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 Medicin)

O-1　Development of drug screening system using iPS cells derived from a patient with Gaucher disease type II.

○Jun Kido[1], Tadahiro Numakawa[2], Shirou Matsumoto[1], Haruki Odaka[2], Minami Soga[2]
Ryutarou Kajihara[2], Fumio Endo[1], Kimitoshi Nakamura[1], Takumi Era[2]
[1]Department of Pediatrics, Kumamoto University
[2]Cell 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 Liao[1], Hsiao-Jan Chen[1], Chuan-Chi Chiang[1], Michael H. Gelb[2]
[1]Chinese Foundation of Health, newborn screening center
[2]University of Washington, Department of Chemistry

O-5　Newborn screening for Mucopolysaccharidosis type II in Taiwan: A One-and-a-half year Report.

○Hsiao-Jan Chen[1], Min-Ju Chan[1], Mei-Ying Liu[1], Shuan-Pei Lin[2], Arun Babu Kumar[3]
Michael H. Gelb[3], Chuan-Chi Chiang[1]
[1]Neonatal Screening Center, The Chinese Foundation of Health
[2]MacKay Memorial Hospital, Taipei, Taiwan
[3]Department 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 Soga[1], Jun Kido[2], Takumi Era[1]

[1]Department of Cell Modulation, IMEG, Kumamoto University

[2]Department 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 Maekawa[1], Isamu Jinnou[2], Aya Narita[3], Hiroaki Yamaguchi[1], Mano Nariyasu[1]

[1]Department of Pharmaceutical Sciences, Tohoku University Hospital

[2]Department of Pharmaceutical Sciences, Tohoku University

[3]Department of Child Neurology, Tottori University, Faculty of Medicine

O-8　Hematopoietic Stem Cell Transplantation for Patients with Mucopolysaccharidosis II

○Shunji Tomatsu[1, 2], Francyne Kubaski[1], Hiromasa Yabe[3], Yasuyuki Suzuki[4], Toshiyuki Seto[5]
Takashi Hamazaki[5], Seiji Yamaguchi[6], Kenji E. Orii[7], Tadao Orii[7]

[1]Nemours/Alfred I. duPont Hospital for Children

[2]Departments of Biomedical Research and Orthopedics

[3]Department of Cell Transplantation and Regenerative Medicine, Tokai University School of Medicine

[4]Medical Education Development Center, Gifu University Medical Education Development Center, Gifu University

[5]Department of Pediatrics, Osaka City University Graduate School of Medicine

[6]Department of Pediatrics, Shimane University

[7]Department of Pediatrics, Graduate School of Medicine, Gifu University

O-9　Analysis of safety of pentosan polysulphate in young patients with mucopolysaccharidosis type VI

○Mahoko Furujo[1], Kenji Orii[2], Shunji Tomatsu[3], Yasuyuki Suzuki[4], Toshiyuki Fukao[5]

[1]Department of Pediatrics, NHO Okayama Medical Center

[2]Division of Neonatal intensive Care Unit, Gifu University Hospital

[3]Nemours/Alfred I, duPont Hospital for Children

[4]Medical education development Center, Gifu University

[5]Department 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 Asakura[1], Yasuyuki Fukuhara[1], Motomichi Kosuga[1], Torayuki Okuyama[2]

[1]Division of Medical Genetics, National Center for Child Health and Development

[2]Department 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 Akahira[1], Yukiko Shimizu[2], Yoshiaki Okuma[3], Tadashi Okamura[2], Toshihiro Ohura[4]

[1]Division of Medical Genetics, Kanagawa Children's Medical Center

[2]Department of Laboratory Animal Medicine, Research Institute, National Center for Global Health and Medicine

[3]Department of Pediatrics, National Center for Global Health and Medicine

[4]Division of Pediatrics, Sendai City Hospital

O-12　Therapeutic potency of teprenone in comprehensive lysosomal storage diseases

○Yohta Shimada[1], Natsumi Ishii[1], Hiroo Hoshina[2], Takashi Higuchi[1], Hiroshi Kobayashi[1]
Hiroyuki Ida[2], Toya Ohashi[1]

[1]Div. of Gene Therapy, Res. Cent. for Med. Sci. Jikei Univ. Sch. of Med.

[2]Dept. of Pediatrics, Jikei Univ. Sch. of Med.

O-13　Mucopolysaccharidosis-plus syndrome: A report of new type of MPS

○Hidehito Kondo[1], Takanobu Otomo[2], Nadezda Maksimova[3], Norio Sakai[1], Keiichi Ozono[1]

[1]Department of Pediatrics, Osaka University Graduate School of Medicine

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

[3]Laboratory 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 Lyosomal 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 Lyosomal 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 Nwborn 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 Endo[1], Shinichiro Yoshida[2], Kousuke Kumeda[2], Kimitoshi Nakamura[3]
[1]Kumamoto Ezuko Ryoiku Iryo Center
[2]Newborn screening center, The Chemo-Sero-Therapeutic Research Institute
[3]Dep 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 LEE[1], Hyunjoo Lee[1], Cheol-Ho Lee[1], Yangrae Cho[2], Jongsun Jung[2], Yoon Hee Ko[1]
[1]Department of Pediatrics, Yonsei University College of Medicine
[2]Syntekabio, Inc., Seoul, KOREA

O-17　Mass screening for VLCAD deficiency in Tokyo; Results and sttudy for the future

