Day 3：Nov. 10（Sat）
Venue 1（2F Hall）

8：45～9：33　Oral 9：Organic Acidemias

Chairperson: Ikue Hata

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

O-37　Nation-wide survey of methylmalonic aciduria

○ Tetsuya Ito[1], Katsuyuki Yokoi[1], Yoko Nakajima[1], Toshiyuki Fukao[2]
[1]Department of Pediatrics, Fujita Health University
[2]Department of Pediatrics, Gifu University

O-38　A 10-year-old girl with propionic acidemia diagnosed from long QT by cardiac screening at school

○ Hideo Sasai[1,2], Hiroko Goto[3], Miwa Kawashiri[4], Hideki Matsumoto[1], Hiroki Otsuka[1,5]
Yasuhiko Ago[1], Ryoji Fujiki[6], Osamu Ohara[6], Toshiyuki Fukao[1,2]
[1]Department of Pediatrics, Graduate School of Medicine, Gifu University
[2]Division of Clinical Genetics, Gifu University Hospital
[3]Department of Pediatric Cardiology, Gifu Prefectural General Medical Center
[4]Department of Pediatrics, Takayama Red Cross Hospital
[5]Department of Neonatology, Gifu Prefectural General Medical Center
[6]Department of Technology Development, Kazusa DNA Research Institute

O-39　A patient with 3-MCC deficiency developed an acute metabolic crisis in the neonatal period

○ Tomohiro Saitou, Asami Okafuji, Tomoko Tando, Yusuke Goto, Takayuki Komai
Department of Pediatrics, Yamanashi Prefectural Central Hospital

O-40　A case with glutaric acidemia type 2 involving FLAD1 mutation detected by newborn screening

○ Kenji Yamada[1], Michinori Ito[2], Hironori Kobayashi[1], Yuki Hasegawa[1], Seiji Yamaguchi[1]
Takeshi Taketani[1]
[1]Department of Pediatrics, Shimane University Faculty of Medicine
[2]Departmental of Metabolism, Shikoku Medical Center for Children and Adults

9：33～10：33　Oral 10：Disorders in Fatty Acid Oxidation or Ketone Body Metabolism

Chairperson: Hideo Sasai

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

O-41　Investigation of beta oxidation process in MCAD-deficient patients who showed normal enzyme activity

○ Miori Yuasa[1], Ikue Hata[1], Keiichi Sugihara[1], Yuko Isozaki[1], Yousuke Shigematsu[1], Yusei Ohshima[1]
Reiko Kagawa[2], Satoshi Okada[2], Keiichi Hara[3], Go Tajima[4]
[1]Department of Pediatrics, Fukui University
[2]Department of Pediatrics, Hiroshima University Graduate School of Biomedical & Health Sciences
[3]Department of Pediatrics, National Hospital Organization Kure Medical Center
[4]Division of Neonatal Screening, Research Institute, National Center for Child Health and Development

O-42 Dietary Therapy in a patient with ECHS1 deficiency

○ Mari Kuwajima[1], Karin Kojima[1], Takahiro Ikeda[1], Masahide Goto[1], Takuya Hushimi[2]
Kei Murayama[2], Yoshihito Kishita[3], Kenichirou Yamada[4], Hitoshi Osaka[1], Takanori Yamagata[1]
[1]Department of Pediatrics
[2]Chiba Children's Hospital
[3]Diagnostics and Therapeutics of Intractable Diseases/Intractable Disease Research Center, Juntendo University, Graduate School of Medicine
[4]Institute for Developmental Desease, Aichi Human Service Center

