Tuesday, April 5

8:00-10:00 Annex 1

CIS11 Concurrent Invited Session 11
"Frontiers of Human Genomics"

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

Jun Wang
iCarbonX, China

In this session, we will hear from four prominent researchers exploring the frontiers of human genomics. Dr. Jun Wang co-founded BGI and has published over 400 original papers in genomic sciences. Last year, he founded a new company, ICarbonX, to establish a health-related big-omics data platform and to manage individual health more effectively. Dr. Richard Durbin co-leads the 1,000 Genomes Project and is well-known for developing many computational sequence analysis programs (e.g., HMMER, BWA, and GeneWise) and biological databases (e.g., WormBase, Pfam, and Ensembl). Dr. Erez Lieberman-Aiden developed the Hi-C method for studying long-range interactions in a genome-wide manner. Hi-C has been widely used to examine the entire three-dimensional structures of genomes, revealing important principles of chromatin looping. Dr. Brian Piening works on the Human Microbiome Project with Dr. Michael Snyder; he analyzes the dynamic aspects of microbiome-host omics during periods of human health and disease. We hope that this session will provide a vision of the future for human genomics.

Tue(3)-CIS11-1
Million Genomes Ahead
Jun Wang
iCarbonX, China

Tue(3)-CIS11-2
New human genome reference structures
Richard Durbin
Wellcome Trust Sanger Institute, UK

Tue(3)-CIS11-3
Reading and Writing Genomes in 3D: The CTCF code and how to hack it
Erez Lieberman Aiden
Molecular & Human Genetics, Baylor College of Medicine, USA

Tue(3)-CIS11-4
An 'omic checkup: longitudinal multi-omics for personalized medicine
Brian D. Piening
Genetics, Stanford University, USA

10:20-12:20 Annex 1

CIS12 Concurrent Invited Session 12
"Variations in Genome Structure"

Conveners: **Hiroki Kurahashi**
Division of Molecular Genetics, Institute for Comprehensive Medical Science, Fujita Health University, Japan
Wigard P. Kloosterman
Dept. of Medical Genetics, Center for Molecular Medicine, University Medical Center Utrecht, The Netherlands

Structural genomic variations (SVs) form a relatively uncharted territory of genetic variations and include both copy neutral rearrangement and copy number changes, such as deletions and duplications. SVs form a major driver of human phenotypic variation and disease. Advances in next-generation sequencing have enabled the discovery of SVs at an unprecedented scale in recent years. This has provided substantial insight into the mechanistic origin of SVs and has also unveiled unexpected complexity of some SVs. Yet, the full spectrum and architecture of SVs will likely only become clear with further improvements in sequencing technology and analysis tools.
This session will highlight our current knowledge on the frequencies and types of SVs in the human genome based on data from population-scale sequencing projects. Another topic concerns the underlying mechanistic origin of SVs, both simple changes as well as complex genomic rearrangements involving chromothripsis, Finally, latest insights into SV discovery from third-generation sequencing methods will be presented, which provides an outlook into future developments in this field.

Speakers:

Tue(3)-CIS12-1

Somatic mosaicism - how much of *de novo* is mitotic?
Pawel Stankiewicz[1,2]
[1]Molecular & Human Genetics, Baylor College of Medicine, USA, [2]Institute of Mother and Child, Warsaw, Poland

Tue(3)-CIS12-2

Palindrome-mediated recurrent translocations in humans
Hiroki Kurahashi
Division of Molecular Genetics, Institute for Comprehensive Medical Science, Fujita Health University, Japan

Tue(3)-CIS12-3

Detection and interpretation of complex structural variations in human genomes
Wigard Kloosterman
Dept. of Medical Genetics, Center for Molecular Medicine, University Medical Center Utrecht, The Netherlands

Tue(3)-CIS12-4

Genome-first detection of variation with single-molecule sequencing
Mark J.P. Chaisson
Genome Sciences, University of Washington, USA

8:00-10:00 Annex 2

CIS13 **Concurrent Invited Session 13**

"Complex Trait Diseases 1: Common Disease"

Conveners: **Michiaki Kubo**
Laboratory for Genotyping Development, Center for Integrative Medical Sciences, RIKEN, Japan
Anne M. Bowcock
National Heart and Lung Institute, Imperial College, London, UK

In the past ten years, widespread genomic mapping of factors contributing to complex traits have led to the identification of thousands of loci, and some specific genes, that harbor non-coding variants that lead to genetic susceptibility. However, how these factors lead to dysregulation of specific genes and pathways is still largely unknown; however, specific studies are providing molecular insight into the biology of their complexity. Such studies exemplify two types of complexity: first, their inheritance and second their biology. This session will use four exemplary study systems to demonstrate how genetic advancements can lead to understanding disease pathophysiology.

Tue(3)-CIS13-1

Statistical Genetics for Autoimmune Diseases and Drug Discovery

Yukinori Okada
Department of Human Genetics and Disease Diversity, Tokyo Medical and Dental University; Laboratory for Statistical Analysis, RIKEN Center for Integrative Medical Sciences, Japan

Tue(3)-CIS13-2

DRIVER GENES OF ORAL CANCER, ITS PROGRESSION AND METASTASIS

Partha P. Majumder
National Institute of Biomedical Genomics, India

Tue(3)-CIS13-3

Transcriptome landscape of chronic traumatic encephalopathy and Alzheimer disease in human brains

Jeong-Sun Seo
Genomic Medicine Institute, Seoul National University, Korea

Tue(3)-CIS13-4

Genetic analysis of psoriasis and psoriatic arthritis

Anne M. Bowcock
National Heart and Lung Institute, Imperial College, London, UK

10:20-12:20 Annex 2

CIS14 Concurrent Invited Session 14
"Complex Trait Diseases 2: Autoimmune Diseases"

Conveners: **Kazuhiko Yamamoto**
Department of Allergy and Rheumatology, Graduate School of Medicine, University of Tokyo, Japan
Soumya Raychaudhuri
Medicine, Harvard Medical School, Divisions of Genetics and Rheumatology, Brigham and Women's Hospital; Medical and Population Genetics, Broad Institute; Institute of Inflammation and Repair, University of Manchester; Department of Medicine, Karolinska Institutet, USA

The majority of autoimmune diseases are complex genetic traits. Since early 1970s, HLA has been identified as the most important genetic factor of many autoimmune diseases, but the precise roles of HLA have not yet clarified. Over the last decade, genome-wide association studies (GWASs) have been employed to identify hundreds of non-HLA susceptibility loci. Recently, extensive GWAS and meta-analyses have been performed for several autoimmune diseases, leading to the discovery of >100 alleles for inflammatory bowel disease and rheumatoid arthritis. In order to promote our understanding on the pathogeneses using our findings from genetic studies, it is vital to determine how disease associated variants function to influence fundamental human immune processes. Integration of clinical information and functional immune data on patients with genetic data has the potential to enable us the discovery of mechanistic clues. On the other hand, in common autoimmune diseases, the driving alleles for the majority of these loci do not map to known coding regions of the genome. Several studies suggest many autoimmune variants are enriched for expression quantitative trait loci (eQTL), which influence the expression level of genes. Further, non-coding chromatin marks that identify regions of the genome with regulatory potential, such as H3K4me3, have been found to localize genetic signals in critical cell types, and may be useful for fine-mapping autoimmune disease loci and eQTL. Therefore, comprehensive integrative approach using such information will unravel the fine mechanisms of complex trait autoimmune diseases. In this session, four speakers will discuss several issues above.

Tue(3)-CIS14-1
Fine-mapping the HLA and other autoimmune loci
Soumya Raychaudhuri
Medicine, Harvard Medical School, Divisions of Genetics and Rheumatology, Brigham and Women's Hospital; Medical and Population Genetics, Broad Institute; Institute of Inflammation and Repair, University of Manchester; Department of Medicine, Karolinska Institutet, USA

Tue(3)-CIS14-2
GWAS and functional genomics of autoimmune diseases
Yuta Kochi
Laboratory for Autoimmune Diseases, IMS, RIKEN, Japan

Tue(3)-CIS14-3
Combining population and clinical biobanks to translate genetic variation into immune function
Cisca Wijmenga
Genetics, University Medical Center Groningen, Netherlands

Tue(3)-CIS14-4
Transcriptome variation in human immunity
Barbara E. Stranger
Section to Genetic Medicine, University of Chicago, USA

8:00-10:00 Room A

CIS15 **Concurrent Invited Session 15**

"Therapy for Genetic Diseases"

Conveners: **Eiji Nanba**
Division of Functional Genomics, Research Center for Bioscience and Technology, Tottori University, Japan
Jeffery W. Kelly
Molecular And Experimental Medicine, The Scripps Research Institute; The Skaggs Institute for Chemical Biology, The Scripps Research Institute, USA

Most genetic disorders unfortunately cannot be cured. For a group of genetic disorders called inborn errors of metabolism, which result from genetic mutations that disrupt the product of specific proteins or enzymes. Molecular understanding on these diseases has been advanced during the last decades and leads to novel therapeutic agents. This session in the *Therapy for genetic diseases* explores in depth many aspects of novel therapeutic approaches on the genetic metabolic diseases as well as neurodegenerative diseases.

Tue(3)-CIS15-1

Understanding the Genetics and Biochemistry of the Transthyretin Amyloid Diseases Afforded a Disease-Modifying Therapy, and Importantly, New Insights about Chaperone Function

Jeffery W. Kelly
Molecular And Experimental Medicine, The Scripps Research Institute; The Skaggs Institute for Chemical Biology, The Scripps Research Institute, USA

Tue(3)-CIS15-2

AUTOPHAGY AND OTHER PATHWAYS THAT PROTECT AGAINST NEURODEGENERATION

David C. Rubinsztein
Cambridge Institute for Medical Research, University of Cambridge, UK

Tue(3)-CIS15-3

Treatment of Friedreich's ataxia and mitochondrial diseases

Jeanne Amiel
Institute Imagine and University Paris Descartes, Paris, France

Tue(3)-CIS15-4

Chaperone therapy for lysosomal storage diseases

Katsumi Higaki
Research Center for Bioscience and Technology, Tottori University, Japan

10:20-12:20 Room A

CIS16 Concurrent Invited Session 16
"Gene Therapy"

Conveners: **Keiya Ozawa**
Division of Genetic Therapeutics, The Advanced Clinical Research Center, Institute of Medical Science, The University of Tokyo, Japan
Michel Sadelain
Center for Cell Engineering, Memorial Sloan Kettering Cancer Center, New York, USA

Gene therapy research remained stagnant for many years due to serious side effects. However, clinical gene therapy has been revived in Western countries, because a number of successful clinical trials have been reported recently. Currently, lentiviral vectors and AAV (adeno-associated virus) vectors are mainly utilized for gene transfer into hematopoietic stem cells and differentiated cells (neurons, muscles, retinal pigment epithelial cells, hepatocytes and so on), respectively. The introduction of safe AAV vectors has expanded the target diseases of gene therapy. Regarding cancer gene therapy, there has been increasing focus on engineered T cell therapy. Especially, CD19-targeted CAR (chimeric antigen receptor)-expressing T cell gene therapy has achieved a great success in the treatment of relapsed/refractory B cell malignancies. Promising technology for gene therapy in the near future is genome editing, and its clinical application has already started for gene therapy of HIV infection and CAR gene therapy. Hereditary diseases will be the next target of genome editing technology. These topics will be presented in this Gene Therapy session.

Tue(3)-CIS16-1
Haematopoietic stem cell- and liver-targeted gene therapy for hereditary disease
Ian E. Alexander
Gene Therapy Research Unit, Sydney Children's Hospitals Network and Children's Medical Research Institute; University of Sydney Medical School, Australia

Tue(3)-CIS16-2
AAV (adeno-associated virus) vector-mediated gene therapy for hereditary and non-hereditory diseases
Keiya Ozawa
Division of Genetic Therapeutics, The Advanced Clinical Research Center, Institute of Medical Science, The University of Tokyo, Japan

Tue(3)-CIS16-3
Gene editing - from modeling diseases to treating patients
Toni Cathomen
Institute for Cell and Gene Therapy, Medical Center - University of Freiburg, Germany

Tue(3)-CIS16-4
CAR Therapy: The CD19 Paradigm
Michel Sadelain
Center for Cell Engineering, Memorial Sloan Kettering Cancer Center, New York, USA

8:00-10:00 Room E

CIS17 Concurrent Invited Session 17
"ELSI"

Conveners: **Masayuki Yoshida**
Department of Life Science and Bioethics, Tokyo Medical and Dental University, Japan
Dina N. Paltoo
Genetics, Health and Society Program, Office of Science Policy/National Institute of Health, USA

Though future advances in genomic research require accessing and sharing of global genomic data, the sharing of such data continues to raise concerns regarding how to protect genomic privacy. The difficulty in balancing the sharing and protection genetic information is partially based on how the genomic data governance and management system or model ensures that cutting edge genomic research is available with a reasonable control of research participant's privacy. Current approaches to overcome this issue are rather heterogeneous depending upon the region. Additionally, the responsibilities of the researchers who access these data should also be considered.
In this workshop, we will learn of genomic data management practices and concerns from diverse geographic regions, in an effort to discuss the practical solutions to confront the issue of sharing and potential risks in doing so. We hope that this session will provide a meaningful vision to all stakeholders that ensures the promise of genetic research and its application to medicine and health.

Tue(3)-CIS17-1

Patient Preferences for Governance of Use of Genomic Information in Research

Sandra S. Lee
Stanford Center for Biomedical Ethics, Stanford University, USA

Tue(3)-CIS17-2

Given a Voice: An Update on the Lacks Family-NIH Partnership on Use of HeLa Genome Data

Dina N. Paltoo
Genetics, Health and Society Program, Office of Science Policy/National Institute of Health, USA

Tue(3)-CIS17-3

ELSI practices and regulations for collaboration and public participation in personal genome research

Kazuto Kato
Biomedical Ethics and Public Policy, Osaka University, Japan

Tue(3)-CIS17-4

Ethical theory and global challenges in the era of -omics and predictive medicine

Ruth F. Chadwick
Centre for Social Ethics and Policy, University of Manchester, UK

Tue(3)-CIS17-5

Ethical and Policy Issues in Human Genome Editing and Germline Modifications

Xiaomei Zhai
Peking Union Medical College and Chinese Academy of Medical Sciences, China

10:20-12:20 Room E

CIS18 Concurrent Invited Session 18
"Genetic Counseling/Education"

Conveners: **Kristine Barlow-Stewart**
Sydney Medical School, The University of Sydney; Royal North Shore Hospital, Australia
Junko Yotsumoto
Natural Science Division, Faculty of Core Research, Ochanomizu University; Showa University, Japan

Genetic counseling is defined as a communication process that aims to help people understand and adapt to the medical, psychological and familial implications of genetic contributions to health conditions (Resta et al JGC, 2006). Necessary for this process is the capacity of the practitioner to assess risk based on personal and/or family histories and/or genetic/genomic test information. They also need to utilize education and counseling skills to promote understanding of the condition and its inheritance, awareness of resources and adaptation to the risk or condition as well as informed choice regarding testing, management and prevention strategies where available, and related research. The session will present the experiences of genetic counselors from Japan, Australia, UK, USA and Malaysia in regard to some of these elements in the process in the genomic era.
An overview of the training and roles of genetic counselors in Japan will be provided, illustrated by the challenges faced when dealing with Variants of Unknown Significance (VUS) in BRCA testing.
Masters level training for genetic counselors is now available in Australia, Canada, China, Cuba, France, Israel, Japan, Malaysia, the Netherlands, Norway, the Philippines, Saudi Arabia, South Africa, Spain, Sweden, Taiwan, United States of America and the United Kingdom and their roles are expanding in many countries. However the rapid developments in genomics have created a knowledge gap in genetic counselors who have not graduated recently and the strategies to up-skill these practitioners in Australia and internationally will be reviewed. Similarly strategies to meet the genetics education needs of health professionals faced with the increasing mainstreaming of genomic medicine will be discussed as well as the resources available to promote education and decision-making of the community in regards to genetic and genomic tests and their implications.
These implications increasingly facing practitioners in the genomic era include addressing the potential for incidental findings and promoting informed choice in this regard as well as managing the return of such unexpected results. Genomic testing can be provided to those seeking a clinical service or to those taking part in research studies, and the views and experience of those utilizing and providing the test in both of these settings will be reviewed.
In regard to the clinical setting, genetic counselors working in the Clinical Sequencing Exploratory Research Consortium in the USA have developed recommendations in regard to best practice for facilitating informed consent in the pre-test counseling session which will be presented.
In terms of expectations of research participants in regard to return of results, the responses of 6944 individuals (both community and professionals) from 75 countries to a cross-sectional web-based survey will be presented.
However there are many additional challenges facing those practitioners in the implementation of genetic and genomic medicine in non-Western middle and low income countries including overcoming health system, access, health and genetic literacy, funding and cultural and psychosocial barriers. The experience of genetic counselors working in the field of cancer genetics Malaysia will be used to illustrate the interventions which can be put in place to address some of these barriers.