○Nobuyuki Ishige[1], Kazuhiro Watanabe[1], Satomi Hasegawa[1], Kaoru Konishi[1]
Michiko Mashita[1], Sera Yasumi[1], Mika Ishige[2], Misao Owada[1]
[1]Division of Newborn Screening, Tokyo Health Service Association
[2]Department 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 Arao[1], Taro Yamazaki[1], Hiroko Harashima[1], Aya Takada[2], Kazuyuki Saito[2]
Yosuke Shigematsu[3], Keiichi Hara[4], Go Tajima[5], Akira Ohtake[1]
[1]Department of Pediatrics, Saitama Medical University Hospital
[2]Department of Forensic Medicine, Saitama Medical University
[3]Department of Health Science, Faculty of Medical Sciences, University of Fukui
[4]Department of Pediatrics, National Hospital Organization Kure Medical Center
[5]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 Numakura[1], Shion Hayashi[2], Osamu Sakamoto[3], Kiyoshi Hayasaka[1, 4]

[1]Department of Pediatrics, Yamagata University Faculty of Medicine

[2]Department of Ophthalmology, Yamagata University Faculty of Medicine

[3]Department of Pediatrics, Tohoku University School of Medicine

[4]Department of Pediatrics, Miyukikai Hospital

O-20　A Case of Combined malonic and methylmalonic aciduria

○Daiki Kondo[1], Wakako Kikuchi[1], Atsuko Noguchi[1], Ikuko takahashi[1], Tsutomu Takahashi[1]
Tomoko Uehara[2], Kenjiro Kosaki[2]

[1]Department of Pediatrics, Akita University School of Medicine

[2]Center for Medical Genetics, Keio University School of Medicine

❀ O-21　Phenotypes and genotypes of 52 Chinese patients with propionic acidemia

○Yi Liu[1], Yuan Ding[1], Dongxiao Li[1], Xiyuan Li[1], Ying Jin[1], Jinqing Song[1], Yao Zhang[1]
Yanling Yang[1], Haixia Li[2], Lili Jiao[2]

[1]Department of Pediatrics, Peking University First Hospital

[2]Department 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

10：48 ～ 11：36　Oral6：Organic acid disorders and fatty acid disorders 2

Chairperson: Hironori Kobayashi
(Department of Pediatrics, Shimane University Faculty of Medicine)

Shigenori Yamamoto
(Shimoshizu National Hospital)

O-23　Insufficient ketogenesis in 3-hydroxybutyrate dehydrogenase (Bdh1) KO mice in fasting test

○Hiroki Otsuka[1], Takeshi Kimura[1], Yasuhiko Ago[1], Mina Nakama[2], Abdelkreem Elsayed[1]
Yuka Aoyama[3], Hideo Sasai[1], Hidenori Ohnishi[1], Masatake Osawa[4], Yusuke Kawashima[5]
Osamu Ohara[5], Seiji Yamaguchi[6], Toshiyuki Fukao[1]

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

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

○Kenji Yamada[1], Hideaki Shiraishi[2], Naoko Asahina[2], Saki Yokoshiki[3], Takashi Miyakoshi[3]
Kota Ohno[3], Toshiyuki Isoe[3], Hiroshi Hayashi[3], Seiji Yamaguchi[1], Norihiro Sato[3]

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

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

○Hideo Sasai[1], Hidenori Ohnishi[1], Shohei Akagawa[2], Kazuhisa Akiba[3], Yukihiro Hasegawa[3]
Masahisa Kobayashi[4], Hiroki Otsuka[1], Yuka Aoyama[1], Toshiyuki Fukao[1]

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

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

○Kenji Yamada[1], Hironori Kobayashi[1], Yuki Hasegawa[1], Akio Takahashi[2], Haruo Shintaku[3]
Hideo Sasai[4], Toshiyuki Fukao[4], Seiji Yamaguchi[1], Takeshi Taketani[1]

[1]Department of Pediatrics, Shimane University Faculty of Medicine
[2]Department of Pediatrics, Morioka Children's Hospital
[3]Department of Pediatrics, Osaka City University Faculty of Medicine
[4]Department 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 Abdenur, 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 stategy 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 Yépez[1], Laura Kremer[2], Daniel Bader[1], Mirjana Gusic[2], Holger Prokisch[2], Julien Gagneur[1]
[1]Technical University Munich, [2]Helmholtz Zentrum München

12：40 ～ 13：40　Luncheon Seminar 2

Chairperson: Torayuki Okuyama
(Department of Clinical Laboratory Medicine,
Center for Lyosomal 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[5], Jeff Castelli[5], Christopher Viereck[5]

[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 functin 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 Katsuta[1], Kazuya Tsuboi[2], Hiroshi Yamamoto[2], Hiromi Goto[2]

[1]Junior Resident, Nagoya Central Hospital

[2]LSD 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 Kuranobu[1], Aya Narita[2], Masaru Shimura[1], Makiko Tajika [1], Keiko Ichimoto[1]

Ayako Matsunaga[1], Tomoko Tsuruoka[1], Masaki Takayanagi [1], Akira Ohtake[3], Kei Murayama[1]

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

[2]Department of Child Neurology, Tottori University

[3]Department 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 Sakurai[1], Junji Azuma[1], Yusuke Hamada[2], Eri Kizima[1], Koji Tominaga[3]

Yoshimi Mizoguchi[1], Tunesuke Shimotuji[1], Takehisa Yamamoto[1], Norio Sakai[3]

[1]Department of Pediatrics, Minoh City Hospital

[2]Department of Pediatrics, Japan Community health care organization Osaka hospital

[3]Department 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 Miyana[1], Hidehito Kondo[1], Keiichi Ozono[1], Norio Sakai[2]

