DAY1: Oct.12 (Thu)
Hall 1

8:50 ~ Opening Remarks

9:00 ~ 10:00 Oral1: Lysosome 1
Chairperson: Norio Sakai
(Child Healthcare and Genetic Science Laboratory,
Division of Health Science, Osaka University Graduate School
of Medicine)
Hiroshi Kobayashi
(Division of Gene Therapy, Research Center of Medical
Genetics, Department of Pediatrics, The Jikei University
School of Medicine)

O-1 Development of drug screening system using iPS cells derived from a patient with Gaucher
disease type II.
○ Jun Kido¹, Tadahiro Numakawa², Shirou Matsumoto¹, Haruki Odaka², Minami Soga²
Ryutarou Kajihara², Fumio Endo¹, Kimitoshi Nakamura¹, Takumi Era²
¹Department of Pediatrics, Kumamoto University
²Cell Modulation, Institute of Molecular Embryology and Genetics

O-2 A pathological study of the bone lesions in Gaucher disease
○ Kazuya Tsuboi, Hiroshi Yamamoto, Hiromi Goto
LSD Center, Nagoya Central Hospital

O-3 Methylation studies of CpG islands in GLA gene: best clinical phenotype predictor for
heterozygous Fabry female
○ MOHAMMAD ARIF HOSSAIN, Chen Wu, Takashi Miyajima, Hiroko Yanagisawa
Keiko Akiyama, Takeo Iwamoto, Yoshikatsu Eto
Advanced Clinical Research Center, Institute of Neurological Disorders

O-4 Tandem mass spectrometry but not fluorimetry readily distinguishes Pompe-affected
patients from pseudodeficiency in dried blood spots.
○ Hsuan-Chieh Liao¹, Hsiao-Jan Chen¹, Chuan-Chi Chiang¹, Michael H. Gelb²
¹Chinese Foundation of Health, newborn screening center
²University of Washington, Department of Chemistry

O-5 Newborn screening for Mucopolysaccharidosis type II in Taiwan: A One-and-a-half year Report.
○ Hsiao-Jan Chen¹, Min-Ju Chan¹, Mei-Ying Liu¹, Shuan-Pei Lin², Arun Babu Kumar³
Michael H. Gelb³, Chuan-Chi Chiang¹
¹Neonatal Screening Center, The Chinese Foundation of Health
²MacKay Memorial Hospital, Taipei, Taiwan
³Department of Chemistry, University of Washington
10:00 ~ 10:48 Oral2: Lysosome 2  
Chairperson: Kimitoshi Nakamura  
(Department of Pediatrics, Graduate School of Medical Sciences, Kumamoto University)  
Motomichi Kosuga  
(Division of Medical Genetics, National Center for Child Health and Development)

O-6 Evaluation of the efficacy and safety of a new drug candidate for Niemann-Pick disease type C
○Minami Soga1, Jun Kido2, Takumi Era1  
1Department of Cell Modulation, IMEG, Kumamoto University  
2Department of Pediatrics, Graduate School of Medical Sciences, Kumamoto University

O-7 Development of chemical diagnosis for Niemann-Pick disease type C by 5 urinary conjugated abnormal bile acids
○Masamitsu Maekawa1, Isamu Jinnou2, Aya Narita2, Hiroaki Yamaguchi1, Mano Nariyasu1  
1Department of Pharmaceutical Sciences, Tohoku University Hospital  
2Department of Pharmaceutical Sciences, Tohoku University  
3Department of Child Neurology, Tottori University, Faculty of Medicine

O-8 Hematopoietic Stem Cell Transplantation for Patients with Mucopolysaccharidosis II
○Shunji Tomatsu1, 2, Francyne Kubaski1, Hiromasa Yabe1, Yasuyuki Suzuki4, Toshiyuki Seto5  
Takashi Hamazaki5, Seiji Yamaguchi5, Kenji E. Orii7, Tadao Orii7  
1Nemours/Alfred I. duPont Hospital for Children  
2Departments of Biomedical Research and Orthopedics  
3Department of Cell Transplantation and Regenerative Medicine, Tokai University School of Medicine  
4Medical Education Development Center, Gifu University Medical Education Development Center, Gifu University  
5Department of Pediatrics, Osaka City University Graduate School of Medicine  
6Department of Pediatrics, Shimane University  
7Department of Pediatrics, Graduate School of Medicine, Gifu University

O-9 Analysis of safety of pentosan polysulphate in young patients with mucopolysaccharidosis type VI
○Mahoko Furujo1, Kenji Orii2, Shunji Tomatsu3, Yasuyuki Suzuki4, Toshiyuki Fukao5  
1Department of Pediatrics, NHO Okayama Medical Center  
2Division of Neonatal intensive Care Unit, Gifu University Hospital  
3Nemours/Alfred I. duPont Hospital for Children  
4Medical education development Center, Gifu University  
5Department of Pediatrics, Gifu graduate school of medicine
10:48 ~ 11:36  Oral3: Lysosome 3  

Chairperson: Tsutomu Takahashi  
(Department of Pediatrics, Akita University Graduate School of Medicine)  

Hiroshi Mochizuki  
(Saitama Children’s Medical Center)  

O-10  Clinical, biochemical and molecular characterization of 8 patients with mucolipidosis  
○Mari Asakura¹, Yasuyuki Fukuhara¹, Motomichi Kosuga¹, Torayuki Okuyama²  
¹Division of Medical Genetics, National Center for Child Health and Development  
²Department of Clinical Laboratory Medicine, National Center for Child Health and Development  

O-11  The clinical profile in Japanese cystinosis patients and the treatment effect of cysteamine.  
○Moe Akahira¹, Yukiko Shimizu², Yoshiaki Okuma³, Tadashi Okamura², Toshihiro Ohura⁴  
¹Division of Medical Genetics, Kanagawa Children’s Medical Center  
²Department of Laboratory Animal Medicine, Research Institute, National Center for Global Health and Medicine  
³Department of Pediatrics, National Center for Global Health and Medicine  
⁴Division of Pediatrics, Sendai City Hospital  

O-12  Therapeutic potency of teprenone in comprehensive lysosomal storage diseases  
○Yohta Shimada¹, Natsumi Ishii¹, Hiroo Hoshina², Takashi Higuchi¹, Hiroshi Kobayashi¹, Hiroyuki Ida², Toya Ohashi¹  
²Dept. of Pediatrics, Jikei Univ. Sch. of Med.  

O-13  Mucopolysaccharidosis-plus syndrome: A report of new type of MPS  
○Hidehito Kondo¹, Takanobu Otomo², Nadezda Maksimova³, Norio Sakai¹, Keiichi Ozono¹  
¹Department of Pediatrics, Osaka University Graduate School of Medicine  
²Department of Pathophysiology and Metabolism, Kawasaki Medical School  
³Laboratory of Genome Medicine, Clinics of Medical Institute, North East Federal University  

11:40 ~ 12:30  Sponsored Seminar 1  

Chairperson: Haruo Shintaku  
(Department of Pediatrics, Osaka City University, Graduate School of Medicine, Osaka Japan)  

Development of Novel Therapy for Central Nervous System Involvement of Lysosomal Storage Diseases  
Torayuki Okuyama  
Department of Clinical Laboratory Medicine, Center for Lysosomal Storage Diseases, National Center for Child Health and Development, Tokyo, Japan
13:50 ~ 14:20  JSIMD Annual General Meeting

14:20 ~ 14:50  JSIMD Award Lecture
   Chairperson: Fumio Endo
   (Kumamoto-Ezuko Medical Center for The Severely Disabled)
   Study on molecular genetics, diagnosis and treatment of trace element disorders
   Hiroko Kodama
   Department of Health and Dietetics, Faculty of Health and Medical Sciences, Teikyo Heisei University

15:00 ~ 16:00  Special Lecture 1
   Chairperson: Akira Ohtake
   (Department of Pediatrics, Saitama Medical University)
   Citrin deficiency -Fateful encounter-
   Takeyori Saheki
   Department of Hygiene and Health Promotion Medicine, Kagoshima University Graduate School
   of Medical and Dental Sciences

16:10 ~ 17:10  Educational Lecture 2
   Chairperson: Torayuki Okuyama
   (Department of Clinical Laboratory Medicine,
   Center for Lysosomal Storage Diseases,
   National Center for Child Health and Development)
   Molecular diagnostics of Inherited Metabolic Diseases via RNA sequencing
   Holger Prokisch
   Technical University Munich, Helmholtz Zentrum München

17:20 ~ 18:50  Symposium 1: Mitochondrial disorder - basic researches for understanding the disorder
   Organizer: Masakazu Kohda
   (Intractable Disease Research Center, Juntendo University
   Division of Translational Research, Research Center for Genomic Medicine, Saitama Medical University)

S1-1  Mitochondrial Dynamics in Aging and Disease
   ○Takeshi Tokuyama, Shigeru Yanagi
   Dept. of life Sci., Tokyo univ. of Pharm. and Life sci.

