# Thursday, April 7

15:00-16:30 Main Hall

**PCS** **Plenary Closing Symposium**

## "New Technology of Single Molecule Genome Sequencing"

Chairs: **Shinichi Morishita**
*Department of Computational Biology and Medical Sciences, Graduate School of Frontier Sciences, The University of Tokyo, Japan*

**Yutaka Suzuki**
*Department of Medical Genome Sciences, Graduate School of Frontier Sciences, The University of Tokyo, Japan*

Thu(5)-PCS-1

**What do you call a complete, contiguous and accurate sequence? A SMRT Sequence!**

**Stephen Turner**
*Founder and CTO, Pacific Biosciences, Menlo Park, CA, USA*

Thu(5)-PCS-2

**Real time, portable DNA sequencing using nanopore sensing**

**Clive G. Brown**
*Chief Technology Officer at Oxford Nanopore Technologies Ltd., UK*

Thu(5)-PCS-3

**Single-molecule Electrical Sequencing of DNA, RNA, and Peptide**

**Tomoji Kawai**
*The Institute of Scientific and Industrial Research, Osaka University, Japan*

12:45-14:45 Annex 1

**CIS25** **Concurrent Invited Session 25**

## "Databases and Data Sharing for Cross-border Genomics"

Conveners: **Yasukazu Nakamura**
*Genome Informatics Laboratory, National Institute of Genetics, Japan*
**Guy Cochrane**
*European Nucleotide Archive/European Bioinformatics Institute (EMBL-EBI), UK*

In this session, we address the existing and emerging infrastructure and practices that serve global genomics data sharing. With speakers representing major bioinformatics data resources, a key knowledge organisation technology, a large-scale national medical genomics programme and a global genomics data initiative, we will explore technical and human aspects of databases and data sharing.

Thu(5)-CIS25-1

### Data coordination in cross-border genomics: A very human challenges

**Guy Cochrane**
*European Nucleotide Archive/European Bioinformatics institute (EMBL-EBI), UK*

Thu(5)-CIS25-2

### Genome graphs: A new kind of reference from human genetic variation

**David Haussler**
*University of California Santa Cruse, Genomics Institute, The Global Alliance for Genomics and Health, USA*

Thu(5)-CIS25-3

### The 100,000 Genome Project, UK

**Mark J. Caulfield**
*William Harvey Research Institute, Barts and The London School of Medicine and Dentistry, Queen Mary, UK*

Thu(5)-CIS25-4

### The Human Phenotype Ontology: A Resource for Clinical Data Sharing and Phenotype-Driven Genomic Diagnostics

**Peter N. Robinson**
*Institute for Medical Genetics and Human Genetics, Charité Universitätsmedizin Berlin; Free University Berlin; Berlin-Brandenburg Center for Regenerative Therapy; Max Planck Institute for Molecular Genetics, Germany*

12:45-14:45 Annex 2

## CIS26 Concurrent Invited Session 26
## "Clinical Sequencing"

Conveners: **Kenjiro Kosaki**
*Center for Medical Genetics, Keio University School of Medicine, Japan*
**Leslie G. Biesecker**
*National Human Genome Research Institute, National Institute of Health, USA*

The first speaker in the session will be Professor Eric E. Schadt from Icahn School of Medicine at Mount Sinai. His twill address the role of engaging patients in long-term relationships to enhance interpretation of genomic testing results. He will also address frontiers of sequencing technology employing next generation sequencers.
The second speaker in the session will be Professor Veltman, from Radboud University in the Netherlands. He will discuss approaches to the future unification of what is now a multimodal approach to molecular diagnosis. Severe intellectual disability (ID) is an excellent model for this challenge because it is genetically and clinically highly heterogeneous. The large number of genes involved in ID, plus the heterogeneity of the molecular lesions challenge us to push the frontiers of sequencing technology to encompass the many lesions which range from substitutions to triplet repeats, to CNVs to trisomy. Moving toward a single test platform is critical for cost efficiencies in the laboratory and the clinic.
The third speaker in the session will be Dr. Biesecker of the U.S. Genome Institute at the NIH. He will address some of the conceptual shifts in approaching genetic testing and screening that need to be addressed and how these changes in clinical testing mirror opportunities in clinical research. These concepts include the hypothesis-generating research design and in clinical medicine genomic screening (opportunistic or primary). These two uses of next-generation sequencing (research and clinical) are connected through secondary findings; which is the practice of opportunistic screening in the context of clinically indicated sequencing.
The final speaker is Professor Kosaki of the Center for Medical Genetics, Keio University School of Medicine. He will address how next-generation sequencing is entering the clinic from two clinical studies; medical exome analysis of singletons and trios. By evaluating hundreds of cases with these two complementary approaches, this review will demonstrate both the power of the approaches both to diagnose disease and for medical discovery, but also the critical need to develop novel approaches to exome analysis, as exemplified by directed searches for retrotransposon and regulatory element mutational events as a solution for currently unsolved cases.

Thu(5)-CIS26-1
**Engaging patients in long-term relationships to enhance interpretation of genomic testing results**
**Eric Schadt**
*Genetics and Genomic Sciences, Mount Sinai Hospital, USA*

Thu(5)-CIS26-2
**Towards single-test genomics**
**Joris A. Veltman**
*Department of Human Genetics, Radboud University Medical Center; Department of Clinical Genetics, Maastricht University Medical Centre, Maastricht, The Netherlands*

Thu(5)-CIS26-3
**Hypothesis-generating research and predictive genomics**
**Leslie G. Biesecker**
*National Human Genome Research Institute, National Institute of Health, USA*

Thu(5)-CIS26-4
**Genome-scale sequencing in clinical settings**
**Kenjiro Kosaki**
*Center for Medical Genetics, Keio University School of Medicine, Japan*

12:45-14:45 Room A

**CIS27** **Concurrent Invited Session 27**

## "Effects of Genetic and Epigenetics, Geno-environmental Interactions on Healthy Aging and Longevity"

Conveners: **Yi Zeng**
*Center for the Study of Aging and Human Development, Medical School of Duke University, Durham, NC, USA; Center for Healthy Aging and Development Studies, National School of Development, Peking University, Beijing, China, China/USA*

**Makoto Suzuki**
*Okinawa Research Center for Longevity Science, Japan*

While human lifespan is increasing and the number of elderly (especially oldest-old) is rapidly growing in almost all countries in the world, is it possible to realize compression of morbidity, or at least dynamic equilibrium, rather than expansion of disability? Why do some people survive to advanced ages with good health but others suffer severe disability and morbidity? So far, there are not many good answers to these critical questions.
Various research have shown that genetics, epigenetics and gene-environment (GxE) interactions play crucial roles in health and longevity, because environmental factors may activate or regulate gene expressions and functions, which then influences health outcomes. Existing literature indicate that because of GxE interaction effects, the positive or negative associations between environmental factors and health outcomes differ significantly among individuals with different genotypes. Consequently, Genetic, epigenetic and gene-environment interaction studies can contribute to yield significantly increased benefits and reduced costs of health promotion, programs which consider individuals' genetic profiles, and enable much more people to enjoy better health while lifespan is increasing. This is the goal of present session and we intend to promote academic discussions on this important topic, and make contributions to improve the quality of life for the elderly and all members of their families and entire society around the world.

Thu(5)-CIS27-1

### Genetic studies of the oldest old: Somatic and germline variation

**Eline Slagboom**
*Molecular Epidemiology Section, Department of Medical Statistics and Bioinfomatics, Leiden University Medical Centre, The Netherlands*

Thu(5)-CIS27-2

### Energy Sensing Genes and Longevity: Novel Findings from Multi-Ethic Populations Energy Sensing Genes and Longevity: Novel Findings from Multi-Ethic Populations

**Bradley J. Willcox and D. Craig Willcox**
*Department of Geriatric Medicine / Department of Research, University of Hawaii / Kuakini Medical Center, USA*

Thu(5)-CIS27-3

### Meta-analysis of 4 genome-wide association studies identify new longevity genes

**Paola Sebastiani and Thomas Perls**
*Department of Biostatistics, Boston University, USA*

Thu(5)-CIS27-4

### Associations of novel loci, pathway-specific polygenic scores and GxE interactions with longevity and cognition in Han Chinese

**Yi Zeng**
*Center for the Study of Aging and Human Development, Medical School of Duke University USA; Center for Healthy Aging and Development Studies, National School of Development, Peking University, Beijing, China*

12:45-14:45 Room E

## CIS28 Concurrent Invited Session 28
## "Current Aspects of Inborn Error of Metabolism"

Conveners: **Shigeo Kure**
*Department of Pediatrics, Tohoku University School of Medicine, Japan*
**Wuh-Liang Hwu**
*Department of Pediatrics and Medical Genetics, National Taiwan University Hospital, Taiwan*

This session includes four current topics in the study of inborn errors of metabolism (IEM), which will be presented by four distinguished speakers. The presenting topics are mitochondrial diseases, IEMs of amino acid, IEMs of metals and the gene therapy. First speaker, Dr Thorburn from Melbourne, has identified many causative genes for mitochondrial disease by using next generation sequencing (NGS), and will present the latest genetic view of mitochondrial diseases. Second speaker, Dr Blom from Heidelberg, is an expert of biochemical and genetic analysis of IEMs, especially one-carbon metabolism. He will review the current topics of IEMs of amino acid, including recent progress in the study of homocysteine metabolism. Third speaker, Dr Yoo from Seoul, will update genetic and metabolic pathogenesis of IEMs of metals, especially of cupper metabolism including Wilson and Menkes diseases. Last speaker, Dr Hwu from Taipei, will present the first successful gene therapy for aromatic acid decarboxylase (AADC) deficiency using adeno-associated virus vector, which has amazingly ameliorated prognosis of patients with AADC. These four lectures would delineate various aspects of current researches on IEM.

Thu(5)-CIS28-1

### Novel mitochondrial diseases identified by NGS

**David R. Thorburn**
*Genetics, Murdoch Childrens Research Institute; University of Melbourne, Dept of Paediatrics; Victorian Clinical Genetics Services, Australia*

Thu(5)-CIS28-2

### Current topics in inborn errors of amino acid metabolism

**Henk Blom**
*Laboratory of Clinical Biochemistry and Metabolism, Department of General Pediatrics, Adolescent Medicine and Neonatology, University Medical Centre Freiburg, Netherlands*

Thu(5)-CIS28-3

### Clinical and molecular spectrum of Wilson disease (WD) patients with understanding of molecular pathophysiology of WD in animal model, Long-Evans Cinnamon rats

**Han-Wook Yoo**
*Pediatrics & Medical Genetics, Asan Medical Center Childrens Hospital, University of Ulsan College of Medicine, Korea*

Thu(5)-CIS28-4

### Gene therapy for aromatic L-amino acid decarboxylase (AADC) deficiency

**Wuh-Liang Hwu**
*Department of Pediatrics and Medical Genetics, National Taiwan University Hospital, Taiwan*

12:45-14:45 Room B-1

## SFS20 Special Focus Session 20
## "HVP (Sharing Human Variant Data Globally - Challenges and Opportunities for 2020)"

Moderator: **Helen M. Robinson**
*Liason-World Health Organization*
*The Human Variome Project International Coordinating Office*
*University of Melbourne, Australia*

This session will focus on recent developments and future challenges faced in the sharing of human genetic and genomic information on variants to support clinical practice across the world. It will discuss issues in relation to current trends in collecting, curating and interpreting global knowledge on variants. HVP has been instrumental in establishing several key projects that are making significant progress in these areas and these projects will be used to report on progress in the past two years. The session will also pay tribute to the work and vision of the late Professor Richard Cotton who was the Funding Patron of HVP until his death in June 2015. Project-wide initiatives of HVP in haemoglobinopathies and some cancers combine with those on gastrointestinal heredity tumours and cystic fibrosis, and form the basis of this work on establishing international consortia to link and harmonise work being done in all parts of the world. The value of accurate assignment of pathogenicity of variants in clinical practice is a challenge currently being addressed. The need to form multi-disciplinary teams across all region of the world to build expertise in consistent interpretation based on the best evidence will be described in this session. How to organize this engagement through a series of initiatives and mechanisms will be described together with lessons learned and implications for the future. Examples include:
Breast cancer is a high profile disease. Understanding the impact of variants carried by patients is of great clinical importance as it directs risk-reducing management strategies that improve survival such as screening and prophylactic surgery and the choice of therapy after diagnosis. Unclassified variants area significant clinical problem that can be addressed through the systematic collection and curation of key data that influencing clinical decision making;
An emphasis on Haemoglobinopathies aims to raise the profile of genomic medicine in low and middle income countries and develop the capacity required for diagnosing, treating carriers in low and middle income countries by applying key developments in human genomics to heamoglobinopathies. Tackling haemoglobinopathies is an ideal entry point for these countries to develop the necessary infrastructure and expertise that can expand into other areas of health-service delivery. Growth in the quality and quantity of curated inputs into internationally recognized genetic databases from low- and middle-income countries requires the harmonization between countries in accordance with international best practice.
Ensuring that the storage, curation and sharing of the relevant DNA variation information is sustainable in the medium and longer term is vital. Only by expanding and strengthening the international network of professionals, including curators, researchers, clinicians, bioinformaticians, counsellors, patient groups and health bureaucrats can cost-effective health care objectives be achieved.

Thu(5)-SFS20-1

### Sharing human variant data globally – What can be achieved on the African Continent by 2020?
**Raj Ramesar**
*Division of Human Genetics, University of Cape Town, South Africa*

Thu(5)-SFS20-2

### "Sharing human variant data globally -challenges and opportunities for 2020" Human Variome Project and the Latin American region
**Aida B. Falcón de Vargas**
*Clinical Genetics Unit, Hospital Vargas de Caracas, Escuela de Medicina JM Vargas, Universidad Central de Venezuela. Hospital de Clinicas Caracas, Venezuela*

Thu(5)-SFS20-3

### Human Variome Project; The Global Globin 2020 Challenge (southeast Asia)
**Zilfalil Alwi**
*Universiti Sains Malaysia, Malaysia*

Thu(5)-SFS20-4

### The value of accurate assignment of pathogenicity of variants in clinical genetic practice
**Ingrid M. Winship**
*University of Melbourne and Melbourne Health, Australia*

8:00-9:30 Room D

**ED7** **Educational Program 7**

## "Education of Genetics: Genetics Education for Undergraduate Medical Students in Asia"

Moderators: **Akihiro Sakurai**
*Sapporo Medical University, Japan*
**Meow-Keong Thong**
*Genetics and Metabolism Unit, Department of Paediatrics, Faculty of Medicine, University of Malaya, Kuala Lumpur, Malaysia*

Until decades ago, clinical genetics remained one of the minor medical subspecialties which cover relatively narrow area of clinical practice (mainly in pediatrics and obstetrics). In this century, however, accumulation of our knowledge in genetic conditions and disorders as well as explosive technical advances of genetic analysis made genetic information fundamental and indispensable for wide range of clinical management. Accordingly, importance of genetics education for medical professionals is growing more than ever. Meanwhile, public perception about issues related to genetics (such as hereditary diseases and reproduction) are thought to be influenced by social, cultural and religious background of the community, of which medical professionals should be aware.
In this session, we discuss genetics education for undergraduate medical students in Asia. Four speakers from Malaysia, Philippines, India and Japan present current status and challenges in their countries. Participation of audience to discussion is welcome.

Thu(5)-ED7-1

**Problem-based learning in genetics education in a developing country**
**Meow-Keong Thong**
*Genetics and Metabolism Unit, Department of Paediatrics, Faculty of Medicine, University of Malaya, Kuala Lumpur, Malaysia*

Thu(5)-ED7-2

**Genetics education in Philippine medical schools**
**Maria Melanie Liberty B. Alcausin**
*Newborn Screening Reference Center, National Institutes of Health-University of the Philippines, Manila, Philippines*

Thu(5)-ED7-3

**Integrating genetics in undergraduate curriculum – Indian perspective**
**Seema Kapoor**
*Department of Pediatrics, Maulana Azad Medical College, New Delhi, India*

Thu(5)-ED7-4

**Genetic Education for Undergraduate Medical Students in Japan**
**Atsushi Watanabe**
*Division of Clinical Genetics, Nippon Medical School, Japan*

9:45-11:15 Room D

## ED8 Educational Program 8
## "Education of Genetics: Genetics Education for Public"

Moderators: **Takahito Wada**
*Kyoto University, Japan*
**Michael J. Dougherty**
*American Society of Human Genetics, USA*

In this session, we would discuss the vital issue of broadening our genetic education activities that are pressingly required in society today, focusing on "What should we do to promote awareness of human genetics among young people, especially pre-college students?", and through this, we would also like to reconsider this increased role of genetic professionals.
The session consists of the five speakers from four countries, Australia, UK, USA, and Japan.
Since 2003, when the complete human genome was sequenced, many new DNA sequencing techniques have been rapidly identifying a lot of susceptibility genes for multifactorial common diseases, as well as for various genes of Mendelian disease. These advances have the huge benefit to our medical care in the diagnosis and treatment of a wide range of disorders. Actually, the President Obama proposed the new conception of "the Precision Medicine Initiative" in State of the Union Address of 2015. On the other hand, for many ordinary people, increased knowledge or information on their genes may present ethical dilemmas between life and treatment styles.
The WHO report in 2003, or "Review of Ethical Issues in Medical Genetics", says that "The goals of medical genetics can be optimally fulfilled only in the context of an educated, informed public. Education about human reproduction and genetics should be part of the educational heritage of every person", and "In the long run, genetics education for the public can best be achieved through education IN SCHOOLS."
We would expect your heated and fruitful discussion on the matter and will be able to find the right direction of our future genetic study to be applied for.

