Day 3：Nov. 9（Sat.）
Venue 2（6F 602ABCD）

8：10～9：00　Morning Seminar 4

Sponsored by Ultragenyx Japan K.K.

Chairperson: Kei Murayama
（Diagnostics and Therapeutics of Intractable Diseases, Juntendo University Faculty of Medicine）

MS4　Protecting Parents and Children with Familial Hypercholesterolemia

○ Keiji Matsunaga
Anti-aging and Vascular Medicine, Kagawa University Hospital

9：20～10：10　Sponsored Seminar 6

Sponsored by Ultragenyx Japan K.K.

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

SPSE6　Current Status and Challenges in the Diagnosis, Treatment, and Long-term management of Long-chain fatty acid oxidation disorders

○ Hironori Kobayashi
Laboratories Division, Shimane University Hospital

10：30～11：20　Oral 10：Patient registry/Mitochondrial disorders 1

Chairpersons: Ayako Matsunaga
（Department of Pediatrics, St. Marianna University School of Medicine）
Chikahiko Numakura
（Department of Clinical Genomics & Pediatrics, Saitama Medical University）

O-47　Report on the activities of JaSMIn

○ Satoko Tsushima[1,2,3], Masumi Omori[2], Taiko Fujioka[2], Masahisa Kobayashi[2,4], Motomichi Kosuga[2,3]
[1]Nursing Department, Specialist Nursing Office, Genetic Coordinator, National Center for Child Health and Development
[2]Committee for Patient Registration, Japanese Society for Inherited Metabolic Diseases
[3]Division of Medical Genetics, National Center for Child Health and Development
[4]Department of Pediatrics, The Jikei University School of Medicine

O-48　A case of Leigh syndrome and SLC25A19 abnormality identified through Mitochondrial disease panel

○ Takanori Onuki[1], Hiromi Nyuzuki[1], Kentaro Sawano[1], Nao Shibata[1], Yohei Ogawa[1], Hitomi Fujii[2] Shigeru Maruyama[3], Akira Ohtake[5], Yasushi Okazaki[4], Kei Murayama[4]
[1]Department of Pediatrics, Niigata University Graduate School of Medical and Dental Sciences
[2]Department of Child Neurology, National Hospital Organization Nishiniigata Chuo Hospital
[3]Department of Pediatrics, Niigata Prefectural Central Hospital
[4]Diagnostics and Therapeutics of Intractable Diseases, Intractable Disease Research Center, Juntendo University, Graduate School of Medicine
[5]Department of Pediatrics & Clinical Genomics, Faculty of Medicine, Saitama Medical University

O-49 Skeletal alterations resembling mucopolysaccharidosis associated with novel variants in *MARS2* gene

○ Hiroyuki Iijima[1,2], Yuko Tsujioka[3], Yoshiyuki Tsutsumi[4], Akira Ohtake[2]
[1]Department of General Pediatrics & interdisciplinary Medicine, National Center for Child Health and Development
[2]Department of Clinical Genomics & Pediatrics, Saitama Medical University
[3]Department of Radiology, Keio University School of Medicine
[4]Department of Radiology, National Center for Child Health and Development

O-50 Functional analysis of mitochondrial disease gene *PNPLA4* and search target lipid using zebrafish

○ Hiromi Nyuzuki[1,2], Ryuichi Hishida[2], Hideaki Matsui[2]
[1]Department of Pediatrics, Center for Medical Genetics, Niigata University Medical and Dental Hospital
[2]Department of Neuroscience of Disease, Brain Research Institute, Niigata University

O-51 Novel mitochondrial disorder developed by epigenetics silencing: MORC2-related disorders

○ Masaru Shimura[1], Tomohiro Ebihara[2], Makiko Tajika[1], Yohei Sugiyama[3], Takuya Fushimi[1]
Keiko Ichimoto[1], Akira Ohtake[4], Yasushi Okazaki[5], Kei Murayama[1,3,5]
[1]Department of Metabolism, Chiba Children's Hospital
[2]Institute of Neurogenomics, Helmholtz Zentrum Munich, Munich, Germany
[3]Department of Pediatrics, Juntendo University Faculty of Medicine
[4]Department of Pediatrics & Clinical Genomics, Faculty of Medicine, Saitama Medical University
[5]Diagnostics and Therapeutics of Intractable Diseases, Intractable Disease Research Center, Juntendo University, Graduate School of Medicine

11：20～12：00 Oral 11：Mitochondrial disorders 2

Chairpersons: Hiromi Nyuzuki
(Nigata University Medical & Dental Hospital)
Chunhua Zhang
(Department of research & development of MILS International)

O-52 ATP7A gene variants cause symptoms of mitochondrial disease including Leigh syndrome in two cases.

