DAY2：Oct.13（Fri）
Hall 1

8：30～ 9：18 Oral7：Mitochondrial disorders 1

Chairperson: Tohru Yorifuji
(Division of Pediatric Endocrinology and Metabolism, Children's Medical Center, Osaka City General Hospital)

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

❀ O-27 Clinical and molecular studies in 203 Chinese patients with mitochondrial disorders

○Dongxiao Li[1, 2], Yi Liu[1], Xiyuan Li[1], Ying Jin[1], Jinqing Song[1], Hezhi Fang[3], Yao Zhang[1]
Hui Dong[1], Yanling Yang[4]

[1]Department of Pediatrics, Peking University First Hospital

[2]Rehabilitation center, Henan Children's Hospital.

[3]Key Laboratory of Laboratory Medicine, Ministry of Education, Zhejiang Provincial Key Laboratory of Medical Genetics, College of Laboratory Medicine and Life Sciences, Wenzhou Medical University, Zhejiang, China.

[4]Department of Pediatrics, Children's Hospital of Shanxi Province, Taiyuan, China

O-28 Biochemical and molecular analysis of Leigh and Leigh-like patients in Japan

○Erika Ogawa[1], Masaru Shimura[2], Takuya Fushimi[2], Makiko Tajika[2], Masato Mori[3]
Masakazu Kohda[4], Yoshihito Kishita[4], Yasushi Okazaki[4], Akira Ohtake[5], Kei Murayama[2]

[1]Department of Pediatrics and Child Health, Nihon University

[2]Department of Metabolism, Chiba Children's Hospital

[3]Department of Pediatrics, Matsudo City Hospital

[4]Research Center for Genomic Medicine, Saitama Medical University

[5]Department of Pediatrics, Saitama Medical University

O-29 Efficacy of 5-aminolevulinic acid and sodium ferrous citrate in treating mitochondrial respiratory chain disorder

○Ayako Matsunaga-Fujinami[1], Masaru Shimura[1], Makiko Tajika[1], Naomi Kuranobu[1]
Takuya Fushimi[1], Naoko Nozawa[2], Kiwamu Takahashi[2], Yasushi Okazaki[3, 4], Akira Ohtake[5]
Kei Murayama[1]

[1]Department of Metabolism, Chiba Children's Hospital

[2]Division of Pharmaceutical Research, SBI Pharmaceuticals Co., Ltd.

[3]Research Center for Genomic Medicine, Saitama Medical University

[4]Intractable Disease Research Center, Juntendo University

[5]Department of Pediatrics, Saitama Medical University

O-30 Mitochonic acid(MA-5) promotes ATP synthase oligomerization and cell survival in various mitochondrial diseases

○Tetsuro Matsuhashi[1], Takehiro Suzuki[2], Takeya Sato[3], Hitoshi Osaka[4], Shigeo Kure[1]
Takaaki Abe[5]

[1]Department of Pediatrics, Tohoku University

[2]Division of Nephrology, Endocrinology and Vascular Medicine, Tohoku University

[3]Department of Molecular Pharmacology, Tohoku University Graduate School of Medicine

[4]Department of Pediatrics, Jichi University

[5]Clinical Biology and Hormonal Regulation, Tohoku University

9：18～ 9：54 Oral8：Mitochondrial disorders 2

Chairperson: Masakazu Mimaki
(Department of Pediatrics, Teikyo University School of Medicine)
Yoko Nakajima
(Fujita Health University School of Medicine, Dept. Pediatrics)

O-31 A case report: mitochondrial disorder,caused by ATAD3 gene cluster deletion

○Kazuyuki Ito [1], Haruka Kuno[1], Yoshihiro Minosaki[1], Kei Murayama[2], Akira Ohtake[3]

[1]Department of Pediatrics and Neonatal Medicine, Kawaguchi Municipal Medical Cente

[2]Department of Metabolism, Chiba Children's Hospital

[3]Department of Pediatrics, Saitama Medical University

O-32 ATAD3 cluster deletions cause altered mtDNA and cholesterol metabolism related mitochondrial disease