[1]Division of Pediatrics, Osaka University Graduate School of Medicine

[2]Division of Health Science, Osaka University Graduate School of Medicine

P-13　Clinical and molecular analysis of six novel GALC 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 Odaka[1], Tadahiro Numakawa[2], Minami Soga[2], Jun Kido[3], Ryutaro Kajihara[4] Toshika Okumiya[4], Hirokazu Furuya[5], Takafumi Inoue[1], Takumi Era[2]

[1]Department of Life Science and Medical Bioscience, School of Advanced Science and Engineering, Waseda University

[2]Department of Cell Modulation, Institute of Molecular Embryology and Genetics, Kumamoto University

[3]Department of Pediatrics, Graduate School of Medical Sciences, Kumamoto University

[4]Department of Biomedical Laboratory Sciences, Faculty of Life Science, Kumamoto University

[5]Department of Neurology, Kochi Medical School, Kochi University

P-16　A summary of the lysosomal enzyme assay for patients with leukodystrophy in our facility

○Sachiko Nakaoka[1], Toko Shibuya[1], Kaori Miyana[1], Hidehito Kondo[1], Norio Sakai[2] Keiichi Ozono[1]

[1]Department of Pediatrics, Osaka University Graduate School of Medicine

[2]Division of health science, Osaka University Graduate School of Medicine

P-17　Newborn screening for pompe disease in northern Kawasaki city

○Ohsuke MIGITA[1], Kojima Takahiro[2], Yusuke Miyaji[2], Yusaku Miyamoto[2], Isamu Hokuto[1] Motomichi Kosuga[3], Torayuki Okuyama[3], Hitoshi Yamamoto[1]

[1]Department of Pediatrics, St. Marianna University School of Medicine

[2]Division of Pediatrics, Kawasaki Municipal Tama Hospital, St. Marianna University School of Medicine.

[3]Center for Lysosomal Storage Diseases, National Center for Child Health and Development

＜ Block2 ＞　17：20 ～ 17：50　Lysosome 4

Chairperson: Yusuke Hamada

(Japan community health care organization Osaka hospital)

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 [1], Kimitoshi Nakamura[2]

[1]Division of Pediatrics, Nishibeppu National Hospital

[2]Department of Pediatrics, Kumamoto University Hospital

P-20　Case report: 10-year enzyme replacement therapy in Japanese siblings with mucopolysaccharidosis VI

○Mahoko Furujo[1], Motomichi Kosuga [2], Torayuki Okuyama[2]

[1]Department of Pediatrics, National Okayama Medical Center

[2]Center 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[1], Takayuki Yokoi[1], Shimada Yohta[2], Higuchi Takashi[2], Hiroshi Kobayashi[2] Toya Ohashi[2], 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

P-23　Clinical features of mucopolysaccharidosis type VII "Case series in Japan"

○Saki Kasuga[1], Kadono Chiho[1], Satoshi Kudoh[2], Hidetomi Terai[3], Seto Toshiyuki[1] Takashi Hamazaki[1], Haruo Shintaku[1]

[1]Department of Pediatrics, Osaka City University Graduate School of Medicine, Osaka City University

[2]ATOX Co.,Ltd

[3]Department 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 Tomatsu[1], Francyne Kubaski[1], Yasuyuki Suzuki[2], Kenji Orii[3], Robert W. Mason[1]
Vũ Chí Dũng[4], Seiji Yamaguchi[5], Hironori Kobayashi[5], Toshiyuki Fukao[3], Tadao Orii[3]

[1]Nemours/Alfred I. duPont Hospital for Children

[2]Medical Education Development Center, Gifu University

[3]Department of Pediatrics, Graduate School of Medicine, Gifu University

[4]Vietnam National Children's Hospital, Department of Medical Genetics, Metabolism & Endocrinology

[5]Department of Pediatrics, Shimane University

P-25　LC/MS/MS measurement of glycosaminoglycans in amniotic fluid of a MPS VII fetus

○Shunji Tomatsu[1], Francyne Kubaski[1], Robert W. Mason[1], Roberto Giugliani[2]

[1]Nemours/Alfred I. duPont Hospital for Children, [2]Medical Genetics Service, HCPA

P-26　Method for measuring mannose 6-phosphate residue contents of enzyme preparations in Lysosomal diseas

○Minori Kanzaki [1], Masashi Shigenaga[1], Yuya Tayama[1], Takahiro Tsukimura[1]
Tadayasu Togawa[1], Hitoshi Sakuraba[2]

[1]Department of Functional Bioanalysis, Meiji Pharmaceutical University

[2]Department 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 Wu[1], Takeo Iwamoto[2], Hiroko Yanagisawa[1], Keiko Akiyama[1], Takashi Miyajima[1]
Junko Igarashi[3], Mohammad Arif Hossain[1], Yoshikatsu Eto[1]

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[3]AnGes, Inc.

