

Day 1 : Oct. 15 (Thu.) 5F Venue1 (501)

8 : 55~9 : 30 President Lecture

Chairperson: Toyojiro Matsuishi
(St. Mary's Hospital)

PL From Clinical Genetics to Inherited Metabolic Disorders: Connections Revealed Along the Way

- Yoriko Watanabe
Division of Medical Mass Spectrometry/Department of Pediatrics and Child Health, Kurume University School of Medicine

9 : 40~10 : 40 Keynote Lecture2

Chairperson: Yoriko Watanabe
(Research Institute of Medical Mass Spectrometry, and Department of Pediatrics and Child Health, Kurume University School of Medicine)

KL2 One Third of a Century Captivated by DNA Sequences

- Naomichi Matsumoto
Department of Human Genetics, Yokohama City University Graduate School of Medicine

10 : 50~11 : 50 Keynote Lecture1

Chairperson: Shoji Yano
(Los Angeles General Medical Center/USC Pediatrics, Los Angeles, CA, USA)

KL1 Find and Treat Congenital Disorders of Glycosylation: Opportunities Abound

- Hudson H. Freeze
Sanford Burnham Prebys Medical Discovery Institute, La Jolla, CA, USA

12 : 00~12 : 50 Luncheon Seminar 1 : Management of Fabry Disease: From Newborn Screening to Follow-up and Treatment

Sponsored by Sanofi K.K.

Chairperson: Takahito Inoue
(Center for Maternal, Fetal and Neonatal Medicine, Fukuoka University Hospital)

LS1-1 Current Status and Challenges of the Follow-up System for Fabry Disease Diagnosed through Newborn Screening

- Takaaki Sawada
Center for Clinical Genetics, Kumamoto University Hospital

LS1-2 Treatment Strategies for Fabry Disease: Perspectives on Long-Term Management

- Hikaru Nishida
Department of Pediatrics, The Jikei University School of Medicine

13 : 30~14 : 30 JSIMD Award Lecture

Chairperson: Kimitoshi Nakamura

(Department of Pediatrics, Kumamoto University Graduate School of Medical Sciences)

AL1 Clinical Drug Discovery: From Concept to Application of Splice-Switching Antisense Oligonucleotides

○ Masafumi Matsuo^{1,2}

¹Graduate School of Science, Technology and Innovation, Kobe University

²Faculty of Health Sciences, Kobe Tokiwa University

AL2 Research and Development of gene therapy for lysosomal storage diseases

○ Hiroshi Kobayashi

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

14 : 40~15 : 30 Sponsored Seminar 1 : Rethinking Gaucher Disease Management: Understanding of Treatment Options and Chronic Inflammation Through Clinical Cases

Sponsored by Sanofi K.K.

Chairperson: Kimitoshi Nakamura

(Department of Pediatrics, Kumamoto University Graduate School of Medical Sciences)

SPSE1-1 Gaucher Disease Management Through Clinical Cases: ERT/SRT Selection and Transitional Care

○ Kazuo Kubota

Department of Pediatrics, Graduate School of Medicine, Gifu University

SPSE1-2 Lyso-GB1–Mediated Chronic Inflammation in Gaucher Disease: Pathophysiological Mechanisms and the Clinical Significance of Eliglustat

○ Shirou Matsumoto

Department of Neonatology, Kumamoto University Hospital

15 : 40~16 : 30 Sweets Seminar 1

Sponsored by OrphanPacific, Inc.

Chairperson: Takashi Hamazaki

(Department of Pediatrics, Osaka Metropolitan University Graduate School of Medicine)

SWSE1-1 Efficacy and Safety of RAVICTI in Urea Cycle Disorders

○ Yoichi Wada

Department of Medical Science and Innovation, SiRIUS Institute of Medical Research, Tohoku University

SWSE1-2 Long-Term Management of Urea Cycle Disorders: The Clinical Role of RAVICTI

○ Kimihiko Oishi

Department of Pediatrics, The Jikei University School of Medicine

16 : 40~17 : 40 Special Lecture 1

Chairperson: Mitsuru Kubota

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

SL1 Genome Sequencing for Critically Ill Newborns: From Diagnosis to Precision Medicine

○ Toshiki Takenouchi

Department of Pediatric Neurology Okayama University Graduate School of Medicine, Dentistry
and Pharmaceutical Sciences

Day 1 : Oct. 15 (Thu.)
4F Venue2 (411+412)

12 : 00~12 : 50 Luncheon Seminar 2

Sponsored by JCR Pharmaceuticals Co., Ltd.

Chairperson: Motomichi Kosuga

(Division of Medical Genetics, National Center for Child Health and Development)

LS2 Early Intervention with Pabinafusp Alfa for Neuroprotection

○ Shirou Matsumoto

Department of Neonatology, Kumamoto University Hospital

14 : 40~15 : 30 Oral 1 : Young Researcher Award Candidates' Presentation

Chairpersons: Kei Murayama

(Department of Pediatrics, Diagnostics and Therapeutics of Intractable Diseases, Juntendo University Faculty of Medicine)

Yoko Nakajima

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

01-1 Conditions for Discontinuation of Blood Purification Therapy in Urea Cycle Disorders

○ Shogo Shioda¹, Keisuke Yoshii¹, Kentaro Ide², Norihiko Tsuboi², Shotaro Matsumoto²

Hajime Uchida³, Akinari Fukuda³, Seisuke Sakamoto³, Mureo Kasahara³, Kenichi Kashimada¹

¹Department of Endocrinology and Metabolism, National Center for Child Health and Development

²Division of Critical Care Medicine, National Center for Child Health and Development

³Organ Transplant Center, National Center for Child Health and Development

01-2 Antisense oligonucleotide-mediated pseudoexon skipping restores PCC activity in propionic acidemia

○ Eriko Totsune¹, Yoichi Wada^{1,2}, Yasuko Mikami-Saito¹, Natsuko Arai-Ichinoi¹, Naoya Saijo¹

Jun Takayama^{3,4,5,6}, Yasuhiro Maeda⁷, Yoko Nakajima⁸, Osamu Ohara⁹, Shigeo Kure^{1,6,10}

¹Department of Pediatrics, Tohoku University Graduate School of Medicine

²Department of Medical Science and Innovation, SiRIUS Institute of Medical Research, Tohoku University

³Department of AI and Innovative Medicine, Tohoku University Graduate School of Medicine

⁴Tohoku Medical Megabank organization, Tohoku University

⁵Statistical Genetics Team, RIKEN Center for Advanced Intelligence Project

⁶Department of Rare Disease Genomics, Tohoku University Graduate School of Medicine

⁷Open Facility Center, Fujita Health University, ⁸Department of Pediatrics, Fujita Health University School of Medicine

⁹Department of Applied Genomics, Kazusa DNA Research Institute, ¹⁰Miyagi Children's Hospital

01-3 Genotype-Phenotype Study of PDHA1-Related PDHc Deficiency in an International Cohort of 891 Patients

○ Hisato Aihara^{1,2}, Tomoki Saito¹, Mai Kashisaka¹, Yohei Sugiyama^{1,3}, Tomohiro Ebihara⁴

Tomoko Tsuruoka⁴, Yasushi Okazaki³, Kei Murayama^{1,3}

¹Department of Pediatrics, Juntendo University

²Department of Pediatrics, Tokyo Metropolitan Toshima Hospital

³Diagnostics and Therapeutics of Intractable Diseases, Intractable Disease Research Center, Juntendo University

⁴Department of Neonatology, Chiba Children's Hospital

01-4 Intracerebral mRNA replacement therapy for sialidosis: a rare central nervous system disorder

○ Jun Tsukimoto¹, Natsumi Shima², Kurumi Higa³, Takashi Watanabe⁴, Yuta Ishizuka⁴

Noriko Tarashima¹, Junko Matsuda⁴, Yusuke Sato³, Kohji Nakamura³, Noriaki Minakawa¹

¹Bioorganic Chemistry, Graduate School of Biomedical Sciences, Tokushima University

²Laboratory for Molecular Design of Pharmaceuticals, Faculty of Pharmaceutical Sciences, Hokkaido University

³Drug Delivery & Science, College of Medical, Pharmaceutical and Health Sciences School of Pharmacy, Kanazawa University

⁴Department of Pathophysiology and Metabolism, Kawasaki Medical School

01-5 Sialyl Glycan Accumulation and Histological Findings in the Brain of a Galactosialidosis Mouse Model

○ Takashi Watanabe¹, Taketo Horino², Yuta Ishizuka¹, Koji Itoh^{1,3}, Jun Tsukimoto⁴, Junko Matsuda¹

¹Department of Pathophysiology and Metabolism, Kawasaki Medical School

²Faculty of Health Science and Technology, Kawasaki University of Medical Welfare

³Department of Pediatrics, Jichi Medical University

⁴Department of Bioorganic Chemistry, Graduate School of Biomedical Sciences, Tokushima University

15 : 40~16 : 30 Sweets Seminar 2

Sponsored by Medical Affairs, Otsuka Pharmaceutical Co., Ltd.

Chairperson: Shirou Matsumoto

(Department of Neonatology, Kumamoto University Hospital)

SWSE2 History and Future Perspectives of Phenylketonuria Treatment

○ Mika Ishige

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

16 : 40~17 : 40 Oral 2 : Lysosomal storage disorders 1

Chairpersons: Hiroshi Kobayashi

(Research Center for Medical Science, The Jikei University School of Medicine)

Junko Matsuda

(Department of Pathophysiology and Metabolism, Kawasaki Medical School)

02-1 Integration of LAL Activity into a Six-Enzyme LSD MS/MS Panel: Evaluation Using LAL-D DBS

- Tetsumin So¹, Keita Matsumoto¹, Misa Tanaka¹, Naoko Fuji², Motomichi Kosuga^{1,2}

¹Department of Medical Genetics, Center for Medical Genetics, National Center for Child Health and Development

²Department of Clinical Laboratory, Advanced Clinical Laboratory, National Center for Child Health and Development

02-2 Modeling Niemann-Pick Disease Type C Using Directly Converted Neurons from Urine-Derived Cells

- Keita Matsumoto¹, Naoki Kato², Maika Itsuno¹, Hideyuki Okano³, Sumihiro Maeda¹

¹Department of Physiology, Keio University School of Medicine

²Department of Electrical Engineering and Biosciences, School of Advanced Science and Engineering, Waseda University

³Keio Regenerative Medicine Research Center

02-3 Characterization of Ocular Pathology and Visual Function in a Mouse Model of Galactosialidosis

- Yuta Ishizuka¹, Hikaru Katsube^{1,2}, Tsugumi Chikamoto¹, Takashi Watanabe¹, Hiroyuki Kamao³
Kohji Itoh^{1,4}, Jun Tsukimoto⁵, Junko Matsuda¹

¹Department of Pathophysiology and Metabolism, Kawasaki Medical School

²Faculty of Health Science and Technology, Kawasaki University of Medical Welfare

³Department of Ophthalmology, Kawasaki Medical School

⁴Department of Pediatrics, Jichi Medical University

⁵Department of Bioorganic Chemistry, Institute for Bioorganic Chemistry, Graduate School of Pharmaceutical Sciences

02-4 Significance and challenge of extended newborn screening for Krabbe disease

- Aya Narita¹, Takuma Shigeyama², Aoi Saito², Kazuya Sakemoto³, Wataru Oboshi⁴

Takashi Hamazaki⁵, Torayuki Okuyama⁶, Norio Sakai⁷

¹The Department of Pediatrics, ISEIKAI International General Hospital

²Sumika Chemical Analysis Service, Ltd., ³Osaka City Environmental Health Association

⁴AnGes Clinical Research Laboratory, AnGes, Inc.

