Program

Presidential Lecture

Presidential Lecture

Date ：Thursday, October 12, 2023　9:20 ～ 9:50　Room A（Cosmos, 3F, Toshi Center Hotel）
Chair ：Thanyachai Sura（Mahidol University, Thailand）

PR

Let us treasure and share our exceptions: Story on PDGFRB activating variants

Kenjiro Kosaki
Center for Medical Genetics, Keio University School of Medicine, Japan

Plenary Lecture

Plenary Lecture 1

Date ：Thursday, October 12, 2023　13:00 ～ 13:50　Room A（Cosmos, 3F, Toshi Center Hotel）
Chair ：Masayuki Yoshida（Tokyo Medical and Dental University, Japan）

PL1

The expanding role of genetics in the care of patients with rare disorders

Robert L. Nussbaum
Invitae Corporation, USA / University of California, San Francisco, USA

Plenary Lecture 2　Nobel Prize Laureate Lecture

Date ：Friday, October 13, 2023　13:10 ～ 14:00　Room A（Cosmos, 3F, Toshi Center Hotel）
Chair ：Katsushi Tokunaga（Genome Medical Science Project, National Center for Global Health and Medicine, Japan）

PL2

About Neandertals, Denisovans, and modern humans

Svante Pääbo
Max Planck Institute for Evolutionary Anthropology, Germany / Okinawa Institute of Science and Technology, Japan

Plenary Lecture 3

Date ：Saturday, October 14, 2023　9:40 ～ 10:20　Room A（Cosmos, 3F, Toshi Center Hotel）
Chair ：Johji Inazawa（Tokyo Medical and Dental University, Japan）

PL3

40 years of human genetics, genomics, genetic variations and precision medicine

Yusuke Nakamura
National Institutes of Biomedical Innovation, Health and Nutrition, Japan

Plenary Lecture 4

Date ：Saturday, October 14, 2023　10:20 ～ 11:00　Room A（Cosmos, 3F, Toshi Center Hotel）
Chair ：Kenjiro Kosaki（Center for Medical Genetics, Keio University School of Medicine, Japan）

PL4

Therapeutic gene editing for cardiovascular and metabolic diseases: From the laboratory bench to the clinic

Kiran Musunuru
Perelman School of Medicine at the University of Pennsylvania, USA

Plenary Lecture 5

Date ：Saturday, October 14, 2023　11:00 ～ 11:40　Room A（Cosmos, 3F, Toshi Center Hotel）
Chair ：Kenjiro Kosaki（Center for Medical Genetics, Keio University School of Medicine, Japan）

PL5

Innovative approach for implementation of Genomic Medicine in Health Systems

Borut Peterlin
Clinical Institute of Genomic Medicine, University Medical Center Ljubljana, Slovenia

Translating human genetics and genomics into the future: Foresights, hurdles and global co-operation

Translating human genetics and genomics into the future: Foresights, hurdles and global co-operation

Date ：Friday, October 13, 2023　14:00 ～ 16:00　Room A（Cosmos, 3F, Toshi Center Hotel）
Chairs ：Poh San Lai（National University of Singapore, Singapore）
Kenjiro Kosaki（Center for Medical Genetics, Keio University School of Medicine, Japan）

Following Professor Pääbo's presentation on human evolution and population genetics, the Global Forum will host a discussion on the future of human genomics. As we learn from ancient DNA, embrace current technological advances, and apply knowledge of human genetics to translational medicine, we also want to take a step forward to anticipate and prepare for a brave new future where genomic technologies and information are accessible to all people in all countries. What would we do then? What would we expect?

JS-1

Kym Boycott
University of Ottawa, Canada

JS-2

Brian H.Y. Chung
Department of Paediatrics and Adolescent Medicine, Li Ka Shing Faculty of Medicine, The University of Hong Kong, Hong Kong

JS-3

Chae Jong Hee
Seoul National University Hospital, Korea

JS-4

Kiran Musunuru
Perelman School of Medicine at the University of Pennsylvania, USA

JS-5

Kaori Muto
Department of Public Policy, HGC, IMSUT, Japan

JS-6

Robert L. Nussbaum
Invitae Corporation, USA

JS-7

Borut Peterlin
Clinical Institute of Genomic Medicine, University Medical Center Ljubljana, Slovenia

JS-8

Zornitza Stark

Australian Genomics, Australia

JS-9

Thanyachai Sura

Mahidol University, Thailand

JS-10

Xianjun Zhu

Sichuan Provincial People's Hospital, China

Symposium

Symposium 1　Therapeutics (DNA, mRNA, genome editing, others)

Date　：Thursday, October 12, 2023　10:10 ～ 11:40　Room A（Cosmos, 3F, Toshi Center Hotel）
Chairs　：Xianjun Zhu（Sichuan Provincial People's Hospital, China）
Tatsushi Toda（Graduate School of Medicine, The University of Tokyo, Japan）

SY1-1

Current status and future prospects of gene therapy for inherited diseases

Masafumi Onodera

National Center for Child Health and Development, Japan

SY1-2

Genome editing and iPS therapy for muscular dystrophies

Akitsu Hotta

Center for iPS Cell Research and Application, Kyoto University, Japan

SY1-3

Precise Gene Editing in Rare Diseases

Sangsu Bae

Seoul National University College of Medicine, Korea

SY1-4

Gene therapy for adult neuromuscular diseases

Tatsushi Toda

Graduate School of Medicine, The University of Tokyo, Japan

Symposium 2　Cancer Genomics, Germline

Date　：Thursday, October 12, 2023　10:10 ～ 11:40　Room B（Orion, 5F, Toshi Center Hotel）
Chairs　：Joanne Ngeow（National Cancer Centre, Singapore / Lee Kong Chian School of Medicine, Nanyang Technological University, Singapore / Cancer Genetics Service, National Cancer Centre, Singapore）
Motohiro Kato（The University of Tokyo, Japan）

SY2-1

Issues related to germline predisposition in adult/AYA patients with haematological disorders

Mizuki Watanabe[1,2]

1　Kyoto University Hospital, Japan
2　National Cancer Center Hospital of Japan, Japan

SY2-2 **Exploring hereditary cancer syndromes in the era of cancer genomic medicine**

Makoto Hirata

National Cancer Center Hospital, Japan

SY2-3 **Cancer Predisposition in Singapore: Insights from the SG10K study**

Joanne Ngeow[1,2,3]

1 National Cancer Centre, Singapore
2 Lee Kong Chian School of Medicine, Nanyang Technological University, Singapore
3 Cancer Genetics Service, National Cancer Centre, Singapore

SY2-4 **Predisposition to secondary cancer**

Motohiro Kato

The University of Tokyo, Japan

Symposium 3 Newborn Screening

Date ：Thursday, October 12, 2023 10:10 ～ 11:40 Room C（606, 6F, Toshi Center Hotel）
Chairs ：Duangrurdee Wattanasirichaigoon（Faculty of Medicine Ramathibodi Hospital, Mahidol University, Thailand）
Torayuki Okuyama（Saitama Medical University, Japan）

SY3-1 **Optional newborn screening for lysosomal storage diseases**

Torayuki Okuyama

Saitama Medical University, Japan

SY3-2 **Newborn screening for primary immunodeficiency in Japan**

Kohsuke Imai

National Defense Medical College, Japan

SY3-3 **Maple syrup urine disease screening in the Philippines**

Catherine Lynn T. Silao

Institute of Human Genetics, National Institutes of Health; College of Medicine and Philippine General Hospital, University of the Philippines Manila, Philippines

SY3-4 **Newborn screening and early detection of inborn errors of metabolism: Lessons from Taiwan**

Yin-Hsiu Chien

Department of Medical Genetics, National Taiwan University Hospital, Taiwan

Symposium 4 COVID-19 Susceptibility

Date ：Thursday, October 12, 2023 10:10 ～ 11:40 Room D（701, 7F, Toshi Center Hotel）
Chairs ：Namkoong Ho（Department of Infectious Diseases, Keio University School of Medicine, Japan）
Yukinori Okada（Graduate School of Medicine, The University of Tokyo, Japan / Osaka University Graduate School of Medicine, Japan / RIKEN Center for Integrative Medical Sciences, Japan）

SY4-1 **Genome analysis of the virus in sewage shows the actual situation of infection epidemic of SARS-CoV-2 strains**

Seiya Imoto

The Institute of Medical Science, The University of Tokyo, Japan

SY4-2

Trans-omics analysis for COVID-19 biology

Yukinori Okada[1,2,3]

1 Graduate School of Medicine, The University of Tokyo, Japan
2 Osaka University Graduate School of Medicine, Japan
3 RIKEN Center for Integrative Medical Sciences, Japan

SY4-3

Severe COVID-19 associate with impaired host immunity

Satoshi Okada

Hiroshima University Graduate School of Biomedical and Health Sciences, Japan

SY4-4

Elucidating the mechanism of COVID-19 severity by integrative analysis of single-cell and host genetics data

Ryuya Edahiro

Osaka University, Japan

Symposium 5　Pediatric Genetics / Congenital Malformation

Date　：Thursday, October 12, 2023　16:00 ～ 17:30　Room A（Cosmos, 3F, Toshi Center Hotel）
Chairs　：Eva Maria Cutiongco-de la Paz（Institute of Human Genetics, National Institutes of Health, University of the Philippines, Philippines）
Yoko Aoki（Tohoku University School of Medicine, Japan）

SY5-1

Transforming neonatal and pediatric care through genomic medicine

Brian H.Y. Chung[1,2]

1 Department of Paediatrics and Adolescent Medicine, Li Ka Shing Faculty of Medicine, The University of Hong Kong, Hong Kong
2 Hong Kong Genome Institute, Hong Kong

SY5-2

Recent progress in RASopathies

Yoko Aoki, Taiki Abe, Tetsuya Niihori

Tohoku University School of Medicine, Japan

SY5-3

Discovery and delineation of EXOC6B-related spondyloepimetaphyseal dysplasia with joint laxity

Katta Girisha[1,3], Hitesh Shah[2], Gandham SriLakshmi Bhavani[1]

1 Department of Medical Genetics, Kasturba Medical College, Manipal, Manipal Academy of Higher Education, Manipal, India
2 Department of Pediatric Orthopedics, Kasturba Medical College, Manipal, Manipal Academy of Higher Education, Manipal, India
3 Department of Genetics, College of Medicine and Health Sciences, Sultan Qaboos University, Muscat, Oman

SY5-4

Goldfish morphological diversity: Elucidate the mechanisms of congenital anomalies using non-human models

Tetsuo Kon

University of Vienna, Austria

Symposium 6　Cancer Genomics, Somatic

Date　：Thursday, October 12, 2023　16:00 ～ 17:30　Room B（Orion, 5F, Toshi Center Hotel）
Chairs　：Young Seok Ju（Korea Advanced Institute of Science and Technology (KAIST), Korea / Genome Insight Inc., Korea）
Issei Imoto（Aichi Cancer Center Research Institute, Japan）

SY6-1

Polygenic germline effects on cancer somatic alterations

Shinichi Namba[1,2]

1　Osaka University Graduate School of Medicine, Japan
2　Graduate School of Medicine, The University of Tokyo, Japan

SY6-2

Mutational signatures and their associations with cancer etiology and phenotypes

Hidewaki Nakagawa

RIKEN Center for Integrative Medical Sciences, Japan

SY6-3

Muliti-omic analyses to detemine molecular characteristics of colorectal cancer

Hiromichi Ebi, Rui Yamaguchi

Aichi Cancer Center Research Institute, Japan

SY6-4

Widespread somatic L1 retrotransposition in normal colorectal epithelium

Young Seok Ju[1,2]

1　Korea Advanced Institute of Science and Technology (KAIST), Korea
2　Genome Insight Inc., Korea

Symposium 7　Aging and Diseases

Date　：Thursday, October 12, 2023　16:00 ～ 17:30　Room C（606, 6F, Toshi Center Hotel）
Chair　：Kouichi Ozaki（Medical Genome Center, Research Institute, National Center for Geriatrics and Gerontology, Japan）

SY7-1

Comparative genomics reveals that a gut ceramidase activity determines the rate of systemic aging

Tohru Ishitani

Osaka University, Japan

SY7-2

Genetics and epigenetics of exceptional longevity: Lessons from centenarians in Japan

Yasumichi Arai, Takashi Sasaki

Keio University School of Medicine, Japan

SY7-3

Susceptibility genes for dementia: Focusing on APOE

Akinori Miyashita, Norikazu Hara, Ai Obinata, Tamao Tsukie, Mai Hasegawa, Kensaku Kasuga, Takeshi Ikeuchi

Brain Research Institute, Niigata University, Japan

SY7-4

Genomic research on dementia in the Japanese population

Daichi Shigemizu[1,2,3], Kouichi Ozaki[1,2,3]

1　National Center for Geriatrics and Gerontology, Japan
2　Hiroshima University Graduate School of Biomedical and Health Sciences, Japan
3　RIKEN Center for Integrative Medical Sciences, Japan

Symposium 8　Hemoglobinpathies

Date　：Thursday, October 12, 2023　16:00 ～ 17:30　Room D（701, 7F, Toshi Center Hotel）
Chairs　：Zilfalil Bin Alwi（School of Medical Sciences, Universiti Sains Malaysia, Malaysia）
Hiroki Kurahashi（Division of Molecular Genetics, Center for Medical Science, Fujita Health University, Japan）

SY8-1

Empowering Asia through the Global Globin Network: Harnessing genomic advancements in hemoglobinopathies

Zilfalil Bin Alwi
School of Medical Sciences, Universiti Sains Malaysia, Malaysia

SY8-2

Hemoglobinopathies in Japan

Yukio Hattori
Saiseikai Yamaguchi General Hospital, Japan

SY8-3

Preimplantation genetic testing (PGT-M) for thalassemia

Canquan Zhou
Former, Reproductive Medicine Center, First Affiliated Hospital of Sun Yat-sen University, China

SY8-4

Roles of transcription factors and chromatin remodelers in regulation of erythroid gene expression

Xiang Guo[1,2,3], Ann Dean[3]
1　School of Medicine, University Electronic Science and Technology China
2　Department of Hematology, Sichuan Academy of Medical Science and Sichuan Provincial People's Hospital, Chengdu, 610072, China
3　Laboratory of Cellular and Developmental Biology, National Institute of Diabetes and Digestive and Kidney Diseases, National Institutes of Health, Bethesda, Maryland, USA

Symposium 9　Omics/ Sequencing Technologies

Date　：Friday, October 13, 2023　10:10 ～ 11:40　Room A（Cosmos, 3F, Toshi Center Hotel）
Chairs　：Hirotomo Saitsu（Department of Biochemistry, Hamamatsu University School of Medicine, Japan）
Akihiro Fujimoto（Department of Human Genetics, Graduate School of Medicine, The University of Tokyo, Japan）

SY9-1

Long read genomics dissecting genetic variants in rare diseases

Naomichi Matsumoto
Yokohama City University Graduate School of Medicine, Japan

SY9-2

Solving variants of unknown significance with deep learning

Kyle Kai-How Farh
Illumina, USA

SY9-3

Spatial analysis to reveal the cancer microenviroments

Yutaka Suzuki
Department of Computational Biology and Medical Sciences, The University of Tokyo, Japan

SY9-4

Dynamics of chromatin organization at enhancers mediated by CTCF and KMT2C/D during cell differentiation

Naoki Kubo[1], Bing Ren[2]
1　Medical Institute of Bioregulation, Kyushu University, Japan
2　Department of Cellular and Molecular Medicine, University of California San Diego School of Medicine, USA

Symposium 10　Prenatal Genetics

Date　：Friday, October 13, 2023　10:10 ～ 11:40　Room B（Orion, 5F, Toshi Center Hotel）
Chairs　：Chanane Wanapirak（Department of Obstetrics and Gynecology, Chiang Mai University, Thailand）
Kiyonori Miura（Department of Obstetrics and Gynecology, Nagasaki University Graduate School of Biomedical Sciences, Japan）

SY10-1

Fetal ultrasonography and genetics

Mayumi Tokunaka
Showa University, Japan

SY10-2

Cell-free fetal nucleic acids

Chen Ming
Changhua Christian Hospital, and National Chung Shing University, Taiwan

SY10-3

NIPT for Thalassemia

Chanane Wanapirak
Department of Obstetrics and Gynecology, Chiang Mai University, Thailand

SY10-4

Prenatal fetal exome sequencing in one medical center in Israel: The Promise and its application

Orit Reish[1,2]

1　Tel Aviv University, Israel
2　Medical Genetics Institute, Shamir Medical Center, Zerifin, Israel

Symposium 11　Non-coding Regulatory Elements / ncRNA / miRNA / other RNAs

Date　：Friday, October 13, 2023　10:10 ～ 11:40　Room C（606, 6F, Toshi Center Hotel）
Chairs　：Piero Carninci（RIKEN Center for Integrative Medical Science(IMS), Japan / Fondazione Human Technopole, Italy）
Masao Nagasaki（Medical Institute of Bioregulation, Kyushu University, Japan）

SY11-1

Understanding the complex patterns of DNA modification in cancer

Genta Nagae
Research Center for Advanced Science and Technology, The University of Tokyo, Japan

SY11-2

Inferring function of lncRNAs through interactome with chromatin

Piero Carninci[1,2], Hazuki Takahashi[1], Masaki Kato[1], Takeya Kasukawa[1], Chi Wai Yip[1], Chung Chau Hon[1], Miki Kojima[1], Jay Shin[1,3], Hiromi Sueki-Nishiyori[1], Harshita Sharma[1], Mitsuyoshi Murata[1], Sachi Kato[1], Aslihan Karabacak Calviello[1,2], Rodrigo Pracana[1,2], Laura Carpen[1,2], Beatrice Bodega[4], Valeria Ranzani[4], Magda Bienko[1,2,5], Wenjing Kang[1,5]

1　RIKEN Center for Integrative Medical Science(IMS), Japan
2　Fondazione Human Technopole, Italy
3　Agency for Science, Technology and Research (A*STAR), Singapore
4　Istituto Nazionale di Genetica Molecolare, Italy
5　Karolinska Institutet, Sweden

SY11-3

Mobile element variation contributes to population-specific genome diversification, gene regulation, and disease risk

Shohei Kojima, Nicholas Parrish
RIKEN IMS, Japan

SY11-4

Functional noncoding variants in 15q26.1 cause familial non-autoimmune thyroid abnormalities

Satoshi Narumi
Keio University, Japan

Symposium 12　Genomic Risk Assessment: Towards Preventive Medicine

Date　：Friday, October 13, 2023　10:10 ～ 11:40　Room D（701, 7F, Toshi Center Hotel）
Chairs　：Hie Lim Kim（Nanyang Technological University, Singapore）
Yukinori Okada（Graduate School of Medicine, The University of Tokyo, Japan / Osaka University Graduate School of Medicine, Japan / RIKEN Center for Integrative Medical Sciences, Japan）

SY12-1　Polygenic risk score for precision medicine of east Asians

Yukinori Okada[1,2,3]

1　Graduate School of Medicine, The University of Tokyo, Japan
2　Osaka University Graduate School of Medicine, Japan
3　RIKEN Center for Integrative Medical Sciences, Japan

SY12-2　Patient-centric stratified drug therapy based on pharmacogenomic testing

Taisei Mushiroda

RIKEN, Japan

SY12-3　The GenomeAsia 100K project: Unrevealing genetic diversity and history of Asian populations

Hie Lim Kim

Nanyang Technological University, Singapore

SY12-4　Can genetic variants be diagnostic discriminatory markers for schizophrenia and bipolar disorder?

Kazutaka Ohi

Gifu University Graduate School of Medicine, Japan

Symposium 13　Databases / Data sharing / Biobank

Date　：Friday, October 13, 2023　16:40 ～ 18:20　Room A（Cosmos, 3F, Toshi Center Hotel）
Chairs　：Zornitza Stark（Australian Genomics, Australia）
Soichi Ogishima（The Advanced Research Center for Innovations in Next-Generation Medicine(INGEM), Japan / Tohoku Medical Megabank Organization, Tohoku University, Japan）

SY13-1　Biobank information platform for R&D of genomic medicine in Japan

Soichi Ogishima[1,2]

1　The Advanced Research Center for Innovations in Next-Generation Medicine(INGEM), Japan
2　Tohoku Medical Megabank Organization, Tohoku University, Japan

SY13-2　Omics analysis and data sharing in Biobank Japan

Koichi Matsuda

The University of Tokyo, Japan

SY13-3　The GenomeAsia 100K project: Advancing human genetic studies in Asian populations

Hie Lim Kim

Nanyang Technological University, Singapore

SY13-4　Challenges, enablers and opportunities in rare disease research

Tiong Yang Tan[1,2,3], Michelle G. de Silva[1,2], Katrina Bell[2,3], Lyndon Gallacher[1,2,3], Rocio Rius[2,3], Kirsten Allan[1,2], Natasha J Brown[1,2,3], Natalie B Tan[1,2,3], Smitha Kumble[1,2,3], Russell Gear[1,2,3], Cas Simons[2,3,4], David Stroud[2,3], David Thorburn[1,2,3], John Christodoulou[1,2,3], Susan M White[1,2,3]

1　Victorian Clinical Genetics Services, Melbourne, Australia
2　Murdoch Children's Research Institute, Melbourne, Australia
3　Department of Paediatrics, University of Melbourne, Melbourne, Australia
4　Centre for Population Genomics, Murdoch Children's Research Institute, Melbourne, Australia and Garvan Institute, Sydney, Australia

SY13-5 **Human Genome Projects in Ethnically-Diverse Indonesian Populations**

Herawati Sudoyo, Pradiptajati Kusuma, Safarina G. Malik, Isabella Apriyana

Genome Diversity and Diseases Division, Mochtar Riady Institute for Nanotechnology, Indonesia

SY13-6 **Integrating whole genome sequencing into national health data warehouse**

Surakameth Mahasirimongkol

Office of Permanent Secretary, Ministry of Public Health, Thailand

SY13-7 **Databases of rare genetic diseases in Vietnam**

Vu Dung

Vietnam National Children's Hospital, Vietnam

SY13-8 **The FILIPINOme: The Filipino Genome Sequencing Project**

Eva Maria Cutiongco-de la Paz

Institute of Human Genetics, National Institutes of Health, Philippine Genome Center University of the Philippines, Philippines

Symposium 14 Comprehensive Management of Differences of Sex Development (DSD); Lessons from Genetics

Date ：Friday, October 13, 2023 16:40 ～ 18:10 Room B（Orion, 5F, Toshi Center Hotel）
Chairs ：Sultana Faradz（Faculty of Medicine, Diponegoro University and Universitas Yarsi, Indonesia / Faculty of Medicine, Diponegoro University and School of Post Graduate Program Universitas YARSI, Indonesia）
Tomonobu Hasegawa（Keio University School of Medicine, Japan）

SY14-1 **Molecular mechanism of sex development and genetic diagnosis of DSD**

Tomohiro Ishii

Keio University School of Medicine, Japan

SY14-2 **Collaborative study of multidisciplinary care for differences of sex development Patients**

Sultana Faradz[1,2], Katie Ayers[3], Stenvert Drop[4], Andrew Sinclair[3]

1 Faculty of Medicine, Diponegoro University and Universitas Yarsi, Indonesia
2 Faculty of Medicine, Diponegoro University and School of Post Graduate Program Universitas YARSI, Indonesia
3 Murdoch Children's Research Institute, Melbourne, Australia
4 Division of Endocrinology, Department of Pediatrics, Sophia Children's Hospital / Erasmus University Medical Centre Rotterdam, The Netherlands

SY14-3 **Individualized care for patients with difference of sex development; Multidisciplinary team approach**

Masanobu Kawai

Osaka Women's and Children's Hospital, Japan

SY14-4 **Current status of uterus transplantation**

Iori Kisu

Department of Obstetrics and Gynecology, Keio University School of Medicine, Japan

Symposium 15　Bioinformatics

Date　：Friday, October 13, 2023　16:40 ～ 18:10　Room C（606, 6F, Toshi Center Hotel）
Chairs　：Poh San Lai（National University of Singapore, Singapore）
Kazuhiko Nakabayashi（National Center for Child Health and Development (NCCHD), Japan）

SY15-1

MOVA: A method of missense variant pathogenicity using AlphaFold2

Tomohiko Ishihara, Yuya Hatano, Osamu Onodera
Department of Neurology, Brain Research Institute, Niigata University, Japan

SY15-2

Large-scale computational genomics: Tools for decoding genomic features

Ryuichiro Nakato
Laboratory of Computational Genomics, Institute for Quantitative Biosciences, The University of Tokyo, Japan

SY15-3

Population-specific reference genome and rapid WGS analyses for rare diseases

Jun Takayama
Tohoku University, Japan

SY15-4

ETCHING: Ultrafast prediction of somatic structural variations by filtering out reads matched to pan-genome k-mer sets

Jin-Wu Nam[1,2,3]
1　BIG Lab, South Korea
2　Department of Life Science, Hanyang University, Seoul, South Korea
3　HY Institute of Bioscience and Biotechnology, Hanyang University, Seoul, South Korea

Symposium 16　Genetics of Complex Diseases

Date　：Friday, October 13, 2023　16:40 ～ 18:10　Room D（701, 7F, Toshi Center Hotel）
Chairs　：Stephen Lam（The Hong Kong Sanatorium and Hospital, Hong Kong）
Yoichiro Kamatani（The University of Tokyo, Japan）

SY16-1

Autoimmunity pathology elucidated by functional genetics studies

Kazuyoshi Ishigaki
RIKEN Center for Integrative Medical Sciences, Japan

SY16-2

Genomic analysis and polygenic score application in cardiovascular diseases

Kaoru Ito
RIKEN Center for Integrative Medical Sciences, Japan

SY16-3

Increased PDGFRB and NF-κB signaling caused by highly prevalent somatic mutations in intracranial aneurysms

Hirofumi Nakatomi[1,2,3]
1　Biomedical Neural Dynamics Collaboration Laboratory, RIKEN Center for Brain Science, Wako, Saitama, Japan
2　Department of Neurosurgery, Faculty of Medicine, Kyorin University, Mitaka, Tokyo, Japan
3　Department of Neurosurgery, Graduate School of Medicine, The University of Tokyo, Tokyo, Japan

SY16-4

Digital Patients and Virtual Trials for Target Identification and Evaluation

Hao Li
China National Research Center for Neurological Diseases, China

Symposium 17　Rare and Undiagnosed Diseases

Date　：Saturday, October 14, 2023　8:00 ～ 9:30　Room A（Cosmos, 3F, Toshi Center Hotel）
Chairs　：Chae Jong Hee（Seoul National University Hospital, Korea）
Toshiki Takenouchi（Keio University School of Medicine, Japan）

SY17-1　Identification of a new causative gene in cardiomyopathy, elucidation of molecular mechanisms, and functional analysis

Yoshihiro Asano[1,2]

1　National Cerebral and Cardiovascular Center, Japan
2　Osaka University Graduate School of Medicine, Japan

SY17-2　Long-read sequencing in rare diseases

Vorasuk Shotelersuk[1,2]

1　Department of Pediatrics, King Chulalongkorn Memorial Hospital, Thailand
2　Center of Excellence for Medical Genomics, Faculty of Medicine, Chulalongkorn University, Thailand

SY17-3　J-RDMM and the study of rare and undiagnosed diseases using zebrafish

Koichi Kawakami[1], Hideyuki Tanabe[1], Yuri Uchiyama[2], Naomichi Matsumoto[2]

1　National Institute of Genetics, Japan
2　Yokohama City University, Japan

SY17-4　The journey of undiagnosed disease program in Korea: From pilot project to K-UDP and beyond

Chae Jong Hee[1,2]

1　Department of Genomic Medicine, Seoul National University Hospital
2　Department of Pediactics, Seoul National University College of Medicine, Seoul Korea

Symposium 18　Ethical, Legal and Social Implications

Date　：Saturday, October 14, 2023　8:00 ～ 9:30　Room B（Orion, 5F, Toshi Center Hotel）
Chairs　：Vajira H.W. Dissanayake（Department of Anatomy, Faculty of Medicine, University of Colombo, Sri Lanka）
Masayuki Yoshida（Tokyo Medical and Dental University, Japan）

SY18-1　Framework for a genomic medicine society

Masayuki Yoshida

Tokyo Medical and Dental University, Japan

SY18-2　Ethical issues involved in explaining the results of genomic information

Kyoko Takashima

National Center for Global Health and Medicine, Japan

SY18-3　Ethical considerations in genomic medicine in South Korea

Ock-Joo Kim[1], Yoon-Jung Chang[2]

1　Department of Medical History and Medical Humanities, Seoul National University College of Medicine, South Korea
2　ELSI branch, Research Institute, National Cancer Center, South Korea

SY18-4　Focusing attention on ancestral diversity in genomic research and service provision

Vajira Dissanayake[1,2]

1　Department of Anatomy, Faculty of Medicine, University of Colombo, Sri Lanka
2　Global Genomic Medicine Collaborative, Sri Lanka

Symposium 19　Neurologic Disorders-Diagnostic Perspectives

Date　：Saturday, October 14, 2023　8:00 ～ 9:30　Room C（606, 6F, Toshi Center Hotel）
Chairs　：Christian Gilissen（Radbound University, The Netherlands）
Hiroyuki Ishiura（Department of Neurology, Okayama University, Japan）

SY19-1　Improving the diagnosis of rare neurodevelopmental disorders

Christian Gilissen
Radbound University, The Netherlands

SY19-2　Whole-genome sequencing provides insights into the aetiology of neurodevelopmental disorders

Kohei Hamanaka[1,2]

1　Kyoto University, Japan
2　Yokohama City University, Japan

SY19-3　Common and rare Parkinson's disease risk variants in East Asians

Jia Nee Foo
Nanyang Technological University, Singapore

SY19-4　Recombination of repeat elements generates somatic complexity in human genomes

Giovanni Pascarella[1], Chung Chau Hon[1], Kosuke Hashimoto[2], Annika Busch[1], Joachim Luginbuhl[1], Callum Parr[1], Wing Hin Yip[1], Kazumi Abe[3], Anton Kratz[4], Alessandro Bonetti[5], Federico Agostini[5], Jessica Severin[1], Shigeo Murayama[6], Yutaka Suzuki[3], Stefano Gustincich[7], Martin Frith[3], Piero Carninci[8]

1　RIKEN, Japan
2　University of Osaka, Japan
3　University of Tokyo, Japan
4　Systems Biology Institute, Tokyo, Japan
5　Karolinska Institutet, Stockholm, Sweden
6　Tokyo Metropolitan Geriatrics and Gerontology Center, Japan
7　Italian Institute of Technology, Italy
8　Human Technopole, Milan, Italy

Symposium 20　HGA 2023 Genetic Counseling Programs Session1: Professional Development Issues including Education and Training Updates

Date　：Saturday, October 14, 2023　8:00 ～ 9:30　Room D（701, 7F, Toshi Center Hotel）
Chairs　：Mercy Laurino（Cancer Genetics and Prevention, USA / MS Genetic Counseling Program, Fred Hutchinson Cancer Center, University of the Philippines Manila, Philippines）
Junko Yotsumoto（Patient Awareness & Diagnosis, Market Access Public Affairs & Patient Experience, Japan Pharma Business Unit, Takeda Pharmaceutical Company, Japan）

SY20-1　Developing a career pathway for genetic counsellors

Breana Cham
KK Women's and Children's Hospital, Singapore

SY20-2　Challenges in implementing an effective Genetic Counseling Programme – Indian Scenario

Q Annie Hasan
Dept. of Genetics and Molecular Medicine Kamineni Hospitals, Hyderabad, India

SY20-3 **Japanese CGC development from the Japanese Association of Certified Genetic Counselors Biennial Survey in 2022**

Manami Matsukawa, Nana Akiyama, Ayumi Abe, Momoko Kato, Mikiko Kaneko, Mari Kikuchi, Sawako Matsuzaki, Mami Morita, Saki Shimada, Eriko Takamine, Asuka Toshida, Mari Tsubata, Junko Yotsumoto

The Japanese Association of Certified Genetic Counselors Survey and Research Committee, Japan

SY20-4 **Genetic counselor workforce: Current status and future perspectives**

Masakazu Nishigaki

International University of Health and Welfare, Japan

Symposium 21 HGA 2023 Genetic Counseling Programs Session2: From clinical practice -Challenges in Sharing Genetic Information within Families and GC practice issues

Date ：Saturday, October 14, 2023 13:40 ～ 15:10 Room A（Cosmos, 3F, Toshi Center Hotel）
Chairs ：Juliana Lee（Genetic Counselling Asia, Professional Society of Genetic Counselors in Asia, Asia Pacific Society of Human Genetics, Genetic Counselling Society Malaysia, Malaysia）
Yasue Horiuchi（Shizuoka Graduate University of Public Health, Japan）

SY21-1 **Communicating genetic information with families: Models, policies, and use of technology**

Peter Abad[1,2]

1 University of the Philippines Manila, Philippines
2 University of Iowa, USA

SY21-2 **Connecting generations: Bridging families with genetic information**

Jingyi Dong[1,2]

1 Tokyo Medical University Hospital, Japan
2 Ochanomizu University, Japan

SY21-3 **Disclosing information about diagnosis and conditions to children with genetic disorders and their siblings**

Mikiko Kaneko

The Jikei University Hospital, Japan

SY21-4 **Enhancing clinical genetic counselling through education: opportunities for collaborative approaches**

Alison McEwen

FHGSA(Genetic Counselling) Graduate School of Health, University of Technology Sydney, Sydney, Australia

Symposium 22 Inherited Metabolic Diseases

Date ：Saturday, October 14, 2023 13:40 ～ 15:10 Room B（Orion, 5F, Toshi Center Hotel）
Chairs ：Meow-Keong Thong（University of Malaya, Malaysia）
Kimihiko Oishi（Department of Pediatrics, The Jikei University School of Medicine, Japan）

SY22-1 **Recent advances in treatment of lysosomal storage diseases**

Motomichi Kosuga

National Center for Child Health and Development, Japan

SY22-2 **Current status of mitochondrial disease clinical practice**

Kei Murayama

Juntendo University Faculty of Medicine, Japan

SY22-3 **Research and development of gene therapy for lysosomal storage diseases**

Hiroshi Kobayashi

The Jikei University School of Medicine, Japan

SY22-4 **Inborn errors of metabolism in adults**

Thanyachai Sura

Mahidol University, Thailand

Symposium 23 Neurologic Diseases-Therapeutic Perspectives

Date ：Saturday, October 14, 2023 13:40 ～ 15:10 Room C（606, 6F, Toshi Center Hotel）
Chairs ：Mariko Taniguchi-Ikeda（Fujita Health Universtiy Hospital, Japan）
Kazuhiro Muramatsu（Jichi Medical University, Japan）

SY23-1 **Development of gene therapy for neuromuscular disorders**

Kazuhiro Muramatsu

Jichi Medical University, Japan

SY23-2 **Recent advances in repeat expansion diseases and the future potential of gene therapy**

Hiroyuki Ishiura

Okayama University, Japan

SY23-3 **Finding cures for Fukuyama muscular dystrophy**

Mariko Taniguchi-Ikeda

Fujita Health Universtiy Hospital, Japan

SY23-4 **A novel dual-targeting antisense oligonucleotide, NS-089/NCNP-02, for exon 44 Skipping in Duchenne muscular dystrophy**

Yoshitsugu Aoki

National Center of Neurology and Psychiatry, Japan

Sponsored Symposium by Myriad Genetics G.K.

Sponsored Symposium by Myriad Genetics G.K.
The large-scale genetic analysis and data sharing in East Asia for hereditary tumors

Date ：Thursday, October 12, 2023 13:50 ～ 15:20 Room A（Cosmos, 3F, Toshi Center Hotel）
Chairs ：Mercy Laurino（Cancer Genetics and Prevention, USA / MS Genetic Counseling Program, Fred Hutchinson Cancer Center, University of the Philippines Manila, Philippines）
Yoshio Miki（University of Tsukuba Research and Development Center for Precision Medicine, Japan）

SS-1 **The importance of large-scale genetic analysis in each area and population for its personalized medicine**

Yukihide Momozawa

RIKEN, Japan

SS-2 **Cancer Genetics and Testing: Importance of Genetic Counseling**

Mercy Laurino[1,2]

1 Fred Hutchinson Cancer Center, USA
2 University of the Philippines Manila, Philippines

SS-3

Genetic on Hereditary Breast and Ovarian Cancers in Hong Kong

Ava Kwong

School of Clinical Medicine, LKS Faculty of Medicine, The University of Hong-Kong, Hong Kong

Oral Presentation Award Session

Oral Presentation Award Session

Date ：Friday, October 13, 2023　8:40 ～ 9:50　Room A（Cosmos, 3F, Toshi Center Hotel）
Chairs ：Brian H.Y. Chung（Department of Paediatrics and Adolescent Medicine, School of Clinical Medicine, The University of Hong Kong, Hong Kong）
Naomichi Matsumoto（Department of Human Genetics, Yokohama City University Graduate School of Medicine, Yokohama, Japan）

BO-1

Body mass index stratification improves polygenic prediction of type 2 diabetes in trans-biobank analysis

Takafumi Ojima[1], Shinichi Namba[1], Ken Suzuki[1,3], Kyuto Sonehara[1,6], Kenichi Yamamoto[1,4,5], Akira Narita[7], The Tohoku Medical Megabank Project Study Group[7], The Biobank Japan Project[8], Yoichiro Kamatani[9], Gen Tamiya[2,7,10], Masayuki Yamamoto[2,7], Toshimasa Yamauchi[3], Takashi Kadowaki[11], Yukinori Okada[1,5,6,12]

1 Department of Statistical Genetics, Osaka University, Japan
2 Graduate School of Medicine, Tohoku University, Japan
3 Department of Diabetes and Metabolic Diseases, The University of Tokyo, Japan
4 Department of Pediatrics, Osaka University, Japan
5 Laboratory of Statistical Immunology, Osaka University, Japan
6 Department of Genome Informatics, The University of Tokyo, Japan
7 Tohoku Medical Megabank Organization, Tohoku University, Japan
8 Institute of Medical Science, The University of Tokyo, Japan
9 Laboratory of Complex Trait Genomics, The University of Tokyo, Japan
10 Center for Advanced Intelligence Project, RIKEN, Japan
11 Toranomon Hospital, Japan
12 Laboratory for Systems Genetics, RIKEN, Japan

BO-2

The insertion of SVA retrotransposon in deep intron of ATP7A as a novel cause of occipital horn syndrome

Naoko Yano[1], Takeshi Yoshida[1], Chong Pin Fee[2,3], Yu Kimura[4], Tomoichiro Miyoshi[5], Masahiko Ajiro[7], Kengo Kora[1], Taisei Kayaki[1], Kinuko Nishikawa[1], Saeko Sasaki[1], Atsushi Yokoyama[1], Masatoshi Hagiwara[6], Teruyuki Kondo[4], Ryutaro Kira[3], Junko Takita[1]

1 Department of Pediatrics, Kyoto University Graduate School of Medicine, Kyoto, Japan
2 Department of Pediatrics, Graduate School of Medical Sciences, Kyushu University, Fukuoka, Kyoto, Japan
3 Department of Pediatric Neurology, Fukuoka Children's Hospital, Fukuoka, Japan
4 Department of Energy and Hydrocarbon Chemistry, Graduate School of Engineering, Kyoto University, Kyoto, Japan
5 Center for Integrative Medical Sciences, RIKEN, Kanagawa, Japan
6 Department of Anatomy and Development Biology, Kyoto University Graduate School of Medicine, Kyoto, Japan
7 Division of Cancer RNA Research, National Cancer Center Research Institute, Tokyo, Japan

BO-3

A cross-population atlas of genome-wide gene-environment interactions between the East Asian and European populations

Shinichi Namba[1], Kyuto Sonehara[1,2,3], Koichi Matsuda[4], Yukinori Okada[1,2,3]

1 Osaka University Graduate School of Medicine, Japan
2 Graduate School of Medicine, The University of Tokyo, Japan
3 RIKEN Center for Integrative Medical Sciences, Japan
4 Graduate School of Frontier Sciences, The University of Tokyo, Japan

BO-4

Large-scale evaluation of germline variants among 11 thousand gastric cancer patients and 44 thousand controls

Yoshiaki Usui[1,2,3], Yukari Taniyama[2], Mikiko Endo[1], Yuriko N. Koyanagi[2], Yumiko Kasugai[2,4], Isao Oze[2], Hidemi Ito[2,4], Issei Imoto[2], Tsutomu Tanaka[2], Masahiro Tajika[2], Yasumasa Niwa[2], Yusuke Iwasaki[1], Tomomi Aoi[1], Nozomi Hakozaki[1], Sadaaki Takata[1], Kunihiko Suzuki[1], Chikashi Terao[1], Masanori Hatakeyama[5,6], Makoto Hirata[7,8], Kokichi Sugano[7,9], Teruhiko Yoshida[7], Yoichiro Kamatani[8], Hidewaki Nakagawa[1], Koichi Matsuda[8], Yoshinori Murakami[8], Amanda B. Spurdle[10], Keitaro Matsuo[2,4], Yukihide Momozawa[1]

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2 Aichi Cancer Center, Japan
3 Okayama University, Okayama, Japan
4 Nagoya University, Nagoya, Japan
5 Institute of Microbial Chemistry, Tokyo, Japan
6 Hokkaido University, Sapporo, Japan
7 National Cancer Center, Tokyo, Japan
8 The University of Tokyo, Tokyo, Japan
9 Kyoundo Hospital, Tokyo, Japan
10 QIMR Berghofer Medical Research Institute, Brisbane, Australia

BO-5

The effect of SOX10 expressions in a complex multifactorial Hirschsprung disease

Khoitul Machis[1], Verell Christopher Amadeus[1], Fiqih Vidiantoro Halim[1], Kristy Iskandar[2], Eko Purnomo[1], Gunadi Gunadi[1]

1 Pediatric Surgery Division, Department of Surgery, Faculty of Medicine, Public Health and Nursing, Universitas Gadjah Mada, Indonesia
2 Department of Child Health, Faculty of Medicine, Public Health and Nursing, Universitas Gadjah Mada, Indonesia

BO-6

SLC10A5 deficiency is a cause of familial intrahepatic cholestasis

Yuqing Xu[1,2,3], Yeqing Qian[1,2,3], Minyue Dong[1,2,3]

