Day 1：Nov. 7（Thu.）
Poster Venue（5F 503ABCD）

18：00〜19：10　Poster 1：Amino acid disorders/Urea cycle disorders

Chairperson: Yoko Nakajima

（Department of Pediatrics, Fujita Health University School of Medicine）

P-1　A case of citrin deficiency diagnosed from short stature and reccurent episode of hypoglycemia

○ Mahoko Furujo[1], Yousuke Higuchi[1], Shintaro Fujiwara[1], Yasuo Nakahara[2]
[1]Department of Pediatrics, NHO Okayama Medical Center
[2]Department of Pediatric Surgery, NHO Okayama Medical Center

P-2　Ischemic Colitis Following Anaphylactic Shock Related to Pegvaliase in a Male with PKU

○ Yuki Hasegawa[1], Shota Tanabe[2], Kazumasa Adachi[3], Kei Ugata[3], Hironori Kobayashi[4]
[1]Department of Pediatrics, Matsue Red Cross Hospital
[2]Emergency and Critical Care Medicine, Matsue Red Cross Hospital
[3]Intensive Care Unit, Matsue Red Cross Hospital
[4]Laboratories Division, Shimane University Hospital

P-3　An adult case of phenylketonuria in which approval of pegvaliase led to resumption of treatment

○ Satoru Meiri[1], Misayo Matsuyama[1], Hirotake Sawada[2], Hiroshi Moritake[1]
[1]Division of Pediatrics, Department of Developmental and Urological-Reproductive Medicine, Faculty of Medicine, University of Miyazaki
[2]Faculty of Medicine School of Nursing, University of Miyazaki

P-4　Introduction of Pegvaliase to a PKU Patient Who Has Continued a Strict Dietary Therapy Since Infanc

○ Naoko Okamura[1], Chika Takano[2,4], Erika Ogawa[2,3], Ichiro Morioka[2], Mika Ishige[2]
[1]Nutritional Management Division, Nihon University Hospital
[2]Department of Pediatrics and Child Health, Nihon University School of Medicine
[3]Department of Pediatrics, Tokyo Metropolitan Hiroo Hospital
[4]Division of Microbiology, Department of Pathology and Microbiology, Nihon University School of Medicine

P-5　A case of CPS1 deficiency in which elective surgery was possible

○ Chinatsu Nobuto[1], Kei Tamai[2], Mariko Hattori[2], Yousuke Higuchi[1], Misao Kageyama[2]
Mahoko Furujo[1]
[1]NHO Okayama Medical Center Pediatrics
[2]NHO Okayama Medical Center Neonatology

P-6　OTC deficiency presenting features consistent with neonatal hemochromatosis diagnostic criteria

○ Yuta Sudo, Yoko Nakajima, Yasuaki Yasuda, Tetsushi Yoshikawa, Tetsuya Ito
Department of Pediatrics, Fujita Health University

P-7　Adolescent complication in late-onset Argininosuccinic Aciduria

○ Yoshimitsu Osawa, Aya Wada, Yoshiaki Ohtsu, Takumi Takizawa
Department of Pediatrics Gunma University Graduate School of Medicine

18：00〜19：10　Poster 2：Organic acid disorders/Fatty acid disorders

Chairperson: Yoriko Watanabe

(Research Institute of Medical Mass Spectrometry/Department of Pediatrics, Kurume University School of Medicine)

P-8　A case of mild propionic acidemia with long QT syndrome requiring beta-blocker

○ Ryosuke Kasai[1], Hideo Sasai[1,2,3], Hiroko Goto[4,5], Mai Mori[1], Hideki Matsumoto[1], Tomohiro Hori[1]
Norio Kawamoto[1], Go Tajima[6], Hidenori Ohnishi[1,3]

[1]Department of Pediatrics, Graduate School of Medicine, Gifu University
[2]Department of Early Diagnosis and Preventive Medicine for Rare and Intractable Pediatric Diseases, Graduate School of Medicine, Gifu University
[3]Clinical Genetics Center, Gifu University Hospital
[4]Department of Pediatric Cardiology, Nagoya Tokushukai General Hospital
[5]Department of Pediatric Cardiology, Gifu Prefectural General Medical Center
[6]Division of Neonatal Screening, Research Institute, National Center for Child Health and Development

