Day 2：Nov. 8（Fri.）
Venue 2（6F 602ABCD）

8：10〜9：00　Morning Seminar 2

Sponsored by Sanofi K.K.

Chairperson: Motomichi Kosuga
（National Center for Child Health and Development）

MS2-1　Long-term results of a new enzyme for Pompe disease

○ Hiroshi Kobayashi
The Jikei Unviersity School of Medicine

MS2-2　Importance and problems of accumulating natural history data on rare diseases—Pompe Disease Registry

○ Keiko Ishigaki
Department of Pediatrics, Tokyo Women's Medical University

9：10〜10：00　Oral 5：Peroxisomal diseases/Mucopolysaccharidoses

Chairpersons: Takanobu Otomo
（Department of Molecular and Genetic Medicine, Kawasaki Medical School）
Motomichi Kosuga
（Division of Medical Genetics, National Center for Child Health and Development）

O-23　Biochemical study on the effect of ursodeoxycholic acid in a patient with Zellweger syndrome.

○ Hiroki Kawai[1,2,3], Kazuo Kubota[2], Shigeo Takashima[1], Hidenori Ohnishi[2], Nobuyuki Shimozawa[1,2]
[1]Division of Genomics Research, Life Science Research Center, Gifu University
[2]Department of Pediatrics, Graduate School of Medicine, Gifu University
[3]Department of Child Neurology, Okayama University Hospital

O-24 Longitudinal changes in urine and CSF GAG levels in MPS II patients treated with pabinafusp alfa

○ Tsubasa Oguni[1], Yoshitomo Notsu[1], Akihiro Kunisawa[2], Noboru Tanaka[3], Motomichi Kosuga[4]
Norio Sakai[5], Hideo Sasai[6], Asako Tajima[7], Yoko Nakajima[8], Koichi Nakanishi[9]
Takashi Hamazaki[10], Kazuhiro Muramatsu[11], Shozo Yano[1], Hironori Kobayashi[1]
[1]Laboratories Division, Shimane University Hospital
[2]Shimadzu Corporation
[3]JCR pharmaceuticals Co., Ltd.
[4]Division of Medical Genetics National Center for Child Health and Development
[5]Child Healthcare and Genetic Science Laboratory, Division of Health Sciences, Osaka University Graduate School of Medicine
[6]Dept of Early Diagnosis and Preventive Medicine for Rare Intractable Pediatric Diseases, Graduate School of Medicine, Gifu University
[7]Division of Endocrinology and Metabolism, Saitama Children's Medical Center
[8]Dept of Pediatrics, Fujita Health University School of Medicine
[9]Dept of Child Health and Welfare (Pediatrics), Graduate School of Medicine, University of the Ryukyus
[10]Dept of Pediatrics, Osaka City University Graduate School of Medicine
[11]Dept of Pediatrics, Jichi Medical University

O-25 Nonclinical safety study for hematopoietic stem cell gene therapy for mucopolysaccharidosis type II

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

O-26 Our experience in treating mucopolysaccharidosis type 2 with Pabinafusp alfa

○ Yusuke Hattori[1], Yusuke Noda[2], Takaaki Sawada[3], Keishin Sugawara[2], Jun Kido[2]
Shirou Matsumoto[4], Kimitoshi Nakamura[2]
[1]Kumamoto University Hospital, The Department of Pediatrics
[2]The Department of Pediatrics, Kumamoto University, Graduate School of Medical Sciences
[3]Kumamoto Iniversity Hospital, Genetic Medical Centor
[4]Kumamoto University Hospital, The Department of Neurology

O-27 A case of MPS1 with early-onset cardiomyopathy diagnosed by expanded newborn screening

○ Shoko Sonehara[1], Ryosuke Bo[1], Shingo Kubo[2], Yumi Sato[3], Nobuyuki Yamamoto[1]
Shohei Yamamoto[3], Tomoko Li[4], Hiroyuki Awano[5], Yasuhiro Takeshima[4], Kandai Nozu[1]
[1]Department of Pediatrics, Kobe University Graduate School of Medicine
[2]Department of Cardiology, Kobe Children's Hospital
[3]Department of Pediatrics, Tokai University School of Medicine
[4]Department of Pediatrics, Hyogo Medical University
[5]Organization for Research Initiative and Promotion, Tottori University

10：00～10：50　Oral 6：Other Lysosomal storage disorders

Chairpersons: Masahisa Kobayashi
(Department of Pediatrics, The Jikei University School of Medicine)
Tsutomu Takahashi
(Department of Pediatrics, Akita University Graduate School of Medicine)

O-28　Impaired lysosomal acidity maintenance in acid lipase-deficient cells leads to defective autophagy