⁵Dept of Pediatrics Graduate School of Medicine Osaka Metropolitan University

⁶Department of Clinical Genomics, Saitama Medical University

⁷Center for Promoting Treatment of Intractable Diseases, ISEIKAI International General Hospital

02-5 Proteomic analysis of fibroblasts derived from patients with neuronal ceroid lipofuscinosis type 1

- Kiyoko Hirano¹, Miyo Munakata¹, Takeo Iwamoto^{1,2}, Keiko Akiyama¹, Yoshikatsu Eto^{1,2}

¹Advanced Clinical Research Center, Southern Tohoku Research Institute for Neuroscience

²The Jikei University School of Medicine

02-6 Defining Optimal Monitoring Strategies for Assessing Migalastat Treatment Response in Fabry Disease

○ Miyo Munakata^{1,2}, Takeo Iwamoto^{1,2,3}, Keiko Akiyama^{1,4}, Kiyoko Hirano^{1,4}, Yoshikatsu Eto^{1,2,3,4}

¹Advanced Clinical Research Center, Institute for Neurological Disorders

²Minamitohoku General Hospital, ³The Jikei University School of Medicine

⁴Shinyurigaoka General Hospital

Day 1 : Oct. 15 (Thu.)
4F Poster Venue1 (409+410)

**17 : 50~19 : 00 Poster 1 : Lysosomal storage disorders I (MPS)/Adrenal disorders/
Other related diseases**

Chairpersons: Aya Narita

(Department of Pediatrics/Center for Promoting Treatment of
Intractable Diseases, ISEIKAI International General Hospital)

Ryuichi Mashima

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

P-1 A case of mucopolysaccharidosis type IVA diagnosed through evaluation of short stature

- Tomonori Sase¹, Takehiro Homma¹, Tasuku Ichikawa², Yuki Naruse², Keiko Ichimoto¹
Masato Mori², Masaru Shimura¹

¹Department of Metabolism, Chiba Children's Hospital

²Department of Pediatrics, Matsudo City General Hospital

P-2 TFE3-Related Neurodevelopmental Disorder Mimicking MPS with Pigmentary Mosaicism

- Shinji Higuchi¹, Nobuhiko Okamoto², Kumiko Yanagi³, Tadashi Kaname³, Gen Nishimura⁴
Torayuki Okuyama⁵

¹Division of Pediatric Endocrinology, Metabolism and Nephrology, Osaka City General Hospital

²Department of Medical Genetics, Osaka Women's and Children's Hospital

³Department of Genome Medicine, National Center for Child Health and Development

⁴Department of Radiology, Musashino-Yowakai Hospital

⁵Department of Clinical Genomics, Saitama Medical University

P-3 Exploratory Study of Cerebrospinal Fluid Biomarkers in Severe Mucopolysaccharidosis Type II

- Tetsumin So¹, Misa Tanaka¹, Satoshi Tanaka², Eiichiro Watanabe², Yusuke Kawashima³
Motomichi Kosuga¹

¹Department of Medical Genetics, Center for Medical Genetics, National Center for Child Health
and Development

²Department of Pediatric Surgery, Kitasato University Hospital

³Laboratory of Applied Proteomics Research, Department of Applied Genomics, Kazusa DNA
Research Institute

P-4 Pancreatic Pseudocyst in a Long-Term Survivor with Severe Mucopolysaccharidosis Type II

- Satoru Meiri¹, Shigenobu Tokunaga¹, Natsuho Yamamoto¹, Misayo Matsuyama¹
Hirotake Sawada², Hiroshi Hatada³, Naomi Uchiyama⁴, Hiroshi Kawakami⁴, Kazuhiko Nakame⁵
Hiroshi Moritake¹

¹Division of Pediatrics, Faculty of Medicine, University of Miyazaki

²Division of Nursing, Faculty of Medicine, University of Miyazaki

³Department of Endoscopy, Faculty of Medicine, University of Miyazaki Hospital

⁴Department of Gastroenterology and Hepatology, University of Miyazaki Hospital

⁵Division of Gastrointestinal, Endocrine and Pediatric Surgery, Department of Surgery, Faculty of
Medicine, University of Miyazaki

P-5 Post-marketing surveillance of pabinafusp alfa for mucopolysaccharidosis type II: an interim report

- Shungo Okamoto¹, Takashi Hamazaki¹, Tetsumin So², Motomichi Kosuga², Minoru Ioroi³
Atsushi Noguchi³, Sakiko Minai³

¹Department of Pediatrics, Graduate School of Medicine, Osaka Metropolitan University

²Division of Medical Genetics, National Center for Child Health and Development

³Development Division, JCR Pharmaceuticals Co., Ltd.

P-6 Efficacy of eliglustat with/without imiglucerase in pediatric Gaucher disease: ELIKIDS study

- Giraldo Pilar¹, Deniz Kor², Isabela Batsu³, Julie Kissell⁴, Ning Li⁵, Sefika Uslu Cil⁶

Guillermo I Drelichman⁷, Christine Xu⁸

¹Hopital Quironsalud Zaragoza and Spanish Foundation for Gaucher Disease and other Lysosomal Disorders, Zaragoza, Spain

²Cukurova University, Pediatric Metabolism and Nutrition Department, Adana, Turkey

³Sanofi, Bridgewater, New Jersey, United States

⁴Sanofi, Cambridge, MA, United States, ⁵Sanofi, Beijing, China

⁶Sanofi, Cambridge, MA, United States

⁷Hopital de Ninos Ricardo Gutierrez, Buenos Aires, Argentina

⁸Sanofi, Bridgewater, New Jersey, United States

P-7 ERT for neurovisceral acid sphingomyelinase deficiency: comparison with an untreated sibling

- Yuta Sudo, Yasuaki Yasuda, Tetsuya Ito, Yoko Nakajima

Department of Pediatrics, Fujita Health University

P-8 Screening HSP Inducers and FDA Approved Drugs for New MLD Therapeutic Candidates

- Taeko Iwamoto^{1,3}, Takashi Miyajima¹, Miki Igarashu², Chen Wu¹, Yoshikatsu Eto^{1,3}

¹Advanced Clinical Research Center, Institute for Neurological Disorders

²Institute of Medical Science, Tokyo Medical University, ³The Jikei University School of Medicine

P-9 Newborn Screening for ADA in the Kumamoto Region

- Shinichiro Yoshida¹, Ryutarou Tsuru¹, Saori Yamshita¹, Miwa Hayashida¹, Akihiro Koube¹

Takaiki Sawada², Fumio Endo³, Kimitoshi Nakamura²

¹Newborn Screening Center, KM Biologics Co., Ltd.

²Department of Pediatrics, Faculty of Life Sciences, Kumamoto University

³Kumamoto-Ezuko Medical Center for The Severely Disabled

P-10 Burden of Metabolic Dysfunction and Hormonal Outcomes in Adults with Congenital Adrenal Hyperplasia

- Biswas Monirujjaman

Department of Internal Medicine, National Institute of Respiratory Diseases, Delhi, India

P-11 Understanding Cardiovascular and Metabolic Outcomes in Patients with Congenital Adrenal Hyperplasia

- Biswas Monirujjaman

Department of Internal Medicine, National Institute of Respiratory Diseases, Delhi, India

17 : 50~19 : 00 Poster 2 : Amino acid disorders (PKU/Urea cycle disorders)

Chairpersons: Shunsuke Hayashi

(Department of Health and Nutrition, Nagasaki International University)

Mahoko Furujo

(Department of Pediatrics, Okayama Medical Center)

P-12 Introduction of Sepiapterin to a PKU girl who has continued a dietary therapy since infancy

- Mahoko Furujo, Yousuke Higuchi, Shintaro Fujiwara
Department of Pediatrics, NPO Okayama Medical Center

P-13 Successful Preconception Use of Pegvaliase in a Patient with Classical Phenylketonuria

- Tetsuya Ito^{1,2}, Yuta Sudo¹, Yasuaki Yasuda¹, Yoko Nakajima¹
¹Department of Pediatrics, Fujita Health University, School of Medicine
²Department of Pediatrics, Aichi Gakuin University Dental Hospital

P-14 Dietary Therapies were Successfully Relieved with Pegvaliase in Two Cases of Classic Phenylketonuria

- Keiko Ichimoto¹, Takehiro Homma¹, Tomonori Sase¹, Kei Murayama², Masaru Shimura¹
¹Department of Metabolism, Chiba Children's Hospital
²Diagnostics and Therapeutics of Intractable Diseases, Intractable Disease Research Center, Graduate School of Medicine, Juntendo University

P-15 Diet liberalization and QoL in sepiapterin-treated subjects with PKU receiving BH4 at study entry

- Masahiko Uchiyama¹, Nicola Longo², Janet A Thomas³, Melissa Lah⁴, Frank Rutsch⁵
Laura Guilder⁶, Liali Aljouda⁷, Alberto Burlina⁸, Neil Smith⁹, Ania C Muntau¹⁰
¹PTC Therapeutics K.K., ²University of California Los Angeles, Los Angeles, USA
³University of Colorado School of Medicine, Aurora, USA
⁴Indiana University School of Medicine, Indianapolis, USA
⁵Munster University Children Hospital, Munster, Germany
⁶University Hospitals Bristol and Weston NHS Foundation Trust, Bristol, UK
⁷The Hospital for Sick Children, Toronto, Canada, ⁸University Hospital of Padua, Padua, Italy
⁹PTC Therapeutics, Inc., Warren, USA
¹⁰University Children Hospital, University Medical Center Hamburg-Eppendorf, Hamburg, Germany

P-16 Efficacy and safety of pegvaliase in adolescents with phenylketonuria: Interim results from PEGASUS

- Kristin Lindstrom¹, Amarilis Sanchez-Valle², Ashley Andrews³, Julia B. Hennermann⁴
Jasmine Knoll⁵, Yuri A. Zarate⁶, Dammie Brown⁷, Konstantia-Maria Chavele⁸
Naomi Rm. Schwartz⁷, Janet A. Thomas⁹
¹BioMarin Pharmaceutical Inc., Novato, CA, USA
²University of South Florida, Tampa, FL, USA, ³University of Utah, Salt Lake City, UT, USA
⁴University Medical Center Mainz, Mainz, Germany
⁵Phoenix Children's Hospital, Phoenix, AZ, USA
⁶University of Kentucky College of Medicine, Lexington, KY, USA
⁷BioMarin Pharmaceutical Inc., Novato, CA, USA, ⁸BioMarin (UK) Ltd., London, UK
⁹University of Colorado School of Medicine, Aurora, CO, USA

P-17 Sepiapterin Responsiveness in people with Phenylketonuria: Pooled Analysis of Clinical Trials

- Tomoko Bessho¹, Melissa Lah², Nicola Longo³, Maria Gizewska⁴, Janet A Thomas⁵
Amaya Belanger-Quintana⁶, Ida Vanessa D Schwartz⁷, Heidi Peters⁸, Neil Smith⁹
Ania C Muntau¹⁰
¹PTC Therapeutics K.K., ²Indiana University School of Medicine, Indianapolis, USA
³University of California, Los Angeles, Los Angeles, USA
⁴Pomeranian Medical University in Szczecin, Szczecin, Poland
⁵University of Colorado School of Medicine, Aurora, USA
⁶Ramon y Cajal University Hospital, Madrid, Spain
⁷Hospital de Clinicas de Porto Alegre, Porto Alegre, Brazil
⁸The Royal Children Hospital, Melbourne, Australia, ⁹PTC Therapeutics, Inc., Warren, USA
¹⁰University Children Hospital, University Medical Center Hamburg-Eppendorf, Hamburg, Germany

P-18 A case of HHH syndrome initially suspected as ornithine transcarbamylase deficiency

- Yasuaki Yasuda, Yuta Sudo, Yoko Nakajima, Tetsuya Ito
Department of Pediatrics, Fujita Health University School of Medicine

P-19 A Girl with Late-Onset Ornithine Transcarbamylase Deficiency Diagnosed after Elevated Liver Enzymes

- Ayaka Moriwaki¹, Aya Nishikiori¹, Yu Nishibayashi¹, Kiiko Iketani¹, Ryosuke Bo²
Takeshi Kimura³, Kayo Ozaki¹
¹Department of Endocrinology and Metabolism, Hyogo Prefectural Kobe Children Hospital
²Department of Pediatrics, Kobe University School of Medicine
³Department of Pediatrics, Osaka University Graduate School of Medicine