P-9　Differences in Clinical Symptoms Between D-2- and L-2-Hydroxyglutaric Aciduria

○ Ayako Matsunaga[1,3], Shiori Komachi[2], Maho Ogiwara[3], Ohsuke Migita[3,4]

[1]Department of Pediatrics, St. Marianna University School of Medicine
[2]Department of Neonatology, St. Marianna University School of Medicine
[3]Department of Clinical Genetics, St. Marianna University Hospital
[4]Department of Laboratory Medicine, St. Marianna University School of Medicine

P-10　Report of an infant with positive NBS for cblC, treated with oral cobalamin supplementation

○ Kaori Fukui[1,2], Yuri Misawa[3], Haruna Tsutsumi[3], Aki Sato[3], Azusa Tamada[3], Kaori Inoue[3]
Ryuta Takase[1], Yoriko Watanabe[1,3]

[1]Department of Pediatric and Child Health, Kurume University School of Medicine
[2]Kitakyushu City Yahata Hospital Children's Medical Center
[3]Research Institute of Medical Mass Spectrometry, Kurume University School of Medicine

P-11　Two cases of inflammatory bowel disease after liver transplantation for methylmalonic acidemia

○ Sadahiro Fukui[1], Hibiki Doi[1], Ichiro Takeuchi[2], Hirotaka Shimizu[2], Keisuke Yoshii[1]
Yasuhiro Naiki[1], Katsuhiro Arai[2], Akinari Fukuda[3], Mureo Kasahara[3], Reiko Horikawa[1]

[1]Division of Endocrinology and Metabolism,National Center for Child Health and Development
[2]Division of Gastroenterology,National Center for Child Health and Development
[3]Center for Organ Transplantation,National Center for Child Health and Development

P-12　Carnitine deficiency induced by oral semaglutide in a patient with MADD and type 2 diabetes mellitus

○ Yasuko Saito[1], Yoichi Wada[1], Masamitsu Maekawa[2], Masahiro Watanabe[2], Eriko Totsune[1]
Natsuko Ichinoi[1], Atsuo Kikuchi[1], Shigeo Kure[1]

[1]Department of Pediatrics, Tohoku University Graduate School of Medicine
[2]Department of Pharmaceutical Sciences, Tohoku University Hospital

P-13　Case Report: Late-Onset CPT2 Deficiency in a 9-Year-Old Female

○ Yuka Nakajima, You Watanabe, Kohei Iwata, Yuuki Yamada, Shinji Higuchi, Jun Mori
Osaka City General Hospital

P-14　Recurrent acute pancreatitis in a case with carnitine palmitoyl transferase-I deficiency

○ Shungo Okamoto, Kana Kitayama, Takashi Hamazaki
Department of Pediatrics, Osaka Metropolitan University Graduate School of Medicine

18：00～19：10　Poster 3：Newborn screening

Chairperson: Ryuichi Mashima

（Department of Clinical Laboratory Medicine, National Center for Child Health and Development）

P-15　Newborn screening for Citrullinemia in Taiwan

○ Chia-Yi Hung, Chen-Chen Liu, Chao-Chuan Liao, Li-Wen Hsu, Yin-Hsiu Chien
National Taiwan University Hospital

P-16　A case of MTHFR deficiency found by a pilot newborn screening for hypomethioninemia

○ Reiko Kagawa[1], Go Tajima[2], Toko Maeda[2], Fumiaki Sakura[1], Miori Yuasa[3], Yosuke Shigematsu[3]
Hideo Sasai[4], Masahisa Kobayashi[5], Takashi Hamazaki[6], Satoshi Okada[1]
[1]Department of Pediatrics, Hiroshima University Graduate School of Biomedical and Health Science
[2]Division of Neonatal Screening, Research Institute, National Center for Child Health and Development
[3]Department of Pediatrics, Faculty of Medical Sciences, University of Fukui
[4]Department of Early Diagnosis and Preventive Medicine for Rare Intractable Pediatric Diseases, Graduate School of Medicine, Gifu University
[5]Department of Pediatrics, The Jikei University School of Medicine
[6]Department of Pediatrics, Osaka Metropolitan University Graduate School of Medicine

