# Day 1：Nov. 8（Thu）
# Venue 1（2F Hall）

## 8：50〜9：00　Opening Remarks

## 9：00〜10：00　English Session 1：Treatment

**Chairpersons: Shunji Tomatsu**
（Alfled I. duPont Hospital for Children）
**Toya Ohashi**
（Department of Pediatrics, Division of Gene Therapy, Research Center for Medical Sciences, The Jikei University School of Meidicine）

### ES-01　Phase 3 Study to Assess Safety and Efficacy of Pegvaliase in Adult Japanese Subjects With Phenylketonuria

○ Haruo Shintaku[1], Joy Olbertz[2], Xinqun Yang[2], Shahid Khan[2], Soumi Gupta[2], Qing Chang[2]
Haoling H Weng[2]
[1]Osaka City University Graduate School of Medicine
[2]BioMarin Pharmaceutical Inc., Novato, CA, USA

### ES-02　Phase 3 PRISM studies of pegvaliase for the treatment of adult phenylketonuria（PKU）

○ Janet Thomas[1], Harvey Levy[2], Stephen Amato[3], Jerry Vockley[4], Roberto Zori[5], David Dimmock[6]
Cary O Harding[7], Debora A Bilder[8], Haoling H Weng[9], Hope Northrup[10]
[1]University of Colorado
[2]Boston Children's Hospital, Boston, MA, USA
[3]University of Kentucky, Lexington, KY, USA
[4]University of Pittsburgh and Children's Hospital of Pittsburgh, Pittsburgh, PA, USA
[5]University of Florida, Gainesville, FL, USA
[6]Medical College of Wisconsin, Milwaukee, WI, USA
[7]Oregon Health and Science University, Portland, OR, USA
[8]University of Utah, Salt Lake City, UT, USA
[9]BioMarin Pharmaceutical Inc., Novato, CA, USA
[10]University of Houston Medical School, Houston, TX, USA

### ES-03　Development of AAV Gene Therapy for Morquio A Syndrome

○ Shunji Tomatsu[1], Kazuki Sawamoto[2], Shaukat Kahn[2], Molly Stapleton[2]
Subha Karumuthil-Melethil[3], Olivier Danos[3]
[1]Nemours/Alfred I. duPont Hospital for Children
[2]Biomedical, Nemours/Alfred I. duPont Hospital for Children, WILMINGTON, USA
[3]REGENXBIO Inc., Rockville, USA

### ES-04　Development of gene therapy for GM2 gangliosidoses by utilizing AAV vector

○ Kohji Itoh[1,2], Yukiya Ohnishi[1], Daisuke Tsuji[1,2], Ryousuke Watanabe[2], Katsuhito Asai[3]
Shin-ichi Muramatsu[4]
[1]Department of Medicinal Biotechnology, Institute for Medicinal Research, Graduate School of Pharmaceutical Scinece, Tokushima University
[2]Department of Medicinal Biotechnology, Faculty of Pharmaceutical Sciences, Tokushima University
[3]Gene Therapy Research. Institution, Co., Ltd.
[4]Division of Neurology, Faculty of Medicine, Jichi Medical University

### ES-05 Correction of aberrant splicing of G6PC by c.648G to T variant using antisense oligonucleotides

○ Go Tajima[1,2], Miyuki Tsumura[2], Satoshi Okada[2], Satoshi Obika[3]
[1]Division of Neonatal Screening, Research Institute, National Center for Child Health and Development
[2]Department of Pediatrics, Hiroshima University Graduate School of Biomedical & Health Sciences
[3]Graduate School of Pharmaceutical Sciences, Osaka University

## 10：00～10：48 English Session 2：Lysosomal Disorders

**Chairpersons: Motomichi Kosuga**
**(Division of Medical Genetics, National Center for Child Health and Development)**
**Masahisa Kobayashi**
**(Department of Pediatrics, The Jikei University School of Medicine)**

### ES-06 Newborn screening for mucopolysaccharidoses by GAG assay with tandem mass spectrometry

○ Shunji Tomatsu[1], Francyne Kubaski[2], Molly Stapleton[2], Hironori Kobayashi[3], Seiji Yamaguchi[3]
[1]Nemours/Alfred I. duPont Hospital for Children
[2]Biomedical, Nemours/Alfred I. duPont Hospital for Children, WILMINGTON, USA
[3]Department of Pediatrics, Shimane University

### ES-07 Ten-year-long ERT shows a poor effect to reduce sphingolipid accumulation in patients with Fabry disease

○ Mohammad Arif Hossain[1], Chen Wu[1], Keiko Akiyama[1], Takashi Miyajima[1], Takeo Iwamoto[2]
Yoshikatsu Eto[1]
[1]Advanced Clinical Research Center
[2]Jikei University School of Medicine

### ES-08 Elevation of biomarkers in Niemann-Pick disease type C-affected individuals

○ Ryuichi Mashima[1], Masamitsu Maekawa[2], Aya Narita[3], Torayuki Okuyama[1], Nariyasu Mano[2]
[1]Department of Clinical Laboratory Medicine
[2]Tohoku University Hospital
[3]Tottori University Faculty of Medicine

### ES-09 Mucopolysaccharidosis-plus syndrome: report of two new cases

○ Filipp Vasilev[1,2,3], Yukiko Kawakami[1], Elizabeta Gurinova[4], Aitalina Sukhomyasova[3,4]
Nadezda Maksimova[3], Junko Matsuda[1], Takanobu Otomo[1]
[1]Department of Pathophysiology and Metabolism, Kawasaki Medical School
[2]JSPS International Postdoctoral Fellow
[3]North-Eastern Federal University, Yakutsk, Russia
[4]Republican Hospital no. 1, Yakutsk, Russia

## 11：00～11：50　Sponsored Seminar 1　Sponsored by Amicus Therapeutics K.K.

**Chairperson: Yoshikatsu Eto**

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

### SS1　Oral Chaperone Therapy for Fabry Disease

○ Roberto Giugliani

Department of Generics, UFRGS/Medical Genetics Service, HCPA, Porto Alegre, Brazil

## 12：00～13：00　Luncheon Seminer 1　Sponsored by Shire Japan KK

**Chairperson: Hiroyuki Ida**

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

### LS1　How to make treatment choices in Gaucher disease in 2018 and beyond

○ Ari Zimran

Gaucher Clinic, Shaare Zedek Medical Center, Jerusalem, Israel

## 13：10～13：40　JSIMD Annual General Assembly

## 13：40～14：10　JSIMD Award Lecture

**Chairperson: Masaki Takayanagi**

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

### JAL　Shifts in the diagnostic paradigm of inherited metabolic diseases

○ Akira Ohtake

Department of Pediatrics, Faculty of Medicine, Saitama Medical University; Center for Intractable Diseases, Saitama Medical University Hospital

## 14：10～15：25　Young Researcher Award Candidates' Presentation

**Chairpersons: Toshihiro Ohura**

(Division of Pediatrics, Sendai City Hospital)