○ Seigo Terawaki, Takahito Moriwaki, Takanobu Otomo
Department of Molecular and Genetic Medicine, Kawasaki Medical School

O-29　A case of acid sphingomyelinase deficiency（ASMD）A/B treated with enzyme replacement therapy

○ Ken Sakurai[1], Masahisa Kobayashi[1], Hiroyuki Ida[2], Kimihiko Oishi[1]
[1]Department of Pediatrics, The Jikei University School of Medicine
[2]The Jikei University

O-30　Calcium dysregulation in Tay-Sachs disease neurons

○ Yumeng Zhang[1,2], Tadahiro Numakawa[1], Ryutaro Kajihara[3], Kiseok Lee[1], Jing Pu[1], Chisato Horita[1] Jun Kido[4], Muneaki Matsuo[2], Takumi Era[1]
[1]Department of Cell Modulation, Institute of Molecular Embryology and Genetics, Kumamoto University
[2]Department of Pediatrics, Faculty of Medicine, Saga University
[3]Department of Biomedical Laboratory Sciences, Faculty of Life Sciences, Kumamoto University
[4]Department of Pediatrics, Graduate School of Medical Sciences, Kumamoto University

O-31　Sphingolipid analysis in mouse models of cuprizone-induced demyelination and Krabbe disease

○ Takashi Watanabe[1], Mao Ueno[2], Yuta Ishizuka[1], Junko Matsuda[1]
[1]Department of Pathophysiology and Metabolism, Kawasaki Medical School
[2]Faculty of Health Science and Technology, Kawasaki University of Medical Welfare

O-32　High-Dose Enzyme Replacement Therapy for Infantile-Onset Pompe Disease Started on Day 31

○ Hikaru Satou, Ryuta Takase, Kouki Kiyomatsu, Kaori Fukui, Kenji Suda, Yoriko Watanebe
The Department of Pediatrics and Child Health Kurume University School of Medicine

11：00～11：50　Sponsored Seminar 4

Sponsored by ReqMed Company, Ltd.

Chairperson: Mitsuyoshi Suzuki
(Department of Pediatrics and Adolescent Medicine, Juntendo University Faculty of Medicine)

SPSE4　CIRCLe: A Nationwide Patient Registry Study of Childhood-Onset Cholestatic Liver Disease —*A platform for diagnostic support and pathophysiological elucidation*—

○ Hisamitsu Hayashi
Graduate School of Pharmaceutical Sciences, The University of Tokyo

12：30～13：20　Lancheon Seminar 4

Sponsored by Daiichi Snkyo Co., Ltd.

Chairperson: Haruo Shintaku

(Endowed Chair for Human Resource Development in Regional Perinatal and Neonatal Medicine Osaka Metropolitan University Graduate School of Medicine)

LS4　Lifelong treatment and management of phenylketonuria in the target range of the Japanese Guidelines 2019

○ Mika Ishige

Department of Pediatrics and Child Health, Nihon University School of Medicine

13：40～14：30　Sponsored Seminar 5

Sponsored by Takeda Pharmaceutical Company Limited

Chairperson: Hiroshi Kobayashi

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

SPSE5-1　How to address anti-drug antibodies in replacement therapy; experiences in Haemophilia therapy

○ Keiji Nogami

Department of Pediatrics, Nara Medical University

SPSE5-2　Clinical issues with enzyme replacement therapy, including anti-drug antibodies

○ Takaaki Sawada

Center for Clinical Genetics, Kumamoto University Hospital

14：40～15：40　Oral 7：Amino acid disorders/Organic acid disorders

Chairpersons: Jun Kido

(Department of Pediatrics, Faculty of Life Sciences, Kumamoto University)

Yuki Hasegawa

(Department of Pediatrics, Matsue Red Cross Hospital)

O-33　A survey of SLC25A13 gene carriers for citrin deficiency using multiplex Real- time PCR assay

○ Hiroyuki Ebinuma[1], Yuzuru Abe[1], Ayaka Takezoe[1], Toshiyuki Tanaka[1], Takeshi Uramoto[2] Masaki Yoshihiro[2], CReARID General Incorporated Association[3]

[1]Tsukuba Research Institute, SEKISUI MEDICAL CO., LTD

[2]Diagnostics Strategy and Development Department, SEKISUI MEDICAL CO., LTD

[3]CReARID

O-34　Comparative analysis of biochemical and genetic profiles in Citrin Deficiency and MASLD adults

○ Hikaru Nishida[1], Eri Imagawa[1], Kaoru Ueda[2], Chisato Saeki[2], Tsunekazu Oikawa[2], Kimihiko Oishi[1]