O-43 A infant with CPT2 deficiency diagnosed at rhabdomyolysis

○ Tomoko Lee[1], Kazuhiro Yamamoto[2], Kenji Yamada[3], Hironori Kobayashi[3], Minori Yuasa[4]
Yosuke Shigematsu[4], Keiichi Hara[5], Go Tajima[6], Yasuhiro Takeshima[1]
[1]Department of pediatrics, Hyogo College of Medicine
[2]Department of pediatrics, Takatsuki General Hospital
[3]Department of pediatrics, Shimane University Graduate School of Medicine
[4]Department of pediatrics, Faculty of medical Science, University of Fukui
[5]Kure Medical center and Chugoku Cancer Center
[6]Division of Neonatal Screening, Research Institute, National Center for Child Health and Development

O-44 Characterization of *HMGCS2* mutations identified in Japanese HMG-CoA synthase deficient patients

○ Yasuhiko Ago[1], Hiroki Otsuka[2], Elsayed Abdelkreem[3], Hideo Sasai[1], Mina Nakama[4]
Yuka Aoyama[1], Yoko Nishimura[5], Yoko Nakajima[6], Tetsuya Ito[6], Toshiyuki Fukao[1],
Yoriko Watanabe[7], Kaori Fukui[7], Kazumasa Akiyama[8], Lee Tomoko[9]
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[2]Gifu prefectural general medical center
[3]Department of Pediatrics, Faculty of Medicine, Sohag University, Sohag, Egypt
[4]Division of clinical genetics, Gifu University hospital
[5]Department of Neuropediatrics, Tottori University
[6]Department of Pediatrics, Fujita Health University
[7]Kurume University
[8]Kitazato University
[9]Hyogo College of Medicine

O-45 Fasting test with infantile 3-hydroxybutyrate dehydrogenase (*Bdh1*) KO mice

○ Hiroki Otsuka[1,2], Takeshi Kimura[1], Yasuhiko Ago[1], Mina Nakama[3], Elsayed Abdelkreem[1]
Yuka Aoyama[4], Hideki Matsumoto[1], Hideo Sasai[1], Hidenori Ohnishi[1], Toshiyuki Fukao[1]
[1]Department of Pediatrics, Graduate School of Medicine, Gifu University
[2]Department of Neonatology, Gifu Prefectural General Medical Center
[3]Division of Clinical Genetics, Gifu University Hospital
[4]Department of Biomedical Sciences, College of Life and Health Sciences, Chubu University

10：40〜11：30 Sponsored Seminar 6 Sponsored by Alexion Pharma GK

Chairperson: Yoshikatsu Eto
(Advanced Clinical Research Center & Institute for the Treatment of Genetic Disease, Institute of Neurological Disorders)

SS6 Tips for Treatment & Diagnosis of Lysosomal Acid Lipase Deficiency

○ Satoshi Watanabe
Department of Pediatrics, Nagasaki University Hospital

11：40～12：40　Luncheon Seminer 5：Fabry disease：Frontiers of diagnosis/ treatment/genetic counseling
Sponsored by Sumitomo Dainippon Pharma Co., Ltd.

Chairperson: Hitoshi Sakuraba
(Department of Clinical Genetics, Meiji Pharmaceutical University)

LS5-1　Dianosisi of Fabry diease

○ Toya Ohashi[1,2]
[1]Division of Gene Therapy, Research Center for Medical Sciences, The Jikie University School of Meidicine
[2]Department of Pediatrics, The Jikei University School of Medicine

LS5-2　Practical and task of genetic counseling in the scene of treatment for Fabry disease

○ Norio Sakai
Child Healthcare and Genetic Science Laboratory, Division of Health Science, Osaka University Graduate School of Medicine

12：50～14：50　Symposium 2：Future Direction of JSIMD

Chairpersons: Hiroyuki Ida
(Department of Pediatrics, The Jikei University School of Medicine)
Fumio Endo
(Kumamoto-Ezuko Medical Center for the Severely Disabled)

SY2-1　Past, present and future of Japanese Society for Inherited Metabolic Disease

○ Hiroyuki Ida
Department of Pediatrics, Jikei University School of Medicine

SY2-2　How we can facilitate the activity of JSIMD?

