

# DAY3 : Oct.14 (Sat)

## Hall 1

8 : 30 ~ 9 : 18 Oral13 : Peroxisomal disorder & others

Chairperson: Nobuyuki Shimozawa

(Division of Genomics Research Life Science Research Center,  
Gifu University)

Masahiko Kawai

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

**O-46 The first Japanese case of rhizomelic chondrodysplasia punctata type 2**

○Hiromi Nyuzuki<sup>1</sup>, Keisuke Nagasaki<sup>1</sup>, Reiko Murakami<sup>2</sup>, Nao Shibata<sup>1</sup>, Sunao Sasaki<sup>1</sup>  
Yohei Ogawa<sup>1</sup>, Hiroto Saito<sup>3</sup>, Gen Nishimura<sup>4</sup>, Shigeo Takashima<sup>5</sup>  
Nobuyuki Shimozawa<sup>5</sup>

<sup>1</sup>Department of Pediatrics, Niigata University

<sup>2</sup>Department of Orthopedic Surgery, Niigata University

<sup>3</sup>Department of Biochemistry, Hamamatsu University School of Medicine

<sup>4</sup>Intractable Disease Center, Saitama Medical University Hospital

<sup>5</sup>Life Science Research Center, Gifu University

**O-47 Simultaneous analysis of peroxisomal metabolites using liquid-chromatography mass-spectrometry**

○Shigeo Takashima, Kayoko Toyoshi, Akiko Ohba, Shoko Takemoto, Nobuyuki Shimozawa  
Division of Genomics Research, Life Science Research Center, Gifu University

**O-48 Quantitative analysis of very long chain fatty acyl-CoA in ABCD1-deficient cells**

○Kotaro Hama, Yuko Fujiwara, Atsushi Yamashita, Kazuaki Yokoyama  
Faculty of Pharmaceutical Sciences, Teikyo University

**O-49 Four Chinese children with congenital generalized lipodystrophy and novel mutations of *BSCL2* and *AGPAT2* genes**

○Yi Liu<sup>1</sup>, Dongxiao Li<sup>1</sup>, Xiyuan Li<sup>1</sup>, Yuan Ding<sup>1</sup>, Ying Jin<sup>1</sup>, Jinqing Song<sup>1</sup>, Haixia Li<sup>2</sup>  
Hongyun Yan<sup>2</sup>, Yanling Yang<sup>1</sup>

<sup>1</sup>Department of Pediatrics, Peking University First Hospital

<sup>2</sup>Department of Clinical Laboratory, Peking University First Hospital

**9 : 20 ~ 10 : 50 Symposium 4: Novel Therapeutic Approach for Inherited and Metabolic Diseases**

**Organizer: Torayuki Okuyama**

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

**Chairperson: Torayuki Okuyama**

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

**Fumio Endo**

(Kumamoto-Ezuko Medical Center for The Severely Disabled)

**S4-1 Chaperone therapy from basic to clinical research**

○Eiji Nanba, Katsumi Higaki

Research Center for Bioscience and Technology, Tottori University

**S4-2 Liver derived cells, stem cells & microparticles to treat inborn errors of metabolism**

Etienne Sokal

Cliniques Universitaires ST Luc, Université Catholique de Louvain and Promethera Biosciences

**S4-3 Phase 3 Long-term Study Evaluating Efficacy and Safety of Pegvaliase Treatment in Adults with PKU**

○Nicola Longo<sup>1</sup>, Jeffery Vockley<sup>2</sup>, Harvey Levy<sup>3</sup>, Stephen Amato<sup>4</sup>, Roberto Zori<sup>5</sup>, Janet Thomas<sup>6</sup>  
Barbara Burton<sup>7</sup>, Cary Harding<sup>8</sup>, John Posner<sup>9</sup>, Deborah Bilder<sup>1</sup>, Joy Olbertz<sup>10</sup>, Zonghua Gu<sup>10</sup>  
Kelly Lau<sup>10</sup>, Mingjin Lin<sup>10</sup>, Kevin Larimore<sup>10</sup>, David Dimmock<sup>11</sup>

<sup>1</sup>University of Utah, Salt Lake City, UT

<sup>2</sup>University of Pittsburgh and Children's Hospital of Pittsburgh, Pittsburgh, PA

<sup>3</sup>Boston Children's Hospital, Boston, MA, <sup>4</sup>University of Kentucky, Lexington, KY

<sup>5</sup>University of Florida, Gainesville, FL, <sup>6</sup>University of Colorado School of Medicine, Aurora, CO

<sup>7</sup>Lurie Children's Hospital of Chicago, Chicago, IL

<sup>8</sup>Oregon Health & Science University, Portland, OR

<sup>9</sup>King's College, London, UK, <sup>10</sup>BioMarin Pharmaceutical Inc., Novato, CA