**Ken Sakurai**

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

### YRC-1　Comprehensive understanding of lysosomal storage disease using genome-editing technology

○ Takanobu Otomo, Yukiko Kawakami, Filipp Vasilev, Junko Matsuda

Department of Pathophysiology and Metabolism, Kawasaki Medical School

### YRC-2　Newborn Screening Program for Pompe Disease in Japan: Towards Earlier Diagnosis for Patients

○ Ken Momosaki

Department of Pediatrics, Kumamoto University

### YRC-3 Gene panel study for target metabolic diseases of newborn mass screening in Japan

○ Hideo Sasai[1,2], Tetsuya Ito[3], Hironori Kobayashi[4], Masahisa Kobayashi[5], Go Tajima[6]
Osamu Sakamoto[7], Kimitoshi Nakamura[8], Takashi Hamazaki[9], Yuki Hasegawa[4]
Toshiyuki Fukao[1,2]
[1]Department of Pediatrics, Graduate School of Medicine, Gifu University
[2]Division of Clinical Genetics, Gifu University Hospital
[3]Department of Pediatrics, School of Medicine, Fujita Health University
[4]Department of Pediatrics, Shimane University Faculty of Medicine
[5]Department of Pediatrics, The Jikei University School of Medicine
[6]Division of Neonatal Screening, NCCHD
[7]Department of Pediatrics, Tohoku University School of Medicine
[8]Department of Pediatrics, Kumamoto University Graduate School of Medical Sciences
[9]Department of Pediatrics, Faculty of Medicine, Osaka City University

### YRC-4 Development of a novel chimeric mouse model using the hepatocytes of patients with OTC deficiency

○ Go Sugahara[1], Chihiro Yamasaki[1], Ami Yanagi[1], Suzue Furukawa[1], Kazuaki Nakamura[2]
Shin Enosawa[2], Yuji Ishida[1,3], Chise Tateno[1,3]
[1]PhoenixBio Co., Ltd.
[2]National Center for Child Health and Development
[3]Research Center for Hepatology and Gastroenterology, Hiroshima University

### YRC-5 AAV-mediated gene therapy for GM2 gangliosidosis model mouse

○ Yukiya Ohnishi[1], Daisuke Tsuji[1,2], Ryosuke Watanabe[2], Shin-ichi Muramatsu[3], Kohji Itoh[1,2]
[1]The Department of Medicinal Biotechnology, Graduate School of Biomedical Science, University of Tokushima
[2]The Faculty of Pharmaceutical Science, University of Tokushima
[3]The Faculty of Medicine, Jichi Medical University

### YRC-6 Gene therapy for *Glut1*-deficient mice using AAV vectors with GLUT1 promoter

○ Sachie Nakamura[1], Hitoshi Osaka[1], Shin-ichi Muramatsu[2,3], Naomi Takino[2], Eriko Jimbo F[1]
Kuniko Shimazaki[4], Tatsushi Onaka[5], Sumio Ohtsuki[6], Tetsuya Terasaki[7], Takanori Yamagata[1]
[1]Department of Pediatrics, Jichi Medical University
[2]Division of Neurology, Jichi Medical University
[3]Center for Gene and Cell Therapy, The Institute of Medical Science, The University of Tokyo
[4]Department of Neurosurgery, Jichi Medical University
[5]Division of Brain and Neurophysiology, Department of Physiology, Jichi Medical University
[6]Department of Pharmaceutical Microbiology, Faculty of Life Sciences, Kumamoto University
[7]Division of Membrane Transport and Drug Targeting, Graduate School of Pharmaceutical Sciences, Tohoku University

## 15：30～16：00 SLEIMPN Recommending Lecture

**Chairperson: Torayuki Okuyama**
**(National Center for Child Health and Development)**

### SLEIMPN Mucopolysaccharidoses: From Understanding to Treatment, a Century of Discoveries

○ Roberto Giugliani
Federal University of Rio Grande do Sul, Brazil

## 16：00～16：30　SIMD Recommending Lecture

**Chairperson: Shigeo Kure**

（Department of Pediatrics, Tohoku University School of Medicine）

### SIMD　Functional genomics screening of variants causing mitochondrial disease

○ Penelope Bonnen

Department of Molecular & Human Genetics, Baylor College of Medicine, USA; NASA Human Research Program and NASA Human Health & Performance Directorate, USA

## 16：30～17：30　English Session 3：Mitochondrial Disorders

**Chairpersons: Kei Murayama**

（Center for Medical Genetics, Department of Metabolism, Chiba Children's Hospital）

**Shoji Yano**

（Genetics Division, Department of Pediatrics, University of South California）

### ES-10　Novel mitochondrial cardiomyopathy with elevated sister chromatid exchanges: TOP3A deficiency

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

[1]Department of Metabolism, Chiba Children's Hospital
[2]Center for Medical Genetics, Chiba Children's Hospital
[3]Intractable Disease Research Center, Juntendo University
[4]Department of Pediatrics, Saitama Medical University

### ES-11　Clinical features of C1QBP deficiency

○ Keiko Ichimoto[1], Takuya Fushimi[2], Naomi Kuranobu[1], Masaru Shimura[1], Ayako Matsunaga[1]
Yoshihito Kishita[2], Masakazu Kouda[2], Yasushi Okazaki[2], Akira Ohtake[3], Kei Murayama[1]

[1]Center for Medical Genetics, Department of Metabolism, Chiba Children's Hospital
[2]Intractable Disease Research Center, Juntendo University
[3]Department of Pediatrics, Saitama Medical University

### ES-12　Analysis of 154 cases with mitochondrial DNA variations in Chinese children with mitochondrial disorders

○ Yuqing Shi, Fang Fang, Zhimei Liu, Danmin Shen, Lifang Dai, Weihua Zhang, Jiuwei Li
Xiaotun Ren, Tongli Han, Changhong Ding

Department of Neurology, Beijing Children's Hospital, Capital Medical University, National Center for Children's Health, Beijing, China

### ES-13　A neurophysiologic model of MELAS disease using human inducible pluripotent stem cell derived excitatory cortical neurons

○ Tamas Kozicz[1], Teun M Klein Gunnewiek[2], David Cassiman[3], Eva Morava[1], Neal Nadif Kasri[4]

[1]Department of Clinical Genomics, Mayo Clinic, Rochester MN, USA
[2]Department of Anatomy, Radboud University Medical Centre, Donders Institute for Brain, Cognition and Behaviour, Nijmegen, The Netherlands
[3]University Hospital Leuven, Leuven, Belguim
[4]Department of Congnitive Neuroscience, Radboud Univesity Medical Centre, Donders Institute for Brain, Congnition and Behaviour, Nijmegen, The Netherlands

### ES-14 Mitochondrial dysfunction is associated with trauma exposure and susceptibility to post traumatic stress disorder（PTSD）in mice

○ Tamas Kozicz[1], Graeme Preston[2], Hans Anderson[2], Tim Emmerzaal[3], Eva Morava[1]
[1]Department of Clinical Genomics, Mayo Clinic, Rochester MN, USA
[2]Hayward Genetics Center, Department of Pediatrics, Tulane University school of Medicine, New Orleans, LA, USA
[3]Department of Anatomy, Radboud University Medical Centre, Donders Institute for Brain, Cognition and Behaviour, Nijmegen, The Netherlands

## 17：35～18：20 Special Lecture 1

**Chairperson: Seiji Yamaguchi**

（Department of Pediatrics, Shimane University School of Medicine）

### SL1 Correction of mitochondrial fatty acid oxidation or respiratory chain defects by drugs or natural compounds

○ Jean Bastin, Fatima Djouadi
INSERM U1124 Université Paris-Descartes, Paris, France

# Day 1：Nov. 8（Thu）
# Venue 2（5F Conference Room）

## 9：10〜9：58　Oral 1：Disorders of Amino Acid Metabolism 1

**Chairperson: Mika Ishige**

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

### O-01　Effectiveness of Pramipexole in On-Off phenomenon of an adolescent patient with PTPS deficiency


○ Mika Ishige[1], Erika Ogawa[1], Chika Takano[1], Yumi Nishimura[1], Yuki Kasuga[1], Haruo Shintaku[2]
Tatsuo Fuchigami[1], Ichiro Morioka[1]