S1-2  Reverse genetic studies on mitochondrial DNAs with pathogenic mutations in mice
   Kazuto Nakada
   Faculty of Life and Environmental Sciences, University of Tsukuba
S1-3 Challenging to mitochondrial gene therapy by MITO-Porter, a nano device for mitochondrial drug delivery

○ Yuma Yamada, Hideyoshi Harashima
Faculty of Pharmaceutical Sciences, Hokkaido University
DAY1：Oct.12（Thu）
Hall 2

9：00～10：00 Oral4：Screening
Chairperson: Nobuyuki Ishige
(Division of Newborn Screening, Tokyo Health Service Association)
Masaki Kanazawa
(Chiba Kaihin Municipal Hospital)

O-14  Multi-plex LSD enzyme assay using mass spectrometry
○ Ryuichi Mashima, Mari Ohira, Akiya Tatsumi, Torayuki Okuyama
National Center for Child Health and Development

O-15  Development of screening test method for Hypophosphatasia using dried blood spot
○ Fumio Endo1, Shinichiro Yoshida2, Kousuke Kumeda2, Kimitoshi Nakamura3
1Kumamoto Ezuko Ryoiku Iryo Center
2Newborn screening center, The Chemo-Sero-Therapeutic Research Institute
3Dep Ped, Grad Schl of Med,Univ Kumamoto

O-16  Clinical applications of NGS for the diagnosis of different genetic diseases and newborn screening
○ JIN-SUNG LEE1, Hyunjoo Lee1, Cheol-Ho Lee1, Yangrae Cho2, Jongsun Jung2, Yoon Hee Ko1
1Department of Pediatrics, Yonsei University College of Medicine
2Syntekabio, Inc., Seoul, KOREA

O-17  Mass screening for VLCAD deficiency in Tokyo; Results and study for the future
○ Nobuyuki Ishige1, Kazuhiro Watanabe1, Satomi Hasegawa1, Kaoru Konishi3
Michiko Mashita1, Sera Yasumi1, Mika Ishige2, Misao Owada1
1Division of Newborn Screening, Tokyo Health Service Association
2Department of Pediatrics and Child Health, Nihon University School of Medicine

O-18  Fatty acid oxidation disorders and sudden unexpected death in infancy:
Is neonatal tandem mass screening system useful?
○ Masato Arao1, Taro Yamazaki1, Hiroko Harashima1, Aya Takada2, Kazuyuki Saito2
Yosuke Shigematsu1, Keiichi Hara4, Go Tajima2, Akira Ohtake1
1Department of Pediatrics, Saitama Medical University Hospital
2Department of Forensic Medicine, Saitama Medical University
3Department of Health Science, Faculty of Medical Sciences, University of Fukui
4Department of Pediatrics, National Hospital Organization Kure Medical Center
Division of Neonatal Screening, National Center for Child Health and Development
10：00～10：48 Oral5：Organic acid disorders and fatty acid disorders 1
Chairperson: Go Tajima
(Division of Neonatal Screening, Research Institute, National Center for Child Health and Development)
Masahisa Kobayashi
(Department of Pediatrics, The Jikei University School of Medicine)

O-19 Methylmalonyl-CoA mutase deficiency with early-onset optic atrophy: A case report
○Chikahiko Numakura1, Shion Hayashi2, Osamu Sakamoto3, Kiyoshi Hayasaka1,4
1Department of Pediatrics, Yamagata University Faculty of Medicine
2Department of Ophthalmology, Yamagata University Faculty of Medicine
3Department of Pediatrics, Tohoku University School of Medicine
4Department of Pediatrics, Miyukikai Hospital

O-20 A Case of Combined malonic and methylmalonic aciduria
○Daiki Kondo1, Wakako Kikuchi1, Atsuko Noguchi1, Ikuko takahashi1, Tsutomu Takahashi1
Tomoko Uehara2, Kenjiro Kosaki2
1Department of Pediatrics, Akita University School of Medicine
2Center for Medical Genetics, Keio University School of Medicine

O-21 Phenotypes and genotypes of 52 Chinese patients with propionic acidemia
○Yi Liu1, Yuan Ding1, Dongxiao Li1, Xiyuan Li1, Ying Jin1, Jinqing Song1, Yao Zhang1
Yanling Yang1, Haixia Li2, Lili Jiao2
1Department of Pediatrics, Peking University First Hospital
2Department of Clinical Laboratory, Peking University First Hospital

O-22 Cluster Seizure in A Patient with Methylenetetrahydrofolate Reductase Deficiency due to Novel MTHFR mutation
○Lina Xie, Qian Chen, Shuo Feng, Shuhua Chen, Yingying Mao, Zhijie Gao, Keming Xu
Children’s Hospital Capital Institute of Pediatrics
**O-23 Insufficient ketogenesis in 3-hydroxybutyrate dehydrogenase (Bdh1) KO mice in fasting test**

- Hiroki Otsuka¹, Takeshi Kimura¹, Yasuhiro Ago¹, Mina Nakama², Abdelkreem Elsayed¹
- Yuka Aoyama², Hideo Sasai¹, Hidenori Ohnishi¹, Masatake Osawa³, Yusuke Kawashima⁵
- Osamu Ohara⁵, Seiji Yamaguchi⁶, Toshiyuki Fukao¹

¹Department of Pediatrics, Graduate School of Medicine, Gifu University
²Division of Clinical Genetics, Gifu University Hospital
³Department of Biomedical Sciences, College of Life and Health Sciences, Chubu University
⁴Department of Regenerative Medicine and Applied Medical Sciences, Graduate School of Medicine, Gifu University
⁵Laboratory for Integrative Genomics, RIKEN Center for Integrative Medical Sciences (IMS)
⁶Department of Pediatrics, Graduate School of Medicine, Shimane University

**O-24 Open-label clinical trial of bezafibrate for fatty acid oxidation disorders in Japan**

- Kenji Yamada¹, Hideaki Shiraishi², Naoko Asahina², Saki Yokoshiki³, Takashi Miyakoshi³
- Kota Ohno³, Toshiyuki Isoe³, Hiroshi Hayashi³, Seiji Yamaguchi¹, Norihiro Sato³

¹Department of Pediatrics, Shimane University Faculty of Medicine
²Department of Pediatrics, Kansai Medical University
³Hokkaido University Hospital Clinical Research and Medical Innovation Center

**O-25 Expression Analysis of the Recombinant Proteins Related with Human HSD10 Disease**

- Hideo Sasai¹, Hidenori Ohnishi¹, Shohei Akagawa², Kazuhisa Akiha³, Yukihiro Hasegawa³
- Masahisa Kobayashi¹, Hiroki Otsuka¹, Yuka Aoyama¹, Toshiyuki Fukao¹

¹Department of Pediatrics, Graduate School of Medicine, Gifu University
²Department of Pediatrics, Kansai Medical University
³Department of Endocrinology and Metabolism, Tokyo Metropolitan Children's Medical Center
⁴Department of Pediatrics, The Jikei University School of Medicine

**O-26 Five Japanese cases with glutaric acidemia type II detected by newborn screening**

- Kenji Yamada¹, Hironori Kobayashi¹, Yuki Hasegawa¹, Akio Takahashi², Haruo Shintaku³
- Hideo Sasai¹, Toshiyuki Fukao¹, Seiji Yamaguchi¹, Takeshi Taketani¹

¹Department of Pediatrics, Shimane University Faculty of Medicine
²Department of Pediatrics, Morioka Children's Hospital
³Department of Pediatrics, Osaka City University Faculty of Medicine
⁴Department of Pediatrics, Gifu University School of Medicine
11:40 ~ 12:30  SLEIMPN Recommending Lecture  
Chairperson: Mitsuru Kubota  
(Department of General Pediatrics & Interdisciplinary Medicine, National Center for Child Health and Development)

NGS in the diagnosis of mitochondrial diseases: novel disorders and unexpected phenotypes.  
○Jose Abdennur, Mariella Simon, Alex Stover, Shaina Efthekarian  
CHOC Children’s, Division of Metabolic Disorders. Orange CA, USA/FESEN Buenos Aires, Argentina

12:40 ~ 13:40  Luncheon Seminar 1  
Chairperson: Yoshikatsu Eto  
(Advanced Clinical Research Center & Institute for the treatment of Genetic Disease, Institute of Neurological Disorders)

Treatment strategy for Gaucher Disease  
-The Results of VPRIV Extension Study and Therapeutic Goals-  
Hiroyuki Ida  
Department of Pediatrics, The Jikei University School of Medicine
DAY1：Oct.12（Thu）
Hall 3

9:00 ~ 11:36  Live broadcast：Hall 2

11:40 ~ 12:10  Educational Lecture 1
Chairperson: Kei Murayama
(Department of Metabolism, Chiba Children's Hospital)

Integrative analysis of transcriptome and cellular bioenergetics profiles
○ Vicente Yépez¹, Laura Kremer², Daniel Bader¹, Mirjana Gusic², Holger Prokisch², Julien Gagneur³
¹Technical University Munich, ²Helmholtz Zentrum München

12:40 ~ 13:40 Luncheon Seminar 2
Chairperson: Torayuki Okuyama
(Department of Clinical Laboratory Medicine,
Center for Lysosomal Storage Diseases,
National Center for Child Health and Development)

“Pathophysiology of Lysosomal Acid Lipase Deficiency (LAL-D)”
John Jay Gargus
Center for Autism Research and Translation/Genetics and Genomics, Pediatrics, Physiology & Biophysics University of California, Irvine
DAY1：Oct.12（Thu）
Poster Exhibition Hall