<sup>11</sup>Rady Children's Institute for Genomic Medicine, San Diego, CA

**11 : 00 ~ 11 : 50 Sponsored Seminar 4**

**Chairperson: Hiroyuki Ida**

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

**Various problems in neurologic type of Gaucher disease.**

Kei Murayama

Department of Metabolism, Chiba Children's Hospital

**Lipid Metabolism Disorders might be associated with Parkinson's Disease**

Taku Hatano

Department of Neurology, Juntendo University School of Medicine

**13 : 10 ~ 14 : 10 Educational Lecture 4**

**Chairperson: Toshihiro Ohura**  
(Sendai City Hospital)

**Mitochondrial Disease Therapies: Translation from simple model animals to bedside**

Marni Falk

The Children's Hospital of Philadelphia and University of Pennsylvania Perelman School of  
Medicine

**14 : 10 ~ Closing Remarks**

# DAY3 : Oct.14 (Sat)

## Hall 2

8 : 30 ~ 9 : 18 Oral14 : Patient Registration & Treatments

Chairperson: Ikue Hata

(Department of Pediatrics, of Faculty Medical Sciences,  
University of Fukui)

**Ken Sakurai**

(Department of Pediatrics, The Jikei University Katsushika  
Medical Center)

**O-50 Characteristics of 28 patients with X-linked adrenoleukodystrophy from the registry system in Japan**

○Ken Sakurai<sup>1</sup>, Toya Ohashi<sup>2</sup>, Seo Joo-Hyun<sup>3</sup>, Torayuki Okuyama<sup>3</sup>, Nobuyuki Shimozawa<sup>4</sup>,  
Hiroyuki Ida<sup>5</sup>

<sup>1</sup>Department of Pediatrics, The Jikei University Katsushika Medical Center

<sup>2</sup>Division of Gene Therapy, Research Center for Medical Science, The Jikei University School  
of Medicine

<sup>3</sup>Department of Laboratory Medicine, National Center for Child Health and Development

<sup>4</sup>Division of Genomics Research, Life Science research Center, Gifu University

<sup>5</sup>Department of Pediatrics, The Jikei University School of Medicine

**O-51 LIVALO Effectiveness and Safety Study in Children with Familial Hypercholesterolemia (LIVES-CH) : Interim report.**

○Akira Ohtake<sup>1</sup>, Takashi Kanno<sup>2</sup>, Shoko Fujii<sup>2</sup>, Wataru Shingaki<sup>2</sup>, Yuji Kurihara<sup>2</sup>  
Ryoji Gunji<sup>2</sup>

<sup>1</sup>Department of Pediatrics, Faculty of Medicine, Saitama Medical University

<sup>2</sup>Post Marketing Surveillance Department, Kowa Company

**O-52 Novel Surgical Reconstruction Rescues Life-threatening Tracheal Obstruction in MPS**

○Shunji Tomatsu, Christian Pizarro, Ryan R. Davies, Lauren Averill, Mary Theroux  
Nemours/Alfred I. duPont Hospital for Children

**O-53 Hematopoietic stem cell targeted gene therapy for bone involvement of murine mucopolysaccharidosis type II**

○Miho Wada<sup>1</sup>, Yohta Shimada<sup>2</sup>, Takashi Higuchi<sup>2</sup>, Kazuhiro Maeda<sup>3</sup>, Mitsuru Saito<sup>3</sup>,  
Hiroyuki Ida<sup>1</sup>, Toya Ohashi<sup>2</sup>, Hiroshi Kobayashi<sup>2</sup>

<sup>1</sup>Department of Pediatrics, Jikei University School of Medicine

<sup>2</sup>Division of Gene Therapy, Research Center for Medicine Sciences, Jikei University School of  
Medicine

<sup>3</sup>Department of Orthopedic Surgery, Jikei University School of Medicine

**11 : 00 ~ 11 : 50 KSIMD Recommending Lecture**

**Chairperson: Tetsuya Ito**

(Fujita Health University, School of Medicine)

**Mitochondrial Diseases in Korea**

Young-Mock Lee

Department of Pediatrics, Yonsei University College of Medicine

**12 : 00 ~ 13 : 00 Luncheon Seminar 5**

**Chairperson: Motomichi Kosuga**

(National Center for Child Health and Development, Tokyo, Japan)

**Early diagnosis & management of MPS IVA**

Julian Raiman

Birmingham Children's Hospital, Birmingham, United Kingdom

**DAY3 : Oct.14 (Sat)**  
**Hall 3**

**8 : 30 ~ 9 : 18 Live broadcast : Hall 2**

**11 : 00 ~ 11 : 50 Live broadcast : Hall 2**

**12 : 00 ~ 13 : 00 Luncheon Seminar 6**

**Chairperson: Fumio Endo**

(Kumamoto-Ezuko Medical Center for The Severely Disabled)

**Latest Therapy for PKU - Aiming the Life-Long Treatment-**

Haruo Shintaku

Department of Pediatrics Osaka City University Graduate School of Medicine