P-17　High-risk screening of neuronal ceroid lpofuscinosis（NCL）type 1 and 2 using DBS

○ Miyo Munakata[1], Ken Suzuki[2], Chen Wu[2], Miki Igarashi[2], Takeo Iwamoto[1,2], Yoshikatsu Etou[1,2,3]
[1]Advanced Clinical Research Center, Southern Tohoku Research Institute for Neuroscience
[2]Advanced Clinical Research Center, Southern Tohoku Research Institute for Neuroscience
[3]The Jikei Univirsity School of Medicine

P-18　A study of neonatal screening for SCID, SMA, and 7 lysosomal storage disease（4th version）

○ Ken Suzuki[1], Miyo Munakata[2], Chen Wu[1], Miki Igarashi[1], Daisuke Hanada[3], Misaki Oosawa[3]
Takeo Iwamotpo[2,5], Yosikatsu Eto[1,2,4,5]
[1]Advanced Clinical Research Center, Institute of Neurological Disorders
[2]Minami Tohoku General Hospital
[3]Sekisui Medical, CO., LTD, SMCL Center
[4]Shinyurigaoka General Hospital
[5]The Jikei University School of Medicine

P-19　Operation considering the decrease in sample enzyme activity during transport in NBS for LSD

○ Shinichiro Yoshida[1], Yoshimi Sakaue[1], Yasuyo Sakamoto[1], Hiroko Nasu[1], Takaaki Sawada[2]
Fumio Endo[3], Kimitoshi Nakamura[2]
[1]KM Biologics Co.,Ltd. Newborn Screening Center
[2]Department of Pediatrics, Faculty of Life Sciences, Kumamoto University
[3]Kumamoto-Ezuko Medical Center for The Severely Disabled

P-20　Genotype-Phenotype Correlation in Mucopolysaccharidosis Type II Among Neonatal Screenings in Taiwan

○ Hui-Ying Yeh, Min-Huei Hu, Yin-Hsiu Chien
National Taiwan University Hospital, Taipei, Taiwan

P-21 High Enzyme Activity of IDS and ASM Enhances Diagnosis of Mucolipidosis II/III in Newborn Screening

○ Kuan-chi Tseng, An-ju Lee, Chao-chuan Liao, Pin-wen Chen, Hui-ying Yeh, Yin-hsiu Chien
Department of Medical Genetics and Pediatrics, National Taiwan University Hospital, Taipei, Taiwan

18：00〜19：10 Poster 4：Mucopolysaccharidoses

Chairperson: Kenji Orii

(Department of Pediatrics, Nagamori Kodomo Clinic)

P-22 IDS gene mutation identified in an adult patient with Marfan syndrome-like symptoms

○ Toshiki Tsunogai[1], Ken Sakurai[1], Masahisa Kobayashi[1], Hiroshi Kobayashi[1,2]
[1]Department of Pediatrics, Jikei University School of Medicine
[2]Division of Gene Therapy, Research Center for Medical Sciences, The Jikei University School of Medicine

P-23 Heparan Sulfate in Cerebrospinal Fluid and Developmental Progression Post-Treatment in MPS II

○ Tetsumin So[1], Misa Tanaka[2], Wataru Oboshi[2], Torayuki Okuyama[2], Motomichi Kosuga[1]
[1]Department of Genetic Medicine, National Center for Child Health and Development
[2]Department of Clinical Genomics, Saitama Medical University

P-24 Non-clinical efficacy of pabinafusp alfa (JR-141) in a mouse model of mucopolysaccharidosis type II

○ Hideto Morimoto, Atsushi Imakiire, Hiroki Morioka, Noboru Tanaka, Ryuji Yamamoto
Kohtaro Minami, Tohru Hirato, Kenichi Takahashi, Hiroyuki Sonoda
JCR Pharmaceuticals Co., Ltd.