Thu(5)-ED8-1

**Educating the Public about Genetics: A Perspective from the U.S.**
**Michael J. Dougherty**
*American Society of Human Genetics, USA*

Thu(5)-ED8-2

**The Australian experience with genetics education for primary and high school students and current challenges**
**Kristine Barlow-Stewart**
*Sydney Medical School Northern, University of Sydney, Australia*

Thu(5)-ED8-3

**The changing face of genomics learning and its drivers in the UK**
**Mat Hickman**
*Wellcome Trust, UK*

Thu(5)-ED8-4

**Genetic Education for Children: A Nagasaki University Initiative**
**Kanako Morifuji and Noriko Sasaki**
*Department of Nursing, Health Sciences, Nagasaki University Graduate School of Biomedical Sciences, Japan*

8:00-9:30 Annex 1

## O34 Concurrent Oral Session 34
## "Cancer Genetics 4"

Chairs: **Anne Goverde**
*Department of Clinical Genetics / Department of Gastroenterology & Hepatology, Erasmus MC, University Medical Center Rotterdam, Netherlands*
**Hideki Makishima**
*Department of Pathology and Tumor Biology, Kyoto University, Japan*

Thu(5)-O34-1

### Germline mutations in familial prostate cancer

**Takahide Hayano**[1], Hiroshi Matsui[2], Hirofumi Nakaoka[1], Nobuaki Ohtake[2], Kazuhiro Suzuki[2], Ituro Inoue[1]
*[1]Division of Human Genetics, National Institute of Genetics, Japan, [2]Department of Urology, Gunma University Graduate School of Medicine*

Thu(5)-O34-2

### Imbalance of miR-194 -CUL4B negative feedback loop favoring CUL4B upregulation enhances the malignancy of non-small-cell lung carcinoma

**Yongxin Zou**, Jun Mi, Xiaohua Lin, Juanjuan Lu, Xiaochen Liu, Hui Zhao, Zhaoyang Wang, Huili Hu, Peishan Li, Hao Dou, Baichun Jiang, Changshun Shao, Yaoqin Gong
*Institute of Medical Genetics, School of Medicine, Shandong University, China*

Thu(5)-O34-3

### Hypomorphic *CYP2C9*2* and **3* alleles associate with improved non-small-cell lung cancer (NSCLC) prognosis

**Lindsay N Sausville**[1], Jorge H Capdevila[2,3], Ambra Pozzi[3,4], Scott M Williams[1,5,6]
*[1]Genetics, Dartmouth College, USA, [2]Medicine, Division of Nephrology, Vanderbilt University, [3]Cancer Biology, Vanderbilt University, [4]Biochemistry, Vanderbilt University, [5]Epidemiology and Biostatistics, Case Western, [6]Institute for Quantitative Biomedical Sciences, Darmouth College*

Thu(5)-O34-4

### Cost-effectiveness of routine screening for Lynch syndrome in endometrial cancer patients up to 70 years of age

**Anne Goverde**[1,2], Manon C.W. Spaander[2], Helena C. van Doorn[3], Hendrikus J. Dubbink[4], Ans M.W. van den Ouweland[1], Carli M. Tops[5], Sjarlot G. Kooi[6], Judith de Waard[7], Robert F. Hoedemaeker[8], Marco J. Bruno[2], Robert M.W. Hofstra[1], Esther W. de Bekker-Grob[9], Winand N.M. Dinjens[4], Ewout W. Steyerberg[9], Anja Wagner[1], The LIMO study group
*[1]Department of Clinical Genetics, Erasmus MC, University Medical Center Rotterdam, Netherlands, [2]Department of Gastroenterology and Hepatology, Erasmus MC, University Medical Center Rotterdam, the Netherlands, [3]Department of Gynaecology, Erasmus MC, University Medical Center Rotterdam, the Netherlands, [4]Department of Pathology, Erasmus MC, University Medical Center Rotterdam, the Netherlands, [5]Department of Clinical Genetics, Leiden University Medical Center, Leiden, the Netherlands, [6]Department of Gynaecology, Albert Schweitzer Hospital, Dordrecht, the Netherlands, [7]Department of Gynaecology, Sint Franciscus Gasthuis, Rotterdam, the Netherlands, [8]Pathology laboratory Pathan, Rotterdam, the Netherlands, [9]Department of Public Health, Erasmus MC, University Medical Center Rotterdam, the Netherlands*

Thu(5)-O34-5

**Universal testing of mismatch repair protein deficiency in colorectal cancer and its usefulness for identification of Lynch syndrome patient**

**Takeshi Nakajima**[1,2], Shigeki Sekine[3], Yoshimi Nakajima[1], Mineko Ushiama[4], Taku Sakamoto[1], Takahisa Matsuda[1], Yutaka Saito[1], Yukihide Kanemitsu[5], Hiromi Sakamoto[4], Teruhiko Yoshida[2,4], Kokichi Sugano[2,6]

*[1]Endoscopy Division, National Cancer Center Hospital, Japan, [2]Department of Genetic Counseling, National Cancer Center Hospital, [3]Pathology Division, National Cancer Center Hospital, [4]Division of Genetics, National Cancer Center Research Institute, [5]Colorectal Surgery, National Cancer Center Hospital, [6]Oncogene Research Unit/Cancer Prevention Unit, Tochigi Cancer Center Research Institute, Tochigi, Japan*

Thu(5)-O34-6

**Comprehensive methylation analysis of imprinting-associated differentially methylated regions in colorectal cancer**

**Hidenori Hidaka**[1,2], Ken Higashimoto[1], Saori Aoki[1], Hidetaka Watanabe[1], Hitomi Yatsuki[1], Kenichi Nishioka[1], Keiichiro Joh[1], Toshiyuki Maeda[3], Yasuo Koga[4], Ryuichi Iwakiri[2], Hirokazu Noshiro[4], Kazuma Fujimoto[2], Hidenobu Soejima[1]

*[1]Division of Molecular Genetics and Epigenetics, Department of Biomolecular Sciences, Faculty of Medicine, Saga University, Saga, Japan, [2]Department of Internal Medicine and Gastrointestinal Endoscopy, Saga Medical School, Saga, Japan, [3]Department of Pediatrics, Faculty of Medicine, Saga University, Saga, Japan, [4]Department of Surgery, Saga University Faculty of Medicine, Saga, Japan*

9:45-11:15 Annex 1

## O35 Concurrent Oral Session 35
## "Cancer Genetics 5"

Chairs: **Stuart MacGregor**
*Statistical Genetics Laboratory, QIMR Berghofer Medical Research Institute, Australia*
**Mitsuru Emi**
*Thoracic Oncology, University of Hawaii Cancer Center, USA*

Thu(5)-O35-1

**Whole-exome Analysis of Hereditary Microsatellite-stable Colorectal cancer in Israel**

**Revital Kariv**[1], Guy Rosner[1], Hana Strul[1], Nathan Gluck[1], Sivan Caspi[1], Leon Raslin[2]

*[1]Tel aviv sourasky Medical center, Israel, [2]Vanderblit Ingram cancer Center, Vanderblit university*

Thu(5)-O35-2

**Hepatitis B Virus HBx Activates Notch Signaling via Delta-like 4/Notch1 in Hepatocellular Carcinoma**

**Pornrat Kongkavitoon**, Pisit Tangkijvanich, Nattiya Hirankarn, Tanapat Palaga

*Chulalongkorn university, Thailand*

Thu(5)-O35-3

***miR-19b* up-regulates *hTERT* expression by inhibition of *PITX1* in melanoma cells**

**Takahio Ohira**[1], Naohiro Sunamura[1], Daigo Inaoka[1], Yuji Nakayama[2], Mitsuhiko Osaki[3], Futoshi Okada[3,4], Mitsuo Oshimura[4], Hiroyuki Kugoh[1,4]

*[1]Division of Molecular Genetics and Biofunction, Tottori University Graduate School of Medical Science, Japan, [2]Division of Functional Genomics, Research Center for Bioscience and Technology, Tottori University, Tottori, Japan, [3]Division of Pathological Biochemistry, Tottori University Faculty of Medicine, Tottori, Japan, [4]Chromosome Engineering Research Center, Tottori University, Tottori, Japan*

Thu(5)-O35-4

**The role of germline genetic variation in Breslow's depth, a predictor of survival after melanoma**

**Matthew H Law**[1], Casey Rowe[2], Anne E Cust[3], John L Hopper[4], Graham J Mann[5], Gemma Cadby[6], Sarah V Ward[6], Eric Moses[6], David C Whiteman[7], Nicholas K Hayward[8], Kiarash Khosrotehrani[2], Stuart Macgregor[1]

*[1]Statistical Genetics, QIMR Berghofer Medical Institute, Australia, [2]The University of Queensland, UQ centre for Clinical Research, Brisbane, Australia., [3]Cancer Epidemiology and Prevention Research, Sydney School of Public Health and Melanoma Institute Australia, [4]Centre for Molecular, Environmental, Genetic and Analytic (MEGA) Epidemiology, Melbourne School of Population Health, University of Melbourne, Melbourne, Australia, [5]Westmead Institute of Cancer Research, University of Sydney at Westmead Millennium Institute and Melanoma Institute Australia, Sydney, Australia, [6]Centre for Genetic Origins of Health and Disease, Faculty of Medicine, Dentistry and Health Sciences, The University of Western Australia, Australia, [7]Cancer Control Group, QIMR Berghofer Medical Research Institute, Brisbane, Australia., [8]Oncogenomics, QIMR Berghofer Medical Research Institute, Brisbane, Australia*

Thu(5)-O35-5

**Founder BAP1 Mutation in Four American Families Predisposes to Malignant Peritoneal Mesothelioma, Uveal Melanoma, and other cancers**

**Mitsuru Emi**, Sandra Pastorino, Masaki Nasu, Erin Froles, Haining Yang, Michele Carbone

*Thoracic Oncology, University of Hawaii Cancer Center, USA*

Thu(5)-O35-6

**Large scale meta-analysis identifies several new risk loci for development of esophageal adenocarcinoma and Barrett's esophagus**

**Stuart MacGregor**[1], Puya Gharahkhani[1], Rebecca Fitzgerald[2], Tom Vaughan[3], Ian Tomlinson[4], Ines Gockel[5], Claire Palles[4], Michael Knapp[6], Markus M Noethen[7,8], Jessica Becker[7,8], Paul Pharoah[9], David Whiteman[10], Janusz Jankowski[11,12], Johannes Schumacher[7,8],
Barrett's and Esophageal Adenocarcinoma Consortium (BEACON) and The Wellcome Trust Case Control Consortium 2 (WTCCC2)

*[1]Statistical Genetics, QIMR Berghofer Medical Research Institute, Australia, [2]Medical Research Council (MRC) Cancer Cell Unit, Hutchison-MRC Research Centre and University of Cambridge, Cambridge, United Kingdom, [3]Division of Public Health Sciences, Fred Hutchinson Cancer Research Center, Seattle, WA, USA, [4]Wellcome Trust Centre for Human Genetics, University of Oxford, Oxford, UK, [5]Department of Visceral, Transplant, Thoracic and Vascular Surgery, University Hospital of Leipzig, Leipzig, Germany, [6]Institute for Medical Biometry, Informatics, and Epidemiology, University of Bonn, Bonn, Germany, [7]Institute of Human Genetics, University of Bonn, Bonn, Germany, [8]Department of Genomics, Life & Brain Center, University of Bonn, Bonn, Germany, [9]Centre for Cancer Genetic Epidemiology, Department of Oncology, University of Cambridge, Cambridge, United Kingdom, [10]Cancer Control, QIMR Berghofer Medical Research Institute, Brisbane, Australia, [11]University Hospitals Coventry & Warwickshire NHS Trust, Warwickshire, United Kingdom, [12]Warwick Medical School, University of Warwick, Warwickshire, United Kingdom*

8:00-9:30 Annex 2

## O36 Concurrent Oral Session 36
## "Ethical, Legal, Social and Policy Issues in Genetics"

Chairs: **Adrian Thorogood**
*Centre of Genomics and Policy, McGill University, Canada*
**Kaori Muto**
*Department of Public Policy, The Institute of Medical Sciences, The University of Tokyo, Japan*

Thu(5)-O36-1

### Care to share? An international comparison of research directives to promote data sharing among decisionally-incompetent adults living with dementia

**Adrian Thorogood**[1], Vasiliki Rahimzadeh[1], Bartha M Knoppers[1], Anna Maki-Petaja-Leinonen[2], Martin Bobrow[3], Ageing and Dementia Task Team, Global Alliance for Genomics and Health
*[1]Centre of Genomics and Policy, McGill University, Canada, [2]Faculty of Law, University of Helsinki, [3]Emeritus Professor of Medical Genetics, Cambridge University*

Thu(5)-O36-2

### Holding Researchers to Account for Responsible Genomic Data Sharing

**Adrian Thorogood**[1], Calvin WL Ho[2], Bartha M Knoppers[1]
*[1]Centre of Genomics and Policy, McGill University, Canada, [2]Yong Loo Lin School of Medicine, National University of Singapore*

Thu(5)-O36-3

### European Legal Perspectives on Cloud Computing in Cross-Border Translational Genome Research

**Fruzsina Molnar Gabor**
*Heidelberg Academy of Sciences and Humanities, Germany*

Thu(5)-O36-4

### Return of individual genomic research results in patient biobanks: Ethical challenges for Biobank Japan

**Kaori Muto**, Hyunsoo Hong
*The Institute of Medical Science, The University of Tokyo, Japan*

Thu(5)-O36-5

### Ethical premises in the prenatal diagnosis of birth defects in Cuba

**Beatriz Marcheco-Teruel**, Iris A Rojas-Betancourt
*National Center of Medical Genetics, Cuba*

Thu(5)-O36-6

### Socialising the Genome

**Anna Middleton**[1], Julian Borra[2], Vivienne Parry[3], Katrina Nevin-Ridley[3], Amy Sanders[4], Julian Rayner[1]
*[1]Wellcome Genome Campus, UK, [2]Thin Air Factory, London, UK, [3]Genomics England, London, UK, [4]Wellcome Trust, London, UK*

9:45-11:15 Annex 2

## O37 Concurrent Oral Session 37
## "Therapy for Genetic Disorders"

Chairs: **Jan P. Kraus**
*Dept. of Pediatrics, University of Colorado School of Medicine, USA*

**Yu-ichi Goto**
*Medical Genome Center, National Center of Neurology and Psychiatry, Japan*

Thu(5)-O37-1

### Disruption of Microtubule Dynamics in Rett Syndrome (RTT): a Possible New Therapeutic Target

**John Christodoulou**[1,2,3], Wendy Gold[1,2], Tamara Lacina[4], Sarah Williamson[1], Laurence Cantrill[5]
*[1]Western Sydney Genetics Program, Children's Hospital at Westmead, Australia, [2]Discipline of Paediatrics and Child Health, Sydney Medical School, University of Sydney, [3]Discipline of Genetic Medicine, Sydney Medical School, University of Sydney, [4]Faculty of Biotechnology, Hochschule Mannheim - University of Applied Sciences, Germany, [5]Microscope Facility, Kids Research Institute, Children's Hospital at Westmead, Sydney, Australia*

Thu(5)-O37-2

### Vosoritide (BMN 111) in Children with Achondroplasia: Initial results from a Phase 2, open-label, sequential cohort, dose-escalation study

**Sagar A. Vaidya**[1], Melita Irving[2], Carlos Bacino[3], Xiaofan Cao[1], Joel Charrow[4], Valerie Cormier-Daire[5], Wolfgang Dummer[1], Paul Harmatz[6], Leonid Katz[1], Kevin Larimore[1], John Phillips[7], Julie Hoover-Fong[8], Ravi Savarirayan[9]
*[1]BioMarin Pharmaceutical Inc., USA, [2]Guy's and St. Thomas' NHS Foundation Trust, Evelina Children's Hospital, London, UK, [3]Baylor College of Medicine, Houston, TX, USA, [4]Ann and Robert H. Lurie Children's Hospital of Chicago, Chicago, IL, USA, [5]Institut Imagine, Universite Paris Descartes, Hopital Necker - Enfants Malades, Paris, France, [6]UCSF Benioff Children's Hospital Oakland, Oakland, CA, USA, [7]Vanderbilt University Medical Center, Nashville, TN, USA, [8]Johns Hopkins University School of Medicine, Baltimore, MD, USA, [9]Murdoch Children's Research Institute, Royal Children's Hospital Victoria, University of Melbourne, Parkville, Victoria, Australia*

Thu(5)-O37-3

### Salbutamol inhibits ubiquitin-mediated survival motor neuron protein degradation in spinal muscular atrophy cells

**Nur Imma Fatimah Harahap**[1], Dian Kesumapramudya Nurputra[1], Mawaddah Ar Rochmah[1], Ai Shima[1], Naoya Morisada[1,2], Toru Takarada[3], Atsuko Takeuchi[3], Yumi Tohyama[4], Shinichiro Yanagisawa[5], Hisahide Nishio[1,2]
*[1]Community Medicine and Social Healthcare Science, Kobe University, Graduate School of Medicine, Japan, [2]Department of Pediatrics, Kobe University Graduate School of Medicine, [3]Analytical Center, Kobe Pharmaceutical University, [4]Division of Biochemistry, Faculty of Pharmaceutical Sciences, Himeji Dokkyo University, [5]Division of Medical Economics, Faculty of Pharmaceutical Sciences, Himeji Dokkyo University*

Thu(5)-O37-4

### Enzyme replacement therapy for homocystinuria

**Jan P. Kraus**[1], Erez M. Bublil[1], Tomas Majtan[1], Insun Park[1], Richard Carrillo[1], June Ereno-Orbea[2], Louis A. Martinez-Cruz[2], Helena Hulkova[3], Viktor Kozich[3], Warren Kruger[4]
*[1]Pediatrics, University of Colorado School of Medicine, USA, [2]Structural Biology Unit, Center for Cooperative Research in Biosciences, Bizkaia, Derio, Spain, [3]Institute of Inherited Metabolic Disorders, Charles University, First Faculty of Medicine and General University Hospital, Czech Republic, [4]Cancer Biology Program, Fox Chase Cancer Center, Philadelphia, Pennsylvania, U.S.A.*