17：20～18：50 Poster Presentation

< Block1 > 17：20～17：50 Lysosome 1
Chairperson: Kazuya Tsuboi
(LSD Center, Nagoya Central Hospital)

P-1 Evidence of Mechanism of Action of Migalastat in the Japanese Subgroup of the Phase 3 ATTRACT trial

○Toya Ohashi\(^1\), Ichiei Narita\(^2\), Norio Sakai\(^3\), Takashi Hamazaki\(^4\), Jay Barth\(^5\), Hjalmar Lagast\(^5\)
Nina Skuban\(^5\), Julie Yu\(^6\), Jeff Castelli\(^6\), Christopher Viereck\(^6\)

\(^1\)Department of Pediatrics, The Jikei University School of Medicine
\(^2\)Division of Clinical Nephrology and Rheumatology, Niigta University
\(^3\)Osaka University Graduate School of Medicine, Division of Health Science, Child Healthcare and Genetic Science Laborator
\(^4\)Department of Pediatrics, Osaka City University Graduate School of Medicine
\(^5\)Amicus Therapeutics

P-2 Comparison of variants causing later-onset type Fabry disease with functional polymorphisms

○Masashi Shigenaga\(^1\), Takahiro Tsukimura\(^1\), Atsuko Sato\(^2\), Seiji Saito\(^3\), Tadayasu Togawa\(^1\)
Hitoshi Sakuraba\(^2\)

\(^1\)Department of Functional Bioanalysis, Meiji Pharmaceutical University
\(^2\)Department of Clinical Genetics, Meiji Pharmaceutical University
\(^3\)Department of Medical Management and Informatics, Hokkaido Information University

P-3 The relationship between vertebral basilar artery findings and hearing function in Fabry disease

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

P-4 High sensitive TroponinI of 45 Fabry patients

○Hiromi Goto, Kazuya Tsuboi, Hiroshi Yamamoto
Lysosome Center, Nagoya Central Hospital

P-5 Association with methylation and autophagic flux in Fabry disease

○Hiroko Yanagisawa\(^1\), Mohammad Arif Hossain\(^1\), Takashi Miyajima\(^2\), Keiko Akiyama\(^1\)
Yoshikatsu Eto\(^1\)

\(^1\)Advanced Clinical Research Center, Institute for Neurological Disorders
\(^2\)Rare Disease Research Center, AnGes
P-6  Study of relations between HDL-Cholesterol and vascular lesions in Fabry disease
○Hiroki Katsuta¹, Kazuya Tsuboi², Hiroshi Yamamoto², Hiromi Goto²
¹Junior Resident, Nagoya Central Hospital
²LSD Center, Nagoya Central Hospital

17:50 ~ 18:15  Lysosome 2
Chairperson: Eiji Nanba
(Research Center for Bioscience and Technology)

P-7  Mitochondrial functional analysis in the Gaucher’s disease patients
○Naomi Kuranobu¹, Aya Narita², Masaru Shimura¹, Makiko Tajika¹, Keiko Ichimoto¹
Ayako Matsunaga¹, Tomoko Tsuruoka¹, Masaki Takayanagi¹, Akira Ohtake³, Kei Murayama¹
¹Department of Metabolism, Chiba Children’s Hospital
²Department of Child Neurology, Tottori University
³Department of Pediatrics, School of Medicine Saitama Medical University

P-8  Enzyme replacement therapy and chronological cytokine profile one patient of perinatal-lethal Gaucher’s disease
○Takeaki Sasamoto, Masaru Shimura, Yusuke Suganami, Junya Nakajima, Shonosuke Nara
Hiroki Ishii, Daisuke Sunohara, Yasuyo Kashiwagi, Hisashi Kawashima
Department of pediatrics, Tokyo medical University

P-9  Drug development for GM1 gangliosidosis
○Ryutaro Kajihara, Takumi Era
Department of Cell Modulation, Institute of Molecular Embryology and Genetics, Kumamoto University

P-10  A case of Tay-Sachs disease difficult to distinguish from acute encephalopathy
○Naoto Iwanami, Daisuke Azuma, Hiroko Suzuki, Asako Arai, Tamotsu Matsunaga
Todachuo General Hospital

P-11  The case of late onset Tay-Sachs disease, subtle hyperacusis led the diagnosis
○Mihoko Sakurai¹, Junji Azuma¹, Yusuke Hamada², Eri Kizima¹, Koji Tominaga³
Yoshimi Mizoguchi¹, Tunesuke Shimotuji¹, Takehisa Yamamoto¹, Norio Sakai³
¹Department of Pediatrics, Minoh City Hospital
²Department of Pediatrics, Japan Community health care organization Osaka hospital
³Department of Pediatrics, Osaka University hospital
18:15 ~ 18:45  Lysosome 3

Chairperson: Toju Tanaka
(Center for Genetics & Metabolic Disorder and Clinical Research
National Hospital Organization Hokkaido Medical Center)

P-12  MLPA analysis for Japanese Krabbe patients without mutation in direct sequence for detection of deletion mutation
○ Kaori Miyana1, Hidehito Kondo1, Keiichi Ozono1, Norio Sakai2
1Division of Pediatrics, Osaka University Graduate School of Medicine
2Division of Health Science, Osaka University Graduate School of Medicine

P-13  Clinical and molecular analysis of six novel GALK mutations identified in 9 Chinese children with Krabbe disease
○ Lifang Dai, Tongli Han, Xinying Yang, Xu Wang, Jiuwei Li, Junlan Lu, Wuchang Zhang
Xiaotun Ren, Fang Fang
Department of Neurology, Beijing Children’s Hospital, Capital Medical University

P-14  Late Infantile Neuronal Ceroid Lipofuscinoses Due to Novel CLN5 Variants In a Chinese Family
○ Xiaotun Ren, Fang Fang, Husheng Wu, Zhimei Liu
Department of Neurology, Beijing Children’s Hospital, Capital Medical University, National Center for Children’s Health, Beijing, China

P-15  Modeling the neuronal dysfunction in sialidosis using patient-derived iPSCs
○ Haruki Odaka1, Tadahiro Numakawa2, Minami Soga3, Jun Kido3, Ryutaro Kajihara4
Toshiya Okumiya4, Hirokazu Furuya5, Takafumi Inoue1, Takumi Era2
1Department of Life Science and Medical Bioscience, School of Advanced Science and Engineering, Waseda University
2Department of Cell Modulation, Institute of Molecular Embryology and Genetics, Kumamoto University
3Department of Pediatrics, Graduate School of Medical Sciences, Kumamoto University
4Department of Biomedical Laboratory Sciences, Faculty of Life Science, Kumamoto University
5Department of Neurology, Kochi Medical School, Kochi University

P-16  A summary of the lysosomal enzyme assay for patients with leukodystrophy in our facility
○ Sachiko Nakaoka1, Toko Shibuya1, Kaori Miyana1, Hidehito Kondo1, Norio Sakai2
Keiichi Ozono1
1Department of Pediatrics, Osaka University Graduate School of Medicine
2Division of Health Science, Osaka University Graduate School of Medicine

P-17  Newborn screening for Pompe disease in northern Kawasaki city
○ Ohsuke MIGITA1, Kojiro Takahiro2, Yusuke Miyaji2, Yusaku Miyamoto2, Isamu Hokuto1
Motomichi Kosuga3, Torayuki Okuyama3, Hitoshi Yamamoto1
1Department of Pediatrics, St. Marianna University School of Medicine
2Division of Pediatrics, Kawasaki Municipal Tama Hospital, St. Marianna University School of Medicine.
3Center for Lysosomal Storage Diseases, National Center for Child Health and Development
P-18  CSF heparan sulfate is a useful CNS biomarker for Mucopolysaccharidosis Type II
Sachiho Kida, Noboru Tanaka, Hideto Morimoto
Research Division, JCR Pharmaceuticals Co., Ltd.

P-19  Case report: Elosulfase alfa enzyme replacement therapy in a Japanese patient with Morquio A syndrome
Misako Hiramatsu, Kimitoshi Nakamura
1Division of Pediatrics, Nishibeppu National Hospital
2Department of Pediatrics, Kumamoto University Hospital

P-20  Case report: 10-year enzyme replacement therapy in Japanese siblings with mucopolysaccharidosis VI
Mahoko Furujo, Motomichi Kosuga, Torayuki Okuyama
1Department of Pediatrics, National Okayama Medical Center
2Center for Lysosomal Storage Diseases, National Center for Child Health and Development

P-21  Influence of mannose-6-phosphate content on iduronate-2-sulfatase in enzyme replacement therapy in mice.
Tomoki Fukatsu, Atsushi Imakiire, Sachiho Kida, Masafumi Kinoshita, Satowa Tanaka, Hiroyuki Sonoda
Biopharmaceutical Innovation Research Institute, Research Division, JCR Pharmaceuticals Co., Ltd.

