Day 2：Nov. 9（Fri）
Venue 1（2F Hall）

8：50～9：20　Presidential Lecture

Chairperson: Hiroyuki Ida

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

PL　Splicing abnormalities have attracted me

○ Toshiyuki Fukao

Department of Pediatrics, Graduate School of Medicine, Gifu University

9：20～10：05　Educational Lecture 1

Chairperson: Nobuyuki Shimozawa

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

EL1　Peroxisomal disorders: new defects, new phenotypes and the continued importance of biochemical studies

○ Ronald Wanders

Departments Clinical Chemistry, Academic Medical Center, University of Amsterdam; Pediatrics, Laboratory Genetic Metabolic Diseases, Emma Children's Hospital, Amsterdam, The Netherlands

10：05～10：50　Special Lecture 2

Chairperson: Tetsuya Ito

（Department of Pediatrics, Fujita Health University）

SL2　A review on congenital disorders of glycosylation and its therapies

○ Eva Morava

Department of Clinical Genomics, Mayo Clinic, USA

11：00～11：50　Sponsored Seminar 3

Sponsored by BioMarin Pharmaceutical Japan

Chairperson: Toshiyuki Fukao

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

SS3　Recognizing MPS in daily practice. Detecting early signs and symptoms of MPSIVA

○ Maurizio Scarpa

Centre for Rare Diseases, Helios Dr. Horst Schmidt Klinik, Wiesbaden, Germany

12：00～13：00 Luncheon Seminer 3：Tips of diagnosis and importance of early diagnosis －Fabry disease－ Sponsored by Sanofi K.K.

Chairperson: Toya Ohashi

(Research Center for Medical Science, The Jikei University School of Medicine)

LS3-1 A family case of Fabry disease which was diagnosed by anhidrosis

○ Atsushi Fukunaga

Division of Dermatology, Department of Internal Related, Kobe University Graduate School of Medicine

LS3-2 Updated review on cornea verticillata in Fabry disease

○ Shizuka Koh

Department of Innovative Visual Science, Osaka University Graduate School of Medicine

13：10～14：10 Special Lecture 3

Chairperson: Toshiyuki Fukao

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

SL3 Glycan structures and biological functions, focussing on heparin, sialoglycans and gangliosides

○ Makoto Kiso

Professor Emeritus, Gifu University

14：10～14：55 Educational Lecture 2

Chairperson: Tsutomu Takahashi

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

EL2 The many faces of tyrosinemia: challenges for the next generation

○ Grant A. Mitchell[1], Hao Yang[1], Pei Weng[1,2], The Quebec NTBC Study Group[3]

[1]Department of Pediatrics, Medical Genetics Division, CHU Ste-Justine, Canada

[2]Central South University, Changsha, Hunan, China

[3]All treatment centers in Quebec, Canada

15：00～16：10 Sponsored Symposium：Diagnosis and Treatment for Hereditary tyrosinemia type1 Disease－Current status in Japanese cases of treatment with Nitisinone and future－ Sponsored by Astellas Pharma Inc.

Chairpersons: Fumio Endo

(Kumamoto-Ezuko Medical Center for the Severely Disabled/ Emeritus Professor, Kumamoto University School of Medicine)

Kimitoshi Nakamura

(Department of Pediatrics, Kumamoto University Graduate School of Medical Sciences)

Commentator: Grant A. Mitchell

(Department of Pediatrics, CHU Sainte-Justine, University of Montreal, Canada)

SSY-1 A case of unclassifiable tyrosine metabolism disorder

○ Yuri Etani[1], Misuzu Yoshida[2], Hideaki Okajima[3]
[1]Departmenet of Pediatric Gastroenterology, Nutrition and Endocrinology, Osaka Women's and Children's Hospital
[2]Department of Neonatal Medicine, Osaka Women's and Children's Hospital
[3]Division of Hepato-Biliary-Pancreatic Surgey and Transplantation/Pediatric Surgery, Graduate School of Medicine Kyoto University

SSY-2 Discontinuation of NTBC plus unrestricted diet post-liver transplantation in hereditary tyrosinemia type 1