Thu(5)-O37-5

**Development of a novel pig model of Duchenne muscular dystrophy and evaluation of antisense-mediated exon skipping**

Kana Hosoki[1], Yusuke Echigoya[1], William Duddy[2], Terence A. Partridge[3,4], Eric P. Hoffman[3,4], Joe N. Kornegay[5], Christopher Rogers[6], Toshifumi Yokota[1,7]

*[1]Department of Medical Genetics, University of Alberta Faculty of Medicine and Dentistry, Canada, [2]Center of Research in Myology, Sorbonne University, UPMC Univ., Paris, France, [3]Research Center for Genetic Medicine, Children's National Medical Center, Washington DC, USA, [4]Department of Integrative Systems Biology, George Washington University School of Medicine, Washington DC, USA, [5]Department of Veterinary Integrative Biosciences, Texas A&M University, TX, USA, [6]Exemplar Genetics, IA, USA, [7]The Friends of Garrett Cumming Research & Muscular Dystrophy Canada HM Toupin Neurological Science Endowed Research Chair, AB, Canada*

Thu(5)-O37-6

**RNA/ENA chimera antisense oligonucleotide (AO85) was safely administered and shown to induce dystrophin exon 45 skipping in Duchenne muscular dystrophy patient: the first clinical study**

Yasuhiro Takeshima[1], Tomoko Lee[1], Hideki Shimomura[1], Yasuhiko Tanaka[1], Hiroyuki Awano[2], Atsushi Nishida[3], Isao Ojima[3], Satoshi Minami[3], Akio Nakagawa[3], Kazumoto Iijima[2], Masafumi Matsuo[3]

*[1]Department of Pediatrics, Hyogo College of Medicine, Japan, [2]Department of Pediatrics, Kobe University Graduate School of Medicine, [3]Department of Physical Rehabilitation, Kobegakuin University*

8:00-9:30 Room A

## O38 Concurrent Oral Session 38
## "Prenatal, Perinatal and Reproductive Genetics 3"

Chairs: **Desheng Liang**
*State Key Laboratory of Medical Genetics, Central South University, China*
**Mayumi Sugiura-Ogasawara**
*Obstetrics and Gynecology, Research Center for Recurrent Pregnancy Loss, Nagoya City University, Graduate School of Medical Sciences, Japan*

Thu(5)-O38-1

**Risk assessment of medically assisted reproduction and advanced maternal ages in the development of Prader-Willi syndrome due to UPD(15)mat**

Keiko Matsubara[1,2], Nobuyuki Murakami[2], Maki Fukami[1], Masayo Kagami[1], Toshiro Nagai[2], Tsutomu Ogata[1,3]

*[1]Department of Molecular Endocrinology, National Research Institute for Child Health and Development, Japan, [2]Department of Pediatrics, Dokkyo Medical University Koshigaya Hospital, [3]Department of Pediatrics, Hamamatsu University School of Medicine*

Thu(5)-O38-2

**SNP Testing before IVF: searching for optimal number and contain**

Andrei V Ivanov

*Human Genetics, University Hospital of Saint-Petersburg State University, Russia*

Thu(5)-O38-3

**The examination of chromosome abnormality in couples with recurrent pregnancy loss**

Hiroaki Aoki[1], Osamu Samura[1], Akiko Konishi[1], Michiko Suzuki[1], Momoko Inoue[1], Madoka Horiya[1], Taizan Kamide[2], Eri Ilkura[3], Tomohiro Tanemoto[1], Rie Tachimoto[1], Takayuki Haino[1], Nozomu Yanaihara[1], Kohei Sugimoto[1], Aikou Okamoto[1]

*[1]Obstetrics and Gynecology, The Jikei University School of Medicine, Japan, [2]Obstetrics and Gynecology, The Jikei University Kashiwa Hospital, [3]Obstetrics and Gynecology, The Jikei University Daisan Hospital*

Thu(5)-O38-4

**Day 5 embryos show reduced aneuploidy rate compared to day 3 embryos in preimplantation genetic diagnosis for reciprocal translocation carriers**

Yoshiharu Nakaoka[1], Michiko Ammae[1], Tatsuya Nakano[1], Kayo Takahashi[1], Kanako Katsu[1], Hiroko Yamauchi[1], Takao Himeno[1], Keijiro Ito[1], Ayumi Yamamoto[2], Ryota Kobayashi[2], Risa Mori[2], Aisaku Fukuda[2], Tomoko Inoue[3], Yoshiharu Morimoto[3]
*[1]IVF Namba Clinic, Japan, [2]IVF Osaka Clinic, [3]HORAC Grand Front Osaka Clinic*

Thu(5)-O38-5

**Preimplantation genetic diagnosis and natural conception: a comparison of live birth rates in patients with recurrent pregnancy loss associated with translocation**

Shinichiro Ikuma[1,2], Takeshi Sato[3], Mayumi Sugiura-Ogasawara[3], Takashi Yamaguchi[2], Tamito Miki[2], Motoi Nagayoshi[2], Atsushi Tanaka[2], Satoru Takeda[1]
*[1]Department of Obstetrics and Gynecology, Juntendo University Faculty of Medicine, Japan, [2]Saint Mother Obstetrics and Gynecology Hospital, [3]Department of Obstetrics and Gynecology, Nagoya City University, Graduate School of Medical Sciences*

Thu(5)-O38-6

**Genetic Counsellor's Preferences for Public Coverage of Preimplantation Genetic Diagnosis: A Discrete Choice Experiment**

Elaine S Goh[1,2], Wendy Ungar[1,2], Deborah Marshall[3], Fiona A Miller[1]
*[1]Institute of Health Policy, Management and Evaluation, University of Toronto, Canada, [2]Child Health Evaluative Sciences, The Hospital for Sick Children Research Institute, Toronto, Canada, [3]Department of Community Health Sciences, University of Calgary, Calgary, Canada*

9:45-11:15 Room A

## O39 Concurrent Oral Session 39
## "Prenatal, Perinatal and Reproductive Genetics 4"

Chairs: **Do Yeong Hwang**
*Department of OB & Gyn, Hamchoon Women's Clinic, Korea, South*
**Haruhiko Sago**
*Center of Maternal-Fetal, Neonatal and Reproductive Medicine, National Center for Child Health and Development, Japan*

Thu(5)-O39-1

**Risk level of intracytoplasmic sperm/spermatid injection for 116 non-mosaic Klinefelter syndrome (KS) patients**

Atsushi Tanaka[1], Motoi Nagayoshi[1], Shinichiro Ikuma[1], Tamito Miki[1], Takashi Yamaguchi[1], Izumi Tanaka[1], Youichi Takemoto[1], Hiroshi Kusunoki[2], Seiji Watanabe[3], Satoru Takeda[4]
*[1]Saint Mother Hospital, Japan, [2]Faunal Diversity Sciences, Graduate School of Agriculture, Kobe University, [3]Department of Anatomical Science, Hirosaki University Graduate school of Medicine, [4]Department of Obstetrics and Gynecology, Juntendo University School of Medicine*

Thu(5)-O39-2

### New candidate genes for NTD and CHD screened from a PiggyBac transgenic mice library have higher mutation rates in human NTD

**Yufang Zheng**[1,2,5], Zhongzhong Chen[1], Yingchun Jing[1], Zhiwen Shi[1], Shuxia Chen[1], Weiqi Liu[1], Jiaojiao Liu[4], Chunyan Wang[4], Hong Xu[4], Tian Xu[2], Ting Zhang[3], Xiaohui Wu[2], Hongyan Wang[1,5]

*[1]State Key Laboratory of Genetic Engineering and Ministry of Education (MOE) Key Laboratory of Contemporary Anthropology, School of Life Science, Fudan University, China, [2]The Institute of Developmental Biology and Molecular Medicine, Fudan University, [3]Capital Institute of Pediatrics, Beijing, China, [4]The children hospital of Fudan University, Shanghai, China, [5]8. bstetrics and Gynecology Hospital, Fudan University*

Thu(5)-O39-3

### Mutations in *CYP11B1* Gene of Vietnamese Patients with 11ß-hydroxylase Deficiency

**Mai T.P. Nguyen**[1,2]

*[1]Human genetics, National Hospital of Pediatrics, Vietnam, [2]Institute of Genome Research*

Thu(5)-O39-4

### Preimplantation Genetic Aneuploidy Screening At Ege University, Izmir, Turkey: 10 Years' Experience

**Burak Durmaz**[1], Emin Karaca[1], Ege N Tavmergen Goker[2], Erol Tavmergen[2], Nilufer Calimlioglu[2], Pelin Yasar[2], Cumhur Gunduz[3], Ferda Ozkinay[1]

*[1]Department of Medical Genetics, Ege University, Faculty of Medicine, Turkey, [2]Department of Obstetrics and Gynecology, IVF Unit, Ege University, Faculty of Medicine, [3]Department of Medical Biology, Ege University, Faculty of Medicine*

Thu(5)-O39-5

### Parental decisions on prenatally diagnosed chromosome abnormalities before 22 weeks of gestation: A Japanese multicenter retrospective study

**Miyuki Nishiyama**[1], Akihiko Sekizawa[2], Hiroaki Nakamura[3], Nobuhiro Suzumori[4], Setsuko Nakayama[5], Takahiro Yamada[6], Masaki Ogawa[7], Yukiko Katagiri[8], Yoko Okamoto[9], Akira Namba[10], Haruka Hamanoue[11], Masanobu Ogawa[12], Kiyonori Miura[13], Shunichiro Izumi[14], Yoshimasa Kamei[10], Haruhiko Sago[1]

*[1]Center of Maternal-Fetal, Neonatal and Reproductive Medicine, National Center for Child Health and Development, Japan, [2]Department of Obstetrics and Gynecology, Showa University School of Medicine, [3]Department of Obstetrics, Osaka City General Hospital, [4]Department of Obstetrics and Gynecology, Nagoya City University Graduate School of Medical Sciences, [5]Department of Obstetrics and Gynecology, Aiiku Clinic, [6]Department of Obstetrics and Gynecology, Hokkaido University Graduate School of Medicine, [7]Department of Obstetrics and Gynecology, Tokyo Women's Medical University, [8]Department of Obstetrics and Gynecology, Toho University Omori Medical Center, [9]Department of Obstetrics, Osaka Medical Center and Research Institute for Maternal and Child Health, [10]Department of Obstetrics and Gynecology, Saitama Medical University, [11]Department of Obstetrics and Gynecology, Yokohama City University Graduate School of Medicine, [12]Department of Obstetrics and Gynecology / Clinical Research Institute, National Kyusyu Medical Center, [13]Department of Obstetrics and Gynecology, Nagasaki University School of Medicine, [14]Department of Obstetrics and Gynecology, Tokai University School of Medicine*

Thu(5)-O39-6

### Isolation of mesenchymal stem cells derived from human placental tissue and their expression of C19MC microRNAs

**Naoki Fuchi**[1], Kiyonori Miura[1], Ai Higashijima[1], Tao-Sheng Li[2], Hideaki Masuzaki[1]

*[1]Department of Obstetrics and Gynecology, Nagasaki University Graduate School of Medicine, Japan, [2]Department of Stem Cell Biology, Atomic Bomb Disease Institute, Nagasaki University*

8:00-9:30 Room E

**O40** **Concurrent Oral Session 40**

## "Psychiatric Genetics, Neurogenetics and Neurodegeneration 3"

Chairs: **Jozef Gecz**
*Paediatrics, The University of Adelaide, Australia*
**Ryota Hashimoto**
*Molecular Research Center for Children's Mental Development, United Graduate School of Child Development, Osaka University, Japan*

Thu(5)-O40-1

### Exome Sequencing of Pakistani Consanguineous Families Identifies 31 Novel Candidate Genes for Recessive Intellectual Disability

**Hans van Bokhoven**[1], Saima Riazuddin[2], Mureed Hussain[1,3,4], Attia Razzaq[1,3,4], Zafar Iqbal[1], M Shahzad[2], Daniel Lopo Polla[1], Y Song[5], A A Khan[4], Joris A Veltman[1,6], Z M Khan[7], Detelina Grozeva[8], Karen Carrs[9], Tjitske Kleefstra[1], S A Riazuddin[10], Muhammad Ansar[1,3,4], F Lucy Raymond[8,9], S N Khan[4], Z M Ahmed[2], Arjan PM de Brouwer[1], Sheikh Riazuddin[3,4]
*[1]Human Genetics 855, Radboud University Medical Center, Netherlands, [2]Department of Otorhinolaryngology-Head & Neck Surgery, University of Maryland, Maryland, USA, [3]Allama Iqbal Medical College, University of Health Sciences, Pakistan, [4]National Center for Excellence in Molecular Biology, University of the Punjab, Pakistan, [5]Institute for Genome Sciences and Program in Personalized and Genomic Medicine, University of Maryland School of Medicine, USA, [6]Department of Clinical Genetics, GROW School for Oncology and Developmental Biology, Maastricht University Medical Centre, Maastricht, The Netherlands, [7]Shaheed Zulfiqar Ali Bhutto Medical University, Pakistan Institute of Medical Sciences, Pakistan, [8]Department of Medical Genetics, Cambridge Institute for Medical Research, University of Cambridge, Cambridge, United Kingdom, [9]Department of Haematology, University of Cambridge, Cambridge, United Kingdom, [10]The Wilmer Eye Institute, Johns Hopkins University School of Medicine, Baltimore, Maryland, USA*

Thu(5)-O40-2

### Protocadherin 19 (PCDH19) epilepsy, intellectual disability and autism limited to females

**Jozef Gecz**[1], Chuan Tan[1,2], Claire C Homan[1,3], Dale McAninch[3], Archa Fox[4], Daniel Pederick[3], Paul Q Thomas[3], Lachlan Jolly[1,2], Raman Kumar[1,2], Duyen Pham[1,2]
*[1]Paediatrics, The University of Adelaide, Australia, [2]Robinson Research Institute, The University of Adelaide, [3]School of Biological Sciences, The University of Adelaide, [4]University of Western Australia*

Thu(5)-O40-3

### Maternal Copy Number Variants (CNV) transmission to their Autism Spectrum Disorder (ASD) sons correlates with phenotypic traits

**Astrid Moura Vicente**[1,2,3,4], Muhammad Asif[1,2,3,5], Ines Conceicao[2,3,4], Katarzyna Kwiatkowska[2], Celia Rasga[2,3], Francisco Couto[1]
*[1]Faculdade de Ciencias da Universidade de Lisboa, Pakistan, [2]Instituto Nacional de Saude Doutor Ricardo Jorge, Lisboa, Portugal, [3]Biosytems and Integrative Sciences Institute, Lisboa, Portugal, [4]Instituto Gulbenkian de Ciencia, Oeiras, Portugal, [5]Department of Biosciences, COMSATS Institute of Information Technology, Sahiwal, Pakistan*

Thu(5)-O40-4

### Massively parallel sequencing in a case control cohort and extended families identifies noncoding risk variants for autism spectrum disorder

**Anthony J Griswold**[1], Holly N Cukier[1], Derek Van Booven[1], Eden R Martin[1,2], Michael L Cuccaro[1,2], John R Gilbert[1,3], Jonathan L Haines[3], John P Hussman[4], Margaret A Pericak-Vance[1,2]
*[1]John P. Hussman Insitute for Human Genomics, University of Miami, USA, [2]Dr. John T. Macdonald Department of Human Genetics, University of Miami, [3]Department of Epidemiology and Biostatistics, Case Western Reserve University, [4]Hussman Institute for Autism*

Thu(5)-O40-5

**Identification of rare risk variants in voltage-gated channel genes (*CACNA1C, CACNA1D, CACNA1S, CACNA1I*) in Japanese population of schizophrenia and autism spectrum disorder using Ion PGM platform**

**Chenyao Wang**, Hiroki Kimura, Jingrui Xing, Itaru Kushima, Branko Aleksic, Norio Ozaki
*Nagoya University, Japan*

Thu(5)-O40-6

**Systematic integration of brain eQTL and GWAS identifies *ZNF323* as a novel schizophrenia risk gene and suggests recent positive selection based on compensatory advantage on pulmonary function**

**Xiong-jian Luo**[1], Manuel Mattheisen[2], Ming Li[3], Liang Huang[4], Marcella Rietschel[5], Anders D Borglum[2], Thomas D Als[2], Edwin J van den Oord[6], Karolina A Aberg[6], Ole Mors[7], Preben Bo Mortensen[8], Zhenwu Luo[9], Franziska Degenhardt[10], Sven Cichon[11], Thomas G Schulze[12], Markus M Nothen[10], Bing Su[13], Zhongming Zhao[14], Lin Gan[15], Yong-gang Yao[16]
*[1]Genetic and Psychiatry, Kunming Institute of Zoology, Chinese Academy of Sciences, China, [2]Department of Biomedicine and Centre for Integrative Sequencing (iSEQ), Aarhus University, Denmark, [3]Lieber Institute for Brain Development, Johns Hopkins Medical Campus, Baltimore, MD, USA, [4]First Affiliated Hospital of Gannan Medical University, Ganzhou, China, [5]Department of Genetic Epidemiology in Psychiatry, Central Institute of Mental Health, Medical Faculty of Mannheim, University of Heidelberg, Mannheim, Germany, [6]Center for Biomarker Research and Personalized Medicine, Virginia Commonwealth University, [7]Centre for Psychiatric Research, Aarhus University Hospital, Risskov, Denmark, [8]National Centre for Register-based Research, Aarhus University, Aarhus, Denmark, [9]Department of Microbiology and Immunology, Medical University of South Carolina, Charleston, SC, USA, [10]Department of Genomics, Life & Brain Center, and Institute of Human Genetics, University of Bonn, Bonn, Germany, [11]Division of Medical Genetics, Department of Biomedicine, University Basel, Basel, Switzerland, [12]Department of Psychiatry and Psychotherapy, University Medical Center Georg-August-University, Goettingen, Germany, [13]State Key Laboratory of Genetic Resources and Evolution, Kunming Institute of Zoology, Chinese Academy of Sciences, Kunming, Yunnan, China, [14]Departments of Biomedical Informatics and Psychiatry, Vanderbilt University School of Medicine, Nashville, TN, USA, [15]Flaum Eye Institute and Department of Ophthalmology, University of Rochester, Rochester, NY, USA, [16]CAS Center for Excellence in Brain Science, Chinese Academy of Sciences, Shanghai, China*

