

## **Day 2 : Nov. 8 (Fri.) Venue 1 (5F 501AB)**

### **8 : 10~9 : 00 Morning Seminar 1**

**Sponsored by Biomarin Pharmaceutical Japan K.K.**  
**Chairperson: Hironori Kobayashi**  
(Clinical Laboratory Division, Shimane University Hospital)

#### **MS1 Progress of the research on next-generation expanded newborn screening supported by AMED and Children and Families Agency**

- Go Tajima  
Division of Neonatal Screening, Research Institute, National Center for Child Health and Development

### **9 : 20~10 : 20 Special Lecture**

**Chairperson: Kimihiko Oishi**  
(Department of Pediatrics, The Jikei University School of Medicine)

#### **SP Leveraging Education in Inborn Errors of Metabolism to Support the Workforce and Patient Care**

- Debra Sue Regier<sup>1</sup>, Christine L Maccia<sup>2</sup>  
<sup>1</sup>Chief, Genetics and Metabolism Interim Director, Children's National Rare Disease Institute  
Children's National Hospital  
<sup>2</sup>Genetics and Metabolism Children's National Rare Disease Institute Children's National Hospital

### **10 : 30~12 : 00 Special Symposium**

**Chairpersons: Mika Ishige**  
(Department of Pediatrics and Child Health, Nihon University School of Medicine)  
**Takashi Hamazaki**  
(Department of Pediatrics, Osaka Metropolitan University Graduate School of Medicine)

#### **SS0 Opening Remarks**

- Mika Ishige  
Department of Pediatrics and Child Health, Nihon University School of Medicine

#### **SS1 Collaborative Management of Inborn Errors of Metabolism: The Integral Role of Dietitians in New York**

- Ilona Ginevic  
Icahn School of Medicine at Mount Sinai, New York, NY

#### **SS2 Dietitian's efforts for patients with inborn error of metabolism at our hospital—Focusing on PKU—**

- Hiroki Fujimoto  
Osaka Metropolitan University Hospital, Department of Nutrition

**SS3 The role of dietitians in the care of patients with phenylketonuria at Nihon university Hospital**

- Naoko Okamura  
Nutritional Management Division, Nihon University Hospital

**SS4 Adding Phe-free amino acid or low Phe peptide to Phe-free formula for treatment of PKU children**

- Erika Ogawa<sup>1,2</sup>, Chika Takano<sup>1,3</sup>, Naoko Okamura<sup>4</sup>, Ichiro Morioka<sup>1</sup>, Mika Ishige<sup>1</sup>  
<sup>1</sup>Department of Pediatrics and Child Health, Nihon University School of Medicine  
<sup>2</sup>Department of Pediatrics, Tokyo Metropolitan Hiroo Hospital  
<sup>3</sup>Division of Microbiology, Department of Pathology and Microbiology, Nihon University School of Medicine  
<sup>4</sup>Nutrition Management Division, Nihon University Hospital

**12 : 30~13 : 20 Lancheon Seminar 3**

**Sponsored by Sumitomo Pharma Co., Ltd.**

**Chairperson: Kimitoshi Nakamura**

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

**LS3 Fabry disease: Intracellular uptake of drugs for enzyme replacement therapy and formation of antidrug antibodies**

- Hitoshi Sakuraba  
Department of Clinical Genetics, Meiji Pharmaceutical University

**13 : 40~14 : 10 SLEIMPN Resommending Lecture**

**Chairperson: Masahisa Kobayashi**

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

**SLEIMPN Machine learning and metabolomics: new approaches for early detection of liver complications in Tyrosinemia Type-1**

- Karen Fuenzalida<sup>1</sup>, Maria Jesus Leal-Witt<sup>1</sup>, Alejandro Acevedo<sup>1</sup>, Carolina Arias<sup>1</sup>  
Juan Francisco Cabello<sup>1</sup>, Giancarlo La Marca<sup>2</sup>, Cristiano Rizzo<sup>3</sup>, Carlo Dionisi-Vici<sup>3</sup>  
Veronica Cornejo<sup>1</sup>  
<sup>1</sup>Institute of Nutrition and Food Technology, University of Chile  
<sup>2</sup>Newborn Screening, Clinical Chemistry and Pharmacology Laboratory, Meyer Children's Hospital IRCCS, Florence, Italy  
<sup>3</sup>Division of Metabolism and Metabolic Diseases Research Unit, Bambino Gesù Children's Hospital, IRCCS, Rome, Italy

## 14 : 20~14 : 50 KSIMD Recommending Lecture

**Chairperson: Yoriko Watanabe**

(Research Institute of Medical Mass Spectrometry, and Department.  
Pediatrics and Child Health Kurume University School of  
Medicine)

### **KSIMD Development of intracerebroventricular recombinant human Heparan-N-Sulfatase enzyme replacement therapy in MPSIIIA**