P-25 Infusion rate adjustment in pabinafusp alfa administration for mucopolysaccharidosis II

○ Kimitoshi Nakamura[1], Norio Sakai[2], Hideaki Hirai[3], Naoko Takasao[3], Tatsuyoshi Yamamoto[3]
Pascal Yoshida[3], Yuji Sato[3]
[1]Department of Pediatrics, Graduate School of Medical Sciences, Kumamoto University
[2]Center for Promoting Treatment of Intractable Diseases, Iseikai International General Hospital
[3]JCR Pharmaceuticals Co., Ltd.

P-26 Resolution of Mongolian spots in a patient with mucopolysaccharidosis type II by pabinafusp alfa

○ Tomomi Nakamura[1], Takahiro Yonekawa[2], Motomichi Kosuga[3], Minehiro Kurai[4]
Hiroyuki Sakatoku[4], Hidemi Toyoda[2], Masahiro Hirayama[2], Tatsuyoshi Yamamoto[5]
Pascal Yoshida[5], Yuji Sato[5]
[1]Mie National Hospital
[2]Mie University School of Medicine
[3]National Centre for Child Health and Development
[4]Sakatoku Pediatric clinic
[5]JCR Pharmaceuticals Co., Ltd.

P-27 Development of modified enzyme for gene therapy of mucopolysaccharidosis type II

○ Masamu Fukagawa, Yohta Shimada, Akihiro Ishii, Moe Tomifuji, Saki Matsushima
Takashi Higuchi, Hiroshi Kobayashi
Division of Gene Therapy, Research Center for Medical Sciences, The Jikei University School of Medicine

P-28 iPSCs-Derived Neurons and Brain Organoids: From Neuronal Development to Lysosomal Storage Disease

○ Chong Kun Cheon[1], Hye Ji Moon[2], Nayeon Lee[2]
[1]Department of Pediatrics, Pusan National University Children's Hospital, Pusan National University School of Medicine, Yangsan, Korea
[2]Convergence Stem Cell Research Center, Pusan National University School of Medicine, Yangsan, Korea

18：00〜19：10 Poster 5：Fabry disease/Gaucher disease/MPS II

Chairperson: Aya Narita

(Department of Pediatrics, Iseikai International General Hospital)

P-29 Exploring the Burdens of Women Living with Fabry Disease in Japan: A Patient Survey

○ Masahisa Kobayashi[1], Ikuko Kaku[2], Nanae Goto[3], Mio Tsuchiya[3,4], Norio Sakai[5]
[1]Department of Pediatrics, The Jikei University School of Medicine
[2]Japan Fabry Disease Patients and Family Association (JFA)
[3]Amicus Therapeutics K.K.
[4]Chiesi Pharma Japan K.K. (current affiliation)
[5]Center for Promoting Treatment of Intractable Diseases, ISEIKAI International General Hospital

P-30 A snapshot analysis of the post-marketing surveillance of migalastat in Japanese Fabry patients

○ Yuko Komiya[1,2], Mio Tsuchiya[1,3], Haoyang Sun[4], Crawford Bruce[4], Kazuki Otani[1]
[1]Amicus Therapeutics KK
[2]CMIC INIZIO
[3]Chiesi Pharma Japan KK
[4]Vista Health Japan

P-31 A phase 4 study to evaluate the safety and tolerability of higher infusion rates of agalsidase beta

○ Shinichi Nakamuta[1], Ozlem Goker-Alpan[2], Maryam Banikazemi[3], Khan Nedd[4], Manish Maski[5] Chase Lee[5]
[1]Rare Disease Medical, Specialty Care, Sanofi K.K.
[2]Lysosomal Disorders Research and Treatment Unit, Fairfax, VA, USA
[3]Department of Human Genetics, Mount Sinai School of Medicine, New York, NY, USA
[4]Infusion Associates, Grand Rapids, MI, USA
[5]Sanofi, Cambridge, MA, USA

P-32 Globotriaosylsphingosine (Lyso-Gb3) from cell as a biomarker for Fabry disease; Electrospary ionization HPLC-MS/MS

○ Hye-Ran Yoon[1], Chong Kun Cheon[2], Jihun Jo[1]
[1]Department of Biomedical & Pharmaceutical Analyses, College of Pharmacy, Duksung Women's University, Seoul, South Korea
[2]Pusan National University School of Medicine, Pusan National University Children's Hospital

P-33 The Impact of the COVID-19 epidemic on Gaucher disease patients in Japan.