1 Zhejiang University, China
2 Women's Hospital, School of Medicine, Zhejiang University, Hangzhou, China
3 Key Laboratory of Reproductive Genetics, Ministry of Education (Zhejiang University), Hangzhou, China

BO-7

Prenatal WES: Experiences from >1400 cases - high diagnostic yield and benefit for pregnancy management and counseling

Stefan Griesbach[1], Martin Ritthaler[1], Florian Battke[1], Constantin von Kaisenberg[2], Holger Lebek[3], Michael Entezami[4], Max Wüstemann[5], Andreas Schröer[3], Saskia Biskup[1], Heinz Gabriel[1]

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2 Perinatal Center, Medizinische Hochschule Hannover, Germany
3 Prenatal Diagnostics Berlin-Lichtenberg, Germany
4 Center for Prenatal Diagnostics and Human Genetics Berlin, Germany
5 Center for Prenatal Medicine Hannover, Germany

Oral Session

Oral Session 1　Complex Diseases and Genomic Risk Assessment

Date　：Thursday, October 12, 2023　10:10 ～ 11:40　Room E（706, 7F, Toshi Center Hotel）
Chairs　：Yukihide Momozawa（RIKEN Center for Integrative Medical Sciences (IMS), Japan）
Kinya Ishikawa（Center for Personalized Medicine for Healthy Aging, Tokyo Medical and Dental University, Japan）

OS-01-1　East Asian-specific variants in RNF213 confer a penetrating risk of vasospastic angina

Keiko Hikino[1], Satoshi Koyama[1], Kaoru Ito[1], Yoshinao Koike[1,2], Masaru Koido[1,3], Takayoshi Matsumura[4], Ryo Kurosawa[1], Kohei Tomizuka[1], Shuji Ito[1,5], Xiaoxi Liu[1,6], Yuki Ishikawa[1], Yukihide Momozawa[1], Takayuki Morisaki[3], Yoichiro Kamatani[1,3], The Biobank Japan Project[3], Taisei Mushiroda[1], Chikashi Terao[1,6,7]

1 RIKEN Center for Integrative Medical Sciences, Japan
2 Hokkaido University Graduate School of Medicine, Japan
3 The University of Tokyo, Japan
4 Jichi Medical University, Japan
5 Shimane University Faculty of Medicine, Japan
6 Shizuoka General Hospital, Japan
7 University of Shizuoka, Japan

OS-01-2　Low carnitine palmitoyltransferase 1 activity is a risk factor for narcolepsy type 1

Taku Miyagawa[1], Yosuke Shigematsu[2], Mihoko Shimada[1,3,4], Yoshiko Honda[1], Katsushi Tokunaga[3,4], Makoto Honda[1,5]

1 Tokyo Metropolitan Institute of Medical Science, Japan
2 University of Fukui, Japan
3 National Center for Global Health and Medicine, Japan
4 The University of Tokyo, Japan
5 Japan Somnology Center and Seiwa Hospital, Japan

OS-01-3　Identification of a gene expression regulatory variant in autoimmune diseases-susceptibility locus CD58

Yuki Hitomi[1], Kazuko Ueno[1], Yoshihiro Aiba[2], Nao Nishida[3], Yosuke Kawai[1], Minae Kawashima[4], Seik-Soon Khor[1], Sanami Takada[1], Chisato Iwabuchi[1], Masao Nagasaki[5], Katsushi Tokunaga[1], Minoru Nakamura[2,6]

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2 Nagasaki Medical Center, Japan
3 Tokyo Medical and Dental University, Japan
4 Research Organization of Information and Systems, Japan
5 Kyushu University, Japan
6 Nagasaki University, Japan

OS-01-4　Association of metabolism-related genes polymorphisms with metabolic syndrome Components in The Balinese Population

Safarina G. Malik, Herawati Sudoyo, Sukma Oktavianthi, Pradiptajati Kusuma

Genome Diversity and Diseases Division, Mochtar Riady Institute for Nanotechnology, Indonesia

OS-01-5　Association analysis between FLG loss of function mutations and allergen sensitization: A birth cohort study

Emiko Noguchi[1], Hisato Suzuki[1], Monami Hara[1], Daisuke Hayashi[1], Tatsuki Fukuie[2], Mayako Saito-Abe[2], Limin Yang[2], Kiwako Yamamoto-Hanada[2], Masami Narita[3], Yukihiro Ohya[2]

1 University of Tsukuba, Japan
2 Allergy Center, National Center for Child Health and Development, Japan
3 Department of Pediatrics, School of Medicine, Kyorin University, Japan

OS-01-6 **Genetic association of LPL intronic variants with metabolic disorder among the Kuwaiti population**

Suzanne Al-Bustan

Kuwait University, College of Science, Kuwait

OS-01-7 **Prediction model with HLA-A*33:03 reveals number of days for liver carcinogenesis**

Nao Nishida[1], Jun Ohashi[2], Yosuke Kawai[3], Takayo Tsuchiura[1], Miyuki Ishikawa[3], Katsushi Tokunaga[3]

1 Tokyo Medical and Dental University, Japan
2 The University of Tokyo, Japan
3 National Center for Global Health and Medicine, Japan

OS-01-8 **Highly prevalent pathogenic somatic or mosaic PDGFRB variants in intracranial aneurysm**

Shota Sasagawa[1], Yasuyuki Shima[2,3], Nakao Ota[2,4], Kenjiro Kosaki[5], Hiroyuki Kamiguchi[6], Shigeo Okabe[7,8], Tadafumi Kato[9], Nobuhito Saito[10], Hirofumi Nakatomi[2,10,11], Hidewaki Nakagawa[1]

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10 Department of Neurosurgery, Graduate School of Medicine, The University of Tokyo, Tokyo, Japan
11 Department of Neurosurgery, Faculty of Medicine, Kyorin University, Mitaka, Tokyo, Japan

OS-01-9 **Study for developing mechanisms of autoimmune encephalitis: contribution of immunomodulatory genes in 43 patients**

Yukitoshi Takahashi[1,2,3,4], Shigeko Nishimura[1], Emiko Takao[1], Risa Kasai[1], Kaoru Enokida[1]

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3 School of Pharmaceutical Sciences, University of Shizuoka, Japan
4 Department of Pediatrics, Gifu city hospital, Japan

Oral Session 2　Neurology

Date　：Thursday, October 12, 2023　16:00 ～ 17:30　Room G（Meeting Room 2, 3F, Zenkoku Toshi Kaikan）
Chairs　：Tatsushi Toda（Department of Neurology, Graduate School of Medicine, The University of Tokyo, Japan）
Yuji Takahashi（Department of Neurology, Department of Genomic Medicine, National Center Hospital, National Center of Neurology and Psychiatry, Japan）

OS-02-1 **Contribution of rare coding variations to microcephaly in patients with neurodevelopmental disorders**

Jihoon Yoon[1], Seungbok Lee[1,2], Se Song Jang[2], Soojin Park[2], Jaeso Cho[1,2], Man Jin Kim[1,3], Soo Yeon Kim[1,2], Woo Joong Kim[2], Anna Cho[4], Jin Sook Lee[5], Murim Choi[6], Byung Chan Lim[2], Jung Min Ko[2], Ki Joong Kim[2], Jong Hee Chae[1,2]

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4 Department of Pediatrics, Seoul National University Bundang Hospital, Seongnam, Republic of Korea
5 Department of Pediatrics, Seoul National University Hospital Child Cancer and Rare Disease Administration, Seoul National University Children's Hospital, Seoul, Republic of Korea
6 Department of Biomedical Sciences, Seoul National University College of Medicine, Seoul, Republic of Korea

OS-02-2

Integrated analysis of DNA methylation, gene expression, and genetic variant data in CD4+/CD8+ T cells of narcolepsy

Mihoko Shimada[1], Yoshiko Honda[2], Makoto Honda[2], Katsushi Tokunaga[1], Taku Miyagawa[2]

1 National Center for Global Health and Medicine (NCGM), Japan
2 Sleep Disorders Project, Department of Psychiatry and Behavioral Sciences, Tokyo Metropolitan Institute of Medical Science, Japan

OS-02-3

Hemizygous EIF1AX variants in male are associated with neurodevelopmental disorders

Kazuyuki Komatsu[1], Atsushi Sugie[2], Yohei Nitta[2], M.Ummul Halilunnisa[1], Petra J.G. Zwijnenburg[3], Melissa T Carter[4], William B. Dobyns[5], Emanuela Argilli[6,7], Mitsuru Kubota[8], Nobuyuki Shimozawa[9,10]

1 Hamamatsu University School of Medicine, Japan
2 Brain Research Institute, Niigata University, Japan
3 Emma Children's Hospital Amsterdam UMC, University of Amsterdam, The Netherlands
4 University of Ottawa, Canada
5 University of Minnesota, USA
6 University of California, USA
7 Pediatrics Institute of Human Genetics and Weill Institute for Neurosciences, University of California, USA
8 National Center for Child Health and Development, Japan
9 Life Science Research Center, Gifu University, Japan
10 Gifu University Hospital, Japan

OS-02-4

Expansion of clinico-genetic spectrum of PRDX3 disease: A literature review with two additional cases in East Asia

Jaeso Cho[1], Jihoon G Yoon[1], Seungbok Lee[1], Sheehyun Kim[1], Soo Yeon Kim[1,2], Man Jin Kim[1,3], Jangsup Moon[1,4], Jong Hee Chae[1,2]

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2 Seoul National University College of Medicine, Department of Pediatrics, Korea
3 Seoul National University Hospital, Department of Laboratory Medicine, Korea
4 Seoul National University Hospital, Department of Neurology, Korea

OS-02-5

SYNGAP1 encephalopathy: Natural history with developmental outcome in single center cohort

Woojoong Kim[1], Hyejin Kim[1], Jong-Hee Chae[1,2]

1 Department of Pediatrics, Division of Pediatric Neurology, Seoul National University Hospital, Korea
2 Department of Genomic Medicine, Seoul National University Hospital, Korea

OS-02-6

Detailed distribution analysis of epilepsy/neurodevelopmental disorder proteins Nav1.1 and Nav1.2

Tetsushi Yamagata[1], Toshimitsu Suzuki[1], Yurina Hibi[1], Ikuo Ogiwara[2], Kazuhiro Yamakawa[1]

1 Nagoya City University Graduate School of Medical Sciences, Japan
2 Nippon Medical School, Japan

OS-02-7

CGG repeat expansion in LRP12 in amyotrophic lateral sclerosis

Kodai Kume[1], Takashi Kurashige[2], Keiko Muguruma[3], Hiroyuki Morino[4], Yui Tada[1], Mai Kikumoto[1], Tatsuo Miyamoto[1], Silvia Natsuko Akutsu[1], Matsuda Yukiko[1], Shinya Matsuura[1], Masahiro Nakamori[5], Ayumi Nishiyama[6], Rumiko Izumi[6], Tetsuya Niihori[6], Masashi Ogasawara[7], Nobuyuki Eura[7], Tamaki Kato[8], Mamoru Yokomura[8], Yoshiaki Nakayama[9], Hidefumi Ito[9], Masataka Nakamura[3], Kayoko Saito[8], Yuichi Riku[10], Yasushi Iwasaki[10], Hirofumi Maruyama[5], Yoko Aoki[6], Ichizo Nishino[7], Yuishin Izumi[4], Masashi Aoki[6], Hideshi Kawakami[1]

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3 Kansai Medical University, Japan
4 Tokushima University, Japan
5 Hiroshima University, Japan
6 Tohoku University, Japan
7 National Institute of Neuroscience, National Center of Neurology and Psychiatry, National Center Hospital, Japan
8 Tokyo Women's Medical University, Japan
9 Wakayama Medical University, Japan
10 Aichi Medical University, Japan

OS-02-8

Circulating microRNAs as disease biomarkers of Fukuyama muscular dystrophy

Mariko Taniguchi-Ikeda[1], Daisuke Masuda[3], Tatsutoshi Inuzuka[3], Tatsushi Toda[2]

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OS-02-9

Impaired gating of γ- and ε-AChR respectively causes Escobar syndrome and fast-channel congenital myasthenic syndrome

Tomohiko Nakata[1], Xin-Ming Shen[2], Seiji Mizuno[3], Issei Imoto[4], Duygu Selcen[2], Andrew G. Engel[2], Kinji Ohno[1]

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2 Mayo Clinic, USA
3 Central Hospital, Aichi Developmental Disability Center, Japan
4 Aichi Cancer Center Research Institute, Japan

Oral Session 3　Inherited Metabolic Diseases and Newborn Screening

Date　：Friday, October 13, 2023　10:10 ～ 11:40　Room E（706, 7F, Toshi Center Hotel）
Chairs　：Kimitoshi Nakamura（Dept. Pediatrics, Kumamoto Univ., Japan）
Kayoko Saito（Institute of Medical Genetics, Tokyo Women's Medical University, Japan）

OS-03-1

Genetic background and long-term prognosis of cardiomyopathy in 313 mitochondrial disease patients

Atsuko Okazaki[1], Ayako Matsunaga[2], Yukiko Yatsuka[1], Yoshihito Kishita[3], Ayumu Sugiura[1], Yohei Sugiyama[1,4], Takuya Fushimi[4], Masaru Shimura[4], Keiko Ichimoto[4], Makiko Tajika[4], Tomohiro Ebihara[5], Tesuro Matsuhashi[4], Tomoko Tsuruoka[5], Tomoko Hirata[6], Atsuhito Takeda[7], Akira Ohtake[8,9], Kei Murayama[1,4,10], Yasushi Okazaki[1,6]

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5 Department of Neonatology, Chiba Children's Hospital, Chiba, Japan
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OS-03-2

UPLC-MS/MS analysis of urinary oligosaccharides for the diagnosis of mucopolysaccharidosis and glycoproteinosis

Parith Wongkittichote, Se Hyun Cho, Rebecca Ahrens-Nicklas, Can Ficicioglu, Sarah Schmidt, Xinying Hong

Children's Hospital of Philadelphia, USA

OS-03-3

Efforts aimed at quick genetic diagnosis of suspected mitochondrial diseases: Application of whole blood RNA sequencing

Yukiko Yatsuka[1], Yoshihito Kishita[1,2], Takuya Fushimi[5,6], Atsuko Okazaki[1], Ayumu Sugiura[1], Akira Ohtake[3,4], Kei Murayama[1,5,6], Yasushi Okazaki[1,7]

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4 Center for Intractable Diseases, Saitama Medical University Hospital, Japan
5 Center for Medical Genetics, Chiba Children's Hospital, Japan
6 Department of Metabolism, Chiba Children's Hospital, Japan
7 Laboratory for Comprehensive Genomic Analysis, RIKEN Center for Integrative Medical Sciences, Japan

OS-03-4

Distribution of variants and genotypes of CYP21A2 in congenital adrenal hyperplasia over a period of 15 years in Iran

Bahareh Rabbani[1], Nejat Mahdieh[2], Ali Rabbani[1], Mahin Hashemipour[3], Zahra Razavi[4], Mahtab Ordouei[5], Parastoo Rostami[1], Aria Setoudeh[1], Kobra Shiasi Arani[6], Reza Tavakolizadeh[7], Reihaneh Mohsenipour[1], Fatemaeh Sayarifard[8], Naserali Mirhosseini[6], Fahimeh Soheilipour[9]

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2 Rajaie Cardiovascular Research Center, Iran
3 Isfahan University of Medical Science, Iran
4 Hamedan University of Medical Science, Iran
5 Shahid Sadoughi University of Medical Sciences, Iran
6 Kashan University of Medical Sciences, Iran
7 Tehran University of Medical Science, Iran
8 Children's Medical Center, Iran
9 Iran University of Medical Sciences, Iran

OS-03-5

Calcitonin gene-related peptide (CGRP) receptor component (CRCP) deficiency in atypical hemolytic uremic syndrome

Ching Wan Lam

Department of Pathology, School of Cinical Medicine, The University of Hong Kong, Hong Kong, China

OS-03-6

Streamlined determination of 3-O-methyldopa in dried blood spots: prospective screening for AADC deficiency

Yin-Hsiu Chien, Pin-Wen Chen, Wuh-Liang Hwu, Ni-CHung Lee

National Taiwan University Hospital, Taiwan

OS-03-7

Future perspectives of NBS: experience from cord blood transplantation for the infants with athymic CHARGE syndrome

Motoshi Sonoda[1], Masataka Ishimura[1], Hirosuke Inoue[1], Katsuhide Eguchi[1], Masayuki Ochiai[1], Yasunari Sakai[1], Takehiko Doi[2], Kyoko Suzuki[3], Takeshi Inoue[4], Tomoyuki Mizukami[5], Hidetoshi Takada[6], Shouichi Ohga[1]

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3 Department of Pediatrics, Juntendo University, Urayasu Hospital, Japan
4 Division of Neonatology, Perinatal Center, Kumamoto City Hospital, Japan
5 Department of Pediatrics, National Hospital Organization Kumamoto Medical Center, Japan
6 Department of Child Health, Faculty of Medicine, University of Tsukuba, Japan

OS-03-8

Acceptability of dried blood spot collection by caregivers of Filipino patients with maple syrup urine disease and PKU

Roxanne Janica Merencilla[1], Leniza de Castro-Hamoy[2], Ebner Bon Maceda[1,2], Jeanne Ruth Basas[1], Rufus Thomas Adducul[1]

1 Institute of Human Genetics, National Institute for Health - University of the Philippines, Manila, Philippines
2 Department of Pediatrics- Philippine General Hospital, Philippines

OS-03-9

Experiences and perceptions of Filipino mothers on the disclosure of positive maple syrup urine disease (MSUD) Newborn Screening and confirmatory test results with supplemental data from healthcare workers

Kevina Mariz Dajoyag, Catherine Lynn Silao, Mercy Laurino, Ma-Am Joy Tumulak, Mary Anne Chiong, Manuel Victor Sapitula

University of the Philippines, Manila, Philippines

Oral Session 4　Rare Diseases 1

Date　：Friday, October 13, 2023　10:10 ～ 11:40　Room G（Meeting Room 2, 3F, Zenkoku Toshi Kaikan）
Chairs　：Maki Fukami（National Research Institute for Child Health and Development, Japan）
Shoji Tsuji（International University of Health and Welfare, Japan）

OS-04-1 The comprehensive genetic analyses of cystic kidney disease in Japan

Naoya Morisada[1], Ming Juan Ye[2], Yu Tanaka[2], Eri Okada[3], Kazumoto Iijima[2], Kandai Nozu[2]

1 Hyogo Prefectural Kobe Children's Hospital, Japan
2 Department of Pediatrics, Kobe University Graduate School of Medicine, Japan
3 Department of Nephrology, Faculty of Medicine, University of Tsukuba, Japan

OS-04-2 Risk of assisted reproductive technology and maternal childbearing age for the development of imprinting disorders

Kaori Hara[1], Keiko Matsubara[1], Akie Nakamura[1,2], Shinichiro Sano[1,3], Takanobu Inoue[1], Sayaka Kawashima[1], Tomoko Fuke[1], Kazuki Yamazawa[1,4], Maki Fukami[1], Tsutomu Ogata[1,5,6], Masayo Kagami[1]

1 National Research Institute for Child Health and Development, Japan
2 Department of Pediatrics, Hokkaido University School of Medicine, Japan
3 Department of Endcrinology and Metabolism, Shizuoka Children's Hospital, Japan
4 Medical Genetics Center, National Hospital Organization Tokyo Medical Center, Japan
5 Department of Biochemistry, Hamamatsu University School of Medicine, Japan
6 Department of Pediatrics, Hamamatsu Medical Center, Japan

OS-04-3 Impaired hematopoiesis in mice harboring a disease-associated MECOM (EVI1) mutation

Koki Nagai[1], Tetsuya Niihori[1], Akihiko Muto[2], Yoshikazu Hayashi[3], Taiki Abe[1], Kazuhiko Igarashi[2], Yoko Aoki[1]

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2 Department of Biochemistry, Tohoku University Graduate School of Medicine, Japan
3 Division of Functional Structure, Department of Morphological Biology, Fukuoka Dental College, Japan

OS-04-4 PMEPA1 mutation found in 3 Japanese families with MFS/LDS like features

Hiroko Morisaki[1,2,3], Itaru Yamanaka[3], Takako Ohata[4], Tomoki Kosho[5,6], Keiko Wakui[5,6], Mitsuo Masuno[7], Tetsuro Watabe[8], Yukihide Watanabe[9], Takayuki Morisaki[2,3]

1 Sakakibara Heart Institute, Japan
2 IMSUT Hospital, Inst Med Sci, Univ Tokyo, Japan
3 Natl Cerebr Cardiovasc Ctr, Japan
4 Okinawa Pref Hosp, Japan
5 Dept Med Genet, Shinshu Univ Med Sch, Japan
6 Ctr Med Genet, Shishu Univ Hosp, Japan
7 Kawasaki Med Sch, Japan
8 Dept Biochem, Tokyo Med Dent Univ, Japan
9 Facult Med & Grad Sch Coprehens Hum Sci, Univ Tsukuba, Japan

OS-04-5 Inpatient morbidity, mortality, and healthcare utilisation of hospitalised patients with Prader-Willi syndrome

Jirat Chenbhanich[1], Patompong Ungprasert[2], Paul Kroner[3]

1 Department of Genetics and Genomic Sceinces, Case Western Reserve University, Cleveland, Ohio, USA
2 Department of Rheumatic and Immunologic Diseases, Cleveland Clinic, Cleveland, Ohio, USA
3 Division of Gastroenterology and Hepatology, Department of Medicine, Mayo Clinic, Jacksonville, FL, USA

OS-04-6 Biallelic null variants in PNPLA8 cause microcephaly by reducing the number of basal radial glia

Yuji Nakamura[1], Issei Shimada[1], Reza Maroofian[2], Akihiko Miyauchi[3], Eriko Koshimizu[4], Satoko Miyatake[4], Yuko Arioka[5], Mizuki Honda[6], Takayoshi Higashi[7], Fuyuki Miya[8], Kazuhiro Haginoya[9], Naomichi Matsumoto[4], Norio Ozaki[5], Yasuyuki Ohkawa[10], Shinya Oki[6], Tatsuhiko Tsunoda[11], Yoshitaka Taketomi[7], Makoto Murakami[7], Yoichi Kato[1], Shinji Saitoh[1]

1 Nagoya City University Graduate School of Medical Sciences, Japan
2 UCL Queen Square Institute of Neurology, University College London, UK
3 Jichi Medical University, Japan
4 Yokohama City University Graduate School of Medicine, Japan
5 Nagoya University Graduate School of Medicine, Japan
6 Kyoto University Graduate School of Medicine, Japan
7 Center for Disease Biology and Integrative Medicine, Graduate School of Medicine, The University of Tokyo, Japan
8 Keio University School of Medicine, Japan
9 Miyagi Children's Hospital, Japan
10 Medical Institute of Bioregulation, Kyushu University, Japan
11 School of Science, The University of Tokyo, Japan

OS-04-7 Gain-of-function MYCN causes a megalencephaly-polydactyly syndrome manifesting mirror phenotypes of Feingold syndrome

Yosuke Nishio[1,2,3], Kohji Kato[1,2,3], Frederic Tran Mau-Them[4,5], Hiroshi Futagawa[6], Chloé Quélin[7], Saori Masuda[8], Antonio Vitobello[4,5], Shiomi Otsuji[1], Hossam H. Shawki[9], Hisashi Oishi[9], Christel Thauvin-Robinet[4,5,10], Toshiki Takenouchi[11], Kenjiro Kosaki[12], Yoshiyuki Takahashi[2], Shinji Saitoh[1]

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3 Department of Genetics, Research Institute of Environmental Medicine, Nagoya University, Japan
4 Unité Fonctionnelle d'Innovation en Diagnostique Génomique des Maladies Rares, Pôle de Biologie, CHU Dijon Bourgogne, France
5 INSERM UMR GAD, Université de Bourgogne, France
6 Department of Clinical Genetics, Tokyo Metropolitan Children's Medical Center, Japan
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9 Department of Comparative and Experimental Medicine, Nagoya City University Graduate School of Medical Sciences and Medical School, Japan
10 Centre de Référence Maladies Rares "Anomalies du développement et syndromes malformatifs", Centre de Génétique, FHU TRANSLAD et Institut GIMI, CHU Dijon Bourgogne, France
11 Department of Pediatrics, Keio University School of Medicine, Japan
12 Center for Medical Genetics, Keio University School of Medicine, Japan

OS-04-8 Phenotypic heterogeneities in five patients with ZMYND11-related syndromic intellectual disability

Qiaowei Liang[1], Yuri Uchiyama[1,2], Naomi Tsuchida[1,2], Yuta Inoue[1], Hiromi Aoi[1], Rie Seyama[1], Isabel Furquim[3], Chong Ae Kim[3], Hirotaka Motoi[4], Ikumi Moriyama[5], Mariko Taniguchi-Ikeda[5], Chikahiko Numakura[6], Eriko Koshimizu[1], Atsushi Fujita[1], Kazuharu Misawa[1], Satoko Miyatake[1,7], Takeshi Mizuguchi[1], Naomichi Matsumoto[1]

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3 Genetics Unit, Instituto da Crianca, Faculdade de Medicina, Universidade de São Paulo, São Paulo, Brazil
4 Department of Pediatrics, Yokohama City University Medical Center, Japan
5 Department of Clinical Genetics, Fujita Health University Hospital, Japan
6 Department of Clinical Genomics, Saitama Medical University, Japan
7 Department of Clinical Genetics, Yokohama City University Hospital, Japan

OS-04-9 Molecular pathogenesis underlying genetic diseases with abnormalities of DNA methylation

Motoko Unoki[1], Shuhei Uemura[2], Hiroyuki Sasaki[2]

1 The University of Tokyo, Japan
2 Kyushu University, Japan

Oral Session 5　Rare Diseases 2

Date　：Friday, October 13, 2023　16:20 ～ 18:10　Room G（Meeting Room 2, 3F, Zenkoku Toshi Kaikan）
Chairs　：Mariko Eguchi（Department of Pediatrics, Ehime University Graduate School of Medicine, Japan）
Yoichi Matsubara（National Center for Child Health and Development, Japan）

OS-05-1 Heterozygous loss-of-function DHX9 variants are associated with neurodevelopmental disorders

Mamiko Yamada[1], Yohei Nitta[2], Tomoko Uehara[1], Hisato Suzuki[1], Toshiki Takenouchi[3], Masaru Tamura[4], Shinya Ayabe[5], Atsushi Yoshiki[5], Yumiko Saga[6], Nobuhiko Okamoto[7], Atsushi Sugie[2], Kenjiro Kosaki[1]

1 Keio University, School of Medicine, Center for Medical Genetics, Japan
2 Brain Research Institute, Niigata University, Japan
3 Department of Pediatrics, Keio University School of Medicine, Japan
4 Mouse Phenotype Analysis Division, RIKEN BioResource Research Center, Japan
5 Experimental Animal Division, RIKEN BioResource Research Center, Japan
6 Mammalian Development Laboratory, Department of Gene Function and Phenomics, National Institute of Genetics, Japan
7 Department of Medical Genetics, Osaka Women's and Children's Hospital, Japan

OS-05-2 Genome-wide association and HLA fine-mapping analysis of Hunner-type interstitial cystitis identify risk HLA variants

Kyuto Sonehara[1,2,3], Yoshiyuki Akiyama[1], Daichi Maeda[4], Hiroto Katoh[1], Tatsuhiko Naito[1,2,3], Kenichi Yamamoto[2], Biobank Japan Project[6], Takayuki Morisaki[1], Shumpei Ishikawa[1], Tetsuo Ushiku[1], Haruki Kume[1], Yukio Homma[5], Yukinori Okada[1,2,3]

1 The University of Tokyo, Japan
2 Osaka University, Japan
3 RIKEN Center for Integrative Medical Sciences, Japan
4 Kanazawa University, Japan
5 Kyorin University, Japan
6 Biobank Japan Project, Japan

OS-05-3 Genotype and phenotype landscape of 283 Japanese patients with tuberous sclerosis complex

Yo Niida[1], Sumihito Togi[2], Hiroki Ura[2]

1 Kanazawa Medical University Hospital, Japan
2 Medical Research Institute, Kanazawa Medical University, Japan

OS-05-4 Diagnostic yield and utility of exome sequencing on 18,994 patients with suspected rare genetic disorders

Go Hun Seo, Seong-In Hyun, Kisang Kwon, Hane Lee

3billion, Korea

OS-05-5 Rapid generation of transgenic mouse mimicking variant of uncertain significance (VUS) clarifies its pathogenicity

Shin Hayashi[1], Yasuyo Suzuki[1], Daisuke Fukushi[1], Kenichiro Yamada[1], Hiroaki Miyahara[2]

1 Institute for Developmental Research, Aichi Developmental Disability Center, Japan
2 Department of Neuropathology, Institute for Medical Science of Aging, Aichi Medical University, Japan

OS-05-6 A disease model using iPSs from patients with Kosaki overgrowth syndrome reveals abnormal migration of myofibroblasts

Hironobu Okuno[1], Miki Sato[2], Naoko Yamamoto[2], Kenjiro Kosaki[2], Hideyuki Okano[2]

1 Tokyo Medical University, Japan
2 Keio University School of Medicine, Japan

OS-05-7 **Impact of clonal hematopoiesis in chronic thromboembolic pulmonary disease: involvement of neutrophil extracellular trap**

Mizuki Momoi[1], Yoshinori Katsumata[1], Hiroyoshi Kunimoto[2], Yoshiki Shinya[1], Takahiro Hiraide[1], Fuyuki Miya[1], Kenjiro Kosaki[1], Hideaki Nakajima[2], Masaharu Kataoka[1,3]

1 Keio University School of Medicine, Japan
2 Yokohama City University Graduate School of Medicine, Japan
3 University of Occupational and Environmental Health, Japan

OS-05-8 **Exome sequencing identifies novel genes and variants in patients with Hirschsprung disease**

. Gunadi[1], Alvin Santoso Kalim[1], Kristy Iskandar[1], Marcellus Marcellus[1], Dyah Ayu Puspitarani[1], Rizki Diposarosa[2], Akhmad Makhmudi[1], Galuh Dyah Nur Astuti[3]

1 Faculty of Medicine, Public Health, and Nursing, Universitas Gadjah Mada, Indonesia
2 Faculty of Medicine, Universitas Padjadjaran, Indonesia
3 Radboud University Medical Center, Nijmegen, The Netherlands

OS-05-9 **The clinical spectrum and genetic variability of limb-girdle muscular dystrophy in a cohort of Indonesian patients**

Kristy Iskandar, . Sunartini, Gabriele Ivana, . Gunadi

Universitas Gadjah Mada, Indonesia

OS-05-10 **Diverse clinical manifestations due to pathogenic DHX37 variants in 46,XY disorders of sex differentiation**

Yuko Katoh-Fukui[1], Daisuke Saito[1], Hiroko Narumi[2], Atsushi Hattori[1], Maki Igarashi[1], Erika Uehara[1,3], Hirohito Shima[4], Junko Kanno[4], Yukihiro Hasegawa[2], Reiko Horikawa[3], Maki Fukami[1]

1 National Research Institute for Child Health and Development, Japan
2 Tokyo Metropolitan Children's Medical Center, Japan
3 National Center for Child Health and Development, Japan
4 Tohoku University School of Medicine, Japan

OS-05-11 **Care4Rare Canada: Network science to solve the unsolved rare genetic diseases**

Kym Boycott[1], Taila Hartley[1], Care4Rare Canada Consortium[1], Francois Bernier[2]

1 University of Ottawa, Canada
2 University of Calgary, Canada

Oral Session 6 Databases / Data sharing / Biobank

Date ：Saturday, October 14, 2023 13:40 ～ 14:40 Room D（701, 7F, Toshi Center Hotel）
Chairs ：Toshiaki Katayama（Database Center for Life Science, Japan）
Mayumi Kamada（Graduate School of Medicine, Kyoto University, Japan）

OS-06-1 **Establishment of a nationwide cardiovascular genomic registry for genomic medicine**

Yohei Miyashita[1,2], Koshiro Kanaoka[1], Yuki Kuramoto[2], Yasuki Ishihara[1], Yuji Sakahashi[1], Yoshihiro Asano[1,2]

1 National Cerebral and Cardiovascular Center, Japan
2 Osaka University Graduate School of Medicine, Japan

OS-06-2 **CaseSharing: A case information management system in PubCaseFinder suitable for sharing rare disease cases**

Toyofumi Fujiwara[1], Eisuke Dohi[3], Jae-Moon Shin[1], Yuka Tateisi[4], Yasunori Yamamoto[1], Atsuko Yamaguchi[5], Atsuo Kikuchi[2]

1 Database Center for Life Science, Japan
2 Department of Pediatrics, Tohoku University Graduate School of Medicine, Japan
3 National Center of Neurology and Psychiatry, Japan
4 Japan Science and Technology Agency Department of NBDC Program, Japan
5 Tokyo City University, Japan

OS-06-3

Real-word data of gynecological malignancies in Japan -Comprehensive analysis of C-CAT database

Michihiro Tanikawa, Masachika Ikegami, Misako Kusakabe, Tatsuya Sato, Aya Osonoi, Fuminori Yoshino, Reiko Nakamura, Nao Kino, Tatsuro Yamaguchi, Toshiharu Yasugi

Tokyo Metropolitan Cancer and Infectious Diseases Center Komagome Hospital, Japan

OS-06-4

Investigating batch effects in whole genome sequencing: A study of paired samples from two sequencing projects

Uladzislau Korzun[1,2,3], Hiroyuki Mishima[1,3,4], Koh-ichiro Yoshiura[1,3,4]

1 Nagasaki University, Japan
2 Medical Science Course, Department of Disaster and Radiation Medical Sciences, Graduate School of Biomedical Science, Nagasaki University, Japan
3 Department of Human Genetics, Genomic Function Analysis Unit, Atomic Bomb Disease Institute, Nagasaki University, Japan
4 Leading Medical Research Core Unit, Graduate School of Biomedical Science, Nagasaki University, Japan

OS-06-5

GestaltMatcher supports lumping and splitting decision-making by facial phenotype descriptors

Tzung-Chien Hsieh[1], Hannah Klinkhammer[1,2], Hellen Lesmann[1,3], Shahida Moosa[4], Alexander Hustinx[1], Behnam Javanmardi[1], Jing-Mei Li[1], Martin M.C. Chui[5], Christopher C.Y. Mak[5], Luisa Averdunk[6], Felix Distelmaier[6], Brian H.Y. Chung[5], Peter Krawitz[1]

1 Institute for Genomic Statistics and Bioinformatics, University Hospital of Bonn, Bonn, Germany
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3 Institute of Human Genetics, University Hospital of Bonn, Bonn, Germany
4 Division of Molecular Biology and Human Genetics, Stellenbosch University, Stellenbosch, South Africa
5 Department of Paediatrics and Adolescent Medicine, School of Clinical Medicine, LKS Faculty of Medicine, University of Hong Kong, Hong Kong
6 Department of General Pediatrics, Neonatology and Pediatric Cardiology, University Children's Hospital, Heinrich-Heine-University Düsseldorf, Düsseldorf, Germany

OS-06-6

GestaltMatcher Database - a FAIR database for medical imaging data of rare disorders

Hellen Lesmann[1,2], Gholson Lyon[3], Pilar Caro[4], Ibrahim Abdelrazek[5], Shahida Moosa[6], Jean Tori Pantel[7], Jing-Mei Li[2], Merle ten Hagen[2], Tom Kamphans[8], Wolfgang Meiswinkel[8], Benjamin D. Solomon[9], Rebekah Waikel[9], Ebtesam Abdalla[5], Markus M. Nöthen[1], Peter Krawitz[2], Tzung-Chien Hsieh[2]

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3 Department of Human Genetics, New York State Institute for Basic Research in Developmental Disabilities, Staten Island, New York, United States of America
4 Institute of Human Genetics, Heidelberg University, Heidelberg, Germany
5 Department of Human Genetics, Medical Research Institute, Alexandria University, Alexandria, Egypt
6 Division of Molecular Biology and Human Genetics, Stellenbosch University and Medical Genetics, Tygerberg Hospital, Stellenbosch, South Africa
7 Institute for Digitalization and General Medicine, University Hospital RWTH Aachen, Aachen, Germany
8 GeneTalk, Bonn, Germany
9 Medical Genomics Unit, Medical Genetics Branch, National Human Genome Research Institute, Bethesda, USA

Oral Session 7　Prenatal Genetics

Date　：Saturday, October 14, 2023　8:00 ～ 9:30　Room E（706, 7F, Toshi Center Hotel）
Chairs　：Osamu Samura（Department of Obstetrics and Gynecology, The Jikei University School of Medicine, Japan）
Aiko Sasaki（National Center for Child Health and Development, Japan）

OS-07-1

The Role of prenatal genetic counselling in parental reproductive decision in rare disease

Rifhan Azwani Mazlan[1], Meow Keong Thong[1,2], Sok Kun Tae[2]

1 Medical Genetics Unit, University Malaya Medical Centre, Malaysia
2 Genetic and Metabolism Unit, Department of Pediatric, Faculty of Medicine, University Malaya, Malaysia

OS-07-2 Low-frequency maternal novel MYH7 mosaicism mutation in recurrent fetal-onset severe left ventricular noncompaction

Hiroshi Kawamura[1], Masamichi Ikawa[2], Keiichi Hirono[3], Junya Kimura[6], Takashi Okuno[4], Masao Kawatani[4], Kunihiro Inai[5], Yoshio Yoshida[1]

1 Department of Obstetrics and Gynecology, University of Fukui, Japan
2 Department of Medical Genetics, University of Fukui Hospital, Japan
3 Department of Pediatrics, University of Toyama, Japan
4 Department of Pediatrics, University of Fukui, Japan
5 Department of Molecular Pathology, University of Fukui, Japan
6 Division of Diagnostic Pathology / Surgical Pathology, University of Fukui Hospital, Japan

OS-07-3 Preimplantation and prenatal genetic diagnosis for mitochondrial DNA disorders: efforts for reassurance

Eri Shijiki[1], Chikahiko Numakura[1,3], Megumi Tsuruoka[1], Hazuki Sato[1,2], Nozomi Uemura[1,2], Sayaka Ajihara[1,3], Ikuma Musha[1,3], Yusuke Narizuka[1,4], Yukiko Yatsuka[5], Yosuke Mizuno[1,4], Kei Murayama[5,6,7], Yasushi Okazaki[5], Naoki Hayashi[8], Akira Namba[1,2], Yoshimasa Kamei[2], Akira Ohtake[1,3]

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3 Department of Pediatrics, Saitama Medical University, Japan
4 Division of Morphological Science, Biomedical Research Center, Saitama Medical University, Japan
5 Intractable Disease Research Center, Juntendo University, Japan
6 Department of Metabolism, Chiba Children's Hospital, Japan
7 Clinical Genetics Center, Chiba Children's Hospital, Japan
8 Women's Clinic Fujimino, Japan

OS-07-4 Japanese multicenter study on fetal CNVs in detected in maternal cell-free DNA in cases of miscarriage and stillbirth

Yuka Yamashita[1], Nahoko Shirato[1], Kiyotake Ichizuka[3], Reina Komatsu[2], Seiji Wada[4], Haruhiko Sagou[4], Yuki Ito[5], Osamu Samura[5], Nobuhiro Suzumori[6], Hideaki Sawai[7], Yuko Tamaki[9], Yukiko Katagiri[9], Yoshinori Maeda[8], Hiroko Morisaki[13], Akira Namba[10], Yoshimasa Kamei[10], Yuri Hasegawa[11], Kiyonori Miura[11], Setsuko Nakayama[12], Akihiko Sekizawa[1]

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4 National Center for Child Health and Development Department of Obstetrics and Gynecology, Japan
5 The Jikei University School of Medicine Department of Obstetrics and Gynecology, Japan
6 Nagoya City University Hospital Department of Obstetrics and Gynecology, Japan
7 Hyogo Medical University Department of Obstetrics and Gynecology, Japan
8 Sakakibara Heart Institute Department of Obstetrics and Gynecology, Japan
9 Toho University Omori Medical Center Department of Obstetrics and Gynecology, Japan
10 Saitama Medical University Hospital Department of Obstetrics and Gynecology, Japan
11 Nagasaki University Department of Obstetrics and Gynecology, Japan
12 Aiiku Hosipital Department of Obstetrics and Gynecology, Japan
13 Sakakibara Heart Institute Department of Clinical Genetics, Japan

OS-07-5 Comparison of full length aneuploidy and segmental aneuploidy in PGT-A mosaic embryos at a private clinic

Atsushi Tanaka[1], Shohei Komemoto[1], Youichi Takemoto[1], Motoi Nagayoshi[1], Yuya Makino[1,2], Daisuke Nakajima[1,3], Seiji Watanabe[4]

1 Saint Mother Clinic, Japan
2 Juntendo University School of Medicine, Japan
3 Occupational and Environmental Health University School of Medicine, Japan
4 Hirosaki University Graduate School of Medicine, Japan

OS-07-6 Accuracy in predicting chromosomal aneuploidy by professional precise ultrasound examination at 1st trimester

Norio Shinozuka, Ayaka Kawabe, Yuto Yamamoto, Michiharu Seto

Seto Hospital, Japan

OS-07-7 **What is needed to discuss regarding termination of pregnancy: lessons we learned at a prenatal diagnostic center**

Chieko Tamura, Chikoto Ihara, Mio Furusawa, Kenji Yamada, Yasushi Yamada

FMC Tokyo Clinic, Japan

OS-07-8 **Establishment of genetic tool for reproductive risk analysis of reciprocal translocation**

Makoto Inaki[1], Tsuyoshi Baba[2], Hiroki Kurahashi[3], Toshiaki Endo[1,2,4]

1 Sapporo Recurrent Miscarriage and Implantation Failure Consortium, Japan
2 Department of Obstetrics and Gynecology, Sapporo Medical University, Japan
3 Division of Molecular Genetics, Institute for Comprehensive Medical Science, Fujita Health University, Japan
4 Ena Asabu Art Clinic, Japan

OS-07-9 **A study on the selection of examination facilities after the start of the NIPT certification system**