Tue(3)-CIS18-1

Introduction and brief overview of genetic counselling in Japan - Role and training of genetic counselors, and a topic, "dealing with BRCA VUS in Japan"

Junko Yotsumoto
Natural Science Division, Faculty of Core Research, Ochanomizu University; Showa University, Japan

Tue(3)-CIS18-2

Genetic Education Strategies to Enhance the Genetic Counselling process in the Genomic Era

Kristine K. Barlow-Stewart
Sydney Medical School, The University of Sydney; Royal North Shore Hospital, Australia

Tue(3)-CIS18-3

Genetic counselors' experiences obtaining informed consent for genomic sequencing: Lessons learned

Barbara A. Bernhardt
Translational Medicine and Human Genetics, University of Pennsylvania, USA

Tue(3)-CIS18-4

Engaging 7,000 people about the return of results from sequencing research

Anna Middleton
Social Science and Ethics, Wellcome Genome Campus, UK

Tue(3)-CIS18-5

Genetic Counselling and hereditary testing in low and middle income Asian setting

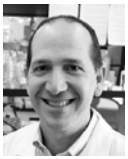
Sook Yee Yoon
Familial Cancer, Cancer Research Malaysia, Malaysia

8:00-10:00 Room B-1

CIS19 Concurrent Invited Session 19
"Psychiatric Genetics"

Conveners: **Norio Ozaki**
Department of Psychiatry, Nagoya University Graduate School of Medicine, Japan
Joseph D. Buxbaum
Icahn School of Medicine at Mount Sinai, USA

Psychiatric genetics explains the role of genes in mental disorders. The answers are not simple and the questions remain numerous and complex, but the results are exciting and have the potential to transform the practice of psychiatry and psychopharmacology. In this session the speakers will present the data about new paradigm for genes and psychiatry that has recently emerged. That paradigm sees genes only as direct causes of mental disorders but also as direct causes of undelaying molecular abnormalities that increase risk for onset of mental disorders. In other words, deleterious mutations do not cause mental disorders but can bias brain circuits toward inefficient information processing, which may lead to mental disorders under certain circumstances. It may be possible to identify critical genes to assess risk for mental illness in individual patients and their families, and this may someday help guide treatment selection as well.



Tue(3)-CIS19-1

Rare and common variation in autism and associated neurodevelopment disorders

Joseph D. Buxbaum
Icahn School of Medicine at Mount Sinai, USA

Tue(3)-CIS19-2

Genetics of schizophrenia and bipolar disorder

Michael J. Owen
MRC Centre for Neuropsychiatric Genetics and Genomics, Cardiff University, UK

Tue(3)-CIS19-3

Shared Genetic Risk Across Psychiatric Disorders

Naomi R. Wray
Queensland Brain Institute, The University of Queensland, Brisbane, Queensland, Australia

Tue(3)-CIS19-4

Pharmacogenomics in Psychiatry

Masashi Ikeda
Psychiatry, Fujita Health University School of Medicine, Japan

10:20-12:20 Room B-1

CIS20 **Concurrent Invited Session 20**

"Neurogenetics and Neurodegeneration"

Conveners: **Tatsushi Toda**
Division of Neurology, Kobe University Graduate School of Medicine, Japan
Bryan J. Traynor
Laboratory of Neurogenetics, National Institute on Aging, USA

This symposium focuses on the major neurological diseases that are mostly sporadic but partially familial with a Mendelian pattern of inheritance, including multiple system atrophy, Parkinson's disease, amyotrophic lateral sclerosis, frontotemporal dementia, and mitochondrial diseases.
There is currently no cure or treatment for those intractable neurological diseases. While the genetic background of the diseases has been partly revealed, much work remains to be done to find additional causative genes. A variety of approaches have been employed to identify these genes ranging from genome-wide association studies that examine common variation to next generation sequencing that is ideal for identifying rare variants.
The groups presenting during this symposium lead the world in applying advanced genomic methods to the intractable neurological disorders to identify genes. Through their analyses, they are aiming to unveil the pathogenesis of these diseases and develop effective cures.

Tue(3)-CIS20-1

Molecular genetic basis of multiple system atrophy

Jun Mitsui
Neurology, The University of Tokyo, Japan

Tue(3)-CIS20-2

Genetics of Parkinson's Disease

Tatsushi Toda
Division of Neurology/Molecular Brain Science, Kobe University Graduate School of Medicine, Japan

Tue(3)-CIS20-3

Genetics of mitochondrial diseases

Patrick F. Chinnery[1,2]
[1]University of Cambridge, UK, [2]MRC Mitochondrial Biology Unit

Tue(3)-CIS20-4

Genomics of amyotrophic lateral sclerosis and frontotemporal dementia

Bryan J. Traynor
Laboratory of Neurogenetics, National Institute on Aging, USA

13:50-15:20 Room A

WS1 **Workshop 1**

"Global Data Sharing in Human Genetics"

Moderators: **Sharon F. Terry**
President and CEO
Genetic Alliance, USA

Kazuto Kato
Graduate School of Medicine, Osaka University, Japan

The issue of sharing human genetic data in any single country is filled with complexity. In a global context it is even more complicated, as culture, context, laws, regulations, directives, and customs vary greatly. For some countries, genetic data can be simply used like other health data, and in others, it represents the essence of being human. Regardless of a country's oversight of the matter, individual and community sensibilities vary a great deal even within national boundaries. This workshop will provide a sampling of some of the issues relative to different countries and cultures. Kazuto Kato and Sharon Terry will set the stage for this presentation with a short overview of the general issues related to sharing genetic and genomic data. They will present this overview from policy, ethics, and consumer perspectives. Bartha Knoppers will present on the work she has led for the Global Alliance for Genomes and Health (GA4GH). In this framework she developed for the GA4GH, she details using a human rights framework to guide data sharing in the countries participating in GA4GH. Victor Penchaszadah will discuss north-south inequities in global sharing of genetic data. The Southern Hemisphere generally is home to emerging nations who are less able to bear the same potential burdens as do citizens of the Northern Hemisphere. It is not clear to what extent global sharing is actually global, and so Charles Rotimi will describe his work in this area. Partha Majumder will present the point of view of individuals working in India and in Asia in general.

Tue(3)-WS1-1

Global Genomic Data Sharing for Health: A Human Rights Approach
Bartha M. Knoppers
Human Genetics, McGill University, Centre of Genomics and Policy, Canada

Tue(3)-WS1-2

North-South inequities in global sharing of human genetic data
Victor B. Penchaszadeh
Latin American Bioethics Network, Argentina

Tue(3)-WS1-3

How Global is "Global Data sharing"
Charles N. Rotimi
Center for Research on Genomics and Global Health, National Human Genome Research Institute, NIH, USA

Tue(3)-WS1-4

Sharing Big Data: A Personal Perspective from a Developing Nation
Partha Majumder
National Institute of Biomedical Genomics, India

8:00-10:00 Room D

ED3 Educational Program 3
"Case Studies in Clinical Genetics: Dysmorphology"

Moderators: **Nobuhiko Okamoto**
Department of Medical Genetics, Osaka Medical Center and Research Institute for Maternal and Child Health, Osaka, Japan
Ritsuko K. Pooh
CRIFM Clinical Research Institute of Fetal Medicine PMC, Osaka, Japan

It has been estimated that about 3% of neonates are born with congenital malformations that are responsible for 20–30% of neonatal and 35% of infantile deaths. Dysmorphic syndromes are defined as the constellation of anomalies that are observed in combination more frequently than they are estimated to occur together by chance. Every syndrome has its characteristic problems in growth, development, and behavior. Correct diagnosis and appropriate medical care is important in these patients. Dysmorphology is the study of syndromes that constitute recognizable patterns of anomalies. We will present educational cases. All attendants are encouraged to consider appropriate diagnosis together.
The prevalence of cerebral palsy has not decreased despite major improvements in clinical care in antenatal/neonatal period as well as intrapartum period. The antepartum risk factors should include fetal brain maldevelopment and intrauterine brain injuries, which are unclassifiable into congenital brain anomalies and may exist unconspicuously during pregnancy and even after birth. Especially, neuronal migration disorder in utero should be responsible for postnatal neurological impairment. Imaging technologies including 3D ultrasound have been remarkably improved and contributed to prenatal evaluation of fetal central nervous system (CNS) development and assessment of CNS abnormalities in utero. Migration takes place in the first and early-second trimesters and phenotype of migration in the cortex appears after 28 weeks. It has been believed that migration disorder such as lissencephaly cannot be detected before 28 weeks. However, recent neuroimaging has enabled us to suspect migration disorder from early-second trimester. Fetal neurology has great responsibility and an important role in perinatal medicine and a new field of fetal neuro-sono-genetics will be established.

Presenters/Discussants:

Tue(3)-ED3-1

Nobuhiko Okamoto
Department of Medical Genetics, Osaka Medical Center and Research Institute for Maternal and Child Health, Osaka, Japan

Tue(3)-ED3-2

Ritsuko K. Pooh
CRIFM Clinical Research Institute of Fetal Medicine PMC, Osaka, Japan

Tue(3)-ED3-3

Louanne Hudgins
Stanford University and Lucile Packard Children's Hospital, USA

10:20-12:20 Room D

ED4 Educational Program 4
"Case Studies in Clinical Genetics: Lysosomal Storage Disease"

Moderators: **Norio Sakai**
Osaka University Graduate School of Medicine, Division of Health Sciences, Japan
Han-Wook Yoo
Medical Genetics Center, Asan Medical Center, University of Ulsan College of Medicine, Seoul, Korea

Recent development of treatment for lysosomal storage disease (LSD) makes challenge for physical doctors who meet these rare diseases. Establishment of enzyme replacement therapy and hematopoietic stem cell transplantation, development of substrate reduction therapy and chaperone therapy are leading the change of prognosis of many LSDs. Physical doctor are expected to diagnose the patients precisely and promptly in order to start the effective treatment and supply the important genetic counseling for the family.
In this session, we pick up four categories of disease within the LSDs. It includes mucopolysaccharidosis, Gaucher disease, leukodystrophy (metachromatic leukodystrophy, Krabbe disease and adrenoleukodystrophy) and Fbry disease. Case presenter shows the case history with clinical hints for diagnosis and the discussant picks up the points and asks several questions to the audience. All attendants are expected to join the voting system to answer these questions. After the voting, presenter answers the questions and talks next clinical scenario including treatment, genetic counseling or prenatal diagnosis etc.
Audience should be involved in considering the best attitude of physical doctors in these setting. Organizers expect all attendants participate positively to this program and hope effective discussion between the presenters and discussants.

Presenters:

Tue(3)-ED4-1
MPS
Motomichi Kosuga
Department of Clinical Laboratory Medicine, National Center for Child Health and Development, Japan

Tue(3)-ED4-2
Gaucher
Han-Wook Yoo
Medical Genetics Center, Asan Medical Center, University of Ulsan College of Medicine, Seoul, Korea

Tue(3)-ED4-3
Luekodystrophy
Norio Sakai
Osaka University Graduate School of Medicine, division of Health Sciences, Japan

Tue(3)-ED4-4
Fabry
Kimitoshi Nakamura
Department of Pediatrics, Kumamoto University, Japan

Discussants:

Norio Sakai for 1 and 2
Osaka University Graduate School of Medicine, division of Health Sciences, Japan

Han-Wook Yoo for 3 and 4
Medical Genetics Center, Asan Medical Center, University of Ulsan College of Medicine, Seoul, Korea

13:50-15:20 Room D

ED5 Educational Program 5
"Case Studies in Clinical Genetics: Cancer"

Moderators: **Takashi Kohno**
National Cancer Center Research Institute, Japan
Yoshinori Murakami
Institute of Medical Science, The University of Tokyo, Japan

This session will focus on two important themes in the clinical practice of cancer, decision making based on identification of somatic mutations (part 1) and genetic counseling for familial cancer (part 2). The clinical information will be first presented, followed by decisions/discussion based on clinical sequencing of tumor and cell-free DNAs by the discussant and the audience. Latest information on somatic mutations in lung and other cancers and selection of therapeutic ways, including the entry into clinical trials, will be presented. In the genetic counseling for breast cancer, the clinical information of cases and their family history suggesting possible hereditary breast and ovarian cancer (HBOC) will be first presented. Various issues on genetic counseling, including the genetic test and its possible application to prophylaxis and treatment of cancer will be discussed.

Part 1: Genomic Medicine of Cancer on the Basis of the Somatic Mutations

Presenters:

Tue(3)-ED5-1

Takashi Kohno
National Cancer Center Research Institute, Japan

Tue(3)-ED5-2

Sadakatsu Ikeda
Center for Personalized Cancer Therapy, University of California San Diego, Moores Cancer Center, USA

Discussant:

Kuniko Sunami
National Cancer Research Center Hospital, Japan

Part 2: Genetic Counseling of Breast Cancer

Presenter:

Tue(3)-ED5-3

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

Discussants:

Seigo Nakamura
Department of Breast Surgical Oncology, Showa University School of Medicine, Japan

Chieko Tamura
Certified Genetic Counselor, Japan / USA; Medical Information & Genetic Counseling Division, FMC Tokyo Clinic, Japan

15:40-17:10 Room D

ED6 Educational Program 6
"Case Studies in Clinical Genetics: Neurology"

Moderators: **Thomas Gasser**
German Center for Neurodegenerative Diseases (DZNE), Germany
Hiroyuki Ishiura
The University of Tokyo, Japan

Genetic analysis has often an important role in clinical neurology. We have to appropriately use sequence analysis technologies for making a molecular diagnosis, appropriate management of patient care including molecular therapies, and providing genetic counseling. In this program, three cases will be presented. After case presentations, presenters provide several choices on further clinical testing required, genetic diagnosis at the first glance, recommended methods of genetic testing, and so on. Participants in the floor will make the online votes with their smartphones or laptop computers. After experts' discussion, presenters provide genetic diagnosis of the patients including an overview and an update of the disease. The program is aimed mainly at neurologists or geneticists including beginners.
Please bring your own smartphones or laptop computers.

Presenters:

Tue(3)-ED6-1
Takashi Matsukawa
Department of Neurology, Graduate School of Medicine, The University of Tokyo, Japan

Tue(3)-ED6-2
Masaki Tanaka
Department of Neurology, The University of Tokyo, Japan

Tue(3)-ED6-3
Yoshio Sakiyama
Department of Neurology, Jichi Medical University, Saitama Medical Center, Japan

Discussants:

Bing-wen Soong
Department of Neurology, National Yand-Ming University, Taipei, Taiwan

Hiroshi Takashima
Neurology and Geriatrics, Kagoshima University, Japan

13:50-15:20 Annex 1

YIA1 Young Investigator Awards Session 1

Chairs: **Brunhilde Wirth**
Institute of Human Genetics, University of Cologne, Germany
Stephen T.S. Lam
Faculty of Medicine, The Chinese University of Hong Kong, China

Tue(3)-YIA1-1

Association between the literacy on genomics and health status; encouraging genomics education in personalized preventive medicine era

Sho Nakamura[1,2], Hiroto Narimatsu[2,3], Kayoko Katayama[2], Ri Sho[3], Ryo Kawasaki[3], Akira Fukao[3], Takashi Yoshioka[1], Takamasa Kayama[4]
[1]Department of Clinical Oncology, Yamagata University Faculty of Medicine, Japan, [2]Cancer Prevention and Control Division, Kanagawa Cancer Center Research Institute, [3]Department of Public Health, Yamagata University Graduate School of Medical Science, [4]Department of Advanced Cancer Science, Yamagata University Faculty of Medicine

Tue(3)-YIA1-2

A molecular study on Stevens-Johnson syndrome patients with ocular manifestations

Sushil Kumari Sangwan[1], Arundhati Sharma[1], Namrata Sharma[2], Neena Khanna[3], Tushar Agarwal[2], Rasik B Vajpayee[4]
[1]Department of Anatomy, All India Institute of Medical Sciences, New Delhi, India, [2]Dr. Rajendra Prasad Centre for Ophthalmic Sciences, AIIMS, New Delhi, [3]Department of Dermatology and Venerology, AIIMS, New Delhi, [4]Center for Eye Research,University of Melbourne, Australia

Tue(3)-YIA1-3

Investigation of Variants within Antipsychotic Pharmacogenes Associated with Treatment Outcome in a South African First Episode Schizophrenia Cohort

Faatiemah Higgins[1], Britt I Drögemöller [5], Galen EB Wright[5], Lize Van der Merwe[3,4], Bonga Chiliza[2], Laila Asmal[2], Dana Niehaus[2], Robin Emsley[2], Louise Warnich[1]
[1]Genetics, Stellenbosch University, South Africa, [2]Psychiatry, Stellenbosch University, [3]Statistics, Stellenbosch University, [4]Molecular Biology and Human Genetics, Stellenbosch University, [5]Paediatrics, University of British Columbia

Tue(3)-YIA1-4

Biallelic truncating mutations in *ALPK3* cause severe pediatric cardiomyopathy

Judith M.A. Verhagen[1], Rowida Almomani[2], Johanna C. Herkert[2], Erwin Brosens[1], Karin Y. van Spaendonck-Zwarts[2,3], Angeliki Asimaki[4], Paul A. van der Zwaag[2], Ingrid M.E. Frohn-Mulder[5], Aida M. Bertoli-Avella[1,6], Ludolf G. Boven[2], Marjon A. van Slegtenhorst[1], Jasper J. van der Smagt[7], Wilfred F.J. van IJcken[8], Bert Timmer[9], Margriet van Stuijvenberg[10], Rob M. Verdijk[11], Jeffrey E. Saffitz[4], Frederik A. du Plessis[5], Michelle Michels[12], Robert M.W. Hofstra[1], Richard J. Sinke[2]
[1]Department of Clinical Genetics, Erasmus University Medical Center, Rotterdam, the Netherlands, Netherlands, [2]University of Groningen, University Medical Center Groningen, Department of Genetics, Groningen, the Netherlands, [3]Department of Clinical Genetics, Academic Medical Center, University of Amsterdam, Amsterdam, the Netherlands, [4]Department of Pathology, Harvard Medical School, Beth Israel Deaconess Medical Center, Boston, USA, [5]Department of Pediatric Cardiology, Erasmus University Medical Center, Rotterdam, the Netherlands, [6]Centogene AG, Rostock, Germany, [7]Department of Medical Genetics, University Medical Center Utrecht, Utrecht, The Netherlands, [8]Center for Biomics, Erasmus University Medical Center, Rotterdam, the Netherlands, [9]University of Groningen, University Medical Center Groningen, Department of Pathology and Medical Biology, Groningen, the Netherlands, [10]University of Groningen, University Medical Center Groningen, Division of Neonatology, Beatrix Children's Hospital, Groningen, the Netherlands, [11]Department of Pathology, Erasmus University Medical Center, Rotterdam, the Netherlands, [12]Department of Cardiology, Erasmus University Medical Center, Rotterdam, the Netherlands