9:45-11:15 Room E

## O41 Concurrent Oral Session 41
## "Psychiatric Genetics, Neurogenetics and Neurodegeneration 4"

Chairs: **Murim Choi**
*Department of Biomedical Sciences, Seoul National University College of Medicine, Korea, South*
**Kazuya Iwamoto**
*Department of Molecular Brain Science, Faculty of Life Sciences, Kumamoto University, Japan*

Thu(5)-O41-1

**Investigating the transcriptome wide impact of expanded polyalanine tract mutations in *ARX* contributing to intellectual disability and seizures**

**Tessa R Mattiske**[1], Kristie PY Lee[1,2], Jozef Gecz[1,2], Cheryl A Shoubridge[1,2]
*[1]School of Medicine, The University of Adelaide, Australia, [2]Robinson Research Institute, The University of Adelaide*

Thu(5)-O41-2

**Maternal Effects and Maternal Factors in OCD and Tourette Disorder**

**Dorothy E Grice**[1], Heidi A Browne[1], Amirhossein Modabbernia[1], Sven T Sandin[1], Eric T Parner[2], Christina Hultman[3], Diana E Schendel[2], Joseph D Buxbaum[1], Avi Reichenberg[1]
*[1]Department of Psychiatry, Icahn School of Medicine at Mount Sinai, New York, NY, USA, [2]Aarhus University, Denmark, [3]Karolinska Institute, Sweden*

Thu(5)-O41-3

### Genome-Wide Analysis of Attention-Deficit/Hyperactivity Disorder in Korean Children

**Hyo-Won Kim**[1], Kukju Kweon[1], Eun-Soon Shin[2], Yeonho Joo[1]
*[1]Department of Psychiatry, University of Ulsan College of Medicine, Asan Medical Center, Korea, South, [2]DNA Link, Inc. Bioinformatics*

Thu(5)-O41-4

### An Ultraconserved Brain-specific Transcriptional Enhancer within the *ADGRL3 (LPHN3)* Gene Underpins ADHD Susceptibility

**Ariel F Martinez**[1], Yu Abe[1], Sung-Kook Hong[1], Kevin Molyneux[1], David Yarnell[1], Heiko Lohr[2], Wolfgang Driever[2], Mauricio Arcos-Burgos[3], Maximilian Muenke[1]
*[1]Medical Genetics Branch, National Institutes of Health, USA, [2]Institute of Biology I, Faculty of Biology, University of Freiburg, [3]John Curtin School of Medical Research, The Australian National University*

Thu(5)-O41-5

### Autistic MeCP2 mutations lost regulation on miR197/ADAM10/NOTCH and affected neural progenitor cells differentiation

**Hongyan Wang**[1,3], Yufang Zheng[1,2], Yumeng Wang[1,2], Yahui Liu[1], Zhangmin Yang[5], Yanqing He[5], Xiaohong Gong[1], Bing Su[6], Keping Hu[7], Zilong Qiu[8], Dong Liu[9], Yasong Du[4]
*[1]State Key Laboratory of Genetic Engineering and Ministry of Education (MOE) Key Laboratory of Contemporary Anthropology, School of Life Science, Fudan University, China, [2]Institute of Developmental Biology & Molecular Medicine, Fudan University, [3]The Obstetrics & Gynecology Hospital of Fudan University, [4]Shanghai Mental Health Center, Shanghai Jiaotong University, [5]Shanxi Normal University School of Life Sciences, [6]Kunming Institute of Zoology, Chinese Academy of Sciences, [7]The Institute of Medicinal Plant Development, Chinese Academy of Medical Sciences, Beijing, [8]The Institute of Neuroscience, Chinese Academy of Sciences, Shanghai, [9]Nantong University, Jiangsu, China*

Thu(5)-O41-6

### A recurrent mutation in γ-aminobutyric acid type B (GABAB) receptor R2 causes a Rett-like phenotype

**Murim Choi**[1], Yongjin Yoo[1], Jane Jung[2], Yuna Lee[3], Youngha Lee[1], Hyosuk Cho[1], Jin S Lee[4], Je S Lee[5], Chansik Hong[6], Sang-Yoon Park[7], Jinhong Wie[6], Ki J Kim[4], Yong S Hwang[4], Seok-Geun Lee[7], Hee-Jung Choi[8], Insuk So[6], Byung C Lim[4], Jae Y Sung[3], Hosung Jung[2], Yong B Shin[5], Jong-Hee Chae[4]
*[1]Department of Biomedical Sciences, Seoul National University, Korea, South, [2]Department of Anatomy, Brain Research Institute, and Brain Korea 21 PLUS Project for Medical Science, Yonsei University College of Medicine, [3]Graduate School of Medicine, Korea University, [4]Department of Pediatrics, Seoul National University College of Medicine, Seoul National University Children's Hospital, [5]Department of Rehabilitation Medicine, Pusan National University College of Medicine, [6]Department of Physiology, Seoul National University College of Medicine, [7]Department of Science in Korean Medicine, Cancer Preventive Material Developmental Research Center, College of Korean Medicine, Kyung Hee University, [8]Department of Biological Sciences, Seoul National University College of Natural Sciences*

8:00-9:30 Room B-1

## O42 Concurrent Oral Session 42
## "Clinical Genetics and Dysmorphology 4"

Chairs: **Reha M. Toydemir**
*Pathology, University of Utah, USA*
**Hiroshi Kawame**
*Division of Genomic Medicine Support and Genetic Counseling, Tohoku University, Japan*

Thu(5)-O42-1

### MICRODELETION OF 12q14.2q14.3 IN THREE MEMBERS OF A FAMILY DETECTED BY SNP-ARRAY ANALYSIS

Rita Fischetto[1,5], Orazio Palumbo[2], Federica Ortolani[1], Pietro Palumbo[2], Maria Pia Leone[2,3], Maria Cristina Di Gilio[4], Leopoldo Zelante[2], Massimo Carella[2], Francesco Papadia[1]
*[1]U.O.C. Malattie Metaboliche-Genetica-Medica, A.O.U. Policlinico Consorziale Bari, Italy, [2]Laboratorio di Genetica Medica, IRCSS Casa Sollievo della Sofferenza, S.Giovanni Rotondo, Italy., [3]Dipartimento di Scienze del suolo, della pianta e degli alimenti, Università degli Studi di Bari "Aldo Moro", Italy, [4]U.O.S Genetica Medica, Ospedale Bambin Gesù, Roma, Italy, [5]Istituto Biologia e Genetica; Medicina e Chirugia; Università degli Studi di Bari*

Thu(5)-O42-2

### Expressive Language Delay and Characteristic Facial Features - A Novel 7p22.3p22.2 Microdeletion Syndrome?

Andrea C Yu[1], Regina M Zambrano[2], Ingrid Cristian[3], Sue Price[4], Christine Armour[1,5]
*[1]Department of Genetics, Children's Hospital of Eastern Ontario, Canada, [2]Division of Clinical Genetics, Department of Pediatrics, Louisiana State University Health Science Center, [3]Division of Genetics and Metabolism, Department of Pediatrics, Nemours Children's Hospital Orlando, [4]Department of Clinical Genetics, Northampton General Hospital, [5]Children's Hospital of Eastern Ontario Research Institute*

Thu(5)-O42-3

### Delineation of the 9q31 microdeletion syndrome

Reha M Toydemir[1,3], Emanuele Panza[2], Sarah L Dugan[3], Lorenzo D Botto[3]
*[1]Pathology, University of Utah, USA, [2]Human Genetics, University of Utah, [3]Pediatrics, University of Utah*

Thu(5)-O42-4

### Noonan syndrome and related disorders associated with coloboma: five case reports and review of literature

Yline Capri[1], Hend Dridi[1], Fabien Guimiot[2,3], Delphine Heron[4], Marianne Till[5], Nicole Philip[6], Helene Dollfus[7,8], Liza Vera[9], Helene Cave[1], Alain Verloes[1,3]
*[1]Clinical Genetics, CHU Robert Debre, France, [2]Foetopathology, CHU Robert Debre, [3]Paris VII University, INSERM UMR1141, [4]Medical genetics, La Pitie-Salpetriere, [5]Cytogenetics, CHU Lyon, [6]Medical genetics, CHU Marseille, [7]Strabourg University, INSERM EA3949, [8]Medical Genetics, CARGO, CHU Strasbourg, [9]Ophtalmology, CHU Robert Debre*

Thu(5)-O42-5

### Sex chromosomal Abnormalities in Egyptian DSD patients

Inas M Mazen[1], Mona M Mekkawi[2], Alaa K Kamel[2], Aya A Elaidy[1]
*[1]Clinical Genetics and Endocrinology, National Research Centre, Egypt, [2]Medical Cytogenetics, National Research Centre*

Thu(5)-O42-6

**Male-to-female (XY) sex reversal and systemic lupus erythematosis: Association of functional Xp disomy including *DAX-1* and *TLR7***

**Rie Kawakita**[1,2], Azumi Sakakibara[1], Yukiko Hashimoto[1], Yuki Hosokawa[1], Rika Fujimaru[1], Nobuyoshi Tamagawa[2], Hiroaki Nakamura[2], Tohru Yorifuji[1,2]
*[1]Department of Pediatric Endocrinology and Metabolism, Osaka City General Hospital, Japan, [2]Department of Genetic Medicine, Osaka City General Hospital*

9:45-11:15 Room B-1

## O43 Concurrent Oral Session 43
## "Clinical Genetics and Dysmorphology 5"

Chairs: **Hsiang-Yu Lin**
*Department of Pediatrics, Mackay Memorial Hospital, Taipei, Taiwan*
**Tomoki Kosho**
*Department of Medical Genetics, Shinshu University School of Medicine, Japan*

Thu(5)-O43-1

**Clinical and Molecular Characterisation of Frontonasal Dysplasia**

**Patrick JJ Yap**[1], Stefanie Eggers[2,3], David J Amor[1,3], George McGillivray[1], Kate Pope[1,2], Martin Delatycki[2,4], Matthew Hunter[5,6], Naomi Baker[2,3], Peter Farlie[2,3], Tiong Y Tan[1,2,3]
*[1]Victorian Clinical Genetics Services, Murdoch Children's Research Institute, Australia, [2]Murdoch Children's Research Institute, Royal Children's Hospital, Melbourne, Australia, [3]Department of Paediatrics, University of Melbourne, Melbourne, Australia, [4]Department of Clinical Genetics, Austin Health, Heidelberg, Australia, [5]Monash Genetics, Monash Medical Centre, Clayton, Australia, [6]Dept of Paediatrics, Monash University, Clayton, Australia*

Thu(5)-O43-2

**Co-occurrence of Sturge-Weber syndrome phenotype and Klippel-Trenaunay-Weber syndrome phenotype in a patient: Molecular evidence of the shared pathological basis of the two conditions**

**Yuri Sakaguchi**[1], Toshiki Takenouchi[1,2], Takao Takahashi[1], Kenjiro Kosaki[2]
*[1]Department of Pediatrics, Keio University School of Medicine, Japan, [2]Center for Medical Genetics, Keio University School of Medicine*

Thu(5)-O43-3

**CDC42 as a new human disease causative gene**

**Tomoko Uehara**[1], Nobuhiko Okamoto[2], Toshiki Takenouchi[1,3], Shinobu Ida[4], Kenjiro Kosaki[1]
*[1]Center for Medical Genetics, Keio University School of Medicine, Japan, [2]Department of Medical Genetics, Osaka Medical Center and Research Institute for Maternal and Child Health, [3]Department of Pediatrics, Keio University School of Medicine, [4]Department of Gastroenterology and Endocrinology, Osaka Medical Center and Research Institute for Maternal and Child Health*

Thu(5)-O43-4

**Two novel mutations in the *FUCA1* gene causing fucosidosis**

**Wipa Panmontha**, Ponghatai Damrongphol, Tayard Desudchit, Vorasuk Shotelersuk, Kanya Suphapeetiporn
*Chulalongkorn University, Thailand*

Thu(5)-O43-5

### Ocular Features in Patients with Mucopolysaccharidosis

**Hsiang-Yu Lin**[1,2,3,4], Chih-Kuang Chuang[2], Wei-Chun Chan[5], Dau-Ming Niu[6], Pao Chin Chiu[7], Wen-Hui Tsai[8], Wuh-Liang Hwu[9], Shuan-Pei Lin[1,2,3,4,10]

*[1]Department of Pediatrics, Mackay Memorial Hospital, Taiwan, [2]Department of Medical Research, Mackay Memorial Hospital, Taipei, Taiwan, [3]Department of Medicine, Mackay Medical College, New Taipei City, Taiwan, [4]Mackay Junior College of Medicine, Nursing and Management, Taipei, Taiwan, [5]Department of Ophthalmology, Mackay Memorial Hospital, Taipei, Taiwan, [6]Department of Pediatrics, Taipei Veterans General Hospital, Taipei, Taiwan, [7]Department of Pediatrics, Kaohsiung Veterans General Hospital, Kaohsiung, Taiwan, [8]Department of Pediatrics, Chi Mei Medical Center, Tainan, Taiwan, [9]Department of Pediatrics, National Taiwan University Hospital, Taipei, Taiwan, [10]Department of Infant and Child Care, National Taipei University of Nursing and Health Sciences, Taipei, Taiwan*

Thu(5)-O43-6

### Two-dimensional Speckle Tracking Echocardiography in 53 Patients with Mucopolysaccharidosis

**Hsiang-Yu Lin**[1,2,3,4,5], Chih-Kuang Chuang[2], Ming-Ren Chen[1,3,4], Dau-Ming Niu[5,6], Chung-Lieh Hung[3,4,7], Shuan-Pei Lin[1,2,3,4,8]

*[1]Department of Pediatrics, Mackay Memorial Hospital, Taiwan, [2]Department of Medical Research, Mackay Memorial Hospital, Taipei, Taiwan, [3]Department of Medicine, Mackay Medical College, New Taipei City, Taiwan, [4]Mackay Junior College of Medicine, Nursing and Management, Taipei, Taiwan, [5]Institute of Clinical Medicine, National Yang-Ming University, Taipei, Taiwan, [6]Department of Pediatrics, Taipei Veterans General Hospital, Taipei, Taiwan, [7]Division of Cardiology, Department of Internal Medicine, Mackay Memorial Hospital, Taipei, Taiwan, [8]Department of Infant and Child Care, National Taipei University of Nursing and Health Sciences, Taipei, Taiwan*

8:00-9:30 Room B-2

## O44 Concurrent Oral Session 44
## "Complex Traits and Polygenic Disorders 4"

Chairs: **Andrew P. Morris**
*Department of Bioinfomatics, University of Liverpool, UK*
**Akira Hata**
*Department of Public Health, Chiba University Graduate School of Medicine, Japan*

Thu(5)-O44-1

### Trans-ethnic meta-analysis and genomic annotation reveals novel loci and effector genes for kidney function in diverse populations

**Andrew P Morris**[1,2], Anubha Mahajan[2], Kyle Gaulton[2], Jeffrey Haessler[3], Yukinori Okada[4], Adrienne Stilp[5], John Whitfield[6], Cathy Laurie[5], Nora Franceschini[7]

*[1]Department of Biostatistics, University of Liverpool, UK, [2]Wellcome Trust Centre for Human Genetics, University of Oxford, [3]Public Health Sciences Division, Fred Hutchinson Cancer Research Center, [4]Graduate School of Medical and Dental Sciences, Tokyo Medical and Dental University, [5]Department of Biostatistics, University of Washington, [6]QIMR Berghofer Medical Research Institute, [7]Department of Epidemiology, University of North Carolina*

Thu(5)-O44-2

### MicroRNA Transcriptome Changes in Multiple Brain Regions of Subjects with Alcohol Use Disorders

**Huiping Zhang**[1], Hongyu Zhao[2], Joel Gelernter[1]

*[1]Psychiatry, Yale University School of Medicine, USA, [2]Biostatistics, Yale University School of Medicine*

Thu(5)-O44-3

**Targeted-bisulfite sequence analysis of the methylation of CpG islands in the *PNPLA3*, *SAMM50*, and *PARVB* of patients with nonalcoholic fatty liver disease: relationship to their mRNA expression and rs738409 genotype**

Kikuko Hotta[1], Yuji Ogawa[2], Yasushi Honda[2], Kento Imajo[2], Satoru Saito[2], Masato Yoneda[2], Atsushi Nakajima[2]

*[1]Department of Medical Innovation, Osaka University Hospital, Japan, [2]Department of Gastroenterology and Hepatology, Yokohama City University Graduate School of Medicine*

Thu(5)-O44-4

**Genome-wide multi-phenotype and eQTL analyses detect novel signals for omega fatty acids and provide insights into their biology**

Annique J. Claringbould[1,2], Fiona Haagenbeek[2,3], Reedik Magi[4], Pasi Soininen[5], Marjo-Riitta Jarvelin[6,7,8,9], BIOS Consortium[1], Marika Kaakinen[2], Inga Prokopenko[2]

*[1]Department of Genetics, University Medical Centre Groningen, Netherlands, [2]Department of Genomics of Common Disease, Imperial College London, United Kingdom, [3]Department of Biological Psychology, VU University Amsterdam, The Netherlands, [4]Estonian Genome Center, University of Tartu, Estonia, [5]Computational Medicine, University of Oulu, Finland, [6]Center for Life Course Epidemiology and Systems Medicine, University of Oulu, Finland, [7]Department of Epidemiology and Biostatistics, Imperial College London, UK, [8]Biocenter Oulu, University of Oulu, Finland, [9]Unit of Primary Care, Oulu University Hospital, Finland*

Thu(5)-O44-5

**Genome-wide study for circulating metabolites identifies 62 loci and reveals novel systemic effects for *LPA***

Johannes Kettunen[1,2,3], MAGNETIC Consortium

*[1]Computational medicine, University of Oulu, Finland, [2]National Institute for Health and Welfare, Helsinki, Finland, [3]NMR Metabolomics Laboratory, School of Pharmacy, University of Eastern Finland, Kuopio, Finland*