P-29 Glycosaminoglycan Assay as First-tier for MPS Newborn Screening

○Shunji Tomatsu[1], Francyne Kubaski[1], Akiko Nakatomi[2], Haruo Shintaku[3]
Hironori Kobayashi[4], Seiji Yamaguchi[4], Yasuyuki Suzuki[5], Tadao Orii[6], Toshiyuki Fukao[6]
Adriana M. Montaño[7]

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[2]Department of Pediatrics, Nagasaki University
[3]Department of Pediatrics, Osaka City University Graduate School of Medicine
[4]Department of Pediatrics, Shimane University
[5]Medical Education Development Center, Gifu University
[6]Department of Pediatrics, Gifu University, [7]Department 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 Momosaki[1], Shinichiro Yoshida[2], Kousuke Kumeda[2], Fumio Endo[3]
Kimitoshi Nakamura[1]

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

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

○Takashi Miyajima[1], Jyunko Igarashi[2], Chen Wu[3], Hiroko Yanagisawa[1]
Mohammad Arif Hossain[1], Keiko Akiyama[1], Takashi Hamazaki[4], Takeo Iwamoto[5]
Yoshikatsu Eto[1]

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

○Rina Itagaki[1], Keiko Yaginuma[1], Masahiro Endo[1], Keiko Akiyama[1], Hiroko Yanagisawa[1]
Takeo Iwamoto[2], Haruo Shintaku[3], Yoshikatsu Eto[1, 4]

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[3]Department of Pediatrics Osaka City University Graduate School of Medicine
[4]The Jikei University School of Medicine

P-33 Factors to influence Plasma LysoGb3 Levels in Fabry Disease- Age, Sex and Antibody against ERT

○Chen Wu[1], Takeo Iwamoto[2], Mohammad Arif Hossain[1], Takashi Miyajima[1]
Keiko Akiyama[1], Hiroko Yanagisawa[1], Junko Igarashi[3], Yoshikatsu Eto[1]
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[3]AnGes, 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 Sasai[1], Fujiki Ryoji[2], Ohara Osamu[2], Nakajima Yoko[3], Testuya Ito[3]
Masahisa Kobayashi[4], Go Tajima[5], Osamu Sakamoto[6], Matsumoto Shiro[7]
Kimitoshi Nakamura[7], Takashi Hamazaki[8], Hironori Kobayashi[9], Yuki Hasegawa[9]
Toshiyuki Fukao[1]
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[2]Department of Technology Development, Kazusa DNA Research Institute
[3]Department of Pediatrics, School of Medicine, Fujita Health University
[4]Department of Pediatrics, The Jikei University School of Medicine
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[6]Department of Pediatrics, Tohoku University School of Medicine
[7]Department of Pediatrics, Kumamoto University Graduate School of Medical Sciences
[8]Department of Pediatrics, Osaka City Graduate School of Medicine
[9]Department 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 Nagamatsu[1], Akira Ohtake[2], Nobuyuki Ishige[3], Hironori Kobayashi[4], Toshiyuki Fukao[5]
Yukihiro Hasegawa[1]
[1]Department of Endocrinology and Metabolism, Tokyo Metropolitan Children's Medical Center
[2]Department of Pediatrics, Saitama Medical University
[3]Tokyo Health Service Association
[4]Department of Pediatrics, Shimane University Faculty of Medicine
[5]Department 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 Nakajima[1], Katsuyuki Yokoi[1], Yoshimi Sakai[2], Tetsushi Yoshikawa[1], Tetsuya Ito[1]

[1]Department of Pediatrics, Fujita Health University

[2]Aichi Health Promotion Public Interest Foundation

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

○Yuki Omura-Hasegawa[1], Hideo Sasai[2], Osamu Sakamoto[3], Hironori Kobayashi[1]
Hiroki Otsuka[2], Ryoji Fujiki[4], Osamu Ohara[4], Toshiyuki Fukao[2]

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[2]Department of Pediatrics, Graduate School of Medicine, Gifu University

[3]Department of Pediatrics, Tohoku University School of Medicine

[4]Department of Technology Development, Kazusa DNA Research Institute

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

○Somporn Liammongkolkul[1], Boonchai Boonyawat[2], Kasinat Sanomcham[1]
Achara Sathienkijkanchai[1], Pornswan Wasant[1], Nithiwat Vatanavicharn[1]

[1]Division of Medical Genetics, Department of Pediatrics, Faculty of Medicine Siriraj Hospital, Mahidol University

[2]Division 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 Shibata[1], Yoriko Watanabe[1, 2], Kaori Fukui[2], Hiromi Isii[1], Misa Inaba[1]
Kyoko Tashiro[1], Kumiko Aoki[1], Yoko Nakajima[3], Takahiro Inokuchi[1], Naohisa Uchimura[1, 4]

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[4]Department 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 Hamada[1], Toko Shibuya[2], Hidehito Kondo[2], Kanako Kishimoto[1], Yasuhiro Maeda[3], Noriyuki Namba[1], Norio Sakai[2]

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[2]Department of Pediatrics, Osaka University hospital

[3]Department of Hospital Pharmacy, Graduate School of Pharmaceutical Sciences, Nagoya City University

P-43　Effectiveness of carglumic acid on ammonia detoxification in methylmalonic acidemia

○Aiko Gishi[1], Saki Kasuga[2], Makiko Fuyuki[2], Takashi Hamazaki[2], Shintaku Haruo[2]

[1]Department of Pediatrics, Osaka City University

[2]Department 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 Tashiro[1], Yoriko Watanabe[1, 2], Kaori Fukui[2], Yuichi Ochi[2], Shinichiro Nakagawa[2] Shuichi Ozono[2], Yushiro Yamashita[2]

[1]Research Institute of Medical Mass Spectrometry, Kurume University School of Medicine

[2]Department 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 Nishiyama, 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 Maeda[1], Kana Gotoh[1], Yoko Nakajima[2], Go Tajima[3], Yuji Hotta[1], Tomoya Kataoka[4] Kazunori Kimura[4], Tetsuya Ito[2]

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[3]National Center for Child Health and Development

[4]Department 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 Kim[1], Wung Joo Song[2], William Nyhan[3]

[1]Korea Genetics Research Center/KSZ Children's Hospital

[2]Korea Genetics Research Center/KSZ Children's Hospital, Cheongju, South Korea

[3]2UCSD Biochemical Genetics Laboratory, La Jolla, CA, USA

P-48　Classic organic acidurias in north-eastern Thai children: a seven-year experience.