○ Nanako Kashima[1], Ken Sakurai[2], Kimihiko Oishi[2]
[1]Department of Pediatrics, The Jikei University Katsushika Medical Center
[2]Department of Pediatrics, The Jikei University School of Medicine

P-34 Pathological and biochemical studies of an autopsy case with Gaucher disease

○ Hayato Naruse[1], Nei Fukasawa[2], Masayuki Shimoda[2], Masahisa Kobayashi[1], Yota Shimada[3]
Hiroshi Kobayashi[3], Keiko Hirano[4], Kimihiko Oishi[1]
[1]Department of Pediatrics, The Jikei University School of Medicine
[2]Department of Pathology, The Jikei University School of Medicine
[3]Division of gene therapy, Research Center for Medical Sciences, The Jikei University School of Medicine
[4]Department of Pediatrics, Iwata City Hospital

P-35 Development of intracerebroventricular recombinant human Heparan-N-Sulfatase enzyme replacement therapy in MPSIIIA

○ Young Bae Sohn[1], Aram Yang[2], Jinsup Kim[3], Ah-ra Ko[3], Yu Yeongju[3], Inyoung Jo[3]
Hyeongseok Uhm[3], Sujeong Kim[4], Sora Kim[4], Dong-Kyu Jin[3]
[1]Department of Medical Genetics, Ajou University Hospital, Ajou University School of Medicine, Suwon, Republic of Korea
[2]Department of Pediatrics, Kangbuk Samsung Hospital, Sungkyunkwan University, School of Medicine, Seoul, Republic of Korea
[3]Novel Pharma, Inc., Seoul, Republic of Korea
[4]GC Biopharma Corp., Yongin, Republic of Korea

18：00〜19：10 Poster 6：Other Lysosomal storage disorders

Chairperson: Yohta Shimada
(Division of Gene Therapy, Research Center for Medical Sciences, The Jikei University School of Medicine)

P-36 Enzyme replacement therapy for Pompe disease: Alglucosidase alfa PMS additional analysis

○ Mitsunobu Ikeda[1], Yoshinori Sunaga[2], Tatsuro Sakashita[3], Tadashi Koga[3], Takayuki Sawada[3]
Shiho Yamane[4]
[1]Rare Diseases Medical, Specialty Care Medical, Sanofi K.K.
[2]Medical affairs, Real world evidence generation partnering, Sanofi K.K.
[3]Clinical Study Support, Inc.
[4]Medical Affairs, Post-Authorization Regulatory Studies, Sanofi K.K.

P-37 Development of proxy-reported outcome for inherited metabolic leukodystrophy

○ Yuta Koto[1], Masami Tanaka[1], HyeSook Kim[1], Mitsuyo Ishiura[1], Atsushi Ohashi[1], Aya Narita[2]
Norio Sakai[2]
[1]Faculty of Nursing, Kansai Medical University
[2]ISEIKAI International General Hospital

P-38 Phenotypic analyses and drug screening using NGN2-induced neurons derived from NPC-iPSCs

○ Miki Igarashi[1], Chen Wu[1], Ken Suzuki[1], Miyo Munakata[1], Takeo Iwamoto[1], Yoshikatsu Eto[1,2]
[1]Southern TOHOKU Research Institute for Neuroscience
[2]The Jikei University

P-39 Overexpression of Prosaposin Induces Photoreceptor-Specific Apoptosis and Visual Function Impairment

○ Yuta Ishizuka[1], Daiki Hosomi[1,2], Takashi Watanabe[1], Hiroyuki Kamao[3], Junko Matsuda[1]
[1]Department of Pathophysiology and Metabolism, Kawasaki Medical School
[2]Graduate School of Health Science and Technology, Kawasaki University of Medical Welfare
[3]Department of Ophthalmology, Kawasaki Medical School

P-40 Analysis of sialidosis /galactosialidosis mice and comparison of AAV vectors for gene therapy