○ Yoichi Matsubara
The Japan Society of Human Genetics

SY2-3　How to enhance a sense of presence of JSIMD?

○ Toshiyuki Fukao
Department of Pediatrics, Graduate School of Medicine, Gifu University

SY2-4　Basic research of inborn errors of metabolism

○ Shigeo Kure
Department of Pediatrics, Tohoku University

SY2-5　Education for young doctors

○ Norio Sakai
Child Healthcare and Genetic Science Laboratory, Division of Health Science, Osaka University Graduate School of Medicine

14：50～　Closing Remarks

Day 3：Nov. 10（Sat）
Venue 2（5F Conference Room）

8：45〜9：45　Oral 11：Mitochondrial Disorders・Peroxisomal Disorders

Chairperson: Nobuyuki Shimozawa

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

O-46　IARS deficiency in Japan and China －Collaboration of rare diseases in Asia－

○ Ayako Matsunaga-Fujinami[1], Zhimei Liu[1], Reiko Itoh[2], Masaru Shimura[1], Ayano Inui[3]
Tomoo Fujisawa[3], Yasushi Okazaki[4], Akira Ohtake[5], Fang Fang[6], Kei Murayama[1]
[1]Center for Medical Genetics, Department of Metabolism, Chiba Children's Hospital
[2]Division of General Pediatrics and Interdisciplinary Medicine, National Center for Child Health and Development
[3]Department of Pediatric Hepatology and Gastroenterology, Saiseikai Yokohama Tobu Hospital
[4]Intractable Disease Research Center, Juntendo University
[5]Department of Pediatrics, Saitama Medical University
[6]Department of Neurology, Beijing Children's Hospital, Beijing, China

O-47　A case of MELAS/Leigh overlap syndrome with m.T10158C mutation in MTND3 gene

○ Hidehito Kondo[1,2], Mikihiro Akagi[3], Tohko Shibuya[2], Sachiko Nakaoka[2,4], Kohji Tominaga[2]
Shin Nabatame[2], Norio Sakai[5], Keiichi Ozono[2]
[1]Department of Pediatrics, Japanese Red Cross Kyoto Daiichi Hospital
[2]Department of Pediatrics, Osaka University Graduate School of Medicine
[3]Department of Pediatrics, Nissei Hospital
[4]Department of Pediatrics, Kouseiren Takaoka Hospital
[5]Department of Health Science, Child Healthcare and Genetic Science, Osaka University Graduate School of Medicine

O-48　Pathological study of Zellweger syndrome using disease model fish

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

O-49　Sibling cases of Zellweger syndrome

○ Rieko Sakamoto[1], Shiro Matsumoto[2], Jun Kido[2], Kimitoshi Nakamura[2]
[1]Perinatal medical center Kumamoto University Hospital
[2]Department of Pediatrics Division of Life Science Kumamoto University

O-50　Diagnostic process and neuroimages in ALD children with initial symptoms of higher brain dysfunction

○ Kazuo Kubota[1], Hiroki Kawai[1], Shigeo Takashima[2], Toshiyuki Fukao[1], Nobuyuki Shimozawa[2]
[1]Department of Pediatrics, Gifu University Graduate School of Medicine
[2]Division of Genomics Research, Life Science Research Center, Gifu University

9：45～10：33　Oral 12：Patient Registration and Others

Chairperson: Yoko Nakajima

(Fujita Health University School of Medicine)

O-51　Epidemiology in Japanese Fabry patients: data from the Jikei Fabry Registry

○ Toshiki Tsunogai[1,2], Mio Sakuma[4], Asako Morita[3], Toya Ohashi[1,3], Hiroyuki Ida[1,3]

[1]Department of Pediatrics, The Jikei University School of Medicine

[2]Department of Pediatrics, Fuji City General Hospital

[3]Division of Gene therapy, Research Center for Medical Science, The Jikei University School of Medicine