[1]Department of Pediatrics and Child Health, Nihon University School of Medicine
[2]Department of Pediatrics, Osaka City University Graduate School of Medicine


### O-02　Neonatal methionine adenosyltransferase I/III deficiency with brainstem lesions


○ Takaaki Sawada, Jun Kido, Rieko Sakamoto, Ken Momosaki, Yosuke Suzuki, Hiroshi Mitsubuchi
Kimitoshi Nakamura, Shiro Matsumoto

Department of Pediatrics, Kumamoto University


### O-03　Asparagine synthetase deficiency: functional analysis of mutations identified in Japanese patients


○ Hideki Matsumoto, Takahiro Yamamoto, Yasuhiko Ago, Hideo Sasai, Kazuo Kubota
Takeshi Kimura, Michio Ozeki, Norio Kawamoto, Hidenori Ohnishi, Toshiyuki Fukao

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


### O-04　Characterization of infants with abnormal newborn screening for mild hypersuccinylacetonemia


○ Kimihiko Oishi[1,2], Neal Cody[1], Amy Williamson[1], Lissette Estrella[1], Chunli Yu[1], George Diaz[1]

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


## 9：58〜10：34　Oral 2：Disorders of Amino Acid Metabolism 2

**Chairperson: Kenji Yamada**

（Department of Pediatrics, Shimane University Faculty of Medicine）

### O-05　Plasma nitric oxide and oxidative stress markers in lysine urinary protein intolerance


○ Atsuko Noguchi[1], Daiki Kondo[1,2], Wakako Kikuchi[1], Yuhei Takasago[3], Hirokazu Tsukahara[4]
Tsutomu Takahashi[1]

[1]Department of Pediatrics, Akita University Graduate School of Medicine
[2]Hiraka General Hospital
[3]Morioka Children's Hospital
[4]Okayama University Graduate School of Medicine

### O-06 Medium-chain triglycerides supplement with a low-carbohydrate formula for type II citrullinemia

○ Kiyoshi Hayasaka[1,2], Chikahiko Numakura[1], Mitsunori Yamakawa[3], Tetsuo Mitsui[1]
Hisayoshi Watanabe[4], Masahide Yazaki[5], Hiromasa Ohira[6], Toshiyuki Tahara[7]
Takahiro Nakayama[8], Hiroshi Mitsubuchi[9], Hiroshi Yoshida[10]
[1]Department of Pediatrics, Yamagata University School of Medicine
[2]Department of Pediatrics, Miyukikai Hospital
[3]Department of Pathological Diagnostics, Yamagata University School of Medicine
[4]Department of Gastroenterology, Yamagata University School of Medicine
[5]Department of Biological Sciences for Intractable Neurological Disorders, Institute for Biomedical Sciences, Shinshu University
[6]Department of Gastroenterology, Fukushima Medical University School of Medicine
[7]Department of Gastroenterology, Saiseikai Utsunomiya Hospital
[8]Division of Internal Medicine, Nihonkai General Hospital
[9]Department of Pediatrics, Graduate School of Medical Sciences, Kumamoto University
[10]Department of Pediatrics, Tsuruoka Municipal Shonai Hospital

### O-07 Citrin-KO mice show hyperammonemia under high hepatic cytosolic NADH conditions

○ Takeyori Saheki[1], Aki Funahashi[1], Eishi Kuroda[1], Izumi Yasuda[1], Yoshiko Setogawa[1]
Qinghua Gao[1], Momoko Hamano[1], Miharu Ushikai[1], Masahisa Horiuchi[1], Mitsuaki Moriyama[2]
[1]Department of Hygiene and Health Promotion Medicine
[2]Laboratory of Integrative Physiology in Veterinary Sciences, Osaka Prefecture University

## 11：00～11：50 Sponsored Seminar 2 Sponsored by OrphanPacific, Inc.

**Chairperson: Kimitoshi Nakamura**
**(Department of Pediatrics, Kumamoto University Graduate School of Medical Sciences)**

### SS2 Experiences of urea cycle disorder treatment

○ Yoko Nakajima
Department of Pediatrics, Fujita Health University School of Medicine

## 12：00～13：00 Luncheon Seminer 2

**Sponsored by JCR Pharmaceuticals Co., Ltd.**

**Chairperson: Toshiyuki Fukao**
**(Department of Pediatrics, Graduate School of Medicine, Gifu University)**

### LS2-1 Experience in using biosimilars in enzyme replacement treatment of Fabry disease

○ Hideo Sasai[1,2], Kazuo Kubota[1,2], Yasuhiko Ago[1], Hideki Matsumoto[1], Takahiro Yamamoto[3]
Norio Kawamoto[1], Minako Kawamoto[1], Hidenori Ohnishi[1], Toshiyuki Fukao[1]
[1]Department of Pediatrics, Graduate School of Medicine, Gifu University
[2]Division of Clinical Genetics, Gifu University Hospital
[3]Department of Disability medicine, Graduate School of Medicine, Gifu University

### LS2-2 Early diagnosis of adrenoleukodystrophy（ALD）

○ Nobuyuki Shimozawa
Division of Genomics Research, Life Science Research Center, Gifu University

## 15：30〜16：18　Oral 3：Disorders of Metal Metabolism

**Chairperson: Norikazu Shimizu**

(Department of Pediatrics, Toho University Ohashi Medical Center)

### O-08　Mutational analysis of 11Wilson disease patientsin our center and its clinical correlation

○ Narutoshi Yamazaki[1,2], Yasuyuki Fukuhara[1], Asami Hirakiyama[3], Ai Miura[3], Tetsumin So[4]
Reiko Ito[5], Juhyon So[3], Akira Ishiguro[2], Motomichi Kosuga[1,3], Torayuki Okuyama[3]
Akinari Fukuda[6]

[1]National Center for Child Health and Development, Division of Medical Genetics
[2]National Center for Child Health and Development, Postgraduate Education and Training Center
[3]National Center for Child Health and Development, Department of Clinical Laboratory Medicine
[4]National Center for Child Health and Development, Department of Critical Care of Medicine
[5]National Center for Child Health and Development, Department of General Medicine
[6]Organ Transplantation Center, National Center for Child Health and Development

### O-09　A Wilson Disease patient with acute liver failure due to poor medical compliance

○ Kouki Sasamoto, Shinji Utsunomiya, Chieko Ito, Norikazu Shimizu, Tsugutoshi Aoki
Department of Pediatrics, Toho University Ohashi Medical Center

### O-10　A Wilson disease patient with hypoxemia due to hepatopulmonary syndrome

○ Miku Hattori, Shoko Nakazawa, Chieko Ito, Norikazu Shimizu, Tsugutoshi Aoki
Pediatrics, Toho University Ohashi Medical Center

### O-11　A 43-year-old female of beta-propeller protein-associated neurodegeneration by the *WDR45* mutation

○ Mariko Yagi[1], Yoko Matsumoto[1], Mio Nishimura[1], Mariko Takata[2,3], Wataru Satake[2,3]
Kenji Sekiguchi[2], Tatsushi Toda[4], Yoko Kawasaki[1]

[1]Department of Pediatrics, Nikoniko House Medical and Welfare Center
[2]Division of Neurology, Kobe University Graduate School of Medicine
[3]Division of Molecular Brain Science, Kobe University Graduate School of Medicine
[4]Department of Neurology, The University of Tokyo

## 16：20〜18：20　Symposium 1：Treatment Plan for Mucopolysaccharidosis

**Chairpersons: Torayuki Okuyama**

(National Center for Child Health and Development)

**Shunji Tomatsu**

(Nemours/Alfred I. duPont Hospital for Children)