P-20 Arginine-Induced Paradoxical Hyperammonemia in a Female Patient with OTC Deficiency

- Yasutsugu Chinen¹, Sadao Nakamura^{1,2}, Shogo Nakada¹, Noriko Nakayama¹, Takuya Kanashi¹
Koichi Nakanishi¹
¹Department of Child Health and Welfare, Graduate School of Medicine, University of the Ryukyus
²Genetic Counseling Unit, University of the Ryukyus Hospital

P-21 Chronic Management of Late-Onset OTC Deficiency Without Developmental Delay Diagnosed at Age 8

- Sakura Kawahara¹, Atsushi Iwabuchi^{1,2}, Kazuo Imagawa^{1,2}, Kennta Morimoto¹, Honoka Koutoku¹
Hajime Okuwaki¹, Takahiro Kido^{1,2}, Hidetoshi Takada^{1,2}
¹University of Tsukuba Hospital Department of Pediatrics
²Department of Child health, Faculty of Medicine, University of Tsukuba

P-22 Glycerol phenylbutyrate in Japanese urea cycle disorder patients: 24-month Phase 3 extension results

- Yoichi Wada^{1,2}, Kenichi Kashimada³, Hiromi Nyuzuki⁴, Takashi Hamazaki⁵, Mahoko Furujo⁶
Keiko Ichimoto⁷, Toshihiko Kakiuchi⁸, Chiho Ono⁹, Hiroiku Furukawa⁹, Kimitoshi Nakamura¹⁰
¹Department of Medical Science and Innovation, SiRIUS Institute of Medical Research, Tohoku University
²Department of Pediatrics, Tohoku University Hospital
³Division of Endocrinology and Metabolism, National Center for Child Health and Development
⁴Department of Pediatrics, Niigata University Medical and Dental Hospital
⁵Department of Pediatrics, Osaka Metropolitan University
⁶Department of Pediatrics, Okayama Medical Center
⁷Department of Metabolism, Chiba Children's Hospital, ⁸Department of Pediatrics, Saga University
⁹OrphanPacific Inc., ¹⁰Department of Pediatrics, Kumamoto University

Day 1 : Oct. 15 (Thu.)
4F Poster Venue2 (413+414)

17 : 50~19 : 00 Poster 3 : Fatty acid disorders/Mitochondrial disorders/Other related diseases

Chairpersons: Yuki Hasegawa

(Department of Pediatrics, Matsue Red Cross Hospital)

Masaru Shimura

(Department of Metabolism, Chiba Children's Hospital)

P-23 A case of a boy in whom malnutrition was considered the cause rather than a metabolic disorder

- Yu Nishibayashi, Aya Nishikiori, Ayaka Moriwaki, Kiiko Iketani, Kayo Ozaki
Department of Metabolism and Endocrinology, Hyogo Prefectural Children's Hospital

P-24 A 10-Year-Old Girl with CPT2 Deficiency Presenting with Hypoglycemia Triggered by Influenza A

- Hironori Kobayashi¹, Tsubasa Oguni^{1,2}, Yosihitomo Notsu², Yuki Hasegawa³, Yusuke Fujita²
Keisuke Wada², Takeshi Taketani²
¹Laboratories Division, Shimane University Hospital
²Department of Pediatrics, Shimane University Faculty of Medicine
³Division of Pediatrics, Matsue Red Cross Hospital

P-25 Triheptanoin for Adult Mitochondrial Trifunctional Protein Deficiency in Japan: Treatment Initiation

- Miki Matsui¹, Tomoko Lee¹, Yoko Yokoyama¹, Ryosuke Bo², Hikaru Nishida³
Yasuhiro Takeshima¹, Kimihiko Oishi³
¹Department of Pediatrics, Hyogo Medical University
²Department of Pediatrics, Kobe University Graduate School of Medicine
³Department of Pediatrics, The Jikei University School of Medicine

P-26 Triheptanoin for Adult Mitochondrial Trifunctional Protein Deficiency in Japan: Clinical Improvement

- Hikaru Nishida¹, Miki Matsui², Yoshimi Fujita¹, Toshiki Tsunogai¹, Tomoko Lee², Kimihiko Oishi¹
¹Department of Pediatrics, The Jikei University School of Medicine
²Department of Pediatrics, Hyogo Medical University

P-27 Longitudinal Motor Development in a Girl with Neonatal-onset cbIC and Hydrocephalus

- Yuki Hasegawa¹, Mie Uotani², Daisuke Koike¹, Keiichiro Iwasaki¹, Marie Tokushima¹
Miku Nakamura¹, Akiyoshi Horie¹, Takehisa Fujiwaki¹
¹Department of Pediatrics, Matsue Red Cross Hospital, ²Tobu Shimane Medical and Welfare Center

P-28 Cobalamin C Deficiency Stabilized by Daily High-Dose Hydroxocobalamin After Metabolic Decompensation

- Kanako Yoshizaki¹, Masahiro Miwa¹, Yuushin Yamagata¹, Takayuki Hamabata¹, Rie Kawakita¹
Mariko Sawada¹, Yuuki Hosokawa¹, Akihito Takahashi¹, Yoichi Wada^{2,3}, Kunihiro Aya¹
¹Kurashiki Central Hospital, ²Department of Pediatrics, Tohoku University Hospital
³Department of Medical Science and Innovation, SiRIUS Institute of Medical Research, Tohoku University

P-29 A familial case of transient hypermethioninemia with a heterozygous *MAT1A* variant

- Chikahiko Numakura^{1,2}, Moeko Tanaka^{1,2}, Sayaka Suzuki-Ajihara^{1,2}, Ikuma Musha^{1,2}
Midori Kimura¹, Megumi Saito-Tsuruoka¹, Junko Yotsumoto¹, Michiko Okajima³, Akira Ohtake^{1,2}
¹Department of Clinical Genomics, Saitama Medical University
²Department of Pediatrics, Saitama Medical University
³Department of Pediatrics, Kanazawa University

P-30 Ketogenic Diet for Metabolic Stabilization in a Neonate with Fetal-Onset PDHC Deficiency

- Yusuke Miyagi¹, Ryuta Takase¹, Kaori Fukui¹, Toshiki Takeuchi³, Yoriko Watanabe^{1,2}
Tatsuki Mizuochi¹
¹Department of Pediatrics and Child Health, Kurume University
²Research Institute of Medical Mass Spectrometry, Kurume University
³Department of Pediatric Neurology, Okayama University Graduate School of Medicine, Dentistry and Pharmaceutical Sciences

P-31 Multidisciplinary support for patients and families with maternally inherited mitochondrial disease

- Kasumi Kamimaru¹, Ayako Matsunaga¹, Chika Nakamura², Kei Murayama^{1,3,4}, Naoki Shimizu¹
¹Department of Pediatrics, St. Marianna University School of Medicine
²Department of Psychological, Mental Health, and Welfare Services, St. Marianna University School of Medicine
³Department of Pediatrics, Graduate School of Medicine, Juntendo University
⁴Diagnosis and Therapeutics of Intractable Diseases, Intractable Disease Research Center, Graduate School of Medicine, Juntendo University

P-32 ESTIMATING PREVALANCE OF LONG CHAIN FATTY ACID OXIDATION DISORDERS IN JAPAN

- Kimimasa Tobita¹, Yu Funakoshi¹, Yang Erru Christy²
¹Ultragenyx Japan, ²Ultragenyx Pharmaceuticals, Novato, CA, US

P-33 Rare Disease Genes: A web-based platform for genetic variant databases linked to rare diseases

- Kimimasa Tobita¹, Yu Funakoshi¹, Sean Daugherty², Robyn Hyland², Omid Khazaie Japaraghi²
Michael Parfenov², Richbourg Heather², Madoka Chinen¹, Vanessa Rangel Miller²
¹Ultragenyx Japan, ²Ultragenyx Pharmaceuticals, Novato, CA, US

17 : 50~19 : 00 Poster 4 : Other Inherited metabolic diseases/Diagnosis/Related research

Chairpersons: Hideo Sasai

(Department of Early Diagnosis and Preventive Medicine for Rare Intractable Pediatric Diseases, Graduate School of Medicine, Gifu University)

Chunhua Zhang

(Department of research & development of MILS International)

P-34 MRI-based hepatic steatosis in citrin deficiency without a history of hyperammonemia

- Miki Honto, Eri Imagawa, Hikaru Nishida, Tomoko Kasanuki, Kimihiko Oishi
Department of Pediatrics, The Jikei University School of Medicine

P-35 Patient-Reported Experience Informing Research and Outcomes in Citrin Deficiency

- Yurika Asami¹, Kuek Li Eon², Su Kit Chew¹, Holly Robinson¹, Barbara Yu^{1,2}
¹Citrin Foundation UK, London, UK, ²Citrin Foundation Singapore, Singapore

- P-36 Intensive oral anti-copper therapy prevented liver transplantation in a patient with Wilson disease**
 ○ Taku Kimura, Kazuki Homma, Naoya Tsumura, Tatsuki Mizuochi
 Department of Pediatrics and Child Health, Kurume University School of Medicine
- P-37 Clinical course of five cases of Menkes disease treated with subcutaneous copper histidinate**
 ○ Erika Ogawa^{1,2}, Chika Takano-Asai¹, Rintaro Furuta¹, Aki Kawakami³, Satoko Kumada⁴
 Yu-Hao Chen⁵, Ichiro Morioka¹, Mika Ishige¹
¹Department of Pediatrics and Child Health, Nihon University School of Medicine
²Department of Pediatrics, Tokyo Metropolitan Hiroo Hospital
³Department of Home Medical Care, Tokyo Metropolitan Children's Medical Center
⁴Department of Neuropediatrics, Tokyo Metropolitan Neurological Hospital
⁵Department of General Pediatrics, Shizuoka Children's Hospital
- P-38 Expression and enzymatic activity assay of variant-type recombinant phosphoserine phosphatase**
 ○ Hideki Matsumoto^{1,2}, Hideo Sasai^{2,3}, Mai Mori^{1,2}, Hiroki Kawai^{1,2}, Hiroki Otsuka^{1,2}
 Yuka Aoyama^{1,4}, Tomohiro Hori^{1,2}, Michio Ozeki^{1,2}, Norio Kawamoto^{1,2}, Hidenori Ohnishi^{1,2}
¹Department of Pediatrics, Graduate School of Medicine, Gifu University
²Laboratory of Intractable and Rare Diseases, Graduate School of Medicine, Gifu University
³Department of Early Diagnosis and Preventive Medicine for Rare Intractable Pediatric Diseases, Graduate School of Medicine, Gifu University
⁴Department of Clinical Engineering, College of Life and Health Sciences, Chubu University
- P-39 Profiles of metabolites in pyrimidine *de novo* pathway and phospholipids in CAD deficiency patients**
 ○ Yasuhiro Maeda¹, Yuta Sudo², Yasuaki Yasuda², Tetsuya Ito², Yoko Nakajima²
¹Open Facility Center, Fujita Health University, ²Department of Pediatrics, Fujita Health University
- P-40 Clinical Significance of Discordant Creatinine Measurements Between GC/MS and Enzymatic Assays**
 ○ Kaori Inoue¹, Yuri Misawa¹, Haruna Tsutsumi¹, Azusa Tamada¹, Mina Narahashi¹
 Sanae Matsufuji¹, Kaori Fukui², Ryuta Takase², Kei Fukami¹, Yoriko Watanabe^{1,2}
¹Research Institute of Medical Mass Spectrometry, Kurume University School of Medicine
²Department of Pediatrics and Child Health, Kurume University School of Medicine
- P-41 Rapid and Simple Differential Diagnosis of Xanthoma Using Metabolome Analysis**
 ○ Chunhua Zhang¹, Fengyun Li²
¹Department of Research & Development of MILS International
²MILS (Beijing) medical laboratory, Beijing, China
- P-42 Citrin deficiency-causing gene variant detection performance by improved multiplex RT-PCR assay**
 ○ Hiroyuki Ebinuma¹, Takuto Imaizumi¹, Ayaka Takezoe¹, Toshiyuki Tanaka¹, Shinya Okuyama²
 Clinical & Research Association for Rare, Intractable Diseases General Incorporated Association³
¹SEKISUI MEDICAL CO., LTD. Tsukuba Research Institute
²SEKISUI MEDICAL CO., LTD. Diagnostic Product Development Department, ³CRARID