Tue(3)-YIA1-5

Genetic association study identifies common variation in *PHACTR1* to associate with fibromuscular dysplasia

Nabila Bouatia-Naji[1,2,3], Soto Romuald Kiando[1,2,3], Nathan R Tucker[4], Cyrielle Treard[1,2,3], Luis J Castro-Vega[1,2,3], Cristina Barlasina[8], Daniele Cusi[8], Pilar Galan[9], Jean-Philippe Empana[1,2,3], Xavier Jouven[1,2,3,10], Jeffrey W Olin[6], Heather L Gornik[11], Pierre-Francois Plouin[12], Iftikhar J Kullo[7], David J Milan[4], Santhi K Ganesh[5], Pierre Boutouyrie[1,2,3,13], Jason Kovacic[6], Xavier Jeunemaitre[1,2,3,14]

[1]Paris Cardiovascular Research Center, INSERM, France, [2]INSERM UMR970, Paris, France, [3]Faculty of medicine, Paris-Descartes University, Sorbonne Paris Cite, [4]Cardiovascular research Center, Massachusetts General Hospital, Charlestown, MA, USA, [5]Department of Internal Medicine and Department of Human Genetics, University of Michigan, Ann Arbor, MI, USA., [6]Zena and Michael A. Wiener Cardiovascular Institute & Marie-Josee and Henry R. Kravis Center for Cardiovascular Health, Icahn School of Medicine at Mount Sinai, New York, NY, USA, [7]Department of Medicine, Division of Cardiovascular Diseases, Mayo Clinic, Rochester, Minnesota, USA, [8]Dept. of Health Sciences, Genomic and Bioinformatics Unit, School of Nephrology, University of Milano, Institute of Biomedical Technologies, Italian National Centre of Research, Italy, [9]Nutritional Epidemiology Research Group, Sorbonne-Paris-Cite, UMR University of Paris, France, [10]AP-HP, Department of Cardiology, Hopital Europeen Georges Pompidou, Paris, France, [11]Cleveland Clinic Heart and Vascular Institute, Cleveland, OH, USA., [12]AP-HP, Department of Hypertension, Hopital Europeen Georges Pompidou, Paris, France, [13]AP-HP, Department of Pharmacology, Hopital Europeen Georges Pompidou, Paris, France, [14]AP-HP, Refferal Center for Rare Vascular Diseases, Hopital Europeen Georges Pompidou, Paris, France

Tue(3)-YIA1-6

Size-based molecular diagnostics using plasma DNA for noninvasive prenatal testing

Stephanie C. Y. Yu[1,2], K. C. Allen Chan[1,2], Yama W. L. Zheng[1,2], Peiyong Jiang[1,2], Gary J. W. Liao[1,2], Hao Sun[1,2], Ranjit Akolekar[3], Tak Y. Leung[4], Attie T. J. I. Go[5], John M. G. van Vugt[5], Ryoko Minekawa[3], Cees B. M. Oudejans[5], Kypros H. Nicolaides[3], Rossa W. K. Chiu[1,2], Y. M. Dennis Lo[1,2]

[1]Department of Chemical Pathology, The Chinese University of Hong Kong, Prince of Wales Hospital, Shatin, NT, Hong Kong SAR, China, Hong Kong, [2]Centre for Research into Circulating Fetal Nucleic Acids, Li Ka Shing Institute of Health Sciences, The Chinese University of Hong Kong, Shatin, NT, Hong Kong SAR, China, [3]Harris Birthright Research Centre for Fetal Medicine, King's College Hospital, London, United Kingdom, [4]Department of Obstetrics and Gynaecology, The Chinese University of Hong Kong, Prince of Wales Hospital, Shatin, NT, Hong Kong SAR, China, [5]Department of Clinical Chemistry, VU University Medical Center, Amsterdam, The Netherlands

15:40-17:10 Annex 1

YIA2 Young Investigator Awards Session 2

Chairs: **Joris A. Veltman**
Human Genetics, Radboud University Medical Centre, The Netherlands
Naomichi Matsumoto
Department of Human Genetics, Yokohama City University Graduate School of Medicine, Japan

Tue(3)-YIA2-1

LAT2 transporter is involved in age-related hearing loss

Meritxell Espino Guarch[1,2,3], Mariona Font[2], Giorgia Girotto[1], Ekaitz Errasti[3], Clara Vilches[2], Silvia Murillo[4], Paolo Gasparini[1], Manuel Palacin[3,5], Virginia Nunes[2,5]

[1]Experimental Genetics, Sidra Medical and Research Center, Qatar, [2]Molecular Genetics Laboratory, Bellvitge Biomedical Research Institute (IDIBELL), Barcelona, Spain, [3]Institute of Research in Biomedicine (IRB), Barcelona, Spain, [4]Unit 761, Center for Biomedical Network Research on Rare Diseases (CIBERER), Madrid, Spain, [5]Biochemistry and Molecular Biology Department, Barcelona University (UB), Barcelona, Spain

Tue(3)-YIA2-2

Whole-genome sequencing of monozygotic twins discordant for schizophrenia

Yu Fan[1,2], Jinsong Tang[3], Qun Xiang[1,2], Hong Ll[3], Yong-Gang Yao[1,2], Xiaogang Chen[3]

[1]Kunming Institution of Zoology, Chinese Academy of Sciences, China, [2]Kunming College of Life Science, University of Chinese Academy of Sciences, [3]Institute of Mental Health, the Second Xiangya Hospital, Central South University

Tue(3)-YIA2-3

Towards clinical accreditation of structural variation calling from HiSeq X whole genome sequencing data

Andre E Minoche[1], Greg B Peters[2], Velimir Gayevskiy[1], Mike Field[3], Claire Horvat[4], Andreas Zankl[2], Diane Fatkin[4], Tony Roscioli[1], Marcel E Dinger[1], Mark J Cowley[1]

[1]Kinghorn Centre for Clinical Genomics, Garvan Institute of Medical Research, Australia, [2]Sydney Genome Diagnostics, Children's Hospital Westmead, [3]NSW Health, Royal North Shore Hospital, [4]Victor Chang Cardiac Research Institute

Tue(3)-YIA2-4

Thousands of novel translated open reading frames in humans inferred by ribosome footprint profiling

Anil Raj[1], Sidney H Wang[2], Heejung Shim[6], Arbel Harpak[4], Yang I Li[1], Brett Engelmann[2], Matthew Stephens[2,3], Yoav Gilad[2], Jonathan K Pritchard[1,4,5]

[1]Department of Genetics, Stanford University, USA, [2]Department of Human Genetics, University of Chicago, [3]Department of Statistics, University of Chicago, [4]Department of Biology, Stanford University, [5]Howard Hughes Medical Institute, [6]Department of Statistics, Purdue University

Tue(3)-YIA2-5

Visualizing structural variation at the single cell level to explore human genome heterogeneity

Ashley D Sanders[1], Mark Hills[1], David Porubsky[3], Victor Guryev[3], Ester Falconer[1], Peter M Lansdorp[1,2,3]

[1]BC Cancer Agency, University of British Columbia, Canada, [2]Division of Hematology, Department of Medicine, University of British Columbia, [3]European Research Institute for the Biology of Ageing, University of Groningen, University Medical Centre Groningen

Tue(3)-YIA2-6

Genome-wide multi-phenotype analysis of rare variants boosts power for locus discovery and indicates novel rare variant effects from a known common variant locus on omega fatty acids

Marika Kaakinen[1], Annique Claringbould[2], Reedik Magi[3], Krista Fischer[3], Mika Ala-Korpela[4,5,6], Marjo-Riitta Jarvelin[4,6,7,8], Andrew P. Morris[9], Inga Prokopenko[1]

[1]Genomics of Common Disease, Imperial College London, UK, [2]Department of Genetics, University Medical Centre Groningen, [3]Estonian Genome Center, University of Tartu, [4]Center for Life Course Epidemiology and Systems Medicine, University of Oulu, [5]Computational Medicine, School of Social and Community Medicine and the Medical Research Council Integrative Epidemiology Unit, University of Bristol, [6]Unit of Primary Care, Oulu University Hospital, [7]Biocenter Oulu, University of Oulu, [8]Department of Epidemiology and Biostatistics, MRC-PHE Centre for Environment and Health, Imperial College London, [9]Department of Biostatistics, University of Liverpool

13:50-15:20 Annex 2

O13 Concurrent Oral Session 13
"Cancer Genetics 2"

Chairs: **Stacey Edwards**
Genetics and Computational Biology, QIMR Berghofer Medical Research Institute, Australia
Hiroyuki Aburatani
Research Center for Advanced Science and Technology, The University of Tokyo, Japan

Tue(3)-O13-1

Non-random occurrence and early age of onset of diverse lymphoid cancers in families supports the existence of genetic risk factors for multiple lymphoid cancers

Samantha Jones[1,2]
[1]*Medical Genetics, University of British Columbia, Canada,* [2]*Cancer Genetics, British Columbia Cancer Agency*

Tue(3)-O13-2

Targeted TET oxidase activity through methyl-CpG binding domain extensively suppresses cancer cell proliferation

Shinichi Fukushige, Yasuhiko Mizuguchi, Kanchan Chakma, Yuriko Saiki, Akira Horii
Department of Molecular Pathology, Tohoku University School of Medicine, Japan

Tue(3)-O13-3

Testing of Deletions or Excess Homozygosity for Head and Neck Cancer Association in Whole Genome SNP Genotyping Studies

Chih-Chieh Wu[1], Sanjay Shete[2]
[1]*College of Medicine, Department of Environmental and Occupational Health, National Cheng Kung University, Taiwan,* [2]*Department of Biostatistics, The University of Texas MD Anderson Cancer Center*

Tue(3)-O13-4

Five independent breast cancer risk variants at 6q25 display genotype-phenotype correlations and regulate *ESR1* and *RMND1*

Stacey Edwards[1], Alison Dunning[2], Kyriaki Michailidou[3], Karoline Kuchenbaecker[3], Deborah Thompson[3], Juliet French[1], Jonathan Beesley[1], Catherine Healy[2], Siddhartha Kar[2], Richard Sallari[4], Elena Lopez-Knowles[5,6], Mitch Dowsett[5,6], Paul Pharoah[2,3], Jacques Simard[7], Per Hall[8], Montserrat Garcia-Closas[9,10], Celine Vachon[11], Georgia Chenevix-Trench[1], Antonis Antoniou[3], Douglas Easton[2,3]
[1]*QIMR Berghofer Medical Research Institute, Australia,* [2]*Department of Oncology, University of Cambridge, UK,* [3]*Department of Public Health and Primary Care, University of Cambridge, UK,* [4]*Computer Science and Artificial Intelligence Laboratory, Massachusetts Institute of Technology, Cambridge, MA, USA,* [5]*Breast Cancer Research, Breakthrough Breast Cancer Research Centre, UK,* [6]*Academic Biochemistry, Royal Marsden Hospital, UK,* [7]*Centre Hospitalier Universitaire de Québec Research Center, Laval University, Canada,* [8]*Department of Medical Epidemiology and Biostatistics, Karolinska Institutet, Sweden,* [9]*Division of Cancer Studies, Breakthrough Breast Cancer Research Centre, Institute of Cancer Research, UK,* [10]*Division of Genetics and Epidemiology, Institute of Cancer Research, UK,* [11]*Department of Health Sciences Research, Mayo Clinic, Rochester, USA*

Tue(3)-O13-5

Breast cancer pedigree exome sequencing reveals inherited RAD52 truncation mutation implicated in breast cancer susceptibility

Helio A Costa[1,2], Martin Sikora[3], Kedar Hastak[2], James M Ford[2], Louise C Laurent[4], Carlos D Bustamante[2]
[1]*Genetics, Stanford University, USA,* [2]*Stanford University School of Medicine, Department of Genetics, Stanford, CA, USA,* [3]*Natural History Museum of Denmark, Centre for GeoGenetics, Copenhagen, Denmark,* [4]*University of California, San Diego, Department of Reproductive Medicine, La Jolla, CA, USA*

Tue(3)-O13-6

High miR-30d expression associates with improved breast cancer survival

Maral Jamshidi, Rainer Fagerholm, Sippy Kaur, Sofia Khan, Eliisa Ollikainen, Johanna Kiiski, Kristiina Aittomaki, Paivi Heikkila, Ralf Butzow, Carl Blomqvist, Heli Nevanlinna
University of Helsinki and Helsinki University Hospital, Finland

15:40-17:10 Annex 2

O14 Concurrent Oral Session 14 "Cancer Genetics 3"

Chairs: **Denise A.S. Batista**
Department of Pathology, Johns Hopkins University, USA
Seigo Nakamura
Division of Breast Surgical Oncology, Showa University, Japan

Tue(3)-O14-1

Clinical analysis of founder mutations of *BRCA1* and *BRCA2* in the Japanese population

Reiko Yoshida[1], Shiro Yokoyama[1], Chie Watanabe[2], Mayuko Inuzuka[1], Junko Yotsumoto[4], Masami Arai[3], Seigo Nakamura[1], The registration committee of The Japanese HBOC consortium
[1]Breast center, Showa University, Japan, [2]Sophia University, Faculty of Human Sciences, [3]Cancer Institute Hospital, Division of Clinical Genetic Oncology, [4]Ochanomizu University, Natural Science Division, Faculty of Core Research

Tue(3)-O14-2

Exome sequencing reveals new potential markers of therapy efficacy and safe cancellation of targeted therapy in patients with chronic myeloid leukemia

Sergey I Kutsev[1,2], Svetlana A Smirnikhina[1], Elmira P Adilgereeva[1], Ekaterina Y Chelysheva[3], Oleg A Shukhov[3], Anna G Turkina[3], Alexander V. Lavrov[1,2]
[1]Laboratory of Mutagenesis, Research Center for Medical Genetics, Russia, [2]Russian National Research Medical University, [3]National Research Center for Hematology

Tue(3)-O14-3

Next-generation sequencing to analyze ABL1 tyrosine kinase domain mutations in targeted therapeutic chronic myelogenic leukemia patients

Chinh Q Duong, Hang T Pham, Trang T Nguyen, Hoang C Tran, Tuong Q Le, Khanh Q Bach, Tri A Nguyen
Department of Genetics and Molecular Biology, National Institute of Haematology and Blood Transfusion, Vietnam

Tue(3)-O14-4

Increasing diagnostic yield: Addition of next generation sequencing panel to chromosome microarray and karyotype in myeloid leukemia

Denise A.S. Batista[1], Elizabeth Wohler[2], Kerry Powell[2], Victoria Stinnett[2], Yi Ning[1]
[1]Pathology, Johns Hopkins University School of Medicine, USA, [2]Pathology, Johns Hopkins Hospital

Tue(3)-O14-5

The Effects Of *JAK2V617F, MPL* and *CALR* Mutations On Diagnosis, Classification, Frequency, Laboratory Results and Clinical Status In Myeloproliferative Neoplasms

Hatice Akar, Deniz Torun, Yusuf Tunca
Medical Genetics, Gulhane Military Medical Academy, Turkey

Tue(3)-O14-6

Germline variants in pediatric leukemia detected by next generation sequence

Akira Shimada[1], Hiromu Narasaki[1], Takae Hanada[1], Ritsuo Nisiuchi[2]
[1]*Pediatrics, Okayama University Hospital, Japan,* [2]*Pediatrics, Kouchi Medical Center*

15:40-17:10 Room A

O15 Concurrent Oral Session 15
"Prenatal, Perinatal and Reproductive Genetics 2"

Chairs: **Marieke Joosten**
Clinical Genetics, Erasmus MC, Rotterdam, Netherlands
Takahiro Yamada
Department of Obstetrics, Hokkaido University Graduate School of Medicine, Japan

Tue(3)-O15-1

A case report of management including perinatal genetic counseling for May Hegglin Anomaly in pregnancy that low platelets counts made the opportunity to diagnose

Yuka Yamashita[1], Rei Matsuura[1], Yoshie Oikawa[2], Shoko Hamada[1], Hirotugu Ariizumi[2], Kei Odawara[1], Maya Koyano[1], Shogo Nishii[1], Tsutomu Muramoto[1], Shin Takenaka[1], Ken Nakayama[1], Kaori Matsumoto[1], Mitsuyoshi Ichihara[1], Yasushi Sasaki[1], Nahoko Shiroto[4], Ryu Matsuoka[4], Kouichi Ogawa[1], Akihiko Sekizawa[3], Shinji Kunishima[5]
[1]*Department of Obstetrics and Gynecology, Showa University Fujigaoka Hospital, Japan,* [2]*Departnebt of Clinical Laboratory, Showa University Fujigaoka Hospital,* [3]*Department of Hematology, Showa University Fujigaoka Hospital,* [4]*Department of Obstetrics and Gynecology, Showa University Hospital,* [5]*Department of Hemostasis and Thrombosis, Clinical Research Center, National Hospital Organization Nagoya Medical Center*

Tue(3)-O15-2

Uniparental disomy (UPD) 14 diagnosed by SNP microarray at 16 week amniotic fluid showed distinctive ultrasonic finding from early 2nd trimester; a case report