P-22  Brain targeted high dose enzyme replacement therapy with immune tolerance induction in a murine model of MPS II
Kentaro Yokoi, Takayuki Yokoi, Shimada Yohta, Higuchi Takashi, Hiroshi Kobayashi, Toya Ohashi, Hiroyuki Ida
1Department of Pediatrics, The Jikei University School of Medicine
2Division of Gene Therapy, Research Center for Medical Sciences, The Jikei University School of Medicine

P-23  Clinical features of mucopolysaccharidosis type VII "Case series in Japan"
Saki Kasuga, Kadono Chihoo, Satoshi Kudoh, Hidetomi Terai, Seto Toshiyuki
Takashi Hamazaki, Haruo Shintaku
1Department of Pediatrics, Osaka City University Graduate School of Medicine, Osaka City University
2ATOX Co., Ltd
3Department of Orthopedic Surgery, Osaka City University Graduate School of Medicine, Osaka City University
17:50 ~ 18:20  Lysosome 5+Screening 1
Chairperson: Satoshi Ishii
(Department of Matrix Medicine, Faculty of Medicine, Oita University)

P-24  Glycosaminoglycan Levels in Dried Blood Sopts of Patients of MPS and ML
○Shunji Tomatsu¹, Francyne Kubaski¹, Yasuyuki Suzuki², Kenji Orii³, Robert W. Mason¹
Vũ Chí Dũng⁴, Seiji Yamaguchi⁵, Hironori Kobayashi³, Toshiyuki Fukao³, Tadao Orii³
¹Nemours/Alfred I. duPont Hospital for Children
²Medical Education Development Center, Gifu University
³Department of Pediatrics, Graduate School of Medicine, Gifu University
⁴Vietnam National Children’s Hospital, Department of Medical Genetics, Metabolism & Endocrinology
⁵Department of Pediatrics, Shimane University

P-25  LC/MS/MS measurement of glycosaminoglycans in amniotic fluid of a MPS VII fetus
○Shunji Tomatsu¹, Francyne Kubaski¹, Robert W. Mason¹, Roberto Giugliani²
¹Nemours/Alfred I. duPont Hospital for Children, ²Medical Genetics Service, HCPA

P-26  Method for measuring mannose 6-phosphate residue contents of enzyme preparations in Lysosomal diseas
○Minori Kanzaki¹, Masashi Shigenaga¹, Yuya Tayama¹, Takahiro Tsukimura¹
Tadayasu Togawa¹, Hitoshi Sakuraba²
¹Department of Functional Bioanalysis, Meiji Pharmaceutical University
²Department of Clinical Genetics, Meiji Pharmaceutical University

P-27  Sphingoglycolipid analysis in biological samples using a chiral culumn
○Yuko Fujiwara , Kotaro Hama, Kazuaki Yokoyama
Faculty of Pharmaceutical Sciences, Teikyo University

P-28  Diagnosis of Niemann-Pick disease type C using new potential biomarkers integrating with multivariate analysis
○Chen Wu¹, Takeo Iwamoto², Hiroko Yanagisawa¹, Keiko Akiyama¹, Takashi Miyajima¹
Junko Igarashi³, Mohammad Arif Hossain¹, Yoshikatsu Eto¹
¹Advanced Clinical Research Center, Institute of Neurological Disorders
²Core Research Facilities for Basic Science, Molecular Cell Biology, The Jikei University School of Medicine, Tokyo, Japan
³AnGes, Inc.
P-29  Glycosaminoglycan Assay as First-tier for MPS Newborn Screening

○Shunji Tomatsu1, Francyne Kubaski1, Akiko Nakatomi2, Haruo Shintaku3
Hironori Kobayashi1, Seiji Yamaguchi4, Yasuyuki Suzuki5, Tadao Orii6, Toshiyuki Fukao6
Adriana M. Montaño7

1Nemours/Alfred I. duPont Hospital for Children
2Department of Pediatrics, Nagasaki University
3Department of Pediatrics, Osaka City University Graduate School of Medicine
4Department of Pediatrics, Shimane University
5Medical Education Development Center, Gifu University
6Department of Pediatrics, Gifu University, 7Department of Pediatrics, Saint Louis University

18:20 ~ 18:45  Screening 2
Chairperson: Yasutsugu Chinen
(Child Health and Welfare (Pediatrics), Faculty of Medicine, University of Ryukyus)

P-30  Pilot study of newborn screening for mucopolysaccharidosis type I and II

○Ken Momosaki1, Shinichiro Yoshida2, Kousuke Kumeda2, Fumio Endo3
Kimitoshi Nakamura1

1Department of Pediatrics, Kumamoto University
2Kaketsuken (The Chemo-Sero-Therapeutic Research Institute)
3Kumamoto Ezuko Ryoiku Iryo Center

P-31  Enzymatic screening in Dried Blood Spot (DBS) of Mucopolysaccharidosis type VII

○Takashi Miyajima1, Jyunko Igarashi2, Chen Wu3, Hiroko Yanagisawa1
Mohammad Arif Hossain1, Keiko Akiyama1, Takashi Hamazaki1, Takeo Iwamoto5
Yoshikatsu Eto1

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2AnGes, Inc. Rare Disease Research Center
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5Division of Molecular Cell Biology, Core Research Facilities for Basic Science, The Jikei University School of Medicine

P-32  Newborn Screening of Neuronal Ceroid Lipofuscinosis (NCL) I and II by Dried Blood Spots (DBS)

○Rina Itagaki1, Keiko Yaginuma1, Masahiro Endo1, Keiko Akiyama1, Hiroko Yanagisawa1
Takeo Iwamoto5, Haruo Shintaku3, Yoshikatsu Eto1, 4

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2Division of Molecular Cell Biology, Core Research Facilities for Basic Science, The Jikei University School of Medicine
3Department of Pediatrics Osaka City University Graduate School of Medicine
4The Jikei University School of Medicine
P-33  **Factors to influence Plasma LysoGb3 Levels in Fabry Disease- Age, Sex and Antibody against ERT**

○ Chen Wu¹, Takeo Iwamoto², Mohammad Arif Hossain¹, Takashi Miyajima¹
   Keiko Akiyama¹, Hiroko Yanagisawa¹, Junko Igarashi³, Yoshikatsu Eto¹
   ¹Advanced Clinical Research Center, Institute of Neurological Disorders
   ²Core Research Facilities for Basic Science, Molecular Cell Biology, The Jikei University School of Medicine, Tokyo, Japan
   ³AnGes, Inc.

P-34  **Metabolomics-screening of diseases with multiple disturbed organic, amino acid, sugar metabolism**

○ Tomiko Kuhara, Morimasa Ohse
   Japan Clinical Metabolomics Institute

< Block3 >  17:20 ~ 17:55  Screening 3  
Chairperson: Yuki Omura-Hasegawa  
(Department of Pediatrics, Shimane University Faculty of Medicine)

P-35  **Molecular Diagnosis for Target Metabolic Diseases of Newborn Screening Using a Gene Panel in Japan**

○ Hideo Sasai¹, Fujiki Ryoji², Ohara Osamu², Nakajima Yoko³, Testuya Ito³
   Masahisa Kobayashi⁴, Go Tajima⁴, Osamu Sakamoto⁵, Matsumoto Shiro⁷
   Kimitoshi Nakamura⁷, Takashi Hamazaki⁸, Hironori Kobayashi⁹, Yuki Hasegawa⁹
   Toshiyuki Fukao¹
   ¹Department of Pediatrics, Graduate School of Medicine, Gifu University
   ²Department of Technology Development, Kazusa DNA Research Institute
   ³Department of Pediatrics, School of Medicine, Fujita Health University
   ⁴Department of Pediatrics, The Jikei University School of Medicine
   ⁵Division of Neonatal Screening, NCCHD
   ⁶Department of Pediatrics, Tohoku University School of Medicine
   ⁷Department of Pediatrics, Kumamoto University Graduate School of Medical Sciences
   ⁸Department of Pediatrics, Osaka City Graduate School of Medicine
   ⁹Department of Pediatrics, Shimane University Faculty of Medicine

P-36  **A case of carnitine deficiency, systemic primary with two previously reported PCCA gene mutation in cis form**

○ Fusa Nagamatsu¹, Akira Ohtake², Nobuyuki Ishige³, Hironori Kobayashi⁴, Toshiyuki Fukao⁵
   Yukihiro Hasegawa¹
   ¹Department of Endocrinology and Metabolism, Tokyo Metropolitan Children’s Medical Center
   ²Department of Pediatrics, Saitama Medical University
   ³Tokyo Health Service Association
   ⁴Department of Pediatrics, Shimane University Faculty of Medicine
   ⁵Department of Pediatrics, Graduate School of Medicine, Gifu University
P-37  **PCCB monoallelic mutation (Y435C) and low activity of PCC in a Japanese girl with mild form of propionic acidemia**

○ Daisuke Sugawara, Ko Ichihashi

Department of Pediatrics, Saitama Medical Center Jichi Medical University

P-38  **High C3 and C3/C2 levels at NBS with increased methylmalonate excretion due to maternal antibody to intrinsic factor**

○ Yoko Nakajima¹, Katsuyuki Yokoi¹, Yoshimi Sakai², Tetsushi Yoshikawa¹, Tetsuya Ito¹

¹Department of Pediatrics, Fujita Health University
²Aichi Health Promotion Public Interest Foundation

P-39  **Genetic background of the cases with high C5-OH level**

○ Yuki Omura-Hasegawa¹, Hideo Sasai², Osamu Sakamoto³, Hironori Kobayashi¹

Hiroki Otsuka³, Ryoji Fujiki¹, Osamu Ohara¹, Toshiyuki Fukao²

¹Department of Pediatrics, Shimane University Faculty of Medicine
²Department of Pediatrics, Graduate School of Medicine, Gifu University
³Department of Pediatrics, Tohoku University School of Medicine
⁴Department of Technology Development, Kazusa DNA Research Institute