P-43 ADA-Specific Newborn Screening Identifies a Case Missed by Conventional SCID Screening

○ Yuya Kinoshita¹, Jun Kido¹, Takaaki Sawada^{1,2}, Keishin Sugawara¹, Kimitoshi Nakamura¹

¹Department of Pediatrics, Faculty of Life Sciences, Kumamoto University

²Center for Clinical Genetics, Kumamoto University Hospital

P-44 Current operation of JaSMIn (Japan Registration System for Metabolic & Inherited Diseases) in 2026

○ Satoko Tsushima^{1,2,3}, Masahisa Kobayashi^{2,4}, Motomichi Kosuga^{2,3}

¹Nursing Department, Specialist Nursing Office, Certified Nurse Specialist in Genetic Nursing, National Center for Child Health and Development

²Committee for Patient Registration, Japanese Society for Inherited Metabolic Diseases

³Division of Medical Genetics, National Center for Child Health and Development

⁴Department of Pediatrics, The Jikei University School of Medicine

Day 2 : Oct. 16 (Fri.) 5F Venue1 (501)

8 : 10~9 : 00 Morning Seminar 1 Sponsored by Sumitomo Pharma Co., Ltd.

Chairperson: Fumio Takada

(Department of Medical Genetics, The Kitasato University
Graduate School of Medical Sciences)

MS1-1 The Importance of Biosimilars: Future Prospects for Rare Diseases, Including Fabry Disease

○ Shigeki Yamada

Department of Pharmacotherapeutics and Informatics, Fujita Health University School of Medicine

MS1-2 Essentials for best practices in the management of Fabry disease : Approach to treatment options

○ Masahisa Kobayashi

Department of Pediatrics, The Jikei University School of Medicine

9 : 10~10 : 10 Special Program

Chairperson: Junko Matsuda

(Department of Pathophysiology and Metabolism, Kawasaki
Medical School)

SP The Role of Lipid Metabolism in Parkinson's Disease

○ Taku Hatano

Department of Neurology, Juntendo University Graduate School of Medicine

10 : 20~11 : 50 Sponsored Symposium

Sponsored by BioMarin Pharmaceutical Japan K.K.

Chairpersons: Yoriko Watanabe

(Division of Medical Mass Spectrometry/Department of Pediatrics
and Child Health, Kurume University School of Medicine)

Takashi Hamazaki

(Department of Pediatrics, Osaka Metropolitan University
Graduate School of Medicine)

SPSY-1 The Importance of Phe Control for Neuropsychiatric and Cognitive Symptoms in PKU

○ Kristin Lindstrom

Enzyme Conditions BioMarin Pharmaceutical Inc., Novato, CA, USA

SPSY-2 Nutritional Management in PKU: Current Challenges and Future Perspectives

○ Yoko Nakajima

Department of Pediatrics, School of Medicine Fujita Health University

SPSY-3 Pharmacological Treatment of PKU in Clinical Practice: Considerations for Treatment Selection

○ Mika Ishige

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

12 : 00~12 : 50 Luncheon Seminar 3 Sponsored by PTC Therapeutics K.K.
Chairpersons: Tetsuya Ito
(Department of Pediatrics, Aichi Gakuin University Dental Hospital)

LS3 Real World Experience—Short Term Outcomes of Sephience

- Mika Ishige¹, Mahoko Furujo², Takashi Hamazaki³
 - ¹Department of Pediatrics and Child Health, Nihon University School of Medicine
 - ²Department of Pediatrics, NHO Okayama Medical Center
 - ³Department of Pediatrics, Graduate School of Medicine, Osaka Metropolitan University

13 : 00~13 : 30 SIMD Recommending Lecture
Chairperson: Masahisa Kobayashi
(Department of Pediatrics, The Jikei University Hospital)

SIMD Metabolomic and glycomic analysis of a human cell model of Krabbe disease reveal treatable deficits in glycosylation and serine metabolism

- Rodrigo T. Starosta¹, Hannah Saeger¹, Johanna Ten Hoeve², Sj Kim¹, Johan L.K. Van Hove³
Xuntian Jiang⁴, Miao He⁵, Neal Bennett¹
 - ¹Department of Molecular and Medical Genetics, Oregon Health & Science University, Portland, OR, USA
 - ²Department of Molecular and Medical Pharmacology, University of California Los Angeles, CA, USA
 - ³Department of Pediatrics, University of Colorado Anschutz, Aurora, CO, USA
 - ⁴Department of Internal Medicine, Washington University in Saint Louis, Saint Louis, MO
 - ⁵Department of Pathology and Laboratory Medicine, Children's Hospital of Philadelphia, Philadelphia, PA, USA

13 : 30~14 : 00 SLEIMPN Recommending Lecture

SLEIMPN

- Soledad Kleppe
Hospital Italiano, Buenos Aires, Argentina

14 : 10~15 : 00 Sponsored Seminar 3
Sponsored by Recordati Rare Diseases Japan K.K.
Chairperson: Shirou Matsumoto
(Department of Neonatology, Kumamoto University Hospital)

SPSE3-1 Management of Organic Acidurias in Japan: 2026 Guidelines and Clinical Practice

- Tomoko Lee
Department of Clinical Genetics/Department of Pediatrics, Hyogo Medical University

SPSE3-2 Expanding Real-World Understanding of OA Management: Updated Results from the PROTECT Study

- Sufin Yap
Sheffield Children's NHS Foundation Trust, Clontarf, Ireland

15 : 10~15 : 40 KSIMD Recommending Lecture

Chairperson: Kimihiko Oishi

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

KSIMD C3a accumulation drives endothelial-mesenchymal transition in Fabry disease despite ERT

- Jun-Hong Park¹, Hyo-Sang Do², Jong Bin Choi³, Soyoung Kim², Sun Hee Heo², Minji Kang²
Soojin Hwang⁴, In Hee Choi⁵, Gu-Hwan Kim⁶, Gabbine Wee⁷, Yong-Mahn Han³, Heounjeong Go⁸
Beom Hee Lee⁴

¹Department of Pediatrics, Asan Medical Center Children's Hospital, University of Ulsan College of Medicine, Seoul, Republic of Korea

²Asan Institute for Life Sciences, Asan Medical Center, University of Ulsan College of Medicine, Seoul, Republic of Korea

³Department of Biological Sciences, KAIST, Daejeon, Republic of Korea

⁴Department of Pediatrics, Asan Medical Center Children's Hospital, University of Ulsan College of Medicine, Seoul, Republic of Korea

⁵Department of Genetic Counseling, University of Ulsan College of Medicine, Seoul, Republic of Korea

⁶Medical Genetics Center, Asan Medical Center, University of Ulsan College of Medicine, Seoul, Republic of Korea

⁷Laboratory Animal Center, Daegu-Gyeongbuk Medical Innovation Foundation (DGMIF), Daegu, Republic of Korea

⁸Department of Pathology, Asan Medical Center, University of Ulsan College of Medicine, Seoul, Republic of Korea

15 : 50~16 : 40 Special Lecture 2

Chairperson: Norio Sakai

(Center for Promoting Treatment of Intractable Diseases, ISEIKAI International General Hospital)

SL2 Gene- and Cell-Based Strategies for Skeletal Disease in MPS

- Shunji Tomatsu
Nemours Children's Health, Wilmington, DE, USA

16 : 50~17 : 40 Sweets Seminar 4 Sponsored by Daiichi Sankyo Co., Ltd.

Chairperson: Mika Ishige

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

SWSE4 Recent Advances in the Management of Maternal Phenylketonuria

- Hiromi Nyuzuki
Department of Pediatrics and Center for Medical Genetics, Niigata University Medical and Dental Hospital

Day 2 : Oct. 16 (Fri.) 4F Venue2 (411+412)

9 : 10~10 : 10 Oral 3 : Carbohydrate metabolism disorders

Chairpersons: Atsuo Kikuchi

(Department of Pediatrics, Tohoku University School of Medicine)

Tokiko Fukuda

(Hamamatsu Child Health and Developmental Medicine, Hamamatsu University School of Medicine)

03-1 Beyond a myopathic disorder: underrecognized ketotic hypoglycemia in GSD type 0b

- Takako Yokota^{1,2}, Eri Imagawa², Miki Honto², Toshiki Tsunogai², Tomoko Kasanuki²
Wataru Murasaki¹, Kimihiko Oishi²

¹Department of Pediatrics, The Jikei University School of Medicine Kashiwa Hospital

²Department of Pediatrics, The Jikei University School of Medicine

03-2 Early SGLT2 inhibitor initiation for neutropenia in an infant with glycogen storage disease type Ib

- Takehiro Homma, Tomonori Sase, Keiko Ichimoto, Masaru Shimura
Chiba Children's Hospital

03-3 Fluctuating cholestasis in glycogen storage disease type IXc triggered by exanthema subitum

- Masayoshi Nagao¹, Ayaka Sunaga¹, Azusa Kawaguchi¹, Jiro Abe^{1,2}

¹Department of Pediatrics, Center for Genetic Metabolism, NHO Hokkaido Medical Center

²Center for Neuromuscular Disease, Child Health & Development, NHO Hokkaido Medical Center

03-4 Challenges in long-term management in fructose 1,6-bisphosphatase deficiency

- Yoshimitsu Osawa^{1,2}, Junko Naganuma², Shunpei Suda³, Masaharu Shimada¹, Kanako Kurata⁴
Yoshiaki Ohtsu¹

¹Department of Pediatrics, Gunma University Graduate School of Medicine

²Department of Endocrinology and Metabolism, Gunma Children's Medical Center

³Department of Pediatrics, Isesaki Municipal Hospital

⁴Department of Pediatrics, Takasaki General Medical Center

03-5 A Case of McArdle Disease Demonstrating a Characteristic Pattern of Muscle Involvement on MRI

- Ryosuke Bo¹, Shoko Sonehara¹, Yoshiki Asagai¹, Hiroaki Hanafusa¹, Yoshinori Nambu¹
Hiroyuki Awano²

¹Department of Pediatrics, Kobe University Graduate School of Medicine

²Research Initiative Center, Organization for Research Initiative and Promotion, Tottori University

03-6 ENO3 Gene Mutation Detected After Exercise Related Illness: A Case Report

- Shungo Okamoto, Takashi Hamazaki

Department of Pediatrics, Graduate School of Medicine, Osaka Metropolitan University

10 : 20~11 : 10 Sponsored Seminar 2 Sponsored by Ultragenyx Japan K.K.
Chairperson: Masahisa Kobayashi
(Department of Pediatrics, The Jikei University Hospital)

SPSE2-1 Screening for Familial Hypercholesterolemia Initiated through Pediatric Lifestyle Disease Prevention Health Checkups

- Tetsuo Minamino
Department of CardioRenal and CerebroVascular Medicine, Faculty of Medicine, Kagawa University

SPSE2-2 Early Recognition of HoFH: From Diagnosis to LDL Receptor-Independent Therapies

- Yukari Kinjo¹, Kazushige Dobashi², Hayato Tada^{3,4}, Junko Igaki¹
¹Department of Pediatrics, Endocrinology and Metabolism, Okinawa Prefectural Nanbu Medical Center and Children's Medical Center
²Department of Pediatrics, School of Medicine, University of Yamanashi
³Department of Cardiology, Kanazawa University Hospital
⁴Department of Cardiovascular Medicine, Graduate School of Advanced Preventive Medicine, Kanazawa University