Thu(5)-O44-6

**Exome chip meta-analysis identifies novel low-frequency variants contributing to central body fat distribution**

Tugce Karaderi[1], Anne E Justice[2], Kristin L Young[2,3], Heather M Highland[2], Mariaelisa Graff[2], Valerie Turcot[4], Paul Auer[5], Nancy L Heard-Costa[6,7], Claudia Schurmann[8], Yingchang Lu[8], L Addriene Cupples[6,9], Caroline S Fox[6], Thomas W Winkler[10], Niels Grarup[11], Robert A Scott[12], Mark McCarthy[13], Karen Mohlke[14], Ruth JF Loos[8], Ingrid Borecki[15], Kari E North[2], Cecilia M Lindgren[1], on the behalf of BBMRI, GOT2D, CHARGE and GIANT Consortia

*[1]Wellcome Trust Centre for Human Genetics, University of Oxford, UK, [2]Department of Epidemiology, University of North Carolina at Chapel Hill, USA, [3]Carolina Population Center, University of North Carolina at Chapel Hill, USA, [4]Montreal Heart Institute, University of Montreal, Canada, [5]Department of Biostatistics, University of Wisconsin-Milwaukee, USA, [6]The Framingham Heart Study, National Heart, Lung, and Blood Institute, USA, [7]Department of Neurology, Boston University School of Medicine, USA, [8]The Genetics of Obesity and Related Metabolic Traits Program, The Charles Bronfman Institute for Personalized Medicine, Icahn School of Medicine at Mount Sinai, USA, [9]Department of Biostatistics, School of Public Health, Boston University, USA, [10]Department of Genetic Epidemiology, Institute of Epidemiology and Preventive Medicine, University of Regensburg, Regensburg, Germany, [11]The Novo Nordisk Foundation Center for Basic Metabolic Research, University of Copenhagen, Denmark, [12]MRC Epidemiology Unit, University of Cambridge, UK, [13]Oxford Centre for Diabetes Endocrinology and Metabolism, University of Oxford, UK, [14]Department of Genetics, University of North Carolina at Chapel Hill, USA, [15]Department of Genetics Division of Statistical Genomics, Washington University School of Medicine, USA*

9:45-11:15 Room B-2

## O45 Concurrent Oral Session 45
## "Complex Traits and Polygenic Disorders 5"

Chairs: **Derek M. Dykxhoorn**
*John P. Hussman Institute for Human Genomics, Universty of Miami Miller School of Medicine, USA*
**Michiaki Kubo**
*Center for Integrative Medical Sciences, RIKEN, Japan*

Thu(5)-O45-1

### Pathogen lineage based analysis of host genetic risk factor in young onset tuberculosis

**Yosuke Omae**[1], Surakameth Mahasirimongkol[2], Licht Toyo-oka[1], Hideki Yanai[3], Supalert Nedsuwan[4], Sukanya Wattanapokayakit[2], Nat Smittipat[5], Prasit Paliittapongarnpim[5], Pathom Sawanpanyalert[6], Nuanjun Wichukchinda[2], Ekawat Pasomsub[5], Taisei Mushiroda[7], Michiaki Kubo[8], Katsushi Tokunaga[1]
*[1]Faculty of Medicine, The University of Tokyo, Japan, [2]Medical Genetics Center, Medical Life Sciences institute, Department of Medical Sciences, Ministry of Public Health, Thailand, [3]Fukujuji Hospital, Japan Anti-tuberculosis Association, Kiyose, Japan, [4]Chaing Rai Prachanukroh Hospital, Ministry of Public Health, Thailand, [5]Department of Microbiology, Faculty of Science, Mahidol University, Thailand, [6]Food and Drug Administration, Ministry of Public Health, Thailand, [7]Research Group for Pharmacogenomics, RIKEN Center for Integrative Medical Sciences, Yokohama, Japan, [8]RIKEN Center for Integrative Medical Sciences, Yokohama, Japan*

Thu(5)-O45-2

### Transcriptome analysis reveals autism-specific convergent molecular pathways during neurogenesis

**Derek M. Dykxhoorn**[1,2], Brooke A. DeRosa[1], Kinsley Belle[1], Catherine Garcia-Serje[1], Holly N. Cukier[1], Joycelyn M. Lee[1], Michael L. Cuccaro[1,2], Jeffery M. Vance[1,2], Margaret A. Pericak-Vance[1,2]
*[1]John P. Hussman Institute for Human Genomics, Universty of Miami Miller School of Medicine, USA, [2]Dr. John T. Macdonald Foundation Department of Human Geneics, University of Miami Miller School of Medicine*

Thu(5)-O45-3

### Analysis of the planar cell polarity regulator gene *PTK7* in neural tube defects

**Richard H Finnell**[1,2], Gary M Shaw[3], Elizabeth Ross[4]
*[1]Nutritional Sciences and Chemistry, The University of Texas at Austin, USA, [2]Fudan University, [3]Stanford University School of Medicine, [4]Weill Cornell Medical College*

Thu(5)-O45-4

### Rare variants in the *COL5A1* gene are associated with risk for keratoconus, a blinding eye disease

**Kathryn P Burdon**[1], Sionne EM Lucas[1], Richard A Mills[2], Nicholas B Blackburn[1,3], Paul Leo[4], Jac C Charlesworth[1], Matthew A Brown[4], Jamie E Craig[2]
*[1]Menzies Institute for Medical Research, University of Tasmania, Australia, [2]Department of Ophthalmology, Flinders University, [3]South Texas Diabetes and Obesity Institute, University of Texas Rio Grande Valley, [4]Diamantina Institute, University of Queensland and Translational Research Institute, Princess Alexandra Hospital*

Thu(5)-O45-5

### Examining the Genetic Architecture of Age-related Macular Degeneration (AMD) in the Amish

**Jonathan L Haines**[1], Rebecca J Sardell[2], Joshua Hoffman[1], Jessica N Cooke Bailey[1], Srinivas R Sadda[3], William K Scott[2], Dwight Stambolian[4], Margaret A Pericak-Vance[2]
*[1]Epidemiology & Biostatistics, Case Western Reserve University, USA, [2]Hussman Institute for Human Genomics, Miller School of Medicine, University of Miami, [3]Department of Ophthalmology, Doheny Eye Institute, [4]Departments of Ophthalmology and Genetics, University of Pennsylvania*

Thu(5)-O45-6

**Familial insight: Identifying glaucoma susceptibility variants by exome sequencing in extended pedigrees**

**Jac Charlesworth**[1], Kathryn Burdon[1], Juan Peralta[2], Nicholas Blackburn[1,2], Joanne Curran[2], Mary Wirtz[3], David Mackey[4], John Blangero[2]
*[1]University of Tasmania, Menzies Institute for Medical Research, Australia, [2]South Texas Diabetes and Obesity Institute, University of Texas Rio Grande Valley, USA, [3]Casey Eye Institute, Oregon Health and Science University, USA, [4]Lions Eye Institute, University of Western Australia*

8:00-9:30 Room C-1

## O46 Concurrent Oral Session 46
## "Molecular Basis of Mendelian Disorders 4"

Chairs: **Christian T. Thiel**
*Institute of Human Genetics, Friedrich-Alexander University of Erlangen-Nuremberg, Germany*
**Tadashi Kaname**
*Genome Medicine, National Center for Child Health and Development, Japan*

Thu(5)-O46-1

**Identifying a splice site mutation in *RAB3GAP1* in Martsolf Syndrome by whole exom sequencing and revealing the function of the novel mutation**

**Mustafa Ozen**[1,2,3], Asuman Koparir[2], Omer F. Karatas[3,4], Emre Kirat[2], Seda S. Yılmaz[2], Bugra Ozer[5], Betul Yuceturk[2,5], Mahmut S. Sagiroglu[5], Adnan Yuksel[1]
*[1]Department of Medical Genetics, Biruni University, Istanbul, Turkey, [2]Department of Medical Genetics, Istanbul University Cerrahpasa Medical School, Istanbul, Turkey, [3]Department of Pathology and Immunology, Baylor College of Medicine, Houston, TX, USA, [4]Molecular Biology and Genetics Department, Erzurum Technical University, Erzurum, Turkey, [5]Advanced Genomics and Bioinformatics Research Center, The Scientific and Technological Research Council of Turkey (TUBITAK-BILGEM), Kocaeli, Turkey*

Thu(5)-O46-2

**Adult mice expressing a Braf Q241R mutation on an ICR/CD-1 background exhibit a cardio-facio-cutaneous syndrome phenotype**

**Shin-ichi Inoue**[1], Mitsuji Moriya[1,2], Sachiko Miyagawa-Tomita[3], Yasumi Nakashima[4], Daiju Oba[1], Tetsuya Niihori[1], Misato Hashi[5], Hiroshi Ohnishi[5], Shigeo Kure[2], Yoichi Matsubara[1,6], Yoko Aoki[1]
*[1]Department of Medical Genetics, Tohoku University School of Medcine, Japan, [2]Department of Pediatrics, Tohoku University School of Medicine, [3]Department of Veterinary Technology, Yamazaki gakuen University, [4]Department of Pediatrics, Seirei Hamamatsu General Hospital, [5]Department of Laboratory Sciences, Gunma University Graduate School of Health Sciences, [6]National Research Institute for Child Health and Development*

Thu(5)-O46-3

**Massively parallel sequencing of a targeted panel for the diagnosis of Disorders of Sex Development**

**Andrew H Sinclair**, Stefanie Eggers
*Molecular Development, Murdoch Children's Research Institute, Australia*

Thu(5)-O46-4

### Systematic evaluation of patients with idiopathic short stature using whole exome sequencing

**Christian T Thiel**[1], Nadine N Hauer[1], Sarah Schuhmann[1], Eva Schoeller[1], Marie T Wittmann[1], Steffen Uebe[1], Arif B Ekici[1], Heinrich Sticht[2], Helmuth-Guenther Doerr[3], Andé Reis[1]

*[1]Institute of Human Genetics, Friedrich-Alexander-University of Erlangen-Nuremberg, Germany, [2]Institute of Biochemistry Friedrich-Alexander-University of Erlangen-Nuremberg, [3]Department of Pediatrics and Adolescent Medicine Friedrich-Alexander-University of Erlangen-Nuremberg*

Thu(5)-O46-5

### Novel candidate gene for congenital alveolar proteinosis with hypogammaglobulinemia identified by whole exome sequencing analysis

**Kazutoshi Cho**[1], Takuma Akimoto[1], Itaru Hayasaka[1], Hisanori Minakami[1], Tadashi Ariga[2], Masafumi Yamada[2], Masahiro Ueki[2], Naomichi Matsumoto[3], Noriko Miyake[3], Atsushi Fujita[3], Hirokazu Kanegane[4], Satoshi Miyamoto[4], Satoru Ikemoto[5], Kazunaga Agamatsu[6], Norimoto Kobayashi[6]

*[1]Maternity and Perinatal Care Center, Hokkaido University Hospital, Japan, [2]Department of Pediatrics, Hokkaido University Graduate School of Medicine, Sapporo, Japan, [3]Department of Human Genetics, Yokohama City University Graduate School of Medicine, Yokohama, Japan, [4]Department of Pediatrics and Developmental Biology, Tokyo Medical and Dental University, Tokyo, Japan, [5]Division of General Pediatrics, Saitama Children's Medical Center, Saitama, Japan, [6]Department of Pediatrics, Shinshu University, School of Medicine, Nagano, Japan*

Thu(5)-O46-6

### Whole exome sequencing identifies homozygous mutation in *ERCC1* in three sibling with a complex phenotypic disorder

**Zeynep Ocak**[1], Tulay Ozlu[2], Tarik Ocak[1], Yavuz Bayram[3], Davut Pehlivan[3,4], Ender Karaca[3], Richard A. Gibbs [5], James R. Lupski[3,5,6,7]

*[1]Medical Genetics, Kanuni Sultan Suleyman Research and Training, Turkey, [2]Department of Obstetrics and Gynecology, Abant izzet Baysal University Medical Faculty, Bolu, Turkey., [3]Department of Molecular and Human Genetics, Baylor College of Medicine, Houston, TX, USA, [4]Section of Neurology, Department of Pediatrics, Baylor College of Medicine, One Baylor, [5]Human Genome Sequencing Center, Baylor College of Medicine, Houston, TX, USA, [6]Department of Pediatrics, Baylor College of Medicine, Houston, TX, USA, [7]Texas Childrens Hospital, Houston, TX, USA*

9:45-11:15 Room C-1

## O47 Concurrent Oral Session 47
## "Molecular Basis of Mendelian Disorders 5"

Chairs: **Vanessa Sancho-Shimizu**
*Department of Virology and Paediatrics, Imperial College London, UK*
**Yoko Aoki**
*Department of Medical Genetics, Tohoku University School of Medicine, Japan*

Thu(5)-O47-1

### Study on molecular mechanism of episodic pain with $Na_v1.9$ channel mutations

**Jing Yu Liu**[1], Luyao Yang[1], Xiangyang Zhang[1], Jingmin Wen[1], Wei Yang[2], Cheng Wang[1], Lunan Gao[1], Junyu Luo[3], Jing Yao[4], Xue Zhang[2]

*[1]Key Laboratory of Molecular Biophysics of the Ministry of Education, School of Life Science and Technology, Huazhong University of Science and Technology, China, [2]McKusick-Zhang Center for Genetic Medicine and State Key Laboratory of Medical Molecular Biology, Institute of Basic Medical Sciences, Chinese Academy of Medical Sciences & Peking Union Medical College, [3]School of Chemical Engineering and Pharmacy, Wuhan Institute of Technology, [4]College of Life Sciences, Wuhan University*

Thu(5)-O47-2

**Assembling the complex immune region haplotypes using Long Read Single Molecule Real-Time Sequencing**

**Swati S Ranade**[1], Richard Hall[1], Kevin Eng[1], Chul-woo Pyo[2], Dave Roe[3], Primo Baybayan[1], Lawrence Hon[1], Daniel E Geraghty[2], Cynthia Vierra-Green[3], Steve Kujawa[1], Martin Maiers[3]
*[1]Pacific Biosciences, USA, [2]Fred Hutchinson Cancer Research Center, Seattle, USA, [3]Center for International Blood and Marrow Transplant Research, Minneapolis, USA*

Thu(5)-O47-3

**Mutations in *MECOM*, encoding oncoprotein EVI1, cause radioulnar synostosis with amegakaryocytic thrombocytopenia**

**Tetsuya Niihori**[1], Meri Ouchi-Uchiyama[2,3], Yoji Sasahara[2], Takashi Kaneko[4], Yoshiko Hashii[5], Masahiro Irie[2,3], Atsushi Sato[3], Yuka Saito-Nanjo[2,3], Ryo Funayama[6], Takeshi Nagashima[6], Shin-ichi Inoue[1], Keiko Nakayama[6], Keiichi Ozono[5], Shigeo Kure[2], Yoichi Matsubara[1,7], Masue Imaizumi[3], Yoko Aoki[1]
*[1]Department of Medical Genetics, Tohoku University School of Medicine, Japan, [2]Department of Pediatrics, Tohoku University School of Medicine, [3]Department of Hematology and Oncology, Miyagi Children's Hospital, [4]Department of Hematology-Oncology, Tokyo Metropolitan Children's Medical Center, [5]Department of Pediatrics, Osaka University Graduate School of Medicine, [6]Division of Cell Proliferation, United Centers for Advanced Research and Translational Medicine, Tohoku University Graduate School of Medicine, [7]National Research Institute for Child Health and Development*

Thu(5)-O47-4

**Beta-globin haplotypes in Hemoglobin E and normal individuals from seven minority groups of Yunnan province, China**

**Zhaoqing Yang**, Hongxian Liu, Kai Huang, Shuyan Liu, Hao Sun, Keqin Lin, Xiaoqin Huang, Jiayou Chu
*Institute of Medical Biology, Chinese Academy of Medical Sciences, China*

Thu(5)-O47-5

**Heterozygous mutations in *NFKB1* cause immunodeficiency and autoinflammatory episodes**

**Meri Kaustio**[1], Emma Haapaniemi[2,3], Helka Nurkkala[4], Giljun Park[5], Elisabet Einarsdottir[3,6], Fitsum Tamene[4], Luca Trotta[1], Ekaterina Morgunova[3], Kaarel Krjutskov[3], Jaana Syrjanen[7], Anssi Lagerstedt[8], Merja Helminen[9], Timi Martelius[10], Timo Hautala[11], Satu Mustjoki[5,12], Janna Saarela[1], Juha Kere[2,3,6], Markku Varjosalo[4], Mikko Seppanen[10,13]
*[1]Institute for Molecular Medicine Finland, University of Helsinki, Helsinki, Finland, [2]Folkhalsan Institute of Genetics, Helsinki, Finland, [3]Department of Biosciences and Nutrition, Karolinska Institutet, Stockholm, Sweden, [4]Institute of Biotechnology, University of Helsinki, Helsinki, Finland, [5]Hematology Research Unit Helsinki, Department of Clinical Chemistry and Hematology, University of Helsinki, Helsinki, Finland, [6]Center for Innovative Medicine, Karolinska Institutet, Stockholm, Sweden, [7]Department of Internal Medicine, Tampere University Hospital, Tampere, Finland, [8]Fimlab Laboratories, Tampere University Hospital, Tampere, Finland, [9]Tampere Center for Child Health Research, Tampere University Hospital, Tampere, Finland, [10]Adult Immunodeficiency Unit, Infectious Diseases, Inflammation Center, University of Helsinki and Helsinki University Hospital, Helsinki, Finland, [11]Department of Internal Medicine, Oulu University Hospital, Oulu, Finland, [12]Helsinki University Central Hospital Comprehensive Cancer Center, Helsinki, Finland, [13]Rare Diseases Center, Children's Hospital, University of Helsinki and Helsinki University Hospital, Helsinki, Finland*