Nahoko Shirato[1], Akihiko Sekizawa[1], Tatsuko Hirose[1,2], Shin Ikebukuro[1], Takeshi Nakamura[1], Keiko Miyagami[1], Takahiro Yamada[3]

1 Showa University School of Medicine, Japan
2 Showa University Graduate School of Health Sciences, Japan
3 Division of Clinical Genetics, Hokkaido University Hospital, Japan

Oral Session 8 Technological Advanced, Wet and Dry

Date ：Saturday, October 14, 2023 13:40 ～ 15:10 Room E（706, 7F, Toshi Center Hotel）
Chairs ：Masayo Kagami（Department of Molecular Endocrinology, National Research Institute for Child Health and Development, Japan）
Kenichiro Hata（Dept. Hum. Mol. Genet., Gunma Univ., Japan / National Center for Child Health and Development, Japan）

OS-08-1 **Region-specific gene isoforms in the human brain using long-read sequencing and their correlation with DNA methylation**

Mihoko Shimada[1], Yosuke Omae[1], Akiyoshi Kakita[2], Ramil Gabdulkhaev[2], Taku Miyagawa[3], Makoto Honda[3], Akihiro Fujimoto[4], Katsushi Tokunaga[1]

1 National Center for Global Health and Medicine (NCGM), Japan
2 Department of Pathology, Brain Research Institute, Niigata University, Japan
3 Sleep Disorders Project, Department of Psychiatry and Behavioral Sciences, Tokyo Metropolitan Institute of Medical Science, Japan
4 Department of Human Genetics, Graduate School of Medicine, The University of Tokyo, Japan

OS-08-2 **Functional and dynamic profiling of transcript isoforms reveals roles of isoform switching in interferon response**

Mahoko Ueda[1], Jun Inamo[2,3], Fuyuki Miya[4], Kensuke Yamaguchi[5,6], Yuta Kochi[1,6]

1 Medical Research Institute, Tokyo Medical and Dental University, Japan
2 Division of Rheumatology, University of Colorado School of Medicine, Aurora, CO, USA
3 Department of Biomedical Informatics, Center for Health Artificial Intelligence, University of Colorado School of Medicine, Aurora, CO, USA
4 Center for Medical Genetics, Keio University School of Medicine, Japan
5 Biomedical Engineering Research Innovation Center, Institute of Biomaterials and Bioengineering, Tokyo Medical and Dental University, Tokyo, Japan
6 Laboratory for Autoimmune Diseases, RIKEN Center for Integrative Medical Sciences, Yokohama, Kanagawa, Japan

OS-08-3 **Identification of DNA methylation sites by a trans-ethnic epigenome-wide meta-analysis for circulating adiponectin levels**

Masahiro Nakatochi[1], Sahoko Ichihara[2], Ken Yamamoto[3], Tatsuaki Matsubara[4], Mitsuhiro Yokota[5]

1 Nagoya University, Japan
2 Department of Environmental and Preventive Medicine, Jichi Medical University School of Medicine, Shimotsuke, Japan
3 Department of Medical Biochemistry, Kurume University School of Medicine, Kurume, Japan
4 Faculty of Human Sciences, Aichi Mizuho College, Nagoya, Japan
5 Kurume University School of Medicine, Kurume, Japan

OS-08-4 **Japanese reference genome elucidated chromosomal rearrangements with structural variations at long-read sequencing**

Yukiko Kuroda, Yoko Saito, Yumi Enomoto, Takuya Naruto, Kenji Kurosawa

Kanagawa Children's Medical Center, Japan

OS-08-5 **Demonstrating the variability of variant classification through external quality assessment**

Zandra Deans, Dave Cregeen, Jenni Fairley, Farrah Khawaja, Mark Sales, Melody Tabiner, Rebecca Treacy, Rosalind Hastings

GenQA, UK

OS-08-6 **PDIVAS: Pathogenicity predictor for Deep-Intronic Variants causing Aberrant Splicing**

Ryo Kurosawa[1], Kei Iida[2], Masahiko Ajiro[3], Tomonari Awaya[1], Mamiko Yamada[4], Kenjiro Kosaki[4], Masatoshi Hagiwara[1]

1 Kyoto University, Japan
2 Kindai University, Japan
3 National Cancer Center Research Institute, Japan
4 Center for Medical Genetics, Keio University School of Medicine, Japan

OS-08-7 **Quantification of escape from X chromosome inactivation with the million cell-scale single-cell RNA-seq datasets**

Yoshihiko Tomofuji, Ryuya Edahiro, Yuya Shirai, Kyuto Sonehara, Atsushi Kumanogoh, Yukinori Okada

Osaka University, Japan

OS-08-8 **From Uncertain to Likely: The impact of an update on variant classification**

Norafiza Mohd Yasin, Faidatul Syazlin Abdul Hamid, Nur Aishah Aziz, Ezzanie Suffya Zulkefli, Syahzuwan Hassan, Azian Naila Md Nor, Ermi Neiza Mohd Sahid, Yuslina Mat Yusoff, Ezalia Esa

Hematopathologist, Haematology Unit, Cancer Research Centre, Institute for Medical Research, Malaysia

OS-08-9 **Improving CNV detection performance in microarray data using a machine learning-based approach: Validation with 29,508 Korean population**

Sam Martin, Hyuk-Jung Kwon

EDGC, Korea

Oral Session 9　Cancer Genomics, Somatic

Date　：Saturday, October 14, 2023　8:00 ～ 9:30　Room F（Meeting Room 1, 3F, Zenkoku Toshi Kaikan）
Chairs　：Akihiro Fujimoto（Department of Human Genetics, Graduate School of Medicine, The University of Tokyo, Japan）
Hidenobu Soejima（Division of Molecular Genetics & Epigenetics, Department of Biomolecular Sciences, Faculty of Medicine, Saga University, Japan）

OS-09-1 **Molecular characterization of URST1 as a novel prognostic biomarker and therapeutic target for lung cancer**

Atsushi Takano[1], Yohei Miyagi[3], Yataro Daigo[1,2]

1 Shiga University of Medical Science, Japan
2 Institute of Medical Science, The University of Tokyo, Japan
3 Kanagawa Cancer Center, Japan

OS-09-2 **Pan-cancer analysis of HRD score using whole-genome sequencing for non-pathogenic variant cases**

Akane Naruoka, Masakuni Serizawa, Keiichi Ohshima, Yasue Horiuchi, Takeshi Nagashima, Keiichi hatakeyama, Yuji Shimoda, Sumiko Ohnami, Shumpei Ohnami, Kenichi Urakami, Yasuto Akiyama, Ken Yamaguchi

Shizuoka Cancer Center Research Institute, Japan

OS-09-3

Oncogenic EGFR signal induces histone deacetylation at a putative common enhancer region near CXCL9/10/11 gene loci

Hidetoshi Sumimoto[1], Atsushi Takano[1,2], Koji Teramoto[1], Yataro Daigo[1,2]

1 Shiga University of Medical Science, Japan
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OS-09-4

Genomic nomenclature for the accurate description of pathogenic variation

Rosalind Hastings[1], Melody Tabiner[2], Mark Sales[1], Fiona Morgan[1], Zandra Deans[1]

1 NHS Lothian, UK
2 Oxford University Hospitals NHS Foundation Trust, UK

OS-09-5

Expression of HJURP regulates oral cancer cell proliferation through regulation of downstream genes

Bayarbat Tsevegjav[1], Atsushi Takano[1,2], Yoshihiro Yoshitake[3], Masanori Shinohara[3], Yataro Daigo[1,2]

1 Shiga University of Medical Science, Japan
2 The University of Tokyo, Japan
3 Kumamoto University, Japan

OS-09-6

Molecular characterization of URST7 as a new prognostic biomarker and therapeutic target for breast cancer subtypes

Regina Mbugua[1], Atsushi Takano[1,2], Bayarbat Tsevegjav[1], Yohei Miyagi[3], Yataro Daigo[1,2]

1 Shiga University of Medical Science, Japan
2 The University of Tokyo, Japan
3 Kanagawa Cancer Center, Japan

OS-09-7

The regulation of transforming growth factor-beta by hypoxia in tumor-associated macrophages

Koji Teramoto, Hidetoshi Sumimoto, Yataro Daigo

Shiga University of Medical Science, Japan

OS-09-8

Genome-wide analysis shows liver cancers lacking defined etiologies are enriched for tobacco exposure related mutations

Todd Johnson[1,2], Shota Sasagawa[1,2], Kazuhiro Maejima[1,2], Masaki Ueno[3], Kazuaki Chayama[4], Atsushi Ono[4], Hisashi Kosaka[5], Masashi Kaibori[5], Hidewaki Nakagawa[1,2]

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3 Second Department of Surgery, Wakayama Medical University, Wakayama, Japan
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5 Department of Surgery, Kansai Medical University, Osaka, Japan

OS-09-9

The epigenetic and cancer precision medicine: Plasticity of noncoding RNA in nasopharyngeal carcinoma in Indonesia

Sofia Harjana[1], Dicka Setyosari[1], Tirta Wardana[2], Cita Herawati[3]

1 Faculty of Medicine Public Health and Nursing, Universitas Gadjah Mada, Yogyakarta, Indonesia
2 Faculty of Medicine UNSOED, Purwokerto, Indonesia
3 Faculty of Medicine Universitas Gunadarma, Jakarta, Indonesia

Oral Session 10　Cancer Genomics, Germline

Date　：Saturday, October 14, 2023　13:40 ～ 15:10　Room F（Meeting Room 1, 3F, Zenkoku Toshi Kaikan）
Chairs　：Akira Hirasawa（Department of Clinical Genomic Medicine, Okayama University, Japan）
Arisa Ueki（Cancer Institute Hospital, Clinical Genetic Oncology, Japan）

OS-10-1　Case-control analysis for germline PTEN variants indicates their associations with endometrial and breast cancer risk

Yuki Kanazashi[1,3], Yusuke Iwasaki[4], Yoichiro Kamatani[5], Koichi Matsuda[6], Yoshinori Murakami[7], Hidewaki Nakagawa[2], Yukihide Momozawa[4]

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4 Laboratory for Genotyping Development, RIKEN Center for Integrative Medical Sciences, Yokohama, Japan
5 Laboratory of Complex Trait Genomics, Department of Computational Biology and Medical Sciences, Graduate School of Frontier Sciences, The University of Tokyo, Tokyo, Japan
6 Laboratory of Clinical Genome Sequencing, Department of Computational Biology and Medical Sciences, Graduate School of Frontier Sciences, The University of Tokyo, Tokyo, Japan
7 Division of Molecular Pathology, Department of Cancer Biology, Institute of Medical Science, The University of Tokyo, Tokyo, Japan

OS-10-2　Mismatch repair deficient endometrial cancers and Lynch syndrome in a large cohort study

Mayuko Goda, Shingo Miyamoto, Saki Aota, Asumi Misawa, Hirofumi Inaba, Sho Mizuno, Akira Kawata, Yurina Suzuki, Koji Horie, Kiwamu Akagi

Saitama Prefectual Cancer Center, Japan

OS-10-3　Utility of multi-gene panel testing for patients with breast / ovarian cancer

Yuko Minoura[1], Keika Kaneko[1], Hiromi Arakawa[1], Eri Habano[1], Asami Kuga[1], Naomi Hayashi[1], Akito Dobashi[2], Takahiro Kogawa[1], Arisa Ueki[1]

1 The Cancer Institute Hospital, Japanese Foundation for Cancer Research, Japan
2 The Cancer Institute, Japanese Foundation for Cancer Research, Japan

OS-10-4　The pathogenic role of the BRCA2 c.7847C>T (p.Ser2616Phe) variant in breast and ovarian cancer predisposition

Kazuki Yamazawa[1], Kokichi Sugano[2,3], Kohji Tanakaya[4], Satomi Inoue[1], Haruka Murakami[1], Moeko Nakashima[1], Masataka Adachi[11], Shinya Oki[11], Takeshi Makabe[11], Hiroshi Yamashita[11], Arisa Ueki[5], Tatsuo Matsunaga[1], Takayuki Kinoshita[12], Masami Arai[6], Seigo Nakamura[7], Hiroaki Miyata[8], Masachika Ikegami[9,10], Hiroyuki Mano[9], Shinji Kohsaka[9], Akira Matsui[12]

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2 Department of Genetic Medicine, Sasaki Foundation, Kyoundo Hospital, Japan
3 Department of Genetic Medicine and Services, National Cancer Center Hospital, Japan
4 Department of Surgery, National Hospital Organization Iwakuni Clinical Center, Japan
5 Division of Clinical Genetic Oncology, Cancer Institute Hospital, Japanese Foundation for Cancer Research, Japan
6 Department of Clinical Genetics, Juntendo University, Graduate School of Medicine, Japan
7 Division of Breast Surgical Oncology, Department of Surgery, Showa University School of Medicine, Japan
8 Department of Health Policy and Management, Keio University School of Medicine, Japan
9 Division of Cellular Signaling, National Cancer Center Research Institute, Japan
10 Department of Musculoskeletal Oncology, Tokyo Metropolitan Cancer and Infectious Diseases Center Komagome Hospital, Japan
11 Department of Obstetrics and Gynecology, National Hospital Organization Tokyo Medical Center, Japan
12 Department of Breast Surgery, National Hospital Organization Tokyo Medical Center, Japan

OS-10-5

Pan-cancer and cross-population genome-wide association studies dissect shared genetic basis underlying carcinogenesis

Go Sato[1], Yuya Shirai[1,2], Shinichi Namba[1], Ryuya Edahiro[1], Kyuto Sonehara[1,3,4], Tsuyoshi Hata[1], Mamoru Uemura[1], Biobank Japan Project[5], Koichi Matsuda[6], Yuichiro Doki[1], Hidetoshi Eguchi[1], Yukinori Okada[1,2,3,4]

1 Osaka University Graduate School of Medicine, Japan
2 Immunology Frontier Research Center (WPI-IFReC), Osaka University, Japan
3 Graduate School of Medicine, The University of Tokyo, Japan
4 RIKEN Center for Integrative Medical Sciences, Japan
5 Institute of Medical Science, The University of Tokyo, Japan
6 Graduate School of Frontier Sciences, The University of Tokyo, Japan

OS-10-6

BRCA testing and Japanese testing criteria for hereditary breast and ovarian cancer covered by public health insurance

Kouji Ohta, Tomoko Itoh, Jun Katoh, Ikue Hata

Fukui Prefectural Hospital, Japan

OS-10-7

Functional analysis of the 1p34 risk locus implicates GNL2 in high-grade serous ovarian cancer

Koji Nakamura, Reina Komatsu, Nao Wakui, Airi Kuruma, Sakaaki Machimura, Mariya Kobayashi, Mai Koizumi, Hitomi Sakaguchi, Tadashi Oride, Saori Tsuji, Sohmi Kin, Takeshi Goto, Tadashi Kimura

Osaka University, Japan

OS-10-8

Profiles of cases showing constitutive hypermethylation of the MLH1 promoter region

Kokichi Sugano[1], Miho Ando[1], Sayuri Hiraoka[1], Yasuyuki Miyakura[2], Moriya Iwaizumi[3], Takeshi Kuwata[4], Tadashi Nomizu[5], Yosuke Katsube[5], Shozo Osumi[6], Noriko Tanabe[7], Tomoko Watanabe[7], Makoto Hirata[7], Teruhiko Yoshida[7]

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2 Saitama Medical Center, Jichi Medical University, Japan
3 Clinical and Molecular Genetics Center, Hamamatsu University School of Medicine, Japan
4 Department of Pathology and Clinical Laboratories, National Cancer Center Hospital East, Japan
5 Department of Surgery, Breast Cancer Center, Familial Tumor Clinic, Hoshi General Hospital, Japan
6 Department of Hereditary Tumors, National Hospital Organization, Shikoku Cancer Center, Japan
7 Department of Genetic Medicine and Services, National Cancer Center Hospital, Japan

OS-10-9

Identification of novel genetic risk factors for pancreatic cancer by SNP functional annotation

Federico Canzian, on behalf of the PANDoRA consortium

Genomic Epidemiology Group German Cancer Research Center(DKFZ), Germany

Oral Session 11　Asian Genetics

Date　：Saturday, October 14, 2023　8:00 ～ 9:30　Room G（Meeting Room 2, 3F, Zenkoku Toshi Kaikan）
Chairs　：Taisei Mushiroda（RIKEN Center for Integrative Medical Sciences, Japan）
Emiko Noguchi（University of Tsukuba, Japan）

OS-11-1

Divergence of haplogroup C clade in Y-chromosome among the Japanese population group

Motoki Osawa[1], Zhang Ruogu[1], Eriko Ochiai[2], Atsushi Ueda[1]

1 Tokai University School of Medicine, Japan
2 Kitasato University School of Medicine, Japan

OS-11-2

Withdrawn

OS-11-3

Distinct clinical effects of two RP1L1 hotspots of Miyake disease; Identification of genotype by deep learning

Yu Fujinami-Yokokawa[1,2,3,4], Kwangsic Joo[5], Xiao Liu[1,6,7], Lizhu Yang[13], Kazushige Tsunoda[8], Mineo Kondo[9], Seong Joon Ahn[10], Satomi Inoue[11], Kazuki Yamazawa[11], Tatsuo Matsunaga[11], Izumi Naka[12], Jun Ohashi[12], Hisateru Tachimori[14], Hiroaki Miyata[2], Ruifang Sui[13], Se Joon Woo[5], Kaoru Fujinami[1,3,15]

1 National Institute of Sensory Organs, National Hospital Organization, Tokyo Medical Center, Japan
2 Department of Health Policy and Management, Keio University School of Medicine, Tokyo, Japan
3 UCL Institute of Ophthalmology, London, UK
4 Division of Public Health, Yokokawa Clinic, Suita, Japan
5 Department of Ophthalmology, Seoul National University Bundang Hospital, Seoul National University College of Medicine, Seongnam, Republic of Korea
6 Southwest Hospital, Army Medical University, Chongqing, China
7 Key Lab of Visual Damage and Regeneration & Restoration of Chongqing, China
8 Division of Vision Research, National Institute of Sensory Organs, National Hospital Organization Tokyo Medical Center, Tokyo, Japan
9 Department of Ophthalmology, Mie University Graduate School of Medicine, Mie, Japan
10 Department of Ophthalmology, Hanyang University Hospital, Hanyang University College of Medicine, Seoul, Republic of Korea
11 Department of Medical Genetics, National Hospital Organization Tokyo Medical Center, Tokyo, Japan
12 Department of Biological Sciences, Graduate School of Science, The University of Tokyo, Japan
13 Department of Ophthalmology, Peking Union Medical College Hospital, Peking Union Medical College and Chinese Academy of Medical Sciences, Beijing, China
14 Endowed Course for Health System Innovation, Keio University School of Medicine, Tokyo, Japan
15 Moorfields Eye Hospital, London, UK

OS-11-4

CXCL12-CXCR4 pathway as an novel therapeutic target for RNF213-associated vasculopathy

Takahiro Hiraide[1], Hisato Suzuki[2], Mizuki Momoi[1], Yoshiki Shinya[1], Kenjiro Kosaki[1], Masaharu Kataoka[3]

1 Keio University School of Medicine, Japan
2 Tsukuba University School of Medicine, Japan
3 University of Occupational and Environmental Health, Japan

OS-11-5

Yayoi Genome from the Doigahama site provides insights into the origins of immigrants to the Japanese archipelago

Jonghyun Kim[1], Fuzuki Mizuno[2], Izumi Naka[1], Masami Matsushita[3], Takayuki Matsushita[4], Shintaroh Ueda[1,2], Kunihiko Kurosaki[2], Jun Ohashi[1]

1 Department of Biological Sciences, Graduate School of Science, The University of Tokyo, Tokyo, Japan
2 Department of Legal Medicine, Toho University School of Medicine, Japan
3 The Organization of Anthropological Research, Japan
4 Doigahama Site Anthropological Museum, Japan

OS-11-6

Gene expression profile of cytokines (IL-6, TNF-alpha and TGF-beta1) in Malay male subjects with chronic HCV infection

Imran Ahmad[1], Ismail Che Noh[2], Richard Avoi[2], Nurul Asma Abdullah[1], Ruzilawati Abu Bakar[1]

1 Universiti Sains Malaysia, Malaysia
2 Universiti Malaysia Sabah, Malaysia

OS-11-7

Occult Macular Dysfunction Syndrome: Identification of multiple causative genes of macular dysfunction with normal fundus

Kaoru Fujinami[1,2,3], Yu Fujinami-Yokokawa[1,3,4], Lishu Yang[5], Kwangsic Joo[6], Kazushige Tsunoda[1], Xiao Liu[1,9], Mineo Kondo[7], Izumi Naka[8], Jun Ohashi[8], Satomi Inoue[2], Kazuki Yamazawa[2], Tatsuo Matsunaga[1,2], Hisateru Tachimori[10], Hiroaki Miyata[4], Se Joon Woo[6], Ruifang Sui[5]

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3 UCL Institute of Ophthalmology, London, UK
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7 Department of Ophthalmology, Mie University Graduate School of Medicine, Mie, Japan
8 Department of Biological Sciences, Graduate School of Science, The University of Tokyo, Tokyo, Japan
9 Southwest Hospital, Army Medical University, Chongqing, China
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OS-11-8

Establishment of the pharmacogene variation database in 967 Japanese individuals using corePGseq panel

Koya Fukunaga, Taisei Mushiroda

RIKEN, Japan

OS-11-9

The characterisation of clinical genomic variants in an Asian population and implications for genetic counselling

Yasmin Bylstra[1], Sock Hoai Chan[2], Jing Xian Teo[1], Sonia Davila[1,3], David Amor[4,5], Melody Menezes[4,6], Jan Hodgson[4], Joanne Ngeow[2,8], Patrick Tan[1,7,9], Saumya S. Jamuar[1,10,12], Weng Khong Lim[1,11]

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2 Cancer Genetics Service, National Cancer Centre, Singapore
3 Cardiovascular and Metabolic Disorders, Duke-NUS Medical School, Singapore
4 Department of Paediatrics, The University of Melbourne, Victoria, Australia
5 Murdoch Children's Research Institute, Victoria, Australia
6 Monash Ultrasound for Women, Victoria, Australia
7 Genome Institute of Singapore, Singapore
8 Lee Kong Chian School of Medicine, Nanyang Technological University, Singapore
9 Precision Health Research Singapore (PRECISE), Singapore
10 Genetics Service, KK Women's and Children's Hospital, Singapore
11 Cancer and Stem Cell Biology, Duke-NUS Medical School, Singapore
12 Paediatric Academic Clinical Programme, Duke-NUS Medical School, Singapore

Oral Session 12　Pediatric Genetics

Date　：Saturday, October 14, 2023　13:40 ～ 15:00　Room G（Meeting Room 2, 3F, Zenkoku Toshi Kaikan）
Chairs　：Wuh-Liang Hwu（National Taiwan University Hospital and China Medical University Hospital, Taiwan）
Noriko Miyake（Department of Human Genetics, Research Institute, National Center for Global Health and Medicine, Japan）

OS-12-1

Exploring the clinical utility of targeted MECP2 testing in real-world practice

Soo Yeon Kim[1], Seoyun Jang[2], Jae So Cho[1], Jihoon Yoon[1], Seungbok Lee[1], Man Jin Kim[1], Hyewon Woo[3], Byung Chan Lim[2], Jong Hee Chae[1,2]

1 Department of Genomic Medicine, Seoul National University Hospital, Korea
2 Department of Pediatrics, Seoul National University Children's Hospital, Korea
3 Department of Pediatrics, Chungbuk National University Hospital, Korea

OS-12-2

The impact of LECT2, α-SMA, and COL1A1 expressions on liver fibrogenesis in biliary atresia patients post Kasai surgery

Fiqih Vidiantoro Halim[1], Gunadi Gunadi[1], Diaz Adi Pradana[1], Tiara Putri Leksono[1], Laudria Stella Eryvinka[1], Adisrasti Rejeki Amaragati[1], Kristy Iskandar[2], Akhmad Makhmudi[1]

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2 Department of Child Health, Faculty of Medicine, Public Health and Nursing, Universitas Gadjah Mada / UGM Academic Hospital, Yogyakarta, Indonesia

OS-12-3

Male-dominant SHOX expression in cartilage tissues: implications for sex differences in adult height

Atsushi Hattori[1], Atsuhito Seki[2], Kazuhiko Nakabayashi[1], Yasuhiro Naiki[2], Akie Nakamura[1], Keisuke Ishiwata[1], Kenji Matsumoto[1], Kohji Okamura[1], Katoh-Fukui Yuko[1], Ogata Tsutomu[3,4], Kagami Masayo[1], Maki Fukami[1]

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2 National Center for Child Health and Development, Japan
3 Hamamatsu University School of Medicine, Japan
4 Hamamatsu Medical Center, Japan

OS-12-4

Development of an automatic interpretation tool for copy-number variants

Chiaki Hosaka[1,2], Kenichiro Kori[1,2], Satoshi Yuhara[1,2]

1 SRL, Inc., Japan
2 H.U. Group Research Institute, LLC, Japan

OS-12-5

Comprehensive genetic analysis of 9 families with hereditary Wolff-Parkinson-White syndrome

Hidenori Yamamoto[1], Hidehito Inagaki[2], Kiyotaka Go[1], Yoshihito Morimoto[1], Yoshie Fukasawa[1], Hiroko Goto[3,4], Sayaka Mii[5], Hiroki Kurahashi[2], Taichi Kato[1]

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3 Department of Pediatric Cardiology, Nagoya Tokushukai General Hospital, Japan
4 Department of Pediatric Cardiology, Gifu Prefectural General Medical Center, Japan
5 Department of Pediatric Cardiology, Japanese Red Cross Aichi Medical Center Nagoya Daiichi Hospital, Japan

OS-12-6

Rare sequence variants associated with the risk of non-syndromic biliary atresia

Satoshi Tamaoka[1,3], Akinari Fukuda[2], Yuki Muranishi[1], Kazuhiko Nakabayashi[1], Keiko Matsubara[1,2], Hiroko Ogata-Kawata[1], Kenichiro Hata[1], Yuko Katoh-Fukui[1], Seisuke Sakamoto[2], Mureo Kasahara[2], Maki Fukami[1]

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2 National Center for Child Health and Development, Japan
3 Keio University School of Medicine, Japan

OS-12-7

Comprehensive molecular and clinical analysis in 30 cases with multilocus imprinting disturbance

Tatsuki Urakawa[1,2], Kaori Yamoto[3], Kaori Hara-Isono[1], Keiko Matsubara[1], Maki Fukami[1], Shinji Saitoh[4], Hidenobu Soejima[5], Tsutomu Ogata[1,3], Masayo Kagami[1]

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3 Dept. of Biochem., Hamamatsu Univ. Sch. of Med., Japan
4 Dept., of Pediatr. and Neonatol., Nagoya City Univ. Grad. Sch. of Med., Japan
5 Division of Mol. Genetics and Epigenetics, Dept. of Biomol. Sciences, Faculty of Med., Saga Univ., Japan

OS-12-8

GATA2 expressions and methylation pattern in multifactorial Hirschsprung disease

Kevin Eliezer Ferdinandus[1], Alvin Santoso Kalim[1], Nova Yuli Prasetyo Budi[1], Laudria Stella Eryvinka[1], Setiani Silvy Nurhidayah[1], Kristy Iskandar[2], Dwi Aris Agung Nugrahaningsih[3], Gunadi Gunadi[1]

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3 Department of Pharmacology and Therapy / Genetics Working Group, Faculty of Medicine, Public Health and Nursing, Universitas Gadjah Mada, Yogyakarta, Indonesia

Oral Session (Japanese)

Oral Session (Japanese)　Gentic Counseling in Asia

Japanese Session

Date　：Thursday, October 12, 2023　10:10 ～ 11:40　Room G（Meeting Room 2, 3F, Zenkoku Toshi Kaikan）
Chairs　：Shinji Kosugi（Kyoto University, Japan）
Akihiro Sakurai（Department of Medical Genetics and Genomics, Sapporo Medical University School of Medicine, Japan）

OJ-1 Preliminary screening for hereditary breast and ovarian cancer using a Chatbot: Interview survey in a Clinical Setting

Sato Ann[1], Eri Haneda[1], Yukihiko Hiroshima[2,3], Hiroto Narimatsu[1,4,5]

1 Department of Genetic Medicine, Kanagawa Cancer Center, Yokohama, Kanagawa, Japan
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4 Cancer Prevention and Cancer Control Division, Kanagawa Cancer Center Research Institute, Yokohama, Kanagawa, Japan
5 Graduate School of Health Innovation, Kanagawa University of Human Services, Kawasaki, Kanagawa, Japan

OJ-2 Changes in trends in genetic nursing practice in breast and gynecologic oncology over the past five

Hiromi Moriya[1], Masako Shomura[1], Naho Yaguchi[1], Banri Tsuda[1], Tetsuya Urano[1], Yoshiro Yamamoto[2]

1 Tokai University School of Medicine, Japan
2 Tokai University School of Science, Japan

OJ-3 A case of hyperkalemic periodic paralysis with self-interpretation of the illness

Hisatsugu Tachibana[1], Yuka Hattori[2], Yuka Yotsumoto[2], Tomoko Tamaoki[3]

1 Department of Neurology, Takatsuki General Hospital, Japan
2 Department of Pediatrics, Takatsuki General Hospital, Japan
3 Center for Clinical and Molecular Genetics, Takatsuki General Hospital, Japan

OJ-4 Challenges in supporting perinatal decision for parents of children with de novo monogenic disorders

Fumi Kurebayashi[1,2], Kousuke Yamada[1], Takakazu Kawamura[3], Kenji Shimizu[1]

1 Shizuoka Children's Hospital, Japan
2 Department of Medical Genetics, Shizuoka General Hospital, Japan
3 Perinatal Medical Center, Shizuoka Children's Hospital, Japan

OJ-5 Charcot-Marie-Tooth disease type IA revealed by chromosomal microarray testing in a patient with Miller-Dieker syndrome

Reimi Sogawa[1], Tomoyuki Akiyama[2], Eriko Eto[3], Mashu Futagawa[1], Fumino Kato[1], Hideki Yamamoto[1,4], Akira Hirasawa[1,4], iori Ohmori[5], Katsuhiro Kobayashi[2]

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3 Department of Obstetrics and Gynecology, Okayama University Graduate School of Medicine, Dentistry and Pharmaceutical Sciences, Japan
4 Department of Clinical Genomic Medicine, Okayama University Graduate School of Medicine, Dentistry and Pharmaceutical Sciences, Japan
5 Section of Developmental Physiology and Pathology, Faculty of Education, Okayama University, Japan

OJ-6 A case of pre-symptomatic testing and genetic counseling on infantile metachromatic leukodystrophy

Motomichi Kosuga, Tetsumin So, Satoko Tsushima, Yasuyuki Fukuhara, Rika Kosaki, Torayuki Okuyama, Haruhiko Sago

National Center for Child Hearth and Development, Japan

OJ-7 **Parents' knowledge, anxiety, and understanding regarding genetic testing for children with hearing loss**

Kayono Yamamoto[1], Yumiko Kobayashi[1], Akimune Fukushima[1], Mari Urano[2], Fumie Aizawa[1]

1 Iwate Medical University, Japan
2 Tokyo Women's Medical University, Japan

OJ-8 **A case of an unaffected female BRCA1 pathogenic variant carrier (previvor) who expressed guilty feeling to the proband**

Hiromi Sugawara, Megumi Mukai, Sadia Matsutani, Miyuki Kawamura, Takashi Shibutani, Saki Hinoshita, Kazuo Tamura, Koji Matsumoto

Hyogo Cancer Center, Japan

Poster Session

Poster Session 1-01 Complex Diseases and Genomic Risk Assessment 1

Date ：Thursday, October 12, 2023 17:30 ～ 18:00 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P1-01-1 **Family history of developmental dysplasia of the hip is a risk factor for the progression of hip osteoarthritis**

Soichiro Yoshino[1,2], Ryosuke Yamaguchi[2], Hidenao Tanaka[2], Shiro Ikegawa[1], Yasuharu Nakashima[2], Chikashi Terao[1]

1 RIKEN, Japan
2 Graduate School of Medical Sciences, Kyushu University, Japan

P1-01-2 **The impact of Collagen gene cluster expressions on liver fibrogenesis in biliary atresia**

Donny Aditia, Dyah Ayu Puspitarani, Khanza Adzkia Vujira, Fadila Dyah Trie Utami, Fiqih Vidiantoro Halim, Kristy Iskandar, Akhmad Makhmudi, Gunadi Gunadi

Universitas Gadjah Mada, Indonesia

P1-01-3 **High frequency of MYBPC3 copy number variations in patients with hypertrophic cardiomyopathy**

Seiko Ohno[1], Koichi Kato[2], Erina Ozaki[3], Keiko Sonoda[1], Shigehiro Miyazaki[3], Mariko Eguchi[3], Yoshihiko Ikeda[1], Osamu Yamaguchi[3], Takeshi Aiba[1]

1 National Cerebral and Cardiovascular Center, Japan
2 Shiga University of Medical Science, Japan
3 Ehime University Graduate School of Medicine, Japan

P1-01-4 **A glance into the genetics of body dysmorphic disorder (BDD)**

Mohammad Hilal Atthariq Ramadhan[1], Ziske Maritska[2,3], Bintang Arroyantri Prananjaya[4], Nita Parisa[5]

1 Faculty of Medicine, Universitas Sriwijaya, Indonesia
2 Department of Biology Medicine, Faculty of Medicine, Universitas Sriwijaya, Palembang, Indonesia
3 Indonesian Society of Genetic Counselors (ISGC), Indonesia
4 Department of Psychiatry, Faculty of Medicine, Universitas Sriwijaya, Palembang, Indonesia
5 Department of Pharmacology, Faculty of Medicine, Universitas Sriwijaya, Palembang, Indonesia

P1-01-5 **Molecular genetic analysis of dyslipidemia in Iran**

Nejat Mahdieh[1], Bahareh Rabbani[2], Mohadesh Alimoghadam[1], Shiva Esmaeili[2], Sara Nematolahi[2], Leila Hejazi[1]

1 Rajaie Cardiovascular Research Center, Iran
2 Growth and Development Research Center, Ogene, Iran

P1-01-6 **Genome-wide association study identifies candidate loci associated with postoperative nausea and/or vomiting**

Daisuke Nishizawa[1], Ryozo Morino[2], Miyuki Yokota[3,4], Seii Ohka[1], Shinya Kasai[1], Junko Hasegawa[1], Yuko Ebata[1], Kyoko Nakayama[1], Kazutaka Ikeda[1]

1 Tokyo Metropolitan Institute of Medical Science, Japan
2 Koujinkai Daiichi Hospital, Japan
3 Cancer Institute Hospital, Japan
4 East Hokkaido Hospital, Japan

P1-01-7 **Clinical characteristics & hearing impairment in mitochondrial DNA mutation**

Natsumi Uehara, Takeshi Fujita, Hikari Shimoda, Sayaka Katsunuma, Ryosuke Bo, Akinobu Kakigi, Akiharu Kubo, Ken-ichi Nibu

Kobe University Graduate School of Medicine, Japan

P1-01-8 **Eosinophilic gastroenteritis in a Noonan syndrome patient with PTPN11 variation**

Nobuhiko Koga, Takahito Inoue, Kei Kubota, Toshikazu Niimi, Shuuichi Yatsuga, Shinichirou Nagamitsu

Fukuoka University, Japan

P1-01-9 **Characterization of a dicentric Y chromosome due to complex rearrangements in pseudoautosomal region 1**

Yasuko Ogiwara[1,2], Atsushi Hattori[1,3], Mami Miyado[1], Yoshitomo Kobori[5,6,7], Yoko Kuroki[3,4,8], Maki Fukami[1,3]

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P1-01-10 **Genome-to-genome analysis to identify specific host-pathogen genetic interaction points in tuberculosis**

Yosuke Omae[1], Licht Toyo-Oka[2], Hideki Yanai[3], Reiko Miyahara[1], Jody Phelan[4], Paula Josefina Gomez-Gonzalez[4], Nuria Andreu[4], Supalert Nedsuwan[5], Paola Florez de Sessions[6], Susana Campino[4], Neneh Sallah[4], Julian Parkhill[7], Nat Smittipat[8], Prasit Palittapongarnpim[8], Taisei Mushiroda[9], Michiaki Kubo[9], Surakameth Mahasirimongkol[10], Martin L. Hibberd[4], Taane G. Clark[4], Katsushi Tokunaga[1]

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3 Japan Anti-Tuberculosis Association, Japan
4 London School of Hygiene and Tropical Medicine, UK
5 Chiangrai Prachanukroh Hospital, Thailand
6 Genomics Institute Singapore, Singapore
7 University of Cambridge, UK
8 National Science and Technology Development Agency, Thailand
9 RIKEN Center for Integrative Medical Sciences, Japan
10 Ministry of Public Health, Japan

P1-01-11 **Situation analysis of access to genetic services among female Thai patients with breast Cancer**

Chayaluck Siripukdeekan, Acharaporn Seeherunwong

Mahidol University, Thailand

P1-01-12 **Evaluating mediation effect of obesity on the relationship between polygenic scores and comorbidities: TMM CommCohort**

Yoichi Sutoh, Tsuyoshi Hachiya, Yayoi Otsuka-Yamasaki, Shohei Komaki, Shiori Minabe, Hideki Ohmomo, Makoto Sasaki, Atsushi Shimizu

Iwate Medical University, Japan

P1-01-13 **Genome-wide association analysis and polygenic risk score model for predicting paroxysmal atrial fibrillation**

Megumi Shiomi[1], Yuki Nagata[1,2], Takeaki Sudo[3], Takamasa Ichikawa[2], Kensuke Ihara[4], Ken Asada[5], Yasuaki Tanaka[6], Yasuteru Yamauchi[7], Takeshi Sasaki[8], Hitoshi Hachiya[9], Yasushi Imai[10], Hideo Fujita[11], Tetsuo Sasano[12], Tetsushi Furukawa[4], Toshihiro Tanaka[1,2]

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7 Department of Cardiology, Japan Red Cross Yokohama City Bay Hospital, Kanagawa, Japan
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Poster Session 1-02 Inherited Metabolic Diseases and Newborn Screening 1

Date ：Thursday, October 12, 2023 17:30 ～ 18:00 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P1-02-1 **Withdrawn**

P1-02-2 **Successful diagnosis of Sengers Syndrome using comprehensive genomic analysis**

Kohta Nakamura[1], Yukiko Yatsuka[1], Atsuko Imai-Okazaki[1], Sachie Naito[3], Akira Hasegawa[4], Takeya Kasukawa[4], Atsushi Kondo[4], Yohei Suigyama[2,6], Tomoko Tsuruoka[5], Tomohiro Ebihara[6], Takanori Onuki[6], Keiko Ichimoto[6], Akira Ohtake[7,8], Kei Murayama[1,6], Yasushi Okazaki[1,4]

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6 Department of Metabolism, Chiba Children's Hospital, Japan
7 Department of Pediatrics & Clinical Genomics Diseases, Saitama Medical University, Japan
8 Center for Intractable Diseases, Saitama Medical University Hospital, Japan

P1-02-3 **Next generation sequencing in Japanese patients with Maturity-Onset Diabetes of the young**

Satoshi Tanaka, Naoko Iwasaki, Kenko Azuma, Sayaka Higuchi, Hiroyuki Akagawa, Shohei Mitani

Tokyo Women's Medical University, Japan

P1-02-4 **Discrepancy in insurance covered LDLR genetic testing in familial hypercholesterolemia: A case report**

Keiko Goto-Hirano[1,2], Hidetaka Eguchi[3], Yasushi Okazaki[3], Atsuko Okazaki[3], Masami Arai[1], Takao Kato[2], Hirotoshi Ohmura[2], Hiroyuki Daida[2,4], Tohru Minamino[2]

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4 Faculty of Health Sciences, Juntendo University, Japan

P1-02-5 **Phenotype and genotype of vietnamese patients with combined pitutary hormone deficiency**

Thu Ha Nguyen, Chi Dung Vu, Phuong Thao Bui, Ngoc Khanh Nguyen, Thi Bich Ngoc Can

Vietnam National Children's Hospital, Department of Pediatric Endocrinology and Diabetes, Center for Endocrinology, Metabolism, Genetics / Genomics and Molecular Therapy, Vietnam

P1-02-6 **Pre-liver transplantation rapid genetic diagnosis in a patient with Wilson disease**

Tomomi Yamaguchi[1,2,3], Tomomi Fujikawa[3], Yuri Takiguchi[3], Akiko Sakyu[1], Atsuyoshi Mita[4], Yuji Soejima[4], Tomoki Kosho[1,2,3,5]

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Poster Session 1-03 Prenatal Genetics 1

Date ：Thursday, October 12, 2023 17:30 ～ 18:00 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P1-03-1 **Chromosome analysis of 3PN and 2.1PN embryos by NGS**

Daisuke Nakajima[1,2], Shohei Komemoto[1], Youichi Takemoto[1], Motoi Nagayoshi[1], Yuya Makino[1,3], Izumi Tanaka[1], Atsushi Tanaka[1]

1 Saint Mother Clinic, Japan
2 Occupational and Environmental Health University School of Medicine, Japan
3 Juntendo University School of Medicine, Japan

P1-03-2 **Analysis of clients' evaluation after launching an online disclosure of prenatal counseling results**

Koichiro Kido, Miho Aoki, Noriko Nakabayashi, Chikara Kihira, Miki Nishizawa, Keita Yatsuki, Hedeo Kamata, Haruko Hiraike, Yukifumi Sasamori, Eiji Ryo, Hidemi Okishio, Masakazu Mimaki, Kiyotaka Watanabe, Kazunori Nagasaka

Teikyo University, Japan

P1-03-3 **Transitions of background for non-invasive perinatal testing; a single center analysis**

Kyoko Kumagai[1], Nobuhiro Suzumori[1], Eri Takeda[1], Ayano Otani[1], Shinobu Goto[1], Iku Taguchi[1,2], Kiwa Yamaoka[1,3], Rin Sato[1,3], Ayako Tanabe[1,4], Mayumi Sugiura[1]

1 Nagoya City University Hospital, Japan
2 National Hospital Organization Nagoya Medical Center, Japan
3 Nagoya Ekisaikai Hospital, Japan
4 Toyota Memorial Hospital, Japan

P1-03-4 **Prenatal diagnosis of true fetal mosaicism: Four cases report**

Nobuhiko Hayashi, Momoko Kato

The Fetal Clinic Tokyo-bay Makuhari, Japan

P1-03-5 **Gene signatures in genetically homogeneous trisomy 21 and euploid at the induced pluripotent stem cell level**

Ryotaro Hashizume[1], Hiroshi Imai[1], Sachiko Wakita[1], Mari Hara[1], Hiroki Kurahashi[2]

1 Mie University, Japan
2 Fujita Health University, Japan

P1-03-6 **The contribution of monogenic and oligogenic causes to the etiology of idiopathic non-obstructive azoospermia**

Yuki Muranishi[1], Yoshitomo Kobori[2], Yuko Katoh-Fukui[1], Satoshi Tamaoka[1], Atsushi Hattori[1], Kazuhiko Nakabayashi[1], Hiroko Ogata-Kawata[1], Maki Fukami[1]

1 National Research Institute for Child Health and Development, Japan
2 Dokkyo Medical University Saitama Medical Center, Japan

P1-03-7 **Analysis of 19 cases of autosomal recessive genetic disorders consulted for PGT-M**

Yoshiharu Nakaoka[1], Michiko Ammae[1], Naoya Tsuji[1], Haruhisa Konishi[1], Sho Fujiwara[1], Hiroko Yamauchi[1], Naoharu Morimoto[1], Kanako Katsu[1], Yoshiharu Morimoto[2]

1 IVF Namba Clinic, Japan
2 HORAC Grant Front Osaka Clinic, Japan

P1-03-8

A case of osteogenesis imperfecta with possible maternal somatic cell mosaicism detected by deep sequencing

Fuyuki Hasegawa[1], Asuka Hori[2,3], Kousuke Taniguchi[4], Jin Muromoto[1,5], Rika Sugibayashi[1,5], Katsusuke Ozawa[1,5], Seiji Wada[1,5], Haruhiko Sago[1,5], Kenichiro Hata[4]

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P1-03-9

PGT-M of duplicated Duchenne muscular dystrophy suspected to be germline mosaicism

Michiko Ammae[1], Tatsuya Nakano[1], Hiroko Yamauchi[1], Yoshiharu Nakaoka[1], Yoshiharu Morimoto[2]

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2 Sunkaky Medical Corporation HORAC Grand Front Osaka Clonic, Japan

P1-03-10

First successful preimplantation genetic testing for the m.8993T>G mutation in Japan

Yuki Mizuguchi[1,2], Kou Sueoka[1], Suguru Sato[1], Mamoru Tanaka[1]

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2 Nasu Red Cross Hospital, Japan

P1-03-11

Presence of small 21p-21p chromosome in mosaic rob(21;21) carrier

Rie Kawamura[1], Ikumi Moriyama[2], Shunsaku Fujii[3], Takeshi Iwasa[4], Akira Kuwahara[4], Minoru Irahara[4], Ei Yuzawa[5], Hiroki Kurahashi[1,2]

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5 Yuzawa Ladies Clinic, Hirosaki, Japan

P1-03-12

Does application of chromosomal analysis in products of miscarriage effect on mother's grief ?