Norio Shinozuka[1,2], Sena Eda[2], Akinori Taguchi[1,2], Hiroshi Seto[1], Shoji Okajima[3]
[1]*OBGYN, Seto Hospital, Japan,* [2]*Clin. Genetics, Seto Hospital,* [3]*LabCorp Japan*

Tue(3)-O15-3

Prenatal whole genome SNP array diagnosis: relevance of incidental findings in pregnancies with and without ultrasound anomalies

Marieke Joosten[1], Karin EM Diderich[1], Diane Van Opstal[1], Lutgarde CP Govaerts[1], Sam R Riedijk[1], Krista Prinsen[2], Femke AT de Vries[1], Robert-Jan H Galjaard[1], Malgorzata I Srebniak[1]
[1]*Clinical Genetics, Erasmus MC, Netherlands,* [2]*Gynaecology and Obstetrics, Erasmus MC*

Tue(3)-O15-4

Postnatal and prenatal diagnosis for neonatal intrahepatic cholestasis caused by citrin deficiency

Nguyen T.M Huong[1], Nguyen P.A Hoa[2], Ngo D Ngoc[1], Nguyen T.P Mai[1], Ly T.T Ha[1], Ngo M Tien[1], Le T Hai[1,2], Vu D Quang[1]
[1]*Human Genetics Department, National Hospital of Pediatrics, Vietnam,* [2]*Hepatology Department*

Tue(3)-O15-5

Prenatal Counseling and Diagnosis of Gaucher Disease In Egypt: 15 Years Experience

Ahmed A.L. Aboulnasr[1], Ekram A.M. Fateen[2]
[1]*Obstetrics and Gynecology, Faculty of Medicine, Cairo University, Egypt,* [2]*Biochemical Genetics Department, National Research Centre, Cairo, Egypt*

Tue(3)-O15-6

Serious complex-heart disease is hardly predicted by prenatal genetic screening

Mika Saito[1], Taku Ishii[1], Yuuji Hamamichi[1], Akio Inage[1], Yuuki Nakamoto[1], Tomomi Ueda[1], Satoshi Yazaki[1], Tadahiro Yoshikawa[1], Ryo Suzuki[2], Yoshinori Maeda[2], Ikuno Kawabata[2], Atsushi Yoshida[2], Shinji Katsuragi[2], Gengi Satomi[3]

[1]Pediatric Cardiology, Sakakibara Heart Institute, Japan, [2]Obstetric and gynecology, Sakakibara Heart Institute, [3]Satomi Clinic

13:50-15:20 Room E

O16 Concurrent Oral Session 16
"Bioinformatics and Genomic Technology 2"

Chairs: **Elizabeth Hauser**
Duke Molecular Physiology Institute, Duke University, USA
Tatsuhiko Tsunoda
Department of Medical Science Mathematics, Medical Research Institute, Tokyo Medical and Dental University, Japan

Tue(3)-O16-1

Coding and non-coding transcriptomic landscape of human brain: preliminary analysis of RNA sequencing data

Chao Chen[1], Yan Xia[1], Chuan Jiao[1], Amber Thomas[2], Yongjun Wang[3], Lijun Cheng[3], Xiyao Long[3], Miguel Brown[2], Jason Grundstad[2], Annie Shieh[2], Kevin P. White[2], Chunyu Liu[3]

[1]The State Key Lab of Medical Genetics, Central South University, Changsha, China, [2]Institute for Genomics and Systems Biology, the University of Chicago, Chicago, USA, [3]Department of Psychiatry, University of Illinois at Chicago, Chicago, USA

Tue(3)-O16-2

Pleiotropic landscape of anthropometry inferred from 359 novel and 297 established loci discovered in 270,000 individuals

Xia Shen[1,2], Zheng Ning[1], Yakov Tsepilov[4,5,6], Xiao Wang[10], Peter K. Joshi[2], Masoud Shirali[2], Blair H. Smith[2,7], Lynne J. Hocking[2,8], Sandosh Padmanabhan[2,9], Caroline Hayward[2], David J. Porteous[2], James F. Wilson[2], Yudi Pawitan[1], Chris S. Haley[2], Yurii S. Aulchenko[2,3,4,5], Generation Scotland

[1]Medical Epidemiology and Biostatistics, Karolinska Institutet, Sweden, [2]University of Edinburgh, [3]PolyOmica, [4]Novosibirsk State University, [5]Institute of Cytology and Genetics SB RAS, [6]Helmholtz Zentrum Munchen - German Research Center for Environmental Health, [7]University of Dundee, [8]University of Aberdeen, [9]University of Glasgow, [10]Stockholm University

Tue(3)-O16-3

From Paris to Kyoto or from Dermatoglyphics to Exome Sequencing

Regina M. Zambrano, Yves Lacassie

Department of Pediatrics, Louisiana State University Health Sciences Center and Children's Hospital of New Orleans, USA

Tue(3)-O16-4

QTR1: An Enhanaced, Population-Centric Reference Genome Based on GRCh37 to Facilitate Precision Medicine in Qatar and the Middle East

Khalid Fakhro[1,2], Michelle Staudt[3], Amal Robay[2], Charbel Abi-Khalil[2], Ramin Badii[4], Ajayeb Al-Nabet[4], Jason Mezey[3,5], Ronald Crystal[3], Juan Rodriguez-Flores[3]

[1]Translational Medicine, Sidra Medical Research Center, Qatar, [2]Weill Cornell Medical College in Qatar, [3]Weill Cornell Medical College in New York, [4]Hamad Medical Corporation, [5]Cornell University

Tue(3)-O16-5

HOT or not: redefining the origin of high-occupancy target regions

Altuna Akalin, Katarzyna Wreczycka, Vedran Franke, Bora Uyar, Ricardo Wurmus
BIMSB, Max Delbrueck Center, Germany

Tue(3)-O16-6

Short inversion detection by splitting and re-aligning poorly mapped and unmapped next-generation sequencing reads

Ruoyan Chen, Yan Zhang, Wanling Yang
Paediatrics and Adolescent Medicine, The University of Hong Kong, China

15:40-17:10 Room E

O17 Concurrent Oral Session 17 "Bioinformatics and Genomic Technology 3"

Chairs: **Davide Cittaro**
Center for Translational Genomics and Bioinformatics, San Raffaele Hospital, Italy
Zhaoming Wang
Department of Computational Biology, St. Jude Children's Research Hospital, USA

Tue(3)-O17-1

Imputation of KIR types from SNP variation data

Damjan Vukcevic[1,2], James A. Traherne[3,4], Sigrid Næss[5,6], Eva Ellinghaus[7], Yoichiro Kamatani[8,9], Alexander Dilthey[10], Mark Lathrop[8,11], Tom H. Karlsen[5,12], Andre Franke[7], Miriam Moffatt[13], William Cookson[13], John Trowsdale[3,4], Gil McVean[10], Stephen Sawcer[14], Stephen Leslie[1,2]
[1]Statistical Genetics, Murdoch Childrens Research Institute, Australia, [2]School of Mathematics and Statistics, University of Melbourne, Australia, [3]Cambridge Institute for Medical Research, University of Cambridge, Cambridge, UK, [4]Division of Immunology, Department of Pathology, University of Cambridge, Cambridge, UK, [5]Research Institute of Internal Medicine, Department of Cancer Medicine, Surgery and Transplantation, Oslo University Hospital Rikshospitalet, Oslo, Norway, [6]Norwegian PSC Research Center, Division of Cancer, Surgery and Transplantation, Oslo University Hospital, Oslo, Norway, [7]Institute of Clinical Molecular Biology, Christian-Albrechts-University of Kiel, Schittenhelmstr, Germany, [8]Fondation Jean Dausset-CEPH, Paris, France, [9]RIKEN Center for Integrative Medical Sciences, Kanagawa, Japan, [10]Wellcome Trust Centre for Human Genetics, University of Oxford, UK, [11]McGill University and Génome Québec Innovation Centre, Montreal, Canada, [12]K.G. Jebsen Inflammation Research Centre, Institute of Clinical Medicine, University of Oslo, Oslo, Norway, [13]National Heart and Lung Institute, Imperial College London, Royal Brompton Campus, UK, [14]Department of Clinical Neurosciences, University of Cambridge, Cambridge, UK

Tue(3)-O17-2

Diagnostic Role of Exome Sequencing in Immune Deficiency Disorders

Steven E Brenner[1], Aashish N Adhikari[1], Jay P Patel[2], Alice Y Chan[3], Divya Punwani[3], Haopeng Wang[3], Antonia Kwan[3], Theresa A Kadlecek[3], Morton J Cowan[3], Marianne Mollenauer[3], John Kuriyan[1], Shu Man Fu[4], Uma Sunderam[5], Sadhna Rana[5], Ajithavalli Chellappan[5], Kunal Kundu[5], Arend Mulder[6], Frans HJ Claas[6], Joseph A Church[7], Arthur Weiss[3], Richard Gatti[8], Jennifer Puck[3], Rajgopal Srinivasan[5]
[1]University of California, Berkeley, USA, [2]Children's Hospital of Los Angeles, [3]University of California, San Francisco, [4]University of Virginia School of Medicine, [5]Innovation Labs, Tata Consultancy Services, [6]Leiden University Medical Centre, [7]University of Southern California, [8]University of California, Los Angeles

Tue(3)-O17-3

Using patterns of somatic mutations in cancer to predict disease genes

Davide Cittaro[1], Dejan Lazarevic[1], Paolo Provero[2]
[1]Center for Translational Genomics and Bioinformatics, San Raffaele Hospital, Italy, [2]University of Turin, Dept. of Molecular Biotechnology and Life Sciences, Torino, Italy

Tue(3)-O17-4

Telomere Length and Accelerated Aging in Adult Survivors of Childhood Cancer: a report from the St. Jude Lifetime Cohort

Zhaoming Wang, Yutaka Yasui, Kirsten K Ness, Carmen L Wilson, DeoKumar Srivastava, Michael Rusch, Andrew Thrasher, Melissa M Hudson, Jinghui Zhang, Leslie L Robison
St. Jude Children's Research Hospital, USA

Tue(3)-O17-5

CentoMD®, the largest variant database for rare diseases

Daniel Trujillano
Centogene AG, Germany

Tue(3)-O17-6

Patient Archive: An integrated solution for deep phenotyping of clinical cases

Tudor Groza[1,2], Mark Cowley[1,2], Tony Roscioli[1,2], Gareth Baynam[3,4,5,6,7], Hugh Dawkins[5], Melissa Haendel[8], Chris Mungall[9], Nicole Washington[9], Damian Smedley[10], Peter N Robinson[11,12,13,14], Marcel Dinger[1,2], Andreas Zankl[1,15,16]
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13:50-15:20 Room B-1

O18 Concurrent Oral Session 18
"Clinical Genetics and Dysmorphology 2"

Chairs: **Kimihiko Oishi**
Genetics and Genomic Sciences, Pediatrics, Icahn School of Medicine at Mount Sinai, USA
Nobuhiko Okamoto
Department of Medical Genetics, Osaka Medical Center and Research Institute for Maternal and Child Health, Izumi, Osaka, Japan

Tue(3)-O18-1

***XRCC4,* a novel gene associated with Seckel syndrome and increased genomic instability**

Nadine Rosin[1,2,3], Nursel H. Elcioglu[4], Filippo Beleggia[1,2,3], Pinar Isgueven[5], Janine Altmueller[1,6], Holger Thiele[6], Katharina Steindl[7], Pascal Joset[7], Anita Rauch[7], Peter Nuernberg[2,3,6], Bernd Wollnik[1,2,3,8], Goekhan Yigit[1,2,3]
[1]Institute of Human Genetics, University of Cologne, Cologne, Germany, [2]Center for Molecular Medicine Cologne (CMMC), University of Cologne, Cologne, Germany, [3]Cologne Excellence Cluster on Cellular Stress Responses in Aging-Associated Diseases (CECAD), University of Cologne, Cologne, Germany, [4]Department of Pediatric Genetics, Marmara University School of Medicine, Istanbul, Turkey, [5]Department of Pediatric Endocrinology, Sakarya University Medical Faculty, Sakarya, Turkey, [6]Cologne Center for Genomics, University of Cologne, Cologne, Germany, [7]Institute of Medical Genetics, University of Zurich, Zurich-Schlieren, Switzerland, [8]Institute of Human Genetics, University Medical Center Goettingen, Goettingen, Germany

Tue(3)-O18-2

Systematic Cellular Disease Models Reveal Synergistic Interactions of Trisomy 21 and GATA1 Mutations in Hematopoietic Abnormalities

Kimihiko Banno, Yasuji Kitabatake, Sayaka Omori, Keiichi Ozono
Department of Pediatirics, Graduate School of Medicine, Osaka University, Japan

Tue(3)-O18-3

Novel MCA/ID syndrome with *ASH1L* mutation

Nobuhiko Okamoto[1], Fuyuki Miya[2,3], Kenichi Nishioka[4], Hidenobu Soejima[4], Tatsuhiko Tsunoda[2,3], Mitsuhiro Kato[5], Shinji Saitoh[6], Mami Yamasaki[7], Yonehiro Kanemura[8,9], Kenjiro Kosaki[10]
[1]Department of Medical Genetics, Osaka Medical Center and Research Institute for Maternal and Child Health, Japan, [2]Department of Medical Science Mathematics, Medical Research Institute, Tokyo Medical and Dental University, [3]Laboratory for Medical Science Mathematics, Center for Integrative Medical Sciences, RIKEN, [4]Division of Molecular Genetics and Epigenetics, Department of Biomolecular Sciences, Faculty of Medicine, Saga University, [5]Department of Pediatrics, Showa University School of Medicine, [6]Department of Pediatrics and Neonatology, Nagoya City University Graduate School of Medical Sciences, [7]Department of Pediatric Neurosurgery, Takatsuki General Hospital, [8]Division of Regenerative Medicine, Institute for Clinical Research, Osaka National Hospital, National Hospital Organization, [9]Department of Neurosurgery, Osaka National Hospital, National Hospital Organization, [10]Center for Medical Genetics, Keio University School of Medicine

Tue(3)-O18-4

Novel Splicing Mutation in the *ASXL3* gene causing Bainbridge-Ropers Syndrome

Ikumi Hori[1], Fuyuki Miya[2], Kei Ohashi[1], Yutaka Negishi[1], Ayako Hattori[1], Naoki Ando[1], Nobuhiko Okamoto[3], Mitsuhiro Kato[4], Tatsuhiko Tsunoda[2], Mami Yamasaki[5], Yonehiro Kanemura[6,8], Kenjiro Kosaki[7], Shinji Saitoh[1]
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Tue(3)-O18-5

Further characterization of Coffin-Siris syndrome caused by a novel variant in *SMARCB1*

Kimihiko Oishi[1], Lisa Karger[1], Noriko Miyake[2], Lakshmi Mehta[1], Naomichi Matsumoto[2]
[1]Pediatrics, Genetics and Genomic Sciences, Icahn School of Medicine at Mount Sinai, USA, [2]Human Genetics, Yokohama City University Graduate School of Medicine

Tue(3)-O18-6

Broadening the phenotypic spectrum of *ANKRD11*-related syndrome [S1] [S1] 62 characters <255 characters

Satoko Miyatake[1], Nobuhiko Okamoto[2], Zornitza Stark[3], Yoshinori Tsurusaki[1,4], Mitsuko Nakashima[1], Hirotomo Saitsu[1], Noriko Miyake[1], Akira Ohtake[5], Naomichi Matsumoto[1]
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15:40-17:10 Room B-1

O19 Concurrent Oral Session 19
"Clinical Genetics and Dysmorphology 3"

Chairs: **Susan H. Blanton**
Dr. John T. Macdonald Department of Human Genetics, University of Miami, USA
Hiroki Kurahashi
Division of Molecular Genetics, Institute for Comprehensive Medical Science, Fujita Health University, Japan

Tue(3)-O19-1

An unique case of a mosaic genome-wide uniparental isodisomy in a newborn with Beckwith-Wiedemann syndrome

Lars T. van der Veken[1], PFR Hochstenbach[1], A.A. Verrijn Stuart[2], J.C. Giltay[1], S.M.J. Hopman[1]
[1]Dept of Genetics, University Medical Center Utrecht, Utrecht, The Netherlands, Netherlands, [2]Dept. of Endocrinology, University Medical Center Utrecht, Utrecht, The Netherlands

Tue(3)-O19-2

The comprehensive genetic analysis of congenital anomalies of the kidney and urinary tract (CAKUT) in Japan

Naoya Morisada[1,2], Akemi Shono[2], Mariko Taniguchi-Ikeda[2], Kandai Nozu[2], Koichi Kamei[3], Kenji Ishikura[3], Shuichi Ito[4], Ryojiro Tanaka[5], Hisahide Nishio[1], Kazumoto Iijima[2]
[1]Department of Community Medicine and Social Healthcare Science, Kobe University Graduate School of Medicine, Japan, [2]Department of Pediatrics, Kobe University Graduate School of Medicine, [3]Division of Nephrology and Rheumatology, National Center for Child Health and Development, [4]Department of Pediatrics, Yokohama City University Graduate School of Medicine, [5]Department of Nephrology, Hyogo Prefectural Kobe Children's Hospital

Tue(3)-O19-3

MOLECULAR DIAGNOSIS OF RWANDAN CHILDREN WITH UNEXPLAINED INTELLECTUAL DISABILITY/NEURODEVELOPMENTAL DELAY BY a-CGH AND WHOLE EXOME SEQUENCING

Leon Mutesa[1], Annette Uwineza[1,2], Jean Hubert Caberg[2], Vincent Bours[2]
[1]Human Genetics, University of Rwanda, Rwanda, [2]Human Genetics, University of Liege, Belgium