○ Kosuke Kurihara[1,2], Junya Hirayama[1], Keishirou Amano[1], Hidemi Toyoda[1], Mikihiro Inoue[3] Keiichi Uchida[3], Hiroyuki Sakurai[4], Akinobu Hayashi[5], Masahiro Hirayama[1]
[1]Department of Pediatrics, Mie University Hospital
[2]Department of Pediatrics, Yokkaichi Hazu Medical Center
[3]Department of Pediatric Surgery, Mie University Hospital
[4]Department of Hepatobiliary Pancreatic and Transplant Surgery, Mie University Hospital
[5]Department of Pathology, Mie University Hospital

SSY-3 A Japanese Case of Tyrosinaemia type I and Treatment with NTBC (2-(2-nitro-4-trifluoromethylbenzoyl)-1, 3-cyclohexanedione) After Transplantation

○ Shirou Matsumoto[1], Takanobu Yoshida[1], Tatsuya Kawasaki[1], Rieko Sakamoto[1], Jun Kido[1] Hidekazu Yamamoto[2], Hirotake Sawada[3], Maiko Utoyama[3], Hiroshi Moritake[3], Yukihiro Inomata[4] Fumio Endo[5], Kimitoshi Nakamura[1]
[1]Department of Pediatrcis, Faclty of Life Sciences, Kumamoto University
[2]Department of Pediatric Surgery and Transplantation, Faclty of Life Sciences, Kumamoto University
[3]Department of Pediatrics, Faculty of Medicine, University of Miyazaki
[4]Kumamoto Rosai Hospital
[5]Kumamoto-Ezuko Medical Center for Development and Disability

16：10〜16：58 English Session 4：Organic Acidemias

Chairpersons: Kimihiko Oishi
(Departments of Pediatrics, Genetics & Genomic Sciences, Icahn School of Medicine at Mount Sinai)
Nobuyuki Ishige
(Division of Newborn Screening, Tokyo Health Service Association)

ES-15 Heterogeneous phenotypes, genotypes, treatment and prevention of 1003 patients with methylmalonic acidemia in the mainland of China

○ Yi Liu[1], Yupeng Liu[1], Yao Zhang[1], Jinqing Song[1], Yanyan Ma[2], Xiyuan Li[3], Yuan Ding[4] Dongxiao Li[5], Ying Jin[1], Yanling Yang[1]
[1]Peking University First Hospital
[2]Department of Pediatrics, Affiliated Hospital of Qinghai University
[3]Precision Medicine Center, General Hospital of Tianjin Medical University
[4]Center for Endocrine and genetic Metabolism, Beijing Children's Hospital, Capital Medical University
[5]Laboratory for genetic metabolic diseases, Henan Children's Hospital

ES-16 Detection of methylmalonic acid, methylcitric acid and total homocysteine in dried blood spots by tandem mass spectrometry: clinical verification in large patient samples

○ Yi Liu[1], Lulu Kang[1], Junjuan Wang[2], Ying Jin[1], Mengqiu Li[1], Jinqing Song[1], Haixia Li[2]
Yanling Yang[1]
[1]Peking University First Hospital
[2]Zhejiang Biosan Biochemical Technologies Co., Ltd

ES-17 Maternal beta-ketothiolase deficiency detected by expanded newborn screening

○ Somporn Liammongkolkul, Kasinat Sanomcham, Wasitporn Faksrimuang, Suwit Budda
Pornswan Wasant, Nithiwat Vatanavicharn
Division of Medical Genetics, Department of Pediatrics, Faculty of Medicine Siriraj Hospital, Mahidol University, Thailand

ES-18 Inverse association between blood ammonia and glutamine level in MMA and PA

○ Yoko Nakajima[1], Katsuyuki Yokoi[1], Yasuhiro Maeda[2], Tetsushi Yoshikawa[1], Tetsuya Ito[1]
[1]Department of Pediatrics, Fujita Health University School of Medicine
[2]Graduate School of Pharmaceutical Sciences, Nagoya City University

17：00〜17：30 KSIMD Recommending Lecture

Chairperson: Akira Ohtake
(Department of Pediatrics, Center for Intractable Diseases, Saitama Medical University)

KSIMD Molecular study of neurological Wilson disease

○ Boem Hee Lee
Department of Pediatrics, Medical Genetics Center, Asan Medical Center Children's Hospital, Seoul, Korea

17：40〜18：30 Sponsored Seminar 4

Sponsored by Recordati Rare Diseases Japan K.K./POLA PHARMA INC.