Chenghua Zhu[1], Yodo Sugishita[2], Ohsuke Migita[3]

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P1-03-13

Chromosome analysis using next generation sequencing (NGS) of vanishing twin after term delivery

Saori Tsuji, Tsuyoshi Takiuchi, Hidemine Honda, Mika Handa, Takeshi Goto, Nao Wakui, Sakaaki Machimura, Airi Kuruma, Aiko Okada, Tatsuya Miyake, Mahiru Kawano, Tadashi Kimura

Osaka University, Japan

Poster Session 1-04 Neurology 1

Date ：Thursday, October 12, 2023 17:30 ～ 18:00 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P1-04-1

Topologically associating domains define the impact of de novo promoter variants on autism spectrum disorder risk

Shota Mizuno[1], Takumi Nakamura[1], Junko Ueda[1], Kurara Honda[1], An-a Kazuno[1], Hirona Yamamoto[1,2], Tomonori Hara[1,3], Atsushi Takata[1,4]

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P1-04-2

Is MECP2 the causative gene of MECP2 duplication syndrome?

Keiko Akahoshi[1], Eiji Nakagawa[2], Jun Minato[1], Yumiko Oono[1], Keiko Wada[1], Michiko Makino[1], Yu-ichi Goto[3,4], Ken Inoue[3]

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2 Department of Child Neurology, National Center of Neurology and Psychiatry, Japan
3 Department of Mental Retardation and Birth Defect Research, Japan
4 Medical Genome Center, National Center of Neurology and Psychiatry, Japan

P1-04-3

Missense and nonsense compound heterozygous ATP1A2 variants in a viable patient with FARIMPD

Shogo Furukawa[1], Mitsuhiro Kato[2], Toshihiro Nomura[4], Noriko Sumitomo[3], Shota Yoneno[5], Mitsuko Nakashima[1], Hirotomo Saitsu[1]

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3 National Center of Neurology and Psychiatry, Japan
4 JA Toride Medical Center, Japan
5 Mie University, Japan

P1-04-4

Cases of Becker muscular dystrophy with nonsense mutations in the dystrophin gene

Yoshiyuki Katayama[1], Tomoko Lee[1], Miki Matsui[1], Sachi Tokunaga[1], Naoko Taniguchi[1], Hideki Shimomura[1], Yoko Yokoyama[2], Yukihiro Noda[3], Jun Matsui[4], Katsuhiko Yoshii[5], Yasuhiko Takeshima[1]

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5 Department of Pediatrics, Chibune General Hospital, Osaka, Japan

P1-04-5

Long-read sequencing analysis of Japanese myotonic dystrophy type 2

Satomi Mitsuhashi[1], Keiji Tachikawa[1], Takashi Kimura[2], Ruriko Kitao[3], Shigehisa Ura[4], Yui Sanpei[8], Homare Funasaka[8], Yoshihisa Yamano[1], Yuta Kochi[9], Laura P.W. Ranum[5], Kinji Ohno[6], Tohru Matsuura[7]

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4 Japanese Red Cross Asahikawa Hospital, Japan
5 McKnight Brain Institute, USA
6 Nagoya University Graduate School of Medicine, Japan
7 Jichi Medical University, Japan
8 Akita University Graduate School of Medicine, Japan
9 Tokyo Medical and Dental University, Japan

P1-04-6

Identification of a new ITPR1 mutation for autosomal dominant cerebellar ataxia

Kinya Ishikawa

Tokyo Med. & Dental Univ., Japan

P1-04-7

The Role of RNA and genome analysis in unraveling a deep intronic variant in a family with Duchenne muscular dystrophy

Sok Kun Tae[1], Meow Keong Thong[1,2], Rifhan Azwani Mazlan[2]

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Poster Session 1-05　Technological Advances, Wet and Dry 1

Date　：Thursday, October 12, 2023　17:30 ～ 18:00　Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P1-05-1 Methylation quantitative trait loci (mQTL) of umbilical cord tissue DNA in a Japanese cohort

Tomoko Takahashi[1,2], Kenichi Sakurai[3], Midori Yamamoto[4], Rieko Takatani[4], Yoichiro Kamatani[2,5], Chisato Mori[4,6]

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5 Laboraotry of Complex Trait Genomics, Department of Computational Biology and Medical Sciences, Graduate School of Frontier Sciences, The University of Tokyo, Japan
6 Department of Bioenvironmental Medicine, Graduate School of Medicine, Chiba University, Japan

P1-05-2 Genotype imputation performance of 3.5KJPNv2 from the Tohoku Medical Megabank Project in a distinct genome cohort

Sachiko Ishida[1], Kaname Kojima[2,3], Ryuichiro Kurata[1], Satoru Ishida[1], Kengo Kinoshita[2,4,5,6]

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2 Tohoku Medical Megabank Organization, Tohoku University, Japan
3 RIKEN Center for Advanced Intelligence Project, Japan
4 Advanced Research Center for Innovations in Next-Generation Medicine, Tohoku University, Japan
5 Graduate School of Information Sciences, Tohoku University, Japan
6 Institute of Development, Aging and Cancer, Tohoku University, Japan

P1-05-3 Efficient dimensionality reduction method for large-scale genomic data analysis

Kazuharu Misawa[1,2]

1 Yokohama City University, Japan
2 Riken AIP Center, Japan

P1-05-4 Exploration of blood-based biomarkers to predict the progression of Alzheimer's disease by RNA-sequencing data analysis

Akiko Yamakawa[1], Risa Mitsumori[1], Mutsumi Suganuma[1], Shintaro Akiyama[1], Shumpei Niida[2], Kouichi Ozaki[1,3], Daichi Shigemizu[1,3]

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3 RIKEN Center for Integrative Medical Sciences, Japan

P1-05-5 Co-infection of polyomavirus and SARS-CoV-2 detected by enrichment-based whole-genome sequencing: A case report

Esensi Geometri[1], Marcellus Marcellus[1], Dyah Ayu Puspitarani[1], Fadila Dyah Trie Utami[1], Kristy Iskandar[5], Hendra Wibawa[4], Mohamad Saifudin Hakim[3], Gunadi Gunadi[2]

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4 Disease Investigation Center Wates, Directorate General of Livestock and Animal Health Services, Ministry of Agriculture, Yogyakarta, Indonesia
5 Department of Child Health / Genetics Working Group, Faculty of Medicine, Public Health and Nursing, Universitas Gadjah Mada / UGM Academic Hospital, Yogyakarta, Indonesia

P1-05-6 miRNAs profiling in a complex multifactorial Hirschsprung disease

. Gunadi[1], Dyah Ayu Puspitarani[1], . Puspitarani[1], William Widitjiarso[1], Dwiki Afandy[1], . Afiahayati[2], Kristy Iskandar[1]

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P1-05-7 **Microbiome of the reproductive tract in the menstrual cycle correlates with IVF outcome**

Mio Fukuoka[1], Mitsutoshi Yamada[1], Reina Ooka[1], Yuichi Matsuzawa[1], Maki Iwai[1], Shintaro Kamijo[1], Jumpei Sasabe[2], Kenji Miyado[3], Wataru Yamagami[1], Mamoru Tanaka[1]

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3 Center for Regenerative Medicine, National Center for Child Health and Development, Japan

P1-05-8 **Identification of pathogenic deep intronic variant and exonic LINE-1 insertion in a patient with Meckel syndrome**

Sachiko Miyamoto[1], Kazuyuki Nakamura[2], Mitsuhiro Kato[3], Mitsuko Nakashima[1], Hirotomo Saitsu[1]

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2 Yamagata University, Japan
3 Showa University School of Medicine, Japan

P1-05-9 **Withdrawn**

P1-05-10 **Uncovering deafness-causing single nucleotide variants in STRC through long-read nanopore sequencing**

Hideaki Moteki[1,2], Shin-ya Nishio[1], Shin-ichi Usami[1]

1 Department of Hearing Implant Sciences, Shinshu University School of Medicine, Japan
2 Department of Clinical Genetics, Aizawa Hospital, Japan

P1-05-11 **Novel non-invasive preimplantation genetic testing for aneuploidy algorithm based on cell-free ncRNA expression profiles**

Tasuku Mariya[1], Takeshi Sugimoto[2], Akira Yanagihara[3], Rie Kawamura[2], Makiko Tsutsumi[2], Hidehito Inagaki[2], Tsuyoshi Saito[1], Akihiro Sakurai[1], Hiroki Kurahashi[2]

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3 OVUS Corporation, Japan

P1-05-12 **Novel genetic variants in pulmonary hypertension via exome sequencing in Koreans**

Jungmin Choi, Moonyoung Lee

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Poster Session 1-06 Hemoglobinpathies

Date ：Thursday, October 12, 2023 17:30 ～ 18:00 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P1-06-1 **Molecular characterization of Alpha-globin genes in the Filipino population**

Catherine Lynn T. Silao[1], Maria Liza T. Naranjo[3,4], Ronnette Anne E. Davila[2], Terence Diane F. Fabella[2], Angelika Claudia A. Balitaan[2], Edward Niño P. Garcia[2], Michael Aeron DT. Cruzat[2], Mark John Girasol[5], Mayceemae M. Barnuevo[2], Carmencita D. Padilla[1], Ernesto dJ. Yuson[4]

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4 Lung Center of the Philippines, Quezon City, Philippines
5 College of Medicine, University of the Philippines, Manila, Philippines

P1-06-2

Prevalence of the most common Beta-Globin gene mutations in Filipino beta-thalassemia patients

Catherine Lynn T. Silao[1], Maria Liza T. Naranjo[3,4], Ronnette Anne E. Davila[2], Terence Diane F. Fabella[2], Thomas Gabriel H. Desengaño[2], Carl Angelo S. Estrada[2], Carmencita D. Padilla[1], Ernesto dJ. Yuson[4]

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3 National Children's Hospital, Quezon City, Philippines
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P1-06-3

Frequency of thalassemia and hemoglobinopathies in the Philippines screened by high performance liquid chromatography

Ronnette Anne Davila[1], Catherine Lynn Silao[1,2], Carl Angelo Estrada[1], Ma. Liza Naranjo[3,4], Ernesto Yuson[4]

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3 National Children's Hospital, Quezon City, Philippines
4 Lung Center of the Philippines, Quezon City, Philippines

P1-06-4

Clinical application of targeted long read sequencing in prenatal beta-thalassemia testing and genetic counselling

Hui-Lin Chin[1,2], Miles C Benton[3], Lin Yang[3], Kok Siong Poon[4], Karen ML Tan[4], Saumya S Jamuar[5], Roger Foo[6], Hai Yang Law[7], Denise Li-meng Goh[1,2], Samuel S Chong[2,4,8], Paola Florez de Sessions[3]

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2 Department of Paediatrics, Yong Loo Lin School of Medicine, National University of Singapore, Singapore
3 Oxford Nanopore Technologies, Singapore
4 Department of Laboratory Medicine, National University Hospital, Singapore
5 Genetics Service, Department of Paediatrics, KK Women's and Children's Hospital, Singapore
6 Cardiovascular Research Institute, Yong Loo Lin School of Medicine, National University of Singapore, Singapore
7 DNA Diagnostic and Research Laboratory, KK Women's and Children's Hospital, Singapore
8 Department of Obstetrics and Gynaecology, Yong Loo Lin School of Medicine, National University of Singapore, Singapore

P1-06-5

A rare variant haemoglobin mimicking Hb Q-Thailand (NG_000006.1: g.37919G>C)

Norafiza Mohd Yasin[1], Faidatul Syazlin Abdul Hamid[1], Yuslina Mat Yusoff[1], Ermi Neiza Mohd Sahid[1], Azian Naila Md Noor[1], Nor Syazana Jamali[2], Ezalia Esa[1]

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P1-06-6

Knowledge and awareness of thalassemia carrier screening among Thai medical students

Pawitchaya Jariyapongpaiboon[1], Boodchiya Rojsuriyawong[1], Suwijak Meenapa[1], Pacharapan Surapolchai[2], Prapasri Kulalert[2,3], Kitiwan Rojnueangnit[2]

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3 Department of Epidemiology, Faculty of Medicine, Thammasat University, Thailand

P1-06-7

Identification of novel 2-gene deletion by Multiplex Ligation-dependent Probe Amplification of the α globin gene cluster

Naomi Goh, Wendy Low, Alexis Wang, Hai Yang Law

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Poster Session 1-07　Cancer Genomics, Germline 1

Date　：Thursday, October 12, 2023　17:30 ～ 18:00　Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P1-07-1 A case of MEN1 with pulmonary NET diagnosed by cancer multi-gene panel testing of scalp metastases

Yoshihiro Takahashi[1,2,3], Yukio Horikawa[1,2,3], Yumi Matsuyama[3], Kimiko Asai[3], Junki Endo[4], Mayu Sakai[1,2], Takehiro Kato[1,2], Ken Takao[1,2], Masami Mizuno[1,2], Takuo Hirota[1,2], Daisuke Yabe[1,2,5,6,7]

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P1-07-2 BARD1 nonsense variant c.334C>T in a patient with recurrent breast cancer associated with cervical cancer

Hiroyuki Maeda, Kazumi Ikeda, Masamichi Ikawa, Mizuho Takahashi, Hiroko Kohno, Yoshiaki Imamura, Yasushi Matsuda, Yasuo Hirono, Goi Takanori

University of Fukui, Japan

P1-07-3 The impact of medical expenses on the decision to undergo confirmatory germline testing of secondary findings

Chiaki Inagaki[1,2,3], Itsuki Oda[2], Atsuko Ikegawa[2], Hisato Kawakami[1], Yuzuki Nakagawa[3], Naoki Shiraishi[3], Takayuki Takahama[1,3], Kimio Yonesaka[1,3], Kazuhiko Nakagawa[1,3], Yoshiaki Nakamura[4,5], Takeshi Kuwata[6,7], Kazumasa Saigoh[2], Kazuo Tamura[2,8]

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P1-07-4 Factors influencing clinical decision-making for risk-reducing Salpingo-oophorectomy among female BRCA mutation carriers

Akiko Abe, Hidetaka Nomura, Atsushi Fusegi, Mayu Yunokawa, Sanshiro Okamoto, Terumi Tanigawa, Makiko Omi, Sachiho Netsu, Yoichi Aoki, Motoko Kanno, Satoki Misaka, Teruyuki Yoshimitsu, Yusuke Butsuhara, Hiroyuki Kanao

The Cancer Institute Hospital of Japanese Foundation for Cancer Research, Japan

P1-07-5 Current status of hereditary breast cancer medical treatment by public medical insurance at our hospital

Fuminori Aki[1], Ippei Kamidi[2], Shinzo Ozaki[2], Iyo Nakamura[2], Takanori Kawamura[3], Tomohiro Okazoe[3]

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2 Hosogi Hospital Surgery, Japan
3 Kouchi Seikyou Hospital Surgery, Japan

P1-07-6

A retrospective analysis of presumed germline pathogenic variants in the comprehensive genomic profiling test for cancer

Haruka Murakami[1], Satomi Inoue[1], Tatsuo Matsunaga[1], Kohei Nakamura[2], Hiroshi Nishihara[2], Yasutaka Sukawa[3], Yoshitaka Oyamada[4], Takayuki Kinoshita[5], Kazuki Yamazawa[1]

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P1-07-7

The real-world universal screening for Lynch Syndrome in a public tertiary hospital in Japan

Yozo Suzuki, Masakazu Ikenaga, Kiyotaka Hagihara, Yasufumi Sato, Toshiki Noma, Yoshitomo Yanagimoto, Yasufumi Yamashita, Aki Kobayashi, Junzo Shimizu, Tomono Kawase, Takashi Iwazawa, Naohiro Tomita, Hiroshi Imamura

Toyonaka Municipal Hospital, Japan

P1-07-8

Clinical management of patients with attenuated familial adenomatous polyposis under long-term endoscopic surveillance

Naoyuki Toyota[1], Keiko Makishima[2], Kyoko Takai[2], Takanori Akama[2], Masatsugu Ishii[3], Masaru Takemae[7], Tomoka Toyota[4], Maki Konno[5], Jun Konishi[5], Kokichi Sugano[2,6]

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P1-07-9

Clinical features of the BRCA1 and BRCA2 pathogenic variant carriers in the population-based cohort study in Japan

Yoichi Suzuki[1,2], Kinuko Ohneda[1], Yohei Hamanaka[1,3], Nobuo Fuse[1,4], Fuji Nagami[1,4], Hiroshi Kawame[1,5], Masayuki Yamamoto[1,4]

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P1-07-10

A study of Li-Fraumeni syndrome that led to diagnosis after tumor-only comprehensive genomic profiling at our hospital

Eri Habano[1], Keika Kaneko[1], Hiromi Arakawa[1], Yuko Minoura[1], Asami Kuga[1], Naomi Hayashi[1], Akito Dobashi[2], Takahiro Kogawa[1], Ippei Fukada[1], Seiichi Mori[3], Shunji Takahashi[1], Arisa Ueki[1]

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2 The Cancer Institute, Japanese Foundation for Cancer Research, Japan
3 The Cancer Precision Medicine Center, Japanese Foundation for Cancer Research, Japan

P1-07-11 **Returning genomic results to population cohort study participants with pathogenic variants in hereditary cancer genes**

Kinuko Ohneda[1], Yoichi Suzuki[1,2], Yohei Hamanaka[1,3], Nobuo Fuse[1,5], Fuji Nagami[1,5], Tomoko Kobayashi[1,6], Hiroshi Kawame[1,4], Masanobu Takahashi[7], Muneaki Shimada[1,5,8], Masayuki Yamamoto[1,5]

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7 Department of Clinical Oncology, Tohoku University Graduate School of Medicine, Sendai, Miyagi, Japan
8 Department of Gynecology and Obstetrics, Tohoku University Graduate School of Medicine, Sendai, Miyagi, Japan

P1-07-12 **Clinical characterization of patients with gBRCA1/2 mutation-positive unresectable pancreatic cancer**

Tomohiro Kubo[1], Joji Muramatsu[1], Yohei Arihara[1], Ayako Murota[2,3], Kazuma Ishikawa[1], Makoto Yoshida[1], Hiroyuki Nagashima[4], Yuki Ikeda[5], Makoto Usami[6], Hajime Nakamura[1,7], Daichi Watanabe[8], Takanori Shibata[9], Kaoru Kasahara[10], Akihiro Sakurai[2], Kohichi Takada[1]

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8 Department of Gastroenterology, Japanese Red Cross Date Hospital, Japan
9 Department of Gastroenterology, Rumoi City Hospital, Rumoi, Japan
10 Department of Gastroenterology, Hakodate Goryoukaku Hospital, Japan

P1-07-13 **Screening for Lynch syndrome using the MSI test at a regional core hospital in Japan**

Takahiro Yoshioka, Eri Takeda, Kento Kumon, Ryo Inada, Ryo Yoshioka, Kazuyuki Ooishi

Kochi Health Sciences Center, Japan

P1-07-14 **A case of triple negative breast cancer after ipsilateral malignant lymphoma of the breast with BRCA2 pathogenic variant**

Keiko Miyazato[1], Kaname Kurashita[1], Kyu-ichiro Miyara[2], Rie Tanaka[1], Shigemi Murayama[2], Kiyomi Kimura[2]

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2 Miyara Clinic, USA

Poster Session 1-08 Cancer Genomics, Somatic 1

Date ：Thursday, October 12, 2023 17:30 ～ 18:00 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P1-08-1 **The management system for BRCA1/2 gene analysis and clinical result in our institute**

Yuichiro Inagaki, Takeshi Amemiya, Daijuro Hayashi, Takahiro Suzuki, Satoshi Kurokawa, Junko Arima

Anjo Kosei Hospital, Japan

P1-08-2 **Development of a DNA Chip for MLH1 Methylation Detection in Sporadic MSI-High Tumors**

Takeshi Nagasaka

Kawasaki Medical School Hospital, Japan

P1-08-3

Comprehensive cancer genomic profiling tests in metastatic castration-resistant prostate cancer-patient

Hideyasu Tsumura[1], Naomi Araki[2], Rika Kawata[2], Hironao Shirai[2], Sachiko Ohori[2], Mina Waraya[2], Masao Araki[2], Tsutomu Yoshida[3], Jiichiro Sasaki[3], Masatsugu Iwamura[1], Fumio Takada[4]

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3 Kitasato University School of Medicine New Century Medical Development Center Cross-sectional Medical Area Development, Japan
4 Kitasato University Graduate School of Medicine, Japan

P1-08-4

Clinical applications of comprehensive genomic profiling tests for castration-resistant prostate cancer

Mari Kikuchi[1,2], Kouki Ohtsuka[2,3], Yu Nakamura[2,4], Junji Kitamura[4], Jimpei Miyakawa[4], Chiharu Doutsu[5,6], Tomohiko Taki[2,7], Wataru Ogura[1,2], Takatsugu Okegawa[4], Junji Shibahara[8], Shuichi Hironaka[6,9], Hiroshi Fukuhara[4,6], Yaeko Ichikawa[2,10], Hiroaki Ohnishi[1,2,3]

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5 Nursing Department, Kyorin University Hospital, Japan
6 Cancer Center, Kyorin University Hospital, Japan
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8 Department of Diagnostic Pathology, Faculty of Medicine, Kyorin University, Japan
9 Department of Medical Oncology, Faculty of Medicine, Kyorin University, Japan
10 Department of Neurology, Faculty of Medicine, Kyorin University, Japan

P1-08-5

Current status of the Comprehensive Genome Profiling (CGP) tests in urology at our hospital

Mizuho Okawa[1], Mio Wakai[2], Yuko Tamaki[2], Midori Shuhara[2], Kota Arakawa[5], Shino Hasegawa[6], Shunsuke Hori[1], Masoto Uetani[1], Nahomi Umemura[2], Yuko Hayashi[2], Fumito Yamabe[1], Yozo Mitsui[1], Hideyuki Kobayashi[1], Naobumi Tochigi[3], Shinji Ujiie[4], Eiyu Nozawa[7], Yoshie Murakami[2], Koichi Nagao[1], Yukiko Katagiri[2], Koichi Nakajima[1]

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6 Genetic Counseling Outpatient, Japanese Red Cross Mito Hospital, Japan
7 Department of Urology, Japanese Red Cross Mito Hospital, Japan

P1-08-6

Current status and issues of HRD testing and BRCA gene testing in ovarian cancer cases at our hospital

Takashi Nagai, Yasuhiro Matsuyama, Mikako Asai, Misaki Nobata, Arina Sato, Hirofumi Akita, Keita Kuroda, Kei Hattori, Yuko Suzuki, Ayako Osafune, Tomokazu Umezu

Kariya Toyota General Hospital, Japan

Poster Session 1-09　Pediatric Genetics 1

Date　：Thursday, October 12, 2023　17:30 ～ 18:00　Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P1-09-1 Molecular analysis of case with Beckwith-Wiedemann syndrome, placental mesenchymal dysplasia, and hepatoblastoma

Tomoe Ogawa[1,2], Yohei Kosugi[3], Toshihiro Matsui[4], Yasuto Suzuki[5], Ryoji Kobayashi[6], Maki Fukami[1], Masatoshi Tateno[7], Rika Kosaki[8], Masayo Kagami[1]

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6 Department of Pediatrics, Sapporo Hokuyu Hospital, Japan
7 Department of Pathology, Kushiro Red Cross Hospital, Japan
8 Department of Medical Genetics, National Center for Child Health and Development, Japan

P1-09-2 Autosomal dominant diseases ascertained by microarray chromosome analysis; report of four Japanese patients

Hidefumi Tonoki[1], Tohru Ohta[2], Hisato Suzuki[3], Mamiko Yamada[3], Kenjiro Kosaki[3]

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2 Health Sciences University of Hokkaido, Japan
3 Center for Medical Genetics, Keio University School of Medicine, Japan

P1-09-3 Clinical phenotypes of 10 patients with genetical diagnosed as ciliopathy

Tomoko Uehara, Natsuki Nakamura, Seiji Mizuno, Mie Inaba

Aichi Developmental Disability Center, Japan

P1-09-4 Glial cell line-derived neurotrophic factor and GDNF family receptor alpha-1 expressions effect in patients with Hirschsprung disease

Setiani Nurhidayah

Genetics Working Group / Translational Research Unit, Faculty of Medicine, Public Health and Nursing, Universitas Gadjah Mada, Yogyakarta, Indonesia

P1-09-5 CCND2 and PIK3R3 expression effect on liver fibrogenesis in biliary atresia infants

Rahaditya Hanggoro[1,2], Kevin Eliezer Ferdinandus[2], Fiqih Vidiantoro Halim[2], Laudria Stella Eryvinka[2], Kristy Iskandar[3], Akhmad Makhmudi[2], Gunadi Gunadi[2]

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2 Pediatric Surgery Division, Department of Surgery, Faculty of Medicine, Public Health and Nursing, Universitas Gadjah Mada / Dr.Sardjito Hospital, Yogyakarta, Indonesia
3 Department of Child Health, Faculty of Medicine, Public Health and Nursing, Universitas Gadjah Mada / UGM Academic Hospital, Yogyakarta, Indonesia

P1-09-6 De novo SCN8A variant identified in a patient with infantile epileptic encephalopathy and congenital nephrotic syndrome

Shinsuke Ninomiya[1], Kunihiko Aya[4], Satoko Tokumasu[4], Mariko Sawada[4], Yoshiyuki Hanaoka[4], Hisato Suzuki[2,3], Mamiko Yamada[2], Toshiki Takenouchi[5], Kenjiro Kosaki[2]

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P1-09-7

12 patients with ML in our hospital and their risk of respiratory distress after viral infection

Yuki Sekido[1], Yasuyuki Fukuhara[1,2], Tetsumin So[2], Rinshu Shimabukuro[3], Akihiro Umezawa[1], Motomichi Kosuga[2]

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P1-09-8

Genome and RNA sequencing reveal structural rearrangements in ATAD3 gene cluster

Yoshihito Kishita[1], Yukiko Yatsuka[2], Tomohiro Ebihara[3], Takuya Fushimi[3], Yohei Sugiyama[2,3], Atsuko Imai-Okazaki[2], Akira Ohtake[4], Kei Murayama[2,3], Yasushi Okazaki[2,5]

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3 Chiba Children's Hospital, Japan
4 Saitama Medical University, Japan
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P1-09-9

A splice site pathogenic variant in *Dystrophin* detected by whole-exome sequencing modifies clinical manifestation of a male patient with DMD into a milder one: A case report

Inna Faradina Putri[1], Gunadi[1], Kristy Iskandar[2], Ery Kus Dwianingsih[3], Sunartini[2]

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3 Department of Anatomical Pathology Faculty of Medicine, Public Health and Nursing, Universitas Gadjah Mada, Yogyakarta, Indonesia

P1-09-10

Intrachromosomal copy number gain adjacent to a terminal loss: Points to consider for interpreting CNVs

Keiko Wakui[1,2], Mariko Eguchi[3], Naoki Harada[4]

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P1-09-11

Generation of isogenic models of sex chromosome aneuploid hiPS cells via improved microcell-mediated chromosome transfer

Hitomaru Miyamoto[1], Masaya Egawa[2], Narumi Uno[2], Kyotaro Yamazaki[3], Teruhiko Suzuki[4], Shusei Hamamichi[1], Kazuma Tomizuka[2], Yasuhiro Kazuki[1,3]

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3 National Institutes of Natural Sciences, Japan
4 Tokyo Metropolitan Institute of Medical Science, Japan

P1-09-12

A deep intronic TCTN2 variant activating a cryptic exon predicted by SpliceRover in a patient with Joubert syndrome

Takuya Hiraide[1], Kenji Shimizu[2], Yoshinori Okumura[2], Sachiko Miyamoto[1], Mitsuko Nakashima[1], Tsutomu Ogata[1,3], Hirotomo Saitsu[1]

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2 Shizuoka Children's Hospital, Japan
3 Hamamatsu Medical Center, Japan

P1-09-13 **A case of congenital hypothyroidism with NKX2-1 and DUOX2 variants**

Erika Uehara[1], Naoaki Hori[2], Kanako Nakao[1], Kazuhisa Akiba[1,3], Hidefumi Sueoka[4], Keiko Matsubara[1,5], Satoshi Narumi[1,6]

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4 Dept. of Pediatr., Sapporo Medical Univ., Japan
5 Div. of Diversity Research, National Research Institute for Child Health and Development, Japan
6 Dept. of Pediatr., Keio Univ. Sch. of Med., Japan

P1-09-14 **Analysis of a single exon deletion that was not easily found in the autosomal recessive Bardet-Biedl syndrome**

Takuya Naruto[1], Yukiko Kuroda[2], Yoko Saito[2], Yumi Enomoto[1], Kenji Kurosawa[2]

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2 Division of Medical Genetics, Kanagawa Children's Medical Center, Yokohama, Japan

P1-09-15 **A heterozygous MMP13 mutation in a girl with metaphyseal anadysplasia, showing a skeletal phenotype mimicking rickets**

Midori Motokawa

Nagasaki Univ. Sch. of Med., Japan

P1-09-16 **A patient with cystic fibrosis revealed by maternal uniparental disomy of chromosome seven**

Hayate Masubuchi[1], Kaori Hara[1], Rika Kosaki[2], Goro Koinuma[3], Kenjiro Kosaki[4], Maki Fukami[1], Masayo Kagami[1]

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P1-09-17 **Left main coronary artery ostial atresia in a Noonan-like patient with CBL mutation and RNF213 polymorphism p.R4810K**

Ayako Chida-Nagai[1], Hidefumi Tonoki[2], Naomasa Makita[3], Hiroyuki Ishiyama[3], Masafumi Ihara[3], Yuji Maruo[1], Takao Tsujioka[1], Daisuke Sasaki[1], Gaku Izumi[1], Hirokuni Yamazawa[1], Nobuyasu Kato[1], Masaki Ito[1], Miki Fujimura[1], Osamu Sasaki[2], Atsuhito Takeda[1]

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P1-09-18 **Pulmonary stenosis in a female with Börjeson-Forssman-Lehmann syndrome**

Yoko Saito, Yukiko Kuroda, Yumi Enomoto, Kenji Kurosawa

Kanagawa Children's Medical Center, Japan

P1-09-19 **A distinct genotype and phenotypes in pediatric patients with biventricular noncompaction**

Keiichi Hirono[1], Yukiko Hata[1], Kaori Tsuboi[1], Shinya Takarada[1], Mako Okabe[1], Hideyuki Nakaoka[1], Keijiro Ibuki[1], Sayaka Ozawa[1], Naoki Nishida[1], Fukiko Ichida[2]

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2 International University of Health and Welfare, Japan

P1-09-20 **Two cases of genetic anomalies with major aortopulmonary collateral arteries**

Ayako Matsunaga[1], Minami Ozawa[1], Shotaro Kaku[1], Yosuke Osada[1], Noriko Udagawa[1], Yusaku Miyamoto[1], Ohsuke Migita[2], Kentaro Aso[1], Keishi Yoshida[3], Yoshio Shima[3], Fuyuki Miya[4], Mitsuhiro Kato[5], Naoki Shimizu[1]

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P1-09-21 **16p13.11 microduplication with growth retardation and developmental disorders**

Daisuke Watanabe, Hideaki Yagasaki, Hiromune Narusawa, Takeshi Inukai

Yamanashi University, Japan

P1-09-22 **Lessons learned from a case of Dopa-Responsive Dystonia (Segawa Disease) that took 5 years to diagnose**

Hiroko Taniai[1,3], Kaname Ishii[1], Mihoko Mizuno[2]

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2 Daido Hospital, Japan
3 Nagoya City Child Welfare Center, Japan

P1-09-23 **Understanding of the neurological manifestation in pediatric patients with 16p11.2 deletion**

Ji Yoon Han

Department of Pediatrics, Daejeon St. Mary's Hospital, Korea, College of Medicine, The Catholic University of Korea, Korea

P1-09-24 **Molecular genetics of osteogenesis imperfecta: Data updated from Vietnam national children's hospital**

Ngoc Can, Thao Bui, Khanh Nguyen, Mai Do, Ha Nguyen, Hang Nguyen, Lan Nguyen, Giang Dang, Son Do, Huyen Dang, Thuong Tran, Dung Vu

Center of Endocrinology, Metabolism, Genetics, and Molecular Therapy, Vietnam National Children's Hospital, Vietnam

P1-09-25 **A family with brachydactyly type C, GDF5-related -an experience of genetic counseling**

Tomoyo Yamashita[1], Junko Hotta[1], Noriko Nakano[1], Eri Sakai[1], Chie Ono[1], Haruka Bamba[1], Kanako Yamashita[3], Shiro Ikegawa[2], Gen Nishimura[2], Takashi Hamazaki[1], Toshiyuki Seto[1]

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Poster Session 1-10 Aging and Diseases

Date ：Thursday, October 12, 2023 17:30 ～ 18:00 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P1-10-1 **Knock-in of late-onset Alzheimer's disease-risk variant SHARPIN G186R lessens NF-κB pathway and accelerates Aβ secretion**

Yuya Asanomi[1], Tetsuaki Kimura[1], Nobuyoshi Shimoda[1], Daichi Shigemizu[1,2], Shumpei Niida[3], Kouichi Ozaki[1,2]

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2 RIKEN Center for Integrative Medical Sciences, Yokohama, Japan
3 Center for Core Facility Administration, Research Institute, National Center for Geriatrics and Gerontology, Obu, Japan

P1-10-2 **Vineland-II adaptive behavior profile of adults with genetic disorders and intellectual disability**

Miho Osako, Satoshi Kobayashi, Kouko Asai, Yu Iijima, Yoko Kanbara, Yoko Mochizuki

Tokyo Metropolitan Kita Medical and Rehabilitation Center for the Disabled, Japan

P1-10-3 **Age-dependent metabolic shifts differentiate sarcopenic responses in mice**

Masaki Mori

NCVC Research Institute, Japan

P1-10-4 **A genome wide association study identifies an East Asian-specific risk variant for Lewy bodies dementia in Japanese**

Risa Mitsumori[1], Yuya Asanomi[3], Daichi Shigemizu[2,3], Sintaro Akiyama[3], Takashi Morizono[3], Shumpei Niida[4], Kouichi Ozaki[2,3]

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4 Core Facility Administration, Research Institute, NCGG, Obu, Japan

P1-10-5 **Functional analysis of MFSD3 associated with dementia with Lewy bodies**

Tetsuaki Kimura[1], Mutsumi Suganuma[1], Tohru Hosoyama[2], Kayoko Sawamura[1], Nobuyoshi Shimoda[1], Noboru Ogiso[3], Shumpei Niida[3], Kouichi Ozaki[1,4], Daichi Shigemizu[1,4]

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3 Center for Core Facility Administration, Research Institute, National Center for Geriatrics and Gerontology, Japan
4 RIKEN Center for Integrative Medical Sciences, Japan

P1-10-6 **Identification of potential blood-based biomarkers for frailty by using an integrative approach**

Mutsumi Suganuma[1], Motoki Furutani[2], Tohru Hosoyama[3], Shintaro Akiyama[1], Risa Mitsumori[1], Rei Otsuka[4], Marie Takemura[5], Yasumoto Matsui[5], Yukiko Nakano[2], Shumpei Niida[6], Kouichi Ozaki[1,2,7], Shosuke Satake[8], Daichi Shigemizu[1,7]

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8 Department of Frailty Research, Center for Gerontology and Social Science, National Center for Geriatrics and Gerontology, Japan

P1-10-7 **Genome-wide association study for non-specific chronic pain in Japanese elderly**

Daichi Shigemizu, Yoshihito Sakai, Ken Honjo, Norimitsu Wakao, Hiroki Matsui, Hiroyuki Shimada, Risa Mitsumori, Kouichi Ozaki, Ken Watanabe

National Center for Geriatrics and Gerontology, Japan

P1-10-8 **Potential involvement of a rare variant in a gene encoding a novel ferroptosis-related transporter in osteoarthritis**

Kiyoaki Ishii

National Center for Geriatrics and Gerontology, Japan

Poster Session 1-11 Therapeutics (DNA, mRNA, genome editing, others)

Date ：Thursday, October 12, 2023 17:30 ～ 18:00 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P1-11-1 **Study to improve clinical results of ROSI (Round Spermatid Injection) histone deacetylase inhibitor**

Atsushi Tanaka[1], Youichi Takemoto[1], Motoi Nagayoshi[1], Yuya Makino[1,2], Daisuke Nakajima[1,3], Seiji Watanabe[4]

1 Saint Mother Clinic, Japan
2 Juntendo University School of Medicine, Japan
3 Occupational and Environmental Health University School of Medicine, Japan
4 Hirosaki University Graduate School of Medicine, Japan

P1-11-2 **In vivo glucose lowering effect of MSC derived secretome**

Dwi Aris Agung Nugrahaningsih[1], Sofia Mubarika[1], Pamungkas Bagus Satriyo[1], Purwadi Purwadi[2], Is Sarifin[2], Indra Bachtiar[3], Sunarto Sunarto[2], Ubaidillah Ubaidillah[2], Ines Larasati[2], Dicka Setiasari[1], Jarir At-thobari[1]

1 Faculty of Medicine, Public Health and Nursing, Universitas Gadjah Mada, Indonesia
2 Kepolisian Negara Republik Indonesia, Indonesia
3 PT. Tristem Medika Indonesia, Indonesia

P1-11-3 **Correction of MYH9 R702C by CRISPR/Cas9 system in iPSCs model**

Amornrat Tangprasittipap[1], Pawarit Innachai[1], Alisa Tusuwan[2], Nongnuch Srirachainan[3], Suradej Hongeng[3]

1 Research Center, Faculty of Medicine Ramathibodi Hospital, Mahidol University, Bangkok, Thailand
2 Institute of Molecular Biosciences, Mahidol University, Nakhon Pathom, Thailand
3 Department of Pediatrics, Faculty of Medicine Ramathibodi Hospital, Mahidol University, Bangkok, Thailand

P1-11-4 **Disruption of *ZBTB7A/LRF* or *BCL11A* binding site to reactivate fetal hemoglobin in healthy donor and β^0-thalassemia/HbE**

Chokdee Wongborisuth[1], Amornrat Tangprasittipap[1], Pawarit Innachai[1], Chonticha Saisawang[2], Natee Jearawiriyapaisarn[2], Alisa Tubsuwan[3], Suradej Hongeng[4], Duantida Songdej[4]

1 Research Center, Faculty of Medicine, Ramathibodi Hospital, Bangkok, Thailand
2 Institute of Molecular Biosciences, Mahidol University, Nakhon-Pathom, Thailand
3 Thalassemia Research Center, Institute of Molecular Biosciences, Mahidol University, Nakhon-Pathom, Thailand
4 Hematology and Oncology, Department of Pediatrics, Faculty of Medicine, Ramathibodi Hospital, Bangkok, Thailand

Poster Session 1-12 Rare Diseases 1

Date ：Thursday, October 12, 2023 17:30 ～ 18:00 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P1-12-1 **Detailed analysis of single cell transcriptome using Sturge-Weber syndrome specific on-chip vasculature**

Kimihiko Banno[1,2], Jered Myslinski[3], Junko Yoshida[2], Yoshikazu Kameda[4], Maneesha Shaji[4], Takashi Hato[3], Ryuji Yokokawa[4], Kyoji Horie[2]