Tue(3)-O19-4

Screening of copy number variants in 450 Japanese subjects presenting with intellectual disability (ID) and multiple congenital anomalies (MCA) by SNP array unveiling rare small variants and *PPFIA2* as a novel candidate gene for ID

Daniela T. Uehara[1], Shin Hayashi[1,2,3], Yoshio Makita[4], Akira Hata[5], Issei Imoto[6], Johji Inazawa[1,2,7]
[1]Department of Molecular Cytogenetics, Medical Research Institute, Tokyo Medical and Dental University, Japan, [2]Hard Tissue Genome Research Center, Tokyo Medical and Dental University, Tokyo, Japan, [3]Department of Neurobiology, Yale University School of Medicine, New Haven, Connecticut, USA, [4]Education Center, Asahikawa Medical College, Asahikawa, Japan, [5]Department of Public Health, Chiba University Graduate School of Medicine, Chiba, Japan, [6]Department of Human Genetics, Institute of Biomedical Sciences, Tokushima University Graduate School, Tokushima, Japan, [7]Bioresource Research Center, Tokyo Medical and Dental University, Tokyo, Japan

Tue(3)-O19-5

Family-based association analysis of whole exome sequencing data identifies evidence for major role of focal adhesion pathway

Susan H. Blanton[1,2], Steven Lang[2], Paul Dillingham[2], Jacqueline T. Hecht[3]
[1]Department of Human Genetics, University of Miami, USA, [2]John P. Hussman Institute for Human Genomics, University of Miami, [3]University of Texas Health Medical School and School of Dentistry

Tue(3)-O19-6

Clinical utility of next generation sequencing for undiagnosed syndromic disorder in pediatric patients with short stature or overgrowth

Yoo-Mi Kim[1], Yun-Jin Lee[1], Jae Hong Park[1], Hyoung Doo Lee[1], Chong Kun Cheon[1], Su-Young Kim[1], Gu-Hwan Kim[2], Han-Wook Yoo[2], Eun Hae Cho[3], Ja-Hyun Jang[3]
[1]pediatrics, Pusan National University Children's hospital, Korea, South, [2]Medical Genetics Center, Asan Medical Center, University of Ulsan College of Medicine, Seoul, Korea, [3]Green Cross Laboratories, Green Corss Genome

13:50-15:20 Room B-2

O20 Concurrent Oral Session 20
"Complex Traits and Polygenic Disorders 2"

Chairs: **Guillaume Lettre**
Department of Medicine, Montreal Heart Institute and Université de Montréal, Canada
Toshihiro Tanaka
Department of Human Genetics and Disease Diversity, Graduate School of Medical and Dental Sciences, Tokyo Medical and Dental University, Japan

Tue(3)-O20-1

Imputation analysis using reference panel of 1,070 Japanese individuals (1KJPN) and *in silico* / *in vitro* functional analyses identified functional variants for primary biliary cirrhosis (PBC) susceptibility

Yuki Hitomi[1,11], Kaname Kojima[2,3,11], Minae Kawashima[1,4], Yosuke Kawai[2,3], Nao Nishida[1,5], Yoshihiro Aiba[6], Michio Yasunami[7], Masao Nagasaki[2,3,8], Minoru Nakamura[6,9,10], Katsushi Tokunaga[1]
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Tue(3)-O20-2

The Latent Low Rank Model to Colocalize Genetic Risk Variants in Multiple GWAS

Jin Liu[1], Can Yang[2]
[1]Center of Quantitative Medicine, Duke NUS Graduate Medical School, Singapore, [2]Hong Kong Baptist University

Tue(3)-O20-3

Leveraging Characteristics of Common Genetic Variants to Improve Power of Gene Discovery in Genome-wide Association Study of Neuroticism

Min-Tzu Lo[1], Yunpeng Wang[1,2], Chun-Chieh Fan[1,3], Olav Smeland[2], Aree Witoelar[2], Andrew Schork[1,3], Wesley K. Thompson[5], 23andMe co-authors[7], Srdjan Djurovic[2,4], Ole A. Andreassen[2], Anders M. Dale[1,5,6], Chi-Hua Chen[1]
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Tue(3)-O20-4

Dominant Genetic Variation and Missing Heritability for Human Complex Traits - Insights from Twin versus Genome-wide Common SNP Models

Xu Chen[1], Ralf Kuja-Halkola[1], Iffat Rahman[2], Johannes Arpegard[3,4], Alexander Viktorin[1], Robert Karlsson[1], Sara Hagg[1], Per Svensson[3,4], Nancy L Pedersen[1], Patrik K.E Magnusson[1]
[1]Department of Medical Epidemiology and Biostatistics, Karolinska Institutet, Sweden, [2]Institute of Environmental Medicine, Karolinska Institutet, [3]Department of Medicine-Solna, Karolinska Institutet, [4]Department of Emergency Medicine, Karolinska University Hospital

Tue(3)-O20-5

Diagnostic whole exome sequencing: clinically relevant variants in two thirds of the families

Daniel Trujillano
Centogene AG, Germany

Tue(3)-O20-6

The role of rare and low-frequency coding variants in adult height, a classic polygenic human trait

Guillaume Lettre[1], Eirini Marouli[2], Mariaelisa Graff[3], Carolina Medina-Gomez[4], Ken Sin Lo[1], Claudia Schurmann[5], Kevin Lu[5], Nancy Heard-Costa[6], Joel N. Hirschhorn[7,8], Ruth J.F. Loos[5], Timothy M. Frayling[9], Fernando Rivadeneira[4], Panos Deloukas[2], GIANT Consortium
[1]Medicine, Universite de Montreal, Montreal. Canada, [2]Queen Mary University of London, London, [3]University of North Carolina at Chapel Hill, Chapel Hill, USA, [4]Erasmus Medical Center, Rotterdam, The Netherlands, [5]Icahn School of Medicine at Mount Sinai, New York, USA, [6]Boston University School of Medicine, Boston, USA, [7]Broad Institute, Cambridge, USA, [8]Childrens Hospital Boston, Boston, USA, [9]University of Exeter, Exeter, UK

15:40-17:10 Room B-2

O21 Concurrent Oral Session 21
"Complex Traits and Polygenic Disorders 3"

Chairs: **Swapan K. Nath**
Arthritis and Clinical Immunology Program, Oklahoma Medical Research Foundation, USA
Katsushi Tokunaga
Department of Human Genetics, University of Tokyo, Graduate School of Medicine, Japan

Tue(3)-O21-1

Gene-based analysis of regulatory variants identifies P2RY14 as a new asthma risk gene

Manuel AR Ferreira[1], Rhiannon Werder[2], Melanie Matheson[3], Jennie Hui[4,10], Joyce Tung[5], Svetlana Baltic[6], Peter Le Souef[7], Joseph Powell[8], Grant Montgomery[1], Colin Robertson[9], Alan James[4,10,11,12], Philip Thompson[6], Nicholas Martin[1], John Hopper[3], David Hinds[5], Simon Phipps[2], Australian Asthma Genetics Consortium
[1]QIMR Berghofer Medical Research Institute, Australia, [2]School of Biomedical Sciences, University of Queensland, Brisbane, Australia, [3]Melbourne School of Population and Global Health, The University of Melbourne, Melbourne, Australia, [4]PathWest Laboratory Medicine of Western Australia (WA), Nedlands, Australia, [5]23andMe, Inc., Mountain View, California, USA, [6]Institute for Respiratory Health, University of WA, Perth, Australia, [7]School of Paediatrics and Child Health, Princess Margaret Hospital for Children, Perth, Australia, [8]The Institute for Molecular Bioscience, University of Queensland, Brisbane, Australia, [9]Respiratory Medicine, Murdoch Childrens Research Institute, Melbourne, Australia, [10]Busselton Population Medical Research Foundation, Sir Charles Gairdner Hospital, Perth, Australia, [11]School of Medicine and Pharmacology, University of Western Australia, Nedlands, Australia, [12]Department of Pulmonary Physiology and Sleep Medicine, West Australian Sleep Disorders Research Institute, Nedlands, Australia

Tue(3)-O21-2

Association analysis of the HLA-DRB1 locus in Immune-mediated necrotizing myopathy

Yuko Ohnuki[1], Shingo Suzuki[1], Atsuko Shigenari[1], Shigeaki Suzuki[2], Ichizo Nishino[3], Takashi Shiina[1]
[1]Department of Molecular Life Science, Division of Basic Medical Science and Molecular Medicine, Tokai University School of Medicine, Japan, [2]Department of Neurology, Keio University School of Medicine, [3]Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry

Tue(3)-O21-3

The high comorbidity of inflammatory bowel disease in primary sclerosing cholangitis is only partly explained by shared genetic risk factors

Sun-Gou Ji[1], Brian D Juran[2], Konstantinos N Lazaridis[2], Carl A Anderson[1], International PSC Study Group
[1]Wellcome Trust Sanger Institute, Wellcome Genome Campus, Hinxton, Cambridge, UK, [2]Center for Basic Research in Digestive Diseases, Division of Gastroenterology and Hepatology, Mayo Clinic College of Medicine, Rochester, Minnesota, United States of America

Tue(3)-O21-4

Genotyping of relapsing polychondritis for classical HLA genes identified novel susceptibility HLA alleles and distinct genetic characteristics from other rheumatic diseases

Chikashi Terao[1,2,3,4], Hajime Yoshifuji[3], Yoshihisa Yamano[5], Hiroto Kojima[6], Kimiko Yurugi[7], Yasuo Miura[7], Taira Maekawa[7], Hiroshi Handa[8], Koichiro Ohmura[3], Hiroh Saji[6], Tsuneyo Mimori[3], Fumihiko Matsuda[2]
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Tue(3)-O21-5

Fine-mapping analysis of *TNFSF15* across leprosy, Crohn's disease and primary biliary cirrhosis

Astrid Irwanto[1], Yonghu Sun[4], Yuki Hitomi[2], Licht Toyooka[2], Hyunchul Choi[3], Furen Zhang[4,5], Kyuyoung Song[3], Katsushi Tokunaga[2], Jianjun Liu[1], Anand Kumar Andiappan[6], Olaf Rotzschke[6]
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Tue(3)-O21-6

High-density genotyping of immune-related loci and follow-up genetic association study identified ten novel SLE susceptibility genes in individuals with Asian ancestry

Swapan K. Nath[1], Celi Sun[1], Julio Molineros[1], Loren Looger[2], Xu-Jie Zhou[3], Kwangwoo Kim[4], Yukinori Okada[5], Yuta Kochi[6], Kazuhiko Yamamoto[7], Nan Shen[8], John Harley[9], Kek Heng[10], Hong Zhang[3], Sang-Cheol Bae[4]

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13:50-15:20 Room C-1

O22 Concurrent Oral Session 22
"Molecular Basis of Mendelian Disorders 2"

Chairs: **Tiong Yang Tan**
Victorian Clinical Genetics Services, Murdoch Children's Research Institute, Melbourne, Australia / Department of Paediatrics, University of Melbourne, Melbourne, Australia / Department of Paediatrics and Adolescent Medicine, University of Hong Kong, Hong Kong

Fuki Marie Hisama
Division of Medical Genetics, Department of Medicine, University of Washington, USA

Tue(3)-O22-1

Novel mutations in *ZNF335* broaden the phenotypic spectrum including less severe microcephaly and survivability into childhood

Tiong Yang Tan[1,2,3], Maie Walsh[1,2], Naomi Baker[1,2,3], Mai Raabus[2], Andrew J Kornberg[3,4], David Tickell[3,5,6], Natasha J Brown[1,7], Lavinia Gordon[2,8], Peter G Farlie[2,3]

[1]Victorian Clinical Genetics Services, Australia, [2]Murdoch Childrens Research Institute, Melbourne, Australia, [3]Department of Paediatrics, University of Melbourne, Melbourne, Australia, [4]Neurology Department, Royal Childrens Hospital, Parkville, Australia, [5]Ballarat Health Services, Ballarat, Australia, [6]Deakin University, Melbourne, Australia, [7]Department of Clinical Genetics, Austin Health, Heidelberg, Australia, [8]Australian Genome Research Facility, Walter and Eliza Hall Institute, Parkville, Australia

Tue(3)-O22-2

***ADCY5*-Related Dyskinesia is Likely Under-recognized: Genotype-Phenotype Correlations and Broadening the Spectrum**

Fuki Marie Hisama[1,4], Dong-Hui Chen[1], Jennifer R Friedman[2], Aurelie Meneret[3], Emmanuel Roze[3], Thomas D Bird[1,4], Wendy H Raskind[1]

[1]Medical Genetics/Medicine, Univ of Washington, USA, [2]Departments of Neurosciences and Pediatrics, University of California, San Diego, [3]Departements de Neurologie et de Genetique, Hopital de la Pitie Salpetriere, [4]Department of Neurology, Univ of Washington

Tue(3)-O22-3

Mutations in *HACE1* cause an autosomal-recessive neurodevelopmental disorder

Ronja Hollstein[1], David A. Perry[2], Lisa Nalbach[1], Clare V. Logan[2], Tim M. Strom[3,4], Verity L. Hartill[2,5], Ian M. Carr[2], Georg C. Korenke[6], Sandeep Uppal[2], Mushtaq Ahmed[5], Thomas Wieland[4], Alexander F. Markham[2], Christopher P. Bennett[5], Gabriele Gillessen-Kaesbach[7], Eamonn G. Sheridan[2,5], David T. Bonthron[2,5], Frank J. Kaiser[1]

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Tue(3)-O22-4

Severe CNS involvement in *WWOX* mutations: Description of five new cases

Amal M Alhashem[1], Saad Alsahwan[1], Fowzan S Alkuraya[2], Abdulla M Alhashem[1], Giulio Zuccoli[4], Satyanarayana Gedela[3]

[1]Pediatrics, Prince Sultan Military Medical City, Saudi Arabia, [2]King Faisal Specialist Hospital and Research Center, [3]Nationwide Children Hospital, [4]Children Hospital of Pittsburgh of UPMC, University of Pittsburgh

Tue(3)-O22-5

Genetic studies on a Portuguese Parkinson disease patient cohort

Gabriel Miltenberger-Miltenyi[1], Leonor Guedes[2], Tiago Soeiro[2], Marcos Gomes[1], Joaquim J Ferreira[2], Tiago F Outeiro[3]

[1]Instituto de Medicina Molecular, Faculdade de Medicina, Universidade de Lisboa, Portugal, [2]Neurology Department, Hospital de Santa Maria - Centro Hospitalar Lisboa Norte, Lisbon, [3]Department of NeuroDegeneration and Restaurative Research, University Medical Center Goettingen

Tue(3)-O22-6

Neuron-specific *Cul4b* knockout mice recapture the cognitive impairment phenotype in human X-linked mental retardation patients

Baichun Jiang, Wei Zhao, Shuqian Zhang, Huili Hu, Changshun Shao, Yaoqin Gong

Department of Genetics, School of Medicine, Shandong University, China

15:40-17:10 Room C-1

O23 Concurrent Oral Session 23
"Molecular Basis of Mendelian Disorders 3"

Chairs: **Claude Ferec**
Director of the INSERM UMR1078 "Genetic, Functional Genomic & Biotechnologies", INSERM, Brest University, Brest Hospital, France

Noriko Miyake
Department of Human Genetics, Yokohama City University Graduate School of Medicine, Japan

Tue(3)-O23-1

The utility of medical exome-based virtual gene panel in the molecular diagnosis of genetically heterogeneous sensorineural hearing loss

Qiaoning Guan[1], Kajia Cao[1], Zhiqian Fan[1], Ian Slack[2], Sawona Biswas[2], Sarah Noon[2], Matthew Dulik[1], Elizabeth DeChene[1], Mahdi Sarmady[1], Zhenming Yu[1], Surabhi Mulchandani[1], Jin Yun Chen[1], Elizabeth Denenberg[1], Jinbo Fan[1], Jorune Balciuniene[1], Avni Santani[1], Ian Krantz[2], Nancy Spinner[1], Laura Conlin[1], Minjie Luo[1]

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Tue(3)-O23-2

Targeted screening of 187 genes involved in ocular development increases mutation detection rate by 10 % in individuals with anophthalmia-microphthalmia spectrum

Nicolas Chassaing[1,2,3,6], Nicola Ragge[4,5,6], Patrick Calvas[1,2,3]
[1]Medical Genetics, CHU Toulouse, France, [2]Inserm U1056, [3]EA-4555, Universite Toulouse III, [4]School of Life Sciences, Oxford Brookes University, Oxford, UK, [5]Clinical Genetics Unit, Birmingham Women s Hospital, Birmingham, UK, [6]These authors contributed equally to this work

Tue(3)-O23-3

The p.S178L mutation in *TBC1D24* lead to dominant, non-syndromic hearing impairment through a gain-of-function mechanism

Tao Yang, Luping Zhang, Linxiang Hu, Xiuhong Pang, Penghui Chen, Hao Wu
Xinhua Hospital, Shanghai Jiaotong University School of Medicine, China