P-40  **Maternal glutaric acidemia type I detected by expanded newborn screening: First case in Thailand**

○ Somporn Liammongkolkul¹, Boonchai Boonyawat², Kasinat Sanomcham¹

Achara Sathienkijkanchai¹, Pornswan Wasant¹, Nithiwat Vatanavicharn¹

¹Division of Medical Genetics, Department of Pediatrics, Faculty of Medicine Siriraj Hospital, Mahidol University
²Division of Genetics, Department of Pediatrics, Phramongkutklao Hospital/College of Medicine, Bangkok, Thailand

P-41  **Incidence and natural history of β-ureidopropionase deficiency based on a NBS study at Kurume University.**

○ Natsuko Shibata¹, Yoriko Watanabe¹,², Kaori Fukui³, Hiromi Isii¹, Misa Inaba¹

Kyoko Tashiro¹, Kumiko Aoki¹, Yoko Nakajima³, Takahiro Inokuchi¹, Naohisa Uchimura¹,⁴

¹Research Institute of Medical Mass Spectrometry, Kurume University School of Medicine
²Department of Pediatrics and Child Health, Kurume University School of Medicine
³Department of Pediatrics, Fujita Health University
⁴Department of Neuropsychiatry, Kurume University School of Medicine
17:55 ~ 18:20  Organic acid disorders and fatty acid disorders 1
Chairperson: Ayako Matsunaga
(Department of Metabolism, Chiba Children’s Hospital)

P-42  A case of methylmaronic acidemia diagnosed by neonatal screening without any symptoms
○Yusuke Hamada¹, Toko Shibuya², Hidehito Kondo², Kanako Kishimoto¹, Yasuhiro Maeda³, Noriyuki Namba¹, Norio Sakai²
¹Department of Pediatrics, Osaka hospital, Japan Community Healthcare Organization
²Department of Pediatrics, Osaka University hospital
³Department of Hospital Pharmacy, Graduate School of Pharmaceutical Sciences, Nagoya City University

P-43  Effectiveness of carglumic acid on ammonia detoxification in methylmalonic acidemia
○Aiko Gishi¹, Saki Kasuga², Makiko Fuyuki², Takashi Hamazaki², Shintaku Haruo²
¹Department of Pediatrics, Osaka City University
²Department of Pediatrics, Osaka City University Graduate School of Medicine

P-44  A report of a patient with Methylmalonic acidemia who developed CML 9 years after a liver transplantation
○Kyoko Tashiro¹, Yoriko Watanabe¹, ², Kaori Fukui², Yuichi Ochi², Shinichiro Nakagawa², Shuichi Osano³, Yushiro Yamashita²
¹Research Institute of Medical Mass Spectrometry, Kurume University School of Medicine
²Department of Pediatrics and Child Health, Kurume University School of Medicine

P-45  long-term clinical course of two sibling cases with methylmalonic acidemia
○Ryosuke Bo, Hiroyuki Awano, Masashi Nagai, Masaaki Matsumoto, Kazumi Tomioka, Kaori Maeyama, Tsukasa Tanaka, Masahiro Nishiya, Hiroaki Nagase, Kazumoto Iijima
Department of Pediatrics, Kobe university Graduate School of Medicine

P-46  Association between propionyl-CoA carboxylase activity and severity of propionic acidemia
○Yasuhiro Maeda¹, Kana Gotoh¹, Yoko Nakajima², Go Tajima¹, Yuji Hotta¹, Tomoya Kataoka¹, Kazunori Kimura¹, Tetsuya Ito²
¹Department of Hospital Pharmacy, Graduate School of Pharmaceutical Sciences Nagoya City University
²Department of Pediatrics, Fujita Health University
³National Center for Child Health and Development
⁴Department of Clinical Pharmacy, Graduate School of medical Sciences Nagoya City University
18:20 ~ 18:45 Organic acid disorders and fatty acid disorders 2
Chairperson: Kenji Yamada
(Department of Pediatrics, Shimane University Faculty of Medicine)

P-47 Biotin Responsiveness in 10 Korean Patients with Propionic Acidemia
○Sook Za Kim¹, Wung Joo Song², William Nyhan³
  ¹Korea Genetics Research Center/KSZ Children’s Hospital
  ²Korea Genetics Research Center/KSZ Children’s Hospital, Cheongju, South Korea
  ³UCSD Biochemical Genetics Laboratory, La Jolla, CA, USA

○Khunton Wichajarn
Khon Kaen University

P-49 Elevated 3-hydroxyisovalerylcarnitine on newborn screening suggests mitochondrial disease
○Keisuke Okada¹, Ikuma Mushia¹, Masato Arao¹, Yamazaki Taro¹, Toru kikuchi¹
  Megumi Saito⁵, Hiroshi Mochizuki³, Kei Murayama¹, Yasushi Okazaki⁵, Akira Ohtake¹
  ¹Department of Pediatrics Faculty of Medicine, Saitama Medical University
  ²Intractable Disease Center, Saitama Medical University
  ³Department of Metabolism and Endocrinology, Saitama Children’s Medical Center
  ⁴Department of Metabolism, Chiba Children’s Hospital
  ⁵Research Center for Genomic Medicine, Saitama Medical University

P-50 Glutaric Aciduria type II in Thailand: Phenotypic spectrum, Biochemical, and Molecular features
○Nithiwat Vatanavicharn¹, Somporn Liammongkolkul¹, Boonchai Boonyawat²
  Achara Sathienkijkanchai¹, Pornswan Wasant¹, Seiji Yamaguchi³
  ¹Department of Pediatrics, Faculty of Medicine Siriraj Hospital, Mahidol University, Bangkok, Thailand
  ²Department of Pediatrics, Phramongkutklao College of Medicine, Bangkok, Thailand
  ³Department of Pediatrics, Shimane University School of Medicine, Shimane, Japan

P-51 Blood purification therapy and clinical course at metabolic crisis of organic acidemia
○Kaname Hirashima¹, Shiro Matsumoto², Kido jun³, Rieko Sakamoto³, Hiroshi Mitsubuchi¹
  Fumio Endo¹, Kimitoshi Nakamura²
  ¹Department of Pediatrics, Kumamoto University School of Medicine
  ²Department of Pediatrics, Kumamoto University School of Medicine
  ³Lake Kumamoto Ezu nursing medical center
17:20 ~ 17:55 Organic acid disorders and fatty acid disorders
Chairperson: Yoshiaki Ohtsu
(Gunma University Graduate School of Medicine, Department of Pediatrics)

P-52 CPT-2 deficiency diagnosed by recurrent rhabdomyolysis from early childhood
○Tae Kimura¹, Eiichi Wake¹, Tomotaka Kono¹, Katsuya Aizu¹, Tomonosuke Someya²
Keiichi Hara³, Go Tajima¹, Mitsuru Kubota⁵, Akira Ohtake⁶, Hiroshi Mochizuki¹
¹Division of Endocrinology and Metabolism, Saitama Children’s Medical Center
²Toyosu Medical Home for Children
³Department of Pediatrics, National Hospital Organization Kure Medical Center and Chugoku Cancer Center
⁴Division of Neonatal Screening, Research Institute, National Center for Child Health and Development
⁵Department of General Pediatrics and Interdisciplinary Medicine, National Center for Child Health and Development
⁶Department of Pediatrics, Saitama Medical University

P-53 An attempt of cooperation of local healthcare to glutaric aciduria type1 complicated by acute subdural hematoma.
○Akiko Yamada¹, Atsushi Iwabuchi¹, Kaori Kiyoki¹, Takahiro Kido¹, Tatsuyuki Ohto¹
Takashi Fukushima¹, Tomohiro Kamoda¹, Ryo Sumazaki¹, Kana Tamai²
Michinobu Jogamoto²
¹Department of Pediatrics, University of Tsukuba Hospital
²Department of Pediatrics, Moriya Daiichi General Hospital

P-54 2 cases of Short-chain acyl-CoA dehydrogenase deficiency discovered by neonatal metabolic screening
○Chunhua Zhang¹, Ning Zhao¹, Dan Yu², Xiaoli Tang³
¹Department of research & development of MILS International
²Department of Pediatrics, West China Second University Hospital, Sichuan University
³Department of Pediatrics, Chengdu Angel Women’s and Children’s Hospital

P-55 Newborn screening for VLCAD deficiency: risk assessment of positive subjects by genetic and enzymatic study
○Keiichi Hara¹, Go Tajima², Reiko Kagawa³, Satoshi Okada³, Nobuo Sakura⁴
¹Department of pediatrics, NHO Kure Medical Center
²Division of neonatal screening, National Center for Child Health and Development
³Department of Pediatrics. Hiroshima University Graduate School of Biomedical & Health Sciences
⁴Nursing Home for Severe Disabilities “Suzugamine”.