11 : 20~12 : 00 Oral 4 : Peroxisomal disorders

Chairpersons: Kenji Orii

(Nagamori Kodomo Clinic)

Kazuo Kubota

(Department of Pediatrics, Gifu University Graduate School of Medicine)

04-1 ABCD1-deficient THP-1 cells with VLCFA accumulation show enhanced inflammatory cytokine production

- Shohei Azuma¹, Kotaro Hama^{1,2}, Yuko Fujiwara¹, Kazuaki Yokoyama¹
¹Faculty of Pharmaceutical Sciences, Teikyo University
²Advanced Comprehensive Research Organization (ACRO), Teikyo University

04-2 Development of a cerebral ALD risk assessment method integrating C26:0-LysoPC

- Hiroki Kawai^{1,2,4}, Kazuo Kubota^{1,3}, Kalayi Tandishabo⁴, Shigeo Takashima⁴
Nobuyuki Shimozaawa^{1,4}, Hidenori Ohnishi^{1,2,3}
¹Department of Pediatrics, Gifu University Graduate School of Medicine
²Laboratory of Intractable and Rare Diseases, Graduate School of Medicine, Gifu University
³Center for Genomics and Genetic Medicine, Gifu University Hospital
⁴Linkage of Innovative Knowledge and Skills (LINKs), Life Science Research Center, Institute for Advanced Study, Gifu University

04-3 LPLAT10 synthesizes C26:0-phosphatidylcholine as a precursor of C26:0-lysophosphatidylcholine

- Kotaro Hama^{1,2}, Yuko Fujiwara¹, Koko Imai³, Yoshio Kusakabe^{1,7}, Yasuhiro Hayashi^{1,8}
Shigeo Takashima^{4,5}, Shohei Azuma¹, Masaru Kondo³, Hiroki Kawai⁶, Atsushi Yamashita¹
Ryo Takita³, Nobuyuki Shimozawa^{5,6}, Kazuaki Yokoyama¹
¹Faculty of Pharmaceutical Sciences, Teikyo University
²Advanced Comprehensive Research Organization, Teikyo University
³School of Pharmaceutical Sciences, University of Shizuoka
⁴Institute for Glyco-core Research (iGCORE), Gifu University
⁵Center for Scientific Research Facilities, Institute for Advanced Study, Gifu University
⁶Department of Pediatrics Graduate School of Medicine, Gifu University
⁷Center for the Development of Pharmacy Education, Faculty of Pharmacy, Meijo University
⁸Department of Agriculture, University of Miyazaki

12 : 00~12 : 50 Luncheon Seminar 4 Sponsored by Ultragenyx Japan K.K.
Chairperson: Kimihiko Oishi
(Department of Pediatrics, The Jikei University School of Medicine)

LS4 When Energy Metabolism Fails: Addressing LC-FAOD with Triheptanoin

- Kei Murayama
Department of Pediatrics, Diagnostics and Therapeutics of Intractable Diseases, Juntendo University Faculty of Medicine

13 : 00~14 : 00 Oral 5 : New treatments
Chairpersons: Kazuhiro Muramatsu
(Department of Pediatrics, Jichi Medical University, School of Medicine)
Yoichi Wada
(Department of Medical Science and Innovation, SiRIUS Institute of Medical Research, Tohoku University)

05-1 Evaluation of T-cell gene therapy as a potential treatment for mucopolysaccharidosis type II

- Wakaba Nakano^{1,2}, Yohta Shimada¹, Shawichi Iwamuro², Saki Matsushima¹, Takashi Higuchi¹
Hiroshi Kobayashi¹
¹Division of Gene Therapy, Research Center for Medical Sciences, The Jikei University School of Medicine
²Department of Biology, Faculty of Science, Toho University

05-2 Efficacy of gene therapy for a patient with AADC deficiency moderate type using AAV2-AADC vector

- Karin Kojima¹, Marina Mizobe¹, Akihiko Miyauchi¹, Yoshiyuki Onuki², Takeshi Nakajima²
Hitoshi Osaka¹, Shin-Ichi Muramatsu³, Takanori Yamagata^{1,4}
¹Department of Pediatrics, Jichi Medical University
²Department of Neurosurgery, Jichi Medical University
³Division of Neurological Gene Therapy, Jichi Medical University
⁴Tochigi Prefectural Rehabilitation Center

05-3 Extended Follow-up of DTX301 in Adults with Late-onset Ornithine Transcarbamylase Deficiency (OTCD)

- Kimimasa Tobita¹, Laura Konczal², Janet Thomas³, Melinda Peters⁴, Margo Breilyn⁵
Maria Luzcouce⁶, Tarekegn Geberhiwot⁷, Nathalie Guffon⁸, Leticia Hualde⁹, Aneal Khan¹⁰
¹Ultragenyx Japan, ²University Hospital of Cleveland Medical Center, Cleveland, OH, US
³University of Colorado Children's Hospital of Colorado, Aurora, CO, US
⁴Boston Children's Hospital, Harvard Medical School, Boston, MA, US
⁵Icahn School of Medicine at Mount Sinai, New York, NY, US
⁶University of Santiago de Compostela, Santiago de Compostela, Spain
⁷Birmingham University, Birmingham, UK
⁸Reference Centre in Inherited Metabolic Diseases, HCL, Lyon, France
⁹Cruces University Hospital, Biocruces Bizkaia Health Research Institute, Barakaldo, Spain
¹⁰M.A.G.I.C. Clinic Ltd., Calgary, Canada

05-4 Ex Vivo Gene Therapy Restores Behavioral Performance in a Mouse Model of GM1 Gangliosidosis

- Toshiki Tsunogai¹, Yohta Shimada², Saki Matsushima², Takashi Higuchi², Shunsuke Iizuka³
Masafumi Kinoshita³, Hiroyuki Sonoda³, Toya Ohashi⁴, Hiroshi Kobayashi^{1,2}
¹Jikei University School of Medicine, Department of Pediatrics
²Jikei University School of Medicine, Center for Medical Research, Division of Gene Therapy
³JCR Pharmaceuticals Co., Ltd. Research Division
⁴The Jikei University School of Nursing, Department of Human Health Science and Therapeutics

05-5 Development of a novel oligonucleotide drug targeting a splice variant of G6PC prevalent in Japan

- Go Tajima¹, Hidefumi Nakamura², Fumiaki Sakura³, Satoshi Okada³, Masafumi Matsuo⁴
¹Division of Neonatal Screening, Research Institute, National Center for Child Health and Development
²Department of Research and Development Supervision, National Center for Child Health and Development
³Department of Pediatrics, Hiroshima University Graduate School of Biomedical and Health Sciences
⁴Graduate School of Science, Technology and Innovation, Kobe University

05-6 RESULTS FROM A PIVOTAL, PHASE 3, DOUBLE-BLIND, PLACEBO-CONTROLLED TRIAL OF DTX401 FOR GSDIA

- Yoko Nakajima¹, Jun Mori², Shiro Matsumoto³, Anne Blake⁴, Andrew Grimm⁴, Serap Sankoh⁴
Syeda Rahman⁴, Diane Turner-Bowker⁴, Collis Richard⁴, Kimimasa Tobita⁵
¹Department of Pediatrics, School of Medicine Fujita Health University
²Department of Ped Endocrinology and Metabolic Diseases, Osaka City General Hospital
³Department of Pediatrics, School of Medicine Kumamoto University
⁴Ultragenyx Pharmaceutical Inc., Novato, US, ⁵Ultragenyx Japan

14 : 10~15 : 00 Sweets Seminar 3 **Sponsored by KM Biologics Co.,Ltd.**
Chairperson: Kimitoshi Nakamura
(Department of Pediatrics, Graduate School of Medical Sciences,
Kumamoto University)

SWSE3 Expanded Newborn Screening in Fukuoka: The Development, Current Status, and Future Perspectives

- Takahito Inoue
Center for Maternal, Feetal and Neonatal Medicine, Fukuoka University Hospital

15 : 10~15 : 50 Oral 6 : Organic acid disorders/Fatty acid disorders

Chairpersons: Hironori Kobayashi

(Clinical Laboratory Division, Shimane University Hospital)

Go Tajima

(Division of Neonatal Screening, Research Institute, National, Center for Child Health and Development)

O6-1 Metabolic phenotypes of *Acat1* knockout mice: analysis in juvenile and young adult periods

○ Mai Mori^{1,2,3}, Hideo Sasai^{2,3,4}, Hiroki Otsuka^{1,2}, Ryosuke Kasai^{1,3}, Hideki Matsumoto^{1,3}

Tomohiro Hori^{1,2,3}, Yuka Aoyama^{1,5}, Miki Matsui⁶, Hironori Kobayashi⁷, Hidenori Ohnishi^{1,2,3}

¹Department of Pediatrics, Graduate School of Medicine, Gifu University

²Clinical Genetics Center, Gifu University Hospital

³Laboratory of Intractable and Rare Diseases, Graduate School of Medicine, Gifu University

⁴Department of Early Diagnosis and Preventive Medicine for Rare Intractable Pediatric Diseases, Graduate School of Medicine, Gifu University

⁵Department of Clinical Engineering, College of Life and Health Sciences, Chubu University

⁶Department of Pediatrics, Hyogo Medical University

⁷Laboratories Division, Shimane University Hospital

O6-2 Postmortem Diagnosis of Fatal Neonatal CACT Deficiency with a Novel *SLC25A20* Variant

○ Hiromi Nyuzuki^{1,2}, Aya Miura³, Takuma Yamamoto³, Akihito Koyama^{4,5}, Kazuhisa Funayama^{4,5}

Satoru Takami⁶, Hisakazu Takatsuka^{4,5}, Hajime Nishio³, Takeshi Ikeuchi¹

¹Center for Medical Genetics, Niigata University Medical and Dental Hospital

²Department of Pediatrics, Niigata University School of Medicine

³Department of Legal Medicine, Hyogo Medical University

⁴Division of Forensic Medicine, Niigata University Graduate School of Medicine, Dentistry and Health Sciences

⁵Center of Cause of Death Investigation, Niigata University Graduate School of Medicine, Dentistry and Health Sciences

⁶Department of Pediatrics, Saiseikai Niigata Hospital

O6-3 Long-Term Stable Clinical Course in Four Patients with Mild Methylmalonic Acidemia

○ Shoko Sonehara¹, Yoshitaka Asagai¹, Hiroaki Hanafusa¹, Yoshinori Nambu¹, Hiroyuki Awano²

Ryosuke Bo¹

¹Department of Pediatrics, Kobe University Graduate School of Medicine

²Organization for Research Initiative and Promotion, Tottori University

O6-4 A study of nine children with Neuromyopathy Caused by MTP Deficiency in China

○ Huafang Jiang¹, Fang Fang¹, Chaolong Xu¹, Tongyue Li¹, Danmin Shen¹, Zhimei Liu¹

Ruoyu Duan¹, Minhan Song¹

¹Department of Neurology, Beijing Children's Hospital, Capital Medical University, Beijing, China

²Department of Pediatrics, Weifang Maternal and Child Health Hospital, Weifang, Shandong, China

16 : 00~16 : 40 Oral 7 : Amino acid disorders

Chairpersons: Kenji Ihara

(Department of Pediatrics, Oita University Faculty of Medicine)

Jun Kido

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

07-1 Dimethyl alpha-ketoglutarate partially ameliorates ammonia-induced inhibition of protein synthesis

- Makoto Yoshino, Tomoyuki Takahashi, Toshihiro Morisaki
Division of Gene Therapy and Regenerative Medicine, Cognitive and Molecular Research Institute of Brain Diseases, Kurume University

07-2 The current social status in adult patients with urea cycle disorders in Japan

- Yusuke Noda^{1,2}, Jun Kido^{1,2}, Keishin Sugawara², Yusuke Hattori^{1,2}, Kimitoshi Nakamura^{1,2}
¹Department of Pediatrics, Kumamoto University Hospital
²Department of Pediatrics, Faculty of Life Sciences, Kumamoto University