1 Nara Medical University, Japan
2 Department of Physiology II, Nara Medical University, Japan
3 Department of Medicine, Indiana University School of Medicine, USA
4 Department of Micro Engineering, Graduate School of Engineering, Kyoto University, Japan

P1-12-2 **Narrowing down the A/B-DMR methylation maintenance region based on a deletion in familial Pseudohypoparathyroidism 1A**

Hiromune Narusawa[1,2], Yukie Nakagawa[2], Sayaka Isobe[2], Kyoichiro Tsuchiya[2], Hideaki Yagasaki[2], Kazuhiko Nakabayashi[1], Maki Fukami[1], Masayo Kagami[1]

1 National Center for Child Health and Development, Japan
2 University of Yamanashi, Japan

P1-12-3 **Experience with advance care planning for a patient with connatal Pelizaeus-Merzbacher disease**

Fumihito Nozaki[1], Kei Shiraishi[1], Shohei Eto[1], Mariko Ishihara[1], Atsushi Mori[1], Sayoko Haruyama[2]

1 Shiga Medical Center for Children, Japan
2 Department of Medical Ethics and Medical Genetics, Kyoto University School of Public Health, Japan

P1-12-4 Efficient identification of causative genes of hearing loss by phenotype similarity analysis

Hideki Mutai[1], Fuyuki Miya[2], Kiyomitsu Nara[1], Reiko Muramatsu[1], Satomi Inoue[3], Haruka Murakami[3], Shujiro Minami[4], Atsuko Nakano[5], Yukiko Arimoto[5], Noriko Morimoto[6], Taiji Kawasaki[7], Koichiro Wasano[4,7,8], Hirokazu Sakamoto[9,10], Sayaka Katsunuma[10], Sawako Masuda[11], Kazuki Yamazawa[3], Kenjiro Kosaki[2], Tatsuhiko Tsunoda[12,13,14], Tatasuo Matsunaga[1,3,4]

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2 Center for Medical Genetics, Keio University School of Medicine, Japan
3 Department of Medical Genetics, National Hospital Organization Tokyo Medical Center, Japan
4 Department of Otolaryngology, National Hospital Organization Tokyo Medical Center, Japan
5 Otorhinolaryngology, Chiba Children's Hospital, Japan
6 Otorhinolaryngology, National Center for Child Health and Development, Japan
7 Otolaryngology-Head and Neck Surgery, Red Cross Shizuoka Hospital, Japan
8 Otorhinolaryngology-Head and Neck Surgery, Tokai University School of Medicine, Japan
9 Otorhinolaryngology-Head and Neck Surgery, Osaka Metropolitan University School of Medicine, Japan
10 Otorhinolaryngology, Kobe Children's Hospital, Japan
11 Otorhinolaryngology, National Hospital Organization Mie National Hospital, Japan
12 Medical Science Mathematics, RIKEN Center for Integrative Medical Sciences, Japan
13 Medical Science Mathematics, Department of Biological Sciences, School of Science, The University of Tokyo, Japan
14 Medical Science Mathematics, Department of Computational Biology and Medical Sciences, Graduate School of Frontier Sciences, The University of Tokyo, Japan

P1-12-5 Development of isolated nephrogenic diabetes insipidus in a girl with contiguous gene deletion involving AVPR2 and L1CAM

Shoma Saito[1], Shigeru Suzuki[1], Takuya Kamiyama[2], Takahide Kokumai[1], Akiko Furuya[1], Genya Taketazu[3], Yoshio Makita[4], Satoru Takahashi[1]

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2 Department of Medicine, Asahikawa Medical University, Japan
3 Department of Pediatrics, Asahikawa-Kosei General Hospital, Japan
4 Department of Genetic Counseling, Asahikawa Medical University Hospital, Japan

P1-12-6 A case report on MELAS/Leigh Syndrome overlap with an MT-ND1 mutation

Catrina Yang, Ma. Jesusa Rachelle Vicencio, Leniza De Castro-Hamoy, Ignacio Rivera

The Medical City, Philippines

P1-12-7 A case report on RAB27A variants in a patient presenting with hemophagocytic lymphohistiocytosis

Catrina Yang, Florentina Uy

The Medical City, Philippines

P1-12-8 Two pediatric cases of primary ciliary dyskinesia caused by OFD1 variants

Kazuhiko Takeuchi[1], Tsubasa Matsumoto[2], Yuki Tsurinaga[3], Yifei Xu[1], Taichi Yano[1], Hiroshi Sakaida[1], Sawako Masuda[4], Koki Ueda[1], Guofei Feng[1], Shimpei Gotoh[5], Satoru Ogawa[1], Makoto Ikejiri[1], Kaname Nakatani[6], Mizuho Nagao[4], Masaki Tanabe[1]

1 Mie University, Japan
2 Fukuoka Children's Hospital, Japan
3 Osaka Habikino Medical Center, Japan
4 National Hospital Organization Mie National Hospital, Japan
5 Kyoto University, Japan
6 IGA City General Hospital, Japan

P1-12-9 A girl with multiple autoimmune disorders harboring two rare Mendelian autoimmunity syndromes

Yu-Ming Chang, Yu-Wen Pan, Meng-Che Tsai, Yen-Yin Chou

National Cheng Kung University Hospital, Taiwan

P1-12-10 **Familial pseudohypoparathyroidism type IB associated with an SVA retrotransposon insertion in the GNAS locu**

Masayo Kagami[1], Sayaka Kawashima[1,2], Akiko Yuno[3], Shinichiro Sano[1,4], Akie Nakamura[1,5], Keisuke Ishiwata[1], Tomoyuki Kawasaki[1], Kazuyoshi Hosomichi[6], Kazuhiko Nakabayashi[1], Hidenori Akutsu[1], Hirotomo Saitsu[7], Maki Fukami[1], Takeshi Usui[8,9], Tsutomu Ogata[1,7,10]

1 National Research Institute for Child Health and Development, Japan
2 Tohoku University Graduate School of Medicine, Japan
3 Kin-ikyo Chuo Hospital, Japan
4 Shizuoka Children's Hospital, Japan
5 Hokkaido University School of Medicine, Japan
6 Kanazawa University, Japan
7 Hamamatsu University School of Medicine, Japan
8 Shizuoka General Hospital, Japan
9 Shizuoka Graduate University of Public Health, Japan
10 Hamamatsu Medical Center, Japan

P1-12-11 **Craniofacial and dental characteristics of 3 Japanese individuals with genetically diagnosed SATB2-associated syndrome**

Hiroshi Kurosaka

Osaka University, Japan

P1-12-12 **Family experience of living with children and adults with 1q duplicationsyndrome**

Mikiko Kaneko, Kana Harada, Chisen Takeuchi, Hiroshi Kawame

The Jikei University Hospital, Japan

P1-12-13 **Novel TBCK variant and importance of appropriate VUS interpretation**

Chinmayee Bhimarao Nagaraj[1], Eileen Broomall[1], Lisa Reebals[1], Cuixia Tian[1,2]

1 Cincinnati Children's Hospital, USA
2 University of Cincinnati, USA

P1-12-14 **Two McCune-Albright Syndrome cases with early onset breast cancer**

Akiko Matsutani, Go Yamamoto, Miho Kakuta, Natsuki Naka, Akemi Takahashi, Yumi Ikeda, Yukiko Osanai, Tomomi Hirata, Katsuya Iuch, Tomokazu Wakatsuki, Kiwamu Akagi

Saitama Cancer Center, Japan

P1-12-15 **A case of autosomal dominant junctional epidermolysis bullosa with multiple squamous cell carcinoma**

Mayumi Komine[1], Fuminori Yamamoto[2], Fuminori Katsumata[2], Mamitaro Ohtsuki[2], Ken Natsuga[3], Hideyuki Ujiie[3]

1 Department of Dermatology, Center for Career Support, Jichi Medical University, Japan
2 Department of Dermatology, Jichi Medical University, Japan
3 Department of Dermatology, Hokkaido University, Japan

P1-12-16 **TBX5 pathogenic variant in a patient with congenital heart defect and tracheal stenosis**

Kaori Yamoto[1], Fumiko Kato[1], Masaya Yamoto[2], Koji Fukumoto[2], Kenji Shimizu[2], Hirotomo Saitsu[1], Tsutomu Ogata[1]

1 Hamamatsu Medical University, Japan
2 Shizuoka Children's Hospital, Japan

P1-12-17 **A case of trisomy 14 mosaicism presenting with multiple congenital anomalies in a neonate**

Maria Gabriela Katigbak, Julianne Racoma, Marcelle Reyes-Tiu

Makati Medical Center, Philippines

P1-12-18 **Utilization of chromosomal microarray to elucidate syndromic intellectual disability in Indonesia**

Nydia Sihombing[1], Ferdy Kurniawan Cayami[1], Nani Maharani[1], Agustini Utari[1,2], Tri Indah Winarni[1]

1 Center for Biomedical Research (CEBIOR), Faculty of Medicine, Universitas Diponegoro, Indonesia
2 Department of Pediatrics, Faculty of Medicine, Universitas Diponegoro, Indonesia

P1-12-19 Proposal of genetic diagnosis approach for Prader-Willi syndrome and Kagami-Ogata syndrome

Tsutomu Oata[1], Shinji Saitoh[2], Hidenobu Soejima[3], Masayo Kagami[4]

1 Hamamatsu University School of Medicine, Japan
2 Nagoya City University Gradual School of Medical Sciences, Japan
3 Saga University School of Medicine, Japan
4 National Research Institute for Child Healh and Development, Japan

P1-12-20 Biallelic structural variations within FGF12 detected by long-read whole genome sequencing in epilepsy

Sachiko Ohori[1,2], Akihiko Miyauchi[3], Hitoshi Osaka[3], Naohiro Arakaki[4,5], Toru Sengoku[6], Kazuhiro Ogata[6], Satomi Mitsuhashi[7], Martin Frith[8,9,10], Rie Seyama[1,11], Naomi Tsuchida[1,12], Yuri Uchiyama[1,12], Eriko Koshimizu[1], Kohei Hamanaka[1], Kazuharu Misawa[1], Satoko Miyatake[1,13], Takeshi Mizuguchi[1], Kuniaki Saito[4,5], Atsushi Fujita[1], Naomichi Matsumoto[1]

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3 Department of Pediatrics, Jichi Medical School, Japan
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5 Graduate Institute for Advanced Studies, SOKENDAI, Japan
6 Department of Biochemistry, Yokohama City University Graduate School of Medicine, Japan
7 Department of Neurology, St. Marianna University School of Medicine, Japan
8 Artificial Intelligence Research Center, National Institute of Advanced Industrial Science and Technology (AIST), Japan
9 Graduate School of Frontier Sciences, The University of Tokyo, Japan
10 Computational Bio Big-Data Open Innovation Laboratory (CBBD-OIL), AIST, Japan
11 Department of Obstetrics and Gynecology, Juntendo University, Japan
12 Department of Rare Disease Genomics, Yokohama City University Hospital, Japan
13 Department of Clinical Genetics, Yokohama City University Hospital, Japan

P1-12-21 Schaaf-Yang syndrome: Insights from a nationwide epidemiological study in Japan

Keiko Matsubara[1,2], Yutaka Negishi[3,4], Kenji Kurosawa[5], Kyoko Takano[6], Takeshi Nishiyama[7], Shinji Saito[4]

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3 Dept. of Pediatr., Gifu Prefectural Tajimi Hosp., Japan
4 Dept. of Pediatr. and Neonatol., Nagoya City Univ. Grad. Sch. of Med. Sci., Japan
5 Div. of Med. Genet., Kanagawa Children's Med. Center, Japan
6 Center for Medical Genetics, Shinshu University Hospital, Japan
7 Dept. of Public Health, Nagoya City Univ. Grad. Sch. of Med. Sci., Japan

P1-12-22 Revisiting single nucleotide variants of whole-exome sequencing data involving aberrant splicing for Mendelian diseases

Yasuhiro Utsuno[1], Kohei Hamanaka[1], Yuri Uchiyama[1,2], Naomi Tsuchida[1,2], Eriko Koshimizu[1], Atsushi Fujita[1], Satoko Miyatake[1,3], Kazuharu Misawa[1,4], Takeshi Mizuguchi[1], Naomichi Matsumoto[1]

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3 Department of Clinical Genetics, Yokohama City University Hospital, Japan
4 RIKEN Center for Advanced Intelligence Project, Japan

P1-12-23 The efficient methods of multiple genetic analyses for rare coagulation deficiencies and rare bleeding disorders

Yuri Uchiyama[1,2], Yoshiyuki Ogawa[3], Kunio Yanagisawa[4], Akira Matsumoto[2,3], Hideki Uchiumi[3], Eriko Koshimizu[2], Kohei Hamanaka[2], Atsushi Fujita[2], Kazuharu Misawa[2], Satoko Miyatake[2,5], Takeshi Mizuguchi[2], Hiroshi Handa[3], Naomichi Matsumoto[2]

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2 Department of Human Genetics, Yokohama City University Graduate School of Medicine, Yokohama, Japan
3 Department of Hematology, Gunma University Graduate School of Medicine, Gunma, Japan
4 Infection Control and Prevention Center, Gunma University Hospital, Maebashi, Japan
5 Clinical Genetics Department, Yokohama City University Hospital, Yokohama, Japan

P1-12-24 Pathogenicity of nucleotide deletions at exon-intron borders: Lesson from two cases

Yuta Inoue[1], Naomi Tsuchida[1,2], Ayumi Yoshimura[3], Ayumi Itano[3], Tetsuya Kibe[3], Chan Mei Yan[4], Keng Wee Teik[4], Yuri Uchiyama[1,2], Kohei Hamanaka[1], Eriko Koshimizu[1], Atsushi Fujita[1], Kazuharu Misawa[1], Satoko Miyatake[1,5], Takeshi Mizuguchi[1], Naomichi Matsumoto[1]

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4 Department of Genetics, Hospital Kuala Lumpur, Kuala Lumpur, Malaysia
5 Clinical Genetics Department, Yokohama City University Hospital, Yokohama, Kanagawa, Japan

P1-12-25 On-line group clinics for children with genetic syndromes: A 2022 report at Saitama Children's Medical Center

Yuki Sawada[1], Miwako Kizumi[1], Miwa Kobayashi[1], Kenta Hasumi[1,2], Miyu Fukushima[1,2], Sayuri Oda[1,2], Mariko Sagara[1,2], Daiju Oba[1], Hirofumi Ohashi[1]

1 Department of Genetics, Saitama Children's Medical Center, Japan
2 Department of Clinical Laboratory, Saitama Children's Medical Center, Japan

P1-12-26 Challenges in secondary findings disclosure in facilities offering comprehensive genetic testing for rare diseases

Kana Hiromoto[1,8], Takahiro Yamada[2,8], Mio Tsuchiya[3,8], Hiroshi Kawame[4,5,8], Eiji Nanba[6,8], Yuichi Goto[7,8], Shinji Kosugi[2,8]

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3 Amicus Therapeutics K.K., Japan
4 Tohoku University Tohoku Medical Megabank Organization, Japan
5 Department of Clinical Genetics, Jikei University, Japan
6 Organization for Research Initiative and Promotion, Tottori University, Japan
7 Medical Genome Center, National Center of Neurology and Psychiatry, Japan
8 Research Project on Ethical, Legal, and Social Issues Supported by the Health, Labour and Welfare Sciences Research Grants "Extraction of ethical and social issues and improvement of social environment toward the realization of a society where people can benefit from genome medicine without anxiety"

P1-12-27 Non-SGA girl with chromosome 6q24-related diabetes mellitus

Shinji Higuchi[1,2], Kohei Iwata[1], Yoh Watanabe[1], Yuki Yamada[1], Jun Mori[1], Hiroaki Nakamura[1,2], Tohru Yorifuji[1]

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2 Department of Genetic Medicine, Osaka City General Hospital, Japan

Poster Session 1-13 COVID-19 Susceptibility

Date ：Thursday, October 12, 2023 17:30 ～ 18:00 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P1-13-1 Clustering analysis reveals variable genetic association patterns in COVID-19 clinical subgroups

Nikolaos Avramidis[1,2], Nicola Pirastu[3], Sohan Seth[4], Erola-Pairo Castineira[1], Konrad Rawlik[1], Kenneth Baillie[1,2,5,6]

1 Pandemic Science Hub, Centre for Inflammation Research and Roslin Institute, University of Edinburgh, Edinburgh, United Kingdom
2 Roslin Institute, The University of Edinburgh, Edinburgh, United Kingdom
3 Human Technopole, Milan, Italy
4 Institute for Adaptive and Neural Computation, School of Informatics, The University of Edinburgh, Edinburgh, United Kingdom
5 MRC Human Genetics Unit, Institute of Genetics, Edinburgh, United Kingdom
6 Intensive Care Unit, Royal Infirmary of Edinburgh, Edinburgh, United Kingdom

P1-13-2 **Comparison of COVID-19 patients' outcomes between Omicron and Delta variants infection**

Laudria Stella Eryvinka[1], Mohamad Saifudin Hakim[2], Hendra Wibawa[3], Khanza Adzkia Vujira[1], Dyah Ayu Puspitarani[1], Fadila Dyah Trie Utami[1], Marcellus Marcellus[1], Gunadi Gunadi[1]

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3 Disease Investigation Center Wates (Balai Besar Veteriner Wates), Yogyakarta, Directorate General, and Livestock Services, Ministry of Agriculture Indonesia

P1-13-3 **Influence of germline variants of IGHV3-53 and IGHV3-66 genes on antibody responses to BNT162b2 mRNA COVID-19 vaccine**

Yoichi Mashimo[1], Keiko Yamazaki[1], Takahiro Kageyama[2], Shigeru Tanaka[2], Toshibumi Taniguchi[3,4], Kazuyuki Matsushita[5], Hidetoshi Igari[3,4], Hideki Hanaoka[6], Koutaro Yokote[7], Hiroshi Nakajima[2,4], Yoshihiro Onouchi[1]

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3 Department of Infectious Diseases, Chiba University Hospital, Japan
4 Chiba University Hospital COVID-19 Vaccine Center, Japan
5 Division of Laboratory Medicine, Chiba University Hospital, Japan
6 Clinical Research Center, Chiba University Hospital, Japan
7 Department of Endocrinology, Hematology and Gerontology, Chiba University Graduate School of Medicine, Japan

P1-13-4 **Severe clinical manifestations in an extremely low birth weight preterm baby with vascular Ehlers-Danlos syndrome**

Shujiro Hayashi

Dokkyo Medical University, Japan

Poster Session 1-14 Genetic Counseling in Asia 1

Date ：Thursday, October 12, 2023 17:30 ～ 18:00 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P1-14-1 **The road to recognition: Current genetic counseling practice in Indonesia**

Ziske Maritska[1,2], Sultana MH Faradz[2,3]

1 Department of Biology Medicine, Faculty of Medicine, Universitas Sriwijaya, Palembang, Indonesia
2 Indonesian Society of Genetic Counselors (ISGC), Indonesia
3 Diponegoro National Hospital, Semarang, Indonesia

P1-14-2 **Quick survey on awareness of genetic counselor profession among the first year medical students in Palembang, Indonesia**

Ziske Maritska

Universitas Sriwijaya, Indonesia

P1-14-3 **Withdrawn**

P1-14-4 **A case of severe fetal ARPKD with oligohydramnios in the second trimester of pregnancy, treated with genetic counseling**

Chikara Kihira, Kouichiro Kido, Yumi Suzuki, Kakushou Takahashi, Daejoon Yoon, Tsuyoshi Terashima, Keita Yatsuki, Miki Nishizawa, Hideo Kamata, Haruko Hiraike, Yukifumi Sasamori, Ryo Eiji, Kazunori Nagasaka

Teikyo University, Japan

P1-14-5 **Development of the professional standards and the pathway to certification of genetic counsellors in Malaysia**

Sook-Yee Yoon[1], Suzanah Abd Hamid[2], Yee Ling Cheah[9], Gaik Siew Ch'ng[3], Tiara Hassan[4], Wee Teik Keng[5], Juliana Mei Har Lee[6], Huey Yin Leong[5], Lip Hen Moey[5], Rifhan Azwani Mazlan[7], Winnie Pei Tee Ong[5], Sharifah Azween Syed Omar[8], Meow Keong Thong[7], Shing Yiing Tiong[9]

1 Genetic Counselling Society Malaysia, Malaysia
2 Sabah Women and Children Hospital, Malaysia
3 Department of Genetics, Penang Hospital, Malaysia
4 Genetic Counselling Unit, Cancer Research Malaysia, Malaysia
5 Department of Genetics, Hospital Kuala Lumpur, Malaysia
6 Genetic Counselling Asia, Malaysia
7 Genetics Medicine Unit, University Malaya Medical Centre, Malaysia
8 Department of Paediatrics, Hospital Canselor Tuanku Muhriz, Malaysia
9 Loh Guan Lye Specialists Centre, Malaysia

P1-14-6 **Enhancing genetic counseling in Thailand: Challenges, global lessons, and collaborative strategies**

Naravut Suvannang

The Secret Lab, Thailand

P1-14-7 **Genetic counseling and CGP testing for the patient with advanced gastric cancer and pre-existing FAP: A case report**

Mizuki Takatsu[1], Junko Kimura[1], Mika Tsukahara[1], Mitsutoshi Nomura[1], Hisashi Shimojo[1,2], Masato Nakamura[1,3], Hideaki Moteki[1]

1 Aizawa Hospital, Japan
2 Department of Pathology, Aizawa Hospital, Japan
3 Aizawa Comprehensive Cancer Center, Aizawa Hospital, Japan

P1-14-8 **Identifying patient factors related to genetic counseling visits for von Hippel-Lindau syndrome**

Masako Torishima[1], Akiko Yoshida[1], Akira Inaba[2], Shinji Kosugi[1]

1 Kyoto University School of Public Health, Japan
2 Clinical Genetics Unit, Kyoto University Hospital, Japan

P1-14-9 **A novel compound hetero mutation of PDZD7 associated with moderate non-syndromic hearing loss in a Japanese family**

Yumiko Kobayashi[1], Kayono Yamamoto[1], Akimune Fukushima[1], Shin-ya Nishio[2], Shin-ichi Usami[2]

1 Iwate Medical University, Japan
2 Shinshu University, Japan

Poster Session 2-01 Complex Diseases and Genomic Risk Assessment 2

Date ：Friday, October 13, 2023 18:10 ～ 18:40 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P2-01-1 **Genome-wide association studies identify a susceptibility locus to pelvic organ prolapse in the Japanese**

Minako Imamura[1,2], Masatoshi Matsunami[1], Asuka Ashikari[3], Xiaoxi Liu[4], Rikako Nakamoto[1], Masahiko Isa[1], Azeem Javed[1], Masahiro Yoshida[1], Noriko Ohyama[1], Naoko Miyagawa[4], Kohei Tomizuka[4], Keiko Hikino[5], The Biobank Japan Project[6], Koichi Matsuda[7], Chikashi Terao[4], Minoru Miyazato[8], Shiro Maeda[1,2]

1 Department of Advanced Genomic and Laboratory Medicine, Graduate School of Medicine, University of the Ryukyus, Japan
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3 Department of Urology, Graduate School of Medicine, University of the Ryukyus, Japan
4 Laboratory for Statistical and Translational Genetics, RIKEN Center for Integrative Medical Sciences, Japan
5 Laboratory for Pharmacogenomics, RIKEN Center for Integrative Medical Sciences, Japan
6 Institute of Medical Science, The University of Tokyo, Japan
7 Department of Computational Biology and Medical Sciences, Graduate School of Frontier Sciences, The University of Tokyo, Japan
8 Department of Systems Biology, Graduate School of Medicine, University of the Ryukyus, Japan

P2-01-2

An evaluation of polygenic risk score for atrial fibrillation in a working-age cohort

Koki Tanaka[1], Naoki Itokawa[1], Tomoko Takahashi[2], Masaru Koido[1], Naoki Asanoma[3], Nagisa Shiomi[4], Akinori Fujino[4], Yoichiro Kamatani[1], Michiko Seyama[5], Toru Suzuki[2], Yoshinori Murakami[2,6]

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2 Project Division of Genomic Medicine and Disease Prevention, The Institute of Medical Science, The University of Tokyo, Japan
3 NTT Smart Data Science Center and NTT Computer and Data Science Laboratories, Nippon Telegraph and Telephone Corporation, Japan
4 Bio-Medical Informatics Research Center and NTT Basic Research Laboratories, Nippon Telegraph and Telephone Corporation, Japan
5 Medical Business Planning Office, Nippon Telegraph and Telephone Corporation, Japan
6 Division of Molecular Pathology, The Institute of Medical Science, The University of Tokyo, Japan

P2-01-3

Genome-wide association studies by anti-hypertensive drug classes reveal complex pathogenesis of resistant hypertension

Keiko Yamazaki[1], Chikashi Terao[3], Atsushi Takahashi[4], Yoichiro Kamatani[5], Koichi Matsuda[5], Yasuo Takahashi[2]

1 Chiba University, Japan
2 Nihon University School of Medicine, Japan
3 RIKEN Center for Integrative Medical Sciences, Japan
4 National Cerebral and Cardiovascular Center, Japan
5 The University of Tokyo, Japan

P2-01-4

Genome-wide association study of narcolepsy type 1 in multiple populations

Taku Miyagawa[1,2], Hanna Ollila[3], Hiromi Toyoda[2], Seik-Soon Khor[2,4], Mihoko Shimada[1,2,4], Emmanuel Mignot[3], Katsushi Tokunaga[2,4]

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2 The University of Tokyo, Japan
3 Stanford University, USA
4 National Center for Global Health and Medicine, Japan

P2-01-5

Exome sequencing reveals rare genetic variants associated with the risk of paroxysmal atrial fibrillation

Kanji Tabata[1,2], Takeaki Sudo[3], Yuki Nagata[1,4], Takamasa Ichikawa[1,4], Kensuke Ihara[5], Ken Asada[13], Yasuaki Tanaka[6], Yasuteru Yamauchi[7], Takeshi Sasaki[8], Hitoshi Hachiya[9], Yasushi Imai[10], Hideo Fujita[11], Tetsuo Sasano[12], Tetsushi Furukawa[5], Takanori Iwata[2], Toshihiro Tanaka[1,4]

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11 Division of Cardiovascular Medicine, Saitama Medical Center, Jichi Medical University, Saitama, Japan
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13 Cancer Translational Research Team, RIKEN Center for Advanced Intelligence Project and an External Research Staff of Medical AI Research and Development, National Cancer Center Research Institute, Japan

P2-01-6

Association analysis of HLA-B and KIR genes in ankylosing spondylitis

Aya Kawasaki[1], Ikue Ito-Naito[1,2], Kurisu Tada[3], Makio Kusaoi[3], Keita Yamashita[1,4], Kumiko Shimoyama[5], Hajime Kono[6], Noriyoshi Ogawa[5], Naoto Tamura[3], Naoyuki Tsuchiya[1]

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6 Department of Internal Medicine, Teikyo University School of Medicine, Japan

P2-01-7 Withdrawn

P2-01-8 Acceleration of tau aggregation by synergistic effect of CD40 and CD48 gene in Alzheimer's disease

Sung-Hyun Kim[1], Sumin Yang[1], Key-Hwan Lim[2], Jae-Yeol Joo[1]

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2 College of Pharmacy, Chungbuk National University, Korea

P2-01-9 Development of bias correction methods for cord blood-based epigenome-wide association study

Hideki Ohmomo[1,2,3], Akira Takashima[2], Shiori Minabe[2,3], Yoichi Sutoh[2,3], Kanako Ono[2], So Umekage[2], Shohei Komaki[2,3], Yayoi Otsuka-Yamasaki[2,3], Tsuyoshi Hachiya[2,3], Rie Oyama[4], Tsukasa Baba[4], Makoto Sasaki[2,5], Atsushi Shimizu[2,3]

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5 Division of Ultrahigh Field MRI, Institute for Biomedical Sciences, Iwate Medical University, Japan

P2-01-10 Investigating the association of FOXE1 variant in the etiology of non-syndromic orofacial clefts in a Kuwaiti cohort

Amani AL-Adsani, Nada Abdelhafez, Lateefa Al-Kharafi, Suzanne Al-Bustan

Kuwait University, Kuwait

P2-01-11 Establishment of an immune evaluation system using omics analysis data

Akari Suzuki, Matteo Guerrini, Kazuyoshi Ishigaki, Hiroaki Hatano, Kazuhiko Yamamoto

RIKEN, Japan

P2-01-12 Development of a cross-ethnic polygenic risk scoring method: Taking the Taiwan Biobank and the UK Biobank as examples

Ai-Ru Hsieh, Pin-Hsuan Chiang

Department of Statistics, Tamkang University, Taiwan

P2-01-13 A middle-aged woman with 45, X/46, XX/47, XXX mosaicism: A case report

Keiko Goto-Hirano[1], Yuri Kitamura[1,2], Yoshiteru Arai[2], Ito Kawakami[3], Masaki Nishioka[3], Masami Arai[1]

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3 Department of Psychiatry & Behavioral Science, Juntendo University, Japan

P2-01-14 Detection of copy number variation using digital PCR

Akane Inomoto, Junichi Hosokawa, Keisuke Miyako, Rina Kaneko, Tomomi Tajino, Sakae Itoga, Osamu Ohara

Kazusa DNA Research Institute, Japan

P2-01-15 Application of genetic testing in heritable connective tissue disorders to disease management

Hirokuni Yamazawa, Takahiro Yamada, Ayako Chida-Nagai, Itsumi Sato, Atsuhito Takeda, Yuka Shibata-Ishizaka, Yuna Sasaki, Mio Mukainakano

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Poster Session 2-02　Inherited Metabolic Diseases and Newborn Screening 2

Date　：Friday, October 13, 2023　18:10 ～ 18:40　Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P2-02-1 Newborn screening for acid sphingomyelinase deficiency in Taiwan

An-Ju Lee, Kuan-Chi Tseng, Chao-Chuan Liao, Pin-Wen Chen, Yin-Hsiu Chien, Wuh-Liang Hwu

Department of Medical Genetics and Pediatrics, National Taiwan University Hospital, Taipei, Taiwan

P2-02-2 Identification of a novel homozygous NAXE variant in 6 native Sabahan Malaysian children: A founder mutation?

Winnie Peitee Ong[1], Hock Sin Heng[2], Jun Xiong Lee[2], Elyssa Milus Majawit[2], Mei Yan Chan[1], Nor Azimah Abdul Azize[3], Yusnita Yakob[3], Eugene Lee[4], Rin Khang[4], Go Hun Seo[4], Lock-Hock Ngu[1]

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4 Division of Medical Genetics, 3billion Inc., Seoul, South Korea

P2-02-3 Novel 28 bp deletion variant of the ATP7B gene, c.4022-24_4025del, in a large Thai family with Wilson disease

Dhipsukon Pongborriboon[1], Nattaphon Wansom[2], Noppadol Kietsiriroje[1], Chanin Limwongse[3], Oradawan Plong-On[4], Areerat Hnoonual[4,5], Pornprot Limprasert[4,5]

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4 Department of Pathology, Faculty of Medicine, Prince of Songkla University, Songkhla, Thailand
5 Genomic Medicine Center, Faculty of Medicine, Prince of Songkla University, Songkhla, Thailand

P2-02-4 Lysine metabolites as biomarkers for disorders of mitochondrial iron-sulfur clusters assembly

Parith Wongkittichote, Cassandra Pantano, Matthew Demczko, Amy Goldstein, Xinying Hong, Miao He, Rebecca Ganetzky

Children's Hospital of Philadelphia, USA

P2-02-5 Clinical utility of urinary mulberry bodies/cells testing in the diagnosis of Fabry disease

Katsuya Nakamura[1,2], Saki Mukai[3], Yuka Takezawa[3], Yuika Natori[3], Akari Miyazaki[3], Yuichiro Ide[3], Mayu Takebuchi[3], Kana Nanato[3], Mizuki Katoh[3], Harue Suzuki[3], Akiko Sakyu[1], Tomomi Kojima[1], Emiko Kise[1], Hiroaki Hanafusa[1], Tomoki Kosho[1,4], Koichiro Kuwahara[5], Yoshiki Sekijima[2]

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3 Department of Laboratory Medicine, Shinshu University Hospital, Japan
4 Department of Medical Genetics, Shinshu University School of Medicine, Japan
5 Department of Cardiovascular Medicine, Shinshu University School of Medicine, Japan

P2-02-6 Comprehensive functional annotation of VUS in Japanese pediatric mitochondrial diseases

Ayumu Sugiura[1], Yoshihito Kishita[2], Takanori Onuki[3], Tomohiro Ebihara[4], Tetsuro Matsuhashi[3], Masaru Shimura[3], Takuya Fushimi[3], Noriko Ichino[1], Yukiko Yatsuka[1], Atsuko Imai-Okazaki[1], Kokoro Ozaki[5], Yuma Yamada[6], Hideyoshi Harashima[6], Akira Ohtake[7,8], Kei Murayama[1,3,9], Yasushi Okazaki[1,5]

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4 Department of Neonatology, Chiba Children's Hospital, Japan
5 Laboratory for Comprehensive Genomic Analysis, RIKEN Center for Integrative Medical Sciences, Japan
6 Laboratory for Molecular Design of Pharmaceutics, Faculty of Pharmaceutical Sciences, Hokkaido University, Japan
7 Department of Pediatrics & Clinical Genomics, Faculty of Medicine, Saitama Medical University, Japan
8 Center for Intractable Diseases, Saitama Medical University Hospital, Japan
9 Center for Medical Genetics, Chiba Children's Hospital, Japan

P2-02-7 **Investigating common mutations and prevalence of Wilson's disease in Thai population using whole-genome population data**

Paravee Own-eium[1], Donniphat Dejsuphong[1], Prin Vathesatogkit[2], Thanyachai Sura[3], Bhoom Suktitipat[4], Jakris Eu-ahsunthornwattana[5]

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Poster Session 2-03 Prenatal Genetics 2

Date ：Friday, October 13, 2023 18:10 ～ 18:40 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P2-03-1 **Current status of NIPT initiated at our hospital and results of questionnaire**

Shiori Tsuge, Sanae Shinone, Satoshi Matsukawa, Hiromi Nakamura

Gifu Prefectural Tajimi Hospital, Japan

P2-03-2 **Three pregnant patients with Osteogenesis imperfecta**

Yuya Tanaka[1], Yoshifumi Kasuga[1], Takeshi Arimitsu[2], Takeshi Sato[2], Takane Kinn[2], Satsuki Nakano[2], Moe Kusakawa[3], Mamiko Yamada[4], Keisuke Akita[1], Kunio Tanaka[1], Yuka Fukuma[1], Junko Tamai[1], Keita Hasegawa[1], Toshimitsu Otani[1], Satoru Ikenoue[1], Mariko Hida[2], Gen Nishimura[5], Tomonobu Hasegawa[2], Mamoru Tanaka[1]

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3 Department of Pediatric, Saitama City Hospital, Japan
4 Center for Medical Genetics, Keio University School of Medicine, Japan
5 Department of Radiology, Hospital Musashino Youwa Hospital, Japan

P2-03-3 **A case report of trisomy 16 only in the placenta with small omphalocele**

Momoko Kato, Nobuhiko Hayashi

The Fetal Clinic Tokyo Bay Makuhari, Japan

P2-03-4 **A case with a small supernumerary marker chromosome where CVS and ultrasonography revealed different sexes**

Haruna Okubo[1], Yuki Ito[1], Kana Harada[2], Mikiko Kaneko[2], Yuto Tsuruoka[1], Takeshi Nagao[1], Akihiro Hasegawa[1], Michihiro Yamamura[1], Momoko Inoue[1], Ken Takahashi[1], Michiko Miya[1], Hiroshi Kawame[2], Osamu Samura[1], Aikou Okamoto[1]

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2 The Jikei University Hospital, Department of Clinical Genetics, Japan

P2-03-5 **Changes in awareness of prenatal testing after implementation of the NIPT certification system**

Takeshi Nakamura[1], Nahoko Shirato[1], Tatsuko Hirose[1,2], Shin Ikebukuro[1], Keiko Miyagami[1], Takahiro Yamada[3], Akihiko Sekizawa[1]

1 Showa University School of Medicine, Japan
2 Showa University Graduate School of Health Sciences, Japan
3 Hokkaido University Hospital Clinical Genetics, Japan

P2-03-6 **Is chromosome testing using amniocentesis essential to confirm the diagnosis in NIPT-positive cases?**

Yasushi Nakamura[1], Chieko Tamura[1], Kenji Yamada[1], Chikoto Ihara[1], Seiji Kanazawa[1], Chikara Kihira[2], Mihyon Song[3]

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2 Department of Obstetrics & Gynecology, Teikyo University School of Medicine, Japan
3 Marunouchi-no-mori Ladies' Clinic, Japan

P2-03-7 **Six cases with the indeterminate results in NIPT at Hiroshima Red Cross Hospital & Atomic-bomb Survivors Hospital**

Miho Kodama, Norio Miharu, Itsuka Kai, Takayo Shoji, Minako Hikita, Rikako Nakamae, Kenjiro Date

Hiroshima Red Cross Hospital & Atomic-bomb Survivors Hospital, Japan

P2-03-8 **Non-Invasive prenatal testing for hotspot mutations in Chinese population by droplet digital PCR**

Kai Yan, Yeqing Qian, Bei Liu, Na Chen, Xiaoyang Gao, Minyue Dong

Department of Reproductive and Genetics, Women's Hospital, School of Medicine, Zhejiang University, China

P2-03-9 **Mutation-free baby born from a Vici Syndrome carrier after preimplantation genetic testing (PGT)**

Yuki Mizuguchi[1,2], Kou Sueoka[1], Suguru Sato[1], Mamoru Tanaka[1]

1 Keio University School of Medicine, Japan
2 Nasu Red Cross Hospital, Japan

P2-03-10 **A case of fetal thanatophoric dysplasia type 1 diagnosed with fetal ultrasound and genetic test**

Yoshiki Maeda[1], Asumi Ohmori[2], Ryo Suzuki[2], Miho Izawa[2], Ryoko Ono[2], Masahiro Nakao[2,3], Hiroko Morisaki[2], Chinami Horiuchi[2]

1 Kuwana City Medical Center, Sakakibara Heart Institute, Japan
2 Sakakibara Heart Institute, Japan
3 Auckland University, New Zealand

P2-03-11 **Chromosomal analysis of parental chromosomal aberrations with recurrent pregnancy loss – single institution analysis**

Ikuno Kawabata[1], Tomoko Sahara[1], Sayuri Hiraoka[1], Tomoko Ichikawa[1], Shunji Suzuki[1], Hidehiko Miyake[2], Takeshi Yamada[1]

1 Nippon Medical School Hospital, Japan
2 Department of Genetic Counseling, Graduate School of Humanities and Science, Ochanomizu University, Japan

P2-03-12 **Detection of embryos with balanced reciprocal translocation using breakpoint-specific polymerase chain reaction**

Gen Furukawa[1,2], Rie Kawamura[3], Hidehito Inagaki[3], Yoshihiko Sakakibara[4], Yoshimasa Asada[4], Tetsuaki Hara[5], Takeshi Iwasa[6], Akira Kuwahara[6], Minoru Irahara[6], Hiroki Kurahashi[3]

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4 Asada Ladies Clinic, Nagoya, Aichi, Japan
5 Division of Reproductive Medicine, Hiroshima Prefectural Hospital, Hiroshima, Japan
6 Department of Obstetrics and Gynecology, Graduate School of Biomedical Sciences, Tokushima University, Tokushima, Japan

P2-03-13 **PGT-SR using aCGH and FISH analysis for detecting unbalanced chromosome segments involving less than 5Mb: A Case Report**

Keiichi Kato[1], Nami Kawasaki[1], Hiroko Hayashi[1], Kazuki Ohata[1], Tetsuya Miki[1], Akemi Usami[1], Toshiyuki Yamamoto[2], Tomoko Kuroda[1]

1 Kato Ladies Clinic, Japan
2 Institute of Clinical Genetics, Tokyo Women's Medical University Hospital, Japan

P2-03-14 **Three cases of embryos diagnosed with PGT-A abnormality that resulted in live births**

Maki Kusumi, Chisa Tabata, Osamu Tsutsumi

Sanno Hospital, Center for Human Reproduction and Gynecologic Endoscopic Surgery, Japan

P2-03-15 **Psycho-social and genetic counseling issues in reproductive genetic counselling in Malaysia from 2018 to 2022**

Juliana Lee[1,2,3,4]

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2 Genetic Counselling Society of Malaysia, Malaysia
3 National University of Malaysia, Malaysia
4 Professional Society of Genetic Counselors in Asia, Singapore

P2-03-16 **Utility of haplophasing by nanopore sequencing in preclinical setup for PGT-M**

Yui Shichiri[1], Yuri Murase[2], Hidehito Inagaki[1], Ikumi Moriyama[3], Haruki Nishizawa[4], Eiji Sugihara[5], Hiroki Kurahashi[1]

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Poster Session 2-04 Neurology 2

Date ：Friday, October 13, 2023 18:10 ～ 18:40 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P2-04-1 **Complete SAMD12 repeat expansion sequencing in a four-generation BAFME1 family with anticipation**

Takeshi Mizuguchi[1], Tomoko Toyota[2], Eriko Koshimizu[1], Shinichi Kameyama[1], Hiromi Fukuda[1,3], Naomi Tsuchida[1,4], Yuri Uchiyama[1,4], Kohei Hamanaka[1], Atsushi Fujita[1], Kazuharu Misawa[1], Satoko Miyatake[1,5], Hiroaki Adachi[2], Naomichi Matsumoto[1]

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4 Department of Rare Disease Genomics, Yokohama City University Hospital, Japan
5 Department of Clinical Genetics, Yokohama City University Hospital, Japan

P2-04-2 **Features of pathogenic variants in dysfelrin gene in Japan**

Toshiaki Takahashi[1], Naoki Suzuki[2], Rumiko Izumi[2,3], Chikako Yaginuma[4], Naoko Shimakura[2], Yasuko Shimosegawa[5], Tomoko Totsune[1], Yoko Sugimura[1], Takahiko Sasaki[6], Masaru Yoshioka[1], Toru Baba[1], Hideki Oizumi[1], Hiroyasu Tanaka[1], Hitoshi Warita[2], Tetsuya Niihori[3], Atsushi Takeda[1], Yoko Aoki[3], Masashi Aoki[2]