Tue(3)-O23-4

Submicroscopic deletions at 13q32.1 cause congenital microcoria

Lucas Fares Taie[1], Sylvie Gerber[1], Akihiko Tawara[2], Arturo Ramirez-Miranda[3], Jean-Yves Douet[4], Hannah Verdin[5], Juan C Zenteno[3], Hiroyuki Kondo[2], Bruno Passet[6], Ken Yamamoto[7], Masaru Iwai[8], Toshihiro Tanaka[9], Yusuke Nakamura[10], Wataru Kimura[11], Arnold Munnich[1], Elfride De Baere[5], Isabelle Raymond-Letron[4], Josseline Kaplan[1], Patrick Calvas[12], Olivier Roche[13], Jean-Michel Rozet[1]
[1]Imagine - Institute of Genetic Diseases, France, [2]University of Occupational & Environmental Health, Kitakyushu, Japan, [3]Instituto de Oftalmologia Conde de Valenciana. UNAM, Mexico City, Mexico, [4]Veterinary School of Toulouse, University of Toulouse, France, [5]Center for Medical Genetics, Ghent University, Belgium, [6]Institut Nationale de la Recherche Agronomique, Jouy-en-Josas, France, [7]Institute of Bioregulation, Kyushu University, Fukuoka, Japan, [8]Ehime University Graduate School of Medicine, Japan, [9] Graduate School of Medical and Dental Sciences, Tokyo Medical and Dental University, Japan, [10]University of Chicago, USA, [11]Kimura Eye Clinic, Kure, Japan, [12]Hopital Purpan, Toulouse, France, [13]IHU Necker-Enfants Malades, University Paris-Descartes, Paris, France

Tue(3)-O23-5

Biallelic *NUP107* mutations cause early childhood-onset steroid resistant Nephrotic syndrome

Noriko Miyake[1], Hiroyasu Tsukaguchi[2], Eriko Koshimizu[1], Akemi Shono[3], Satoko Matsunaga[4], Masaaki Shiina[5], Yasuhiro Mimura[6], Shintaro Imamura[7], Tomonori Hirose[8], Koji Okudela[9], Hae Il Cheong[10,11,12], Kenichi Ohashi[9], Naoko Imamoto[6], Akihide Ryo[4], Kazuhiro Ogata[5], Kazumoto Iijima[3], Naomichi Matsumoto[1]
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Tue(3)-O23-6

Genic and allelic variability in polycystic kidney disease :impact in the erea of precision medicine

Claude Ferec[1], Marie-Pierre Audrezet[1], Emilie Cornec-LeGall[1,2], Yannick Le Meur[2], Jian-Min Chen[1]
[1]Inserm/university, France, [2]Service de Nephrologie University/Hospita

13:50-15:20 Room C-2

O24 **Concurrent Oral Session 24**

"Psychiatric Genetics, Neurogenetics and Neurodegeneration 2"

Chairs: **William K. Scott**
Dr. John T. Macdonald Foundation Department of Human Genetics and John P. Hussman Institute for Human Genomics, University of Miami, USA

Takeshi Ikeuchi
Molecular Genetics, Brain Research Institute, Niigata University, Japan

Tue(3)-O24-1

Genome-wide interaction study of Parkinson disease and vitamin D deficiency implicates autoimmunity pathways

William K. Scott[1,2], Lizmarie Maldonado[1], Gary W. Beecham[1,2], Eden R. Martin[1,2], Marian L. Evatt[3], James C. Ritchie[4], Jonathan L. Haines[5], Cyrus P. Zabetian[6,7], Haydeh Payami[8,9], Margaret A. Pericak-Vance[1,2], Jeffery M. Vance[1,2], Liyong Wang[1,2]
[1]John P. Hussman Institute for Human Genomics, University of Miami, USA, [2]Dr. John T. Macdonald Foundation Department of Human Genetics, University of Miami, [3]Department of Neurology, Emory University, [4]Department of Pathology, Emory University, [5]Department of Epidemiology and Biostatistics and Institute for Computational Biology, Case Western Reserve University, [6]Veterans Affairs Puget Sound Health Care System, [7]Department of Neurology, University of Washington, [8]Departments of Neurology and Genetics, University of Alabama-Birmingham, [9]HudsonAlpha Institute for Biotechnology

Tue(3)-O24-2

ABCA7 Frameshift Deletion Associated with Alzheimer's Disease in African Americans

Holly N Cukier[1,2], Brian W Kunkle[1], Badri N Vardarajan[3], Sophie Rolati[1], Kara L Hamilton-Nelson[1], Patrice L Whitehead[1], Derek Van Booven[1], Rosalyn Lang[4], Derek M Dykxhoorn[1,5], Lindsay A Farrer[6], Michael L Cuccaro[1,5], Jeffery M Vance[1,2,5], John R Gilbert[1,5], Gary W Beecham[1,5], Eden R Martin[1,5], Regina M Carney[1,5], Richard Mayeux[1,5], Gerard Schellenberg[7], Goldie S Byrd[4], Jonathan L Haines[8], Margaret A Pericak-Vance[1,2,5], Alzheimer's Disease Genetics Consortium (ADGC)
[1]John P. Hussman Institute for Human Genomics, University of Miami Miller School of Medicine, USA, [2]Department of Neurology, University of Miami Miller School of Medicine, [3]Taub Institute for Research on Alzheimer Disease and the Aging Brain, Gertrude H. Sergievsky Center, Departments of Neurology, Psychiatry, and Epidemiology, Columbia University, [4]Department of Biology, North Carolina A&T State University, [5]Dr. John T. Macdonald Foundation Department of Human Genetics, University of Miami Miller School of Medicine, [6]Departments of Medicine, Neurology, Ophthalmology, Genetics & Genomics, Epidemiology, and Biostatistics, Boston University, [7]Department of Pathology and Laboratory Medicine, University of Pennsylvania Perelman School of Medicine, [8]Department of Epidemiology and Biostatistics, Institute for Computational Biology, Case Western Reserve University School of Medicine

Tue(3)-O24-3

Structural variants and neurodegenerative diseases in aging: regulatory and causality consequences

Ornit Chiba-Falek[1], Michael W Lutz[1], Robert Saul[2], Lidia Tagliafierro[1], Allen D Roses[1,3]
[1]Neurology, Duke University, USA, [2]Polymorphic DNA Technologies, Alameda, CA, USA, [3]Zinfandel Pharmaceuticals, Chapel Hill, NC, USA

Tue(3)-O24-4

Novel candidate genes for early-onset Alzheimer disease identified using whole-exome sequencing

Gary W Beecham[1], Brian W Kunkle[1], Badri Vardarajan[2], Patrice L Whitehead[1], Sophie Rolati[1], Eden R Martin[1], John R Gilbert[1], Richard P Mayeux[2], Jonathan L Haines[3], Margaret A Pericak-Vance[1]
[1]John P Hussman Institute for Human Genomics, University of Miami Miller School of Medicine, USA, [2]Taub Institute of Research on Alzheimer's Disease, Columbia University, [3]Institute for Computational Biology, Case Western Reserve University

Tue(3)-O24-5

Transethnic Genome-Wide Meta-Analysis for Alzheimer Disease identifies Novel Genes

Gyungah R Jun[1,2], Jaeyoon Chung[1], Giuseppe Tosto[3], Badri Vardarajan[3], Christiane Reitz[3], Kathryn L Lunetta[4], Jennifer Manly[3], Goldie Byrd[5], Jonathan L Haines[6], Margaret A Pericak-Vance[7], Ryozo Kuwano[8], Richard Mayeux[3], Gerard D Schellenberg[9], Lindsay A Farrer[1,3,10,11,12], The Alzheimer's Disease Genetics Consortium

[1]Medicine, Boston University, USA, [2]Integrated Neurogenetics, Eisai Inc, [3]Neurology, Columbia University, [4]Biostatistics, Boston University, [5]Biology, North Carolina A&T State University, [6]Epidemiology and Biostatistics, Case Western Reserve University, [7]The John P. Hussman Institute for Human Genomics, University of Miami, [8]Molecular Genetics, Brain Research Institute, Niigata University, [9]Pathology and Laboratory Medicine, University of Pennsylvania, [10]Ophthalmology, Boston University, [11]Epidemiology, Boston University, [12]Neurology, Boston University

Tue(3)-O24-6

CFH variants affect structural and functional brain changes and genetic risk of Alzheimer's disease

Deng-Feng Zhang[1,6], Jin Li[2], Huan Wu[3], Yue Cui[2], Rui Bi[1,6], He-Jiang Zhou[1], Hui-Zhen Wang[1], Chen Zhang[4], Dong Wang[1], Qing-Peng Kong[3], Tao Li[5], Yiru Fang[4], Tianzi Jiang[2,7], Yong-Gang Yao[1,6,7], Alzheimer's Disease Neuroimaging Initiative (ADNI)

[1]Key Laboratory of Animal Models and Human Disease Mechanisms, Kunming Institute of Zoology, Chinese Academy of Sciences, China, [2]Brainnetome Center and National Laboratory of Pattern Recognition, Institute of Automation, Chinese Academy of Sciences, Beijing, China, [3]State Key Laboratory of Genetic Resources and Evolution, Kunming Institute of Zoology, Chinese Academy of Sciences, Kunming, Yunnan, China, [4]Division of Mood Disorders, Shanghai Mental Health Center, Shanghai Jiao Tong University School of Medicine, Shanghai, China, [5]The Mental Health Center & Psychiatric Laboratory, West China Hospital, Sichuan University, Chengdu, Sichuan, China, [6]Kunming College of Life Science, University of Chinese Academy of Sciences, Kunming, Yunnan, China, [7]CAS Center for Excellence in Brain Science, Chinese Academy of Sciences, Shanghai, China

15:40-17:10 Room C-2

O25 Concurrent Oral Session 25
"Metabolic Disorders 2"

Chairs: **Dong Hwan Lee**
Soon Chun Hyang University Hospital, Korea, South
Naoko Iwasaki
Diabetes Center, Institute of Medical Genetics, Tokyo Women's Medical University, Japan

Tue(3)-O25-1

Prevalence of MODY subtype and clinical characteristics in patients with early onset diabetes in Japanese

Naoko Iwasaki[1,2,3], Miho Takizawa[1], Makiko Ogata[1], Risa Ide[1], Yasuko Uchigata[1], Kayoko Saito[1,2,3]

[1]Diabetes Center, Tokyo Women's Medical University, Japan, [2]Institute of Medical Genetics, Tokyo Women's Medical University, [3]Institute of Integrated Medical Science, Tokyo Women's Medical University

Tue(3)-O25-2

Relationship between haplotype diversity of the HLA-G gene with type 1 diabetes mellitus and its expression pattern on dendritic cells DC-10

Rafael de Albuquerque[1], Norma Lucena[2], Eduardo Donadi[1], Celso Mendes-Junior[1], Silvia Gregori[3]

[1]USP, Brazil, [2]Fiocruz, [3]TIGET

Tue(3)-O25-3

Protein tyrosine phosphatase 1B (PTP1B) gene polymorphism is associated with obesity and resistance to weight reduction therapy in the Japanese

Noriko Satoh-Asahara[1], Hajime Yamakage[1], Masashi Tanaka[1], Shinya Masuda[1], Kazuya Muranaka[1], Akira Shimatsu[1], Kikuko Hotta[2], Yoshihiro Miyamoto[3], Hiroko Morisaki[4], Takayuki Morisaki[4]

[1]Division of Diabetic Research, Clinical Research Institute, National Hospital Organization Kyoto Medical Center, Japan, [2]Medical Center for Translational Research, Osaka University Hospital, [3]Department of Preventive Cardiology, National Cerebral and Cardiovascular Center, [4]Department of Bioscience and Genetics, National Cerebral and Cardiovascular Center Research Institute

Tue(3)-O25-4

The Incidence of Congenital Hypothyroidism and Study of Endorine Disruptors in Korea

Dong Hwan Lee[1], Ken Suzuki[2]

[1]Department of Pediatrics, Soonchunhyang University Hospital, Korea, South, [2]Deparmtnet of Neonatal Screening, Tokyo Health Service Association

Tue(3)-O25-5

Natural course of congenital hypothyroidism by dual oxidase 2 mutations from the neonatal period through puberty

Yoshihiro Maruo[1], Keisuke Nagasaki[2], Katsuyuki Matsui[1], Yu Mimura[1], Asami Mori[1], Maki Fukami[3]

[1]Pediatrics, Shiga University of Medical Sicence, Japan, [2]Pediatrics, Niigata University, [3]Molecular Endocrinology, National Research Institute for Child Health and Development

Tue(3)-O25-6

Biallelic Truncating Mutations in *TANGO2* Cause Infancy-Onset Recurrent Metabolic Crises with Rhabdomyolysis, Cardiac Arrhythmias, and Progressive Neurodegeneration

Laura S Kremer[1], Felix Distelmaier[2], Bader Alhaddad[3], Maja Hempel[4], Arcangela Iuso[3], Clemens Kuepper[5], Chris Muehlhausen[6], Reka Kovacs-Nagy[3], Robin Satanofskij[3], Elisabeth Graf[1], Riccardo Berutti[3], Gertrud Eckstein[3], Richard Durbin[7], Sascha Sauer[8], Georg F Hoffmann[7], Tim M Strom[1,3], Rene Santer[6], Thomas Meitinger[1,3], Thomas Klopstock[5], Holger Prokisch[1,3], Tobias B Haack[1,3]

[1]Institute of Human Genetics, Helmholtz Zentrum Muenchen, Neuherberg, Germany, [2]Department of General Pediatrics, University Childrens Hospital, Heinrich-Heine-University Duesseldorf, Germany, [3]Institute of Human Genetics, Technische Universitaet Muenchen, Germany, [4]Institute of Human Genetics, University Medical Center Hamburg-Eppendorf, Hamburg, Germany, [5]Department of Neurology, Friedrich-Baur-Institute, Ludwig-Maximilians-University, Munich, Germany, [6]Department of Pediatrics, University Medical Center Hamburg-Eppendorf, Hamburg, Germany, [7]Wellcome Trust Sanger Institute, Hinxton, Cambridge, United Kingdom, [8]CU Systems Medicine, University of Wuerzburg, Wuerzburg, Germany

13:50-15:20 Room I

O26 Concurrent Oral Session 26
"Clinical Genetic Testing 1"

Chairs: **Ian G. Campbell**
Peter MacCallum Cancer Centre, University of Melbourne, Australia
Shin-ichi Usami
Department of Otorhinolaryngology, Shinshu University School of Medicine, Japan

Tue(3)-O26-1

Mutation spectrum of Japanese Lynch syndrome patients diagnosed by universal tumor screening for colorectal cancer

Kiwamu Akagi[1], Miho Kakuta[1], Akemi Takahashi[1], Tetsuhiko Tachikawa[1], Gou Yamamoto[1], Yoshiko Arai[1], Shiho Kobayasi[1], Kenji Fujiyoshi[1,3], Yoshito Akagi[3], Takashi Takenoya[1,2], Yoji Nishimura[2], Yoshiyuki Kawashima[2], Hirohiko Sakamoto[2]
[1]Molecular Diagnosis and Cancer Prevention, Saitama Cancer Center, Japan, [2]Digestive Surgery, Saitama Cancer Center, [3]Surgery, Kurume University

Tue(3)-O26-2

Breast and Ovarian cancer prevention: Is it time for population screening for BRCA1 and BRCA2 mutations?

Ian G Campbell, Ella Thompson, Simone Rowley, Mary_Anne Young, Alison Trainer, Na Li, Lisa Devereux, Gillian Mitchell, Paul James, Lifepool
Research DIvision, Peter MacCallum Cancer Centre, Australia

Tue(3)-O26-3

Differences of Clinical Characteristics among Heterozygous Familial Hypercholesterolemia Based on Genetic Diagnosis

Atsushi Nohara[1], Masa-aki Kawashiri[2], Hayato Tada[2], Mie Yoshida[1], Mika Mori[2], Chiaki Nakanishi[2], Kunimasa Yagi[3], Akihiro Inazu[4], Takeshi Kobayashi[1], Masakazu Yamagishi[2], Hiroshi Mabuchi[1], The Hokuriku FH Study Group
[1]Department of Advanced Research in Community Medicine, Kanazawa University Graduate School of Medical Sciences, Japan, [2]Department of Cardiovascular Medicine, Kanazawa University Graduate School of Medical Sciences, [3]Medical Education Research Center, Kanazawa University Graduate School of Medical Sciences, [4]Division of Health Sciences, Kanazawa University Graduate School of Medical Sciences

Tue(3)-O26-4

Target resequencing of neuromuscular disease-related genes using next-generation sequencing for patients with undiagnosed early-onset neuromuscular disorders

Yuri Kitamura[1,2], Eri Kondo[1,3], Mari Urano[1], Ryoko Aoki[1], Kayoko Saito[1,2]
[1]Institute of Medical Genetics, Tokyo Women's Medical University, Japan, [2]Affiliated Field of Medical Genetics, Division of Biomedical Engineering and Science, Graduate School of Tokyo Women's Medical University Tokyo, [3]Imperial Gift Foundation AIIKU Maternal and Child Health Center, AIIKU, Clinic Department of Pediatrics

Tue(3)-O26-5

Deafness gene variations in a 1,120 nonsyndromic hearing loss cohort: Molecular epidemiology and deafness mutation spectrum of patients in Japan

Shin-ya Nishio, Shin-ichi Usami
Department of Otorhinolaryngology, Shinshu University School of Medicine, Japan

Tue(3)-O26-6

Improved Performance of Whole Genome Sequencing detects a *SYNGAP1* Mutation in siblings with Epilepsy with Myoclonic-Atonic seizures and photosensitivity

Mark J Cowley[1,2], Yu-Chi Liu[3], Karen L Oliver[3,5], Gemma Carvill[4], Candace Myers[4], Velimir Gayevskiy[1], Marin Delatycki[6], Ying Zhu[7], Kevin Ying[1], David Miller[1], Paula Morris[1], Aaron L Statham[1], Heather Mefford[4], Michael F Buckley[8], Samuel F Berkovic[5,6], Melanie Bahlo[3], Ingrid E Scheffer[5,6,9,10], Marcel E Dinger[1,2], Tony Roscioli[1,2,11]

[1]Kinghorn Centre for Clinical Genomics, Garvan Institute of Medical Research, Australia, [2]St Vincents Clinical School, University of New South Wales, Darlinghurst, Australia, [3]Population Health and Immunity Division, Walter and Eliza Hall Institute, Melbourne, Australia, [4]University of Washington Department of Pediatrics, Genome Sciences, Seattle, USA, [5]Epilepsy Research Centre, Department of Medicine, University of Melbourne, Austin Health, Heidelberg, Australia, [6]Austin Health, Melbourne, Australia, [7]Department of Medical Genetics, Royal North Shore Hospital, [8]SEALS laboratory, Prince of Wales Hospital, Randwick, NSW, Australia, [9]Florey Institute, Melbourne, Australia, [10]Department of Paediatrics, University of Melbourne, Royal Childrens Hospital, Australia, [11]Department of Medical Genetics, Sydney Childrens Hospital, NSW, Australia

15:40-17:10 Room I

O27 Concurrent Oral Session 27
"Clinical Genetic Testing 2"

Chairs: **Michael Buckley**
Kinghorn Centre for Clinical Genomics, Garvan Institute of Medical Research, Sydney, Australia
Toshiyuki Yamamoto
Institute for Integrated Medical Sciences, Tokyo Women's Medical University, Japan

Tue(3)-O27-1

The use of custom-designed NGS panels and CGH array for population-specific clinical testing

Filip Zembol[1], Filip Lhota[1], Bara Honysova[1], Leona Cerna[1], David Stejskal[1], Marie Trkova[2], Monika Koudova[1], Martina Bittoova[1], Martina Putzova[1]

[1]Laboratory of molecular genetics, Gennet, Czech Republic, [2]Laboratory of Clinical Cytology

Tue(3)-O27-2

aCGH ANALYSIS AND ITS IMPLICATIONS IN THE CASE OF A r(X) CHROMOSOME; ADVANTAGES IN AN ERA OF DIAGNOSTIC ODISSEY

Ciprian D. Ion[1], Georgeta Cardos[2], Lucian Oprea[2], Viorica Radoi[1,2]

[1]Medical Genetics, Carol Davila University of Medicine and Pharmacy, Bucharest, Romania, [2]Synevo Central Laboratory, Cytogenetics, Chiajna, Ilfov County, Romania

Tue(3)-O27-3

Childhood-onset peripheral neuropathy: gene panel or whole exome?