○ Yohei Sugiyama[1,2], Yukiko Yatsuka[2], Kouta Nakamura[2], Yoshihito Kishita[2,3], Takuya Fushimi[4]
Masaru Shimura[4], Taira Toki[5], Akira Ohtake[6], Yasushi Okazaki[2,7], Kei Murayama[1,2]
[1]Department of Pediatrics, Juntendo University Faculty of Medicine
[2]Diagnostics and Therapeutics of Intractable Disease Research Center, Graduate School of Medicine, Juntendo University
[3]Laboratory of Genome Sciences, Department of Life Science, Faculty of Science and Engineering, Kindai University
[4]Department of Metabolism, Chiba Children's Hospital
[5]Department of Pediatrics, Faculty of medicine, Kitasato University
[6]Department of Pediatrics & Clinical Genomics, Faculty of Medicine, Saitama Medical University
[7]Laboratory for Comprehensive Genomic Analysis, RIKEN Center for Integrative Medical Sciences

O-53 A study of 23 children with refractory epilepsy and mitochondrial tRNA synthase deficiency in China

○ Danmin Shen[1], Fang Fang[1], Xiaodi Han[1], Xin Duan[1], Chaolong Xu[1], Zhimei Liu[1], Yang Liu[1]
Weixing Feng[1], Jie Deng[2]
[1]Department of Neurology, Beijing Children's Hospital, Capital Medical University, Beijing, China
[2]Department of Neurology, National Center for Children's Health, Beijing Children's Hospital, Capital Medical University, Beijing, China

O-54 Enhancing Molecular Diagnosis yield through transcriptomic disruption discovery for Mitochondrial Disorders in China

○ Xin Duan[1], Zhimei Liu[1], Jia Wang[2], Chaolong Xu[1], Ying Zou[1], Junling Wang[3], Tianyu Song[1] Xiaodi Han[1], Manting Xu[1], Fang Fang[1]

[1]Department of Neurology, Beijing Children's Hospital, Capital Medical University, National Center for Children's Health, Beijing 100045, China

[2]Cipher Gene Ltd, Beijing 100871, China

[3]Department of Pediatrics, Third Affiliated Hospital of Zhengzhou University, Zhengzhou, Henan 450052, China

O-55 Clincal, Metabolic, and Genetic Analysis and Follow-Up of 42 Patients With Short-chain enoyl-CoA hydratase deficiency in China

○ Yang Liu, Fang Fang, Xin Duan, Tianyu Song, Danmin Shen, Ruoyu Duan, Minhan Song Chaolong Xu

Beijing Children's Hospital, Children's National Medical Center

12：20～13：10 Lancheon Seminar 6

Sponsored by OrphanPacific, Inc.

Chairperson: Shirou Matsumoto

(Department of Neonatology, Kumamoto University Hospital)

LS6 The Present and Future of Urea Cycle Disorders

○ Yoichi Wada

Department of Pediatrics, Tohoku University Graduate School of Medicine

13：20～14：10 Sponsored Seminar 7

Sponsored by Clinigen K.K.

Chairperson: Satoshi Okada

(Graduate School of Biomedical and Health Sciences, Hiroshima University)

SPSE7-1 Current status of hematopoietic stem cell transplantation for inborn errors of metabolism

○ Hiromasa Yabe

Department of Pediatrics, Tokai University School of Medicine

SPSE7-2 A Case of Mucopolysaccharidosis-Type-II who Underwent Allogeneic Bone Marrow Transplantation Combined with Intraventricular Enzyme Replacement Therapy

○ Asahito Hama

Department of Hematology and Oncology, Children's Medical Center, Japanese Red Cross Aichi Medical Center Nagoya First Hospital