

**Day 3 : Nov. 9 (Sat.)  
Venue 1 (5F 501AB)**

**8 : 10~9 : 00 Morning Seminar 3**

**Sponsored by KM Biologics Co., Ltd.**

**Chairperson: Fumio Endo**

(Kumamoto-Ezuko Medical Center for The Severely Disabled CEO)

**MS3 Challenges and perspectives for the future of expanded newborn screening**

○ Kimitoshi Nakamura

Department of Pediatrics, Kumamoto University Graduate School of Medical Sciences

**9 : 20~10 : 20 Educational Lecture 2**

**Chairperson: Mitsuru Kubota**

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

**EL2 Pediatric Home Medical Care and Intravenous Enzyme Replacement Therapy in Home**

○ Hirotohi Maeda

Medical Incorporated Foundation Harutaka Kai

**10 : 30~12 : 00 Symposium 2**

**Chairpersons: Torayuki Okuyama**

(Genomic Medicine, Saitama Medical University)

**Yasunori Ishigaki**

(Coral Clinic)

**S2 Home-based enzyme replacement therapy for patients with LSDs (from the perspective of specialist)**

○ Motomichi Kosuga

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

**12 : 20~13 : 10 Lancheon Seminar 5**

**Sponsored by Sanofi K.K.**

**Chairperson: Norio Sakai**

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

**LS5 Practical management for Fabry disease: Newborn screening and follow-up**

○ Masahisa Kobayashi

The Jikei University Hospital

**13 : 20~14 : 10 Oral 12 : Urea cycle disorders/Others**

**Chairpersons: Mahoko Furujo**

(Department of Pediatrics, NHO Okayama Medical Center)

**Atsuko Noguchi**

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

**O-56 Expression and enzymatic activity assay of recombinant protein for variants of glutamine synthetase.**

○ Hideki Matsumoto<sup>1</sup>, Hideo Sasai<sup>1,2,3</sup>, Yue Xiao<sup>1</sup>, Mai Mori<sup>1,3</sup>, Hiroki Otsuka<sup>1,3</sup>, Yuka Aoyama<sup>1,4</sup>

Tomohiro Hori<sup>1,3</sup>, Michio Ozeki<sup>1</sup>, Norio Kawamoto<sup>1</sup>, Hidenori Ohnishi<sup>1,3</sup>

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

<sup>2</sup>Department of Early Diagnosis and Preventive Medicine for Rare Intractable Pediatric Diseases, Graduate School of Medicine, Gifu University

<sup>3</sup>Clinical Genetics Center, Gifu University Hospital

<sup>4</sup>Department of Clinical Engineering, College of Life and Health Sciences, Chubu University

**O-57 Arginine Bolus Necessity in Acute phase of Urea Cycle Disorders while taking arginine or citrulline.**

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

Department of Pediatrics, Fujita Health University Graduate School of Medicine

**O-58 Determination of metabolites related to proximal urea cycle disorders by LC/MS/MS**

○ Yasuhiro Maeda<sup>1</sup>, Yoko Nakajima<sup>2</sup>, Yuta Sudo<sup>2</sup>, Tetsuya Ito<sup>2</sup>

<sup>1</sup>Open Facility Center, Fujita Health University

<sup>2</sup>Department of Pediatrics, Fujita Health University

**O-59 Efforts to identify patients with primary low HDL-cholesterolemia and investigate their prognosis**

○ Masayuki Kuroda<sup>1</sup>, Yoshiro Maezawa<sup>2</sup>, Koutaro Yokote<sup>3</sup>

<sup>1</sup>Center for Advanced Medicine, Chiba University Hospital

<sup>2</sup>Department of Endocrinology, Hematology and Gerontology, Chiba University Graduate School of Medicine

<sup>3</sup>Chiba University

**O-60 Evinacumab Long-Term Effect in Pediatric Homozygous Familial Hypercholesterolemia (HoFH)**

- Kimimasa Tobita<sup>1</sup>, Wiegman Albert<sup>2</sup>, Greber-Platzer Susanne<sup>3</sup>, M.doortje Reijman<sup>1</sup>, Eliot Briton<sup>4</sup>  
Min-Ji Charng<sup>5</sup>, Shubha Srinivasan<sup>6</sup>, Carissa Baker-Smith<sup>7</sup>, Seth Baum<sup>8</sup>, Julie Brothers<sup>9</sup>  
Jacob Hartz<sup>10</sup>, Patrick M. Moriarty<sup>11</sup>, Richard T. George<sup>12</sup>, Robert Pordy<sup>12</sup>

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<sup>6</sup>Institute of Endocrinology and Diabetes, The Children's Hospital at Westmead, Sydney, New South Wales, Australia and The University of Sydney, Sydney, Australia

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<sup>9</sup>Division of Cardiology, The Children's Hospital of Philadelphia, Philadelphia, USA

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