Chairperson: Fumio Endo
(Kumamoto-Ezuko Medical Center for the Severely Disabled)

SS4 Clinical Experience of Carglumic Acid in Hyperammonaemia

○ Sufin Yap
Department of Inherited Metabolic Diseases, Sheffield Children's NHS Foundation Trust, Sheffield, UK

Day 2：Nov. 9（Fri）
Venue 2（5F Conference Room）

9：30〜10：30　Oral 4：Lysosomal Disorders（Mucopolysaccharidosis）

Chairperson: Aya Narita

（Department of Child Neurology, Tottori University Hospital）

O-12　5-plex LC-MS/MS method to measure simultaneously MPS I, II, IIIB, IVA and VI enzyme activities in DBS

○ Tsubasa Oguni[1], Hironori Kobayashi[1,2], Misa Tanaka[3], Kenji Orii[4], Toshiyuki Fukao[4]
Dung Chi Vu[5], Can Thi Bich Ngoc[5], Shunji Tomatsu[2,6], Michael H. Gelb[7], Atsushi Nagai[1]

[1]Clinical Laboratory Division, Shimane University Hospital
[2]Department of Pediatrics, Shimane University Faculty of Medicine
[3]Shimadzu Corporation
[4]Department of Pediatrics, Graduate School of Medicine, Gifu University
[5]Department of Medical Genetics and Metabolism; Center for rare disease and newborn screening, National Children's Hospital, Hanoi, Vietnam
[6]Nemours/Alfred I. duPont Children's Hospital, Wilmington, America
[7]Depts. of Chemistry and Biochemistry, Univ. of Washington, Seattle, USA

O-13　NBS method for MPS II of measuring enzyme activity by LC-MS/MS could reduce false positive

○ Hironori Kobayashi[1,2], Tsubasa Oguni[2], Kimitoshi Nakamura[3], Ken Momose[3], Kenji Orii[4]
Toshiyuki Fukao[4], Can Thi Bich Ngoc[5], Dung Chi Vu[5], Michael H Gelb[6], Shunji Tomatsu[1,7]
Misa Tanaka[8], Kenji Yamada[1], Yuki Hasegawa[1], Jun Watanabe[8], Yoshitomo Notsu[2],
Seiji Yamaguchi[1], Atsushi Nagai[2], Takeshi Taketani[1]

[1]Department of Pediatrics, Shimane University Faculty of Medicine
[2]Clinical Laboratory Division, Shimane University Faculty of Medicine
[3]Department of Pediatrics, Kumamoto University, Graduate School of Medical Sciences
[4]Department of Pediatrics, Graduate School of Medicine, Gifu University
[5]Department of Medical Genetics and Metabolism, Center for rare disease and newborn screening, National Children's Hospital, Hanoi, Vietnam
[6]Depts. of Chemistry and Biochemistry, Univ. of Washington, Seatle, USA
[7]Nemours/Alfred I. duPont Children's Hospital, Wilmington, USA
[8]SHIMADZU Co.

O-14　Investigation of neonatal screening for mucopolysaccharidosis types I and II

○ Kenji Orii[1,2], Hironori Kobayashi[3], Shunji Tomatsu[4], Dung Vu Chi[5], Toshiyuki Fukao[2]

[1]Nagamori children's clinic
[2]Department of Pediatrics, Gifu graduate school of medicine
[3]Department of Pediatrics, Shimane University
[4]Nemours/Alfred I. DuPont Children's Hospital, Wilmington, U.S.A.
[5]Department of Medical Genetics and Metabolism, National Hospital of Pediatrics, Hanoi, Vietnam

O-15　Establishment of immunodeficient murine model of mucopolysaccharidosis type II

○ Yohta Shimada[1], Natsumi Ishii[1], Motohito Goto[2], Takashi Higuchi[1], Hiroshi Kobayashi[1,3]
Toya Ohashi[1,3]

[1]Div. of Gene Therapy, Res. Cent. for Med. Sci., The Jikei Univ. Sch. of Med.
[2]Animal Reso. Tech. Res., Cent. Insti. for exp. animals
[3]Dept. of Pediatrics, The Jikei Univ. Sch. of Med.