### SY1-1　10 years of experience creating options and possibilities for treatments for the next 10 years

○ Takeyuki Akiyama
The Japanese MPS Society

### SY1-2　Challenges in diagnosis and treatment for MPS

○ Motomichi Kosuga
Division of Medical Genetics, National Center for Child Health and Development

### SY1-3　Treatment of Mucopolysacchridosis: Current and Future

○ Torayuki Okuyama
Nationl Center for Child Health and Development

## SY1-4 Effect and limitation of hematopoietic stem cell transplantation for mucopolysaccharidoses

○ Shunji Tomatsu[1,2,3]

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

[2]Departments of Orthopedics and BioMedical, Skeletal Dysplasia

[3]Department of Pediatrics, Sidney Kimmel Medical College at Thomas Jefferson University

## SY1-5 Newborn Screening for Mucopolysaccharidosis

○ Kimitoshi Nakamura

Department of Pediatrics, Kumamoto University Graduate School of Medical Sciences

# Day 1：Nov. 8（Thu）
# Poster Presentations

## 18：30〜18：55　Poster 1：Lysosomal Disorders・Peroxisomal Disorders 1
## （Poster Venue 1 : 4F Room 1）

**Chairperson: Kazuya Tsuboi**

（LSD Center, Nagoya Central Hospital）

### P-01　Infantile – onset Pompe disease（IOPD）: Phenotype, genotype and outcome of 14 Vietnamese patients with ERT

○ Khanh Nguyen, Dung Chi Vu, Mai Thanh Do, Ngoc Thi Bich Can, Vuong Minh Nguyen
Shu-Chuan Chiang, Wuh-Liang Hwu
National Children's Hospital

### P-02　A case of late-onset Pompe disease followed up as hepatic dysfunction of unknown origin

○ Go Takei[1,2], Tetsuharu Kamioka[2,3], Hiroshi Terashima[2,4], Masaya Kubota[2], Reiko Ito[5]
Motomichi Kosuga[1], Torayuki Okuyama[1]
[1]National Center for Child Health and Development, Department of Clinical Laboratory Medicine
[2]National Center for Child Health and Development, Division of Neurology
[3]International University of Health and Welfare, Atami Hospital, Department of Pediatrics
[4]Department of Reproductive, Development and Aging Sciences, the Graduate School of Medicine, the University of Tokyo
[5]National Center for Child Health and Development, Department of General Pediatrics

### P-03　Elevation of LysoGb3 and its analogues in Fabry disease

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

### P-04　Two Male Fabry Patients treated by ERT for 13years －Clinical Efficacy and Skin Biopsied Findings－

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

### P-05　A Case of Hunter Syndrome Diagnosed by Exome Sequencing in Infancy

○ Ji-Eun Lee[1], Ari Song[2], Minji Im[2], Jin Sung Lee[3], Dong-Kyu Jin[2], Sung Yoon Cho[2]
[1]Department of Pediatrics, Inha University Hospital, Inha University School of Medicine
[2]Department of Pediatrics, Samsung Medical Center, Sungkyunkwan University School of Medicine, Seoul, Republic of Korea
[3]Department of Clinical Genetics, Yonsei University School of Medicine, Seoul, Republic of Korea

## 18：30〜18：50　Poster 2：Lysosomal Disorders・Peroxisomal Disorders 2（Poster Venue 1：4F Room 1）

**Chairperson: Hiroshi Mitsubuchi**

（Department of Neonatology, Kumamoto University Hospital）

### P-06　Pilot study of newborn screening for mucopolysaccharidosis type 1, 2 and Diagnosis case report

○ Shinichiro Yoshida[1,2], Ken Momosaki[1], Kousuke Kumeda[2], Fumio Endo[3], Kimitoshi Nakamura[1]
[1]Department of Pediatrics, Faculty of Life Sciences, Kumamoto University
[2]Newborn Screening Center, KM Biologics Co., Ltd.
[3]Kumamoto-Ezuko Medical Center for The Severely Disabled

### P-07　A Japanese boy with Morquio A syndrome treated by Elosulfase alfa since 2 years old: one-year report

○ Akari N.Uutsunomiya[1], Reiko Kagawa[1], Shuhei Karakawa[1], Chihiro Tani[2], Keiko Matsubara[2]
Toshio Nakamae[3], Takeshi Ishino[4], Hiroaki Kimura[5], Go Tajima[6], Masao Kobayashi[1]
[1]Department of Pediatrics, Hiroshima University Hospial
[2]Department of Diagnostic Radiology, Hiroshima University Hospital
[3]Department of Orthopaedic Surgery, Hiroshima University Hospital
[4]Department of Otolaryngology, Hiroshima University Hospital
[5]Department of Rehabilitation, Hiroshima University Hospital
[6]Division of Neonatal Screening, Research Institute, National Center for Child Health and Development

### P-08　Glycosaminoglycans analysis in blood and urine of mucopolysaccharidoses patients

○ Shunji Tomatsu[1], Shaukat Kahn[2]
[1]Nemours/Alfred I. duPont Hospital for Children
[2]Biomedical, Nemours/Alfred I. duPont Hospital for Children, United States

### P-09　Substrate degradation enzyme therapy（SDET）for mucopolysaccharidosis

○ Shunji Tomatsu[1], Kazuki Sawamoto[2], Tokiko Sakai[3], Ikue Kitazawa[3], Hideyuki Futatsumori[3]
[1]Nemours/Alfred I. duPont Hospital for Children
[2]Biomedical, Nemours/Alfred I. duPont Hospital for Children, United States
[3]Seikagaku Co.

## 18：55〜19：15　Poster 3：Lysosomal Disorders・Peroxisomal Disorders 3（Poster Venue 1：4F Room 1）

**Chairperson: Kenji Ihara**

（Department of Pediatrics, Oita University Faculty of Medicine）

### P-10　Novel Surgical Reconstruction Rescues Life-threatening Severe Tracheal Obstruction in Mucopolysaccharidosis IVA

○ Shunji Tomatsu, Christian Pizarro, Lauren Averill, Mary Theroux, William Mackenzie
Nemours/Alfred I. duPont Hospital for Children

### P-11 Analysis of interaction between lysosomal enzymes and mannose 6-phosphate receptor by SPR

○ Minori Kanzaki[1], Yurie Namai[1], Yasunori Chiba[2], Masatoki Katayama[1], Takahiro Tsukimura[1]
Tadayasu Togawa[1], Hitoshi Sakuraba[3]
[1]Department of Functional Bioanalysis, Meiji Pharmaceutical University
[2]Department of Life Science and Biotechnology, National Institute of Advanced Industrial Science and Technology
[3]Department of Clinical Genetics, Meiji Pharmaceutical University

### P-12 LC-MS analysis of Glycosphingolipids containing 2-hydroxy fatty acid with a chiral column

○ Yuko Fujiwara, Kotaro Hama, Kazuaki Yokoyama
Faculty of Pharmaceutical Sciences, Teikyo University

### P-13 Clinical course of 7 year old girl with juvenile/adult type galactosialidosis

○ Ryosuke Bo[1], Hiroyuki Awano[1], Masashi Nagai[1], Kazumi Tomioka[1], Tsukasa Tanaka[1]
Masahiro Nishiyama[1], Hiroaki Nagase[1], Aya Narita[2], Kazumoto Iijima[1]
[1]Departmen of Pediatrics, Kobe University Graduate School of Medicine
[2]Division of Child Neurology, Institute of Neurological Sciences, Faculty of Medicine, Tottori University