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3 Department of Medical Genetics, Tohoku University School of Medicine, Japan
4 Departments of Clinical Laboratory and Division of Clinical Research, National Hospital Organization Sendai-Nishitaga Hospital, Japan
5 Departments of Neurosurgery, National Hospital Organization Sendai-Nishitaga Hospital, Japan
6 Departments of Internal Medicine and Division of Clinical Research, National Hospital Organization Sendai-Nishitaga Hospital, Japan

P2-04-3 **A Japanese boy with muscle weakness and elevated serum CK was detected with compound heterozygous variants in POMGNT2**

Hiroaki Hanafusa[1], Yoshinori Nambu[1], Shoko Sonehara[1], Ryosuke Bo[1], Kandai Nozu[1], Hiroyuki Awano[1,2]

1 Kobe University Graduate School of Medicine, Japan
2 Organization for Research Initiative and Promotion, Tottori University, Japan

P2-04-4 **Generation of a model cell system to track trisomy correction during reprogramming in aneuploidy syndromes**

Silvia Akutsu, Risa Matsumura, Takaki Asano, Shinya Matsuura

Hiroshima University, Japan

P2-04-5 **Investigation of the clinical course of SMN2 gene 4-copy cases: In relation to the "4-copy problem" in newborn screening**

Mamoru Yokomura, Tamaki Kato, Mari Urano, Mayuri Ito, Kayoko Saito

Tokyo Women's Medical University, Japan

P2-04-6 **Comprehensive genetic analysis in Japanese Parkinson's disease**

Hiroyo Yoshino[1], Yuanzhe Li[1,2], Aya Ikeda[2], Arisa Hayashida[2], Kensuke Daida[2], Mayu Ishiguro[2], Manabu Funayama[1,2], Kenya Nishioka[2], Nobutaka Hattori[1,2,3]

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2 Juntendo University School of Medicine, Japan
3 RIKEN Center for Brain Science, Japan

P2-04-7 **Clinical and epidemiological study of CANVAS (cerebellar ataxia, neuropathy, and vestibular areflexia syndrome) in Japan**

Yuka Hama[1], Hidetoshi Date[1], Hidehiro Mizusawa[2], Yuji Takahashi[1], J-CAT (Japan Consortium of Ataxias)[3]

1 National Center Hospital, National Center of Neurology and Psychiatry, Japan
2 National Center of Neurology and Psychiatry, Japan
3 J-CAT (Japan Consortium of Ataxias), Japan

P2-04-8 **Long-read whole-genome sequencing and phasing analyses of a patient alone revealed complex biallelic CC2D2A variants**

Kumiko Yanagi[1], Kazuhito Satou[1], Arisa Igarashi[1], Tomomi Hidai[1], Taiga Aoki[1], Takahiko Iida[1], Masahiko Yamamori[1], Yoichi Matsubara[2], Tadashi Kaname[1]

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Poster Session 2-05 Differences of Sex Development

Date ：Friday, October 13, 2023 18:10 ～ 18:40 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P2-05-1 **Blepharophimosis-ptosis-epicanthus inversus syndrome diagnosed with uterine cancer**

Sana Yokoi[1], Ryoko Suzuki[1], Reiko Ohara[2]

1 Chiba Cancer Center, Japan
2 Kimitsu Chuo Hospital, Japan

P2-05-2 **Microdeletion within ESR1 constitutes a susceptibility factor for the development of undermasculinized genitalia**

Yohei Masunaga[1], Yasuko Fujisawa[1], Maki Fukami[2], Hirotomo Saitsu[1], Naoyuki Kamatani[3], Tsutomu Ogata[4]

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2 National Research Institute for Child Health and Development, Japan
3 StaGen Co., Ltd., Japan
4 Hamamatsu Medical Center, Japan

P2-05-3 **Karyotypes of 20 cases of Turner syndrome experienced in our department**

Ruriko Maruyama, Masatoshi Yamaguchi, Koutarou Doi, Shinji Katsuragi

Miyazaki University, Japan

P2-05-4

A rare case report of primary amenorrhea-associated turner syndrome and mullerian agenesis

M. P. Budyandini Dyah Pramesti[1,2], Ria Margiana[1,3,4,5], Tjahjo Djojo Tanodjo[1,5]

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3 Department of Anatomy, Faculty of Medicine, Universitas Indonesia, Jakarta, Indonesia
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P2-05-5

Phenotype and genotype of lipoid congenital adrenal hyperplasia due to StAR gene mutation

Thu Ha Nguyen, Chi Dung Vu, Phuong Thao Bui, Ngoc Khanh Nguyen, Thi Bich Ngoc Can

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Poster Session 2-06 Technological Advances, Wet and Dry 2

Date ：Friday, October 13, 2023 18:10 ～ 18:40 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P2-06-1

Enhancing Aspergillus IgG with biomarkers and deep learning for chronic pulmonary aspergillosis diagnosis and outcomes

Chia-Ni Hsiung[1], Meng-Rui Lee[2]

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2 Department of Internal Medicine, National Taiwan University Hospital, Taiwan

P2-06-2

Whole transcriptome RNA sequencing reveals distinct gene set enrichment profile in eosinophilic chronic rhinosinusitis

Tomomitsu Hirota[1], Natsuki Inoue[1,2], Daiki Nakashima[1,3], Eri Mori[3], Kazuhiro Omura[3], Tsuguhisa Nakayama[3,4], Nobuyoshi Otori[3], Hiromi Kojima[3], Mayumi Tamari[1]

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3 Department of Otorhinolaryngology, The Jikei University School of Medicine, Japan
4 Department of Otorhinolaryngology, Head and Neck Surgery, Dokkyo Medical University, Japan

P2-06-3

Unraveling Zika virus impact on the CNS: Systems biology insights into immune-mediated neurodevelopmental changes

Tojo Nakayama[1,2], Kimino v[1,3], Amanda Guise[1], Christoph Schlaffner[1], Anais Meziani[1], Mukesh Kumar[1], Long Cheng[1], Dylan Vaughan[1], Andrew Kodani[4], Simon Van Haren[1], Kenneth Parker[5], Ofer Levy[1,8], Ann Durbin[6,9], Irene Bosch[6,9], Lee Gehrke[6,9], Hanno Steen[1], Ganeshwaran Mochida[1,7], Judith Steen[1]

1 Boston Children's Hospital, USA
2 Tokyo Medical and Dental University, Japan
3 Keio University School of Medicine, Japan
4 St. Jude Children's Research Hospital, USA
5 SimulTOF Systems, USA
6 Harvard Medical School, USA
7 Massachusetts General Hospital, USA
8 Broad Institute of Massachusetts Institute of Technology and Harvard, USA
9 Massachusetts Institute of Technology, USA

P2-06-4

Characteristics of sequences and variants of cancer-related genes focusing on codon usage and 2AA patterns：Part1

Yuta Hamano, Nao Kamae, Mika Mizoguti, Takesi Kumagai, Takuya Sugimoto, Nami Ota, Sawako Minami

Wakayama Medical University Hospital, Japan

P2-06-5

eQTL analysis for full length transcripts using long-read technology reveals a lot of splicing variant-specific eQTLs

Yuya Nagura[1], Mihoko Shimada[2], Akihiro Fujimoto[1]

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2 Genome Medical Science Project (Toyama), National Center for Global Health and Medicine (NCGM), Japan

P2-06-6

Genomic and transcriptomic analysis using long-read data by sequence reconstruction

Ko Ikemoto, Akihiro Fujimoto

School of Medicine, The Universisty of Tokyo, Japan

P2-06-7

Investigating the role of repeat elements in promoting the transition of MCF-7 cells to LTED cells

Shengliang Ni[1], Xufeng Shu[1,4], Martin C Frith[1,2,3]

1 Graduate School of Frontier Sciences, The University of Tokyo, Japan
2 Artificial Intelligence Research Center, AIST, Tokyo, Japan
3 Computational Bio Big-Data Open Innovation Laboratory, AIST, Tokyo, Japan
4 RIKEN Center for Integrative Medical Sciences, Yokohama, Japan

P2-06-8

Target-capture long-read sequencing revealed novel intron retention in patient with tuberous sclerosis complex

Hiroki Ura[1], Sumihito Togi[1,2], Yo Niida[1,2]

1 Kanazawa Medical University Hospital, Japan
2 Kanazawa Medical University, Japan

P2-06-9

Full-genome analysis in diagnoses of patients suspicious of genetic disorders

Ni-Chung Lee[1], Hsiao-Jung Kao[3], Hung-Lun Chiang[4], Hsiao-Huei Chen[3], Yen-Yin Chou[5], Hsueh-Wen Hsueh[6], Sung-Tsang Hsieh[6], Pi-Chuang Fan[1], Yi-Fang Tu[5], Ru-Li Lin[7], Yin-Hsiu Chien[1,2], Wuh-Liang Hwu[1,2], Chien-Ling Lin[4], Pui-Yan Kwok[3,4,8]

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P2-06-10

Evaluation of a novel on-demand genetic testing method, targeted RNA long-amplicon sequencing (rLAS)

Sumihito Togi[1,2], Hiroki Ura[1,2], Hisayo Hatanaka[2], Yo Niida[1,2]

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P2-06-11

Long-read sequencing revealing intragenic deletions in exome-negative spastic paraplegias

Hiromi Fukuda[1,2], Takeshi Mizuguchi[1], Hiroshi Doi[2], Shinichi Kameyama[1,3], Misako Kunii[2], Hideto Joki[2,4], Tatsuya Takahashi[4], Hiroyasu Komiya[2], Mei Sasaki[5], Yosuke Miyaji[2], Sachiko Ohori[1,6,7], Eriko Koshimizu[1], Yuri Uchiyama[1,8], Naomi Tsuchida[1,8], Atsushi Fujita[1], Kohei Hamanaka[1], Kazuharu Misawa[1,9], Satoko Miyatake[1,6], Fumiaki Tanaka[1], Naomichi Matsumoto[1]

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9 RIKEN Center for Advanced Intelligence Project, Japan

P2-06-12 **Analysis of variants of secondary findings from clinical sequencing at our hospital**

Naoko Sato[1], Masaki Tanaka[1], Junko Nomoto[1], Kanako Fukushima[2], Masakazu Nishigaki[2], Shoji Tsuji[1,2]

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Poster Session 2-07 Cancer Genomics, Germline 2

Date ：Friday, October 13, 2023 18:10 ～ 18:40 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P2-07-1 **A case of hereditary paraganglioma-pheochromocytoma syndrome with pathogenic variant in SDHA**

Natsuki Naka, Miho Kakuta, Katsuya Iuchi, Goh Yamamoto, Kiwamu Akagi

Saitama Cancer Center, Japan

P2-07-2 **Characteristics of pathogenic germline variants with BRCA1/2 in unselected Japanese patients with ovarian cancer**

Ayaka Saito[1,2], Akira Hirasawa[2,3], Maki Tanioka[4], Kanako Tsukamoto[1], Kazuyo Kiribayashi[1], Naofumi Watanabe[1,5], Mizuki Takano[1], Kako Kuroiwa[1], Rioko Iida[1], Yurie Sato[1], Orie Kobayashi[1], Kazuya Tamura[1], Satoshi Umezawa[1]

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4 Medical AI project, Dentistry and Pharmaceutical Sciences, Okayama University, Okayama, Japan
5 Department of Genetic Medicine, Fukushima Medical University Hospital, Japan

P2-07-3 **Hereditary tumor-specific chatbot system with Large Language Model and LangChain**

Mashu Futagawa[1,2], Ryuuichi Nakahara[3], Maki Tanioka[4], Akira Hirasawa[1,2]

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P2-07-4 **A Family with BAP1 tumor predisposition syndrome identified by cancer genome profiling test and confirmatory testing**

Haruka Yamamoto[1], Motoko Sasaki[1,2], Tomohiro Nakayama[1], Sachio Tsuchida[1], Katsuhiro Miura[1], Hiroshi Umemura[1], Masahiko Tanabe[3], Katsutoshi Oda[3]

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3 The University of Tokyo Hospital, Japan

P2-07-5

Open-ended responses to a multicenter survey of the secondary finding disclosure process for cancer genome profiling

Saki Shimada[1,2], Takahiro Yamada[1,3,15], Akari Minamoto[4], Manami Matsukawa[1,5,15], Ichiro Yabe[3,15], Yoko Aoki[6,15], Katsutoshi Oda[7,15], Arisa Ueki[8,15], Satomi Higashigawa[9,15], Maki Morikawa[10,15], Yuki Sato[11,15], Akira Hirasawa[12,15], Masanobu Ogawa[1,13,15], Tomohiro Kondo[14,15], Masahiro Yoshioka[14,15], Masashi Kanai[14,15], Manabu Muto[14], Shinji Kosugi[1,15]

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7 Division of Integrative Genomics, The University of Tokyo, Japan
8 Center for Medical Genetics, Keio Cancer Center, Keio University School of Medicine, Japan
9 Division of Clinical Genetics, Shizuoka Cancer Center, Japan
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P2-07-6

Current status and issues of genetic medicine after cancer gene panel testing in our hospital

Naomi Araki[1], Hideyasu Tsumura[1,2], Sachiko Ohori[1], Rika Kawata[1], Masao Araki[1], Segi Furukawa[1,3], Mina Waraya[1], Jiichiro Sasaki[4], Fumio Takada[1,5]

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P2-07-7

Challenges and opportunities in cancer genomics practice in a cooperative hospital for cancer genomic medicine

Shogo Watari[1,2], Akira Hirasawa[3], Hiromasa Shiraishi[2], Moto Tokunaga[2], Risa Kubota[2], Norihiro Kusumi[2], Takaharu Ichikawa[2], Tomoyasu Tsushima[2], Yoko Shinno[4], Tomohiko Mannami[5], Haruhiro Yamashita[6], Ichiro Akiyama[7], Kiichiro Kanamitsu[8], Mutsuko Yamashita[9], Kazutaka Sunami[10]

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P2-07-8

Disclosure of secondary findings in comprehensive genomic profiling (CGP) at Sapporo Medical University Hospital

Yumi Tanaka[1], Kohichi Takada[2], Tomohiro Kubo[2], Yohei Arihara[2], Ayako Murota[2], Tasuku Mariya[2], Masashi Idogawa[2], Sachiko Miyazaki[2], Aki Ishikawa[2], Akihiro Sakurai[2]

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P2-07-9 **Factors related to the selection of blood relatives to receive the examination in BRACAnalysis-positive individuals**

Ryoko Suzuki, Reiko Ohara, Sana Yokoi

Chiba Cancer Center, Japan

P2-07-10 **Familial cases of Rhabdoid Tumor Predisposition Syndrome caused by germline mosaicism of SMARCB1 mutation**

Ryojun Takeda[1,2,3], Takashi Kurata[4], Hirokazu Morokawa[4], Kazutoshi Komori[4], Kazuo Sakashita[3,4], Tomomi Yamaguchi[5,6,7], Tomoki Kosho[2,5,6,7,8]

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8 Research Center for Advanced Science and Technology, Shinshu University, Japan

P2-07-11 **Association between patient' background factors and contralateral risk-reduction mastectomy**

Ai Motoyoshi, Runa Sugiyama, Mizuho Tazo, Yasuyuki Kojima, Maho Ogiwara, Minami Ozawa, Yuki Suzuki, Yodo Sugishita, Ohsuke Migita, Koichiro Tsugawa

St. Marianna University School of Medicine, Japan

P2-07-12 **Prevalence of BRCA1/2 pathogenic variants in triple negative breast cancer : A single center retrospective study**

Yuichi Ueda[1], Hiroshi Kiyohara[1], Mayumi Funagayama[1], Naoko Ikeda[1], Akiko Ishikawa[2], Eri Seike[2], Hiromi Koyama[2], Tsugumi Nagatomo[2], Megumi Mitsumatsu[2], Katsunori Abe[3], Suguru Uwai[3], Masatoshi Yamaguchi[4], Minayo Iwai[4], Shinya Makino[4], Junko Kawano[5], Shugo Tamada[1]

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P2-07-13 **A Case of RRM for maintaining CR after bone recurrence in BRCA mutation-positive asynchronous bilateral breast cancer**

Michiko Tsuneizumi[1,2], Ryoichi Matsunuma[1], Shoko Sato[1], Sae Imada[1], Rousuke Hayami[1], Tatsunori Sato[2], Yuji Kanazawa[2], Hiroyuki Ariyasu[2], Kouhei Saito[2], Rei Gou[2], Masayo Ukita[2], Rieko Kosugi[2], Masashi Harasaki[2], Takeshi Usui[2]

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P2-07-14 **A Case of natural pregnancy following prophylactic total colectomy for familial adenomatous polyposis**

Yoko Aoyagi[1], Kentaro Kai[1], Hidefumi Shiroshita[2], Takashi Masuda[2], Eiji Kobayashi[1]

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2 Department of Gastroenterological and Pediatric Surgery, Oita University, Japan

P2-07-15 **Status of tumor/germline BRCA testing and genetic counseling in patients with advanced ovarian cancer at our institution**

Yoshikazu Nagase, Yumi Nakao, Fuyuki Ichikawa, Naoko Komura, Asuka Tanaka, Akihiko Yoshimura, Kumi Masuda, Fujihiro Oka, Takeshi Yokoi

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Poster Session 2-08　Pediatric Genetics 2

Date　：Friday, October 13, 2023　18:10 ～ 18:40　Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P2-08-1　A case of 9q22.32q31.2 deletion involving ZNF462

Ikuko Ohashi[1,2], Misao Kageyama[3], Miho Nagata[4], Yasutaka Ishihara[4], Yohei Miyashita[4], Yoshihiro Asano[4], Yasuko Yamanouchi[2,6], Kayo Takao[2], Kazumi Tawa[2], Takanobu Otomo[2,5], Mitsuo Masuno[6]

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5 Department of Molecular and Genetic Medicine, Kawasaki Medical School, Japan
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P2-08-2　Global developmental delay with abnormal brain MRI and feeding difficulties in a child with DYRK1A mutation

Takato Akiba[1], Shino Shimada[1], Shimpei Matsuda[1], Natsuki Okawa[1], Yosuke Baba[1], Naoya Saijo[2], Atsuo Kikuchi[2], Shigeo Kure[2], Toshiaki Shimizu[2]

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2 Department of Pediatrics, Tohoku University Graduate School of Medicine, Japan

P2-08-3　Pericarditis as a complication of Sotos syndrome

Yu Yamaguchi, Shigeru Nomura

Gunma Children's Medical Center, Japan

P2-08-4　Multi-population meta-analysis implicates immune dysregulation in pediatric steroid sensitive nephrotic syndrome

Makiko Nakayama[1], Alexandra Barry[2,3], Michelle T. McNulty[2,3], Xiaoyuan Jia[1,4], Yask Gupta[5], Hanna Debiec[6], China Nagano[2,3,7], Tomoko Horinouchi[7], Seulgi Jung[8], Yosuke Kwai[1], Kyuyoung Song[8], Hae Il Cheong[9], Prayong Vachvanichsanong[10], Kandai Nozu[7], Katsushi Tokunaga[1], Simone Sanna-Cherchi[5], Pierre Ronco[6,11], Kazumoto Iijima[12,13], Matthew G. Sampson[2,3,14,15]

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12 Hyogo Prefectural Kobe Children's Hospital, Kobe, Japan
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14 Department of Pediatrics, Harvard Medical School, Boston, MA, USA
15 Division of Renal Medicine, Department of Medicine, Brigham and Women's Hospital, Harvard Medical School, Boston, MA, USA

P2-08-5　Genetic autopsy for deceased patients with unknown cause of death: A report from Saitama Children's Medical Center

Mariko Sagara, Sayuri Oda, Kenta Hasumi, Miyu Fukushima, Miwako Kizumi, Yuki Sawada, Miwa Kobayashi, Kei Tonezawa, Takuya Hayashi, Riki Nishimura, Chika Kanno, Kayoko Ichimura, Atsuko Nakazawa, Daiju Oba, Akira Oka, Hirofumi Ohashi

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P2-08-6 **Semi-rapid-NGS diagnosis for severely ill patients: a report from Saitama Children's Medical Center**

Sayuri Oda, Daiju Oba, Satoshi Tonezawa, Takuya Hayashi, Riki Nishimura, Chika Kanno, Kenta Hasumi, Miyu Fukushima, Mariko Sagara, Yuki Sawada, Miwako Kizumi, Masahiro Koyama, Atsuko Nakazawa, Akira Oka, Hirofumi Ohashi

Saitama Children's Medical Center, Japan

P2-08-7 **CHD7, the causative gene of CHARGE syndrome, has a DSB repair function coupled with morphogenesis in fetal development**

Asao Noda, Kaori Muramoto, Shuji Mishima

Radiation Effects Research Foundation, Japan

P2-08-8 **Lower diagnostic rate for undiagnosed multiple malformations and intellectual disability in sibling patients compared to solitary cases by NGS analysis**

Seiji Mizuno[1], Mie Inaba[1], Tomoko Uehara[1], Natsuki Nakamura[1], Shin Hayashi[2]

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2 Institute for Developmetal Research, Aichi Developmental Disability Center, Japan

P2-08-9 **Presymptomatic genetics testing in an infant with a father with the SCN5A-positive Brugada syndrome**

Noriko Onishi[1,2], Masafumi Utsumi[3], Tomomi Yamaguchi[1,4,5], So Nagai[1,2,5], Tomomi Kojima[1], Yoko Yoshida[6], Ramon Brugada[7,8], Takeshi Aiba[9], Tomoki Kosho[1,4,5,10]

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P2-08-10 **IFIH1 gain of function variants in three individuals: A continuum of multiple facets of type I Interferonopathy**

Shino Shimada[1,2,3], Joshi Stephen[3], Sheela Nampoothiri[2,4], Hirotsugu Oda[6], Linnea Laudh[2], Lynne A. Wolfe[2], Camilo Toro[2,5], Cynthia J. Tifft[2,5], David R. Adams[2,3,5], William A. Gahl[2,3], May Christine V. Malicdan[2,3,5]

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P2-08-11 **Exploration of a responsible sequence for aberrant hypermethylation at maternal H19-ICR and BWS-like phenotypes in mice**

Satoshi Hara, Fumikazu Matsuhisa, Shuji Kitajima, Hitomi Yatsuki, Musashi Ichimaru, Ken Higashimoto, Hidenobu Soejima

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P2-08-12 **A case of epilepsy caused by SLC6A1 disruption by de novo balanced chromosomal translocation**

Masamune Sakamoto[1,2,4], Tatsuo Mori[3,5], Takahiro Tayama[3,5], Aya Goji[3,5], Yoshihiro Toda[3,5], Atsushi Fujita[1], Takeshi Mizuguchi[1], Maki Urushihara[3], Naomichi Matsumoto[1]

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P2-08-13 **Clinical phenotype of japanese infants with CEP290-associated leber congenital amaurosis**

Hazuki Morikawa[1,2], Sachiko Nishina[1], Kaoruko Torii[3], Masakazu Takayama[3], Katsuhiro Hosono[3], Tomoka Kanbe[4], Hirotomo Saitsu[3], Maki Fukami[1], Yuichi Hori[2], Yoshihiro Hotta[3]

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3 Hamamatsu University School of Medicine, Japan
4 Saitama Prefectural Children's Medical Center, Japan

P2-08-14 **Identification of different pathogenic variants in siblings with intellectual disability**

Wataru Tanikawa[1], Kenichi Kinjo[1], Yohei Masunaga[1], Yasuko Fujisawa[1], Yoko Masui[2], Konosuke Otaka[2], Tsutomu Ogata[1,3]

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P2-08-15 **Genetic testing averts unnecessary pharmacological treatment in a case of MODY2**

Yumi Matsuyama[1,2], Kumiko Kato[3], Sayaka Ishikawa[4], Kimiko Asai[2], Takenori Ogawa[2], Yukio Horikawa[2]

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4 Department of Pediatrics, Fukui-ken Saiseikai Hospital, Japan

P2-08-16 **A case of infantile spasms with three possible pathogenic de novo missense variants in NF1 and GABBR1**

Kazuki Watanabe[1], Kazuo Kubota[2,3], Mitsuko Nakashima[1], Hirotomo Saitsu[1]

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2 Gifu University Graduate School of Medicine, Japan
3 Gifu University Hospital, Japan

P2-08-17 **Biallelic loss-of-function variants of EZH1 cause a novel developmental disorder with central precocious puberty**

Nobuhiko Okamoto[1], Sayaka Yoshida[2], Yuri Etani[3], Kumiko Yanagi[4], Tadashi Kaname[4]

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4 Department of Genome Medicine, National Center for Child Health and Development, Tokyo, Japan

P2-08-18 **Epigenetic signatures help interpret a nonsense variant with uncertain significance in the last exon of the KMT2A gene**

Kyoko Takano[1,2], Tomoko Kawai[3], Tomomi Yamaguchi[1,2,4], Kazuhiko Nakabayashi[3], Kenichiro Hata[3,5], Shinji Saitoh[6], Tomoki Kosho[1,2,4]

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P2-08-19 A novel GNAO1 variant identified in a patient with clinically diagnosed as cerebral palsy

Osamu Machida[1,2], Taichi Imaizumi[3], Yusaku Miyamoto[3], Rina Shimomura[1,2], Tomoe Yanagishita[2], Keiko Shimojima Yamamoto[4,5], Miho Nagata[6], Yasuki Ishihara[6,7], Yohei Miyashita[6,7], Yoshihiro Asano[6,7], Toshiyuki Yamamoto[1,5]

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P2-08-20 A novel nonsense mutation of the BCL11A gene in a girl with high fetal hemoglobin

Hiroko Kashiwagi[1], Kaoru Ueyama[1], Daisuke Harada[1], Naomichi Matsumoto[3], Noriyuki Namba[2], Yoshiki Seino[1]

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P2-08-21 A novel FBN1 variant associated with mild cardiac phenotype of neonatal Marfan syndrome

Kentaro Shirai[1], Rina Shimomura[2], Senri Kameyama[1], Tsutomu Kondo[3], Toshiyuki Yamamoto[4]

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P2-08-22 Autoimmune disease in Kabuki syndrome

Keisuke Kato[1], Ai Yoshimi[1], Koh-ichiro Yoshiura[2], Yoko Saito-Nakamura[3], Satoru Matsushima[4], Hiroyuki Miyahara[3], Akimitsu Watanabe[3], Masahiro Tsuchida[1]

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P2-08-23 Aberrant RET expressions effect in multifactorial Hirschsprung Disease

Fadila Utami[1], Laudria Stella Eryvinka[1], Verell Christopher Amadeus[1], Setiani Silvi Nurhidayah[1], Kristy Iskandar[2], Eko Purnomo[1], Gunadi[1]

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P2-08-24 A rare FHL inheritance scenario in half-brothers

Chinmayee Bhimarao Nagaraj[1], Shannon Lozinsky[2], Kristina Emeghebo[2], Indira Sahdev[2], Carolyn Levy[2], Teresa Smolarek[1,3], Wenying Zhang[1,3]

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P2-08-25 Response to sandostatin-LAR in a patient with CHI caused by a maternally inherited ABCC8 heterozygous variant

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P2-08-26 A case of osteogenesis imperfecta Type II managed with pamidronate Infusion

Maria Margarita Santiago, Marie Julianne Racoma

The Medical City, Institute of Pediatrics, Philippines

Poster Session 2-09　Rare Diseases 2

Date　：Friday, October 13, 2023　18:10 ～ 18:40　Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P2-09-1 A sibling of mid Xq28 microduplication syndrome with developmental delay diagnosed by chromosomal microarray analysis

Kosuke Yamada[1], Ayumi Kurebayashi[1], Tomomi Murakami[2], Shigeichi Kobayashi[3], Kensuke Otsubo[4], You Fujimoto[4], Atsushi Hamano[5], Hirotsugu Kitayama[6], Kenji Shimizu[1]

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4 Department of Orthopedics, Shizuoka Children's Hospital, Shizuoka, Japan
5 Department of Urology, Shizuoka Children's Hospital, Shizuoka, Japan
6 Department of Nephrology, Shizuoka Children's Hospital, Shizuoka, Japan

P2-09-2 Whole-genome sequencing analysis in 96 families with rare or undiagnosed diseases

Tadashi Kaname[1], Kumiko Yanagi[1], Takaya Iida[1], Taiga Aoki[1], Arisa Igarashi[1], Masahiko Yamamori[1], Kazuhito Satou[1], NCCHD IRUD Cooperative Hospitals[2], Yoichi Matsubara[1]

1 National Center for Child Health and Development, Japan
2 IRUD Cooperative Hospitals of NCCHD, Japan

P2-09-3 Establishment of a mouse model of Sotos syndrome and its phenotypic analysis

Ken Higashimoto[1,5], Keizo Takao[6], Fumikazu Matsuhisa[2], Yoshichika Yoshioka[7], Yuzo Murata[3], Takehisa Sakumoto[4], Satoshi Hara[5], Musashi Ichimaru[5], Hitomi Yatsuki[5], Shuji Kitajima[2], Shigehisa Aoki[4], Koh-ichiro Yoshiura[8], Hidenobu Soejima[5]

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2 Division of Biological Resources and Development, Analytical Research Center for Experimental Sciences, Saga University, Japan
3 School of Health Sciences at Fukuoka, International University of Health and Welfare, Japan
4 Division of Pathology, Department of Pathology and Microbiology, Faculty of Medicine, Saga University, Japan
5 Division of Molecular Genetics and Epigenetics, Department of Biomolecular Sciences, Faculty of Medicine, Saga University, Japan
6 Department of Behavioral Physiology, Faculty of Medicine, University of Toyama, Japan
7 Graduate School of Frontier Biosciences, Osaka University, Japan
8 Department of Human Genetics, Atomic Bomb Disease Institute, Nagasaki University, Japan

P2-09-4 Identification of a candidate drug for the treatment of Facioscapulohumeral muscular dystrophy

Takahiro Yoshizawa[1,2], Yoshitaka Tomiyama[3], Naoko Shiba[4], Tomohide Takaya[5], Daigo Miyazaki[6,10], Tsutomu Nakada[2], Yuji Shiba[2,4], Akinori Nakamura[6,7], Tomoki Kosho[2,8,9,11]

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4 Department of Regenerative Science and Medicine, Shinshu University School of Medicine, Japan
5 Department of Agriculture, Graduate School of Science and Technology, Shinshu University, Japan
6 Department of Medicine (Neurology and Rheumatology), Shinshu University School of Medicine, Japan
7 Department of Neurology, National Hospital Organization Matsumoto Medical Center, Japan
8 Department of Medical Genetics, Shinshu University School of Medicine, Japan
9 Center for Medical Genetics, Shinshu University Hospital, Japan
10 Shinshu Medical Care Collaboration Center (Division of Support for Intractable Disease), Shinshu University Hospital, Japan
11 Division of Clinical Sequencing, Shinshu University School of Medicine, Japan

P2-09-5 Pathophysiological investigation on skeletal manifestations of Musculocontractural Ehlers Danlos Syndrome

Yuki Takahashi[1], Takahiro Yoshizawa[2], Fumiko Ono[1], Shuji Mizumoto[3], Shuhei Yamada[3], Tomoki Kosho[1,4,5,6]

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5 Research Center for Advanced Science and Technology, Shinshu University, Japan
6 Division of Clinical Sequencing, Shinshu University School of Medicine, Japan

P2-09-6

Japanese siblings of cartilage-hair hypoplasia with a novel compound heterozygous variant in RMRP

Naonori Kumagai[1], Yusuke Funato[1], Manabu Wakamatsu[2], Hideki Muramatsu[2], Hiroki Takao[1], Hiroki Kurahashi[1], Haruo Mizuno[1]

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2 Nagoya University, Japan

P2-09-7

NOTCH2NLC GGC repeat expansion in Japanese patients with undiagnosed leukoencephalopathy

Ikuko Mizuta[1], Hiraku Matsuura[1], Chisato Tamai[2], Rei Yasuda[1], Akiko Watanabe-Hosomi[1], Daiki Fukunaga[1], Takashi Koizumi[1,3], Mao Mukai[1], Tomoyuki Ohara[1], Tomokatsu Yoshida[1,4], Jun Sone[2], Toshiki Mizuno[1]

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3 Department of Anatomy and Neurobiology, Graduate School of Medical Science, Kyoto Prefectural University of Medicine, Japan
4 Department of Neurology, Japan Community Health care Organization Kobe Central Hospital, Japan

P2-09-8

Detection of somatic variants in epileptogenic brain lesions

Atsushi Fujita[1], Mitsuhiro Kato[2], Hidenori Sugano[3], Yasushi Iimura[3], Hiroharu Suzuki[3], Jun Tohyama[4], Masafumi Fukuda[5], Yosuke Ito[5], Shimpei Baba[6], Tohru Okanishi[7], Hideo Enoki[8], Ayataka Fujimoto[9], Akiyo Yamamoto[10], Kentaro Kawamura[10], Shinsuke Kato[10], Ryoko Honda[11], Tomonori Ono[12], Hideaki Shiraishi[13], Kiyoshi Egawa[13], Kentaro Shirai[14], Shinji Yamamoto[15], Itaru Hayakawa[16], Hisashi Kawawaki[17], Ken Saida[1], Naomi Tsuchida[1,18], Yuri Uchiyama[1,18], Kohei Hamanaka[1], Satoko Miyatake[1,19], Takeshi Mizuguchi[1], Mitsuko Nakashima[1,20], Hirotomo Saitsu[1,20], Noriko Miyake[1,21], Akiyoshi Kakita[22], Naomichi Matsumoto[1]

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9 Comprehensive Epilepsy Center, Seirei Hamamatsu General Hospital, Japan
10 Department of Pediatrics, Sapporo Medical University School of Medicine, Japan
11 Department of Pediatrics, National Hospital Organization Nagasaki Medical Center, Japan
12 Epilepsy Center, National Hospital Organization Nagasaki Medical Center, Japan
13 Department of Pediatrics, Hokkaido University Graduate School of Medicine, Japan
14 Department of Pediatrics, Tsuchiura Kyodo General Hospital, Japan
15 Department of Neurosurgery, Tsuchiura Kyodo General Hospital, Japan
16 Division of Neurology, National Center for Child Health and Development, Japan
17 Department of Pediatric Neurology, Children's Medical Center, Osaka City General Hospital, Japan
18 Department of Rare Disease Genomics, Yokohama City University Hospital, Japan
19 Department of Clinical Genetics, Yokohama City University Hospital, Japan
20 Department of Biochemistry, Hamamatsu University School of Medicine, Japan
21 Department of Human Genetics, Research Institute, National Center for Global Health and Medicine, Japan
22 Department of Pathology, Brain Research Institute, Niigata University, Japan

P2-09-9

Autophagy enhancement induces steatosis in the developing liver of lars-knock-in zebrafish

Masanori Inoue[1], Wulan Sebastian[1], Hiroaki Miyahara[2], Nobuyuki Shimizu[3], Hiroshi Shiraishi[3], Miwako Maeda[1], Reiko Hanada[4], Toshikatsu Hanada[3], Kenji Ihara[1]

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3 Department of Cell Biology, Oita University Faculty of Medicine, Japan
4 Department of Neurophysiology, Oita University Faculty of Medicine, Japan

P2-09-10 **a-Synuclein pathology is exacerbated by haploinsufficiency of Rop, the STXBP1 homolog in Drosophila melanogaster**

Taro Matsuoka[1], Hideki Yoshida[2], Takashi Kasai[3], Takenori Tozawa[1], Tomohiro Chiyonobu[1,4]

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3 Department of Neurology, Graduate School of Medical Science, Kyoto Prefectural University of Medicine, Kyoto, Japan
4 Department of Molecular Diagnostics and Therapeutics, Graduate School of Medical Science, Kyoto Prefectural University of Medicine, Kyoto, Japan

P2-09-11 **Functional analysis of RRAS2 pathogenic variants presenting Noonan-like phenotype**

Takaya Iida[1], Arisa Igarashi[1], Kai Fukunaga[1,2], Taiga Aoki[1], Kumiko Yanagi[1], Tomomi Hidai[1], Nana Kobayashi[1], Yukimi Abe[1], Kazuhito Satou[1], Yoichi Matsubara[1], Tomoki Kosho[3], Hayato Go[4], Tadashi Kaname[1]

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3 Department of Medical Genetics, Shinshu University School of Medicine, Japan
4 Department of Pediatrics, Fukushima Medical University School of Medicine, Japan

P2-09-12 **Zebrafish model for Loeys-Dietz syndrome**

Rie Chida, Genri Kawahara, Mami Nakayashiki, Hisashi Kawashima, Gaku Yamanaka, Yukiko Hayashi

Tokyo Medical University, Japan

P2-09-13 **Novel deletion and splice-site variant in the ADAMTS3 gene found in patients with Hennekam syndrome**

Arisa Igarashi[1], Yoshio Makita[2], Kumiko Yanagi[1], Tomomi Hidai[1], Makiko Omata[1], Taiga Aoki[1], Takaya Iida[1], Nana Kobayashi[1], Yukimi Abe[1], Kazuhito Satou[1], Yoichi Matsubara[1], Takashi Kaname[1]

1 Dept. of Genome Medicine, National Research Institute for Child Health and Development, Japan
2 Asahikawa Medical University Education Center, Japan

P2-09-14 **Paternal chromosome 6q24 triplication as a cause of Neonatal diabetes mellitus**

Shigeru Suzuki[1], Yuichi Nishikado[2], Kanayo Ochiai[2], Makoto Oshiro[2]

1 Asahikawa Medical University, Japan
2 Japanese Red Cross Aichi Medical Center Nagoya Daiichi Hospital, Japan

P2-09-15 **Diagnostic and clinical utility of genome sequencing in patients with single gene diseases**

Miao-Zi Hung, Ni-Chung Lee, Yin-Hsiu Chien, Wuh-Liang Hwu, Yi-Lin Lin, Ching Hsu, Yu-Hsuan Huang

National Taiwan University Hospital, Taiwan

P2-09-16 **A familial case of Kikuchi-Fujimoto disease across three generations**

Cheryl Weiqi Tan[1], Khadijah Rafi'ee[1], Mark Koh[2], Ene-Choo Tan[1]

1 KK Research Centre, KK Women's and Children's Hospital, Singapore
2 Department of Dermatology, KK Women's and Children's Hospital, Singapore

P2-09-17 **DeSanto-Shinawi syndrome caused by novel missense *WAC* variant: A case report**

Kiwook Jung, Hee Sue Park

Department of Laboratory Medicine, Chungbuk National University Hospital, Cheongju, Korea

P2-09-18 Mutation analysis of the *MECP2* gene in Thai girls referred for suspected Rett Syndrome

Jirakit Sattayapornpipat[1,2], Oradawan Plong-On[1], Supapon Tanpor[3], Chariyawan Charalsawadi[1,2], Areerat Hnoonual[1,2], Pornprot Limprasert[1,2]

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2 Genomic Medicine Center, Faculty of Medicine, Prince of Songkla University, Thailand
3 Graduate Program in Molecular Biology and Bioinformatics, Faculty of Science, Prince of Songkla University, Thailand

P2-09-19 All for One: Laying the Foundation for Precision Health in Canada

Francois Bernier[1,10], Kym Boycott[2,3], Magda Price[2], Kathy Gratton[10], Dennis Bulman[1,10], Jacques Michaud[4,5], Jordan Lerner-Ellis[6,8], Christian Marshall[6,7], Ma'n Zawati[9], Vincent Ferretti[4,5], Meredith Gillespie[2]

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2 Children's Hospital of Ontario Research Institute, Canada
3 University of Ottawa, Canada
4 CHU Sainte-Justine Research Center, Canada
5 Université de Montréal, Canada
6 University of Toronto, Canada
7 The Hospital for Sick Children, Canada
8 Mount Sinai Hospital, Canada
9 McGill University, Canada
10 Alberta Children's Hospital Research Institute, Canada

P2-09-20 DUOX2 frameshift variant in a Japanese case with diffuse goiter and schizophrenia

Kazuyuki Oishi, Takahiro Yoshioka

Kochi Health Science Center, Japan

P2-09-21 Efficient detection of somatic UBA1 variants in patients with clinically suspected VEXAS syndrome

Naomi Tsuchida[1,2,3], Yuri Uchiyama[1,2], Ayaka Maeda[3], Yohei Kirino[3], Hideaki Nakajima[3], Naomichi Matsumoto[2]

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3 Department of Stem Cell and Immune Regulation, Yokohama City University Graduate School of Medicine, Yokohama, Japan

P2-09-22 A family case of metaphyseal anadysplasia type 1 caused by an autosomal dominant variant in exon 2 of MMP13 gene

Tomohiro Hori[1,2], Hideki Matsumoto[1], Mai Mori[1], Hideo Sasai[1,2], Michio Ozeki[1], Norio Kawamoto[1], Hidenori Ohnishi[1,2]

1 Department of Pediatrics, Graduate School of Medicine, Gifu University, Japan
2 Clinical Genetics Center, Gifu University Hospital, Japan

P2-09-23 A first Japanese case with Bryant-Li-Bhoj neurodevelopmental syndrome associated with a novel variant in the H3-3A gene

Sumito Dateki[1,3], Takafumi Yanagi[3], Midori Motokawa[3], Hiroyuki Mishima[4], Koh-ichiro Yoshiura[4], Hiroyuki Moriuchi[2,3]

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3 Department of Pediatrics, Nagasaki University Hospital, Japan
4 Department of Human Genetics, Nagasaki University Graduate School of Biomedical Sciences, Japan

P2-09-24 **Exome analysis of short root anomaly in Japanese population**

Yuki Sagawa[1,2], Takuya Ogawa[1], Takeaki Sudo[3], Yuki Nagata[4,5], Keiji Moriyama[1], Toshihiro Tanaka[4,5]

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3 Institute of Education, Tokyo Medical and Dental University (TMDU), Tokyo, Japan
4 Department of Human Genetics and Disease Diversity, Graduate School of Medical and Dental Sciences, Tokyo Medical and Dental University (TMDU), Tokyo, Japan
5 BioResource Research Center, Tokyo Medical and Dental University (TMDU), Tokyo, Japan

P2-09-25 **Homozygous exon 6-7 deletion of TMEM260 identified in a Japanese family with truncus arteriosus**

