Day 1：Nov. 7（Thu.）
Venue 2（6F 602ABCD）

9：50～10：40　Oral 2：Newborn screening

Chairpersons: Nobuyuki Ishige
（Tokyo Health Service Association）
Go Tajima
（Division of Neonatal Screening, Research Institute, National Center for Child Health and Development）

O-6　A pilot study of newborn screening for late-onset OTC/CPS1 deficiency based on low citrulline levels

○ Tomoko Lee[1,2,3]
[1]Department of Pediatrics, Hyogo Medical University
[2]Department of Pediatrics, Kobe University Graduate School of Medicine
[3]Research Initiative Center, Organization for Research Initiative and Promotion, Tottori University

O-7　LCMS method for amino acids, organic acids and acylcarnitines using serum: Pediatric reference value

○ Yasushi Ueyanagi[1], Shinya Matsumoto[1], Akiyoshi Fujishima[1], Daiki Setoyama[1,2]
Yuichi Mushimoto[3], Vlad Tocan[3], Taeko Hotta[1], Yuya Kunisaki[1,2]
[1]Department of Clinical Chemistry and Laboratory Medicine, Kyushu University Hospital
[2]Department of Clinical Chemistry and Laboratory Medicine, Graduate School of Medical Sciences, Kyushu University
[3]Department of Pediatrics, Kyushu University Hospital

O-8　Pilot newborn screening for hypophosphatasia in Japan

○ Yusuke Noda[1], Jun Kido[1,2], Takaaki Sawada[1,2], Kosuke Kumeda[3], Shinichiro Yoshida[3]
Keishin Sugawara[2], Kimitoshi Nakamura[1,2]
[1]Department of Pediatrics, Kumamoto University Hospital
[2]Department of Pediatrics, Faculty of Life Sciences, Kumamoto University
[3]KM Biologics Co, Ltd, Kumamoto

O-9　Screening of MLD by quantification of N-C16:0-sulfatide using UPLC-MS/MS-Updated

○ Chen Wu[1], Ken Suzuki[1], Miki Igarashi[1], Takeo Iwamoto[1], Miyo Munakata[1], Yoshikatsu Eto[1,2]
[1]Advance Clinical Research Center, Institute of Neurological Disorders
[2]The Jikei University School of Medicine

O-10　Recommendation of NBS- IEM Disorder in India based on 20 Years' Experience using Mass Spectrometry

○ Usha Pinakin Dave
Director, MILS International India

10：50～11：40　Sponsored Seminar 2

Sponsored by Recordati Rare Diseases Japan

Chairperson: Kimihiko Oishi

(Department of Pediatrics, The Jikei University School of Medicine)

SPSE2-1　Dietary Management of Methylmalonic Acidemia and Propionic Acidemia

○ Yoko Nakajima

Department of Pediatrics, Fujita Health University School of Medicine

SPSE2-2　Therapeutic strategy for UCD/OA treated with Carglumic acid based on pathophysiology

○ Shirou Matsumoto

Department of Pediatrics, Kumamoto University

12：00～12：50　Lancheon Seminar 2

Sponsored by Clinigen K.K.

Chairperson: Torayuki Okuyama

(Department of Genomics, Saitama Medical University)

LS2-1　How to treat CNS manifestations of Mucopolysaccharidosis Type II

○ Torayuki Okuyama

Department of Genomics, Saitama Medical University

LS2-2　Intracerebroventricular Enzyme Replacement Therapy for a MPS II Case

○ Yoshimitsu Osawa

Department of Pediatrics, Gunma University Hospital

14：10～15：00　Sponsored Seminar 3

Sponsored by JCR Pharmaceuticals Co., Ltd.

Chairperson: Kimihiko Oishi

(Department of Pediatrics, The Jikei University School of Medicine)

SPSE3　Safety and Efficacy of Pabinafusp Alfa in MPS II: Crossing BBB with an Intravenously Administered Fusion Protein

○ Nicole Muschol

University Medical Center Hamburg-Eppendorf

15：10〜16：20　Oral 3：Phenylketonuria（PKU）

Chairpersons: Tetsuya Ito
（Department of Pediatrics, Fujita Health University School of Medicine）
Tomoko Lee
（Department of Pediatrics, Hyogo Medical University）

O-11　Effect of age and BH_4 bioavailability on BH_4 loading test for phenylketonuria.