Maie I Walsh[1], Katrina Bell[1], Belinda Chong[1], Gemma R Brett[1,2], Paul A James[3], Natalie P Thorn[1,2,4], Alicia Oshlack[1,4], Simon Sadedin[1], Peter Georgeson[4], Ivan Maccocia[1], Clara Gaff[2,4], Eppie M Yiu[1,5], Zornitza Stark[1], Monique M Ryan[1,4,5], Melbourne Genomics Health Alliance

[1]Clinical Genetics, Murdoch Childrens Research Institute, Melbourne, Australia, [2]Melbourne Genomics Health Alliance, Melbourne, Australia, [3]Royal Melbourne Hospital, Melbourne, Australia, [4]University of Melbourne, Australia, [5]Royal Childrens Hospital, Melbourne, Australia

Tue(3)-O27-4

Australian Renal Gene Panels: A National Program of Diagnostic Gene Panels For Multiple Renal Phenotypes Utilising Massively Parallel Sequencing With Multi-Disciplinary Team Reporting

Hugh J McCarthy[1,2,3], Amali C Mallawaarachchi[1], Gladys Ho[4], Katherine Holman[4], Chirag Patel[5], Jeff Fletcher[6], Stephen I Alexander[2,3], Bruce Bennetts[4], Andrew J Mallett[7,8,9]
[1]Department of Clinical Genetics, The Children's Hospital at Westmead, Australia, [2]Department of Paediatric Nephrology, The Children's at Westmead, Sydney, Australia, [3]Centre for Kidney Research, University of Sydney, Sydney, Australia, [4]Department of Molecular Genetics, The Children's Hospital at Westmead, Sydney, Australia, [5]Genetic Health Queensland, Royal Brisbane and Women's Hospital, Brisbane, Australia, [6]Department of Paediatrics, The Canberra Hospital, Canberra, Australia, [7]Kidney Health Service and Conjoint Kidney Research Laboratory, Royal Brisbane and Women's Hospital, Brisbane, Australia, [8]Centre for Kidney Disease Research, The University of Queensland, Brisbane, Australia, [9]Centre for Rare Diseases Research, Institute for Molecular Bioscience, The University of Queensland, Brisbane, Australia

Tue(3)-O27-5

Evaluating Whole-Genome Sequencing as a General Purpose Genetic Screen

Mark Pinese[1], Marcel E Dinger[1,2], Mark J Cowley[1,2]
[1]Kinghorn Centre for Clinical Genomics, Garvan Institute of Medical Research, Australia, [2]St Vincent's Clinical School, Faculty of Medicine, UNSW Australia

Tue(3)-O27-6

Clinical Exome in Consanguineous Population Provides Higher Detection Rate

Abdul Ali Zada[1], Majid Alfadhel[2], Soha Tashkandi[1], Saud Alsahli[2], Iram Alluhaydan[2], Fuad Almutairi[2], Ali Alothaim[2], Seham Alameer[3], Eissa Faqeeh[1], Ali Alasmari[1], Abdulaziz Alsamman[1], Abdulaziz Alghamdi[4], Amal Alhashem[4], Amir Babiker[5], Sarar Mohamed[5], Wafaa Eyaid[2], Ahmed Alfares[2,6]
[1]King Fahad Medical City, Saudi Arabia, [2]King Abdulaziz Medical City, National Guard Hospital, Riyadh, Saudi Arabia, [3]King Khaled National Guard Hospital, Jeddah, Saudi Arabia, [4]Prince Sultan Military Medical City, Riyadh, Saudi Arabia, [5]King Saud University Medical City and College of Medicine, Riyadh, Saudi Arabia, [6]Qassim University, Qassim, Saudi Arabia

13:50-15:20 Room J

O28 Concurrent Oral Session 28
"Epigenetics 1"

Chairs: **Andrea Riccio**
CNR, Institute of Genetics and Biophysics, Italy
Kenichiro Hata
Department of Maternal Fetal Biology, National Research Institute for Child Health and Development, Japan

Tue(3)-O28-1

Identification of genetic determinants of monocyte epigenetic plasticity across differing innate immune stimuli

Benjamin Fairfax, Evelyn Lau, Esther Ng, Sara Danielli, Seiko Makino, Julian Knight
Wellcome Trust Centre for Human Genetics, University of Oxford, UK

Tue(3)-O28-2

Novel epigenetic loci associated with Beckwith Wiedemann Syndrome

Izabela Krzyzewska[1], Marielle Alders[1], Saskia M. Maas[2], Faisal I. Rezwan[3], Karin van der Lip[1], Adri N. Mul[1], Andrea Venema[1], Deborah Mackay[3], Marcel M.A.M. Mannens[1], Peter Henneman[1],
Novel epigenetic loci associated with Beckwith Wiedemann Syndrome
[1]Clinical Genetics, Academic Medical Center, Netherlands, [2]Department of Pediatrics, Academic medical Center, Amsterdam, the Netherlands, [3]Faculty of Medicine, University of Southampton, Southampton, UK

Tue(3)-O28-3

Genome-Wide DNA Methylation and Gene Expression Analyses in Blood and Dermal Fibroblasts from Twin Pairs Discordant for Systemic Sclerosis Reveals Distinct Signatures Between Disease Subsets

Paula S Ramos[1], Thomas A Medsger Jr[2], Carol A Feghali-Bostwick[1]
[1]Medical University of South Carolina, USA, [2]University of Pittsburgh

Tue(3)-O28-4

Deletion of the Williams syndrome region in human and mouse causes systemic, genome-wide changes in DNA methylation

Emma Strong[1], Rajat Singhania[2], Daniel De Carvalho[2], Luis A Perez-Jurado[3,4,5], Victoria Campuzano[3,4,5], Lucy R Osborne[1,6]
[1]Molecular Genetics, University of Toronto, Toronto, Canada, [2]Princess Margaret Cancer Centre, University Health Network, Toronto, Canada, [3]Genetics Unit, Department of Experimental and Health Sciences, Universitat Pompeu Fabra, Barcelona, Spain, [4]Hospital del Mar Research Institute (IMIM), Barcelona, Spain, [5]Centro de Investigacion Biomedica en Red de Enfermedades Raras (CIBERER), Barcelona, Spain, [6]Department of Medicine, University of Toronto, Toronto, Canada

Tue(3)-O28-5

The zinc-finger protein ZFP57 controls imprinted and non-imprinted genes through different types of *cis*-acting regulatory elements

Andrea Riccio[1,2], Vincenzo Riso[1,2], Marco Cammisa[1,2], Harpreet Kukreja[1,2], Zahra Anvar[1,2], Shraddha Lad[1], Annalisa Fico[1], Angela Sparago[1,2], Claudia Angelini[3], Grimaldi Grimaldi[1]
[1]CNR, Institute of Genetics and Biophysics, Italy, [2]2nd University of Naples, DiSTABiF, Caserta, [3]Istituto per le Applicazioni del Calcolo Mauro Picone (IAC), CNR, Napoli

Tue(3)-O28-6

RNF12 is essential for X-inactivation in female mouse embryonic stem cells, is required for female mouse development, and might be a target for future therapies to treat X-linked disorders in females: evidence from a mouse knockout model

Stefan Barakat[1,2], Joost Gribnau[2]
[1]MRC Centre for Regenerative Medicine, University of Edinburgh, UK, [2]Erasmus MC-University Medical Center, Rotterdam

15:40-17:10 Room J

O29 Concurrent Oral Session 29
"Epigenetics 2"

Chairs: **Hiroyuki Sakai**
Division of Epigenomic and Development, Department of Molecular Genetics, Medical Institute of Bioregulation, Kyushu University, Japan

Melanie A. Carless
Genetics, Texas Biomedical Research Institute, USA

Tue(3)-O29-1

Longitudinal changes in DNA methylation influence type 2 diabetes

Melanie A Carless[1], Jack W Kent[1], Hemant Kulkarni[2], Michael C Mahaney[2], Anthony G Comuzzie[1], John B[2]
[1]Genetics, Texas Biomedical Research Institute, USA, [2]South Texas Diabetes and Obesity Institute

Tue(3)-O29-2

Genome-wide and targeted analysis of DNA methylation in disease discordant amyotrophic lateral sclerosis (ALS) cohorts

Kelly L Williams[1], Beben Benyamin[2], Emily P McCann[1], Anjali K Henders[2], Sonia Shah[2], Dominic B Rowe[1], Garth A Nicholson[1], Naomi Wray[2], Ian P Blair[1]
[1]Faculty of Medicine and Health Sciences, Macquarie University, Sydney, Australia, [2]Queensland Brain Institute, University of Queensland, Brisbane, Australia

Tue(3)-O29-3

Genome-wide analysis of neuron specific DNA methylation in Alzheimer's disease

Tatsuo Mano[1], Kenichi Nagata[2], Shigeo Murayama[3], Takaomi C. Saido[2], Shoji Tsuji[1], Atsushi Iwata[1,4]
[1]Department of Neurology, Graduate School of Medicine, The University of Tokyo, Japan, [2]Laboratory for Proteolytic Neuroscience, RIKEN BSI, [3]Department of Neuropathology, Tokyo Metropolitan Geriatric Hospital, [4]Japan Science and Technology Agency, PRESTO

Tue(3)-O29-4

Metabolomic changes fine-map the DNA methylation signature of cigarette smoking

Yan V. Sun[1,2], Yunfeng Huang[1], Qin Hui[1], Douglas Walker[3], Dean Jones[3], Jack Goldberg[4], Viola Vaccarino[1]
[1]Department of Epidemiology, Rollins School of Public Health, Emory University, USA, [2]Department of Biomedical Informatics, Emory University School of Medicine, Atlanta, GA, USA, [3]Division of Pulmonary, Allergy and Critical Care Medicine, Emory University School of Medicine, Atlanta, GA, USA, [4]Vietnam Era Twin Registry and Department of Epidemiology, University of Washington School of Public Health, Seattle, WA, USA

Tue(3)-O29-5

Deciphering the role of DNA methylation in SLE pathogenesis through integrative analysis of different types of genomic data

Mengbiao Guo, Tingyou Wang, Wanling Yang
University of Hong Kong, China

Tue(3)-O29-6

DNA methylation profiling of Crohn's disease in peripheral blood and CD14+ cells in women

Andrew Y.F. Li Yim[1,4], Jing Zhao[2], Nicolette N.W. Duijvis[2], Wouter J. de Jonge[2], Menno P.J. de Winther[3], Adri N. Mul[1], Marcel M.A.M. Mannens[1], Anje A. te Velde[2], Peter Henneman[1]
[1]Clinical Genetics, Academic Medical Center, Netherlands, [2]Tytgat Institute for Live & Intestinal Research, Academic Medical Center, Amsterdam, The Netherlands, [3]Medical Biochemistry, Academic Medical Center, Amsterdam, The Netherlands, [4]Department of Epigenetics, GlaxoSmithKline, Stevenage, United Kingdom

13:50-15:20 Room K

O30 Concurrent Oral Session 30
"Genome structure, variation and function 1"

Chairs: **Andrew H. Sinclair**
Deputy Director, Murdoch Children's Research Institute, Melbourne, Australia
Itsuro Inoue
Division of Human Genetics, National Institute of Genetics, Japan

Tue(3)-O30-1

Mechanism of transcriptome abnormalies in Cornelia de Lange syndrome: Disturbance of trnascriptional elongation

Kazuhiro Akiyama[1,2], Masashige Bando[1], Ian D Krantz[3], Kosuke Izumi[1,3], Katsuhiko Shirahige[1]
[1]Research Center for Epigenetic Disease, Institute for Molecular and Cellular Biosciences, The University of Tokyo, Japan, [2]Japan Society for the Promotion of Science, [3]Division of Human Genetics, Children's Hospital of Philadelphia, Philadelphia, Pennsylvania, USA

Tue(3)-O30-2

Comprehensive analyses of the regulatory sequences derived from human endogenous retroviruses

Jumpei Ito[1], Shiro Yamada[1], Ryota Sugimoto[1], Hirofumi Nakaoka[1], Ituro Inoue[1,2]
[1]Human Genetics, National Institute of Genetics, Japan, [2]The Graduate University For Advanced Studies (SOKENDAI)

Tue(3)-O30-3

RNA splicing is a primary link between genetic variation and disease

Yang I Li[1], Bryce van de Geijn[2], Anil Raj[1], David Knowles[1], Allegra Petti[3], David Golan[1], Yoav Gilad[2], Jonathan K Pritchard[1,4]
[1]Stanford University, USA, [2]University of Chicago, [3]Washington University in St. Louis, [4]Howard Hughes Medical Institute, Stanford University

Tue(3)-O30-4

Global patterns of copy number variation in humans from a population-based analysis

Jean Monlong[1,2], Caroline Meloche[3], Guy Rouleau[4], Patrick Cossette[3], Simon Girard[1,2], Guillaume Bourque[1,2]
[1]Human Genetics, McGill University, Montreal, Canada, [2]McGill University and Genome Quebec Innovation Center, Montreal, Canada, [3]Centre de Recherche du Centre Hospitalier de l'Universite de Montreal, Notre Dame Hospital, University of Montreal, Montreal, Quebec, Canada, [4]Montreal Neurological Institute and Hospital, McGill University, Montreal, Quebec, Canada

Tue(3)-O30-5

Identification and analysis of two novel enhancers of human SOX9: Implications for Disorders of Sex Development

Andrew H Sinclair[1], Thomas Ohnesorg[1], Jacqueline Tan[1], Jo Bowles[2], Peter Koopman[2], Vincent Harley[3]

[1]Molecular Development, Murdoch Children's Research Institute, Australia, [2]Institute for Molecular Bioscience, Queensland, [3]Hudson Institute of Medical Research, Victoria

Tue(3)-O30-6

First genome-wide CNV association meta-analysis on anthropometric traits in 71,288 adults

Aurelien Mace[1,2], Ruth JF Loos[3,4], Jacques S Beckmann[2], Sebastien Jacquemont[5], Andres Metspalu[6], Lude Franke[7], Timothy M Frayling[8], Alexandre Reymond[9], Zoltan Kutalik[2,10], GIANT Consortium

[1]Department of Medical Genetics, University of Lausanne, Switzerland, [2]Swiss Institute of Bioinformatics, University of Lausanne, Lausanne, Switzerland, [3]The Charles Bronfman Institute for Personalized Medicine, Icahn School of Medicine at Mount Sinai, New York, USA, [4]The Genetics of Obesity and Related Metabolic Traits Program, Icahn School of Medicine at Mount Sinai, New York, USA, [5]Service de Genetique Medicale, Centre Universitaire Hospitalier Vaudois, Lausanne, Switzerland, [6]Estonian Genome Center, University of Tartu, Tartu, Estonia, [7]University of Groningen, University Medical Center Groningen, Department of Genetics, Groningen, the Netherlands, [8]Genetics of Complex Traits, University of Exeter Medical School, Exeter, UK, [9]Center of Integrative Genomics, University of Lausanne, Lausanne, Switzerland, [10]Institute of Social and Preventive Medicine, University Hospital of Lausanne, Lausanne, Switzerland

15:40-17:10 Room K

O31 Concurrent Oral Session 31
"Genome structure, variation and function 2"

Chairs: **Michael A. Hauser**
Departments of Medicine and Ophthalmology, Duke Molecular Physiology Institute, Duke University, USA
Shinya Matsuura
Department of Genetics and Cell Biology, Research Institute for Radiation Biology and Medicine, Hiroshima University, Japan

Tue(3)-O31-1

Unraveling the role of genomic imprinting at 16q24.1 in pathogenetics of alveolar capillary dysplasia with misalignment of pulmonary veins and maternal uniparental disomy 16