- Young Bae Sohn<sup>1</sup>, Aram Yang<sup>2</sup>, Jinsup Kim<sup>3</sup>, Ah-ra Ko<sup>3</sup>, Yeongju Yu<sup>3</sup>, Inyoung Jo<sup>3</sup>

Hyeongseok Uhm<sup>3</sup>, Sujeong Kim<sup>4</sup>, Sora Kim<sup>4</sup>, Dong-Kyu Jin<sup>3</sup>

<sup>1</sup>Department of Medical Genetics, Ajou University Hospital, Ajou University School of Medicine, Suwon, Republic of Korea

<sup>2</sup>Department of Pediatrics, Kangbuk Samsung Hospital, Sungkyunkwan University, School of Medicine, Seoul, Republic of Korea

<sup>3</sup>Novel Pharma, Inc., Seoul, Republic of Korea

<sup>4</sup>GC Biopharma Corp., Yongin, Republic of Korea

## 15 : 00~16 : 30 CD Symposium

**Chairpersons: Kimitoshi Nakamura**

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

**Kimihiko Oishi**

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

### **CD-S1 Advancements in Novel Therapies and New Cellular Models for Citrin Deficiency**

- Barbara Yu, Li Eon Kuek  
Citrin Foundation

### **CD-S2 The roles of NAD<sup>+</sup> concentration and redox state in a mouse model of citrin deficiency**

- Joseph Anthony Baur, David W Frederick, Thato T'solo, Rishith Ramamurthy, James G Davis  
Department of Physiology and Institute for Diabetes, Obesity, and Metabolism, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA, USA

### **CD-S3 The latest development in messenger RNA therapy and its application for CD**

- Julien Baruteau<sup>1,2</sup>  
<sup>1</sup>University College London, London, UK  
<sup>2</sup>Great Ormond Street Hospital for Children, London, UK

### **CD-S4 New cellular models for better understanding of therapeutic interventions in CD**

- Jun Kido<sup>1</sup>, Johannes Häberle<sup>2</sup>, Kimitoshi Nakamura<sup>1</sup>  
<sup>1</sup>Department of Pediatrics, Faculty of Life Sciences, Kumamoto University  
<sup>2</sup>University Children's Hospital Zurich and Children's Research Centre, University of Zurich

## 16 : 40~17 : 10 SIMD Recommending Lecture

Chairperson: Tetsuya Ito

(Department of Pediatrics, Fujita Health University)

### **SIMD CERAMIDE – THE UNMASKED DRIVER OF HEART FAILURE IN VERY LONG-CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY (VLCADD)**

- Marie Kristine Norris<sup>1</sup>, Melanie B. Gillingham<sup>2</sup>, Nicola Longo<sup>1</sup>, Christina Lam<sup>3</sup>, Matthew P Yim<sup>1</sup>  
Mary C Playdon<sup>1</sup>, Ralph J DeBerardinis<sup>4</sup>, Jerry Vockley<sup>5</sup>, William L. Holland<sup>1</sup>, Scott A. Summers<sup>1</sup>  
<sup>1</sup>University of Utah  
<sup>2</sup>Oregon Health and Science University  
<sup>3</sup>Seattle Children's Hospital  
<sup>4</sup>University of Texas Southwestern  
<sup>5</sup>University of Pittsburgh

## 17 : 20~18 : 10 Oral 9 : Tomatsu session

Chairperson: Ken Sakurai

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

### **T-1 Accelerating Medicines Partnership Bespoke Gene Therapy Consortium for Rare Disorders: mucopolysaccharidosis IVA**

- Shunji Tomatsu, Shunji Mackenzie, Kimberly Klipner, Allison Bradford  
Nemours Children's Health

### **T-2 Assessment of an iron oxide-coupled CRISPR/nCas9 gene editing in a mucopolysaccharidosis IVA mouse model**

- Shunji Tomatsu, Andres Leal, Fnu Nidhi, Khan Shaukat  
Nemours Children's Health

### **T-3 Lentiviral Vector-Mediated *Ex Vivo* Hematopoietic Stem Cell Gene Therapy for mucopolysaccharidosis IVA Murine Model**

- Shunji Tomatsu, Celik Betul, Fnu Nidhi, Khan Shaukat  
Nemours Children's Health

### **T-4 Immune tolerance to GALNS enhances the therapeutic efficacy of AAV gene therapy**

- Shunji Tomatsu, Sampurna Saikia, Yasuhiko Ago, Khan Shaukat  
Nemours Children's Health

### **T-5 Adeno-associated virus-based gene therapy delivering combinations of two growth-associated genes to MPS IVA mice**

- Shunji Tomatsu, Estera Rintz, Celik Betul, Khan Shaukat  
Nemours Children's Health