O-16 Successful Treatment by Early HSCT for Central Nervous System in Patient with MPS type I

○ Narutoshi Yamazaki[1,4], Yasuyuki Fukuhara[1], Jyohyon So[2], Akira Ishiguro[4], Hiromasa Yabe[3]
Torayuki Okuyama[2], Motomichi Kosuga[1,2]
[1]National Center for Child Health and Development, Division of Medical Genetics
[2]National Center for Child Health and Development, Department of Clinical La boratory Medicine
[3]Center for Matrix Biology and Medicine, Tokai University Graduate School of Medicine
[4]National Center for Child Health and Development, Department of Postgraduate Education and Training Center

11：00～11：50 Sponsored Seminar 5 Sponsored by ReqMed Company, Ltd.

Chairperson: Yasuyuki Suzuki
(Medical Education Development Center, Gifu University)

SS5 Mucopolysaccharidoses Update

○ Shunji Tomatsu[1,2,3]
[1]Nemours/Alfred I. duPont Hospital for Children
[2]Departments of Orthopedics and BioMedical, Skeletal Dysplasia
[3]Department of Pediatrics, Sidney Kimmel Medical College at Thomas Jefferson University

12：00～13：00 Luncheon Seminer 4

Sponsored by DAIICHI SANKYO COMPANY, LIMITED
Chairperson: Shintaku Haruo
(Osaka City University Graduate School of Medicine)

LS4 Diagnosis and Management Guidelines for PKU

○ Takashi Hamazaki
Department of Pediatrics, Faculty of Medicine, Osaka City University

14：20～15：44 Oral 5：Lysosomal Disorders (Fabry Disease)

Chairpersons: Hitoshi Sakuraba
(Department of Clinical Genetics, Meiji Pharmaceutical University)
Mahoko Furujo
(Department of Pediatrics, Okayama Medical Center, National Hospital Organization)

O-17 Investigation of characteristics of heterozygous Fabry disease

○ Kazuya Tsuboi, Hiroshi Yamamoto
LSD Center, Nagoya Central Hospital

O-18 Molecular and biochemical basis of Japanese patients with Fabry disease

○ Hitoshi Sakuraba[1], Takahiro Tsukimura[2], Tadayasu Togawa[2], Tomoko Shiga[1], Seiji Saito[3]
Kazuki Ohno[4], Toshie Tanaka[1], Atsuko Sato[1], Tomoko Otsuka[1]
[1]Department of Clinical Genetics, Meiji Pharmaceutical Univertsity
[2]Department of Functional Bioanalysis, Meiji Pharmaceutical University
[3]Department of Medical Management and Informatics, Hokkaido Information University
[4]Catalyst, Inc.

O-19 Mutation spectrum of Japanese families with Fabry disease: genotype-phenotype correlation

○ Masahisa Kobayashi[1], Eiko Kaneshiro[1], Toya Ohashi[2], Yoshikatsu Eto[3], Hiroyuki Ida[1]
[1]Department of Pediatrics, The Jikei University School of Medicine
[2]Division of Gene Therapy, Research Center for Medical Sciences, The Jikei University School of Medicine
[3]Advanced Clinical Research Center, Institute of Neurological Disorders

O-20 Correlation of lysoGb3 levels with antibody, age and clinical severities in Fabry diseases

○ Chen Wu[1,2,3], Takeo Iwamoto[4], Mohammad Arif Hossain[1,3], Takashi Miyajima[1,2], Keiko Akiyama[1] Hiroko Yanagisawa[1], Junko Igarashi[2], Yoshikatsu Eto[3]
[1]Advanced Clinical Research Center, Institute of Neurological Disorders
[2]Rare disease research center, AnGes Inc.
[3]The Jikei University School of Medicine
[4]Core Research Facilities for Basic Science, Molecular Cell Biology, The Jikei University School of Medicine