○ Jun Tsukimoto[1], Rin Fukuike[1], Mizuki Miyoshi[1], Yuto Horii[1], Yoshie Takeuchi[1], Nijiho Kamori[1]
Junko Matsuda[2], Noriko Tarashima[1], Noriaki Minakawa[1], Kohji Itoh[1,3]
[1]Graduate School of Pharmaceutical science, Tokushima University
[2]Kawasaki Medical School
[3]Jichi Medical University School of Medicine

P-41 Two Cases of Early Diagnosis in Infantile and Juvenile/Adult-Onset Galactosialidosis

○ Tetsumin So[1], Kazuki Yamasawa[2], Tomoko Saito[3], Motomichi Kosuga[1,4]
[1]Department of Genetic Medicine, National Center for Child Health and Developmen
[2]Clinical Genetics Center, Tokyo Medical Center
[3]Departoment of neonatology, Kanagawa Childrens Medical Center
[4]Lysosomal Disease Center, National Center for Child Health and Developmen

P-42 Evaluation of AAV vectors with tissue-specific or ubiquitous promoters for mucopolysaccharidosis type IV

○ Shunji Tomatsu, Khan Shaukat, Fnu Nidhi, Eliana Florez
Nemours Children's Health

P-43 Glycosaminoglycans in mucopolysaccharidoses and other disorders

○ Shunji Tomatsu, Khan Shaukat, Fnu Nidhi, Eliana Florez
Nemours Children's Health

18：00～19：10 Poster 7：Mitochondrial disorders

Chairperson: Masaru Shimura
(Department of Metabolism, Chiba Children's Hospital)

P-44 Fatal pulmonary hypertension in a patient with neonatal-onset mitochondrial disease

○ Vlad Tocan[1], Yuichi Mushimoto[1], Kazuaki Yasuoka[1], Wakato Matsuoka[1,2], Yusaku Nagatomo[1]
Noriyuki Kaku[1,2], Hirokuni Hazama[3], Kei Murayama[4], Yoshinao Oda[3], Shouichi Ohga[1]
[1]Department of Pediatrics, Kyushu University Hospital
[2]Emergency and Critical Care Center, Kyushu University Hospital
[3]Department of Anatomic Pathology, Graduate School of Medical Sciences, Kyushu University
[4]Diagnosis and Therapeutics of Intractable Diseases, Intractable Disease Research Center, Juntendo University Faculty of Medicine

P-45 A case report of sisters with Leigh syndrome carrying mutations in the TPK1 gene

○ Taiju Hayashi[1], Kaishu Oda[1], Rei Urushibata[1], Hidetoshi Ishigaki[1], Takuya Hiraide[1]
Takeshi Miyamoto[2], Hirotomo Saitsu[3], Tokiko Fukuda[4]
[1]Department of Pediatrics, Hamamatsu University School of Medicine
[2]Department of Pediatrics, Hamamatsu Medical Center
[3]Department of Biochemistry, Hamamatsu University School of Medicine
[4]Department of Hamamatsu Child Health and Development, Hamamatsu University School of Medicine

P-46 Clinical and Molecular Characteristics of Mitochondrial Leukoencephalopathy in 41 Children from China

○ Minhan Song[1], Fang Fang[1], Xin Duan[2], Chaolong Xu[1], Huafang Jiang[1], Minzhao Wang[1]
Yang Liu[1], Hua Cheng[1]
[1]Beijing Children Hospital
[2]Beijing Children's Hospital, Children's National Medical Center

P-47 [Canceled]

P-48 Cardiac involvement and long-term prognosis in pediatric mitochondrial disease patients

○ Xin Duan[1], Fang Fang[1], Wang Mingzhao[1], Xu Chaolong[1], Zou Ying[1], Dai Sisi[2], Liu Yang[1]
[1]Department of Neurology, Beijing Children's Hospital, Capital Medical University, National Center for Children's Health, Beijing 100045, China
[2]Department of Pediatrics, Hangzhou Children's Hospital

P-49 Non-Diabetic Ketoacidosis in Patients with Pyruvate Dehydrogenase Deficiency on Classic Ketogenic Diet: A Case Report

○ Rongrong Li, Mingsheng Ma, Wei Chen, Zhengqing Qiu
Peking Union Medical College Hospital, Beijing, China