Yumi Enomoto, Yukiko Kuroda, Yoko Saito, Takuya Naruto, Kenji Kurosawa

Kanagawa Children's Medical Center, Japan

P2-09-26 **A biallelic missense variant in the GSDMD gene in a patient of atypical Gorham-Stout disease in a consanguineous family**

Yoichi Ezura[1], Daniela Tiaki Uehara[2], Tomoki Muramatsu[2], Senichi Ishii[3], Hidetsugu Suzuki[3], Kazuyuki Fukushima[3], Yasuhiro Arasaki[4], Tadayoshi Hayata[4], Johji Inazawa[2]

1 Teikyo Heisei University, Japan
2 Tokyo Medical and Dental University, Japan
3 Saku Central Hospital Advanced Care Center, Japan
4 Tokyo University of Science, Japan

P2-09-27 **A patient with Coffin-Siris syndrome caused by a novel splice-disruptive variant that appeared to be a missense variant**

Taiga Aoki[1], Mutsumi Akaishi[2], Arisa Igarashi[1], Kumiko Yanagi[1], Takaya Iida[1], Tomomi Hidai[1], Nana Kobayashi[1], Yukimi Abe[1], Kazuhito Satou[1], Hiroki Yonemoto[2], Yoichi Matsubara[1], Takashi Kaname[1]

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P2-09-28 **Diagnostic Yield of Whole Genome Sequencing in 210 Undiagnosed Patients Suspected of Rare Genetic Disorders**

Khunton Wichajarn[1,2], Aree Rattanathongkom[1,2], Kanda Sornkayasit[2]

1 Division of Medical Genetics, Department of Pediatrics, Faculty of Medicine, Khon Kaen University, Thailand
2 Center of Excellent in Precision Medicine, Srinagarind Hospital, Khon Kaen University, Thailand

Poster Session 2-10 Ethical, Legal and Social Implications

Date ：Friday, October 13, 2023 18:10 ～ 18:40 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P2-10-1 **Preliminary study to develop a guide for genomic medicine and research press releases**

Misaki Arakawa, Fuji Nagami

Tohoku University, Japan

P2-10-2 **Japanese Translation of the International Genetics Literacy and Attitudes Survey (iGLAS)**

Akiko Yoshida[1,2], Tomoharu Tokutomi[1,2], Akimune Fukushima[1,2], Robert Chapman[3], Fatos Selita[3], Yulia Kovas[3], Makoto Sasaki[1]

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3 Department of Psychology, Goldsmiths, University of London, UK

P2-10-3 **The balance between duty and death: Thanatophoric dysplasia a case of a lethal congenital malformation**

Rendz Mark Tuazon, Marie Julianne Racoma, James Albert Edward Benitez, Glenn Tolentino

Bataan General Hospital and Medical Center, Philippines

P2-10-4 **Needs survey for materials to obtain informed assents from children participating in whole-genome analysis research**

Tomoko Kobayashi[1], Atsushi Asai[2]

1 Tohoku Medical Megabank Organization (ToMMo), Tohoku University, Japan
2 Department of Medical Ethics, Tohoku University, Japan

P2-10-5 **Policies for the protection of genetic information in Korea and implications for Japan ～Focusing on Ethical Issues**

Hyunsoo Hong, Ayako Kamisato, Kazuyo Arisawa, Yoshiko Takahashi

The University of Tokyo,The Institute of Medical Science, Division of Bioethics, Japan

Poster Session 2-11　Databases / Data sharing / Biobank

Date　：Friday, October 13, 2023　18:10 ～ 18:40　Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P2-11-1 **JOB:Japan Omics Browser provides integrative visualization of multi-omics data**

Yugo Takahashi[1], Qingbo Wang[2], Yukinori Okada[2,3,4], Japan COVID-19 Task Force

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2 Department of Genome Informatics, Graduate School of Medicine, The University of Tokyo, Tokyo, Japan
3 Department of Statistical Genetics, Osaka University Graduate School of Medicine, Suita, Japan
4 Laboratory for Systems Genetics, RIKEN Center for Integrative Medical Sciences, Yokohama, Japan

P2-11-2 **Study towards utilization of intractable disease patient data by data linkage between Shouman and Nanbyo DBs in Japan**

Chisato Yamasaki[1], Saburo Takatsu[1], Akinori Moriguchi[2], Ryuichi Sakate[1]

1 National Institutes of Biomedical Innovation, Health and Nutrition (NIBIOHN), Japan
2 National Center for Child Health and Development (NCCHD), Japan

P2-11-3 **TogoVar 2023: Enhancement and update of publicly available variant data in collaboration with genome projects in Japan**

Nobutaka Mitsuhashi[1], Toshiaki Katayama[1], Minae Kawashima[1], Licht Toyo-Oka[2], Yuki Moriya[1], Shuichi Kawashima[1], Toshihisa Takagi[2]

1 Research Organization of Information and Systems, Japan
2 Toyama University of International Studies, Japan

P2-11-4 **The issues of each database and ontology from the viewpoints of clinicians**

Eisuke Dohi[1], Yuka Tateishi[2], Jae-moon Shin[3], Toyofumi Fujiwara[3], Yasunori Yamamoto[3]

1 National Center of Neurology and Psychiatry, Japan
2 Japan Science and Technology Agency Department of NBDC Program, Japan
3 Research Organization of Information and Systems, Database Center for Life Science, Japan

P2-11-5 **Enhancing Clinical Genetic Analysis through the Medical Genomics Japan Variant Database (MGeND)**

Yosuke Kawai[1], Kazuko Ueno[1], Mayumi Kamada[2], Kenjiro Kosaki[3], Yasushi Okuno[2], Katsushi Tokunaga[1]

1 National Center for Global Health and Medicine, Japan
2 Kyoto University, Japan
3 Keio University School of Medicine, Japan

P2-11-6 **Investigate the differences in the questionnaires used by each NC in the National Centre Biobank Network**

Yukiko Nakashima[1], Satoshi Suzuki[2], Tatsuya Kanto[1], Masaya Sugiyama[1]

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2 Center Hospital, Japan

Poster Session 2-12 Genetic Counseling in Asia 2

Date ：Friday, October 13, 2023 18:10 ～ 18:40 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P2-12-1 **Survey on requests for information on prenatal testing in Japan**

Tatsuko Hirose[1,2], Shin Ikebukuro[2], Keiko Miyagami[2], Takahiro Yamada[3], Osamu Samura[4], Haruhiko Sago[5], Akihiko Sekizawa[2], Nahoko Shirato[2]

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4 Department of Obstetrics and Gynecology, The Jikei University School of Medicine, Japan
5 Center for Medical Genetic, National Center for Child Health and Development, Japan

P2-12-2 **Family trees created during busy outpatient work in a hospital having no specialized department of clinical genetics**

Koji Kumagai, Masahiro Sakai

Osaka Railway Hospital, Japan

P2-12-3 **The group clinic for babies with Down syndrome and their families at SCMC: Annual report of 2022**

Miwako Kizumi, Yuki Sawada, Miwa Kobayashi, Daiju Oba, Mariko Sagara, Sayuri Oda, Miyu Fukushima, Kenta Hasumi, Hirofumi Ohashi

Saitama Children's Medical Center, Japan

P2-12-4 **Survey highlights in 2022: The Japanese Association of Certified Genetic Counselors Biennial Survey**

Nana Akiyama, Manami Matsukawa, Ayumi Abe, Momoko Kato, Mikiko Kaneko, Mari Kikuchi, Sawako Matsuzaki, Mami Morita, Saki Shimada, Eriko Takamine, Asuka Toshida, Mari Tsubata, Junko Yotsumoto

The Japanese Association of Certified Genetic Counselors Survey and Research Committee, Japan

P2-12-5 **Genetic counseling for a hereditary pancreatic cancer patient with PALB2 mutation**

Eri Sakai[1], Chie Ono[1], Haruka Bamba[1], Kenjiro Kimura[2], Masakazu Yashiro[3]

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2 Department of Hepato-Biliary-Pancreatic Surgery, Osaka Metropolitan University Graduate School of Medicine, Osaka, Japan
3 Department of Molecular Oncology and Therapeutics, Osaka Metropolitan University Graduate School of Medicine, Osaka, Japan

P2-12-6 **Role of genetic counselors in oncofertility in Japan: A Nationwide survey**

Yuko Tamaki[1], Yukiko Katagiri[1], Kumiko Oseto[2], Yukiko Yoshimoto[3], Sanae Numata[4], Kuniaki Ota[5], Akemi Kataoka[6], Seido Takae[7], Nao Suzuki[7]

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3 Department of Breast Surgery, Kitano Hospital, Japan
4 Department of Cancer Center / Genetic Division, Kurume University Hospital, Japan
5 Department of Obstetrics and Gynecology, Tokyo Rosai Hospital, Japan
6 Department of Surgical Oncology, Breast Oncology Center, Cancer Institute Hospital of the Japanese Foundation for Cancer Research, Japan
7 Department of Obstetrics and Gynecology, St. Marianna University School of Medicine, Japan

P2-12-7 **A FAP patient and his family dynamics and psychological care : An approach from the Family Image Technique (FIT)**

Sayuri Hiraoka[1,2,3], Akiko Kameyama[3], Makiko Dazai[4,5], Kokichi Sugano[2], Takeshi Yamada[1], Kenji Kameguchi[6]

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2 Department of Genetic Medicine, Sasaki Foundation, Kyoundo Hospital, Japan
3 Department of Clinical Psychology, International University of Health and Welfare, Japan
4 Specified Nonprofit Corporation HBOC Patients Association Clavis Arcus, Japan
5 Genetic Alliance JP, Japan
6 The University of Tokyo, Japan

P2-12-8 **Genetic counseling on at-risk individuals of Spinocerebellar ataxia: A single institution-based study**

Akiko Sakyu[1], Katsuya Nakamura[1,2], Emiko Kise[1,3], Tomomi Kojima[1], Tomoki Kosho[1,4,5]

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3 Department of Nursing, Shinshu University Hospital, Japan
4 Department of Medical Genetics, Shinshu University School of Medicine, Japan
5 Division of Clinical Sequencing, Shinshu University School of Medicine, Japan

P2-12-9 **The issues of prenatal genetic counseling for foreigners in Japan**

Miyako Mizukami[1,2]

1 Sapporo Maternity Women's Hospital, Japan
2 Department of Medical Genetics and Genomics, Sapporo Medical University School of Medicine, Japan

P2-12-10 **Genetic counseling for a patient with Mitochondrial myopathy and ataxia**

Kazumi Kawato, Yuiko Hasegawa, Nobuhiko Okamoto

Osaka Women's and Children's Hospital, Izumi, Osaka, Japan

Poster Session 3-01 Complex Diseases and Genomic Risk Assessment 3

Date ：Saturday, October 14, 2023 13:00 ～ 13:30 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P3-01-1 **Three cases of Cowden's syndrome/ PTEN Hamartoma Tumor Syndrome: Three different clinical courses to reach the diagnosis**

Ryuta Takase[1], Kaori Fukui[1], Naoya Tsumura[1], Ken Kato[1], Munetsugu Hara[1], Tatsuki Mizuochi[1], Tomoya Sudo[1], Yoriko Watanabe[1,2]

1 Kurume University School of Medicine, Japan
2 Research Institute of Medical Mass Spectrometry, Kurume University School of Medicine, Japan

P3-01-2 **IL-1 beta gene polymorphisms and the susceptibility of hepatitis C infection in Malay male drug abusers**

Ruzilawati Abu Bakar[1], Jalviana Lansayan[1], Nur Salwani Bakar[1], Ismail Che Noh[1,2], Abdah Karimah Che Md Nor[1], Imran Ahmad[1]

1 Universiti Sains Malaysia, Malaysia
2 Universiti Malaysia Sabah, Malaysia

P3-01-3 **Utility of genetic analysis for dilated phase of hypertrophic cardiomyopathy, a high-risk group**

Natsuko Inagaki[1], Takeharu Hayashi[2], Shinji Suzuki[1], Yasuyoshi Takei[1], Akinori Kimura[3]

1 Tokyo Medical University, Japan
2 Tokai University School of Medicine, Japan
3 Medical Research Institute Tokyo Medical and Dental University, Japan

P3-01-4 **A balanced de novo t(2;3)(q31;p13) disrupting ATXN7 in a spinocerebellar ataxia young man**

Kai Yan, Fan Jin, Minyue Dong

Department of Reproductive and Genetics, Women's Hospital, School of Medicine, Zhejiang University, China

P3-01-5 Polygenic risk scores, lifestyle factors, and risk of carotid atherosclerosis

Liao Li-Na

Department of Public Health, China Medical University, Taiwan

P3-01-6 Long-term impacts of childhood maltreatment: A mendelian randomization study

Nicole Ng[1], Jane Zhao[2], Christopher Mak[1], Brian H.Y. Chung[1]

1 Department of Paediatrics and Adolescent Medicine, School of Clinical Medicine, LKS Faculty of Medicine, The University of Hong Kong, Hong Kong
2 School of Public Health, LKS Faculty of Medicine, The University of Hong Kong, Hong Kong

P3-01-7 Gaining insights into genetic variation underlying non-immune hydrops fetalis through trio exome sequencing

Patrick Yap[1,2], Chew Jasmine[1,3], Heller Raoul[2], Jenny Eaton[2], Monique Stein de-Laat[4], Candice Feben[2], Mark Greenslade[3], Peter Tsai[1], Cristin Print[1], Polona Le Quesne-Stabej[1]

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2 Genetic Health Service New Zealand (Northern Hub), New Zealand
3 Dignostic Genetics, LabPlus, New Zealand
4 Maternal-Fetal Medicine Services, Auckland District Health Board, New Zealand

P3-01-8 Multi-ancestry GWAS of Fuchs endothelial corneal dystrophy in US veterans highlights roles of laminins and collagen

Michael Francis[1,2], Bryan Gorman[1,2], Cari Nealon[3], Christopher Halladay[4], Nalvi Duro[1,2], Kyriacos Markianos[1], Giulio Genovese[5,6,7], Pirro Hysi[8,9,10], United States Veterans Affairs Million Veteran Program, Paul Greenberg[11,12], Saiju Pyarajan[1], Jonathan Lass[13], Neal Peachey[14,15,16], Sudha Iyengar[14,17,18]

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16 Department of Ophthalmology, Cleveland Clinic Lerner College of Medicine of Case Western Reserve University, Cleveland, OH, USA
17 Cleveland Institute for Computational Biology, Case Western Reserve University, Cleveland, OH, USA
18 Department of Population and Quantitative Health Sciences, Case Western Reserve University School of Medicine, Cleveland, OH, USA

P3-01-9 Case report: Spinocerebellar ataxia type 8 and 31

Risa Goto[1], Ayako Miyazaki[1,2], Chiho Okada[1], Chinatsu Kinjo[1], Mina Kashima[1], Mikako Miyata[1], Mako Ueda[1,3], Hideaki Sawai[1,3]

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3 Department of Obstetrics and Gynecology, Hyogo Medical University Hospital, Japan

P3-01-10 ***RNF213*** **p.Arg4810Lys heterozygote is associated with early onset and bilateral cerebrovascular events in Moyamoya disase**

Satoru Miyawaki[1], Daiichiro Ishigami[1], Hideaki Imai[2], Masahiro Shimizu[3], Hiroki Hongo[1], Shogo Dofuku[1], Kenta Ohara[1], Yu Teranishi[1], Daisuke Shimada[4], Satoshi Koizumi[1], Hideaki Ono[5], Yudai Hirano[1], Masafumi Segawa[1], Hirofumi Nakatomi[4], Nobuhito Saito[1]

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3 Department of Neurosurgery, Kanto Neurosurgery Hospital, Japan
4 Department of Neurosurgery, Kyorin University, Japan
5 Department of Neurosurgery, Fuji Brain Institute and Hospital, Japan

P3-01-11 **The current status and Issues of genetic counseling and testing with Inherited cardiac arrhythmias at our hospital**

Mariko Komine[1], Yusuke Ebana[1,2], Tetsuro Sasano[3], Hiroko Kobata[2], Sayako Takahashi[1], Masayuki Yoshida[1,2]

1 Tokyo Medical and Dental University Hospital the Department of Genetic Medicine, Japan
2 Tokyo Medical and Dental Uninersity the Life Science and Bioethics, Japan
3 Tokyo Medical and Dental Uninersity Hospital the Department of Cardiovascular Medicine, Japan

P3-01-12 **Genomic foundation of sensorineural hearing loss**

June-Young Koh[1], Sang-Yeon Lee[2,3,4], Seungbok Lee[2,5], Seongyeol Park[1], Sung Ho Jung[3], So Min Lee[3], Won Hoon Choi[3], Yejin Yun[3], Ju Hyuen Cha[3], Hongseok Yun[2], Myung-Whan Shu[3], Moo Kyun Park[3], Jae-Jin Song[6], Byung Yoon Choi[6], Jun Ho Lee[3], Seung Ha Oh[3]

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7 Graduate School of Medical Science and Engineering, Korea Advanced Institute of Science and Technology, Daejeon, South Korea

P3-01-13 **Preoperative genetic testing impacts decision-making for risk-reducing mastectomy in breast cancer patients**

Chanchira Sriraksasin, Prasit Phowthongkum

Excellence Center for Genomics and Precision Medicine, King Chulalongkorn Memorial Hospital, Thai Red Cross Society, Bangkok, Thailand

P3-01-14 **The first case of segmental UPD of chromosome 7 not involving *MEST* in a patient with Silver-Russell syndrome features**

Da Hye Lee[1], Jung Min Ko[2], Jee-Soo Lee[3,4], Moon-Woo Seong[3,4], Jae Hyeon Park[3]

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4 Department of Laboratory Medicine, Seoul National University College of Medicine, Republic of Korea

Poster Session 3-02 Inherited Metabolic Diseases and Newborn Screening 3

Date ：Saturday, October 14, 2023 13:00 ～ 13:30 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P3-02-1 **Neonatal Screening for SCID: The Taiwan Experience**

Yu-Han Chen[1], Li-Wen Hsu[1], Shu-Chuan Chiang[1], Yin-Hsiu Chien Chien[1,2]

1 Department of Medical Genetics, National Taiwan University Hospital, Taipei, Taiwan
2 Department of Pediatrics, National Taiwan University Hospital, Taipei, Taiwan

P3-02-2 **A Content analysis of Facebook Groups on congenital adrenal hyperplasia**

Ma-Am Joy Tumulak[1], Peter James Abad[1], Rizza Kaye Cases[2], Bubbles Beverly Asor[2], Cheryll Magbanua-Calalo[1], Ebner Bon Maceda[1], Kia Anarna[1], Renchillina Joy Supan[2], Patricia Carla Asuncion[1], Theodore Delfin Vesagas[1]

1 University of the Philippines, Manila, Philippines
2 University of the Philippines Diliman, Philippines

P3-02-3 **The impact of storage conditions on the concentrations of screening markers in newborn dried blood spots**

Li-Chu Chen[1], Chen-Chen Liu[1], Chin-Ting Liu[1], Yin-Hsiu Chien[1,2]

1 Department of Medical Genetics, National Taiwan University Hospital, Taipei, Taiwan
2 Department of Pediatrics, National Taiwan University Hospital, Taipei, Taiwan

P3-02-4 **Neonates in non-mosaic Klinefelter syndrome: A systematic review and meta-analysis**

Reina Kuraki[1], Keiko Goto[2], Yuri Kitamura[2], Ippei Hiramatsu[3], Hiromasa Goto[4], Eri Shimizu[2], Anna Sato[2], Fumi Murakami[2], Motoko Watanabe[2], Miho Isaka[2], Masami Arai[2]

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3 Department of Urology, Juntendo University, Graduate School of Medicine, Japan
4 Department of Metabolism & Endocrinology Medicine, Juntendo University, Graduate School of Medicine, Japan

P3-02-5 **An intronic variant of *SLC6A8* identified functionally critical residues of the creatine transporter**

Toshiki Tsunogai[1], Eri Imagawa[1], Thomas P Naidich[2], Nicola Longo[3], Kimihiko Oishi[1]

1 Department of Pediatrics, The Jikei University School of Medicine, Japan
2 Department of Radiology, Icahn School of Medicine at Mount Sinai, USA
3 Division of Medical Genetics, Department of Pediatrics, University of Utah, USA

P3-02-6 **Molecular relevance of citrullinemia type II and liver cancer**

Phuong Thanh Nguyen[1,2], Ituro Inoue[1]

1 Laboratory of Human Genetics, National Institute of Genetic, Japan
2 Department of Genetics, The Graduate University for Advanced Studies (SOKENDAI), Japan

P3-02-7 **Attenuated MPS II detected by newborn screening whose treatment was discontinued based on family genetic test**

Yoko Nakajima, Katsuyuki Yokoi, Yuta Sudou, Yasuaki Yasuda, Tetsushi Yoshikawa, Tetsuya Ito

Fujita Health University School of Medicine, Japan

Poster Session 3-03 Prenatal Genetics 3

Date ：Saturday, October 14, 2023 13:00 ～ 13:30 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P3-03-1 **A case report of incidental finding of confirmed placental mosaicism (CPM) for prenatal diagnosis of Hunter syndrome**

Hiroaki Nakamura, Yoshiko Matsuda, Michiko Watanabe, Sakika Yanai, Hiroko Katayama, Sachiyo Nishimoto, Rintaro Abe, Yuko Araki, Naoki Yamada, Kazuki Tanimura, Hiroko Akaishi, Kenji Yoshimura, Jun Mori, Shinji Higuchi

Department of Genetic Medicine, Osaka City General Hospital, Japan

P3-03-2

A case of trisomy 13 false positive of NIPT in a pregnant woman who had a history of trisomy 13

Yuuka Abe[1], Shin Onota[4], Akiyo Onota[4], Shiya Kaori[4], Kanami Saito[3], Satoshi Kosugi[3], Satoshi Takakura[3], Kohei Sugimoto[1,2], Yoshinobu Hamada[1,2]

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3 Obstetrics and Gynecology, Dokkyo Medical University Saitama Medical Center, Japan
4 Onota Women's Clinic, Saitama Omiya, Japan

P3-03-3

Development of a safer and simpler embryo biopsy for PGT-A

Atsushi Tanaka[1], Youichi Takemoto[1], Motoi Nagayoshi[1], Yuya Makino[1,2], Daisuke Nakajima[1,3], Seiji Watanabe[4]

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2 Juntendo University School of Medicine, Japan
3 Occupational and Environmental Health University School of Medicine, Japan
4 Hirosaki University Graduate School of Medicine, Japan

P3-03-4

Factors associated with high mental stress of pregnant women undergoing NIPT and changes after genetic counseling

Eri Takeda[1], Nobuhiro Suzumori[1], Kyoko Kumagai[1], Iku Taguchi[1,2], Kiwa Yamaoka[1,3], Rin Sato[1,3], Ayako Tanabe[1,4], Shinobu Goto[1], Ayano Otani[1], Mayumi Sugiura-Ogasawara[1]

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2 National Hospital Organization, Nagoya Medical Center, Japan
3 Nagoya Ekisaikai Hospital, Japan
4 Toyota Memorial Hospital, Japan

P3-03-5

Validation and clinicals case report in non-invasive prenatal testing for all chromosomes

Hyuk-Jung Kwon, Sam Martin

EDGC, Korea

P3-03-6

Withdrawn

P3-03-7

Comparison of G-banding karyotyping and NGS for chromosome testing of products of conception after spontaneous abortion

Yoshie Nagatakidani[1], Michiko Anmae[2], Yoshiko Asai[1], Tomoko Inoue[1], Yoshiharu Morimoto[1]

1 HORAC GRAND FRONT OSAKA Clinic, Japan
2 IVF Namba clinic, Japan

P3-03-8

Pregnant women's recent preferences for prenatal genetic testing: A single-center study in Japan

Rina Akaishi, Fuyuki Hasegawa, Yuuki Kakinuma, Chihiro Nishino, Taishuke Morita, Shin Mouri, Saho Fujino, Yuuya Fujibe, Saori Unno, Jin Muromoto, Tomo Suzuki, Rika Sugibayashi, Katsusuke Ozawa, Aiko Sasaki, Seiji Wada, Haruhiko Sago

National Center for Child Health and Development, Japan

P3-03-9

Risk figures of reciprocal translocation carriers with imbalanced blastocysts after structural rearrangement testing

Tetsuaki Hara[1,2], Eimi Rai[1], Takashi Kodama[1], Keiko Hara[1], Takahiro Miura[1], Yoko Watanabe[1], Kanako Nishimura[1], Aya Ueda[1]

1 Hiroshima Prefectural Hospital, Japan
2 Hiroshima Chuo-Dori Katsuki Ladies Clinic, Japan

P3-03-10 **Efficiency of PGT-SR in chromosomally balanced translocation couples**

Aya Yamazaki[1], Nami Kawasaki[2], Tomoko Kuroda[2], Keiichi Kato[2], Toshiyuki Takeshita[3], Akira Kuwahara[4], Takeshi Iwasa[4], Minoru Irahara[4], Toshiyuki Yamamoto[1]

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2 Kato Ladies Clinic, Japan
3 Nippon Medical School Obstetrics and Gynecology, Japan
4 Tokushima University Obstetrics and Gynecology, Japan

P3-03-11 **Two cases of trisomy13 positive in NIPT subsequently determined as CPM involving trisomy13**

Makiko Tominaga[1], Ayano Sakurai[1], Ayumi Okuyama[1], Mikiko Izumi[2], Kiyotake Ichizuka[1], Akiko Sakashita[1]

1 Showa University Northern Yokohama Hospital, Japan
2 Showa University Hospital, Japan

P3-03-12 **Do treatment outcomes in ART without PGT-SR differ whether the translocation carrier is male or female?**

Eri Nakahara, Koyu Furuhashi, Shoji Kokeguchi, Eri Okamoto, Masahide Shiotani

Hanabusa Women's Clinic, Japan

P3-03-13 **Novel therapeutic approach for patients with Loss-of-function ACE**

Hang-Jing Tan[1,2], Hong-Mei Xiao[1,2]

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Poster Session 3-04 Neurology 3

Date ：Saturday, October 14, 2023 13:00 ～ 13:30 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P3-04-1 **A case of chromosome 1p36 deletion syndrome diagnosed using Chromosomal Microarray Testing at Age 50**

Misako Kaido[1,2,3], Takuhei Yokoyama[1,4], Yuka Yotsumoto[5,6], Tomoko Hashimoto-Tamaoki[5,6]

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2 Department of Clinical Genetics, Sakai City Medical Center, Japan
3 Department of Neurology, Sakai City Medical Center, Japan
4 Department of Obstetrics and Gynecology, Sakai City Medical Center, Japan
5 Takatsuki General Hospital, Japan
6 Department of Genetic Medicine, Takatsuki General Hospital, Japan

P3-04-2 **Identification of a novel RAPSN variant and electrodiagnostic confirmation of congenital myasthenic syndrome**

Chinmayee Bhimarao Nagaraj[1], Hani Kushlaf[2]

1 Cincinnati Children's Hospital, USA
2 University of Cincinnati, USA

P3-04-3 **RNA sequencing confirms the pathogenicity of a novel FHL1 deletion**

Chinmayee Bhimarao Nagaraj[1], Cuixia Tian[1,2], Hani Kushlaf[1,2]

1 Cincinnati Children's Hospital, USA
2 University of Cincinnati, USA

P3-04-4 **A retrospective chart review evaluating genetic testing approaches for patients with neuromuscular disorders**

Chinmayee Bhimarao Nagaraj[1], Amanda Rosenberg[1,2], Hua He[1], Elizabeth Ulm[1], Kathleen Collins[1], Cuixia Tian[1,2]

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2 University of Cincinnati, USA

P3-04-5 **Regular social events for interaction among patients/families with Fragile X syndrome and Fragile X-Related disorders**

Tetsuya Okazaki[1,2], Tomoki Nonaka[2], Saki Shinzato[3], Chisako Aoki[4], Aya Yamamoto[5], Kaori Adachi[2,6], Eiji Nanba[2,6,7]

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4 Department of Medical Genetics, Kakogawa Central City Hospital, Japan
5 Graduate School of Clinical Psychology, Sapporo Gakuin University, Japan
6 Organization for Research Initiative and Promotion, Tottori University, Japan
7 Otani Hospital, Japan

P3-04-6 **Knowledge, awareness and perception on genetic testing among parents of neuromuscular disorders patients in Malaysia**

Farheen Hakim Zada[1], Ahmad Hazim Syakir Ahmad Azahari[1], Sau Wei Wong[1], Adli Ali[1,2], Noor Akmal Shareela Ismail[2,3]

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2 Research Centre, Hospital Tunku Ampuan Besar Tuanku Aishah Rohani, UKM Specialist Children's Hospital, Universiti Kebangsaan Malaysia, Jalan Yaacob Latif, Cheras, Kuala Lumpur, Malaysia
3 Department of Biochemistry, Faculty of Medicine, Universiti Kebangsaan Malaysia, Jalan Yaacob Latif, Cheras, Kuala Lumpur, Malaysia

P3-04-7 **Recurrent rhabdomyolysis and persistent elevated creatine kinase in a medical cadet during medical training: A case report**

Sethapong Lertsakulbunlue[1], Boonsub Sakboonyarat[2], Boonchai Boonyawat[3], Tim Phetthong[3]

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3 Division of Medical Genetics, Department of Pediatrics, Phramongkutklao Hospital and College of Medicine, Bangkok, Thailand

P3-04-8 **Patient registry system for new treatment choice and newborn screening follow-up in spinal muscular atrophy**

Tamaki Kato, Yumi Ikeda, Mamoru Yokomura, Mari Urano, Akiko Ueda, Kayoko Saito

Tokyo Women's Medical University, Japan

Poster Session 3-05 Technological Advances, Wet and Dry 3

Date ：Saturday, October 14, 2023 13:00 ～ 13:30 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P3-05-1 **Comprehensive gene expression analysis during repression of the transcriptional regulator Zfat in mouse fetal liver**

Keiko Doi, Midori Koyanagi, Yoko Tanaka

Faculty of Medicine, Fukuoka University, Japan

P3-05-2 **Withdrawn**

P3-05-3 **Development of a clinically applicable one-step PCR-based CYP21A2 analysis using long read sequences**

Eriko Adachi[1], Ryuichi Nakagawa[1], Atsumi Tsuji-Hosokawa[1], Maki Gau[1], Shizuka Kirino[1], Analia Yogi[1], Hisae Nakatani[1], Tomomi Yamaguchi[2], Masanori Murakami[3], Toshihiro Tajima[4], Tomonobu Hasegawa[5], Tetsuya Yamada[3], Tomohiro Morio[1], Osamu Ohara[6], Kenichi Kashimada[1]

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3 Department of Molecular Endocrinology and Metabolism, Tokyo Medical and Dental University, Japan
4 Department of Pediatrics, Jichi Medical University, Japan
5 Department of Pediatrics, Keio University, School of Medicine, Japan
6 Department of Applied Genomics, Kazusa DNA Research Institute, Japan

P3-05-4 **Establishment of a novel genotyping method for the double homeobox protein 4 (DUX4) gene**

Zhuang Zhaohui[1], Mahoko Ueda[1], Kensuke Yamaguchi[2], Nao Nishida[1], Satomi Mitsuhashi[1], Yuta Kochi[1]

1 Department of Genomic Function and Diversity, Medical Research Institute, Tokyo Medical and Dental University, Japan
2 Biomedical Engineering Research Innovation Center, Institute of Biomaterials and Bioengineering, Tokyo Medical and Dental University, Japan

P3-05-5 **Structural analysis and prediction of pathogenicity in MLH1 variants using homology modeling**

Mirei Tsuchiyama, Kazuo Tamura, Norihito Kawashita

Kindai University, Japan

P3-05-6 **Simulation of P-values for association tests and consideration for GWAS power imbalance**

Takashi Kido[1], Naoyuki Kamatani[2]

1 Teikyo University, Japan
2 StaGen Co., Ltd., Japan

P3-05-7 **Small Cajal body-specific RNA: Regulatory function in Alzheimer's disease**

Sumin Yang, Sung-Hyun Kim, Eunjeong Yang, Jae-Yeol Joo

Hanyang University, Korea

P3-05-8 **Full-length total RNA sequencing reveals the fraction of cell free RNA in spent embryo culture media**

Takeshi Sugimoto[1,2], Tasuku Mariya[2,3], Hiroki Kurahashi[2]

1 Kobe Motomachi Yume Clinic, Japan
2 Division of Molecular Genetics, Institute for Comprehensive Medical Science, Fujita Health University, Japan
3 Department of Obstetrics and Gynecology, School of Medicine, Sapporo Medical University, Japan

P3-05-9 **Construction of quality-controlled genetic testing system for cardiovascular disease**

Kaori Kugo, Yohei Miyashita, Rieko Osawa, Yoshiyuki Sumita, Koji Takata, Yoshihiro Asano

National Cerebral and Cardiovascular Center Hospital, Japan

P3-05-10 **Unraveling the diversity of lncRNAs in cancer: Spatial and Single-cell profiling across tumor types**

Prakrithi Pavithra[1,2,3], Tuan Vo[2], Ishaan Gupta[3], Quan Nguyen[2]

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2 University of Queensland, Institute of Molecular Biosciences, St. Lucia, QLD, Australia
3 Indian Institute of Technology Delhi, Department of Biochemical Engineering and Biotechnology, Hauz Khas, New Delhi, India

P3-05-11 **Chromosome-wide assessment of skewed X-inactivation by parental phasing and methylome analysis using Nanopore sequencing**

Masayuki Sato[1], Toshiki Takenouchi[2], Hisato Suzuki[1,3], Mamiko Yamada[1], Kenjiro Kosaki[1], Fuyuki Miya[1]

1 Center for Medical Genetics, Keio University School of Medicine, Tokyo, Japan
2 Department of Pediatrics, Keio University School of Medicine, Tokyo, Japan
3 Department of Clinical Medicine, Institute of Medicine, University of Tsukuba, Tokyo, Japan

P3-05-12 **Therapeutic potential of chemically-modified microRNA143 lipoplex for refractory cancer**

Kohei Taniguchi[1], Jun Arima[1], Hiromitsu Tsuchihashi[1], Tomohito Tanaka[1], Sang-Woong Lee[1], Yukihiro Akao[2]

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Poster Session 3-06　Cancer Genomics, Germline 3

Date　：Saturday, October 14, 2023　13:00 ～ 13:30　Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P3-06-1 Risk-reducing salpingo-oophorectomy in BRCA variant carriers: An experience at the regional university hospital

Kentaro Kai[1], Yoko Aoyagi[1], Masakazu Nishida[1], Nobue Tsukatani[2], Kenji Ihara[2], Eiji Kobayashi[1]

1　Department of Obstetrics and Gynecology, Oita University Faculty of Medicine, Japan
2　Division of Genetic Medicine, Oita University Hospital, Japan

P3-06-2 Frequency of secondary findings using CGP especially in childhood, adolescent and young adult-onset cancer

Yoshinaga Okugawa, Ryo Hanaki, Takhito Kitajima, Takumi Fujiwara, Junya Tsuboi, Emi Teramoto, Maki Nakamura, Makoto Ikejiri, Kanako Nishikawa, Ikuyo Mochiki, Ryotaro Hashizume, Hiroshi Imai, Kaname Nakatani, Yuji Toiyama

Mie University Hospital, Japan

P3-06-3 Regional collaboration in hereditary tumors

Megumi Matsumoto[1], Mami Takao[2], Aya Tanaka[2], Shoko Miura[2], Yuri Hasegawa[2], Kenichiro Shibata[1], Hideki Taniguchi[1], Kiyonori Miura[2]

1　Japasene Red Cross Nagasaki Genbaku Hospital, Japan
2　Nagasaki University Hospital, Japan

P3-06-4 Elucidation of genetic factors in pancreatic cancer by integration of molecular pathology and genomic epidemiology

Kodai Abe, Minoru Kitago, Yohei Masugi, Mamiko Yamada, Hisato Suzuki, Kenjiro Kosaki, Yuko Kitagawa

Keio University School of Medicine, Japan

P3-06-5 Genetic analysis in preventive medicine and following medical management

Hisato Suzuki[1], Ohsuke Migita[2], Miwa Arita[1], Ayumi Shikama[1], Hiroko Bando[1], Kazuhiro Takekoshi[1], Hideo Suzuki[1], Toshiaki Narasaka[1]

1　University of Tsukuba, Japan
2　St.Marianna University, Japan

P3-06-6 The system of regional collaboration for HBOC treatment at our hospital and its issues

Yukiko Yoshimoto, Ai Itagaki, Tsuyoshi Tachibana, Sachiko Takahara

Kitano Hospital, Japan

P3-06-7 Rethinking the optimal age to consider RRSO in HBOC

Yusaku Urakawa[1,2], Mashu Futagawa[1], Sayaka Ueno[1], Fumino Kato[1], Reimi Sogawa[1], Hideki Yamamoto[1], Akira Hirasawa[1]

1　Okayama University, Japan
2　Kobe City Medical Center General Hospital, Japan

P3-06-8 Two cases of BRCA1/2 double mutation in patients with breast cancer

Sachiko Miyazaki[1,2,3], Yumi Tanaka[3], Maiko Shiga[2], Kentaro Suda[3], Miyako Mizukami[3], Tasuku Mariya[3,4], Aki Ishikawa[1,2,3], Akihiro Sakurai[1,2,3]

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3　Division of Medical Genetics and Genomics of Sapporo Medical University Hospital, Japan
4　Division of Gynecology of Division of Sapporo Medical University, Japan

P3-06-9 Comparison of survival rates of ovarian cancer patients inherited from first- and non-first-degree relatives

Rachadapan Chaitosa

Gynecology Oncology, Thailand

P3-06-10 The family with high penetrance of glioblastoma in Lynch syndrome

Hiroko Fujita[1], Kazuo Tamura[1,2,3], Akira Hirasawa[1,4], Hideki Yamamoto[1,4], Yusaku Urakawa[4], Mashu Futagawa[4], Shohei Kohno[1], Ayako Ito[1], Haruka Tada[1], Tami Nagatani[1], Kyohei Kai[1]

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2 Kindai University, Japan
3 Sakurabashi-Watanabe Hospital, Japan
4 Okayama University, Japan

P3-06-11 A case of Lynch syndrome with strong familial cancer history that was difficult to diagnose due to hematological disease

Tomomi Oka[1,3], Takeshi Nakajima[1,4], Masako Torishima[1,5], Akiko Yoshida[1,5], Hiromi Murakami[1], Sayaka Honda[1], Akira Inaba[1], Hidenori Kawasaki[1,5], Masanobu Ogawa[1,6], Takahito Wada[1,5], Yasuhito Nanya[2,7], Seishi Ogawa[2,8,9], Akifumi Takaori-Kondo[3], Shinji Kosugi[1,5]

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3 Department of Hematology, Kyoto University Hospital, Japan
4 School of Public Health, Medical Ethics and Medical Genetics, Graduate of School of Medicine, Kyoto University, Japan
5 Department of Genomic Medicine, Graduate School of Medicine, Kyoto University, Japan
6 Ethics Support Unit, Kyoto University Hospital, Japan
7 Division of Hematopoietic Disease Control, The Institute of Medical Science, The University of Tokyo, Japan
8 Institute for the Advanced Study of Human Biology (WPI ASHBi), Kyoto University, Japan
9 Center for Hematology and Regenerative Medicine, Karolinska Institutet, Stockholm, Sweden

P3-06-12 Hereditary breast cancer in ramathibodi hospital: A retrospective study

Kanin Sriudomporn[1], Thanyachai Sura[1], Wiriya Pipatsakulroj[2], Atchara Tunteeratum[1]

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P3-06-13 Current situation of risk-reduction surgeries and surveillance for hereditary tumors

Akimitsu Yamada[1], Mahato Sasamoto[1], Masanori Oshi[1], Mii Takatsuka[2], Tomohiro Sakaguchi[2], Hiroko Kuriki[2], Natsuko Kamiya[3], Yuki Ogawara[3], Kouichi Nagai[3], Yumi Ishidera[3], Haruka Hamanoue[2], Itaru Endo[1]

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P3-06-14 Genetic counselling as part of a multi-disciplinary team management for young breast cancer patient with NF1

Claudia Richard[1], Sook-Yee Yoon[1], Kiley Wei-Jen Loh[2], Gaik-Siew Ch'ng[3]

1 Key Genetics, Malaysia
2 Penang Adventist Hospital, Malaysia
3 Hospital Pulau Pinang, Malaysia

P3-06-15 Current status of BRCA testing for breast cancer cases in our hospital

Tomoi Sato

Niigata Prefectural Central Hospital, Japan

Poster Session 3-07 Cancer Genomics, Somatic 2

Date ：Saturday, October 14, 2023 13:00 ～ 13:30 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P3-07-1 Identification of URST4 as a prognostic biomarker and therapeutic target for breast cancer

Nguyen Hoa[1], Atsushi Takano[1,2], Bayarbat Tsevegjav[1], Regina Mbugua[1], Yohei Miyagi[3], Yataro Daigo[1,2]

1 Shiga University of Medical Science, Japan
2 The University of Tokyo, Japan
3 Kanagawa Cancer Center, Japan

P3-07-2

A case of overlapping lung and cervical cancer with SMAD4 gene mutation identified by gene panel test

Masakazu Nishida[1], Kentaro Kai[1], Yoko Aoyagi[1], Yasushi Kawano[1], Nobue Tsukatani[2], Kenji Ihara[2], Eiji Kobayashi[1]

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2 Division of Genetic Medicine, Oita University Hospital, Japan

P3-07-3

A case report from the blood-based comprehensive genomic profiling (CGP) test for solid tumors

Tomoko Sahara[1], Sayuri Hiraoka[1], Ikuno Kawabata[1,3], Masafumi Toyoshima[1,3], Hidehiko Miyake[1,3,4], Hiroyuki Takei[4], Hiroshi Yoshida[2], Takeshi Yamada[1,2]

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3 Department of Obstetrics and Gynecology, Nippon Medical School, Japan
4 Department of Breast Surgery and Oncology, Nippon Medical School, Japan
5 Departmen of Genetic Counseling, Graduate School of Humanities and Sciences, Ochanomizu University, Japan

P3-07-4

A novel biochemical valuable method for interpretation of variants of unknown significance