○Khunton Wichajarn

Khon Kaen University

P-49　Elevated 3-hydroxyisovalerylcarnitine on newborn screening suggests mitochondrial disease

○Keisuke Okada[1], Ikuma Musha[1], Masato Arao[1], Yamazaki Taro[1], Toru kikuchi[1]

Megumi Saito[2], Hiroshi Mochizuki[3], Kei Murayama[4], Yasushi Okazaki[5], Akira Ohtake[1]

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[2]Intractable Disease Center, Saitama Medical University

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

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

P-50　Glutaric Aciduria type II in Thailand: Phenotypic spectrum, Biochemical, and Molecular features

○Nithiwat Vatanavicharn[1], Somporn Liammongkolkul[1], Boonchai Boonyawat[2]

Achara Sathienkijkanchai[1], Pornswan Wasant[1], Seiji Yamaguchi[3]

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[2]Department of Pediatrics, Phramongkutklao College of Medicine, Bangkok, Thailand

[3]Department of Pediatrics, Shimane University School of Medicine, Shimane, Japan

P-51　Blood purification therapy and clinical course at metabolic crisis of organic acidemia

○Kaname Hirashima[1], Shiro Matsumoto[2], Kido jun[2], Rieko Sakamoto[2], Hiroshi Mitsubuchi[1]

Fumio Endo[3], Kimitoshi Nakamura[2]

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[2]Department of Pediatrics, Kumamoto University School of Medicine

[3]Lake Kumamoto Ezu nursing medical center

＜Block4＞　17：20～17：55　Organic acid disorders and fatty acid disorders 3

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 Kimura[1], Eiichi Wake[1], Tomotaka Kono[1], Katsuya Aizu[1], Tomonosuke Someya[2]
Keiichi Hara[3], Go Tajima[4], Mitsuru Kubota[5], Akira Ohtake[6], Hiroshi Mochizuki[1]

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[2]Toyosu Medical Home for Children

[3]Department of Pediatrics, National Hospital Organization Kure Medical Center and Chugoku Cancer Center

[4]Division of Neonatal Screening, Research Institute, National Center for Child Health and Development

[5]Department of General Pediatrics and Interdisciplinary Medicine, National Center for Child Health and Development

[6]Department of Pediatrics, Saitama Medical University

P-53　An attempt of cooperation of local healthcare to glutaric aciduria type1 complicated by acute subdural hematoma.

○Akiko Yamada[1], Atsushi Iwabuchi[1], Kaori Kiyoki[1], Takahiro Kido[1], Tatsuyuki Ohto[1]
Takashi Fukushima[1], Tomohiro Kamoda[1], Ryo Sumazaki[1], Kana Tamai[2]
Michinobu Jogamoto[2]

[1]Department of Pediatrics, University of Tsukuba Hospital

[2]Department of Pediatrics, Moriya Daiichi General Hospital

P-54　2 cases of Short-chain acyl-CoA dehydrogenase deficiency discovered by neonatal metabolic screening

○Chunhua Zhang[1], Ning Zhao[1], Dan Yu[2], Xiaoli Tang[3]

[1]Department of research & development of MILS International

[2]Department of Pediatrics, West China Second University Hospital, Sichuan University

[3]Department 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 Hara[1], Go Tajima[2], Reiko Kagawa[3], Satoshi Okada[3], Nobuo Sakura[4]

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[2]Division of neonatal screening, National Center for Child Health and Development

[3]Department of Pediatrics. Hiroshima University Graduate School of Biomedical & Health Sciences

[4]Nursing Home for Severe Disabilities "Suzugamine".

P-56　Biochemical change after intravenous carnitine infusion in carnitine-acylcarnitine translocase deficiency

○Yasutsugu Chinen[1], Sadao Nakamura[1], Tadashi Kaname[2], Koichi Nakanishi[1]

[1]Department of Pediatrics, Faculty of Medicine, University of the Ryukyus

[2]Department of Genome Medicine, National Center for Child Health and Development

P-57 A patient with mHMG-CoA synthase defi. with developmental delay presented with acute decompensation

○Kaori Fukui[1], Yoriko Watanabe[1], Kojiro Nagai[1], Kyoko Tashiro[2], Tadateru Yasu[3]
Hideo Sasai[4], Yuki Hasegawa[5], Toshiyuki Fukao[4], Takahiro Inokuchi[2], Yushiro Yamashita[1]
[1]The Department of Pediatrics and Child Health Kurume University School of Medicine
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[3]Department of Pediatrics National Hospital Nagasaki Medical center
[4]Department of Pediatrics, Graduate School of Medicine, Gifu University
[5]Department 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[1], Ikue Hata[1], Takuya Kosaka[1], Yuko Isozaki[1], Yosuke Shigematsu[1]
Yusei Ohshima[1], Yoriko Watanabe[2, 3], Kyoko Tashiro[3], Keiichi Hara[4], Go Tajima[5]
[1]Department of Pediatrics, Faculty of Medial Sciences, University of Fukui
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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[1], Kei Murayama[2], Taro Yamazaki[3], Megumi Saitou[4], Takuya Fushimi[2]
Yukiko Yatsuka[5], Tomoko Hirata[6], Masakazu Kohda[7, 8], Yasushi Okazaki[7, 8], Akira Ohtake[3]
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[3]Department of Pediatrics, Faculty of Medicine, Saitama Medical University
[4]Department of Clinical Genetics, Saitama Medical University
[5]Division of Functional Genomics and Systems Medicine, Saitama Medical University
[6]Division of Translational Research, Research Center for Genomic Medicine, Saitama Medical University
[7]Research Center for Genomic Medicine, Saitama Medical University
[8]Intractable Disease Research Center, Juntendo University