[1]Department of Pediatrics, The Jikei University School of Medicine

[2]Division of Gastroenterology and Hepatology, Department of Internal Medicine, The Jikei University School of Medicine

O-35 Monitoring and management of homocystinuria under betaine therapy

○ Masayoshi Nagao, Toju Tanaka
NHO Hokkaido Medical Center

O-36 Long-term prognosis of severe forms of propionic acidemia in Okinawa, Japan

○ Yasutsugu Chinen, Sadao Nakamura, Shingo Kurokawa, Noriko Nakayama, Koichi Nakanishi
Department of Child Health and Welfare, Graduate School of Medicine, University of the Ryukyus

O-37 Successful pregnancy and delivery in maternal isovaleric acidemia

○ Keiko Ichimoto[1], Sachi Morimoto[2], Shouri Itoyama[2], Makiko Tajika[1], Masaru Shimura[1]
[1]Department of Metabolism, Chiba Children's Hospital
[2]Department of Obstetrics and Gynecology, Matsudo City General Hospital

O-38 mRNA-3927 for propionic acidemia: aggregate interim results of ph1/2 and long-term extension trials

○ Stephanie Grunewald[1], Dwight Koeberl[2], Neal Sondheimer[3], Ayesha Ahmad[4], Gerald Lipshutz[5]
Tarekegn Geber Hiwot[6], Can Ficicioglu[7], Claudia Soler-Alfonso[8], Bernd Schwahn[9], Linh Van[10]
Rosa Real[10], Andreas Schulze[3]
[1]Great Ormond Street Hospital for Children and Institute of Child Health, NIHR Biomedical Research Center, London, UK
[2]Department of Pediatrics, Duke University School of Medicine, Durham, NC, USA
[3]Hospital for Sick Children and University of Toronto, Toronto, ON, Canada
[4]Division of Pediatric Genetics, Metabolism and Genomic Medicine, University of Michigan, Ann Arbor, MI, USA
[5]University of California at Los Angeles (UCLA), Los Angeles, CA, USA
[6]University of Birmingham, Birmingham, UK
[7]Children Hospital of Philadelphia, PA, USA
[8]Baylor College of Medicine, Houston, TX, USA
[9]Manchester Academic Health Sciences Centre, Manchester, UK
[10]Moderna, Inc., Cambridge, MA, USA

15：40～17：00 Oral 8：Fatty acid disorders/ketone body disorder

Chairpersons: Hironori Kobayashi
(Laboratories Division, Shimane University Hospital)
Hideo Sasai
(Department of Early Diagnosis and Preventive Medicine for Rare and Intractable Pediatric Diseases, Graduate School of Medicine, Gifu University Department of Pediatrics, Graduate School of Medicine, Gifu University Clinical Genetics Center, Gifu University Hospital)

O-39 Three patients with CPT2 deficiency diagnosed with rhabdomyolysis

○ Yoko Yokoyama, Tomoko Lee, Miki Matsui, Yasuhiro Takeshima
Department of Pediatrics, Hyodo Medical University

O-40 Retrospective Claims Analysis of Cardiac Manifestations in Long-chain Fatty Acid Oxidation Disorders

○ Kimimasa Tobita[1], Julian Mesa[2], Erru Yang[2], Kate Simmons[2], Deborah Marsden[2]
Kathryn Charfield[3]
[1]Ultragenyx Japan K.K.
[2]Ultragenyx Pharmaceutical Inc., Novato, USA
[3]Department of Pediatrics, University of Colorado, Aurora, USA

O-41 Clinical data and notes during a sick day in 8 patients with MCAD deficiency

○ Hiroki Ohashi[1], Shoko Snoehara[1], Kiko Iketani[1], Yoshinori Nambu[1], Shingo Kanatani[2]
Hiroko Yamamoto[3], Atushi Nishiyama[4], Kayo Ozaki[5], Hiroyuki Awano[6], Ryousuke Bou[1]
[1]Department of pediatrics, Kobe University graduate school of medicine, Hyogo
[2]Japanese Red Cross Society Himeji Hospital
[3]Kitaharima medical center
[4]Kakogawa Central City Hospital
[5]Hyogo Prefectural Kobe Children's Hospital
[6]Organization for Research Initiative and Promotion, Tottori University, Tottori

O-42 Metabolize analysis using with novel model of Glutamic acidemia type II

○ Shirou Matsumoto[1], Yousuke Shigematsu[2], Yusuke Noda[3], Ryosuke Shinohara[4], Kana Uendo[4]
Keigo Tateishi[4], Kumiko Yoshinobu[4], Kimi Araki[4], Masatake Araki[4], Kimitoshi Nakamura[3]
[1]Department of Pediatrics, Kumamoto University
[2]Department of Pediatrics, Faculty of Medical Sciences, University of Fukui
[3]Department of Pediatrics, Graduate School of Life Sciences, Kumamoto University
[4]Kumamoto University, Institute of Resource Development and Analysis