## 18：55〜19：15 Poster 4：Lysosomal Disorders・Peroxisomal Disorder 4 （Poster Venue 1：4F Room 1）

**Chairperson: Tomoko Lee**
（Hyogo College Of Medicine）

### P-14 Four cases of Mucolipidosis II/III with neurological symptoms due to cervical cord compression

○ Sachiko Nakaoka[1,2], Hidehito Kondo[1,3], Toko Shibuya[1], Keiko Matsuoka[4], Norio Sakai[5]
Keiichi Ohzono[1]
[1]Department of Pediatrics, Osaka University Graduate School of Medicine
[2]Department of Pediatrics, Kouseiren Takaoka Hospital
[3]Department of Pediatrics, Japanese Red Cross Kyoto Daiichi Hospital
[4]Department of Pathology, Osaka Women's and Children's Hospital
[5]Division of health science, Osaka University Graduate School of Medicine

### P-15 A case of Metachromatic leukodystrophy complicated with Gall bladder cancer

○ Kiri Oguro[1], Takahiro Ikeda[1], Ayafumi Ozaki[1], Daisuke Tamura[1], Kazuhiro Muramatsu[1]
Hitoshi Osaka[1], Shigeru Ono[2], Kaori Adachi[3], Eiji Nanba[4]
[1]Department of pediatrics, Jichi medical university
[2]Department of pediatric surgery, Jichi medical university
[3]Research Initiative Center, Organization for Research Initiative and Promotion, Tottori University
[4]Research Strategy Division, Organization for Research Initiative and Promotion, Tottori University

### P-16 Clinical efficacy of medium chain triglyceride and tranexamic acid in ARC syndrome

○ Yasutsugu Chinen[1], Sadao Nakamura[1], Hideki Goya[1], Noriko Nakayama[1], Tomohide Yoshida[1]
Kumiko Yanagi[2], Tadashi Kaname[2], Koichi Nakanishi[1]
[1]Department of Child Health and Welfare（Pediatrics）, Graduate School of Medicine, University of the Ryukyus
[2]Department of Genome Medicine, National Center for Child Health and Development

### P-17 Therapeutic effect by bezafibrate, using fibroblasts from patients with peroxisomal diseases

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

## 18：30〜18：55 Poster 5：Others 1（Poster Venue 2 : 4F Room 2）

**Chairperson: Takanobu Otomo**
（Kawasaki Medical School）

### P-18 Current status of Japan Registration System for Metabolic & Inherited Diseases

○ Joohyun Seo[1,2], Makiko Miyairi[1,2], Akira Ohtake[2,3], Torayuki Okuyama[1,2,4]
[1]Clinical Laboratory Medicine, National Center for Child Health and Development
[2]Committee for Patient Registration, Japanese Society for Inherited Metabolic Diseases
[3]Department of Pediatrics, Saitama Medical University
[4]Center for Lysosomal Storage Diseases, National Center for Child Health and Development

### P-19 What patients and the families living with lysosomal storage disease feel about newborn screening

○ Yoko Lee, Nozomi Hadano, Norio Sakai
Osaka University Graduate School of Medicine, Division of Health Sciences

### P-20 Maturation of immune response is associated with improved safety and efficacy of pegvaliase

○ Soumi Gupta, Kelly Lau, Joy Olbertz, Orli Rosen, Karen Gu, Mingjin Li, Kevin Larimore
Haoling H. Weng, Stephen Zoog, Becky Schweighardt
BioMarin Pharmaceutical Inc.

### P-21 Assay of pyridoxal phosphate and related metabolites in hypophosphatasia

○ Tomoyuki Akiyama[1], Takuo Kubota[2], Keiichi Ozono[2], Toshimi Michigami[3], Daisuke Kobayashi[4]
Katsuhiro Kobayashi[1]
[1]Department of Child Neurology, Okayama University
[2]Department of Pediatrics, Osaka University
[3]Department of Bone and Mineral Research, Osaka Women's and Children's Hospital
[4]Department of Food and Chemical Toxicology, Health Sciences University of Hokkaido

### P-22 A case genetically diagnosed Cohen syndrome with a specific face, sociable character and neutropenia

○ Toko Shibuya[1], Hidehito Kondo[2], Yusuke Hamada[3], Keiichi Ohzono[1], Norio Sakai[4]
[1]Department of pediatrics, Osaka University Graduate School of Medicine
[2]Department of Pediatrics, Japanese Red Cross Kyoto Daiichi Hospital
[3]Department of Pediatrics, Toyonaka Municipal Hospital
[4]Division of health science, Osaka University Graduate School of Medicine

## 18：30～18：55　Poster 6：Others 2（Poster Venue 2 : 4F Room 2）

**Chairperson: Ayako Matsunaga-Fujinami**

（Center for Medical Genetics, Department of Metabolism, Chiba Children's Hospital）

### P-23　COASY mutations associated with brain iron accumulation in a Chinese boy

○ Lulu Kang, Yi Liu, Ying Jin, Mengqiu Li, Jinqing Song, Yanling Yang
Department of Pediatrics, Peking University First Hospital

### P-24　Six Chinese patients with spondyloepiphyseal dysplasia congenita and two novel mutations in COL2A1 gene

○ Lulu Kang, Yi Liu, Ying Jin, Mengqiu Li, Jinqing Song, Yanling Yang
Department of Pediatrics, Peking University First Hospital

### P-25　Two Chinese boys with SBBYSS caused by *de novo* mutation in *KAT6B* gene

○ Dongxiao Li[1], Nan Lv[1], Caiyun Ma[1], Jingjie Li[1], Hong Zheng[2], Yanling Yan[3], Qing Shang[1]
[1]Children's Hospital Affiliated to Zhengzhou University
[2]The First Hospital of Henan University of Traditional Chinese Medicine
[3]Peking University First Hospital

### P-26　Clinical and genetic characteristics of congenital myasthenia syndrome with episodic apnea caused by CHAT gene mutation in China

○ Zhimei Liu[1], Fang Fang[1], Changhong Ding[1], Weihua Zhang[1], Jie Deng[1], Jun Liu[2], Zheng Li[2]
Suyun Qian[2]
[1]Department of Neurology, Beijing Children's Hospital, Capital Medical University, National Center for Children's Health
[2]Department of PICU, Beijing Children's Hospital, Capital Medical University, National Center for Children's Health, Beijing, China

### P-27　Misdiagnosis of three boys in one family with Xp21 contiguous gene deletion syndrome

○ Lu Mei[1], Yang Yan ling[2]
[1]Xiamen Maternal and Chilid Care Hospital
[2]Peking University First Hospital, China

## 18：55～19：25　Poster 7：Disorders of Carbohydrate or Nucleotide Metabolisms（Poster Venue 2 : 4F Room 2）

**Chairperson: Tokiko Fukuda**

（Pediatrics, Hamamatsu University School of Medicine）

### P-28　The clinical outcome of galactosemic patients detected by neonatal massscreening

○ Sayaka Ajihara[1], Masato Arao[1], Ikuma Musha[1], Toru Kikuchi[1], Akira Ohtake[1], Hiroshi Mochizuki[2]
Daisuke Matsushita[2]
[1]The pediatrics, Saitama medical university
[2]Saitama childrens medical center

### P-29　Clinical presentation in adult with Glycogen storage diseases

○ P Khemthong, A Tunyatheerathum, D Dejsuphong, M Busabaratana, T Sura
Medical Genetics and Molecular Medicine, Department of Medicine, Ramathibodi Hospital, Thailand