O-21 Analysis of anti-GLA antibodies and plasma Lyso-Gb3 level in Fabry patients received ERT

○ Yuya Tayama[1], Tomoko Shiga[2], Takahiro Tsukimura[1], Tadayasu Togawa[1], Hitosi Sakuraba[2]
[1]Department of Functional Bioanalysis, Meiji Pharmaceutical University
[2]Department of Clinical Genetics, Meiji Pharmaceutical University

O-22 Correlations between serum cholesterol and vascular lesions in Fabry disease patients

○ Hiroki Katsuta[1,2,3], Kazuya Tsuboi[1], Hiroshi Yamamoto[1], Hiromi Goto[1]
[1]LSD Center, Nagoya Central Hospital
[2]Department of Cell Biology and Development, Graduate School of Medicine Nagoya University
[3]Division of Embryology

O-23 Vertebral basilar artery findings and risk of sudden hearing loss in 31 patients with Fabry disease

○ Hiroshi Yamamoto, Kazuya Tsubi
LSD Center, Nagoya Central Hospital

15：44〜16：44 Oral 6：Lysosomal Disorders（Gaucher's Disease and Others）

Chairperson: Hiroshi Kobayashi
(Division of Gene Therapy, Research Center for Medical Sciences, The Jikei University School of Medicine)

O-24 Current status of substrate reduction therapy for Gaucher disease in Japan

○ Ken Sakurai, Hiroyuki Ida
Department of Pediatrics, The Jikei University School of Medicine

O-25 Large soft-tissue masses in a patient with Gaucher disease: Evaluation of SRT

○ Shoji Yano, Neha Mahajan
Department of Pediatrics, University of Southern California, Los Angeles, USA

O-26 High risk screening of Neuronal ceroid lipofuscinosis（NCL）I & II by DBS, and pathologic analysis

○ Rina Itagaki[1], Takeo Iwamoto[3], Arif Hossain Mohammad[1], Hiroko Yanagisawa[1]
Takashi Miyajima[1,2], Chen Wu[1,2], Keiko Akiyama[1], Haruo Shintaku[4], Yoshikatsu Etou[1]
[1]Advanced Clinical Research Center, Institute of Neurological Disorder
[2]Institute of Rare disease, AnGes Co.
[3]Core Laboratory, Institute of Medical Science, Tokyo Jikei University School of Medicine
[4]Department of Pediatrics, Osaka City University Graduate School of Medicine

O-27 LC-MS/MS-based multiplex quantification of LSD enzyme activity

○ Mari Ohira, Ryuichi Mashima, Torayuki Okuyama
National Center for Child Health and Development

O-28 A case of acid sphingomyelinase deficiency（ASMD）: a C133Y mutation in saposin domain

○ Shozo Ota[1], Atsuko Noguchi[2], Yoko Nakajima[3], Tetsuya Ito[3], Daiki Kondo[2], Tsutomu Takahashi[2]
[1]Division of Neonatal Intensive Care Unit, Akita Red Corss Hospital
[2]Department of Pediatrics, Akita University Graduate School of Medicine
[3]Department of Pediatrics, Fujita Health University

16：44〜17：20 Oral 7：Disorders of Carbohydrate Metabolism

Chairperson: Hideo Sugie
(Faculty of Health and Medical Sciences, Tokoha University)

O-29 Analysis of Japanese suspected cases of X-linked glycogenosis type 2

○ Tokiko Fukuda[1], Hidetoshi Ishigaki[1], Taiju Hayashi[1], Rei Urushibata[1], Hideo Sugie[2]
[1]Department of Pediatrics, Hamamatsu University School of Medicine
[2]Faculty of health and Medical Sciences, Tokoha University

O-30 Effect of treatment of severe cardiomyopathy in glucogen storage disease type 3 with Atkins diet

○ Hidehito Kondo[1,2], Jun Narita[2], Takeshi Kimura[2], Toko Shibuya[2], Tokiko Fukuda[3], Hideo Sugie[4]
Keiichi Ozono[2], Norio Sakai[5]
[1]Department of Pediatrics, Japanese Red Cross Kyoto Daiichi Hospital
[2]Department of Pediatrics, Osaka University Graduate school of Medicine
[3]Department of Pediatrics, Hamamatsu University Hospital
[4]Faculitu of Medical and Health Sciences, Tokoha University
[5]Department of Health Science, Child Healthcare and Genetic Science, Osaka University Graduate School of Medicine