07-3 Favorable outcomes in two cases with a prenatal diagnosis of severe urea cycle disorders

- Shuhei Yamaguchi¹, Kenichi Kashimada¹, Keisuke Yoshii¹, Akinari Fukuda², Seisuke Sakamoto², Tetsuya Isayama³, Sachi Koinuma⁴, Takahiro Yamada⁵, Akihiro Umezawa⁶, Mureo Kasahara²
¹Division of Endocrinology and Metabolism, National Center for Child Health and Development
²Center for Organ Transplantation, National Center for Child Health and Development
³Division of Neonatology, National Center for Child Health and Development
⁴Japan Drug Information Institute in Pregnancy, National Center for Child Health and Development
⁵Division of Clinical Genetics, Hokkaido University Hospital
⁶Center for Regenerative Medicine, National Center for Child Health and Development

07-4 Hypermethioninemia detected by NBS: Comparison of HCU1 and methionine adenosyltransferase deficiency

- Reiko Kagawa¹, Fumiaki Sakura¹, Tadashi Moriwaki², Miori Yuasa³, Yosuke Shigematsu³, Mahoko Furujo⁴, Masayoshi Nagao⁵, Go Tajima^{1,6}, Satoshi Okada¹
¹Department of Pediatrics, Hiroshima University Graduate School of Biomedical and Health Sciences
²Department of Pediatrics, National Hospital Organization Iwakuni Clinical Center
³Department of Pediatrics, Faculty of Medical Sciences, University of Fukui
⁴Department of Pediatrics, National Hospital Organization Okayama Medical Center
⁵Department of Pediatrics, National Hospital Organization Hokkaido Medical Center
⁶Division of Neonatal Screening, National Center for Child Health and Development

16 : 50~17 : 40 Sweets Seminar 5

Sponsored by Viatris Pharmaceuticals Japan G.K.

Chairperson: Noriko Miyake

(Department of Pediatrics, Nagasaki University School of Medicine)

SWSE5-1 From Development of a Cystinosis Rat Model to Clinical Research

- Tadashi Okamura
Department of Laboratory Animals, National Institute of Global Health and Medicine, Japan Institute for Health Security (JIHS)

SWSE5-2 Detecting Rare Diseases: From Suspecting Cystinosis to Treatment Strategy

- Yuuka Yamamoto
Department of Ophthalmology, Center Hospital of the National Center for Global Health and Medicine, Japan Institute for Health Security (JIHS)

Day 2 : Oct. 16 (Fri.) 4F Poster Venue1 (409+410)

17 : 40~18 : 30 Poster 5 : Poster Award Presentation

Chairpersons: Mika Ishige

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

Yota Shimada

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

P-45 Gene Therapy for Krabbe Disease Using AAV Vectors

- Takashi Higuchi, Saki Matsushima, Yohta Shimada, Hiroshi Kobayashi
Div. of Gene Thera., The Jikei Univ.

P-46 Dose Escalation of Sebelipase Alpha in Pediatric Lysosomal Acid Lipase Deficiency: Real-World Experience

- Lyudmila Ugolkova¹, Anastasiya Shepina¹, Elizaveta Leontieva¹, Yaroslav Ugolkov²
Victoria Lobanova¹
¹Reference Center for Congenital, Hereditary Diseases, Genetic Abnormalities, Orphan and Other Rare Disorders Morozov's Moscow City Children Clinical Hospital, Moscow, Russia
²Marchuk Institute of Numerical Mathematics of the Russian Academy of Sciences (INM RAS), Moscow, Russia

P-47 Intracerebroventricular mRNA delivery using PEG polymer/peptide nanoparticles for sialidosis therapy

- Taku Ikeguchi¹, Jun Tsukimoto², Taiki Yamaguchi^{1,3,4}, Noriko Tarashima², Noriaki Minakawa²
Takanori Kanazawa^{1,3,4}
¹Department of Clinical Pharmacology, School of Pharmaceutical Sciences, Tokushima University
²Department of Bioorganic Chemistry, Graduate School of Biomedical Sciences, Tokushima University
³Department of Clinical Pharmacology, Graduate School of Biomedical Sciences, Tokushima University
⁴Innovative Research Center for Drug Delivery System, Tokushima University

P-48 Molecular Diagnosis of Argininemia Caused by a Novel Deep-Intronic ARG1 Variant

- Hikaru Nishida¹, Eri Imagawa¹, Tomoko Kasanuki¹, Naoya Saijo², Jun Takayama³, Atsuo Kikuchi²
Kimihiko Oishi¹
¹Department of Pediatrics, The Jikei University School of Medicine
²Department of Pediatrics, Tohoku University Graduate School of Medicine
³Department of AI and Innovative Medicine, Tohoku University Graduate School of Medicine

P-49 Evaluation of recombinant PAH/PAL activity and performance after Red Blood Cell encapsulation

- Ai Shirai¹, Yui Kishitani², Kanade Takahara³, Sohei Ito^{3,4}, Yasuhiro Yamada^{2,5}, Nobuhiko Kojima^{1,2}
¹Department of Science, School of science, Yokohama City University
²Graduate School of Nanobioscience, Yokohama City University
³School of Food and Nutritional Sciences, University of Shizuoka
⁴Graduate School of Integrated Pharmaceutical and Nutritional Sciences, University of Shizuoka
⁵Faculty of Pharmaceutical Sciences, Nihon Pharmaceutical University

P-50 Burden and Quality of Life of Mitochondrial Disease Caregivers: A National Survey in China

○ Dan Zhao^{1,2,3}, Chaolong Xu¹, Tongyue Li¹, Huafang Jiang⁴, Junfang Suo⁵, Minhan Song⁶

Fang Fang¹

¹Department of Neurology, Beijing Children's Hospital, Capital Medical University, National Center for Children's Health, Beijing, China

²Department of Medical Genetics and Developmental Biology, School of Basic Medical Science, Capital Medical University, Beijing, China

³Laboratory for Clinical Medicine, Capital Medical University, Beijing, China

⁴Department of Pediatrics, Weifang Maternal and Child Health Hospital, Weifang, Shandong, China

⁵Department of Neurology, Henan Children's Hospital, Zhengzhou, Henan, China

⁶Department of Clinical Genetics, Children's Hospital of Fudan University, National Center for Children's Health, Shanghai, China

17 : 40~18 : 30 Poster 6 : Mitochondrial disorders

Chairpersons: Hiromi Nyuzuki

(Department of Pediatrics, Niigata University Medical and Dental Hospital)

Chika Takano-Asai

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

P-51 Early Diagnosis of ECHS1 Deficiency in an Infant with Hearing Loss and Developmental Delay

○ Ayuko Usami¹, Tomoko Lee¹, Yoko Yokoyama¹, Eisuke Terasaki^{1,2}, Miki Matsui¹

Naoko Taniguchi¹, Hideki Shimomura¹, Hironori Kobayashi³, Yuki Hasegawa⁴, Kei Murayama^{5,6}

Yasuhiro Takeshima¹

¹Department of Pediatrics, Hyogo Medical University

²Department of Pediatrics, Kawanishi Municipal Hospital

³Laboratories Division, Shimane University Hospital

⁴Department of Pediatrics, Matsue Red Cross Hospital

⁵Diagnostics and Therapeutics of Intractable Disease, Intractable Disease Research Center, Juntendo University Graduate School of medicine

⁶Pediatrics and Adolescent Medicine, Juntendo University Graduate School of medicine

P-52 Efficacy of Multimodal Metabolic Interventions in ECHS1 Deficiency: A Case of a 6-Year-Old Girl

○ Tetsuya Yamashita, Munetsugu Hara, Keigo Matsuyuki, Daisuke Yamashita, Ryuta Ishi

Takayoshi Koike, Ryuta Takase, Kaori Fukui, Yoriko Watanabe, Tatsuki Mizuochi

The Department of Pediatrics and Child Health, Kurume University School of Medicine

P-53 Age-Stratified Blood and Urinary mtDNA Heteroplasmy in Mitochondrial Disease Patients

○ Atsuko Okazaki¹, Yukiko Yatsuka¹, Takuya Fushimi², Takanori Onuki², Yuko Aida², Hisato Aihara³

Toshiyuki Imasawa^{1,4}, Akira Ohtake⁵, Kei Murayama^{1,2}, Yasushi Okazaki¹

¹Department of Diagnostics and Therapeutics of Intractable Diseases, Juntendo University, Graduate School of Medicine

²Department of Metabolism, Chiba Children's Hospital

³Department of Pediatrics, Juntendo University Hospital

⁴Department of Nephrology, Juntendo University Faculty of Medicine

⁵Department of Pediatrics and Clinical Genomics, Saitama Medical University

P-54 Clinical spectrum, heteroplasmy-phenotype correlation, and prognosis of the MT-ND3 m.10191T>C mutation

○ Zimeng He¹, Huafang Jiang², Tongyue Li¹, Yang Liu¹, Ying Zou¹, Chaolong Xu¹, Zhimei Liu¹

Fang Fang¹

¹Department of Neurology, Beijing Children's Hospital, Capital Medical University, National Center for Children's Health, Beijing, China

²Department of Pediatrics, Weifang Maternal and Child Health Hospital, Weifang, Shandong, China

P-55 Phenotypic Spectrum, Genetic Spectrum, Genotype-Phenotype Correlation, and Prognosis of MELAS/LS Overlap Syndrome

○ Zimeng He¹, Tongyue Li², Huafang Jiang¹, Yang Liu¹, Ying Zou¹, Chaolong Xu¹, Zhimei Liu¹

Fang Fang¹

¹Department of Neurology, Beijing Children's Hospital, Capital Medical University, National Center for Children's Health, Beijing, China

²Department of Pediatrics, Weifang Maternal and Child Health Hospital, Weifang, Shandong, China

P-56 Improving the Diagnostic Yield of Mitochondrial Disease Through Genomic Reanalysis: A Single-Center Retrospective Cohort Study

○ Ying Zou, Zhimei Liu, Minhan Song, Zimeng He, Xin Duan, Chaolong Xu, Fang Fang

Department of Neurology, National Center for Children's Health, Beijing Children's Hospital, Capital Medical University, Beijing, China

P-57 Clinical Features and Prognosis of MELAS Based on Multivariate Scoring and Preliminary Modification of Scoring Model

○ Zixuan Zhang, Fang Fang

Beijing Children's Hospital Capital Medical University, Beijing, China

Day 2 : Oct. 16 (Fri.) 4F Poster Venue2 (413+414)

17 : 40~18 : 30 Poster 7 : Lysosomal storage disorders 2 (Fabry diseases)

Chairpersons: Hirotake Sawada

(Department of Nursing, Faculty of Medicine, University of Miyazaki)

Nobuyuki Ishige

(Division of Newborn Screening, Tokyo Health Service Association)

P-58 A Case of Fabry Disease Complicated by Infective Endocarditis

- Minami Ozawa¹, Ohsuke Migita^{2,3}, Satoshi Kinebuchi⁴, Daijun Tomimoto⁴, Kan Nawata⁴
Keisuke Kida⁵

¹Department of Pediatrics, St. Marianna University School of Medicine

²Department of Genetic Medicine, St. Marianna University School of Medicine

³Department of Clinical Laboratory Medicine and Genetic Analysis, St. Marianna University School of Medicine

⁴Department of Cardiovascular Surgery, St. Marianna University School of Medicine

⁵Department of Pharmacology, St. Marianna University School of Medicine

P-59 Dual diagnosis of Fabry disease and Hypophosphatasia in an adult case with generalized body pain

- Masaru Shimura, Tomonori Sase, Takehiro Homma, Keiko Ichimoto
Department of Metabolism, Chiba Children's Hospital

P-60 Improved LVH but persistent proteinuria and eGFR decline in young female heterozygous Fabry disease

- Naho Iijima¹, Takao Kato², Tomohi Ajima², Yu-Sureshuwaru Shin², Ryo Naito², Keiko Goto^{2,3}
Hidemori Hayashi², Tohru Minamino²

¹Clinical Training Center, Juntendo University Hospital

²Department of Cardiovascular Biology and Medicine, Juntendo University Graduate School of Medicine

³Clinical Genetics, Juntendo University Faculty of Medicine

P-61 Newborn screening for Fabry disease in Japan: an additional 3-year report

- Takaaki Sawada¹, Jun Kido², Keishin Sugawara², Shinichiro Yoshida³, Takahito Inoue⁴
Kimitoshi Nakamura²

¹Center for Clinical Genetics, Kumamoto University Hospital

²Department of Pediatrics, Faculty of Life Sciences, Kumamoto University, ³KM Biologics Co., Ltd.