### P-30 Clinical and genetic analysis in three Chinese patients with congenital disorder of glycosylation

○ Changhong Ren[1], Fang Fang[1], Changhong Ding[1], Lifang Dai[1], Hua Cheng[2]
[1]Department of Neurology, Beijing Children's Hospital, Capital Medical University, National Center for Children's Health, Beijing, China
[2]Department of Radiology, Beijing Children's Hospital, Capital Medical University, National Center for Children's Health, Beijing, China

### P-31 A child with liver glycogenosis has psychomotor retardation due to chromosome 14q32 duplication

○ Mei Ikenori[1,3], Mitsuru Kubota[1], Masayo Kagami[2], Akira Ishiguro[3], Yoh Umeda[4]
[1]Department of General Pediatrics & interdisciplinary Medicine, National Center for Child Health and Development
[2]Department of Molecular Endocrinology, National Research Institute for Child Health and Development
[3]Center for Postgraduate Education and Training, National Center for Child Health and Development
[4]Child Medical Center, Showa University Northern Yokohama Hospital

### P-32 Dihydropyrimidinase deficiency in four East Asian patients due to novel DPYS mutations

○ Yoko Nakajima[1], Judith Meijer[2], Doreen Dobritzsch[3], Chunhua Zhang[4], Xu Wang[5]
Yoriko Watanabe[6], Tetsuya Ito[1], Andre Van Kuilenburg[2]
[1]Fujita Health University School of Medicine, Department of Pediatrics
[2]Academic Medical Center, Department of Clinical Chemistry, Laboratory Genetic Metabolic Diseases, Amsterdam, The Netherlands
[3]Uppsala University, Department of Chemistry, Biomedical Center, Uppsala, Sweden
[4]MILS International, Department of Research and Development
[5]Beijing Children Hospital, Beijing Affiliated to Capital University of Medical Sciences, Department of Neurology, Beijing, China
[6]Kurume University, School of Medicine, Department of Pediatrics

### P-33 Development of HPRT activity assay method using dried blood spot and UPLC-MS/MS

○ Reika Miyake[1], Yasuhiro Maeda[1], Yoko Nakajima[2], Tetsuya Ito[2], Kana Gotoh[1], Yuji Hotta[1]
Tomoya Kataoka[3], Kazunori Kimura[1,3]
[1]Graduate School of Phermaceutical Sciences, Nagoya City University
[2]Department of Pediatrics, Fujita Health University
[3]Graduate School of Medical Sciences, Nagoya City University

## 18：30〜18：55 Poster 8：Organic Acidurias and Fatty Acid Oxidation Defects 1 （Poster Venue 3 : 5F Room 1）

**Chairperson: Yoriko Watanabe**

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

### P-34 Urinary hydrogen sulfide concentration in the patients with methylmalonic aciduria

○ Jinqing Song, Yi Liu, Lulu Kang, Ying Jin, Mengqiu Li, Junbao Du, Yanling Yang
Department of Pediatrics, Peking University First Hospital

### P-35 Clinical, biochemical and genetic studies of Chinese patients with cobalamin G or J defect missed by current tandem mass spectrometry

○ Yi Liu[1], Lulu Kang[1], Dongxiao Li[2], Ying Jin[1], Jinqing Song[1], Haixia Li[3], Yanling Yang[1]
[1]Peking University First Hospital
[2]Laboratory for genetic metabolic diseases, Henan Children's Hospital
[3]Department of Clinical Laboratory, Peking University First Hospital

### P-36 Cobalamin C Deficiency: a case report of 16-year-old boy with mild phenotype

○ Yukie Kinoshita[1], Yoriko Watanabe[1,2], Kyoko Tashiro[1], Hiromi Shimizu[1], Misa Inaba[1]
Yuri Misawa[1], Sanae Tashima[1], Kaori Fukui[2], Osamu Sakamoto[3], Naohisa Uchimura[1,4]
[1]Research Institute of Medical Mass Spectrometry, Kurume University School of Medicine
[2]Department of Pediatrics and Child Health, Kurume University School of Medicine
[3]Department of Pediatrics, Tohoku University School of Medicine
[4]Department of Neuropsychiatry, Kurume University School of Medicine

### P-37 A case of beta-ketothiolase deficiency required continuous hemodialysis for ketoacidotic crisis

○ Masaki Minami[1], Mariko Sawada[1], Tomohiro Hayashi[1], Masumi Saito[2], Shinichi Watabe[1]
Kenji Waki[1], Junko Matsuda[3], Go Tajima[4], Miori Yuasa[5], Yoshio Aragaki[1]
[1]Department of Pediatrics, Kurashiki Central Hospital
[2]Department of Clinical Engineering
[3]Department of Pathophysiology and Metabolism, Kawasaki Medical School
[4]Division of Neonatal Screening, National Center for Child Health and Development
[5]Department of Pediatrics, Fukui University School of Medical Science

### P-38 Inborn Metabolic Errors presenting with Ketoacidosis: A Case Series from an Egyptian Medical Center

○ Elsayed Abdelkreem, Rofida Magdy, Abdelrahim Sadek, Mohamed Abdelaal
Department of Pediatrics, Faculty of Medicine, Sohag University

## 18：30〜19：00 Poster 9：Organic Acidurias and Fatty Acid Oxidation Defects 2 （Poster Venue 3：5F Room 1）

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

### P-39 Rhabdomyolysis as a diagnostic cue in sibilings with CPT2 deficiency

○ Tomoko Asada[1], Maiko Utoyama[1], Misayo Matsuyama[1], Hiroshi Moritake[1], Hirotake Sawada[2]
Keiichi Hara[3], Go Tajima[4]
[1]Division of Pediatrics, Department of Reproductive and Developmental Medicine Faculty of Medicine, University of Miyazaki
[2]School of nursing, Faculty of Medicine, University of Miyazaki
[3]Department of pediatrics, National Hospital Organization Kure Medical Center and Chugoku Cancer Center
[4]Division of Neonatal Screening, Reserch Institute, National Center for Child Health and Development

### P-40 Carnitine palmitoyltransferase 2 deficiency, brothers case of infants type and pubertal type

○ Megumi Hashimoto[1], Kazuteru Kitsuda[1], Shigeyuki Ohtsu[2], Hideaki Senzaki[1], Keiichi Hara[3]
Go Tajima[4]
[1]Department of Pediatrics, Kitasato University School of Medicine
[2]Nakanoshima Diabetes Clinic
[3]National Hospital Organization, Kure Medical Center and Chugoku Cancer Center
[4]National Center for Child Health and Development, Division of Neonatal Screening

### P-41 the first case of CPT2 deficiency in Hokkaido detected by new cut off marker

○ Toju Tanaka[1], Miwa Yoshinaga[2], Michiko Tezuka[2], Takao Ishikawa[2], Shosuke Nomachi[2]
Masayoshi Nagao[1]
[1]Center for Genetics & Metabolic Disorder and Clinical Research, National Hospital Organization Hokkaido Medical Center
[2]Sapporo City Institute of Public Health

### P-42 One year old girl with status epilepticus due to carnitine related hypoglycemia

○ Tomo Sawada[1], Yui Kakishita[1], Sinchul Jwa[2], Takahiro Fujii[1], Kotaro Yazaki[1], Kenji Bando[1]
Miori Yuasa[3], Yosuke Shigematsu[3], Seiko Murakami[1]
[1]Department of Pediatrics, IZUMI CITY GENERAL HOSPITAL
[2]Department of Pediatrics, Osaka City Univercity Graduate School of Medicine
[3]Department of Pediatrics, Faculty of Medical Sciences, University of Fukui