O-31 GBE1 mutation analysis via protein expression studies in non-progressive form of glycogenosis type 4

○ Hiroyuki Iijima[1], Reiko Iwano[1], Yukichi Tanaka[2], Koji Muroya[1], Tokiko Fukuda[3], Hideo Sugie[4]
Kenji Kurosawa[5], Masanori Adachi[1]
[1]Department of Endocrinology and Metabolism, Kanagawa Children's Medical Center
[2]Department of Pathology, Kanagawa Children's Medical Center
[3]Department of Pediatrics, Hamamatsu University School of Medicine
[4]Faculty of Health and Medical Sciences, Tokoha University
[5]Division of Medical Genetics, Kanagawa Children's Medical Center

17：20〜18：32　Oral 8：Screening

Chairperson: Yosuke Shigematsu

（Department of Pediatrics, University of Fukui）

O-32　The efficacy of a second-tier test for newborn screening using LC/MS/MS method

○ Nobuyuki Ishige[1], Kazuhiro Watanabe[1], Satomi Hasegawa[1], Kaoru Konishi[1], Yasumi Sera[1]
Mika Ishige[2]

[1]Division of Newborn Screening, Tokyo Health Service Association
[2]Department of Pediatrics and Child Health, Nihon University School of Medicine

O-33　Newborn screening for CPT2 deficiency in Japan using（C16 + C18:1）/C2 and C14/C3

○ Go Tajima[1,2], Keiichi Hara[3], Miyuki Tsumura[2], Reiko Kagawa[2], Satoshi Okada[2], Miori Yuasa[4]
Ikue Hata[4], Yosuke Shigematsu[4], Seiji Yamaguchi[5]

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[2]Department of Pediatrics, Hiroshima University Graduate School of Biomedical & Health Sciences
[3]Department of Pediatrics/Institute for Clinical Research, National Hospital Organization Kure Medical Center
[4]Department of Pediatrics, School of Medical Sciences, University of Fukui
[5]Department of Pediatrics, Shimane University Faculty of Medicine

O-34　Assessment of infants with abnormal newborn screening for systemic primary carnitine deficiency

○ Kimihiko Oishi[1,2], Lissette Estrella[1], Ashley Birch[1], Ruth Kornreich[1], Lisa Edelmann[1]
Pankaj Prasun[1], Chunli Yu[1], George Diaz[1]

[1]Departments of Genetics and Genomic Sciences, Pediatrics, Icahn School of Medicine at Mount Sinai, New York, USA
[2]Department of Pediatrics, Jikei University School of Medicine

O-35　Long-term outcomes in the patients diagnosed by tandem mass screening in trial period

○ Chikahiko Numakura[1], Go Tajima[2], Hironori Kobayashi[3], Osamu Sakamoto[4], Yosuke Shigematsu[5]
Seiji Yamaguchi[3]

[1]Department of Pediatrics, Yamagata University Faculty of Medicine
[2]Division of Neonatal Screening, Research Institute, National Center for Child Health and Development
[3]Department of Pediatrics, Shimane University Faculty of Medicine
[4]Department of Pediatrics, School of Medicine, Tohoku University
[5]Department of Pediatrics, Faculty of Medical Sciences, University of Fukui

O-36　Preliminary study on NBS for inborn errors of cobalamin metabolism by hypomethioninemia

○ Reiko Kagawa[1], Go Tajima[1,2], Takako Maeda[2], Keiichi Hara[3], Yutaka Nishimura[4]
Yosuke Shigematsu[5]

[1]Department of Pediatrics, Hiroshima University Hospital
[2]Division of Neonatal Screening, National Center for Child Health and Development
[3]Department of Pediatrics, National Hospital Organization Kure Medical Center and Chugoku Cancer Center
[4]Maternal and Fetal Medicine Center, Hiroshima City Hiroshima Citizens Hospital
[5]Department of Pediatrics, University of Fukui