Yuji Kubo[1], Misaki Iwasaka[1], Yoichi Makino[1], Hiroshi Ueno[2], Kazuhito Tabata[2], Hiroyuki Noji[2]

1 Technical Research Institute, TOPPAN INC., Japan
2 Department of Applied Chemistry, School of Engineering, The University of Tokyo, Japan

P3-07-5

Usefulness of cancer genome medicine using cancer gene panel for advanced uterine leiomyosarcoma

Takuma Hayashi, Ikuo Konishi

Dept. of Cancer Medicine, National Hospital Organization Kyoto Medical Center, Japan

P3-07-6

Monitoring residual disease using cell free DNA in pediatric malignancies

Mari Kagajo[1], Kyoko Moritani[1], Mayumi Iwamoto[1], Machiko Miyamoto[1], Minenori Ishimae[1], Hisamichi Tauchi[1], Yasushi Ishida[2], Mariko Eguchi[1]

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2 Department of Medical Technology. Faculty of Health Sciences, Ehime Prefectural University of Health Sciences, Japan

P3-07-7

Translocation(8;21) AML–cytomorphological, immunophenotyping and cytogenetic profile of three paediatric cases

Farah Azima Abdul Muttlib[1], Julia Mohd Idris[2], Alia Suzana Asri[1], Woon Lee Yong[1]

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Poster Session 3-08 Pediatric Genetics 3

Date ：Saturday, October 14, 2023 13:00 ～ 13:30 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P3-08-1

A case of autosomal dominant spastic paraplegia-9A (SPG9A) with a novel pathogenic variant in ADLH18A1

Masaharu Moroto[1], Tomoya Yodoi[2], Daisuke Uda[2], Yoshihiro Nitta[2], Takenori Tozawa[3], Tomohiro Chiyonobu[3,4], Masafumi Morimoto[3,5], Naoko Yano[6], Takeshi Yoshida[6]

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4 Department of Molecular Diagnostics and Therapeutics, Graduate School of Medical Science, Kyoto Prefectural University of Medicine, Japan
5 Department of Medical Science, School of Nursing, Kyoto Prefectrural University of Medicine, Japan
6 Department of Pediatrics, Kyoto University Graduate School of Medicine, Japan

P3-08-2

Post-mortem whole exome analysis in sudden unexpected death in infancy and children

Rika Kosaki[1], Yoshiko Uchida[4], Kumiko Yanagi[2], Satoko Tsushima[3], Satoko Uematsu[4], Tadashi Kaname[2]

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2 Department of Genome Medicine, National Research Institute for Child Health and Development, Japan
3 Department of Nursing, National Center for Child Health and Development, Japan
4 Division of Pediatric Emergency and Transport Services, National Center for Child Health and Development, Japan

P3-08-3

A phenotypical variation of a familial Noonan syndrome with RIT1 mutation diagnosed by next generation sequencing

Kazuyoshi Saito[1], Arisa Kojima[1], Takanori Suzuki[1], Hidetoshi Uchida[1], Tadayoshi Hata[1], Tetsushi Yoshikawa[1], Satoru Kawai[2]

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P3-08-4

Broad clinical spectrum and various diagnostic opportunities in patients with Coffin-Siris syndrome

Kohei Haraguchi[1], Midori Motokawa[1], Tatsuharu Sato[1], Mami Takao[2], Hiroyuki Mishima[3], Koh-ichiro Yoshiura[3], Tatsuro Kondoh[4], Hiroyuki Moriuchi[1], Sumito Dateki[1]

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4 Division of Developmental Disabilities, Misakaenosono Mutsumi Developmental, Medical and Welfare Center, Isahaya, Japan

P3-08-5

Clinical features of individuals with Rauch-Steindl syndrome due to NSD2 pathogenic variant

Eriko Nishi[1], Kumiko Yanagi[2], Nobuhiko Okamoto[1], Tadashi Kaname[1,2]

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2 National Research Institute for Child Health and Development, Japan

P3-08-6

A case of NSD2 deletion requiring differential diagnosis from Silver-Russell syndrome

Natsuki Nakamura[1], Yuki Hashimoto[2], Tomoko Uehara[1], Mie Inaba[1], Seiji Mizuno[1]

1 Aichi Developmental Disability Center Central Hospital, Japan
2 Ogaki Municipal Hospital, Japan

P3-08-7

A novel FBN2 mutation in a patient with suspected in congenital contractual arachnodactyly (CCA)

Mina Nakama[1,2], Yuki Miwa[2], Sayaka Manabe[1], Hidenori Ohnishi[2]

1 Kindai University, Japan
2 Gifu University, Japan

P3-08-8

A neonatal case of triple X syndrome with hydrops fetalis due to congenital bilateral chylothorax

Jun Nirei[1,2], Masako Hayashi[1,2], Tomoyuki Tani[1,2], Masato Tanaka[1,2], Keisuke Shoji[1,2], Akira Kobayashi[1,2], Akihiko Saitoh[1]

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2 General Center for Perinatal, Maternal, and Neonatal Medicine, Niigata University Medical and Dental Hospital, Japan

P3-08-9

An infant case with chromosome 1p36 deletion syndrome accompanied by 7p22.1 microduplication

Yuka Hattori[1], Yuka Yotsumoto[1,2], Maki Hamamoto[1], Yoshinori Katayama[3], Kenji Nakamura[4], Tomoko Tamaoki[2]

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3 Department of Neonatology, Takatsuki General Hospital, Japan
4 Department of Neonatology, Japanese Red Cross Otsu Hospital, Japan

P3-08-10

Clinical report of Pierson syndrome with biallelic variants in LAMB2

Yumiko Nishimura[1], Keiko Matsuda[1], Yuiko Hasegawa[1], Eriko Nishi[1], Kazumi Kawato[1], Kayo Inoue[1], Kumiko Yanagi[2], Tadashi Kaname[2], Nobuhiko Okamoto[1]

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2 National Center for Child Health and Development, Tokyo, Japan

P3-08-11

Intractable amenorrhea in a case of anorexia nervosa and diagnosis of exclusion by molecular genetic analysis

Naoko Sato

The University of Tokyo Hospital, Japan

P3-08-12

Nanopore long-read sequencing analysis of a triple translocation t(9;17;20) in a patient with congenital anomalies and developmental delay

Hiroaki Murakami[1,2], Tatsuya Sakashita[1], Yumi Enomoto[2], Kenji Kurosawa[2], Atsushi Imamura[1], Hideo Kaneko[1]

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2 Kanagawa Children's Medical Center, Japan

P3-08-13

Type 2 congenital generalized lipodystrophy by NOTCH2 variant

Taichi Imaizumi[1], Rina Shimomura[2,3], Osamu Machida[2,3], Tomoe Yanagishita[2], Keiko Shimojima Yamamoto[4,5], Miho Nagata[6], Yasuki Ishihara[6,7], Yohei Miyashita[6,7], Yoshihiro Asano[6,7], Toshiyuki Yamamoto[3,5]

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6 Department of Cardiovascular Medicine, Osaka University Graduate School of Medicine, Japan
7 Department of Genomic Medicine, National Cerebral and Cardiovascular Center, Japan

P3-08-14

A female patient of Weiss-Kruszka syndrome with 6 MB interstitial deletions of 9q31.1q32 including a whole ZNF462 gene

Hironao Numabe[1,3,4,5], Tomoko Takamatsu[2,3], Noriko Miyake[6]

1 Tokyo Metropolitan Kita Medical Rehabilitation Center for the Handicapped, Japan
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3 Department of Pediatrics, Japan Community Health care Organization Tokyo Yamate Medical Center, Japan
4 Department of Pediatrics, Tokyo Medical and Dental University Hospital, Japan
5 Department of Pediatrics, National Rehabilitation Center for Children with Disabilities, Japan
6 Department of Human Genetics, Research Institute, National Center for Global Health and Medicine, Japan

P3-08-15 Epidermolysis bullosa with congenital absence of skin in Thai infants: A molecular genetics study

Maolee Bhuwapathanapun[1], Sasitorn Auevirivavit[2], Panini Chetprayoon[2], Amornrat Tangprasittipap[3], Rossukon Kaewkhaw[1,4], Duangrurdee Wattanasirichaigoon[5], Natini Jinawath[1,4,6]

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2 National Nanotechnology Center (NANOTEC), National Science and Technology Development Agency (NSTDA), Pathum Thani, Thailand
3 Office of Research, Academic Affairs and Innovations, Faculty of Medicine Ramathibodi Hospital, Mahidol University, Bangkok, Thailand
4 Chakri Naruebodindra Medical Institute, Faculty of Medicine Ramathibodi Hospital, Mahidol University, Bang Phli, Samut Prakarn, Thailand
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6 Integrative Computational BioScience (ICBS) Center, Mahidol University, Nakhon Pathom, Thailand

P3-08-16 Comparison of the diagnosis of 22q11.2 deletion and Williams syndrome by facial photos between Face2gene and clinicians

Nop Khongthon[1], Midi Theeraviwatwong[1], Khunton Wichajarn[2], Kitiwan Rojnueangnit[3]

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3 Department of Pediatrics, Faculty of Medicine, Thammasat University, Pathumthani, Thailand

P3-08-17 Secular decrease in mosaicism in cultured and uncultured blood cells of six patients with mosaic Down syndrome

Takako Takano[1,2], Tatsuo Masuyama[2]

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2 Department of Pediatrics, Tokyo Metropolitan Tobu Medical Center for Children with Developmental Disabilities, Japan

P3-08-18 A rare mosaic variant of GJA1 in a patient with neurodevelopmental disorder

Rina Shimomura[1,2], Tomoe Yanagishita[2], Kumiko Ishiguro[2], Minobu Shichiji[2], Takatoshi Sato[2], Keiko Shimojima Yamamoto[3,4], Keiko Ishigaki[2], Satoru Nagata[2], Miho Nagata[5], Yoshihiro Asano[5,6], Toshiyuki Yamamoto[1,4]

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P3-08-19 Homozygous KCTD3 nonsense variant due to UPD associated with syndromic developmental epileptic encephalopathy

Toshiyuki Yamamoto[1], Keiko Shimojima Yamamoto[1,3], Ayumi Yoshimura[2], Hitoshi Kanno[3]

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2 Seirei Mikatahara General Hospital, Japan
3 Department of Transfusion Medicine and Cell Processing, Tokyo Women's Medical University, Japan

P3-08-20 A de novo KMT2E gene mutation in a patient with developmental delay : a case report

Joo Hyun Park[1], Ah Yeon Lee[1], Myungshin Kim[2], Seung Bin Lee[3]

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3 Industry-Academic Cooperation Foundation, The Catholic University of Korea, Korea

P3-08-21 **Nonsense mutation of methyl-CpG binding domain protein 5 (MBD5) gene with developmental delay ; A two brothers cases**

Hye Jung Park[1], Hogeon Namgung[1], Myungshin Kim[2], Seung Bin Lee[3], Joo Hyun Park[1]

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P3-08-22 **Transient erythroblastopenia by GATA1 variant in female**

Masatoshi Takagi[1], Motoi Yamashita[1], Takahiro Tomoda[1], Takeshi Isoda[1], Makiko Egawa[2], Masayuki Yoshida[2], Tsutomu Toki[3], Ko Kudou[3], Kiminori Terui[3], Etsuro Ito[3], Tomohiro Morio[1]

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P3-08-23 **Shoulder dysfunction is an expanding phenotype of CHD7 disorder**

Tomoko Uehara, Natsuki Nakamura, Seiji Mizuno, Mie Inaba

Department of Clinical Genetics, Aichi Developmental Disability Center Central Hospital, Aichi, Japan

P3-08-24 **Early diagnosis of a case of Bardet–Biedl syndrome with severe obesity using whole-genome sequencing**

Yuri Kitamura[1,2], Nobutomo Saito[1,3], Naoya Saijyo[4], Atsuo Kikuchi[4,6], Shigeo Kure[4,6], Fumiki Katsuoka[5], Akihito Otsuki[5], Gen Tamiya[6], Jun Takayama[6], Akio Nakamura[1], Kotoko Matsui[1], Yu Hosono[1], Chiharu Miyayama[1], Keiko Goto[2], Masami Arai[2], Toshiaki Shimizu[1]

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5 Tohoku University Graduate School of Medicine and Tohoku University Tohoku Medical Megabank Organization, Japan
6 Department of Rare Disease Genomics, Tohoku University Graduate School of Medicine, Japan

P3-08-25 **A case of 11p13 duplication encompassing *PAX6* and *ELP4* with ocular and neurodevelopmental phenotypes and GH deficiency**

Naoki Hamajima, Sawako Tajiri, Naomi Nishikawa

Center for Genetic and Genomic Medicine, Nagoya City University West Medical Center, Japan

P3-08-26 **Difficulties in clinical diagnosis of Kabuki syndrome in newborns CAKUT as a possible diagnostic clue**

Daisuke Nakato[1], Toshiki Takenouchi[2], Kenjiro Kosaki[1]

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Poster Session 3-09 Asian Genetics

Date ：Saturday, October 14, 2023 13:00 ～ 13:30 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P3-09-1 **Withdrawn**

P3-09-2 **History of non-African human populations inferred from introgressed archaic variants**

Hanako Miwa, Jun Ohashi

The University of Tokyo, Japan

P3-09-3

Whole genome sequencing for Japanese patients with Leber's congenital amaurosis and early onset retinal dystrophy

Kaoruko Torii[1], Sachiko Nishina[2], Hazuki Morikawa[2], Kei Mizobuchi[3], Masakazu Takayama[1], Nobutaka Tachibana[1], Tadashi Yokoi[2], Sachiko Miyamoto[1], Maki Fukami[2], Hiroyuki Kondo[4], Noriyuki Azuma[2,5], Takaaki Hayashi[3], Hirotomo Saitsu[1], Yoshihiro Hotta[1]

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2 National Center for Child Health and Development, Japan
3 The Jikei University School of Medicine, Japan
4 University of Occupational and Environmental Health, Japan
5 Medical Research Institute, Tokyo Medical and Dental University, Japan

P3-09-4

A novel silent variant in the EYA4 gene causing hearing loss by affecting RNA splicing

Min Chen, Min-Yue Dong

Women's Hospital, School of Medicine, Zhejiang University, China

P3-09-5

A case of Protein S deficiency with a novel frameshift variant

Chihiro Okoshi[1], Asako Yosie[2], Toma Fukuda[1], Shun Yasuda[1], Hidekazu Nishigori[3], Takafumi Watanabe[1,2]

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3 Fukushima Medical Center for Children and Women, Fukushima Medical University, Japan

P3-09-6

Prevalence of familial hypercholesterolemia, phenylketonuria, Factor V Leiden mutation in Thai population

Rosalind Lalitkulanant[1], Paravee Own-eium[2], Thanyachai Sura[3], Prin Vathesatogkit[4], Piyamitr Sritara[4], Jakris Eu-ahsunthornwattana[5], Donniphat Dejsuphong[2]

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3 Division of Medical Genetics, Department of Medicine, Faculty of Medicine Ramathibodi Hospital, Mahidol University, Bangkok, Thailand
4 Division of Cardiology, Department of Medicine, Faculty of Medicine Ramathibodi Hospital, Mahidol University, Bangkok, Thailand
5 Department of Community Medicine, Faculty of Medicine Ramathibodi Hospital, Mahidol University, Bangkok, Thailand

P3-09-7

Genome-wide association study by proxy on WGS: Susceptibility loci in dementia via Tohoku Medical Megabank Organization

Makiko Taira[1,2], Nobuo Fuse[1,3], Andrew J. Saykin[4,5,6], Fuji Nagami[1,2], Kengo Kinoshita[1,3,7], Masayuki Yamamoto[1,3]

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3 Tohoku University Advanced Research Center for Innovations in Next-Generation, Japan
4 Indiana University Graduate Schoolof Medicine, USA
5 Center for Neuroimaging and Indiana Alzheimer's Disease Research Center, Indiana University, USA
6 The Genetics Core of the National Institute on Aging (NIA) Alzheimer's Disease Neuroimaging Initiative (ADNI), USA
7 Tohoku University Graduate School of Information Sciences, Japan

Poster Session 3-10　Rare Diseases 3

Date　：Saturday, October 14, 2023　13:00 ～ 13:30　Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P3-10-1 Identification of a novel deep intronic DDC variant in patients with aromatic l-amino acid decarboxylase deficiency

Eriko Koshimizu[1], Satoko Miyatake[1,2], Kazuharu Misawa[1], Yuri Uchiyama[1,3], Naomi Tsuchida[1,3], Kohei Hamanaka[1], Atsushi Fujita[1], Takeshi Mizuguchi[1], Mitsuhiro Kato[4], Naomichi Matsumoto[1]

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3 Department of Rare Disease Genomics, Yokohama City University Hospital, Yokohama, Japan
4 Department of Pediatrics, Showa University School of Medicine, Tokyo, Japan

P3-10-2 Genetic background and clinical features of congenital or early-onset deafblindness in Japan

Tatsuo Matsunaga[1], Kiyomitsu Nara[1], Hideki Mutai[1], Haruka Murakami[1], Satomi Inoue[1], Reiko Muramatsu[1], Kazuki Yamazawa[1], Shujiro Minami[1], Kaoru Fujinami[1], Masato Fujioka[2], Nobuko Yamamoto[3], Noriko Morimoto[3], Nana Tsuchihashi[4], Masatsugu Masuda[5], Yukiko Arimoto[6], Atsuko Nakano[6], Hirokazu Sakamoto[7], Toshiyuki Seto[7], Sayaka Katsunuma[8], Shinji Higuchi[9]

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3 National Center for Child Health and Development, Japan
4 Kyushu University School of Medicine, Japan
5 Kyorin University Faculty of Medicine, Japan
6 Chiba Children's Hospital, Japan
7 Osaka Metropolitan University Faculty of Medicine, Japan
8 Hyogo Prefectural Kobe Children's Hospital, Japan
9 Osaka City General Hospital, Japan

P3-10-3 Bardet-Biedl syndrome caused by a novel homozygous deletion in BBIP1 presenting with kidney failure in one sibling

Rini Rossanti[1,8], Naoya Morisada[2,3], Ahmedz Widiasta[1,8], Yunia Sribudiani[8,9], Purboyo Solek[4], Irawati Irfani[5], Dedi Rachmadi[1,8], Dany Hilmanto[1], Kandai Nozu[2], Kazumoto Iijima[6,7]

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3 Department of Clinical Genetics, Hyogo Prefectural Kobe Children's Hospital, Kobe, Japan
4 Department of Child Health, Pediatric Neurology Division, Dr. Hasan Sadikin General Hospital / Faculty of Medicine, Universitas Padjadjaran, Bandung, Indonesia
5 Department of Ophthalmology, Pediatric Ophthalmology & Strabismus Division, Cicendo Eye Hospital / Faculty of Medicine, Universitas Padjadjaran, Bandung, Indonesia
6 Hyogo Prefectural Kobe Children's Hospital, Kobe, Japan
7 Department of Advanced Pediatric Medicine, Kobe University Graduate School of Medicine, Kobe, Japan
8 Medical Genetic Research Center, Faculty of Medicine, Universitas Padjadjaran, Bandung, Indonesia
9 Department of Biomedical Sciences, Division of Biochemistry and Molecular Biology, Faculty of Medicine, Universitas Padjadjaran, Bandung, Indonesia

P3-10-4 A case of hypogonadism and mitochondrial disease with concomitant variants in FGFR1 and RRM2B

Rieko Kosugi[1], Tatsuo Ogawa[1], Hiroyuki Ariyasu[1], Tatsuhide Inoue[1], Tsutomu Ogata[2], Takeshi Usui[3]

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2 Hamamatsu University School of Medicine, Japan
3 Shizuoka Graduate University of Public Health, Japan

P3-10-5 A case of 19p13.3 microdeletion syndrome associated with growth hormone dyssecretion-induced short stature

Aki Ishikawa, Miyako Mizukami, Akira Ishii, Takeshi Tsugawa, Akihiro Sakurai

Sapporo Medical University, Japan

P3-10-6 Hustle in my muscle: A case of Pompe disease

Marielle Millete Bravo, James Albert Edward Benitez, Marie Julianne Racoma

Bataan General Hospital and Medical Center, Philippines

P3-10-7

A new case of Lysyl hydroxylase 3 deficiency caused by biallelic pathogenic variants in PLOD3

Yuiko Hasegawa[1], Taiga Aoki[2], Kazumi Kawato[1,2], Kumiko Yanagi[2], Tadashi Kaname[2], Nobuhiko Okamoto[1,2]

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P3-10-8

Three patients with classical lissencephaly and PAFAH1B1 deletion

Hiroshi Matsumoto[1], Fumi Hirose[2], Hajime Wakamatsu[2], Eri Takeshita[3], Mitsuhiro Kato[4], Mitsuko Nakashima[5], Hirotomo Saitsu[5], Shigeaki Nonoyama[2]

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4 Department of Pediatrics, Showa University School of Medicine, Japan
5 Department of Biochemistry, Hamamatsu University School of Medicine, Japan

P3-10-9

Novel FBN1 intron variant causes isolated ectopia lentis via in-frame exon skipping

Yoichi Mashimo[1], Norihiro Shimizu[2,3], Hirotaka Yokouchi[2], Yosuke Nishio[4,5], Setsu Sawai[6,7], Tomohiko Ichikawa[7,8], Tomoo Ogi[5,9,10,11], Takayuki Baba[2], Yoshihiro Onouchi[1,7]

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5 Department of Genetics, Research Institute of Environmental Medicine, Nagoya University, Japan
6 Department of Neurology, Chiba Aoba Municipal Hospital, Japan
7 Division of Clinical Genetics, Chiba University Hospital, Japan
8 Department of Urology, Chiba University Graduate School of Medicine, Japan
9 Department of Human Genetics and Molecular Biology, Graduate School of Medicine, Nagoya University, Japan
10 Center for One Medicine Innovative Translational Research (COMIT), Nagoya University Institute for Advanced Study, Japan
11 Division of Molecular Physiology and Dynamics, Institute for Glyco-core Research (iGCORE), Tokai National Higher Education and Research System, Japan

P3-10-10

Experiences and perceptions of the symptoms of patients with Fabry disease during their undiagnosed period

Moeko Isono[1], Minori Kokado[1], Rie Okada[2], Hisao Harada[2], Kazuto Kato[1]

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2 Japan Fabry Disease Patients and Family Association (JFA), Japan

P3-10-11

Detection of copy number variations from whole exome sequencing in skeletal dysplasia patients

Kenichi Yamamoto[1,2,3], Yasuhisa Ohata[3], Makoto Fujiwara[3], Shinji Takeyari[3], Chieko Yamada[3], Yukako Nakano[3], Hirofumi Nakayama[3,5], Ikue Hata[6], Taichi Kitaoka[3], Takuo Kubota[3], Yukinori Okada[4], Keiichi Ozono[3]

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5 Surgery for Oral and Maxillofacial Disease, Osaka University Graduate School of Dentistry, Japan
6 Dept. of Pediatrics, Fukui Prefectural Hospital, Japan

P3-10-12

Five cases of the LIPH gene identified in Japanese patients with autosomal recessive woolly hair

Satoko Minakawa[1,2,3], Yasushi Matsuzaki[2], Eijiro Akasaka[2], Tamio Suzuki[4], Hirofumi Tomita[3], Daisuke Sawamura[2]

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3 Department of Clinical Laboratory, Hirosaki University Hospital, Aomori, Japan
4 Department of Dermatology, Faculty of Medicine, Yamagata University, Yamagata, Japan

P3-10-13 **Genetic landscape of Japanese pediatric patients with neurodevelopmental disorders**

Daiju Oba, Mariko Sagara, Sayuri Oda-Hasuko, Kenta Hasumi, Miyu Fukushima, Yuki Sawada, Miwako Kizumi, Hirofumi Ohashi

Saitama Children's Medical Center, Japan

P3-10-14 **A Japanese pedigree of acral peeling skin syndrome suggesting autosomal-dominant inheritance**

Toshihide Higashino[1], Mayu Konomi[1], Yusuke Kawamura[2], Yoshinori Miura[1]

1 Self-Defense Forces Central Hospital, Japan
2 National Defense Medical College, Japan

P3-10-15 **De novo CLCN3 variants affecting Gly327 cause severe neurodevelopmental syndrome with brain structural abnormalities**

Mitsuko Nakashima[1], Emanuela Argilli[3], Sayaka Nakano[4], Elliott Sherr[3], Mitsuhiro Kato[2], Hirotomo Saitsu[1]

1 Hamamatsu University School of Medicine, Japan
2 Showa University School of Medicine, Japan
3 University of California, USA
4 Itami City Hospital, Japan

P3-10-16 **Long-term survival case with severe infantile Marfan syndrome**

Itsumi Sato, Atsuhito Takeda, Hirokuni Yamazawa, Ayako Chida-Nagai, Daisuke Sasaki, Yuji Maruo, Asuka Takahata

Department of Pediatrics, Hokkaido University Hospital, Japan

P3-10-17 **BUSHY BABY, CAN'T DO POTTY: A case of Cornelia de Lange syndrome with rare case of annular pancreas in a newborn**

Ina Paula Santos

BGHMC, Philippines

P3-10-18 **The genetic and clinical spectrum of Coffin-Siris syndrome-8 due to SMARCC2 variants**

Rika Kawata[1], Naomi Araki[1], Asuka Hori[4], Kenichiro Hata[3,4], Fumio Takada[2]

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2 Kitasato University Graduated School of Medical Sciences, Japan
3 Gunma University Graduate School of Medicine, Japan
4 Research Institute, National Center for Child Health and Development, Japan

P3-10-19 **Clinical retrospective study of fat emulsion, tranexamic acid, and ascorbic acid in 4 patients with ARC syndrome**

Yasutsugu Chinen[1], Sadao Nakamura[1], Noriko Nakayama[1], Hideki Goya[1], Tomohide Yoshida[1], Kumiko Yanagi[2], Tadashi Kaname[2], Kenji Naritomi[3], Koichi Nakanishi[1]

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3 Okinawa Nanbu Habilitation and Medical Center, Japan

P3-10-20 **Adventitial collagen fibrils in mouse model for vascular Ehlers-Danlos syndrome**

Kazuyo Kiribayashi[1], Shinichiro Ohno[2], Natsuko Inagaki[1], Masahiko Kuroda[2]

1 Tokyo Medical University Hospital, Japan
2 Tokyo Medical University, Japan

P3-10-21 **Genetic testing and clinical care for Birt-Hogg-Dubé syndrome: A study of 5 families**

Yuna Sasaki[1], Mitsuko Furuya[1,2], Takahiro Osawa[3], Teruki Yanagi[4], Kaoruko Shimizu[5], Yuka Shibata[1], Masaaki Matsushima[1,6], Ichiro Yabe[1,6], Takahiro Yamada[1]

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4 Department of Dermatology, Hokkaido University Hospital, Japan
5 Department of Respiratory Medicine, Hokkaido University Hospital, Japan
6 Department of Neurology, Hokkaido University Hospital, Japan

P3-10-22 **How to visualize the phenotype diversity: A report with Alexander's disease case reports**

Eisuke Dohi[1], Yuka Tateishi[2], Jae-moon Shin[3], Shinichiro Tago[4], Toyofumi Fujiwara[3], Yasunori Yamamoto[3]

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2 Japan Science and Technology Agency Department of NBDC Program, Japan
3 Research Organization of Information and Systems, Database Center for Life Science, Japan
4 Computing Laboratory, Fujitsu Limited, Japan

P3-10-23 **A case of multiple clinical manifestations leading to the diagnosis of Alstrom syndrome**

Yumi Matsuyama[1,2], Yonehiro Kanemura[3], Hiroyuki Yasojima[4], Tatsuo Matsunaga[5], Hiroshi Nishimura[6]

1 Division of Molecular Medicine, Institute for Clinical Research, National Hospital Organization, Osaka National Hospital, Japan
2 Gifu University Hospital, Clinical Genetics Center, Japan
3 Institute for Clinical Research, National Hospital Organization, Osaka National Hospital, Japan
4 Department of Surgery, Breast Oncology, National Hospital Organization, Osaka National Hospital, Japan
5 National Institute of Sensory Organs Division of Hearing and Balance Research / Medical Genetics Center, National Hospital Organization Tokyo Medical Center, Japan
6 Department of Otorhinolaryngology-Head and Neck Surgery, National Hospital Organization Osaka National Hospital, Japan

P3-10-24 **Comprehensive support for families regarding intellectual developmental disorder by KDM5C variants**

Hiroshi Futagawa[1], Kentar Fukuda[1], Haruka Yamanaka[1], Maho Kuroda[1], Shiho Ito[1], Masataka Honda[1], Mamiko Yamada[2], Hisato Suzuki[2], Toshiki Takenouchi[2], Kenjiro Kosaki[2], Hiroshi Yoshihashi[1]

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2 Center for Medical Genetics, Keio University School of Medicine, Japan

P3-10-25 **Acute encephalopathy with ATP1A2 mutation: Case reports**

Naoki Yamada[1], Ichiro Kuki[1], Kohei Matsubara[1], Risako Ishioka[3], Masataka Fukuoka[1], Megumi Nukui[1,3], Takeshi Inoue[1], Kiyoko Amo[2], Shin Okazaki[1,3]

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2 Department of Pediatric Emergency, Osaka City General Hospital, Japan
3 Department of Pediatric Logopedics, Osaka City General Hospital, Japan

P3-10-26 **Initiative on Rare and Undiagnosed Diseases (IRUD) at Tottori University Hospital**

Tomoki Nonaka[1], Tetsuya Okazaki[1], Noriko Kasagi[1,2], Kaori Adachi[2], Eiji Nanba[2,3], Hiroyuki Awano[1,2], Yoshihiro Maegaki[1]

1 Tottori University Hospital, Japan
2 Tottori University, Japan
3 Otani Hospital, Japan

P3-10-27 **Dual genetic diagnosis contributes to atypical mitochondrial encephalomyopathy, lactic acidosis and stroke-like episodes**

Lip Hen Moey[1], Yusnita Yakob[2]

1 Department of Clinical Genetics, Penang Hospital, Malaysia
2 Unit of Molecular Diagnostics, Specialised Diagnostics Centre, Institute for Medical Research, National Institute of Health, Ministry of Health Malaysia, Malaysia

Poster Session 3-11 Ethical, Legal and Social Implications

Date ：Saturday, October 14, 2023 13:00 ～ 13:30 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P3-11-1 **A qualitative study on the reasons for access restrictions to sensitive medical information, including genetic data**

Mizuho Suzuki, Yuko Ohnuki, Ai Unzaki, Kei Takeshita

Tokai University School of Medicine, Japan

P3-11-2 **Exploratory study for patient and public involvement in genomic researchers**

Suzuka Kato[1], Hiroko Kohbata[1], Yusuke Ebana[1], Kaori Muto[2], Fuji Nagami[3], Masayuki Yoshida[1]

1 Tokyo Medical and Dental University, Japan
2 The University of Tokyo, Japan
3 Tohoku University, Japan

P3-11-3 **Report on genetic testing activities at the section of genetic testing for congenital disorders in CLC at NCCHD**

Yoko Kuroki[1,2,3,4,5], Keiko Matsubara[1,2,4,5,7], Aki Ueda[1,2,4,5,7], Nobutaka Kiyokawa[1,2,6], Maki Fukami[1,5,7]

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2 Clinical Laboratory Center, National Research Institute for Child Health and Development, Japan
3 Dept. of Genome Medicine, National Research Institute for Child Health and Development, Japan
4 Div. of Collaborative Research, National Research Institute for Child Health and Development, Japan
5 Div. of Diversity Research, National Research Institute for Child Health and Development, Japan
6 Dept. of Pediatric Hematology and Oncology Research, National Research Institute for Child Health and Development, Japan
7 Dept. of Molecular Endocrinology, National Research Institute for Child Health and Development, Japan

P3-11-4 **Equity, diversity, and inclusion in genomics: An analysis of the current state of knowledge and initiatives**

Kate Nakasato[1,2], Kazuto Kato[1,2]

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2 Department of Biomedical Ethics and Public Policy, Osaka University Graduate School of Medicine, Japan

P3-11-5 **Withdrawn**

Poster Session 3-12 Genetic Counseling in Asia 3

Date ：Saturday, October 14, 2023 13:00 ～ 13:30 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P3-12-1 **Report on genetic tumor counseling and genomic profiling - A case of pathogenic variant in ATM**

Tomoko Ito, Kouji Ohta, Jun Kato, Ikue Hata

Fukui Prefectural Hospital, Japan

P3-12-2 **Genetic counseling for late-onset neurodegenerative diseases (LONDs): Lessons from Taiwan**

NaiQi Chen, Yih-Ru Cheng, Wuh-Liang Hwu, Ni-Chung Lee, Yin-Hsiu Chien

National Taiwan University Hospital, Taiwan

P3-12-3 **Reliability and validity of the Japanese version multidimensional impact of cancer risk (MICRA) scale**

Tomoko Watanabe[1], Kaori Kimura[2], Minako Kakimoto[3], Yumie Hiraoka[2], Manami Matsukawa[1], Hiroko Nagahashi[3], Saki Horiguchi[3], Miwa Toshima[3], Takeshi Kuwata[2], Teruhiko Yoshida[1], Makoto Hirata[1], Noriko Tanabe[1,4]

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2 Department of Genetic Medicine and Services, National Cancer Center Hospital East, Japan
3 Department of Nursing, National Cancer Center Hospital, Japan
4 Department of Clinical Genetics, Saitama Medical Center, Saitama Medical University, Japan

P3-12-4 **BRCA pathogenic variant carrier who developed ovarian cancer before RRSO**

Ai Itagaki, Yukiko Yoshimoto, Tsuyoshi Tachibana, Sachiko Takahara

Kitano Hospital, Japan

P3-12-5 **Transmission of genetic test results to blood relatives by BRCA variant holders**

Maki Ukita[1], Toru Murata[3], Rieko Fujie[2], Tamae Oe[1]

1 Field of Genetic Counseling, Department of Clinical Laboratory Medicine, Graduate School of Health Sciences, Fujita Health University, Japan
2 Medical Communication, Medical faculty, Fujita Health University, Japan
3 Okazaki Municipal Hospital, Japan

P3-12-6 **Perceptions of Filipino adolescents and emerging adults with IMD and providers in achieving transition to adult care**

Roxanne Janica Merencilla[1], Leniza de Castro-Hamoy[2]

1 Institute of Human Genetics, National Institute for Health - University of the Philippines Manila, Philippines
2 Department of Pediatrics- Philippine General Hospital, Philippines

P3-12-7 **Case of CPS-1 deficiency with a family history: Problems in genetic counseling for neonatal-onset of metabolic disorders**

Yuta Sudo, Yoko Nakajima, Yasuaki Yasuda, Katsuyuki Yokoi, Tetsushi Yoshikawa, Tetsuya Ito

Department of Pediatrics, Fujita Health University School of Medicine, Japan

P3-12-8 **Changes in BRCA genetic testing and information management for breast cancer patients**

Kazuo Matsuura, Ayaka Sakakibara, Ayako Nakame, Yuki Ichinose, Akihiro Fujimoto, Asami Nukui, Kei Yamaguchi, Aya Asano, Hiroko Shimada, Masahiro Ohara, Hiroshi Ishiguro, Akihiko Osaki, Toshiaki Saeki

Department of Breast Oncology, Saitama Medical University International Medical Center, Japan

P3-12-9 **Impact on quality of life in women with hereditary breast and ovarian cancer undergoing risk-reducing mastectomy**

Mayuko Inuzuka[1,2], Chie Watanabe[1,2], Kanae Taruno[1], Takashi Kuwayama[1], Seigo Nakamura[1,3]

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2 Showa University Graduate School of Health Sciences, Japan
3 Showa University Institute for Clinical Genetics and Genomics, Japan

Poster Session 3-13 Others

Date ：Saturday, October 14, 2023 13:00 ～ 13:30 Poster Room（Big Hall, 2F, Zenkoku Toshi Kaikan）

P3-13-1 **Prevalence and characterization of NOTCH2NLC GGC repeat expansions in Koreans**

Seungbok Lee[1,2], Jihoon Yoon[1], Juhyeon Hong[3], Narae Kim[4], Jana Vandrovcova[5], Wai Yan Yau[6], Jae So Cho[1,2], Sheehyun Kim[1], Man Jin Kim[1,7], Soon-Tae Lee[4], Kon Chu[4], Sang Kun Lee[4], Han-Joon Kim[4], Jungmin Choi[3], Jangsup Moon[1,4], Jong Hee Chae[1,2]

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3 Department of Biomedical Sciences, Korea University College of Medicine, Seoul, Korea
4 Department of Neurology, Seoul National University Hospital, Seoul, Korea
5 Department of Neuromuscular Diseases, Institute of Neurology, University College London, London, United Kingdom
6 Perron Institute for Neurological and Translational Science, the University of Western Australia, Nedlands, Western Australia, Australia
7 Department of Laboratory Medicine, Seoul National University Hospital, Seoul, Korea

P3-13-2 **The study of the significance of learning human genetics on self-esteem for high school students in Japan**

Yuka Wada[1], Takahito Wada[2], Sinji Kosugi[1], Takeshi Nakajima[1]

1 Genetic Counselor Course, Graduate School of Medicine, Kyoto University, Japan
2 Department of Genomic Medicine, Graduate School of Medicine, Kyoto University, Japan

P3-13-3 **Impact of modern human-specific genetic changes in skeletal muscle: Insights from a mouse model with humanized *ADSL* gene**

Shin-Yu Lee[1], Xiang-Chun Ju[1], Chika Azama[1], Limin Chen[1], Hugo Zeberg[1,2,3], Srante Pääbo[1,3]

1 Human Evolutionary Genomics Unit, Okinawa Institute of Science and Technology, Okinawa, Japan
2 Department of Physiology and Pharmacology, Karolinska Institutet, Stockholm, Sweden
3 Department of Evolutionary Genetics, Max Planck Institute for Evolutionary Anthropology, Leipzig, Germany

P3-13-4 **Malagasy with Asian roots - Cleft lip and palate surgery**

Yasuyoshi Tosa

Department of Plastic and Reconstructive Surgery, Keio University School of Medicine, Japan

Luncheon Seminar in English

Luncheon Seminar 1

Date ：Thursday, October 12, 2023　12:00 ～ 12:50　Room A（Cosmos, 3F, Toshi Center Hotel）
Chair ：Kenjiro Kosaki（Center for Medical Genetics, Keio University School of Medicine, Japan）
Sponsor：Twist Bioscience, JAPAN

LS1 **Genomics for Lifelong Health Benefits: Applications in Screening, Diagnosis and Prediction**

Sebastian Lunke

Division of Genetics and Genomics, Victorian Clinical Genetics Services, Murdoch Children's Research Institute, Melbourne, Australia

Luncheon Seminar 5

Date ：Thursday, October 12, 2023　12:00 ～ 12:50　Room E（706, 7F, Toshi Center Hotel）
Chair ：Tomohiro Yamamoto（Molecular Systems Marketing Dept. Hitachi High-Tech, Japan）
Sponsor：Hitachi High-Tech

LS5 **A novel system for human whole-genome structural variation analysis**

John Thompson[1], Tateo Nagai[2], Justin Cowling[3]

1 Principal Application Scientist, Nabsys, USA
2 Genomic Bioinformatics, Business Creation Dept. Hitachi High-Tech, Japan
3 VP Sales and Marketing, OmniTier, USA

Luncheon Seminar 10

Date ：Friday, October 13, 2023　12:00 ～ 12:50　Room D（701, 7F, Toshi Center Hotel）
Chair ：Yutaka Suzuki（Laboratory of Systems Genomics, Department of Computational Biology and Medical Sciences, Graduate School of Frontier Sciences, The University of Tokyo）
Sponsor：MGI Tech Co., Ltd.

LS10 **Critical elements for protecting children from severe genetic disorders at the genome medicine era in China.**

Yiping Shen

Division of Genetics and Genomics, Harvard Medical School, Boston, USA

Luncheon Seminar 11

Date ：Friday, October 13, 2023　12:00 ～ 12:50　Room E（706, 7F, Toshi Center Hotel）
Chair ：Masayuki Yoshida（Tokyo Medical and Dental University Hospital, Japan）
Sponsor：FINGGAL LINK CO., LTD.

LS11 **Limiting gene testing in cancer patients deprives them of clinically useful information: The case for universal testing**

Robert L. Nussbaum

Invitae Corporation, USA

Luncheon Seminar 16　Oxford Nanopore: A high-precision approach for exploring human disease

Date ：Saturday, October 14, 2023　12:00 ～ 12:50　Room D（701, 7F, Toshi Center Hotel）
Chair ：Kenjiro Kosaki（Center for Medical Genetics, Keio University School of Medicine, Japan）
Sponsor：Oxford Nanopore Technologies plc

LS16-1 **Long-read Nanopore sequencing identified D4Z4 contractions in patients with facioscapulohumeral muscular dystrophy**

Vorasuk Shotelersuk

Center of Excellence for Medical Genomics, Faculty of Medicine, Chulalongkorn University
Thai Society of Human Genetics (TSHG)

LS16-2 **The high precision of the latest nanopore sequencers and the future of nanopore sequencers.**

Mari Miyamoto

Oxford Nanopore Technologies, Japan

Luncheon Seminar 18　How to get your clinical research published

Date ：Saturday, October 14, 2023　12:00 ～ 12:50　Room F（Meeting Room 1, 3F, Zenkoku Toshi Kaikan）
Chair ：Toshiki Takenouchi（Department of Pediatrics, Keio University School of Medicine, Japan）

LS18-1 **Clinical case value and presentation**

Yiping Shen

Boston Children's Hospital, Medical School, USA

LS18-2 **How to write succinct introduction**
Guidelines for responding to reviewers

Tiong Yang Tan

Victorian Clinical Genetics Services, Murdoch Children's Research Institute, University of Melbourne Department of Paediatrics, Royal Children's Hospital, Australia

LS18-3 **How to produce publication quality figures**

Katta M. Girisha

Sultan Qaboos University, Muscat, Oman and Manipal Academy of Higher Education, Manipal, India

LS18-4 **Using the Elements of Morphology in your case reports**

Brian H.Y. Chung

Department of Paediatrics and Adolescent Medicine, School of Clinical Medicine, The University of Hong Kong, Hong Kong