P-56 Biochemical change after intravenous carnitine infusion in carnitine-acylcarnitine translocase deficiency
○Yasutsugu Chinen¹, Sadao Nakamura¹, Tadashi Kaname², Koichi Nakanishi¹
¹Department of Pediatrics, Faculty of Medicine, University of the Ryukyus
²Department of Genome Medicine, National Center for Child Health and Development
P-57  A patient with mHMG-CoA synthase deficiency with developmental delay presented with acute decompensation

○Kaori Fukui, Yoriko Watanabe, Kojiro Nagai, Kyoko Tashiro, Tadateru Yasu, Hideo Sasai, Yuki Hasegawa, Toshiyuki Fukao, Takahiro Inokuchi, Yushiro Yamashita

1The Department of Pediatrics and Child Health Kurume University School of Medicine
2Research Institute of Medical Mass Spectrometry, Kurume University School of Medicine
3Department of Pediatrics National Hospital Nagasaki Medical center
4Department of Pediatrics, Graduate School of Medicine, Gifu University
5Department of Pediatrics Shimane University School of Medicine

P-58  A case with short chain acyl-CoA dehydrogenase deficiency who presented high creatine kinase during febrile seizure.

○Miori Yuasa, Ikue Hata, Takuya Kosaka, Yuko Isozaki, Yosuke Shigematsu, Yusei Ohshima, Yoriko Watanabe, Kyoko Tashiro, Keiichi Hara, Go Tajima

1Department of Pediatrics, Faculty of Medical Sciences, University of Fukui
2Department of Pediatrics, Kurume University School of Medicine
3Research Institute of Medical Mass Spectrometry, Kurume University School of Medicine
4Department of Pediatrics, National Hospital Organization Kure Medical Center
5Division of Neonatal Screening, Research Institute, National Center for Child Health and Development

17:55 ~ 18:20  Mitochondrial disorder 1

Chairperson: Taro Yamazaki
(Department of Pediatrics, Saitama Medical University)

P-59  Prenatal diagnosis of mitochondrial respiratory chain disorders caused by nuclear gene mutations

○Nana Akiyama, Kei Murayama, Tarou Yamazaki, Megumi Saitou, Takuya Fushimi, Yukiko Yatsuka, Tomoko Hirata, Masakazu Kohda, Yasushi Okazaki, Akira Ohtake

1Chiba Children’s Hospital, 2Department of Metabolism, Chiba Children’s Hospital
3Department of Pediatrics, Faculty of Medicine, Saitama Medical University
4Department of Clinical Genetics, Saitama Medical University
5Division of Functional Genomics and Systems Medicine, Saitama Medical University
6Division of Translational Research, Research Center for Genomic Medicine, Saitama Medical University
7Research Center for Genomic Medicine, Saitama Medical University
8Intractable Disease Research Center, Juntendo University

P-60  A case suspected of mitochondrial complex III deficiency carrying BCS1L mutations presented with Fanconi syndrome

○Kanako Ishii, Tocan Vlad, Noriko Oyama, Naoko Toda, Kazuhiro Ohkubo, Yoshito Ishizaki, Koh-ichiro Yoshiura, Shouichi Ohga

1Department of Pediatrics, Graduate School of Medical Sciences, Kyushu University
2Department of Human Genetics, Nagasaki University Graduate School of Biomedical Sciences
P-61  Identification of disease genes with no previous association with mitochondrial defects in mitochondrial diseases
○Yoshihito Kishita\textsuperscript{1, 2}, Masakazu Kohda\textsuperscript{2, 3}, Yukiko Yatsuka\textsuperscript{1}, Tomoko Hirata\textsuperscript{3}, Yosuke Mizuno\textsuperscript{1}
Atsuko Imai-Okazaki\textsuperscript{1}, Hiroko Harashima\textsuperscript{4}, Kei Murayama\textsuperscript{5}, Akira Ohtake\textsuperscript{1}, Yasushi Okazaki\textsuperscript{1, 2, 3}
\textsuperscript{1}Division of Functional Genomics & Systems Medicine, Research Center for Genomic Medicine, Saitama Medical University, Saitama.
\textsuperscript{2}Intractable Disease Research Center, Graduate School of Medicine, Juntendo University, Tokyo.
\textsuperscript{3}Division of Translational Research, Research Center for Genomic Medicine, Saitama Medical University, Saitama.
\textsuperscript{4}Department of Pediatrics, Saitama Medical University, Saitama.
\textsuperscript{5}Department of Metabolism, Chiba Children’s Hospital, Chiba.

P-62  Investigation of causative genes for mitochondrial oxidative phosphorylation (OXPHOS) disorders
○Sze Chern Lim\textsuperscript{1}, Yoshihito Kishita\textsuperscript{1, 3}, Masakazu Kohda\textsuperscript{2, 3}, Tomoko Hirata\textsuperscript{2}, Yukiko Yatsuka\textsuperscript{1}
Atsuko Imai-Okazaki\textsuperscript{1}, Hiroko Harashima\textsuperscript{4}, Kei Murayama\textsuperscript{5}, Akira Ohtake\textsuperscript{1}, Yasushi Okazaki\textsuperscript{1, 6}
\textsuperscript{1}Division of Functional Genomics & Systems Medicine, Research Center for Genomic Medicine, Saitama Medical University, Saitama.
\textsuperscript{2}Division of Translational Research, Research Center for Genomic Medicine, Saitama Medical University, Saitama.
\textsuperscript{3}Intractable Disease Research Center, Graduate School of Medicine, Juntendo University, Tokyo.
\textsuperscript{4}Department of Pediatrics, Saitama Medical University, Saitama.
\textsuperscript{5}Department of Metabolism, Chiba Children’s Hospital, Chiba.
\textsuperscript{6}Division of Functional Genomics & Systems Medicine and Division of Translational Research, Research Center for Genomic Medicine, Saitama Medical University, Saitama.

P-63  Three cases of lethal infantile mitochondrial disease.
○Masakazu Honda\textsuperscript{1}, Hayato Sakurai\textsuperscript{1}, Tetsuya Kunikata\textsuperscript{1}, Taro Yamazaki\textsuperscript{1}
Hiroko Harashima\textsuperscript{1}, Yoshihito Kishita\textsuperscript{2}, Masakazu Kohda\textsuperscript{2}, Yasushi Okazaki\textsuperscript{2}
Kei Murayama\textsuperscript{3}, Akira Ohtake\textsuperscript{1}
\textsuperscript{1}Department of Pediatrics, Saitama Medical University
\textsuperscript{2}Research Center for Genomic Medicine, Saitama Medical University
\textsuperscript{3}Department of Metabolism, Chiba Children’s Hospital

18:20 ~ 18:50  Mitochondrial disorder 2
Chairperson: Kondo Hidehito
(Osaka University Graduate School of Medicine, Department of Pediatrics)

P-64  Aspartate supplementation for aspartate-glutamate carrier isoform 1 deficiency
○Shoji Yano\textsuperscript{1}, Arthur Partikian\textsuperscript{1}, Stefan Bluml\textsuperscript{1}, Kathryn Moseley\textsuperscript{1}, Yoriko Watanabe\textsuperscript{2}
Hirotomo Saitsu\textsuperscript{1}, Naomichi Matsumoto\textsuperscript{3}
\textsuperscript{1}Pediatrics, University of Southern California, \textsuperscript{2}Pediatrics, Kurume University
\textsuperscript{3}Human Genetics, Yokohama City University
P-65  Eleven novel mutations in six Chinese patients with thiamine metabolism dysfunction syndrome
○Dongxiao Li¹, Xiyuan Li², Yi Liu², Hui Dong², Jinqing Song², Yupeng Liu³, Yao Zhang²
Ying Jin¹, Hanzhou Guan¹, Yanling Yang²
¹Department of Pediatrics, Peking University First Hospital; Rehabilitation centre, Henan Children’s Hospital.
²Department of Pediatrics, Peking University First Hospital
³Department of Pediatrics, Children’s Hospital of Shanxi Province, Taiyuan, China

P-66  Serum GDF15 levels in diseases similar to mitochondrial disorders
○Shuichi Yatsuga, Miyuki Kitamura, Yasutoshi Koga
Department of Pediatrics and Child Health, Kurume University School of Medicine

P-67  A novel mutation in TAZ causes mitochondrial respiratory chain disorder without cardiomyopathy
○Nurun Nahar Borna¹, Yoshihito Kishita¹ ², Kaori Ishikawa³, Kazuto Nakada³, Masakazu Kohda³ ⁴, Takashi Nasu³, Atsuhito Takeda⁶, Kei Murayama⁷, Akira Ohtake⁸, Yasushi Okazaki² ⁹
¹Division of Functional Genomics & Systems Medicine, Research Center for Genomic Medicine, Saitama Medical University, Saitama.
²Intractable Disease Research Center, Graduate School of Medicine, Juntendo University, Tokyo.
³Faculty of Life and Environmental Sciences, University of Tsukuba, Tsukuba.
⁴Division of Translational Research, Research Center for Genomic Medicine, Saitama Medical University, Saitama.
⁵Department of Pediatrics, Obihiro Kosei General Hospital, Obihiro.
⁶Department of Pediatrics, Hokkaido University Graduate School of Medicine, Hokkaido.
⁷Department of Metabolism, Chiba Children’s Hospital, Chiba.
⁸Department of Pediatrics, Saitama Medical University, Saitama.
⁹Division of Functional Genomics & Systems Medicine and Division of Translational Research, Research Center for Genomic Medicine, Saitama Medical University, Saitama.