O-43 Development of a method for measuring VLCAD enzyme activity using LC-MS/MS

○ Hironori Kobayashi[1], Miki Matsui[2], Yoshitomo Notsu[1], Tsubasa Oguni[1], Kenji Yamada[3]
Yuki Hasegawa[4], Takeshi Taketani[3], Shozo Yano[1]
[1]Laboratories Division, Shimane University Hospital
[2]Department of Pediatrics, Hyogo Collage of Medicine
[3]Department of Pediatrics, Shimane University Faculty of Medicine
[4]Division of Pediatrics, Matsue Red Cross Hospital

O-44 Site-specific analysis reveals the functional significance of two novel variants of HMGCS2

○ Yue Xiao[1], Hideo Sasai[1,2,3], Hideki Matsumoto[1], Mai Mori[1,3], Yuka Aoyama[1,4], Yuki Murai[5]
Syuichi Suzuki[6], Yasuhiko Ago[7], Norio Kawamoto[1], Hidenori Ohnishi[1,3]
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[5]Department of Diabetes and Metabolism, Shizuoka Children's Hospital
[6]Department of Pediatrics, National Defense Medical College Hospital
[7]Nemours Children Health, Wilmington, United States of America

O-45 Development and phenotypic analysis of mitochondrial acetoacetyl-CoA thiolase deficient mice

○ Mai Mori[1,2], Hideo Sasai[1,2,3], Yue Xiao[1], Hiroki Otsuka[1,2], Hideki Matsumoto[1], Yuka Aoyama[1,4]
Norio Kawamoto[1], Hironori Kobayashi[5], Masatake Osawa[6], Hidenori Ohnishi[1,2]
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[6]Laboratory of Molecular Design and Synthesis/Department of Cell Signaling, Graduate School of Medicine, Gifu University

O-46 A novel PDX1 variant in a Patient with Atypical Hyperketotic Vomiting and Elevated HbA1c

○ Shuhei Sako[1], Toshiki Tsunogai[1], Eri Imagawa[1], Naoya Saijo[2], Atsuo Kikuchi[2], Jun Takayama[3]
Kimihiko Oishi[1]
[1]The Department of Pediatrics, The Jikei University School of Medicine
[2]The Department of Pediatrics, Tohoku University Graduate School of Medicine
[3]The Department of Rare Disease Genomics, Tohoku University Graduate School of Medicine

17：10〜18：10 Case Study Session

Chairperson: Mitsuru Kubota
(Department of General Pediatrics & Interdisciplinary Medicine in National Center for Child Health and Development)

CS-1 Poorly controlled homocystinuria type 1 despite reintroduction of dietary therapy in school age

○ Nobuhiko Koga, Kanako Kojima, Shinichiro Nagamitsu
Department of pediatrics, Fukuoka University Hospital

CS-2 Neonatal fructose 1,6-bisphosphatase deficiency with hyperlactatemia and high lactate/pyruvate ratio

○ Sanae Naito[1], Ryosuke Bo[1], Kiiko Iketani[1], Hiroki Ohashi[1], Shoko Sonehara[1], Hiroaki Hanafusa[1]
Yoshinori Nambu[1], Sachiyo Fukushima[2], Tomoaki Ioroi[2], Hiroyuki Awano[3]
[1]Department of Pediatrics, Kobe University Graduate School of Medicine
[2]Department of Pediatrics, Japanese Red Cross Society Himeji Hospital
[3]Center for Research Infrastructure, Research Promotion Organization, Tottori University

CS-3 A case of hereditary fructose intolerance due to significant hepatomegaly

○ Miki Matsui[1], Tomoko Lee[1], Yoko Yokoyama[1], Masumi Okuda[1], Hideo Sasai[2], Tokiko Fukuda[3]
Yasuhiro Takeshima[1]
[1]Department of Pediatrics, Hyogo Medical University
[2]Department of Early Diagnosis and Preventive Medicine for Rare Intractable Pediatric Diseases, Gifu University Graduate School of Medicine
[3]Hamamatsu Child Health and Developmental medicine, Hamamatsu University School of Medicine

CS-4 LC-FAOD with Recurrent Consciousness Disturbances, High CK Levels During Fever in a 3-Year-Old Boy

○ Hiromune Narusawa[1], Hideaki Yagasaki[1], Toshimichi Fukao[1], Fumikazu Sano[1], Yoshimi Kaga[1]
Tomohiro Saito[2], Takeshi Inukai[1]
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