○Masaru Shimura[1], Fushimi Takuya[1], Keiko Ichimoto[1], Ayako Matsunaga[1], Tomoko Tsuruoka[1]
Yoshihito Kishita[2, 3], Masakazu Kohda[2, 3], Yasushi Okazaki[2, 3], Akira Ohtake[4], Kei Murayama[1]

[1]Department of Metabolism, Chiba Children's Hospital

[2]Research Center for Genomic Medicine, Saitama Medical University

[3]Intractable Disease Research Center, Juntendo University

[4]Department of Pediatrics, Saitama Medical University

O-33 Dig a little deeper into exome: a new way to find out missing causative genes in recessive disorders

○Masakazu KOHDA[1], Tomoko Hirata[2], Yoshihito Kishita[3], Takuya Fushimi[4]
Hiroko Harashima[5], Kei Murayama[4], Akira Ohtake[5], Yasushi Okazaki[1]

[1]Intractable Disease Research Center, Graduate School of Medicine, Juntendo University

[2]Division of Translational Research, Research Center for Genomic Medicine, Saitama Medical University

[3]Division of Functional Genomics & Systems Medicine, Research Center for Genomic Medicine, Saitama Medical University

[4]Department of Metabolism, Chiba Children's Hospital

[5]Department of Pediatrics, Saitama Medical University

10：00 ～ 11：00　Educational Lecture 3

Chairperson: Shigeo Kure

(Department of Pediatrics, Tohoku University School of Medicine)

Genomic diagnosis of mitochondrial disorders: how good are we & how can we increase diagnostic yield?

David Thorburn

Murdoch Childrens Research Institute

11：00 ～ 11：50　Sponsored Seminar 2: Hyperammonemia that can't be overlooked

Chairperson: Toshihiro Ohura

(Sendai City Hospital)

Treatment experience by long-term management of Carglumic Acid (Carbaglu®) to patients with propionic acidemia in Japan

Atsuko Noguchi

Department of Pediatrics, Akita University Graduate School of Medicine

Adult cases of NAGS deficiency with successful management using N-carbamoyl-L-glutamic acid

Kimihiko Oishi

Department of Genetics and Genomic Sciences, Department of Pediatrics
Icahn School of Medicine at Mount Sinai, New York, USA

13：10 ～ 13：40　Presidential Lecture

Chairperson: Hiroyuki Ida

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

Shifts in the diagnostic paradigm of genetic diseases: from 'one gene-one enzyme' to 'too many genes-diverse phenomena'.

Akira Ohtake

Department of Pediatrics, Saitama Medical University

13：50 ～ 15：20　Symposium 2: Gene therapy for inborn error of metabolism

Organizer: Toya Ohashi

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

Chairperson: Toya Ohashi

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

Torayuki Okuyama

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

S2-1　Current status of Gene Therapy for Geneitc Disease in Japan

Toya Ohashi

Div. of Gene Therapy, Research Center for Medical Sciences, Dep. of Pediatrics, The Jikei Univ. Sch. of Med.

S2-2　Gene Editing for MPS 1 and 2 using Zinc Finger Nuclease Technology

○Edward Conner, Michael Holmes

Sangamo Therapeutics

S2-3　Lentiviral based *ex vivo* Gene Therapy for Fabry Disease

○Nerissa C. Kreher[1], Chris Mason[1, 2]

[1]AVROBIO, Inc., [2]University College London

15：30 ～ 17：10　Symposium 3: Clinical practice in Mitochondrial disease; The quintessence of diversity

Organizer: Kei Murayama
(Department of Metabolism, Chiba Children's Hospital)

Chairperson: Kei Murayama
(Department of Metabolism, Chiba Children's Hospital)
Hitoshi Osaka
(Department of Pediatrics, Jichi Medical University)