P-68  The second case of MELAS with m.5541C>T MT-TW
○Yuko Hirose¹, Hitoshi Osaka¹, Masako Nagashima¹, Hirokazu Yamagishi¹, Sumie Ohhata¹
Jun Odaka¹, Yu-ichi Goto², Takanori Yamagata¹
¹Department of Pediatrics, Jichi Medical University
²Department of Mental Retardation and Birth Defect Research, National

P-69  A 14-year-old Chinese boy with Kearns-Sayer syndrome: a case report
○Yuqing Shi, Fang Fang, Zhimei Liu
Beijing Children’s Hospital, Capital Medical University
P-70  Replacement therapy for selenium and biotin in patients with phenylketonuria
○Yoshiyuki Okano1,2, Miki Okamoto1, Toshiaki Watanabe3, Ryoko Watanabe3, Rika Fujii3
Yosuke Shigematsu4, Tomoko Tamaoki2, Yasuhiro Takeshima2
1Okano Children’s Clinic, 2Hyogo College of Medicine
3Osaka Aoyama University, 4Fukui University

P-71  Successful pregnancy in a PKU woman treated with tetrahydrobiopterin
○Natsuko Arai-Ichinoi1, Osamu Sakamoto1, Shigeo Kure1, Kei Murayama2
1Department of Pediatrics, Tohoku University School of Medicine
2Department of Metabolism, Chiba Children’s Hospital

P-72  Extended tetrahydrobiopterin loading test in responsive phenylketonuria with sapropterin
○Chika Takano, Mika Ishige, Erika Ogawa, Tatsuo Fuchigami, Shori Takahashi
Department of Pediatrics and Child Health, Nihon University School of Medicine

P-73  BH4-responsive hyperphenylalaninemia whose initial phenylalanine level was equivalent to classical phenylketonuria
○Tomotaka Kono1, Tae Kimura1, Eiichi Wake1, Katsuya Aizu1, Akira Ohtake2
Hiroshi Mochizuki1
1Division of Endocrinology and Metabolism, Saitama Children’s Medical Center
2Department of Pediatrics, Saitama Medical University

P-74  Phase 3 Long-term Study Evaluating Efficacy and Safety of Pegvaliase Treatment in Adults with PKU
○Nicola Longo1, Jeffery Vockley2, Harvey Levy3, Stephen Amato4, Roberto Zori5
Janet Thomas6, Barbara Burton7, Cary Harding8, John Posner9, Deborah Bilder1
Joy Olbertz10, Zonghua Gu10, Kelly Lau10, Mingjin Lin10, Kevin Larimore10, David Dimmock11
1University of Utah, Salt Lake City, UT
2University of Pittsburgh and Children’s Hospital of Pittsburgh, Pittsburgh, PA
3Boston Children’s Hospital, Boston, MA, 4University of Kentucky, Lexington, KY
5University of Florida, Gainesville, FL, 6University of Colorado School of Medicine, Aurora, CO
7Lurie Children’s Hospital of Chicago, Chicago, IL
8Oregon Health & Science University, Portland, OR, 9King’s College, London, UK
10BioMarin Pharmaceutical Inc., Novato, CA
11Rady Children’s Institute for Genomic Medicine, San Diego, CA
P-75  Phase 3 PRISM Studies Evaluating Efficacy and Safety of Pegvaliase Treatment in Adults with PKU
Nicola Longo1, Cary Harding2, Stephen Amato3, Jeffery Vockley4, Klaas Wierenga5
Hong Li6, Deborah Bilder1, Barbara Burton7, David Dimmock8, John Posner9
Janet Thomas10, Roberto Zorli11, Orli Rosen12, Zonghua Gu12, Mingjin Lin12
Markus Merilainen12, Haoling H. Weng12, Harvey Levy13
1University of Utah, Salt Lake City, UT, 2Oregon Health & Science University, Portland, OR
3University of Kentucky, Lexington, KY
4University of Pittsburgh and Children’s Hospital of Pittsburgh, Pittsburgh, PA
5University of Oklahoma, Oklahoma City, OK, 6Emory University, Decatur, GA
7Lurie Children’s Hospital of Chicago, Chicago, IL
8Rady Children’s Institute for Genomic Medicine, San Diego, CA
9King’s College, London, UK, 10University of Colorado School of Medicine, Aurora, CO
11University of Florida, Gainesville, FL, 12BioMarin Pharmaceutical Inc., Novato, CA
13Boston Children’s Hospital, Boston, MA

17:50 ~ 18:20  Amino acid disorders 2
Chairperson: Erika Ogawa
(Department of Pediatrics and Child Health, Nihon University)

P-76  A retrospective study of orotic acid value in patient with OTC deficiency
Hironori Kobayashi1, Toshikazu Minohata1, Hiroshi Mitsubuchi2, Kimitoshi Nakamura2
Mitsuru Kubota3, Osamu Sakamoto4, Yoriko Watanabe5, Kaori Fukui6, Furujo Mahoko6
Seiji Yamaguchi1
1Department of Pediatrics, Shimane University Faculty of Medicine
2Department of Pediatrics, Kumamoto University Graduate School of Medical Sciences
3Department of General Pediatrics & Interdisciplinary Medicine, National Center for Child Health and Development
4Department of Pediatrics, Tohoku University School of Medicine
5Department of pediatrics, Kurume University School of Medicine
6Department of pediatrics, Okayama Mediacal Center

P-77  Hypocitrullinemia in newborn MS/MS screening with late-onset OTC deficiency
Tomoko Lee1, Katsuhiko Yoshii2, Satoru Yoshida3, Takenori Suga4, Kimitoshi Nakamura5
Toshiyuki Fukao5, Kei Murayama1, Yuki Hasegawa8, Yasuhiro Takeshima1
1Department of Pediatrics, Hyogo College of Medicine
2Department of pediatrics, Chibune General Hospital
3Department of pediatrics, Seirei Mikatahara General Hospital
4Department of pediatric emergency medicine, Hyogo Prefectural Amagasaki general Medical Center
5Department of Pediatrics, Kumamoto University Graduate School of Medicine
6Department of Pediatrics, Gifu University Graduate School of Medicine
7Department of Metabolism, Chiba Children’s Hospital
8Department of Pediatrics, Shimane University Graduate School of Medicine
P-78  A case of late-onset ornithine transcarbamylase deficiency with reversible CT hepatic images
○Shinpei Kawachi, Takenori Suga
Department of pediatric emergency and intensive care unit, Hyogo Prefectural Amagasaki General Medical Center

P-79  Impact of the 2016 Kumamoto Earthquake on a female patient with OTCD
○Jun Kido¹, Takanobu Yoshida¹, Shirou Matsumoto¹, Hiroshi Mitsubuchi¹, Rieko Sakamoto¹
Fumio Endo², Kimitoshi Nakamura¹
¹Department of Pediatrics, Kumamoto University
²Department of Pediatrics, Kumamoto eduko developmental care center

P-80  Sodium pyruvate treatment for persistent NICCD
○Toju Tanaka, Hiroko Shigetomi, Masayoshi Nagao
Center for Genetics & Metabolic Disorder and Clinical Research, National Hospital Organization Hokkaido Medical Center

P-81  A case of cystinuria occurring with acute renal parenchymal injury due to ureteral stone.
○Ikuma Musha, Hiromi Teranishi, Yutaka Ueda, Taro Yamazaki, Yuko Akioka
Hideo Yamanouchi, Kenichi Tokuyama, Toru Kikuchi, Akira Ohita
Department of Pediatrics, Saitama Medical University

18:20 ~ 18:45  Amino acid disorders 3
Chairperson: Atsuo Kikuchi
(Department of Pediatrics, Tohoku University Hospital)

P-82  Medical care subsidies of intractable diseases in adults with PKU, MSUD and HCU.
○Mika Ishige, Erika Ogawa, Chika Takano, Tatsuo Fuchigami, Shori Takahashi
Department of Pediatrics and Child Health, Nihon University School of Medicine

P-83  Quantification of allo-isoleucine and branched-chain amino acid to make a diagnosis and a follow-up of MSUD
○Reiko Iwano¹, Hideki Nakajima², Junko Hanakawa³, Yumi Asakura³, Masanori Adachi³
Koji Muroya³
¹Clinical Research Institute, Kanagawa Children’s Medical Center
²Division of Neonatal Screening, National Center for Child Health and Development
³Department of Endocrinology and Metabolism, Kanagawa Children’s Medical Center

P-84  A Case of Congenital Biliary Atresia with Difficulties in Differentiation from Congenital Metabolic Disorders
○Shin Hoshino¹, Sumire Kumal¹, Toru Maeda¹, Takenori Adachi¹, Takashi Kawabe¹
Katsuyuki Yokoi², Yoko Nakajima², Tetsuya Ito², Toshihiro Yasui², Tatsuya Suzuki²
¹Department of Pediatrics, Kasugai Municipal Hospital
²Department of Pediatrics, Fujita Health University
P-85  Cobalamin disorder CblC presenting acute pulmonary edema detected by expanded newborn screening
○Shoko Sakamoto¹, Osamu Sakamoto², Takashi Hamazaki³, Haruo Shintaku³
¹Department Pediatrics, Osaka City University, ²Department of Pediatrics, Tohoku University
³Department of Pediatrics, Osaka City University Graduate School of Medicine