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

○Kanako Ishii[1], Tocan Vlad[1], Noriko Oyama[1], Naoko Toda[1], Kazuhiro Ohkubo[1]
Yoshito Ishizaki[1], Koh-ichiro Yoshiura[2], Shouichi Ohga[1]
[1]Department of Pediatrics, Graduate School of Medical Sciences, Kyushu University
[2]Department 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[1, 2], Masakazu Kohda[2, 3], Yukiko Yatsuka[1], Tomoko Hirata[3], Yosuke Mizuno[1]
Atsuko Imai-Okazaki[1], Hiroko Harashima[4], Kei Murayama[5], Akira Ohtake[4], Yasushi Okazaki[1, 2, 3]

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[4]Department of Pediatrics, Saitama Medical University, Saitama.

[5]Department of Metabolism, Chiba Children's Hospital, Chiba.

P-62 Investigation of causative genes for mitochondrial oxidative phosphorylation (OXPHOS) disorders

○Sze Chern Lim[1], Yoshihito Kishita[1, 3], Masakazu Kohda[2, 3], Tomoko Hirata[2], Yukiko Yatsuka[1]
Atsuko Imai-Okazaki[1], Hiroko Harashima[4], Kei Murayama[5], Akira Ohtake[4]
Yasushi Okazaki[3, 6]

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P-63 Three cases of lethal infantile mitochondrial disease.

○Masakazu Honda[1], Hayato Sakurai[1], Tetsuya Kunikata[1], Taro Yamazaki[1]
Hiroko Harashima[1], Yoshihito Kishita[2], Masakazu Kohda[2], Yasushi Okazaki[2]
Kei Murayama[3], Akira Ohtake[1]

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

18：20 ～ 18：50 Mitochondrial disorder 2

Chairperson: Kondo Hidehito
(Osaka Univeristy Graduate School of Medicine, Department of Pediatrics)

P-64 Aspartate supplementation for aspartate-glutamate carrier isoform 1 deficiency

○Shoji Yano[1], Arthur Partikian[1], Stefan Bluml[1], Kathryn Moseley[1], Yoriko Watanabe[2]
Hirotomo Saitsu[3], Naomichi Matsumoto[3]

[1]Pediatrics, University of Southern California, [2]Pediatrics, Kurume University

[3]Human Genetics, Yokohama City University

P-65 Eleven novel mutations in six Chinese patients with thiamine metabolism dysfunction syndrome

○Dongxiao Li[1], Xiyuan Li[2], Yi Liu[2], Hui Dong[2], Jinqing Song[2], Yupeng Liu[2], Yao Zhang[2]
Ying Jin[2], Hanzhou Guan[3], Yanling Yang[2]

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[3]Department 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 Univerisity School of Medicine

P-67 A novel mutation in *TAZ* causes mitochondrial respiratory chain disorder without cardiomyopathy

○Nurun Nahar Borna[1], Yoshihito Kishita[1, 2], Kaori Ishikawa[3], Kazuto Nakada[3], Masakazu Kohda[2, 4], Takashi Nasu[5], Atsuhito Takeda[6], Kei Murayama[7], Akira Ohtake[8], Yasushi Okazaki[2, 9]

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[3]Faculty of Life and Environmental Sciences, University of Tsukuba, Tsukuba.

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[5]Department of Pediatrics, Obihiro Kosei General Hospital, Obihiro.

[6]Department of Pediatrics, Hokkaido University Graduate School of Medicine, Hokkaido.

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[8]Department of Pediatrics, Saitama Medical University, Saitama

[9]Division 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 Hirose[1], Hitoshi Osaka[1], Masako Nagashima[1], Hirokazu Yamagishi[1], Sumie Ohhata[1]
Jun Odaka[1], Yu-ichi Goto[2], Takanori Yamagata[1]

[1]Department of Pediatrics, Jichi Medical University

[2]Department 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

Beijng children's hospital, capital medical university

＜ Block5 ＞　17：20 ～ 17：50　Amino acid disorders 1

Chairperson: Masayoshi Nagao

(National Hospital Organization Hokkaido Medial Center)

P-70　Replacement therapy for selenium and biotin in patients with phenylketonuria

○Yoshiyuki Okano[1, 2], Miki Okamoto[1], Toshiaki Watanabe[3], Ryoko Watanabe[3], Rika Fujii[3]
Yosuke Shigematsu[4], Tomoko Tamaoki[2], Yasuhiro Takeshima[2]
[1]Okano Children's Clinic, [2]Hyogo College of Medicine
[3]Osaka Aoyama University, [4]Fukui University

P-71　Successful pregnancy in a PKU woman treated with tetrahydrobiopterin

○Natsuko Arai-Ichinoi[1], Osamu Sakamoto[1], Shigeo Kure[1], Kei Murayama[2]
[1]Department of Pediatrics, Tohoku University School of Medicine
[2]Department 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 Kono[1], Tae Kimura[1], Eiichi Wake[1], Katsuya Aizu[1], Akira Ohtake[2]
Hiroshi Mochizuki[1]
[1]Division of Endocrinology and Metabolism, Saitama Children's Medical Center
[2]Department of Pediatrics, Saitama Medical University