Thu(5)-O47-6

**Whole exome sequencing of an extended family with invasive meningococcal disease**

**Vanessa Sancho Shimizu**[1], Alberto Lopez-Lera[2,3], Evangelos Bellos[4], Bayarchimeg Mashabt[1], Heidi Makrinioti[5,6], Ross P Walton[5,6], Margarita Lopez-Trascasa[2,3], Michael Levin[1]
*[1]Dept of Paediatrics and Virology, Imperial College London, UK, [2]Immunology Unit, Hospital Universitario La Paz and Hospital La Paz Research Institute (IdiPAZ), Madrid, Spain, [3]Centre for Biomedical Network Research on Rare Diseases (CIBERER), Instituto de Salud Carlos III (ISCIII), Madrid, Spain, [4]The Department of Genomics of Common Disease, School of Public Health, Imperial College London, UK, [5]Airway Disease Infection Section, National Heart and Lung Institute, Imperial College London, UK. Medical Research Council (MRC) and Asthma UK Centre in Allergic Mechanisms of Asthma, London UK, [6]Centre for Respiratory Infection, Imperial College London, UK*

8:00-9:30 Room C-2

## O48 Concurrent Oral Session 48
## "Metabolic Disorders 3"

Chairs: **Roberto Giugliani**
*Department of Genetics, Federal University of Rio Grande Do Sul, Brazil*
**Yoshikatsu Eto**
*Advanced Clinical Research Center & Institute of the Treatment of Genetic Disorders, Southern Tohoku Brain Research Institute, Japan*

Thu(5)-O48-1

### Phenotypic variability in the form of pulmonary manifestations and molecular analysis in two patients with Niemann Pick-disease type C from India

**Krati R Shah**[1], Jayesh Sheth[1], Frenny Sheth[1], Mehul Mistry[1], Harsh Patel[1], Mamta Muranjan[2], Jijo Joseph[3]
*[1]Institute of Human Genetics, India, [2]Seth G.S. Medical College, Mumbai, [3]MGM Medical College, Navi Mumbai*

Thu(5)-O48-2

### The Canadian Inherited Metabolic Diseases Research Network: Initial findings from a pan-Canadian longitudinal study of affected children

**Beth K Potter**[1], Pranesh Chakraborty[2], Monica Lamoureux[2], Kylie Tingley[1], Doug Coyle[1], Jonathan B Kronick[3], Kumanan Wilson[1], Valerie Austin[3], Catherine Brunel[4], Daniela Buhas[5], Maggie Chapman[6], Alicia KJ Chan[7], Sarah Dyack[6], Annette Feigenbaum[3], Michael Geraghty[2], Alette Giezen[8], Jane Gillis[6], Shailly Jain[7], Erica Langley[2], Julian Little[1], Jennifer MacKenzie[9], + B Maranda, A Mhanni, G Mitchell, JJ Mitchell, L Nagy, A Pender, M Potter, C Prasad, K Siriwardena, R Sparkes, S Stockler, Y Trakadis, L Turner, C VanKarnebeek, H Vallance, J Walia, BJ Wilson
*[1]University of Ottawa, Canada, [2]Children's Hospital of Eastern Ontario, [3]University of Toronto/ Hospital for Sick Children, [4]CHU Ste-Justine, [5]Montreal Children's Hospital, [6]Dalhousie University, [7]University of Alberta, [8]BC Children's Hospital, [9]Queen's University*

Thu(5)-O48-3

### Relative Frequency of Lysosomal Storage Diseases in Brazil: 1982-2015 Report from a Reference Center

**Roberto Giugliani**[1,2,3], Kristiane Michelin-Tirelli[2], Jurema F de Mari[2], Fernanda Bender[1,2], Fernanda Medeiros[2], Ana P Scholz[2], Fernanda Bittencourt[2], Regis R Guidobono[2], Maira G Burin[2], MPS Brazil Network, LSD Brazil Network, NPC Brazil Network, IEM Brazil Network
*[1]Department of Genetics, UFRGS - Federal University of Rio Grande do Sul, Brazil, [2]Medical Genetics Service, Hospital de Clinicas de Porto Alegre, Brazil, [3]INAGEMP, National Institute of Population Medical Genetics, Brazil*

Thu(5)-O48-4

### Plasma Oxysterol and Lysosphingomyelin-509 as Potential Biomarkers for Japanese Patients with Niemann-Pick C disease measured by Tandem MS and their Changes with Miglustat Treatment

**Yoshikatsu Eto**[1], Takeo Iwamoto[2], Ayumi Takamura[3], Miwa Fujisaki[1], Masayo Kashiwazaki[1], Kaoru Eto[4], Norio Sakai[5]
*[1]Advanced Clinical Research Center, Institute of Neurological Diseases, Japan, [2]Core Laboratory, Tokyo Jikei University School of Medicine, [3]Department of Biological Regulation, School of Health Science, Tottori University, [4]Department of Pediatrics, Tokyo Womens Medical School, [5]Department of Child Health, School of Health Science, Osaka University*

Thu(5)-O48-5

**Pharmacological chaperones for the cure of metabolic diseasesPharmacological chaperones for the cure of metabolic diseases**

**Maria Vittoria Cubellis**[1], Valentina Citro[1], Vincenzo Riso[1], Rosita Del Prete[1], Enza Di Meo[1], Antonia Paone[1], Chiara Cimmaruta[1], Giuseppina Andreotti[2]
*[1]Biology, University Federico II, Italy, [2]Istituto di Chimica Biomolecolare; CNR, Italy*

Thu(5)-O48-6

**Transthyretin-type Cerebral Amyloid Angiopathy in Post-transplant Patients with Hereditary ATTR Amyloidosis: Correlates between Clinical Findings and Amyloid-PET Imaging**

**Yoshiki Sekijima**[1,2,3], Masahide Yazaki[1,2], Kazuhiro Oguchi[3], Tsuneaki Yoshinaga[1], Shu-Ichi Ikeda[1]
*[1]Department of Medicine (Neurology & Rheumatology), Shinshu University, Japan, [2]Institute for Biomedical Sciences, Shinshu University, [3]Jisenkai Brain Imaging Research Center*

9:45-11:15 Room C-2

## O49 Concurrent Oral Session 49
## "Metabolic Disorders 4"

Chairs: **Amal M. Alhashem**
*Pediatrics- Division of Medical Genetics, Prince Sultan Military Medical City, Saudi Arabia*
**Seiji Yamaguchi**
*Pediatrics, Shimane University School of Medicine, Japan*

Thu(5)-O49-1

**Molecular genetic study of PKU patients from Russia with a view to their subsequent treatment with BH4**

**Polina Gundorova**, Anna A Stepanova, Alexander V Polyakov
*Federal State Budgetary Institution Research Centre for Medical Genetics, Russia*

Thu(5)-O49-2

**Molecular Characterisation of Hyperphenylalaninemia in Korea**

**Yong Hee Hong**[1], Byong Hwa Rho[2], Dong Hwan Lee[3]
*[1]Department of Pediatrics, Soonchunhyang University Bucheon Hospital, Korea, South, [2]Department of Dermatology, Wootaeha's Skin Clinic, [3]Department of Pediatrics, Soonchunhyang University Hospital*

Thu(5)-O49-3

**Treatment of biotin-responsive basal ganglia disease: Open comparative study between the combination of biotin plus thiamine versus thiamine alone**

**Amal M Alhashem**, Brahim Tabarki
*Pediatrics, Prince Sultan Military Medical City, Saudi Arabia*

Thu(5)-O49-4

**Diversity of disease distribution and genetic background of inherited metabolic diuseases of organic and fatty acids in Asian countries**

**Seiji Yamaguchi**[1], Yuki Hasegawa[1], Naoaki Shibata[1], Hironori Kobayashi[1], Kenji Yamada[1], Ryosuke Bo[1], Takeshi Taketani[1], Seiji Fukuda[1], Toshiyuki Fukao[2], Yanling Yang[3], Sunita Bijarnia[4], Iswar Verma[4], Dung Vu Chi[5], Nahn Nguyen Thu[5]

*[1]Pediatrics, Shimane University School of Medicine, Japan, [2]Pediatrics, Gifu University Graduate School of Medicine, [3]Pediatrics, Pekin University 1st Hospital, China, [4]Medical Genetics, Sir Ganga Ram Hospital, [5]Pediatrics, National Hospital of Pediatrics Hanoi, Vietnam*

Thu(5)-O49-5

**Molecular characterization of beta-ketothiolase deficiency in 9 Indians: Discovery of 3 novel mutations in *ACAT1* gene**

**Elsayed Abdelkreem**[1,2], Hideo Sasai[1], Hiroki Otsuka[1], Radha Rama Devi Akella[3], Usha Dave[4], Toshiyuki Fukao[1]

*[1]Department of Pediatrics, Gifu University, Japan, [2]Department of Pediatrics, Sohag University, Egypt, [3]Department of Pediatric Neurology and Metabolic Medicine, Rainbow Hospital for Women and Children, Hyderabad, India, [4]MILS International, India*

Thu(5)-O49-6

**Human thioredoxin-2 deficiency impairs mitochondrial redox homeostasis and causes early-onset neurodegeneration**

**Eliska Holzerova**[1,2], Katharina Danhauser[3], Tobias B. Haack[1,2], Laura S. Kremer[1,2], Irina Ingold[4], Sho Kobayashi[4,5], Caterina Terrile[2], Ertan Mayatepek[3], Jose P. Friedmann Angeli[4], Marcus Conrad[4], Tim M. Strom[1,2], Thomas Meitinger[1,2], Holger Prokisch[1,2], Felix Distelmaier[3]

*[1]Institute of Human Genetics, Technische Universitaet Muenchen, Germany, [2]Institute of Human Genetics, Helmholtz Zentrum Muenchen, Germany, [3]Department of General Pediatrics, Neonatology and Pediatric Cardiology, University Children's Hospital, Heinrich-Heine-University Duesseldorf, Germany, [4]Institute of Developmental Genetics, Helmholtz Zentrum Muenchen, Germany, [5]Division of Animal Production, Specialty of Bioproduction Science, The United Graduate School of Agricultural Sciences, Iwate University, Morioka, Japan*

8:00-9:30 Sakura

## O50 Concurrent Oral Session 50
## "Statistical Genetics and Genetic Epidemiology 2"

Chairs: **Noah Zaitlen**
*Medicine, University of California San Francisco, USA*
**Qihua Tan**
*Department of Public Health and Department of Clinical Research, University of Southern Denmark, Denmark*

Thu(5)-O50-1

**Gene Network: Accurate prediction of gene functions and prioritization of disease variants**

**Juha Karjalainen**, Sipko van Dam, Niek de Klein, Patrick Deelen, Vinod Kumar, Lude Franke, Cisca Wijmenga

*Genetics, University Medical Center Groningen, Netherlands*

Thu(5)-O50-2

**Joint whole-genome analysis of associations between host and hepatitis C virus diversity in a patient cohort**

Vincent Pedergnana[1], Azim M Ansari[2,3], Paul Klenerman[2], Eleanor Barnes[2], Chris Spencer[1], STOP-HCV Consortium
*[1]Wellcome Trust Center for Human Genetics, UK, [2]Nuffield Department of Medicine, University of Oxford, [3]Oxford Martin School, University of Oxford*

Thu(5)-O50-3

**Powerful and efficient association testing in cohorts with large phenotypic collections**

Noah Zaitlen[1], Hugues Aschard[2], Joel Mefford[1], John Witte[1], Peter Kraft[2]
*[1]Medicine, UCSF, USA, [2]Genetic Epidemiology, HSPH.*

Thu(5)-O50-4

**Metabolic and transciptomic associations of change in body fat percentage.Metabolic and transciptomic associations of change in body fat percentage**

Annika Wennerstrom[1,2], Maria Hagnes[3], Jari Jokelainen[3,4], Pekka Jousilahti[1], Johannes Kettunen[3], Markus Perola[1,2], Sirkka Keinanen-Kiukaanniemi[3,4]
*[1]THL, Finland, [2]University of Helsinki, The Institute for Molecular Medicine Finland (FIMM), Biomedicum Helsinki, Finland, Nordic EMBL Partnership for Molecular Medicine, [3]Center for Life Course Health Research, Oulu Finland, [4]MRC and Unit of Primary Health Care, Oulu Univeristy Hospital, Oulu Finland*

Thu(5)-O50-5

**Gene by environment interaction in human longevity as observed in Danish birth cohorts from 1895 to 1915**

Qihua Tan, Rune Lindahl-Jacobsen, Marianne Nygaard, Lene Christiansen, Kaare Christensen
*EBB, Dept of Public Health, University of Southern Denmark, Denmark*

Thu(5)-O50-6

**A linear algebraic method for evaluating the relation between power and the pattern of linkage disequilibrium in multiple testing**

Tapati Basak, Ryo Yamada
*Statistical Genetics, Unit of Statistical Genetics, Graduate School of Medicine, Kyoto University, Kyoto, Japan*

9:45-11:15 Sakura

## O51 Concurrent Oral Session 51
## "Statistical Genetics and Genetic Epidemiology 3"

Chairs: **Jonathan Marchini**
*Department of Statistics, University of Oxford, UK*
**Atsuko Imai**
*Department of Cardiovascular Medicine / Genome Informatics, Osaka University Graduate School of Medicine, Japan*

Thu(5)-O51-1

### A genome wide association study of pathological inflammatory responses in leprosy

**Vinicius M Fava**[1,2], Aurelie Cobat[3,4], Jeremy Manry[1,2], Marianna Orlova[1,2], Nguyen Van Thuc[5], Milton O Moraes[6], Mariane M.A Stefani[7], Ana Carla P Latini[8], Andrea Belone[8], Nguyen Ngoc Ba[5], Vu Hong Thai[5], Laurent Abel[4,5,9], Alexandre Alcais[4,5,9], Erwin Schurr[1,2]
*[1]Infectious Diseases and Immunity in Global Health Program, Research Institute of the McGill University Health Centre, Canada, [2]The McGill International TB Centre, Departments of Human Genetics and Medicine, McGill University, [3]Laboratory of Human Genetics of Infectious Diseases, Necker Branch, Institut National de la Santé et de la Recherche Médicale U1163, [4]University Paris Descartes, Imagine Institute, Paris, France, [5]Hospital for Dermato-Venerology, [6]Laboratório de Hanseníase, Instituto Oswaldo Cruz, FIOCRUZ, [7]Tropical Pathology and Public Health Institute, Federal University of Goiás, Goiânia, [8]Lauro de Souza Lima Institute, [9]St Giles Laboratory of Human Genetics of Infectious Diseases, Rockefeller Branch, Rockefeller University*

Thu(5)-O51-2

### Empirical estimation of genome-wide significance thresholds based on the 1000 Genomes Project dataset

**Masahiro Kanai**[1], Toshihiro Tanaka[1,2], Yukinori Okada[1,3]
*[1]Department of Human Genetics and Disease Diversity, Graduate School of Medical and Dental Sciences, Tokyo Medical and Dental University, Japan, [2]Bioresource Research Center, Tokyo Medical and Dental University, [3]Laboratory for Statistical Analysis, RIKEN Center for Integrative Medical Sciences*

Thu(5)-O51-3

### Family-Control analysis based on Hamming distance for prioritizing candidate pathogenic variants

**Atsuko Imai**[1,2], Akihiro Nakaya[1], Somayyeh Fahiminiya[3], Martine Tetreault[3], Jacek Majewski[3], Yasushi Sakata[2], Seiji Takashima[2,4], Mark Lathrop[3], Jurg Ott[5,6]
*[1]Department of Genome Informatics, Osaka University Graduate School of Medicine, Japan, [2]Department of Cardiovascular Medicine, Osaka University Graduate School of Medicine, [3]McGill University and Genome Quebec Innovation Centre, [4]Department of Medical Biochemistry, Osaka University Graduate School of Medicine, [5]Institute of Psychology, Chinese Academy of Sciences, [6]Laboratory of Statistical Genetics, Rockefeller University*

Thu(5)-O51-4

### Significant impact of miRNA-target gene networks on genetics of human complex traits

**Masahiro Kanai**[1], Yukinori Okada[1,2], Tomoki Muramatsu[3], Naomasa Suita[1,4], Eiryo Kawakami[5], Valentina Iotchkova[6,7], Nicole Soranzo[6,7], Johji Inazawa[3,8], Toshihiro Tanaka[1,8,9]
*[1]Department of Human Genetics and Disease Diversity, Tokyo Medical and Dental University, Japan, [2]Laboratory for Statistical Analysis, RIKEN Center for Integrative Medical Sciences, [3]Department of Molecular Cytogenetics, Tokyo Medical and Dental University, [4]Advanced Medicinal Research Laboratories, Ono Pharmaceutical CO., LTD., [5]Laboratory for Disease Systems Modeling, RIKEN Center for Integrative Medical Sciences, [6]Human Genetics, Wellcome Trust Sanger Institute, [7]Department of Haematology, University of Cambridge, [8]Bioresource Research Center, Tokyo Medical and Dental University, [9]Laboratory for Cardiovascular Diseases, RIKEN Center for Integrative Medical Sciences*

Thu(5)-O51-5

**Tensor decomposition uncovers trans eQTL networks in the multi-tissue EuroBATS study**

Jonathan Marchini[1], Victoria Hore[1], Ana Vinuela[2,3], Alfonso Buil[4], Mark McCarthy[2,5], Kerrin Small[3]
*[1]Department of Statistics, University of Oxford, UK, [2]Wellcome Trust Center of Human Genetics, University of Oxford, [3]Department of Twin Research and Genetic Epidemiology, King's College London, UK, [4]Department of Genetic Medicine and Development, University of Geneva, Switzerland, [5]Oxford Centre for Diabetes, Endocrinology and Metabolism, University of Oxford, UK*

Thu(5)-O51-6

**Estimating the shared genetic basis of complex phenotypes between populations from summary statistics gives evidence for widespread non-additive and rare variant effects**