P-50 Survival analysis and treatment of 142 children with MELAS syndrome in China

○ Fang Fang[1], Chaolong Xu[2], Sisi Dai[1], Huafang Jiang[1], Xin Duan[1], Tianyu Song[1], Zixuan Zhang[1]
Tongyue Li[3], Yunxi Zhang[1]
[1]Beijing children hospital
[2]Peking Union Medical College Hospital, Beijing, China
[3]Department of Neurology, Beijing Children's Hospital, Capital Medical University, National Center for Children's Health, Beijing, China

18：00～19：10 Poster 8：Others

Chairperson: Yoichi Wada
(Department of Pediatrics, Tohoku University Hospital)

P-51 A case report of lipid storage myopathies with COASY mutation presented GA2-like clinical course

○ Chunhua Zhang[1], Zhaoxia Wang[2], Chunzhu Yan[3]
[1]MILS International
[2]The 1st Affilited Hospital of Beijing University, Beijing, China
[3]Qilu Hospital of Shandong University, Jinan, China

P-52 A pediatric case of presymptomatic cystinuria diagnosed by the presence of cystine crystals

○ Tomohiro Hori[1,2], Mai Mori[1,2], Hideki Matsumoto[1], Hideo Sasai[1,2,3], Hidenori Ohnishi[1,2]
[1]Department of Pediatrics, Graduate School of Medicine, Gifu University
[2]Clinical Genetics Center, Gifu University Hospital
[3]Department of Early Diagnosis and Preventive Medicine for Rare Intractable Pediatric Diseases, Graduate School of Medicine, Gifu University

P-53 Two siblings with intermediate galactosemia type III.

○ Eriko Totsune[1], Yoichi Wada[1], Yasuko Saito[1], Naoya Saijo[1], Yu Katata[1,2], Natsuko Ichinoi[1]
Atsuo Kikuchi[1]
[1]Department of Pediatrics, Tohoku University Graduate School of Medicine
[2]MIYAGI CHILDREN'S HOSPITAL

P-54 Quantitative Analysis of Nutrient Intake and LDL-C Levels in Familial Hypercholesterolemia

○ Rongrong Li, Wei Chen
Peking Union Medical College Hospital, Beijing, China

P-55 [Canceled]

P-56 Sustained Fracture Rate Reduction in Patients with OI Treated with Setrusumab: 14 Month Orbit Data

○ Kimimasa Tobita[1], Gary Gottesman[2], Thomas Carpenter[3], Danita Velasco[4], Maegan Wallace[5]
Peter Smith[6], Erik Imel[7], Diana Luca[1], Heather Byers[1], Michael Lewiecki[8]
[1]Ultragenyx Pharmaceutical, Novato, USA
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[3]Yale University School of Medicine, New Haven, USA
[4]Children's Nebraska Pediatric Hospital, Omaha, USA
[5]Phoenix Children's Hospital, Phoenix, USA
[6]Shriner's Hospitals for Children, Chicago, USA
[7]Indiana University School of Medicine, Indianapolis, USA
[8]University of New Mexico Health Scinence Center, Albuquerque, USA

P-57 Tailored diagnostic decision tree resulting from machine learning to improve early diagnosis of ASMD

○ Takashi Kiyono[1], Maurizio Scarpa[2], Maria Cappellini[3], Roberto Giugliani[4], Margaux Törnqvist[5]
Pauline Guilmin[5], Martin Montmerle[6], Alexandra Dumitriu[7], Neha Shah[8], Maja Gasparic[9]
[1]Sanofi
[2]Regional Coordination Center for Rare Diseases, Central Friuli University Health Authority, Udine, Italy
[3]Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Department of Clinical Sciences and Community Health, University of Milan, Italy
[4]UFRGS, HCPA, INAGEMP, DASA and CASA DOS RAROS, Porto Alegre, Brazil
[5]Quinten Health, Paris, France
[6]Sanofi, Neuilly-sur-Seine, France（Former employee of Quinten Health）
[7]Sanofi, Cambridge, MA, USA
[8]Sanofi, Cambridge, MA, USA（Former employee of Sanofi）
[9]Sanofi, Amsterdam, The Netherlands