P-86  Mutation in SLC6A9 encoding a glycine transporter causes a novel form of non-ketotic hyperglycinemia in humans
○MOHAMMAD ARIF HOSSAIN, Marwan Nashabat, Fuad Al Mutairi, Majid Alfadhel
King Abdulaziz Medical City, National Guard Health Affairs

< Block6 >  17:20 ~ 17:45  Metal merabolic abnormality & Carbohydrate metabolism
Chairperson: Tokiko Fukuda
(Pediatrics, Hamamatsu University School of Medicine)

P-87  A case of hepatic-type of Wilson disease with long turn good medical compliance, who died due to subarachnoid hemorrhage
○Miku Tsuruoka¹, Yutaka Fuchinoue², Takao Kuroki², Hiroyuki Hiruta², Akihiko Tateno²
Norikazu Shimizu², Tsugutoshi Aoki²
¹Department of Pediatrics, Toho University Ohashi Medical Center
²Toho University

P-88  An example of liver neural-type Wilson's disease that presented atypical findings
○Shinji Utsunomiya, Miku Turuoka, Syoko Nakazawa, Norikazu Shimizu, Tsugutoshi Aoki
Department of pediatrics, Toho University

P-89  The repetitive observation of urine glycerol level is useful for diagnosis of FBPase deficiency.
○Yoshimitsu Osawa¹, Aya Wada¹, Yoko Tabei¹, Yoshiaki Ohtsu¹, Junko Igaki²
Kenichi Maruyama³, Hirokazu Arakawa¹
¹Gunma University Graduate School of Medicine, Department of Pediatrics, Gunma, Japan
²Department of Endocrinology and Metabolism, Nanbu Medical Center・Nanbu Child Medical Center, Okinawa Japan
³Department of Nephrology, Gunma Children’s Medical Center, Gunma Japan

P-90  A Korean girl with neurologic features diagnosed as Fructose-1,6-bisphosphatase (FBPase) deficiency using clinical exome sequencing
○Hyunjoo Lee, JIN-SUNG Lee, Ah Reum Kwon, Ho-Seoung Kim
Department of Pediatrics, Yonsei University College of Medicine
P-91  Epidemiology of insulin resistance syndromes in Japanese children

○Tohru Yorifuji¹, Hiroko Kadowaki², Yushi Hirota³, Wataru Ogawa⁴, Hideki Katagiri⁴
Yasuhi Ishigaki⁵, Takashi Akamizu⁶
¹Division of Pediatric Endocrinology and Metabolism, Children’s Medical Center, Osaka City General Hospital,
²Department of Pediatrics, Sanno Hospital
³Division of Diabetes and Endocrinology, Department of Internal Medicine, Kobe University Graduate School of Medicine
⁴Division of Diabetes and Metabolism, Department of Internal Medicine, Tohoku University School of Medicine
⁵Division of Diabetology, Department of Internal Medicine, Iwate Medical University
⁶First Department of Internal Medicine, Wakayama Medical University

17:45 ~ 18:20  Peroxisomal disorder & others
Chairperson: Hideo Sasai
(Department of Pediatrics, Graduate School of Medicine, Gifu University)

P-92  Establishment and phenotypic analysis of the disease-model zebrafish for Peroxisomal biogenesis disorder

○Shigeo Takashima¹, Shoko Takemoto¹, Kayoko Toyoshi¹, Akiko Ohba¹, Haruka Fujita²
Kentaro Oh-hashi², Yoko Morita², Nobuyuki Shimozawa¹
¹Division of Genomics Research, Life Science Research Center, Gifu University
²Graduate School of Natural Science and Technology, Gifu University

P-93  Screening for chemical compounds that stimulate peroxisomal fatty acid β-oxidation

○Masashi Morita¹, Kaito Tomita², Sato Hideaki², Shun Matumoto², Shiro Watanabe³
Nobuyuki Shimozawa⁴, Tsuneo Imanaka⁵
¹Department of Biological Chemistry, Graduate School of Medicine and Pharmaceutical Sciences, University of Toyama
²Department of Biological Chemistry, Graduate School of Medicine and Pharmaceutical Sciences, University of Toyama
³Institute of Natural medicine, University of Toyama
⁴Division of Genomic Research, Life Science Research Center, Gifu University
⁵Faculty of Pharmaceutical Sciences, Hiroshima International University

P-94  Two cases of long term survival of Adrenoleukodystrophy after hematopoietic stem cell transplant

○Toshiki Tsunogai¹, Masayoshi Yamaoka¹, Masaharu Akiyama¹, Hiroyuki Ida¹, Toya Ohashi²
¹Department of Pediatrics, The Jikei University School of Medicine
²Department of Gene Therapy, Institute of DNA Medicine, The jikei University School of Medicine
P-95  Ten novel mutations of the ERCC6, ERCC8 genes associated with Cockayne syndrome and prenatal diagnosis for three fetuses
○Dongxiao Li1, Yuan Ding2, Hui Dong2, Jinqing Song2, Xiyuan Li2, Ying Jin2, Yupeng Liu2
Lifang Feng3, Xiaohong Chen4, Hong Zheng4
1Department of Pediatrics, Peking University First Hospital; Rehabilitation centre, Henan Children’s Hospital.
2Department of Pediatrics, Peking University First Hospital
3Department of Endocrinology and Metabolism, Wuhan Medical and Healthcare Center for Women and Children, China
4Department of Pediatrics, The First Hospital of Henan University of Traditional Chinese Medicine, China.

P-96  A de novo novel mutation in COL2A1 leading to spondyloepiphyseal dysplasia congenital in a Chinese family
○Yi Liu1, Qiuhong Xiong2, Xiyuan Li1, Yu Xue2, Jing Wang2, Dongxiao Li1, Ying Jin1
Changxin Wu2, Han Xiao3, Yanling Yang1
1Department of Pediatrics, Peking University First Hospital
2Institute of Biomedical Sciences, Shanxi University

P-97  A case with severe acidosis and unconsciousness suspected of inborn errors in keton body metabolism
○Miki Matsui1, Satoshi Oonishi2, Tomoko Lee3, Yo Okizuka2, Taisuke Hashimoto1
Hironori Kobayashi5
Yuki Hasegawa5, Hideo Sasai6, Toshiyuki Fukao6, Hirotaka Minami1
1Department of pediatrics, Takatsuki General Hospital
2Department of pediatric intensive care, Takatsuki General Hospital
3Department of pediatrics, Hyogo College Of Medicine
4Department of pediatrics, Toyonaka Municipal Hospital
5Department of Pediatrics, Shimane University School of Medicine
6Department of Pediatrics, Gifu University Graduate School of Medicine

P-98  A case of dihydropyrimidinase deficiency accidentally discovered by urinary metabolome analysis
○Hiroki Tsuchiya1, Tomoyuki Akiyama1, Tomiko Kuhara2, Yoko Nakashima3
Takashi Shibata1, Mari Akiyama1, Makio Oka1, Fumika Endo1, Harumi Yoshinaga4
Katsuhiro Kobayashi1
1Department of Child Neurology, Okayama University Graduate School of Medicine, Dentistry and Pharmaceutical Sciences
2Japan Clinical Metabolomics Institute
3Department of Pediatrics, Fujita Health University
4National Hospital Organization Minami-Okayama Medical Center
18:20 ~ 18:40  Patient Registration & Treatments
Chairperson: Kohnosuke Mitani
(Saitama Medical University Research Center for Genomic Medicine, Division of Gene Therapy and Genome Editing)

P-99  Epidemiology of Mucopolysaccharidoses
○Shunji Tomatsu¹,², Shaukat A. Khan³, Hira Peracha³, Diana Ballhausen³, Alfred Wiesbauer⁴
Yasuyuki Suzuki³, Kenji E. Orii⁶, Tadao Orii⁶
¹Departments of Biomedical Research and Orthopedics
²Nemours/Alfred I. duPont Hospital for Children
³Centre for molecular diseases, Service for genetic medicine, University Hospital
⁴Institute of Social and Preventive Medicine, University of Bern
⁵Medical Education Development Center, Gifu University
⁶Department of Pediatrics, Gifu University

P-100  Current status of Japan Registration System for Metabolic & Inherited Diseases
○Joo-Hyun Seo¹, Makiko Miyairi¹, Akira Ohtake², Torayuki Okuyama¹
¹Clinical Laboratory Medicine, National Center for Child Health and Development
²Department of Pediatrics, Saitama Medical University

P-101  Development of Bone Targeting Drugs
○Shunji Tomatsu¹, Molly Stapleton¹, Kazuki Sawamoto¹, Carlos J. Alméciga-Díaz², Tadao Orii³
¹Nemours/Alfred I. duPont Hospital for Children
²Institute for the Study of Inborn Errors of Metabolism, Pontificia Universidad Javeriana
³Department of Pediatrics, Graduate School of Medicine, Gifu University

P-102  Therapeutic strategy for Fabry disease peripheral neuropathy with rAAV vector
○Takashi Higuchi¹, Yohta Shimada¹, Hiroshi Kobayashi¹, Takahiro Fukuda², Fusao Kato³
Toya Ohashi¹
¹Division of Gene Therapy, The Jikei University
²Department of Neuropathology, The Jikei University
³Division of Neuroscience, The Jikei University