Brielin C Brown[1], Alkes Price[3,4], Noah Zaitlen[2]
*[1]Department of Computer Science, University of California Berkeley, USA, [2]Department of Medicine, Lung Biology Center, UC San Francisco, [3]Deparment of Epidemiology, Harvard School of Public Health, [4]Deparment of Biostatistics, Harvard School of Public Health*

8:00-9:30 Room I

## O52 Concurrent Oral Session 52
## "Evolutionary and Population Genetics 1"

Chairs: **Arbel Harpak**
*Department of Biology, Stanford University, USA*
**Yoko Satta**
*Department of Evolutionary Studies of Biosystems, SOKENDAI (The Graduate University for Advanced Studies), Japan*

Thu(5)-O52-1

**Large effects of mutation rate variation and epistasis on the distribution of allele frequencies in humans**

Arbel Harpak[1], Anand Bhaskar[2], Jonathan Pritchard[1,2,3]
*[1]Biology, Stanford University, USA, [2]Genetics, Stanford University, [3]Howard Hughes Medical Institute, Stanford University*

Thu(5)-O52-2

**Whole-genome reference panel of Tohoku Medical Megabank Organization (ToMMo) and allele frequency of pathological variants**

Yumi Yamaguchi-Kabata[1], Yosuke Kawai[1], Kaname Kojima[1], Naoki Nariai[1,2], Yukuto Sato[1], Takahiro Mimori[1], Fumiki Katsuoka[1], Jun Yasuda[1], Masayuki Yamamoto[1], Masao Nagasaki[1]
*[1]Tohoku University, Japan, [2]University of California*

Thu(5)-O52-3

**Touching the limits of being alive: the distribution of genome-wide CNV loads in healthy human cohort is right truncated due to ongoing purifying selection**

Konstantin Popadin[1], Katrin Mannik[1], Aurelien Mace[2], Margit Noukas[3,4], Evelin Mihhailov[3,4], Olga Vakhrusheva[5], Marco Garieri[6], Georgii Bazykin[5], Andres Metspalu[3,4], Zoltan Kutalik[2], Alexandre Reymond[1]
*[1] Center for Integrative Genomics, University of Lausanne, Switzerland, [2]Department of Medical Genetics, Faculty of Biology and Medicine, University of Lausanne, Switzerland, [3]Estonian Genome Center, University of Tartu, Estonia, [4]Institute of Molecular and Cell Biology, University of Tartu, Estonia, [5]Institute for Information Transmission Problems (Kharkevich Institute), Russia, [6]University of Geneva Medical School, University of Geneva, Switzerland*

Thu(5)-O52-4

**Health and population effects of rare gene knockouts in adult humans with related parents**

**Vagheesh M Narasimhan**[1], Konrad J Karczewski[2], John Wright[3], Karen A Hunt[4], Daniel G MacArthur[2], Yali Xue[1], Shane McCarthy[1], Richard Trembath[4], Chris Tyler-Smith[1], Eamonn R Maher[5], Richard Durbin[1], David A van Heel[4]

*[1]Wellcome Trust Sanger Institute, UK, [2]Massachusetts General Hospital, Boston, USA, [3]Bradford Institute for Health Research, [4]Queen Mary University of London, [5]University of Cambridge*

Thu(5)-O52-5

**Spread of reduced activity of STX promoter in modern humans**

**Naoko T. Fujito**[1], Yoko Satta[1], Masaya Hane[2], Atsushi Matsui[3], Ken Kitajima[2], Chihiro Sato[2], Toshiyuki Hayakawa[4]

*[1]School of Advanced Sciences, SOKENDAI (The Graduate University for Advanced Studies), Japan, [2]Bioscience and Biotechnology Center, Nagoya University, [3]Primate Research Institute, Kyoto University, [4]The Graduate School of Systems Life Sciences, Kyushu University*

Thu(5)-O52-6

**Evolution of the 'fused' gene family across primates**

**Hirofumi Nakaoka**, Vanessa Romero, Ituro Inoue, Kazuyoshi Hosomichi,

*National Institute of Genetics - Japan, Ecuador*

9:45-11:15 Room I

## O53 Concurrent Oral Session 53
## "Evolutionary and Population Genetics 2"

Chairs: **Anders Eriksson**
*Biological and Environmental Sciences & Engineering Division, King Abdullah University of Science and Technology, Saudi Arabia*

**Jong Bhak**
*Biomedical Engineering, UNIST (Ulsan National Institute of Science and Technology), Korea, South*

Thu(5)-O53-1

**HUGO-Pan Asian Population Genomics Initiative (PAPGI) project for mapping genomic diversity of Asia**

**Jong Bhak**, HUGO-Pan Asian Population Genomics Initiative (HUGO-PAPGI)

*The Genomics Institute, Ulsan National Institute of Science and Technology (UNIST), Korea, South*

Thu(5)-O53-2

**Spatially explicit models and whole genome analysis for reconstructing the colonisation of Asia**

**Anders Eriksson**[1,2], Kyusang Lee[3], Jong Bhak[3], Andrea Manica[2], Timothy Ravasi[1], Pan-Asian Population Genomics Inititative (PAPGI)

*[1]Integrative Systems Biology Laboratory, King Abdullah University of Science and Technology, Saudi Arabia, [2]Department of Zoology, University of Cambridge, [3]Biomedical Engineering, Ulsan National Institute of Science & Technology*

Thu(5)-O53-3

**Large-scale whole genome sequencing of the Estonian population reveals novel loss-of-function variants and new insights into the population history**

**Reedik Magi**[1], Mart Kals[1], Mario Mitt[1], Kalle Parn[1], Mait Metspalu[2], Lili Milani[1], Tonu Esko[1], Andres Metspalu[1]

*[1]Estonian Genome Center, University of Tartu, Estonia, [2]Estonian Biocentre*

Thu(5)-O53-4

**Fine-Scale Population Structure in Europe**

**Stephen Leslie**[1,2], Garrett Hellenthal[3], Simon Myers[4], Peter Donnelly[4,5], International Multiple Sclerosis Genetics Consortium

*[1]Statistical Genetics, Murdoch Childrens Research Institute, Australia, [2]School of Mathematics and Statistics, University of Melbourne, [3]University College London Genetics Institute, UK, [4]Department of Statistics, University of Oxford, UK, [5]Wellcome Trust Centre for Human Genetics, Oxford, UK*

Thu(5)-O53-5

**SNPs associated for height explain about half of the height difference between two historical subpopulations in Finland**

**Markus Perola**[1,2,3]

*[1]Health, National Institute for Health and Welfare, Finland, [2]University of Helsinki, Institute for Molecular Medicine, Finland (FIMM) and Diabetes and Obesity Research Program, [3]University of Tartu, Estonian Genome Center, Tartu, Estonia*

Thu(5)-O53-6

**Characterization of 20,000 Clinically Relevant Variants in 50,000 Non-European Individuals**

**Eimear E Kenny**[1], Christopher R Gignoux[2], Stephanie Rossi[3], Christopher S Carlson[3], Carlos D Bustamante[2], Noura S Abul-husn[1], The Population Architecture using Genomics and Epidemiology Study

*[1]Icahn School of Medicine at Mo, USA, [2]Stanford University, [3]Fred Hutchinson Center for Cancer Research*

8:00-9:30 Room J

## O54 Concurrent Oral Session 54
## "Clinical Genetic Testing 3"

Chairs: **Roberto Mendoza-Londono**
*Medical Genetics, The Hospital for Sick Children and University of Toronto, Canada*
**Tomohiro Nakayama**
*Division of Laboratory Medicine, Department of Pathology and Microbiology, Nihon University School of Medicine, Japan*

Thu(5)-O54-1

**Genetic causes of Intellectual disability**

**Amal M Mohamed**[1], Alaa K Kamel[1], Nivin A Helmy[1], Sayeda A Hammad[1], Hesham F Kayed[1], Marwa M Shehab[1], Asaad S Gerzawy[1], Maha M Ead[1], Ola M Ead[1], Mona K Mekkawy[1], Maha S Zaki[2], Mona S Aglan[2], Samira M Ismaeel[2], Hala E Bassiouny[2], Mona A Abdel Razek[2], Samia A Temtamy[2]

*[1]Human Cytogenetics, National Research Center, Egypt, [2]Clinical Genetics, National Research Center*

Thu(5)-O54-2

**Triaging of epileptic encephalopathy patients for massive parallel sequencing testing**

**Bruce H. Bennetts**[1,3,4], Kavitha Kothur[2,3], Deepak Gill[2,3], Richard Webster[2,3], Katherine Holman[1], Gladys Ho[1]

*[1]Sydney Genome Diagnostics, The Children's Hospital at Westmead, Australia, [2]Department of Neurology, The Children's Hospital at Westmead, [3]Discipline of Paediatric and Child Health, The University of Sydney, [4]Discipline of Genetic Medicine, The University of Sydney*

Thu(5)-O54-3

**Diagnostic whole exome sequencing of danish families with rare genetic diseases**

**Lotte Risom**, Jakob Ek, Elsebet Ostergaard, Morten Duno

*Dept. of Clinical Genetic, Copenhagen University Hospital, Rigshospitalet, Denmark*

Thu(5)-O54-4

**Diagnosis of Skeletal Dysplasias by Exome Sequencing: The Canadian Experience**

**Roberto Mendoza-Londono**[1], Lucie Dupuis[1,2], Peter Kannu[1,2], Andrew Howard[2,3], Jennifer Stimec[2,4], Jennifer Harrington[2,5], Christian Marshall[6], Tara Paton[6], Michael Brudno[6,7], Taila Hartley[8], Amanda Smith[8], Stephen Scherer[6], Kym Boycott[8], Care4Rare Canada Consortium

*[1]Medical Genetics, The Hospital for Sick Children and University of Toronto, Canada, [2]The Bone Health Centre, The Hospital for Sick Children, [3]Department of Orthopaedic Surgery, [4]Department of Diagnostic Imaging, [5]Division of Endocrinology, [6]The Centre for Applied Genomics and Program in Genetics and Genome Biology, The Hospital for Sick Children, Toronto, Ontario, [7]Computational Biology, [8]Children's Hospital of Eastern Ontario Research Institute, University of Ottawa, Ottawa, Ontario, Canada*

Thu(5)-O54-5

**Clinical validation of a Targeted Massively Parallel Sequencing Panel for Craniosynostosis**

**Tony Roscioli**[1,2,3], Eric Lee[4], Ying Zhu[5], Nicole Snow[3], George Elakis[4], Mark J Cowley[1,2], Velimir Gayevskiy[1], Kevin Ying[1], Corrina Walsh[4], Anne Turner[3], Marcel E Dinger[1,2], Wanda Lattanzi[6], Simeon Boyd[7], Michael F Buckley[4]

*[1]Kinghorn Centre for Clinical Genomics, Kinghorn Centre for Clinical Genomics, Australia, [2]St Vincents Clinical School, University of New South Wales, Darlinghurst, Australia, [3]Department of Medical Genetics, Sydney Childrens Hospital, Randwick, NSW, Australia, [4]SEALS Haematology and Genetics Laboratories, New South Wales Health Pathology, Randwick, Sydney, NSW, Australia, [5]Royal North Shore GOLD Service, Sydney, NSW, Australia, [6]Universita Cattolica del Sacro Cuore, Rome, Italy, [7]UC Davis MIND Institute, Sacramento, USA*

Thu(5)-O54-6

**NGS-based diagnostic DNA analysis in syndromal and nonsyndromal obesity.NGS-based diagnostic DNA analysis in syndromal and nonsyndromal obesity**

**Bert van der Zwaag**[1], Elisabeth F.C. van Rossum[2], Erica L.T. van den Akker[2], Patrick H.A. van Zon[1], Vincent L. Wester[2], Ignace M.C. Janssen[3], Hans Kristian Ploos van Amstel[1], Mieke M. van Haelst[1]

*[1]Department of Genetics, UMC Utrecht, Utrecht, The Netherlands, [2]Department of Internal Medicine, Obesity Center CGG, Erasmus MC, University Medical Center Rotterdam, Rotterdam, The Netherlands, [3]Vitalys Obesitas Centrum, Vitalys Klinieken, Velp, The Netherlands*

9:45-11:15 Room J

## O55 Concurrent Oral Session 55
## "Clinical Genetic Testing 4"

Chairs: **Paul F. Lasko**
*McGill University, Canada*
**Jun Mitsui**
*Department of Neurology, The University of Tokyo, Japan*

Thu(5)-O55-1

### Beyond the ACMG 56: Parental choices and initial results from a comprehensive whole genome sequencing-based search for predictive genomic variants in children

**M Stephen Meyn**[1,2,3,4,5], Nasim Monfared[5], Christian Marshall[6,7], Daniele Merico[1,6], Dmitri J Stavropoulos[7,15], Robin Z Hayeems[5,8], Michael Szego[6,9,10], Rebekah Jobling[2], Marta Girdea[1,11], Gary D Bader[3,12], Michael Brudno[1,11], Ronald D Cohn[1,2,3,4,5], Stephen W Scherer[1,3,5,6,13], Randi Zlotnik Shaul[4,8,14], Cheryl Shuman[3,4], Peter N Ray[1,3,5,6,7], Sarah C Bowdin[2,4,5]
*[1]Program in Genetics and Genome Biology, The Hospital for Sick Children, Canada, [2]Division of Clinical and Metabolic Genetics, The Hospital for Sick Children, [3]Department of Molecular Genetics, University of Toronto, [4]Department of Paediatrics, University of Toronto, [5]Centre for Genetic Medicine, The Hospital for Sick Children, [6]The Centre for Applied Genomics, The Hospital for Sick Children, [7]Department of Paediatric Laboratory Medicine, The Hospital for Sick Children, [8]Program in Child Health Evaluative Services, The Hospital for Sick Children, [9]Joint Centre for Bioethics, University of Toronto, [10]Centre for Clinical Ethics, St. Joseph's Health Centre, [11]Department of Computer Science, University of Toronto, [12]The Donnelly Centre, University of Toronto, [13]The McLaughlin Centre, University of Toronto, [14]Department of Bioethics, The Hospital for Sick Children, [15]Department of Laboratory Medicine and Pathology, University of Toronto*

Thu(5)-O55-2

### EuroGentest Guidelines for Diagnostic Next Generation Sequencing

**Gert Matthijs**[1], Erika Souche[1], Marielle Alders[2], Anniek Corveleyn[1], Sebastian Eck[3], Ilse Feenstra[4], Valerie Race[1], Erik Sistermans[5], Marc Sturm[6], Marjan Weiss[5], Helger Yntema[4], Egbert Bakker[7], Peter Bauer[6], Participants to the EuroGentest workshop on Diagnostic NGS Guidelines
*[1]Center for Human Genetics, University of Leuven, Belgium, [2]Department of Clinical Genetics, Academic Medical Centre (AMC), University of Amsterdam, [3]Center for Human Genetics and Laboratory Medicine Dr. Klein, Dr. Rost and Colleagues, Martinsried, [4]Department of Human Genetics, Radboud University Medical Center, Nijmegen, [5]Department of Clinical Genetics, VU University Medical Center, Amsterdam, [6]University Hospital of Tuebingen, Institute of Medical Genetics and Applied Genomics, [7]Department of Clinical Genetics, Leiden University Medical Center*

Thu(5)-O55-3

### Data sharing improves the diagnostic yield of clinical exome sequencing and identifies new disease genes

**Koen L.I. van Gassen**, Martin Elferink, Marc C. van Tuil, Patrick van Zon, Jacques C. Giltay, Mieke M. van Haelst, Eva H. Brilstra, Nine V. Knoers, Gijs van Haaften, Hans Kristian Ploos van Amstel
*Department Genetics, University Medical Center Utrecht, Netherlands*

Thu(5)-O55-4

### The Undiagnosed Diseases Network International (UDNI): Clinical and Laboratory Research to Meet Patient Needs

**P Lasko**[4], John J Mulvihill[1], G Baynam[2], W Gahl[1], S C Groft[1], K Kosak[3], B Melegh[5], D Taruscio[6], Undiagnosed Diseases Network International
*[1]Division of Genomic Medicine, National Human Genome Research Institute, USA, [2]Princess Margaret and King Edward Memorial Hospitals, [3]Center for Medical Genetics, Keio University, [4]McGill University, [5]Department of Medical Genetics, Pecs, Hungary, [6]Istituto Superiore di Sanita, Rome*

Thu(5)-O55-5

**Need to concern about contamination by circulating fetal DNA**

**Jianli Dong**, Hai Wu, Gengming Huang, Zurina Romay-Penabad
*Pathology, University of Texas Medical Branch, USA*

Thu(5)-O55-6

**Preconception screening results for Mendelian diseases in East Asian populations**

**Michal Golan-Mashiach**, Erez Tzur, Itamar Shamshins, Lital Isaacs
*Dr Gene Honk Kong limited, Hong Kong*

8:00-9:30 Room K

## O56 Concurrent Oral Session 56
## "Cardiovascular Genetics 1"

Chairs: **Elena V. Zaklyazminskaya**
*Medical Genetics Laboratory, Petrovsky Russian Research Centre of Surgery, Russia*
**Hiroko Morisaki**
*Department of Bioscience and Genetics, National Cerebral and Cardiovascular Center Research Institute, Japan*

Thu(5)-O56-1

**TTN truncating mutations double the diagnostic yield for DCM and NCCM patients; three years of experience with a targeted panel for cardiomyopathies**

**Marjon A van Slegtenhorst**[1], Marianne van Tienhoven[1], Judith M.A. Verhagen[1], Jaap I. van Waning[1], Ingrid M.B.H. van de Laar[1], Kadir Caliskan[2], Michelle Michels[2], Marja W. Wessels[1], Danielle F. Majoor-Krakauer[1], Hennie T. Bruggenwirth[1], Rogier A. Oldenburg[1]
*[1]Department of Clinical Genetics, Ee2475, Erasmus Medical Center, Netherlands, [2]Department of Cardiology, Thoraxcenter, Erasmus Medical Center*