P-74　Phase 3 Long-term Study Evaluating Efficacy and Safety of Pegvaliase Treatment in Adults with PKU

○Nicola Longo[1], Jeffery Vockley[2], Harvey Levy[3], Stephen Amato[4], Roberto Zori[5]
Janet Thomas[6], Barbara Burton[7], Cary Harding[8], John Posner[9], Deborah Bilder[1]
Joy Olbertz[10], Zonghua Gu[10], Kelly Lau[10], Mingjin Lin[10], Kevin Larimore[10], David Dimmock[11]
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[2]University of Pittsburgh and Children's Hospital of Pittsburgh, Pittsburgh, PA
[3]Boston Children's Hospital, Boston, MA, [4]University of Kentucky, Lexington, KY
[5]University of Florida, Gainesville, FL, [6]University of Colorado School of Medicine, Aurora, CO
[7]Lurie Children's Hospital of Chicago, Chicago, IL
[8]Oregon Health & Science University, Portland, OR, [9]King's College, London, UK
[10]BioMarin Pharmaceutical Inc., Novato, CA
[11]Rady 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 Longo[1], Cary Harding[2], Stephen Amato[3], Jeffery Vockley[4], Klaas Wierenga[5]
Hong Li[6], Deborah Bilder[1], Barbara Burton[7], David Dimmock[8], John Posner[9]
Janet Thomas[10], Roberto Zori[11], Orli Rosen[12], Zonghua Gu[12], Mingjin Lin[12]
Markus Merilainen[12], Haoling H. Weng[12], Harvey Levy[13]
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[3]University of Kentucky, Lexington, KY
[4]University of Pittsburgh and Children's Hospital of Pittsburgh, Pittsburgh, PA
[5]University of Oklahoma, Oklahoma City, OK, [6]Emory University, Decatur, GA
[7]Lurie Children' s Hospital of Chicago, Chicago, IL
[8]Rady Children's Institute for Genomic Medicine, San Diego, CA
[9]King's College, London, UK, [10]University of Colorado School of Medicine, Aurora, CO
[11]University of Florida, Gainesville, FL, [12]BioMarin Pharmaceutical Inc., Novato, CA
[13]Boston 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 deficnecy

○Hironori Kobayashi[1], Toshikazu Minohata[1], Hiroshi Mitsubuchi[2], Kimitoshi Nakamura[2]
Mitsuru Kubota[3], Osamu Sakamoto[4], Yoriko Watanabe[5], Kaori Fukui[5], Furujo Mahoko[6]
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[3]Department of General Pediatrics & Interdisciplinary Medicine, National Center for Child Health and Development
[4]Department of Pediatrics, Tohoku University School of Medicine
[5]Department of pediatrics, Kurume University School of Medicine
[6]Department of pediatrics, Okayama Mediacal Center

P-77 Hypocitrullinemia in newborn MS/MS screening with late-onset OTC deficiency

○Tomoko Lee[1], Katsuhiko Yoshii[2], Satoru Yoshida [3], Takenori Suga[4], Kimitoshi Nakamura[5]
Toshiyuki Fukao[6], Kei Murayama[7], Yuki Hasegawa[8], Yasuhiro Takeshima[1]
[1]Department of Pediatrics, Hyogo College of Medicine
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[3]Department of pediatrics, Seirei Mikatahara General Hospital
[4]Department of pediatric emergency medicine, Hyogo Prefectural Amagasaki general Medical Center
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[6]Department of Pediatrics, Gifu University Graduate School of Medicine
[7]Department of Metabolism, Chiba Children's Hospital
[8]Department 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 Kido[1], Takanobu Yoshida[1], Shirou Matsumoto[1], Hiroshi Mitsubuchi[1], Rieko Sakamoto[1] Fumio Endo[2], Kimitoshi Nakamura[1]

[1]Department of Pediatrics, Kumamoto University

[2]Department 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 Ohtake

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 Iwano[1], Hideki Nakajima[2], Junko Hanakawa[3], Yumi Asakura[3], Masanori Adachi[3] Koji Muroya[3]

[1]Clinical Research Institute, Kanagawa Children's Medical Center

[2]Division of Neonatal Screening, National Center for Child Health and Development

[3]Department 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 Hoshino[1], Sumire Kumai[1], Toru Maeda[1], Takenori Adachi[1], Takashi Kawabe[1] Katsuyuki Yokoi[2], Yoko Nakajima[2], Tetsuya Ito[2], Toshihiro Yasui[2], Tatsuya Suzuki[2]

[1]Department of Pediatrics, Kasugai Municipal Hospital

[2]Department of Pediatrics, Fujita Health University

P-85 Cobalamin disorder CblC presenting acute pulmonary edema detected by expanded newborn screening

○Shoko Sakamoto[1], Osamu Sakamoto[2], Takashi Hamazaki[3], Haruo Shintaku[3]

[1]Department Pediatrics, Osaka City University, [2]Department of Pediatrics, Tohoku University

[3]Department 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 Tsuruoka[1], Yutaka Fuchinoue[2], Takao Kuroki[2], Hiroyuki Hiruta[2], Akihiko Tateno[2]

Norikazu Shimizu[2], Tsugutoshi Aoki[2]

[1]Department of Pediatrics, Toho University Ohashi Medical Center

[2]Toho 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 Osawa[1], Aya Wada[1], Yoko Tabei[1], Yoshiaki Ohtsu[1], Junko Igaki[2]

