析．日本薬学会北陸支部第** 125 **回例会．金沢，**2013. 11

4) **松本隼，守田雅志，渡邊康春，長井良憲，小林博司，高津聖志，今中常雄：副腎白質ジストロフィー：レンチウイルスベクターを用いた** ABCD1 **遺伝子発現と骨髄移植．日本薬学会北陸支部第** 125 **回例会．金沢，**2013. 11

5) **高﨑満喜子，渡邊雄一，深澤力也，川口甲介，守田雅志，大熊芳明，今中常雄：ペルオキシソーム膜形成因子** Pex3p **と相互作用するタンパク質の検索．日本薬学会北陸支部第** 125 **回例会．金沢,** 2013.11

6) **兵藤沙織，川口甲介，守田雅志，今中常雄：リソソーム膜タンパク質** LMBD1 **の異種発現系の構築．日本薬学会北陸支部第** 125 **回例会．金沢，**2013. 11

7) **岡元拓海，川口甲介，金林峰，守田雅志，今中常雄：** ABC **トランスポーター**ABCD４**のリソソームへの局在化における** LMBD1 **の役割．第** 35 **回生体膜と薬物の相互作用シンポジウム．東京,** 2013. 11

8) Morita M, Kostsin DG, Yamazaki K, Arimura K, Shimozawa N, Imanaka T: Screening of chemical compounds that stabilize ABCD1 protein with missense mutation. The 3rd Asian Congress for Inherited Metabolic Diseases. Tokyo, Nov. 2013

9) Yokoyama K, Hama K, Nagai T, Nishizawa C, Ikeda K, Morita M, Nakanishi, H, Imanaka, T, Shimozawa N, Taguchi R, and Inoue K, Inoue K. Molecular species of phospholipids with very long chain fatty acids in skin fibroblasts of Zellweger syndrome. The 3rd Asian Congress for Inherited Metabolic Diseases. Tokyo, Nov. 2013.

10) Okamoto T, Kawaguchi K, Morita M, Imanaka T: Subcellular localization of ABC transporter ABCD4 is regulated by LMBD1. **第** 36 **回日本分子生物学会年会．神戸，**2013, 12

加我 牧子

【論文発表】

1) Yasumira A, Kokubo N, Kaga M, et al：Neurobehavioral and hemodynamic evaluation of

Stroop and reverse Stroop interference in children with attention-deficit/hyperactivity disorder. Brain & Development. (in press).

2) Tsujimototo S, Yasumura A, Kaga M et al. Increased prefrontal oxygenation related to distractor-resistant working memory. Child Psychiatry Hum Development 44:678-688, 2013.

3) Inoue Y, Ito K, Kaga M, et al, Psychometric properties of Japanese version of the Swanson, Nolan, and Pelham, version-IV Scale-Teacher Form: A study of school children in community samples. Brain & Development. (in press)

【学会発表】

1) **加我牧子，軍司敦子，中村雅子，崎原ことえ，稲垣真澄：聴覚失認の神経生理学．第** 43 **回日本臨床神経生理学会学術大会.高知，** 2013．11

横山 和明

【論文発表】

1) Kotaro Hama, Toru Nagai, Chiho Nishizawa, Kazutaka Ikeda, Masashi Morita, Noriko Satoh, Hiroki Nakanishi, Tsuneo Imanaka, Nobuyuki Shimozawa, Ryo Taguchi, Keizo Inoue, Kazuaki Yokoyama. Lipids 48, 1253-1267 (2013) Molecular Species of Phospholipids with Very Long Chain Fatty Acids in Skin Fibroblasts of Zellweger Syndrome

【学会発表】

1) **第** 55 **回日本先天性代謝異常学会、**Molecular Species of Phospholipids with Very Long Chain Fatty Acids in Skin Fibroblasts of Zellweger Syndrome**、**Kazuaki Yokoyama **他、日本先天性代謝異常学会誌、**29, 193 (2013)

渡邊 順子

【論文発表】

1) Ihara K, Yoshino M, Watanabe Y, et al. : Coagulopathy in patients with late-onset ornithine transcarbamylase deficiency in remission state: a previously unrecognized complication. Pediatrics. 2013 Jan;131(1):e327-30.

2) Okano Y, Yoshino M, Watanabe Y, et al. : Fatigue and quality of life in citrin deficiency during adaptation and compensation stage. Mol Genet Metab. 2013 May;109(1):9-13.