○ Kana Kitayama[1], Tomoko Sakaguchi[1], Noriko Nakano[1], Shungo Okamoto[1], Daijiro Kabata[2]
Takashi Hamazaki[1], Haruo Shintaku[3]
[1]Department of Pediatrics, Osaka Metropolitan University Graduate School of Medicine
[2]Department of Medical Statistics, Osaka Metropolitan University Graduate School of Medicine
[3]Endowed Course "Human Resource Development in Regional Perinatal and Neonatal Medicine

O-12　［Canceled］

O-13　Assessment of the Treatment and Management LAndScape if phenylketonuria: ATLAS Survey Study in Japan

○ Hiromi Nyuzuki[1], Karly S. Louie[2], Ogun Sazova[2], Masayoshi Nagao[3], Akari Muranaka[4]
Chikahiko Numakura[5], Mika Ishige[6], Yoko Nakajima[7], Takashi Hamazaki[8], Kimitoshi Nakamura[9]
[1]Department of Pediatrics, Niigata University Graduate School of Medical and Dental Sciences
[2]BioMarin Pharmaceutical Inc, Novato, CA, USA
[3]Department of Pediatrics, National Hospital Organization Hokkaido Medical Center
[4]Department of Pediatrics, Yamagata University Hospital
[5]Department of Clinical Genomics & Pediatrics, Saitama Medical University
[6]Department of Pediatrics and Child Health, Nihon University School of Medicine
[7]Department of Pediatrics, Fujita Health University School of Medicine
[8]Department of Pediatrics, Osaka Metropolitan University Graduate School of Medicine
[9]Department of Pediatrics, Graduate School of Medical Sciences, Kumamoto University

O-14　Final safety and efficacy of pegvaliase in Japanese adults with phenylketonuria

○ Yoko Nakajima[1], Mika Ishige[2], Tetsuya Ito[1], Takashi Hamazaki[3], Mitsuhiro Kuwahara[4]
Lee Lawrence[5], Haruo Shintaku[3]
[1]Department of Pediatrics, Fujita Health University School of Medicine
[2]Pediatrics and Child Health, Nihon University School of Medicine
[3]Department of Pediatrics, Osaka Metropolitan University Graduate School of Medicine
[4]BioMarin Pharmaceutical Japan K.K.
[5]BioMarin Pharmaceutical Inc, Novato, CA, USA

O-15　A 6 months study of the introduction of pegvaliase in 11 adult patients with phenylketonuria

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

O-16 Phase 3 APHENITY Trial Results: Oral Sepiapterin for the Treatment of Phenylketonuria

○ Kathleen Somera-Molina[1], Tomoko Bessho[2], Nicola Longo[3], Ania Muntau[4]
Amaya Belanger-Quintana[5], Lali Margvelashvili[6], Ida Vanessa Schwartz[7], Drago Bratkovic[8]
Kimberly Ingalls[1], Neil Smith[1]
[1]PTC Therapeutics Inc, Warren, NJ, USA
[2]PTC Therapeutics K.K.
[3]Division of Medical Genetics, University of Utah, Salt Lake City, USA
[4]University Children's Hospital, University Medical Center Hamburg Eppendorf, Germany
[5]Hospital Universitario Ramon y Cajal Centro de Referencia Nacional para Enfermedades Metabolicas Hereditarias, Spain
[6]Pediatric Surgery Center, Tbilisi, Georgia
[7]Hospital de Clinicas de Porto Alegre, Porto Alegre, Brazil
[8]PARC Clinical Research, Adelaide, Australia

O-17 APHENITY Extension Study Preliminary Results: Oral Sepiapterin for the Treatment of Phenylketonuria

○ Kathleen Somera-Molina[1], Tomoko Bessho[2], Ania Muntau[3], Lali Margvelashvili[3], Laura Guilder[4]
Ida Vanessa Schwartz[5], Anita MacDonald[6], Kimberly Ingalls[1], Neil Smith[1], Melissa Lah[7]
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[3]University Children's Hospital, University Medical Center Hamburg Eppendorf, Germany
[4]The Hospital for Sick Children, Toronto, ON, Canada
[5]Hospital de Clinicas de Porto Alegre, Porto Alegre, Brazil
[6]Department of Dietetics, Birmingham Children's Hospital NHS Foundation Trust, Birmingham, UK
[7]Department of Medical and Molecular Genetics, Indiana University School of Medicine Indianapolis, IN, USA

16：30〜18：00 Sponsored Symposium
Sponsored by Takeda Pharmaceutical Company Limited

Chairpersons: Mitsuru Kubota
(Department of General Pediatrics and Interdisciplinary Medicine, National Center for Child Health and Development)
Hiroyuki Ida
(The Jikei University)
Kimitoshi Nakamura
(Kumamoto University, Graduate School of Medical Sciences, Department of Pediatrics)

SPSY-1 Shared decision making starting with the one in a million experience-Development and significance of patient-reported outcome in lysosomal storage diseases

○ Yuta Koto
Faculty of Nursing, Graduate School of Nursing, Kansai Medical University

SPSY-2 For a better future—Assessing and managing Gaucher disease—

○ Aya Narita
ISEIKAI International General Hospital

SPSY-3 The role of PRO (Patient Reported Outcome) for the patient assessment of Fabry disease

○ Norio Sakai
Center for Promoting Treatment of Intractable Diseases, ISEIKAI International General Hospital