S3-1　Clinical practice in Mitochondrial disorders; The quintessence of diversity

Kei Murayama

Department of Metabolism, Chiba Children's Hospital

S3-2　Mitochondrial hepatopathy

Shunsaku Kaji

Department of Pediatrics, Tsuyama Chuo Hospital

S3-3　Mitochondrial cardiomyopathy

Atsuhito Takeda

Department of Pediatrics, Graduate School of Hokkaido University

S3-4　The diversity of Leigh syndrome

Masakazu Mimaki

Department of Pediatrics, Teikyo University School of Medicine

S3-5　Mitochondrial disorders in neonatal period

Taro Nagatomo

Ehime Prefectural Central Hospital

17：10 ～ 18：10　Special Lecture 2

Chairperson: Toshiyuki Fukao

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

The Cutting Edge of Autophagy Study：The Path from Basic to Application

Tamotsu Yoshimori

Department of Genetics, Graduate School of Medicine Osaka University

DAY2：Oct.13（Fri）
Hall 2

8：30～ 9：06 Oral9：Amino acid disorders 1

Chairperson: Yoshiyuki Okano
(Okano Children's Clinic)

Koichi Mizuta
(Department of Pediatric Transplant Surgery, Jichi Children's Medical Center Tochigi)

O-34 An infantile case of OTCD patient diagnosed prenatally and treated safely after birth

○Toko Shibuya[1], Hidehito Kondo[1], Sachiko Nakaoka[1], Mika Hirotsune[1], Ryoko Hayashi[1]
Kazuhiko Bessyo[1], Hitomi Arahori[1], Norio Sakai[2], Yoko Nakajima[3], Keiichi Ohzono[1]
[1]Department of pediatrics, Osaka University Graduate School of Medicine
[2]Division of health science, Osaka University Graduate School of Medicine
[3]Department of Pediatrics, Fujita Health University

O-35 The importance of management during hemodialysis and EAA administration in early onset urea cycle disorder.

○Katsuyuki Yokoi, Yoko Nakajima, Yuri Kawai, Tomomi Kondou, Yuuji Matsumoto
Masahumi Miyata, Youhei Ikezumi, Tetsushi Yoshikawa, Tetsuya Ito
Department of pediatrics, Fujita Health University

O-36 Plasma amino acid analysis in OTC deficiency liver transplantation cases

○Koichi Mizuta, Taizen Urahashi, Yoshiyuki Ihara, Yukihiro Sanada, Noriki Okada,
Naoya Yamada, Yuta Hirata, Takumi Katano
Department of Transplant Surgery, Jichi Medical University

9：06～ 9：54 Oral10：Amino acid disorders 2

Chairperson: Hiroshi Mitsubuchi
(Department of neonatology, Kumamoto university hospital)

Takashi Hamazaki
(Department of Pediatrics, Osaka City University Graduate School of Medicine)

O-37 Serum free carnitine levels reflect the disease state of neonatal intrahepatic cholestasis

○Masato Arao, Sayaka Ajihara, Ikuma Musha, Junya Akatsuka, Taro Yamazaki
Toru Kikuchi, Ohtake Akira
Department of Pediatrics, Saitama Medical University Hospital

O-38 Assessment of citrin deficiency frequency in the US and identification of its novel Ashkenazi Jewish founder variant

○Kimihiko Oishi[1], Ashley H. Birch[2], Neal Cody[2], Ruth Kornreich[2], Lisa Edelmann[2]
George A. Diaz[2]

[1]Departments of Genetics and Genomic Sciences, and Pediatrics, Icahn School of Medicine at Mount Sinai

[2]Icahn School of Medicine at Mount Sinai Department of Genetics and Genomic Sciences

O-39 Study on citrin deficiency using mouse models (1) Creation of hyperammonemic mouse

○Takeyori Saheki[1], Eishi Kuroda[1], Aki FUnahashi[1], Izumi Yasuda[1], Yoshiko Setogawa[1]
Tatsuhiko Furukawa[2], Masahisa Horiuchi [1], Mitsuaki Moriyama[3]

[1]Department of Hygiene and Health Promotion Medicine, Kagoshima University Graduate School of Medical and Dental Sciences

[2]Department of Molecular Oncology, Kagoshima University Graduate School of Medical and Dental Sciences

[3]Laboratory of Integrative Physiology in Veterinary Sciences, Osaka Prefecture University

O-40 Study on citrin deficiency using mouse models (2) Identification of efficacious amino acids

○Takeyori Saheki[1], Eishi Kuroda[1], Aki FUnahashi[1], Izumi Yasuda[1], Yoshiko Setogawa[1]
Tatsuhiko Furukawa[2], Masahisa Horiuchi [1], Mitsuaki Moriyama[3]