3) Hara M, Matsuishi T, Yoshino M, et al.: An adult patient with mucolipidosis III alpha/beta presenting with parkinsonism. Brain Dev. 2013 May;35(5):462-5.

【学会発表】

1) **古賀木綿子、原田なをみ、松石豊次郎、猪口隆洋、芳野 信、松石豊次郎、渡邊順子. オルニチントランスカルバミラーゼ欠損症**（OTCD）**の** at risk **新生児における迅速診断の有用性 第** 473 **回日本小児科学会福岡地方会** 2013.2.9**（福岡市）**

2) Yano S, Miyake N, Watanabe Y, Bartley J, Abdenur JE, Wang RY, Chang R, Goto Y, Shiina M, Ogata K, Matsumoto N: Mitochondrial ubiquinol-cytochrome c reductase core protein II defects may affect multiple metabolic pathways. 12th International Congress of Inborn Errors of

Metabolism 2013.9.3-9.6 (Barcelona)

3) Yoshino M, Harada N, Watanabe Y, Soejima M, Koda Y, Okano Y, Nakamura H, Yorifuji T: Intragenic deletion in ornithine transcarbamylase gene associated with nonhomologous recombination between an AluSx and MER68 repetitive sequences. 12th International Congress of Inborn Errors of Metabolism 2013.9.3-9.6 (Barcelona)

4) Watanabe Y, Seki Y, Yanagi T, Mizuochi T, Iwamoto J, Yoshino M、Inokuchi T, Yano S, Watanabe S,Yoshiura K, Matsuishi T: An infantile case of hepatomegaly, lactic acidosis, hypoglycemia, ketosis, and hyperlipidemia of unknown etiology. Annual Symposium of the American Society of Human Genetics 2013.10.22-26 (Boston)

5) Yoshino M, Harada N, Watanabe Y, Koda Y, Okano Y, Nakamura H, Yorifuji T.：Application of SNP-based haplotype analysis to prenatal monitoring in a pregnancy at risk for ornithine transcarbamylase deficiency. The 55th Annual Meeting of Jte Japanese Society for Inherited Metabolic Disease (JSIMD), The 3rd Asian Congress for Inherited Metabolic Diseases (ACIMD) 2013.11.27-29 (Urayasu, Chiba)

6) Harada N, Yoshida M, Watanabe Y, Soejima M, Koda Y, Okano Y, Nakamaura H, Yorifuji T.：Intragenic deletion in OTC gene associated with nonhomologous recombination between an AluSx and MER68 repetitive sequences. The 55th Annual Meeting of Jte Japanese Society for Inherited Metabolic Disease (JSIMD), The 3rd Asian Congress for Inherited Metabolic Diseases (ACIMD) 2013.11.27-29 (Urayasu, Chiba)

7) Watanabe Y, Seki Y, Yanagi T, Mizuochi T, Takeuchi T, Iwamoto J, Yoshino M、Watanabe S, Inokuchi T, Yano S, Yoshiura K, Matsuishi T.：An infantile case of hepatomegaly, lactic acidosis, hypoglycemia, ketosis, and hyperlipidemia of unknown etiology. The 55th Annual Meeting of Jte Japanese Society for Inherited Metabolic Disease (JSIMD), The 3rd Asian Congress for Inherited Metabolic Diseases (ACIMD) 2013.11.27-29 (Chiba)

8) Okano Y, Kobayashi K, Ihara K, Ito T, Yoshino M, Watanabe Y, Kaji S, Ohura T, Nagao M, Noguchi A, Mushiaki S, Hohashi N, Hashimoto-Tamaoki T.：Fatigue and quality of life in citrin deficiency during adaptation and compensation stage. The 55th Annual Meeting of Jte Japanese Society for Inherited Metabolic Disease (JSIMD), The 3rd Asian Congress for Inherited Metabolic Diseases (ACIMD) 2013.11.27-29 (Urayasu, Chiba)

9) **弓削康太郎、芳野 信、渡邊順子：ゴーシェ病 III 型に対するムコソルバン療法導入の試み 平成 25 年度厚生労働科学研究費補助金　難治性疾患克服研究事業班会議**　2013.9.26（**東京**）

石垣　景子

【論文発表】

1) **石垣景子ら．小児の呼吸管理** 9 **「神経・筋疾患」．小児科．** 2013:54(2):213-221

【学会発表】

1) **石垣　景子：小児型** Pompe **病の診断と治療」第** 30 **回小児神経筋疾患懇話会　於東京国際フォーラム，東京**　2013 **年** 8 **月** 24 **日**