Kenichi Maruyama[3], Hirokazu Arakawa[1]

[1]Gunma University Graduate School of Medicine, Depertment of Pediatrics, Gunma, Japan

[2]Depertment of Endcrinology and Metabolism, Nanbu Medical Center・Nanbu Child Medical Center, Okinawa Japan

[3]Depertment 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 Yorifuji[1], Hiroko Kadowaki[2], Yushi Hirota[3], Wataru Ogawa[3], Hideki Katagiri[4]
Yasuhi Ishigaki[5], Takashi Akamizu[6]

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[6]First 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 Takashima[1], Shoko Takemoto[1], Kayoko Toyoshi[1], Akiko Ohba[1], Haruka Fujita[2]
Kentaro Oh-hashi[2], Yoko Morita[2], Nobuyuki Shimozawa[1]

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[2]Graduate School of Natural Science and Technology, Gifu University

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

○Masashi Morita[1], Kaito Tomita[2], Sato Hideaki[2], Shun Matumoto[2], Shiro Watanabe[3]
Nobuyuki Shimozawa[4], Tsuneo Imanaka[5]

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[3]Institute of Natural medicine, University of Toyama

[4]Division of Genomic Research, Life Science Research Center, Gifu University

[5]Faculty of Pharmaceutical Sciences, Hiroshima International University

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

○Toshiki Tsunogai[1], Masayoshi Yamaoka[1], Masaharu Akiyama[1], Hiroyuki Ida[1], Toya Ohashi[2]

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

[2]Department 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 Li[1], Yuan Ding[2], Hui Dong[2], Jinqing Song[2], Xiyuan Li[2], Ying Jin[2], Yupeng Liu[2]
Lifang Feng[3], Xiaohong Chen[3], Hong Zheng[4]

[1]Department of Pediatrics, Peking University First Hospital; Rehabilitation centre, Henan Children's Hospital.

[2]Department of Pediatrics, Peking University First Hospital

[3]Department of Endocrinology and Metabolism, Wuhan Medical and Healthcare Center for Women and Children, China

[4]Department 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 Liu[1], Qiuhong Xiong[2], Xiyuan Li[1], Yu Xue[2], Jing Wang[2], Dongxiao Li[1], Ying Jin[1]
Changxin Wu[2], Han Xiao[2], Yanling Yang[1]

[1]Department of Pediatrics, Peking University First Hospital

[2]Institute of Biomedical Sciences, Shanxi University

P-97 A case with severe acidosis and unconsciousness suspected of inborn errors in keton body metabolism

○Miki Matsui[1], Satoshi Oonishi[2], Tomoko Lee[3], Yo Okizuka[2], Taisuke Hashimoto[4]
Hironori Kobayashi[5]
Yuki Hasegawa[5], Hideo Sasai[6], Toshiyuki Fukao[6], Hirotaka Minami[1]

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[3]Department of pediatrics, Hyogo College Of Medicine

[4]Department of pediatrics, Toyonaka Municipal Hospital

[5]Department of Pediatrics, Shimane University School of Medicine

[6]Department of Pediatrics, Gifu University Graduate School of Medicine

P-98 A case of dihydropyrimidinase deficiency accidentally discovered by urinary metabolome analysis

○Hiroki Tsuchiya[1], Tomoyuki Akiyama[1], Tomiko Kuhara[2], Yoko Nakashima[3]
Takashi Shibata[1], Mari Akiyama[1], Makio Oka[1], Fumika Endo[1], Harumi Yoshinaga[4]
Katsuhiro Kobayashi[1]

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[2]Japan Clinical Metabolomics Institute

[3]Department of Pediatrics, Fujita Health University

[4]National Hosipital 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 Tomatsu[1, 2], Shaukat A. Khan[2], Hira Peracha[2], Diana Ballhausen[3], Alfred Wiesbauer[4]
Yasuyuki Suzuki[5], Kenji E. Orii[6], Tadao Orii[6]

[1]Departments of Biomedical Research and Orthopedics

[2]Nemours/Alfred I. duPont Hospital for Children

[3]Centre for molecular diseases, Service for genetic medicine, University Hospital

[4]Institute of Social and Preventive Medicine, University of Bern

[5]Medical Education Development Center, Gifu University

[6]Department of Pediatrics, Gifu University

P-100　Current status of Japan Registration System for Metabolic & Inherited Diseases

○Joo-Hyun Seo[1], Makiko Miyairi[1], Akira Ohtake[2], Torayuki Okuyama[1]

[1]Clinical Laboratory Medicine, National Center for Child Health and Development

[2]Department of Pediatrics, Saitama Medical University

P-101　Development of Bone Targeting Drugs

○Shunji Tomatsu[1], Molly Stapleton[1], Kazuki Sawamoto[1], Carlos J. Alméciga-Díaz[2], Tadao Orii[3]

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[2]Institute for the Study of Inborn Errors of Metabolism, Pontificia Universidad Javeriana

[3]Department of Pediatrics, Graduate School of Medicine, Gifu University

P-102　Therapeutic strategy for Fabry disease peripheral neuropathy with rAAV vector

○Takashi Higuchi[1], Yohta Shimada[1], Hiroshi Kobayashi[1], Takahiro Fukuda[2], Fusao Kato[3]
Toya Ohashi[1]

[1]Division of Gene Therapy, The Jikei University

[2]Department of Neuropathology, The Jikei University

[3]Division of Neuroscience, The Jikei University