[4]Department of Clinical Epidemiology, Hyogo College of Medicine

O-52　Urinary titin of 3-year-old children can be used as a biomarker to screen muscle diseases

○ Masafumi Matsuo[1], Taku Shirakawa[1], Hiroyuki Awano[2], Hisahide Nishio[1]

[1]Research Center for Locomotion Biology, Kobe Gakuin University

[2]Division of Pediatrics, Graduate School of Medicine, Kobe Univesity

O-53　Fever induced recurrent acute liver failure due to *NBAS* mutation

○ Sahoko Ono[1], Junko Matsuda[1,2], Norikazu Inamura[1], Etsuko Watanabe[2], Takanobu Otomo[1,2] Hideto Teranishi[1], Ippei Miyata[1], Yoshito Sadahira[3], Hirokazu Kanegane[4], Kazunobu Ouchi[1]

[1]Department of Pediatrics, Kawasaki Medical School

[2]Department of Pathophysiology and Metabolism, Kawasaki Medical School

[3]Department of Pathology, Kawasaki Medical School

[4]Department of Pediatrics and Developmental Biology, Tokyo Medical and Dental University

O-54　Disruption of the responsible gene in a PGM 1 deficiency patient by homozygous chromosomal inversion

○ Katsuyuki Yokoi[1], Yoko Nakajima[1], Tamae Ooe[2], Hidehito Inagaki[2], Yoshinao Wada[3], Hideo Sugie[4] Isao Yuasa[5], Tokiko Fukuda[6], Tetsuya Ito[1], Hiroki Kurahashi[2]

[1]Div Pediatrics, Fujita medical university

[2]Div Molecu, Fujita Medical University

[3]Depart Obstertric Medicine Osaka Hosp

[4]Faculty Health, Tokoha Univ

[5]Div Legal Medicine, Tottori Univ

[6]Depart Pediatrics, Hamamatsu Univ

10：40～11：30　Sponsored Seminar 7：Diagnosis and treatment of Gaucher disease　Sponsored by Sanofi K.K.

Chairperson: Tokiko Fukuda

(Department of Pediatrics, Hamamatsu University School of Medicine)

SS7-1　Clinical Manifestations and Management of Gaucher disease

○ Yoriko Watanabe

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

SS7-2　Relationship between Gaucher disease and Parkinson disease

○ Yoshiki Sekijima

Department of Medicine (Neurology and Rheumatology), Shinshu University School of Medicine

11：40～12：40　Luncheon Seminer 6：Wait! That symptom! Could be MPS?
Sponsored by Sanofi K.K.

Chairperson: Torayuki Okuyama
(Center for Lysosomal Storage Disease and Department of Clinical Laboratory Medicine, National Center for Child Health and Development)

LS6-1　Interval change of brain MR Imaging in mucopolysaccharidosis under enzymatic replacement therapy

○ Yoshiko Matsubara
Department of Diagnostic Radiology, Hiroshima University Hospital

LS6-2　Spinal Disorders in Lysosomal Storage Diseases Patients

○ Hidetomi Terai
Department of Orthopaedic Surgery, Osaka City University Graduate School of Medicine

12：50～13：50　Educational Lecture 3

Chairpersons: Norikazu Shimizu
(Department of Pediatrics, Toho University Ohashi Medical Center)
Tomonobu Hasegawa
(Department of Pediatrics, Keio University School of Medicine)

EL3　How to understand inborn errors of metabolism －at the routine medical practice－

○ Mitsuru Kubota
Department of General Pediatrics, National Center for Child Health and Development

13：50～14：50　Educational Lecture 4

Chairpersons: Norikazu Shimizu
(Department of Pediatrics, Toho University Ohashi Medical Center)
Tomonobu Hasegawa
(Department of Pediatrics, Keio Univercity School of Medicine)

EL4　Metabolic emergency

○ Masaki Takayanagi
Department of Nursing, Faculty of Community Health Care, Teikyo Heisei University