Thu(5)-O56-2

**Characterizing functional regulatory variants in iPSC-derived human cardiomyocytes**

**Paola Benaglio**, Christopher DeBoever, Angelo Arias, Frauke Drees, Hiroko Matsui, He Li, Agnieszka D'Antonio-Chronowska, Kelly Frazer
*Pediatrics, UC San Diego, USA*

Thu(5)-O56-3

**Genomic prediction of coronary heart disease**

**Michael Inouye**[1], Gad Abraham[1], Samuli Ripatti[2], Veikko Salomaa[3], Nilesh Samani[4]
*[1]Centre for Systems Genomics, University of Melbourne, Australia, [2]Institute of Molecular Medicine, University of Helsinki, [3]National Institute of Health and Welfare, Finland, [4]Cardiovascular Research Unit, University of Leicester, UK*

Thu(5)-O56-4

**Fatty Acid Oxidation Genes in Childhood Arrhythmia: A Pathway Forgotten**

**Zahurul A Bhuiyan**[1], Elhadi H Aburawi[2], Lihadh Al-Gazali[2], Harsha D Devalla[3], Abdelaziz Beqqali[4], Arie O Verkerk[4], Zenia Tiang[5], Safar Al-Shahrani[6], Samuel Dudley[7], Arthur A.M. Wilde[4,8], Roger S.Y. Foo[5], Jumana Al-Aama[8], Robert Passier[3]

*[1]Laboratoire de diagnostic moleculaire, University Hospital Lausanne (CHUV), Switzerland, [2]Department of Pediatrics, College of Medicine and Health Sciences, UAE University, Al Ain, United Arab Emirates, [3]Department of Anatomy & Embryology, Leiden University Medical Center, Leiden, the Netherlands, [4]Heart Center, Department of Clinical and Experimental Cardiology, Academic Medical Center, Amsterdam, University of Amsterdam, the Netherlands, [5]Department of Cardiology, National University of Singapore, Kent Ridge, Singapore, [6]Department of Cardiology, King Khaled University School of Medicine, Abha, Saudi Arabia, [7]Lifespan Cardiovascular Institute, Warren Alpert Medical School of Brown University, RI, USA, [8]Princess Al-Jawhara Al-Brahim Centre of Excellence in Research of Hereditary Disorders, Jeddah, Saudi Arabia*

Thu(5)-O56-5

**High prevalence of psycho-neurological complications are associated with mutation in *SCN5A* gene**

**Elena V. Zaklyazminskaya**[1,2], Irena V. Pronicheva[3], Amiran Sh. Revishvili[3]

*[1]Medical Genetics Laboratory, Petrovsky Russian Research Centre of Surgery, Russia, [2]Pirogov Russian National Research Medical University, [3]Bakulev Research Centre of Cardiovascular Surgery*

Thu(5)-O56-6

**Up-regulation of *FLT1* by a novel functional SNP increases risk of coronary artery disease through an inflammatory activation**

**Kouichi Ozaki**[1], Takashi Morizono[2], Tatsuhiko Tsunoda[2], Michiaki Kubo[3], Toshihiro Tanaka[1,4,5]

*[1]Cardiovascular Diseases, RIKEN Center for Integrative Medical Sciences, Japan, [2]Laboratory for Medical Science Mathematics, RIKEN Center for Integrative Medical Science, Yokohama, Japan, [3]RIKEN Center for Integrative Medical Science, Yokohama, Japan, [4]Bioresourse Research Center Tokyo Medical and Dental University, Tokyo, Japan, [5]Department of Human Genetics and Disease Diversity, Tokyo Medical and Dental University Graduate School of Medical and Dental Sciences, Tokyo, Japan*

9:45-11:15 Room K

## O57 Concurrent Oral Session 57
## "Cardiovascular Genetics 2"

Chairs: **Geneviève Galarneau**
*The Charles Bronfman Institute for Personalized Medicine, Icahn School of Medicine at Mount Sinai, USA*
**Takayuki Morisaki**
*Department of Bioscience and Genetics, National Cerebral and Cardiovascular Center, Japan*

Thu(5)-O57-1

**Two common single nucleotide polymorphisms in the Renalase gene increase the susceptibility to essential hypertension**

**Amrita Anand Iyer**[1], Parshuram J Sonawane[1], Kalyani Ananthamohan[1], Lakshmi Subramanian[1], Saurabh Sharma[2], Madhu Khullar[2], Ajit S Mullasari[3], Nitish R Mahapatra[1]

*[1]Department of Biotechnology, Indian Institute of Technology, Madras, India, [2]Department of Experimental Medicine and Biotechnology, Postgraduate Institute of Medical Education and Research, Chandigarh, [3]Institute of Cardiovascular Diseases, Madras Medical Mission, Chennai*

Thu(5)-O57-2

### *APOL1* risk allele is associated with early diagnosis of hypertension and a 2-3 mmHg increase in systolic blood pressure in young African American adults

**Geneviève Galarneau**[1], Girish N Nadkarni[1], Stephen B Ellis[1], Rajiv Nadukuru[1], Stuart A Scott[1], Rongling Li[2], Laura J Rasmussen-Torvik[3], Abel N Kho[3], M Geoffrey Hayes[3], Jennifer A Pacheco[3], Teri A Manolio[2], Rex L Chisholm[3], Dan M Roden[4], Joshua C Denny[4], Eimear E Kenny[1], Erwin P Bottinger[1], The eMERGE Network

*[1]Icahn School of Medicine at Mount Sinai, USA, [2]National Human Genome Research Institute, National Institutes of Health, [3]Feinberg School of Medicine, Northwestern University, [4]Vanderbilt University Medical Center*

Thu(5)-O57-3

### DNA methylation in arteries and peripheral blood of patients with atherosclerosis

**Anton V. Markov**[1,2], Maria S. Nazarenko[1,2], Aleksei A. Sleptcov[1,2], Aleksei V. Frolov[3], Olga L. Barbarash[3], Valery P. Puzyrev[1,2]

*[1]Laboratory of Population Genetics, Research Institute of Medical Genetics, Russia, [2]Tomsk State University, [3]Research Institute for Complex Problems of Cardiovascular Diseases*

Thu(5)-O57-4

### A locus near *GRAMD1B* is associated with serum level of atheroprotective anti-phosphorylcholine: genetic effects shared with chronic lymphocytic leukemia

**Xu Chen**[1], Stefan Gustafsson[2], Robert Karlsson[1], Jie Song[1], Iffat Rahman[3], Jun Su[3], Lars Lind[4], Gunnar Engstrom[5], Kenneth Caidahl[6], Johan Frostegard[3], Patrik K.E Magnusson[1]

*[1]Department of Medical Epidemiology and Biostatistics, Karolinska Institutet, Sweden, [2]Department of Medical Sciences, Molecular Epidemiology and Science for Life Laboratory, Uppsala University, [3]Institute of Environmental Medicine, Karolinska Institutet, Stockholm, Sweden, [4]Department of Medical Sciences, Cardiovascular Epidemiology, Uppsala University, [5]Department of Clinical Sciences, Lund University, Malmo, Sweden, [6]Department of Molecular Medicine and Surgery, Karolinska Institutet, Stockholm, Sweden*

Thu(5)-O57-5

### Familial Thoracic Aortic Aneurysms and Dissections (FTAAD) with *ACTA2* Mutation in Japanese

**Takayuki Morisaki**[1], Akiko Yoshida[1], Tomohiko Watanabe[1], Kazufumi Ida[1], Hiroaki Sasaki[2], Tatsuya Oda[2], Hiroshi Tanaka[2], Kenji Minatoya[2], Hiroko Morisaki[1]

*[1]Bioscience and Genetics, and Medical Genetics, National Cerebral and Cardiovascular Center, Japan, [2]Cardiovascular Surgery, National Cerebral and Cardiovascular Center*

Thu(5)-O57-6

### Context-specific eQTLs implicate potential obesity-related transcriptional control by diet in men

**Arthur Ko**[1,2], Marcus Alvarez[1], Elina Nikkola[1], Rita M Cantor[1], Mete Civelek[3], Aldons J Lusis[1,4], Johanna Kuusisto[5], Michael Boehnke[6], Karen L Mohlke[7], Markku Laakso[5], Paivii Pajukanta[1,2,8]

*[1]Department of Human Genetics, University of California, Los Angeles, USA, [2]Molecular Biology Institute at UCLA, [3]Center for Public Health Genomics, University of Virginia, [4]Department of Medicine, David Geffen School of Medicine at UCLA, [5]Department of Medicine, University of Eastern Finland and Kuopio University Hospital, [6]Department of Biostatistics and Center for Statistical Genetics, School of Public Health, University of Michigan, [7]Department of Genetics, University of North Carolina, Chapel Hill, [8]Bioinformatics Interdepartmental Program, UCLA*

8:00-9:30 Room H

## O58 Concurrent Oral Session 58
## "Genome structure, variation and function 3"

Chairs: **Jian-Min Chen**
*INSERM U1078 and EFS-Bretagne, Brest, France*
**Issei Imoto**
*Department of Human Genetics, Institute of Biomedical Sciences, Tokushima University Graduate School, Japan*

Thu(5)-O58-1

### Alternative Splicing in Response to Ionizing Radiation
**Niema Razavian**, Vivian G Cheung
*University of Michigan, Department of Pediatrics, Howard Hughes Medical Institute, USA*

Thu(5)-O58-2

### RNA sequencing reveals stress responses of iPSC-derived endothelial cells isolated from peripheral blood mononuclear cells of supercentenarians
**Cristine R. Casingal**[1,2], Hirofumi Nakaoka[1,2], Yasumichi Arai[3], Nobuyoshi Hirose[3], Ituro Inoue[1,2]
*[1]Division of Human Genetics, National Institute of Genetics, Japan, [2]Department of Genetics, The Graduate School for Advanced Studies, Kanagawa, Japan, [3]Center for Supercentenarian Study, Keio University School of Medicine, Tokyo, Japan*

Thu(5)-O58-3

### Differential extracellular abundance of *COG5* in synovial fluid following meniscal injury supports its role as a major susceptibility gene for knee osteoarthritis
**Liyong Wang**[1,2], Danica D. Vance[3,4], Arpit Mehta[1], Evadnie Rampersaud[1], Bryson P. Lesniak[4], Jeffery M. Vance[1,2], Margaret A. Pericak-Vance[1,2], Lee D. Kaplan[3]
*[1]John P. Hussman Institute for Human Genomics, University of Miami Miller School of Medicine, USA, [2]Dr. John T. Macdonald Foundation Department of Human Genetics, University of Miami Miller School of Medicine, [3]UHealth Sports Performance and Wellness Institute, University of Miami Miller School of Medicine, [4]Department of Orthopedic Surgery, New York Presbyterian Hospital, Columbia University Medical Center*

Thu(5)-O58-4

### Heterogeneity in the individual transcriptomic response to severe sepsis
**Katie L Burnham**[1], Emma E Davenport[1], Jayachandran Radhakrishnan[1], Peter Humburg[1], Paula Hutton[2], Christopher S Garrard[2], Charles J Hinds[3], Julian C Knight[1]
*[1]Wellcome Trust Centre for Human Genetics, UK, [2]Adult Intensive Care Unit, John Radcliffe Hospital, Oxford, UK, [3]William Harvey Research Institute, Barts and the London School of Medicine, UK*

Thu(5)-O58-5

### Inter-individual Variations in Nature and Diversity of Human Facial Skin Microbiome are Significantly Predicted by Sebum and Hydration Levels in Specific Facial Regions
**Souvik Mukherjee**[1], Rupak Mitra[2], Arindam Maitra[3], Satyaranjan Gupta[2], Srikala Kumaran[2], Amit Chakrabortty[2], Partha P Majumder[3]
*[1]BioMedical Genomics Centre, National Institute of Biomedical Genomics, India, [2]Unilever R&D, Bangalore, Karnataka, India, [3]National Institute of Biomedical Genomics, Kalyani, West Bengal, India*

Thu(5)-O58-6

**Meta-analysis of 1343 small complex mutations causing human inherited disease reveals a new mutational signature characteristic of the action of translesion synthesis DNA polymerases in the human genome**

Jian-Min Chen[1,2,3], Claude Ferec[1,2,3], David N Cooper[4]
*[1]Institut National de la Sante et de la Recherche Medicale (INSERM), U1078, Brest, France, [2]Etablissement Francais du Sang (EFS) Bretagne, Brest, France, [3]Faculte de Medecine et des Sciences de la Sante, Universite de Bretagne Occidentale (UBO), Brest, France, [4]Institute of Medical Genetics, School of Medicine, Cardiff University, Cardiff, United Kingdom*

9:45-11:15 Room H

**O59** **Concurrent Oral Session 59**
**"Health Services Research"**

Chairs: Beatriz Marcheco-Teruel
*National Center of Medical Genetics, Havana, Cuba*
Hiroshi Tanaka
*Tohoku Medical Megabank Organization, Tohoku University, Japan*

Thu(5)-O59-1

**Impact of Genome Sequencing on the Medical Care of Healthy Adults: A Randomized Controlled Trial**

Jason L. Vassy[1,2], Kurt D Christensen[2], Dmitry Dukhovny[3], Carrie Blout[2], Jill Oliver Robinson[4], Joel B. Krier[2], Michael F Murray[5], Amy L McGuire[4], Robert C Green[2,6], for the MedSeq Project
*[1]VA Boston Healthcare System, Harvard Medical School, USA, [2]Brigham and Womens Hospital, Harvard Medical School, [3]Oregon Health & Science University, [4]Baylor College of Medicine, [5]Geisinger Health System, [6]Partners HealthCare Personalized Medicine*

Thu(5)-O59-2

**Assessing the Clinical Utility of Family Health History for Guiding Preventive Care in the General Population**

Lori A Orlando[1], Rachel A Myers[1], Adam H Buchanan[2], R. Ryanne Wu[1], Elizabeth R Hauser[1], Geoffrey S Ginsburg[1]
*[1]Medicine, Duke University, USA, [2]Geinsinger Health System*

Thu(5)-O59-3

**A national program for preventing sickle cell anemia: the 30 years Cuban experience**

Beatriz Marcheco-Teruel
*National Center of Medical Genetics, Cuba*

Thu(5)-O59-4

**Epidemiology and health system impact of true-positive and false-positive newborn screening results for phenylketonuria in Ontario, 2006-2012**

**Beth K Potter**[1], Sara D Khangura[1], Pranesh Chakraborty[1,2,3], Christine Davies[2], Doug Coyle[1], Kumanan Wilson[4,5], Marni Brownell[6], Linda Dodds[7], Annette Feigenbaum[8,9], Deshayne B Fell[10], Astrid Guttmann[5,8], Steven Hawken[5], Robin Hayeems[8], Jonathan B Kronick[8,9], Anne-Marie Laberge[11], Aizeddin Mhanni[12], Meranda Nakhla[13], Cheryl Rockman-Greenberg[12], Rebecca Sparkes[14], Keiko Ueda[15], Hilary Vallance[16], with Brenda J Wilson, University of Ottawa; and on behalf of the Canadian Inherited Metabolic Diseases Research Network

*[1]University of Ottawa, Canada, [2]Newborn Screening Ontario, [3]Children's Hospital of Eastern Ontario, [4]Ottawa Health Research Institute, [5]Institute for Clinical Evaluative Sciences, [6]Manitoba Centre for Health Policy, [7]Dalhousie University, [8]The Hospital for Sick Children, [9]University of Toronto, [10]Better Outcomes Registry and Network Ontario, [11]Hopital Sainte-Justine, [12]University of Manitoba, [13]Montreal Children's Hospital, [14]Alberta Children's Hospital, [15]BC Children's Hospital, [16]University of British Columbia*

Thu(5)-O59-5

**Three Dimentional Motion Capture System for Quantitative Evaluation of Motor Functions applied to Healthy Adult and Spinal Muscular Atrophy Patient with Thyrotropine Releasing Hormone Therapy**

**Naoki Matsumaru**[1,2], Zenichiro Kato[2,3], Katsura Tsukamoto[1], Ryo Hattori[4], Norihito Shimizu[4], Yasutaka Shii[4], Hidenori Ohnishi[3], Norio Kawamoto[3], Toshiyuki Fukao[3], Tadayuki Kato[4], Takaaki Aoki[5], Kei Miyamoto[5], Haruhiko Akiyama[5], Michinori Funato[6]

*[1]Global Regulatory Science, Gifu Pharmaceutical University, Japan, [2]Division of Structural Medicine, The United Graduate School of Drug Discovery and Medical Information Sciences, Gifu University, [3]Department of Pediatrics, Graduate School of Medicine, Gifu University, [4]Department of Rehabilitation, Gifu University Hospital, [5]Department of Orthopedic Surgery, Graduate School of Medicine, Gifu University, [6]Department of Clinical Research, National Hospital Organization, Nagara Medical Center*

Thu(5)-O59-6

**Informing policy and practice: a 360 degree evaluation of the impact of prospective WES in comparison to standard care**

**Clara L. Gaff**[1], Ivan Macciocca[1,2], Melissa R. Martyn[1,3], William J. Wilson[4], Deborah Schofield[5], Susan M. White[2,6], Zornitza Stark[2], Paul James[6,7], Andrew Roberts[7,8], Monique Ryan[9], Tim Day[7], Maie Walsh[2], Patrick Kwan[7], Peiro Perucca[7], Alex Boussioutas[6,7,10], Graham Taylor[2], Alicia Oshlack[3], Natalie Thorne[1], Tim Bakker[1], Evaluation team, Genetic Counselling team and the Melbourne Genomics Health Alliance

*[1]Melbourne Genomics Health Alliance, Australia, [2]Victorian Clinican Genetics Services, Vic, Australia, [3]Murdoch Childrens Research Institute, Vic, Australia, [4]Commonwealth Scientific and Industrial Research Organisation (CSIRO), Australia, [5]University of Sydney, NSW, Australia, [6]University of Melbourne, Vic, Australia, [7]Melbourne Health, Vic, Australia, [8]Walter and Eliza Hall Institute, Vic, Australia, [9]Royal Children's Hospital, Vic, Australia, [10]Peter MacCallum Cancer Centre, Vic, Australia*