### P-43 Four infants of sudden death caused by very long chain acyl-CoA dehydrogenase deficiency

○ Xu Bei[1], Liu Yi[2], Kang Lulu[2], Jin Ying[2], Song Jinqing[2], Yang Yanling[2]
[1]Department of Pediatrics, Baoding First Central Hospital
[2]Department of Pediatrics, Peking University First Hospital

### P-44 The efficacy of Bezafibrate for patients with TFP deficiency follow-up report

○ Tomonori Suyama, Keiko Ichimoto, Takuya Fushimi, Naomi Kuranobu, Masaru Shimura
Makiko Tajika, Ayako Matsunaga, Kei Murayama
Center for Medical Genetics, Department of Metabolism, Chiba Children's Hospital

## 18：55〜19：20 Poster 10：Organic Acidurias and Fatty Acid Oxidation Defects 3 (Poster Venue 3 : 5F Room 1)

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

### P-45 Safety and efficacy of ketogenic diet in a patient with pyruvate dehydrogenase complex deficiency

○ Mahoko Furujo[1], Masako Kinoshita[2], Hiroki Tsuchiya[1], Takushi Inoue[1], Toshihide Kubo[1]
[1]Department of Pediatrics, Okayama Medical Center, National Hospital Organization
[2]Department of Neurology, Utano National Hospital, National Hospital Organization

### P-46 Five novel mutations on MLYCD gene in four patients with malonic aciduria

○ Lulu Kang, Yi Liu, Ying Jin, Mengqiu Li, Jinqing Song, Yanling Yang
Department of Pediatrics, Peking University First Hospital

### P-47 Serum acylcarnitine and urinary organic acids analysis of Glutaric acidaemia type 2 in pregnancy

○ Tomoya Takahashi[1], Kougorou Iwanaga[1], Naoko Nishimura[1], Masahiko Kawai[1]
Hironori Kobayashi[2]
[1]Kyoto University Hospital
[2]Department of Pediatrics Shimane University School of Medicine

### P-48 A case of infantile glutaric acidemia type II detected by newborn screening

○ Fusa Nagamatsu[1], Kentaro Sawano[1], Nobuyuki Ishige[2], Yukihiro Hasegawa[1], Akira Ohtake[1,3]
[1]Division of Endocrinology and Metabolism, Tokyo Metropolitan Children's Medical Center
[2]Division of Newborn Screening, Tokyo Health Service Association
[3]Department of Pediatrics, Saitama Medical University

### P-49 Seven novel mutations in four Chinese patients with MEGDEL syndrome

○ Dongxiao Li[1], Yi Liu[2], Hui Dong[2], Jinqing Song[2], Yupeng Liu[2], Yao Zhang[2], Ying Jin[2]
Yanling Yang[2]
[1]Children's Hospital Affiliated to Zhengzhou University
[2]Peking University First Hospital

## 18：55〜19：20 Poster 11：Disorders of Amino Acid Metabolism 1 (Poster Venue 3 : 5F Room 1)

**Chairperson: Yoshiyuki Okano**
(Okano Children's Clinic)

### P-50 The first Mongolian cases of phenylketonuria in selective screening of inborn errors of metabolism

○ Purevsuren Jamiyan[1], Baasandai Bolormaa[1], Chogdon Narantsetseg[1], Renchindorj Batsolongo[1]
Ochirbat Enkhchimeg[1], Munkhuu Bayalag[1], Yuki Hasegawa[2], Haruo Shintaku[3], Seiji Yamaguchi[2]
Shonkhuuz Enkhtur[1]
[1]National Center for Maternal and Child Health
[2]Department of Pediatrics, Shimane University School of Medicine
[3]Department of Pediatrics, Osaka City University Graduate School of Medicine

### P-51 Introducing dietary treatment to a $BH_4$ responsive HPA pre-adolescent suboptimally controlled by $BH_4$

○ Erika Ogawa, Mika Ishige, Chika Takano, Tatsuo Fuchigami, Ichiro Morioka
Department of Pediatrics and Child Health, Nihon University School of Medicine

### P-52 Development of the Japanese version of PKU-QOL: first report of translation

○ Keiko Yamaguchi[1], Rie Wakimizu[2], Mitsuru Kubota[3]
[1]Department of Nursing Science, Graduate School of Comprehensive Human Sciences, University of Tsukuba
[2]Faculty of Medicine, University of Tsukuba
[3]National Center for Child Health and Development

### P-53 Outcome of Continuous Veno-Venous Hemofiltration (CVVH) on acute crisis of Classic Maple Syrup Urine Disease (MSUD) in Vietnam

○ Khanh Ngoc Nguyen, Dung Chi Vu, Ha Thi Nguyen, Nam Huu Dao, Mai Thi Chi Tran
National Children's Hospital, Hanoi, Vietham

### P-54 Bradycardia and sudden cardiac arrest in a case of aromatic L-amino acid decarboxylase deficiency

○ Lu Mei[1], Yang Yan Ling[2]
[1]Xiamen Maternal and Chilid Care Hospital, Fujian, China
[2]Peking University First Hospital, Beijing, China

## 18：30〜18：55 Poster 12：Disorders of Amino Acid Metabolism 2 (Poster Venue 3 : 5F Room 1)

**Chairperson: Shiro Matsumoto**

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

### P-55 Early liver transplantation in neonatal-onset and moderate UCDs may lead to normal neurodevelopment

○ Jun Kido[1,2], Shirou Matsumoto[1,2], Hiroshi Mitsubuchi[2], Fumio Endo[1,3], Kimitoshi Nakamura[1,2]
[1]Department of Pediatrics, Graduate School of Medical Sciences, Kumamoto University
[2]Department of Pediatrics, Kumamoto University Hospital
[3]Kumamoto Eduko Care Mediacal Center

### P-56 A case of heterozygous female diagnosed after the birth of male baby with OTC deficiency

○ Megumi Saito[1], Nana Akiyama[2], Tomoko Tsuruoka[2,3], Takuya Fushimi[2,4], Kaori Sassa[5]
Ikuma Musha[5], Masato Arao[5], Mureo Kasahara[6], Kei Murayama[2,4], Akira Ohtake[1,5]
[1]Center for Intractable Diseases, Saitama Medical University Hospital
[2]Center for Medical Genetics, Chiba Children's Hospital
[3]Department of Neonatology, Chiba Children's Hospital
[4]Department of Metabolism, Chiba Children's Hospital
[5]Department of pediatrics, Saitama Medical University
[6]Organ Transplantation Center, National Center for Child Health and Development

### P-57 Clinical features of Arginase 1 Deficiency: A Review of Global Literature

○ James Edward Wooldridge[1], George Alfred Diaz[2], Nicola Longo[3], Andreas Schulze[4], Gillian Bubb[5]
Stephen Eckert[5], Kiran Chandrakant Patki[5], John Lawrence Merritt[6]
[1]Aeglea BioTherapeutics
[2]Icahn School of Medicine at Mt. Sinai, New York City, USA
[3]University of Utah, Salt Lake City, USA
[4]University of Toronto and The Hospital for Sick Children, Toronto, Canada
[5]Aeglea BioTherapeutics, Austin, USA
[6]University of Washington, Seattle, USA

### P-58 CITRULLINEMIA- EXPERIENCE FROM FIVE PEADIATRIC PATIENTS

○ Uyen Thi Tu Vu, Cuong duy Le, Dung Chi Vu, Khanh Ngoc Nguyen, Mai Thi Chi Tran
Vietnam National's Children Hospital

### P-59 Two cases of patients with ID due to IEM presented with adjustment disorder in young adulthood

○ Mayumi Urashima[1,2], Kaori Fukui[2], Yoriko Watanabe[2,3]
[1]Pediatrics, Saga-Ken Medical Centre Koseikan
[2]Department of Pediatrics and Child Health, Kurume University School of Medicine
[3]Research Institute of Medical Mass Spectrometry, Kurume University School of Medicine