⁴Center for Maternal, Fetal and Neonatal Medicine, Fukuoka University Hospital

P-62 followME Fabry registry: ≥5-year cardiac, renal and multisystem effectiveness with migalastat

- Yasushi Ozeki¹, Ales Linhart², Antonio Pisani³, Olga Azevedo⁴, Haichen Yang⁵, Jennifer Hiros⁵
Vipul Jain⁵, Biliانا O Veleva-Rotse⁵, Derralynn Hughes⁶, Peter Nordbeck⁷

¹Medical Affairs, Amicus Therapeutics K.K

²Second Department of Internal Cardiovascular Medicine, First Faculty of Medicine, Charles University and General University Hospital, Prague, Czechia

³Department of Public Health, Nephrology Unit, Federico II University Hospital, Naples, Italy

⁴Cardiology Department, Reference Center of Lysosomal Storage Disorders, MetabERN Member, Hospital Senhora da Oliveira, Guimarães, Portugal

⁵Amicus Therapeutics, Inc., Princeton, NJ, USA

⁶Royal Free London NHS Foundation Trust, and University College London, London, UK

⁷Internal Medicine I, University Hospital Würzburg, Würzburg, Germany

P-63 Automated Mulberry Body Detection by Imaging Flow Cytometry with Autofluorescence and Deep Learning

- Kazuya Tsuboi¹, Akinori Masago², Mariko Akita², Honoka Hashimoto², Taichi Fukumori²
Gupta Shubham², Akihito Yashima^{1,3}, Momoko Imakubo²

¹Lysosomal Storage Diseases Center, Nagoya Central Hospital, ²Sysmex Corporation

³Nephrology, Nagoya Central Hospital

P-64 Development and Pilot Testing of a Fabry Disease Screening Tool Using the Health Support App

- Hiroyuki Yamakawa^{1,2}, Yasuyuki Fukuhara³, Naoyuki Kosaka⁷, Eiichi Kiyokawa⁸
Tomoaki Takahashi⁸, Hiroshi Kobayashi⁴, Masahisa Kobayashi⁵, Torayuki Okuyama⁶
Masaki Ieda², Hiromasa Takaishi¹

¹Center for Preventive Medicine, School of Medicine, Keio University

²Department of Cardiology, School of Medicine, Keio University

³National Center for Child Health and Development, Center for Genetic Medicine

⁴Jikei University School of Medicine, Department of Gene Therapy Research

⁵Jikei University School of Medicine, Department of Pediatrics

⁶Saitama Medical University, Department of Genomic Medicine, ⁷Medipal Holdings Co., Ltd.

⁸MTI Co., Ltd.

P-72 Screening and Registry of Hereditary Urolithiasis Using Urinary, GC/MS Analysis

- Seiji Tanaka¹, Tatsuya Takayama³, Yoriko Watanabe^{1,2}

¹Department of Pediatrics and Child Health, Kurume University

²Research Institute of Medical Mass Spectrometry, Kurume University School of Medicine

³Department of Urology, International University of Health and Welfare Hospital

17 : 40~18 : 30 Poster 8 : Lysosomal storage disorders 3 (Pompe diseases)

Chairperson: Tomoko Lee

(Department of Pediatrics, Hyogo Medical University)

P-65 Central nervous system manifestations of Pompe disease detected by newborn screening

- Yu Nakashima¹, Nozomi Abe¹, Kanako Ishii¹, Takaaki Sawada², Kimitoshi Nakamura³
Yasunari Sakai¹

¹Department of Pediatrics, Kyushu University Hospital

²Center for Clinical Genetics, Kumamoto University Hospital

³Department of Pediatrics, Faculty of Life Sciences, Kumamoto University

- P-66 Early ERT before CRIM confirmation in NBS-detected infantile-onset Pompe disease**
 ○ Yoshimi Fujita, Toshiki Tsunogai, Masahisa Kobayashi, Kimihiko Oishi
 The Jikei University School of Medicine, Department of Pediatrics
- P-67 Two Years Follow-Up of Infantile-Onset Pompe Disease Case Treated with Avalglucosidase Alfa**
 ○ Chizuko Nakamura, Hiroaki Tadokoro, Kohei Kurosaki, Kota Takei, Keisuke Nagasaki
 Nagano Children's Hospital
- P-68 Update on FORTIS: A phase 1/2 trial of AT845 gene replacement therapy in late-onset Pompe disease**
 ○ Chieri Hayashi¹, Jordi Diaz-Manera², Nicola Longo³, Mark Walzer¹, Gurunadh Chichili¹, Sean Yiu⁴
 Julie Porter⁵, David Viskochil⁵, Tahseen Mozaffar⁶
¹Astellas Pharma Global Development, Northbrook, IL, USA
²John Walton Muscular Dystrophy Research Centre, Newcastle University and Newcastle Hospitals NHS Foundation Trust, Newcastle upon Tyne, UK
³University of California Los Angeles, Los Angeles, CA, USA
⁴Astellas Pharma Limited, Addlestone, UK, ⁵University of Utah, Salt Lake City, UT, USA
⁶University of California Irvine, Irvine, CA, USA
- P-69 Safety and effectiveness of avalglucosidase alfa in Japanese patients with Pompe disease: PMS result**
 ○ Mitsunobu Ikeda, Hiroyuki Kameda, Takeshi Seto, Makiko Hatanaka
 Sanofi K.K.
- P-70 Comparison of the affinity of M6P receptor for two recombinant human acid alpha-glucosidase drugs**
 ○ Takahiro Tsukimura¹, Koki Morita¹, Tomoko Shiga², Chieri Takashima¹, Ayame Hiwatari¹
 Kiyoharu Kawakami¹, Tadayasu Togawa³, Hitoshi Sakuraba², Kazuhiko Nakadate¹
¹Department of Functional Morphology, Meiji Pharmaceutical University
²Department of Clinical Genetics, Meiji Pharmaceutical University
³Department of Functional Bioanalysis, Meiji Pharmaceutical University
- P-71 Long-term (4-year) efficacy and safety of cipa+mig in adults with LOPD treated from PROPEL baseline**
 ○ Hiroshi Kobayashi¹, Priya S Kishnani², Kristl G Claeys³, Jordi Díaz-Manera⁴, Mark Roberts⁵
 Benedikt Schoser⁶, Antonio Toscano⁷, Nadine A M E Van Der Beek⁸, Fred Holdbrook⁹
 Tahseen Mozaffar¹⁰
¹Division of Gene Therapy, Research Center for Medical Sciences, The Jikei University School of Medicine
²Duke University Medical Center, Durham, NC, USA
³Department of Neurology, University Hospitals Leuven, and Department of Neurosciences, Laboratory for Muscle Diseases and Neuropathies, KU Leuven, Leuven Brain Institute (LBI), Leuven, Belgium
⁴John Walton Muscular Dystrophy Research Centre, Newcastle University, Newcastle upon Tyne, UK
⁵Salford Royal NHS Foundation Trust, Salford, UK
⁶Friedrich-Baur-Institute at the Department of Neurology, LMU University Hospital, LMU Munich, Munich, Germany
⁷ERN-NMD Center for Neuromuscular Disorders of Messina, Department of Clinical and Experimental Medicine, University of Messina, Messina, Italy
⁸Erasmus MC University Medical Center, Rotterdam, Netherlands
⁹Amicus Therapeutics, Inc., Princeton, NJ, USA
¹⁰Department of Neurology, University of California, Irvine, CA, USA

Day 3 : Oct. 17 (Sat.) 5F Venue1 (501)

8 : 10~9 : 00 Morning Seminar 2 Sponsored by Ultragenyx Japan K.K.

Chairperson: Tokiko Fukuda

(Hamamatsu Child Health and Developmental Medicine,
Hamamatsu University School of Medicine)

MS2 Glycogen Storage Disease Type Ia—Characteristics and Disease Management—

○ Kimitoshi Nakamura

Department of Pediatrics, Graduate School of Medical Sciences, Kumamoto University

9 : 10~10 : 00 Special Lecture 3

Chairperson: Hitoshi Osaka

(Department of Pediatrics, Jichi Medical University, School of
Medicine)

SL3 A New Era of Mitochondrial Medicine: From Diagnosis to Precision Medicine

○ Kei Murayama

Department of Pediatrics, Diagnostics and Therapeutics of Intractable Disease, Intractable Disease
Research Center, Juntendo University Faculty of Medicine

10 : 10~12 : 00 JSIMD-JSGCT Joint Session : Gene therapy symposium

Chairpersons: Hiroshi Kobayashi

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

Kazuhiro Muramatsu

(Pediatrics, Jichi Medical University/The Center for Gene Therapy
Research, Jichi Medical University)

GT-1 The Dawn of Gene Therapy in Japan

○ Yoshikatsu Eto

Advanced Clinical Research Center, Southern Tohoku Institute for Neuroscience

GT-2 JSGCT Initiatives Toward the Proper Dissemination of Gene Therapy in Japan

○ Yoshikazu Yonemitsu

Kyushu University Graduate School of Pharmaceutical Sciences

GT-3 Gene and Regenerative Therapies for Inherited Metabolic Disorders: Advances and Perspectives

○ Shirou Matsumoto^{1,2}, Tomoko Nakanishi³

¹Department of Neonatology, Kumamoto University Hospital

²Department of Pediatrics, Graduate School of Life Sciences, Kumamoto University

³Center of Biomedical Research Resources, Juntendo University School of Medicine

GT-4 Research and development of ex vivo gene therapy for lysosomal storage diseases

○ Yohta Shimada

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

GT-5 Gene Therapy for Inherited Metabolic & Neurological Diseases: Advances, Challenges & Next-Gen Models

○ Kazuhiro Muramatsu^{1,2}

¹Pediatrics, Jichi Medical University

²The Center for Gene Therapy Research, Jichi Medical University

GT-6 Patient-Initiated Development and Nucleic Acid Drugs—Who Develops Drugs for Ultra-Rare Diseases?