[1]Department of Hygiene and Health Promotion Medicine, Kagoshima University Graduate School of Medical and Dental Sciences

[2]Department of Molecular Oncology, Kagoshima University Graduate School of Medical and Dental Sciences

[3]Laboratory of Integrative Physiology in Veterinary Sciences, Osaka Prefecture University

10：00 ～ 10：36 Oral11：Metal merabolic abnormality

Chairperson: Norikazu Shimizu
(Department of Pediatrics, Toho University School of Medicine, Ohashi Medical Center)

Kenji Ihara
(Department of Pediatrics, Oita University Faculty of Medicine)

O-41 A infant case of Wilson's disease with intrahepatic cholestasis

○Toju Tanaka, Hiroko Shigetomi, Masayoshi Nagao
Center for Genetics & Metabolic Disorder and Clinical Research, National Hos pital Organization Hokkaido Medical Center

O-42 A Wilson disease patient with decompensated cirrhosis treated by zinc monotherapy

○Norikazu Shimizu, Shoko Nakazawa, Ayako Ogawa, Tsugutoshi Aoki
Department of Pediatrics, Toho University School of Medicine, Ohashi Medical Center

O-43　A novel WDR45 mutation in a 9-month-old male infant with epileptic spasms

○Wanting Liu, Wanting Liu, Zhijie Gao, Xinna Ji, Lina Xie, Yingying Mao, Jianzhao Zhang

Department of Pediatric Neurology, Capital Institute of Pediatrics, Beijing, China

10：36 ～ 11：00　Oral12：Carbohydrate metabolism

Chairperson: Mika Ishige

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

O-44　Diagnosis of congenital hyperinsulinism: Biochemical profiles during hypoglycemia

○Rie Kawakita[1], Azumi Sakakibara[2], Shinji Higuchi[1], Yuki Hosokawa[1], Tohru Yorifuji[1]

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

[2]Department of Pediatrics, Minoh City Hospital

O-45　Enzyme assay of phosphorylase b kinase with p.G991A variant in *PHKA2* gene

○Yasuhiko Ago[1], Hideo Sugie[2], Tokiko Fukuda[3], Hiroki Otsuka[4], Hideo Sasai[1], Mina Nakama[5]

Elsayed Abdelkreem[1], Toshiyuki Fukao[1]

[1]Department of pediatrics, Gifu University

[2]Department of Occupational Therapy, Tokoha University

[3]Department of pediatrics, Hamamatsu University School of Medicine

[4]Department of neonatology, Gifu prefectural general medical center

[5]Division of clinical genetics, Gifu University hospital

11：00 ～ 11：50　SIMD Recommending Lecture（Sponsored Seminar 3）

Chairperson: Masaki Takayanagi

(Department of Nursing, Faculty Health Care and Medical Sports, Teikyo Heisei University)

From Bedside to Bench and Back Again

Kimberly Chapman

Children's National Rare Disease Institute

12：00 ～ 13：00　Luncheon Seminar 3

Chairperson: Yoshikatsu Eto

(Advanced Clinical Research Center, Institute of Neurological Disorders/ The Jikei University School of Medicine)

Fabry Disease : Lessons learned from our experience for decades

R. J. Desnick

Department of Genetics and Genomic Sciences Icahn School of Medicine at Mount Sinai

DAY2：Oct.13（Fri）
Hall 3

8：30～ 9：54 Live broadcast：Hall 2

10：00～11：50 Live broadcast：Hall 2

12：00～13：00 Luncheon Seminar 4: Real world of medical service for the patients with Fabry disease

Chairperson: Norio Sakai
(Division of Health Science,
Osaka University Graduate School of Medicine)

Cardiac manifestation of Fabry disease and the long-term effect of enzyme replacement therapy

Kenichi Hongo

Division of Cardiology, Department of Internal Medicine, The Jikei University School of Medicine

Genetic Counseling for the patients with Fabry disease-Significance of pedigree analysis-

Yoriko Watanabe

Research Institute of Medial Mass Spectrometry/ Department of Pediatrics and Child Health,
Kurume University School of Medicine

15：00～18：00 Introduction & Exhibition of Patient Family Groups