## 18：30〜18：55　Poster 13：Mitochondrial Disorders 1
## （Poster Venue 3 : 5F Room 1）

**Chairperson: Takuya Fushimi**

（Department of Metabolism, Chiba Children's Hospital）

### P-60　Clinical and genetic analysis of progressive cavitating leukoencephalopathy in China

○ Changhong Ren[1], Fang Fang[1], Changhong Ding[1], Lifang Dai[1], Hua Cheng[2], Zhimei Liu[1]
Kei Murayama[3]

[1]Department of Neurology, Beijing Children's Hospital, Capital Medical University, National Center for Children's Health, Beijing, China
[2]Department of Radiology, Beijing Children's Hospital, Capital Medical University, National Center for Children's Health, Beijing, China
[3]Department of Metabolism, Chiba Children's Hospital

### P-61　Phenotype and genotype features of twenty Chinese children with mitochondrial DNA depletion syndromes

○ Fang Fang, Lifang Dai, Zhimei Liu, Danmin Shen, Changhong Ding, Jiuwei Li, Xiaotun Ren
Husheng Wu

Department of Neurology, Beijing Children's Hospital, Capital Medical University, National Center for Children's Health, Beijing, China

### P-62　8 Cases of Rare Mitochondrial Diseases Caused by Mutations in the Genes Encoding Aminoacyl-tRNA Synthetases in China

○ Manting Xu, Fang Fang, Zhimei Liu, Lifang Dai, Danmin Shen, Xiaotun Ren, Hong Jin, Shen Zhang
Department of Neurology, Beijing Children's Hospital affiliated to Capital Medical University

### P-63　Improvement of cardiac function in a case of ACAD9 deficiency after administration of pyruvate

○ Kana Kitayama, Shinji Higuchi, Rie Kawakita, Tohru Yorifuji
Divison of Pediatric Endocrinology and Metabolism, Osaka City General Hospital

### P-64　A neonatal case with poor general condition associated with decreased OCR in fibroblasts

○ Taro Nagatomo
Ehime Prefectural Central Hospital

## 18：55〜19：20　Poster 14：Mitochondrial Disorders 2
## （Poster Venue 3 : 5F Room 1）

**Chairperson: Hiroshi Mochizuki**

（Saitama Children's Medical Center）

### P-65　Genotype, phenotype and Follow-up Study of 80 Children with Leigh Syndrome in China

○ Rui Ban, Fang Fang, Zhimei Liu, Lifang Dai, Jiuwei Li, Shuai Gong, Xinying Yang
Weihua Zhang

Beijing Children's Hospital affiliated to Capital Medical University

### P-66 Clinical and genetic characteristics of 12 cases of Leigh syndrome induced by PDHA1 gene mutation

○ Tianyu Song[1], Danmin Shen[1], Manting Xu[1], Lifang Dai[1], Masaru Shimura[2], Weihua Zhang[1]
Jiuwei Li[1], Na Li[3], Kei Murayama[1], Fang Fang[1]
[1]Department of Neurology, Beijing Children's Hospital, Capital Medical University, National Center for Children's Health, Beijing, China
[2]Department of Metabolism, Chiba Children's Hospital, Chiba, Japan
[3]SinoPath, Beijing, China

### P-67 Genotype and phenotype analysis of Leigh syndrome involving valine metabolism in 8 Chinese children

○ Xiaodi Han, Fang Fang, Danmin Shen, Lifang Dai, Weihua Zhang, Jiuwei Li
Neurology Department, Beijing Children's Hospital, Capital Medical University, National Center for Children's Health, Beijing, China

### P-68 Treatable Leigh syndrome involving mitochondrial cofactor metabolism

○ Zhimei Liu[1], Fang Fang[1], Kei Murayama[2], Masaru Shimura[2], Danmin Shen[1], Weihua Zhang[1]
Jiuwei Li[1], Xiaotun Ren[1], Changhong Ding[1]
[1]Department of Neurology, Beijing Children's Hospital, Capital Medical University, National Center for Children's Health
[2]Department of Metabolism, Chiba Children's Hospital

### P-69 Case of infantile-onset Leigh syndrome with a 9185T>C mutation in the MTATP6 gene

○ Rei Takada[1,2], Takenori Tozawa[2], Hidehito Kondo[1], Zenro Kizaki[1], Chie Harasima[3], Kei Murayama[4]
Akira Otake[5], Tomohiro Chiyonobu[2]
[1]Department of Pediatrics, Japanese Red Cross Kyoto Daiichi Hospital
[2]Department of Pediatrics, Kyoto Prefectural University of Medicine
[3]Department of Pediatrics, The Japan Baptist Hospital
[4]Department of Metabolism, Chiba Children Hospital
[5]Department of Pediatrics, Saitama Medical University Hospital

## 18：55～19：25 Poster 15：Screening（Poster Venue 3 : 5F Room 1）

**Chairperson: Yuki Omura-Hasegawa**
**（Department of Pediatrics, Matsue Red Cross Hospital）**

### P-70 Liquid chromatography tandem-mass spectrometry as follow-up assessment for methylmalonic acidemia

○ Hiroyuki Iijima[1], Nobuyuki Ishige[2], Mitsuru Kubota[1]
[1]Department of General Pediatrics & interdisciplinary Medicine, National Center for Child Health and Development
[2]Division of newborn screening, Tokyo Health Service Association

### P-71 Isovaleric acidemia gene carrier found by investigation from neonatal mass screening

○ Yohei Sugiyama[1], Daisuke Sugawara[1], Go Tajima[2], Ryoji Fujiki[3], Osamu Ohara[3], Hideo Sasai[4]
Toshiyuki Fukao[4], Ko Ichihashi[1]
[1]Jichi Medical University Saitama Medical Center
[2]National Center for Child health and Development
[3]Kazusa DNA research institute
[4]Gifu University Graduate School of Medicine

## P-72 Twenty-three newborn died in inherited metabolic disorders diagnosed by post-mortem studies

○ Liu Xiaoyan[1], Wang Jiaxin[1], Kang Lulu[2], Liu Yi[2], Jin Ying[2], Song Jinqing[2], Li Mengqiu[2]
Zhang Yao[2], Yang Yanling[2]
[1]Department of Pediatrics, The Third Affiliated Hospital of Xinxiang Medical College
[2]Department of Pediatrics, Peking University First Hospital, China

## P-73 Low carnitine level in 1-month tandem mass screening with preterm and very low birth weight infants

○ Hiroki Otsuka[1,2], Yuya Mizuno[1], Yuki Miwa[1], Humiko Iwai[1], Hisashi Fukutomi[1]
Toshinari Kouyama[1], Masashi Kondo[1], Yutaka Yamamoto[1], Yoshinori Kohno[1], Toshiyuki Fukao[2]
[1]Department of Neonatology, Gifu Prefectural General Medical Center
[2]Department of Pediatrics, Graduate School of Medicine, Gifu University

## P-74 Clinical background of the cases with high C5DC level in newborn mass screening

○ Yuki Hasegawa, Kenji Yamada, Hironori Kobayashi, Seiji Yamaguchi, Takeshi Taketani
Shimane Univsersity School of Medicine

## P-75 A pilot study on newborn screening for congenital adrenal hyperplasia in Beijing

○ Yuanyuan Kong, Nan Yang, Jinqi Zhao, Haihe Yang, Gong Lifei
Department of Newborn Screening, Beijing Obstetrics and Gynecology Hospital, Capital Medical University