○ Yoshiyuki Saito

Institute of Science Tokyo

12 : 10~13 : 00 Luncheon Seminar 5

Sponsored by Takeda Pharmaceutical Company Limited

Chairperson: Hiroshi Kobayashi

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

LS5 Current Status and Challenges in Adult Fabry Disease Care: A Cardiologist's Perspective

○ Yasuhiro Izumiya

Department of Cardiovascular Medicine, Graduate School of Medical Sciences, Kumamoto University

13 : 10~14 : 00 Oral 10 : Lysosomal storage disorders 2

Chairpersons: Toshiyuki Seto

(Department of Medical Genetics, Osaka Metropolitan University, Graduate School of Medicine)

Motomichi Kosuga

(Division of Medical Genetics, National Center for Child Health and Development)

O10-1 Therapeutic strategies for Mucopolidosis II and Niemann-Pick C2 using M6PR/IGF2R double KO cells

○ Seigo Terawaki, Takanobu Otomo

Department of Molecular and Genetic Medicine, Kawasaki Medical School

O10-2 LEAP2MONO: Venglustat shows neurologic efficacy with systemic control in Gaucher disease type 3

○ Mengel Eugen¹, Irene Motta², Fatma Derya Bulut³, Cheryl Rockman-Greenberg⁴, Anais Brassier⁵, Zhengqing Qiu⁶, Dmitry Cherkasov⁷, Atef Zaher⁸, Qi Zhang⁹, Pramod K. Mistry¹⁰

¹Institute of Clinical Science for Lysosomal Storage Disorders, SphinCS GmbH, Hochheim, Germany

²Unit of Medicine and Metabolic Disease, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milan, Italy

³Cukurova University, Pediatric Metabolism and Nutrition Department, Adana, Turkey

⁴Department of Pediatrics and Child Health, Rady Faculty of Health Sciences, University of Manitoba, Winnipeg, MB, Canada

⁵Reference Center for Inherited Metabolic Diseases, Necker Enfants Malades University Hospital, Assistance Publique Hopitaux de Paris (AP-HP), Paris Cite University, Paris, France

⁶Peking Union Medical College Hospital, Beijing, China

⁷Sanofi, Chilly-Mazarin, Ile-de-France, France, ⁸Sanofi, Toronto, ON, Canada

⁹Sanofi, Morristown, NJ, United States, ¹⁰Yale University School of Medicine, New Haven, CT, USA

010-3 Cytokine Dynamics during Olipudase Alfa Dose Escalation in Acid Sphingomyelinase Deficiency

- Shuhei Sako¹, Eri Imagawa¹, Yasuyo Kashiwagi², Tomoko Kasanuki¹, Gaku Yamanaka²
Kimihiko Oishi¹

¹Department of Pediatrics, The Jikei University School of Medicine

²Department of Pediatrics and Adolescent Medicine, Tokyo Medical University

010-4 Utility of Urinary Glycosaminoglycan Analysis by LC-MS/MS for Diagnosing Mucopolysaccharidosis II

- Tsubasa Oguni, Hironori Kobayashi, Shozo Yano
Laboratories Division, Shimane University Hospital

010-5 Biomarker-Based Evaluation of GLA Variants Detected by Newborn Screening for Fabry Disease

- Jun Kido¹, Takaaki Sawada^{1,2}, Takahiro Tsukimura³, Keishin Sugawara¹, Tadayasu Togawa³
Hitoshi Sakuraba⁴, Takahito Inoue⁵, Yoriko Watanabe⁶, Junpei Hamada⁷, Kimitoshi Nakamura^{1,2}

¹Department of Pediatrics, Faculty of Life Sciences, Kumamoto University

²Center for Clinical Genetics, Kumamoto University Hospital

³Department of Functional Bioanalysis, Meiji Pharmaceutical University

⁴Department of Clinical Genetics, Meiji Pharmaceutical University

⁵Department of Pediatrics, School of Medicine, Fukuoka University

⁶Department of Pediatrics and Child Health, Kurume University School of Medicine

⁷Department of Pediatrics, Ehime University Graduate School of Medicine

**14 : 15~15 : 55 Long-Term Planning Committee Young Researchers Session
Sponsored by PTC Therapeutics K.K.**

Chairpersons: Yoko Nakajima

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

Motomichi Kosuga

(Division of Medical Genetics, National Center for Child Health and Development)

YR Future Planning Committee Workshop From Guidelines to Table: A Hands-On Workshop on Team-Based Nutritional Management of PKU

- Hiroki Fujimoto¹, Ryosuke Bo², Satoru Meiri³, Ryuta Takase⁴, Toshiki Tsunogai⁵, Miki Matsui⁶
Satoko Tsushima⁷, Kazuhiro Watanabe⁸

¹Department of Nutrition, Osaka Metropolitan University Hospital

²Department of Pediatrics, Hyogo Medical University Hospital

³Department of Pediatrics, School of Medicine Miyazaki University

⁴Department of Pediatrics, Kurume University School of Medicine

⁵Department of Pediatrics, The Jikei University School of Medicine

⁶Department of Pediatrics, Hyogo Medical University

⁷Department of Nursing and Medical Genetics, National Center for Child Health and Development

⁸Division of Newborn Screening, Tokyo Health Service Association

Day 3 : Oct. 17 (Sat.)

4F Venue2 (411+412)

9 : 10~10 : 00 **Sponsored Seminar 4**

Sponsored by Clinigen K.K.

Chairperson: Torayuki Okuyama

(Program for Promotion of Genomic Medicine for Rare Diseases,
Department of Genomics Medicine, Saitama Medical University)

SPSE4-1 The optimal timing of HCT for IEM in the era of expanded NBS

○ Katsuhide Eguchi

Department of Pediatrics, Graduate School of Medical Sciences, Kyushu University

SPSE4-2 From ERT to Hematopoietic Cell Transplantation: New Perspectives in the Treatment of MPS Type II

○ Takuro Nishikawa

Creation of Community Pediatric Medicine, Graduate School of Medical and Dental Sciences,
Kagoshima University

10 : 10~11 : 00 Oral 8 : Mitochondrial disorders

Chairpersons: Ayako Matsunaga

(Department of Pediatrics, St. Marianna University School of
Medicine)

Chikahiko Numakura

(Department of Clinical Genomics & Pediatrics, Saitama Medical
University)

O8-1 A Neonatal Scoring System for Early Genomic Triage in Suspected Mitochondrial Disease

○ Tomohiro Ebihara¹, Taro Nagatomo², Tomoko Tsuruoka¹, Hironori Shimoza³

Yasushi Okazaki^{4,5}, Kei Murayama^{4,6}

¹Department of Neonatology, Chiba Children's Hospital

²Department of Neonatology, Japanese Red Cross Fukuoka Hospital

³Department of Pediatrics, Jichi Medical University

⁴Diagnostics and Therapeutics of Intractable Diseases, Intractable Disease Research Center,
Juntendo University, Graduate School of Medicine

⁵Laboratory for Comprehensive Genomic Analysis, RIKEN Center for Integrative Medical Sciences

⁶Department of Pediatrics, Juntendo University Faculty of Medicine

O8-2 Targeting mitochondrial succinate transport to prevent cardiac ischaemia/reperfusion injury

○ Jiro Abe^{1,2,3}, Hiran Prag³, Mike Murphy³, Thomas Krieg³

¹Pediatric Genetics and Metabolism, Hokkaido Medical Center

²Department of Pediatrics, Graduate School of Medicine, Hokkaido University

³MRC Mitochondrial Biology Unit, University of Cambridge, Cambridge, UK

08-3 Decoding the Dark Genome Region in Mitochondrial Disease

- Kohta Nakamura¹, Yoshihito Kishita¹, Ayumu Sugiura¹, Atsuko Okazaki¹, Yukiko Yatsuka¹
Naoyuki Matsumoto¹, Hiroshi Matsumoto^{2,3}, Akira Ohtake^{2,3}, Kei Murayama¹, Yasushi Okazaki¹
¹Diagnostics and Therapeutics of Intractable Diseases, Intractable Disease Research Center,
Graduate School of Medicine, Juntendo University
²Department of Pediatrics, Saitama Medical University
³Department of Clinical Genomics, Saitama Medical University

08-4 Phenotypic Spectrum and Diagnostic Mimics of POLG-Related Disorders: A Single-Center Experience

- Victoria Lobanova¹, Liliya Khayalutdinova¹, Yulia Martynenko¹, Lyudmila Ugolkova¹, Yulia Itkis²
¹Reference Center for Congenital, Hereditary Diseases, Genetic Abnormalities, Orphan and Other
Rare Disorders Morozov's Moscow City Children Clinical Hospital, Moscow, Russia
²Research Centre for Medical Genetics, Moscow, Russia

08-5 Dysfunction of pyruvate dehydrogenase deficiency zebrafish can be partially rescued by glutamine

- Huafang Jiang^{1,2}, Fang Fang¹, Chaolong Xu¹, Ruoyu Duan¹, Danmin Shen¹, Zhimei Liu¹
Ning Zhang²
¹Department of Neurology, Beijing Children's Hospital, Capital Medical University, Beijing, China
²Department of Pediatrics, Weifang Maternal and Child Health Hospital, Weifang, Shandong, China

11 : 10~12 : 00 Oral 9 : Phenylketonuria

Chairpersons: Tetsuya Ito

(Department of Pediatrics, Aichi Gakuin University Dental Hospital)

Atsuko Noguchi

(Department of Pediatrics, Akita University Graduate School of Medicine)

09-1 Rigidity during Parenteral Nutrition in an Adult with Phenylketonuria

- Chika Takano-Asai¹, Erika Ogawa^{1,2}, Naotoshi Natori³, Tomotaka Mizoguchi³
Masahiro Nishihara⁴, Jun Kizuki⁵, Hirotake Inomata⁴, Masahiro Suzuki⁵, Ichiro Morioka¹
Mika Ishige¹
¹Department of Pediatrics and Child Health, Nihon University School of Medicine
²Department of Pediatrics, Tokyo Metropolitan Hiroo Hospital
³Department of Neurology, Nihon University School of Medicine
⁴Department of Hematology and Rheumatology, Nihon University School of Medicine
⁵Department of Psychiatry, Nihon University School of Medicine

09-2 A Single-Center Retrospective Study of PKU and Hyperphenylalaninemia from Childhood to Adulthood

- Ryuta Takase¹, Yusuke Miyagi¹, Kaori Fukui¹, Yoriko Watanabe^{1,2}
¹Department of Pediatrics and Child Health, Kurume University School of Medicine
²Research Institute of Medical Mass Spectrometry, Kurume University School of Medicine

09-3 Questionnaire survey for new treatment of adult patients with classocal PKU:final report

- Mahoko Furujo, Yousuke Higuchi, Shintaro Fujiwara
Department of Pediatrics, NPO Okayama Medical Center

09-4 Efficacy and safety of pegvaliase treatment in 23 adult patients with PKU: 3 years of experience

○ Erika Ogawa^{1,2}, Chika Takano-Asai¹, Rintaro Furuta¹, Naoko Okamura³, Ichiro Morioka¹

Mika Ishige¹

¹Department of Pediatrics and Child Health, Nihon University School of Medicine

²Department of Pediatrics, Tokyo Metropolitan Hiroo Hospital

³Division of Nutritional Management, Nihon University Hospital

09-5 Effect of long-term sepiapterin treatment in participants with phenylketonuria aged below 2 years

○ Tomoko Bessho¹, Laura Nagy², Suzanne Hollander³, Soraia Poloni⁴, Kimberly Ingalls⁵, Neil Smith⁵

Catalina Hughes⁵, Alexandra Larkin⁵, Anita Macdonald⁶

¹PTC Therapeutics K.K.

²Department of Clinical Dietetics, The Hospital for Sick Children, Toronto, Canada

³Department of Clinical Nutrition, Boston Children's Hospital, Boston, USA

⁴Hospital de Clinicas de Porto Alegre, Porto Alegre, Brazil

⁵PTC Therapeutics, Warren, USA

⁶Dietetic Department, Birmingham Children's Hospital, Birmingham, UK

12 : 10~13 : 00 Luncheon Seminar 6 : From Diagnosis to Treatment in Lysosomal Storage Disorders: Real-World Clinical Perspectives on Pompe Disease and Fabry Disease

Sponsored by Amicus Therapeutics, K.K.

Chairperson: Kimitoshi Nakamura

(Department of Pediatrics, Graduate School of Medical Sciences,
Kumamoto University)

LS6-1 Pompe Disease: Diagnosis and New Therapeutic Advances

○ Keiko Ishigaki

Department of Pediatrics, Tokyo Women's Medical University

LS6-2 Practical management for Fabry disease: Perspective on clinical options

○ Masahisa Kobayashi

Department of Pediatrics, The Jikei University School of Medicine

13 : 10~14 : 00 Sweets Seminar 6

Sponsored by ReqMed Company, Ltd

Chairperson: Tatsuki Mizuochi

(Department of Pediatrics and Child Health, Kurume University)

SWSE6 Recognizing IEBAM in Infantile Cholestasis: From Diagnostic Clues to Precision Therapy

○ Mitsuyoshi Suzuki

Department of Pediatrics, Juntendo University Shizuoka Hospital