Pawel Stankiewicz[1], Avinash V Dharmadhikari[1], Jenny J Sun J Sun[2], Brandi Carofino[1], Kadir Caner Akdemir[3], Claire Langston[4], Edwina Popek[4], Monica J Justice[5], Mary E Dickinson[6], Russell Ray[2], Partha Sen[7,8], Przemyslaw Szafranski[1]

[1]Department of Molecular and Human Genetics, Baylor College of Medicine, USA, [2]Department of Neuroscience, Baylor College of Medicine, Houston, Texas, USA, [3]Genomic Medicine Department, MD Anderson Cancer Center, Houston, Texas, USA, [4]Department of Pathology and Immunology, Baylor College of Medicine, Houston, Texas, USA, [5]Genetics & Genome Biology Program, SickKids, Toronto, Canada, [6]Department of Molecular Physiology & Biophysics, [7]Department of Pediatrics, Baylor College of Medicine, Houston, Texas, USA, [8]Department of Pediatrics, Northwestern University, Chicago, Illinois, USA

Tue(3)-O31-2

Genetic variants and cellular stressors associated with exfoliation syndrome modulate promoter activity of a lncRNA within the LOXL1 locus

Michael A Hauser[1,2,3,4], Inas F Aboobakar[2], Chiea-Chuen Khor[5], Allison E Ashley-Koch[1], Yutao Liu[6], Trevor R Carmichael[7], Susan E.I. Williams[7], Mineo Ozaki[8], Aung Tin[3,4], W. Daniel Stamer[2], R. Rand Allingham[2,3,4]

[1]Medicine, Duke University, USA, [2]Ophthalmology, Duke University, [3]Singapore Eye Research Institute, [4]Singapore National Eye Center, [5]Genome Institute of Singapore, [6]Cellular Biology & Anatomy, Georgia Regents University, [7]Ophthalmology, Neurosciences, University of the Witwatersrand, [8]Ozaki Eye Hospital

Tue(3)-O31-3

Genetic correlations between circulating miRNAs and lipid profiles reveal novel biomarkers of CVD risk

Joanne E Curran[1], Scott M McAhren[2], Satish Kumar[1], Juan Peralta[1], Hemant Kulkarni[1], Gerard Wong[3], Jacquelyn M Weir[3], Christopher K Barlow[3], Mark Kowala[2], Peter J Meikle[3], John Blangero[1], Laura F Michael[2]

[1]South Texas Diabetes and Obesity Institute, School of Medicine, University of Texas Rio Grande Valley, USA, [2]Lilly Research Laboratories, Eli Lilly and Company, Indianapolis IN, [3]Baker IDI Heart and Diabetes Institute, Melbourne AU

Tue(3)-O31-4

miRNA expression quantitative trait loci and parent-of-origin effects in human cell lines

Alexander W Drong[1], Quin Wills[2], Rory Bowden[1,2], George Nicholson[2], Sarah Keildson[1], Mahim Jain[5], Fredrik H Pettersson[1], George Davey Smith[3], Sue Ring[4], Mark I McCarthy[1,6], Chris Holmes[2], Nicholas J Timpson[3], Cecilia Lindgren[1]

[1]Wellcome Trust Centre for Human Genetics, University of Oxford, UK, [2]Department of Statistics, University of Oxford, Oxford, UK, [3]MRC Centre for Causal Analyses in Translational Epidemiology, School of Social and Community Medicine, University of Bristol, UK, [4]School of Social and Community Medicine, University of Bristol, UK, [5]Department of Molecular and Human Genetics, Baylor College of Medicine, Houston, TX, USA, [6]Oxford Centre for Diabetes Endocrinology and Metabolism, University of Oxford, Oxford, UK

Tue(3)-O31-5

Gene co-expression network analysis identifies gene modules associated with clinical phenotype in Williams syndrome

Ryo Kimura[1], Kiyotaka Tomiwa[2], Tomonari Awaya[3], Takeo Kato[3], Masatoshi Nakata[1], Yasuko Funabiki[4], Toshio Heike[3], Masatoshi Hagiwara[1]

[1]Anatomy and Developmental Biology, Kyoto University Graduate School of Medicine, Japan, [2]Todaiji Medical & Educational Center, [3]Pediatrics, Kyoto University Graduate School of Medicine, [4]Human Coexistence, Kyoto University Graduate School of Human and Environmental Studies

Tue(3)-O31-6

Diagnosis of bacterial and viral infection using a minimal host blood RNA expression signature

Myrsini Kaforou[1,2], Jethro A Herberg[1], Victoria J Wright[1], Clive J Hoggart[1], Andrew J Pollard[3], Saul N Faust[4,5], Sanjay Patel[5], Lachlan JM Coin[2,6], Federico Martinon-Torres[7], Jane C Burns[8,9], Michael Levin[1]

[1]Paediatrics, Medicine, Imperial College London, UK, [2]Genomics of Common Disease, School of Public Health, Imperial College London, UK, [3]Paediatrics, University of Oxford and the NIHR Oxford Biomedical Research Centre, Oxford, UK, [4]NIHR Wellcome Trust Clinical Research Facility, University of Southampton UK, [5]University Hospital Southampton NHS Foundation Trust, Southampton, UK, [6]Institute for Molecular Bioscience, University of Queensland, St Lucia, Queensland, Australia, [7]Translational Paediatrics and Infectious Diseases section, Department of Paediatrics, Hospital Clínico Universitario de Santiago, Santiago de Compostela, Galicia, Spain, [8]Department of Paediatrics, University of California San Diego, La Jolla, California, USA, [9]Rady Childrens Hospital San Diego, San Diego, California, USA

13:50-15:20 Room H

O32 Concurrent Oral Session 32
"Pharmacogenetics 1"

Chairs: **Filippo Martinelli Boneschi**
Department of Neuro-rehabilitation & INSPE, Scientific Institute San Raffaele, Italy
Hsin-Chou Yang
Institute of Statistical Science, Academia Sinica, Taiwan

Tue(3)-O32-1

Regulation of Mucocutaneous Inflammation by Cold Medicine-Related Stevens-Johnson Syndrome susceptibility gene, IKZF1

Mayumi Ueta[1,2], Hiromi Sawai[2], Junji Hamuro[4], Yuki Hitomi[2], Chie Sotozono[3], Katsushi Tokunaga[2], Shigeru Kinoshita[1]
[1]Department of Frontier Medical Science and Technology for Ophthalmology, Kyoto Prefectural University of Medicine, Japan, [2]Department of Human Genetics, Graduate School of Medicine, The University of Tokyo, [3]Department of Ophthalmology, Kyoto Prefectural University of Medicine, [4]Kyoto Prefectural University of Medicine

Tue(3)-O32-2

Pharmacogenetic and Epigenetic evaluation of CYP19 in PCOS patients underwent ovulation induction cycles

Parvaneh Afsharian[1], Shahrzad Ghazisaeidi[1,2], Zahra Ghezelayagh[1,2], Ali Asghar Akhlaghi[3], Marzieh Shiva[4], Maryam Shahosseini[1]
[1]Department of Genetics, Reproductive Biomedicine Research Center, Royan Institute for Reproductive Biomedicine, ACECR, Tehran, Iran, [2]Faculty of Basic Sciences and Advanced Technologies in Biology at University of Science and Culture, Tehran, Iran, [3]Department of Epidemiology and Reproductive Health, Reproductive Epidemiology Research Center, Royan Institute, ACECR, Tehran, Iran, [4]Department of Endocrinology and Female Infertility, Reproductive Biomedicine Research Center, Royan Institute for Reproductive Biomedicine, ACECR, Tehran, Iran

Tue(3)-O32-3

Clinical response to Nabiximols (Sativex®) on spasticity and pain is paralleled by a down-regulation of immune-related pathways in Multiple Sclerosis patients

Filippo Martinelli Boneschi[1,2], Melissa Sorosina[2], Laura Ferre'[1], Ferdinando Clarelli[2], Vittorio Martinelli[1], Federica Esposito[1,2], Giancarlo Comi[1,2]
[1]Scientific Institute San Raffaele, Italy, [2]Laboratory of Human Genetics of Neurological Disorders, Scientific Institute San Raffaele

Tue(3)-O32-4

A comprehensive analysis of genetic diversity in important pharmacogenes in the 1000 Genomes Project Phase 3 populations

Galen E.B. Wright[1,2,3], Bruce C. Carleton[2,4], Michael R. Hayden[1,2,3], Colin JD Ross[2,4]
[1]Medical Genetics, University of British Columbia, Canada, [2]Child and Family Research Institute, [3]Centre for Molecular Medicine and Therapeutics, [4]Pediatrics, University of British Columbia

Tue(3)-O32-5

Comprehensive exploration of the high-risk rare variants for the cold medicine-related Stevens-Johnson syndrome/ toxic epidermal necrolysis (CM-SJS/TEN) with Severe Ocular complications by whole-exome sequencing

Seik-Soon Khor[1,4], Yuki Hitomi[1,4], Mayumi Ueta[2,3,4], Hiromi Sawai[1], Khun Zawlatt[1], Chie Sotozono[3], Shigeru Kinoshita[2], Katsushi Tokunaga[1]

[1]Graduate School of Medicine, Department of Human Genetics, The University of Tokyo, Japan, [2]Department of Frontier Medical Science and Technology for Ophthalmology, Kyoto Prefectural University of Medicine, Kyoto, Japan, [3]Department of Ophthalmology, Kyoto Prefectural University of Medicine, Kyoto, Japan, [4]These author contributed equally to the work

Tue(3)-O32-6

Ancestry-informative pharmacogenomic loci

Hsin-Chou Yang, Chia-Wei Chen, Yu-Ting Lin, Shih-Kai Chu

Institute of Statistical Science, Academia Sinica, Taiwan

15:40-17:10 Room H

O33 Concurrent Oral Session 33 "Pharmacogenetics 2"

Chairs: **Mia Wadelius**
Medical Sciences, Clinical Pharmacology, Uppsala University, Sweden
Pei-Chieng Cha
Division of Molecular Brain Science, Kobe University Graduate School of Medicine, Japan

Tue(3)-O33-1

The influence of pharmacogenetics on the time to Acute Coronary Syndromes (ACS) recurrence in a UK cohort study

Peng Yin[1], Andrea Jorgensen[1], Andrew Morris[1], Richard Turner[2], Richard Fitzgerald[2], Rod Stables[3], Anita Hanson[2], Munir Pirmohamed[2]

[1]Department of Biostatistics, University of Liverpool, UK, [2]Department of Molecular & Clinical Pharmacology, University of Liverpool, [3]Liverpool Heart and Chest Hospital

Tue(3)-O33-2

Antithyroid drug-induced agranulocytosis is associated with different human leukocyte antigen alleles in Asia and Europe

Mia Wadelius[1], Niclas Eriksson[2], Luisa Ibanez[3], Emmanuelle Bondon-Guitton[4], Reinhold Kreutz[5], Alfonso Carvajal[6], Maribel Lucena[7], Esther Sancho Ponce[8], Javier Martin[9], Tomas Axelsson[10], Qun-Ying Yue[11], Patrik K Magnusson[12], Par Hallberg[1], EuDAC

[1]Medical Sciences, Clinical Pharmacology, Uppsala University, Sweden, [2]Uppsala Clinical Research Center and Department of Medical Sciences, Uppsala University, Uppsala, Sweden, [3]Fundacio Institut Catala de Farmacologia, Hospital Universitari Vall d'Hebron, Universitat Autonoma de Barcelona, Barcelona, Spain, [4]Service de Pharmacologie Médicale et Clinique, Centre Hospitalier Universitaire, Faculte de Medecine de l'Universite de Toulouse, Toulouse, France, [5]Charite - University Medicine, Institute of Clinical Pharmacology and Toxicology, Berlin, Germany, [6]Centro de Estudios sobre la Seguridad de los Medicamentos, Universidad de Valladolid, Valladolid, Spain, [7]Farmacologia Clinica, IBIMA, H Universitario Virgen de la Victoria, Universidad de Malaga, Malaga, Spain, [8]Capio Hospital General de Cataluna HGC, Sant Cugat del Valles, Spain, [9]Instituto de Parasitologia y Biomedicina Lopez Neyra Avda, Armilla, Granada, Spain, [10]Department of Medical Sciences, Molecular Medicine and Science for Life Laboratory, Uppsala University, Uppsala, Sweden, [11]Medical Products Agency, Uppsala, Sweden, [12]Swedish Twin Registry, Department of Medical Epidemiology and Biostatistics, Karolinska Institutet, Stockholm, Sweden

Tue(3)-O33-3

Neuronal enrichment analysis of treatment response in obsessive-compulsive disorder

Yin Yao[1], Haide Qin[1], Jack Samuels[2], Gerald Nestadt[2]
[1]Statistical Genomics, National Institute of Mental Health, USA, [2]Johns Hopkins Medical School, Department of Psychiatry

Tue(3)-O33-4

Genome-wide association study (GWAS) identifies genetic determinants of response to Zonisamide treatment in Parkinson's disease patients with "wearing-off"

Pei-Chieng Cha[1], Wataru Satake[1], Yuko Ando[1], Ken Yamamoto[2], Miho Murata[3], Tatsushi Toda[1]
[1]Division of Neurology/Molecular Brain Science, Kobe University Graduate School of Medicine, Japan, [2]Department of Medical Chemistry, Kurume University School of Medicine, Fukuoka, Japan, [3]Department of Neurology, National Center Hospital, National Center of Neurology and Psychiatry, Tokyo, Japan

Tue(3)-O33-5

Two component mixture modelling approach integrating genetic and clinical variables in analysis of time to remission in epilepsy

Ben R Francis[1], Andrea Jorgensen[1], Andrew Morris[1], Andres Ingasson[3], Anthony Marson[1], Michael Johnson[2], Graeme Sills[1], EpiPGX consortium
[1]Department of Biostatistics, University of Liverpool, UK, [2]Imperial College London, [3]deCODE

Tue(3)-O33-6

Genome-wide association study of L-asparaginase-induced pancreatitis in pediatric patients

Britt I Drogemoller[1], Hisaki Fujii[2], Shinya Ito[2], Bruce Carleton[1], Colin Ross[1], Canadian Pharmacogenomics Network for Drug Safety
[1]Pediatrics, University of British Columbia, Canada, [2]Clinical Pharmacology and Toxicology, The Hospital for Sick Children

Poster Session

Tuesday, April 5 17:30-18:30

Cancer Genetics 2

Tue(3)-P-1

Reducing genotoxicity emitted from diesel engines fueled with diesel/biodiesel/butanol blends

Yuan-Chung Lin[1,2], Chia-Chi Wang[2,3], Ying-Chi Lin[2,3], Po-Ming Yang[1], Syu-Ruei Jhang[1], Li-Jung Lin[4]
[1]Institute of Environmental Engineering, National Sun Yat-Sen University, Kaohsiung, Taiwan, [2]Ph.D. Program in Toxicology, College of Pharmacy, Kaohsiung Medical University, Taiwan, [3]School of Pharmacy, Kaohsiung Medical University, Taiwan, [4]Department of Biomedical Engineering, Oregon Health and Science University, USA

Tue(3)-P-2

Ultra-sensitive droplet digital PCR for detecting a low-prevalence somatic *GNAQ* mutation in Sturge-Weber syndrome

Yuri Uchiyama[1,2], Masakazu Miyajima[3], Masataka Taguri[4], Mitsuko Nakashima[1], Naomichi Matsumoto[1]
[1]Human Genetics, Yokohama City University Graduate School of Medicine, Japan, [2]Medicine and Clinical Science, Gunma University Graduate School of Medicine, [3]Neurosurgery, Juntendo University Graduate School of Medicine, [4]Biostatistics, Graduate school of Medicine, Yokohama City University

Tue(3)-P-3

MiR-200a, miR-200b, and *miR-429* Are Onco-miRs That Target PTEN Gene in Endometrioid Endometrial Carcinomas of The Uterus

Koichi Yoneyama[1], Osamu Ishibashi[2], Rieko Kawase[3], Akihito Yamamoto[3], Keisuke Kurose[3], Toshiyuki Takeshita[3]
[1]Obstetrics and Gynecology, Nippon Medical School Musashi Kosugi Hospital, Japan, [2]Laboratory of Biological Macromolecules, Graduate School of Life and Enviromental Sciences, Osaka Prefercture Unuversity, [3]Obstetrics and Gynecology, Nippon Medical School Hospital

Tue(3)-P-4

Furin, a pro-protein convertase, is a novel molecular target for c-Myc driven ovarian cancers

Junko Minato, Masafumi Toyoshima, Masumi Ishibashi, Shogo Sigeta, Toshinori Usui, Kazuyuki Kitatani, Nobuo Yaegashi
Tohoku University, Japan

Tue(3)-P-5

An Incidental Finding of Indolent T-PLL During a Routine Fertility Screen: A Case Study

Charmaine E Pollock
Cytogenetics, Sullivan Nicolaides Pathology, Australia

Tue(3)-P-6

Comprehensive genetic analysis of a pediatric pleomorphic myxoid liposarcoma reveals near-haploidization and loss of the *RB1* gene

Jakob P Hofvander[1], Vickie Y Jo[2], Iman Ghanei[3], David Gisselsson[1], Emma Martensson[1]
[1]Department of Clinical Genetics, Lund University, Sweden, [2]Department of Pathology, Brigham and Womens Hospital, [3]Department of Orthopedics, Skane University Hospital

Tue(3)-P-7

RNA-seq analysis of lung adenocarcinomas reveals different gene expression profiles between smoking and nonsmoking patients

Yafang Li[1], Xiangjun Xiao[1], Xuemei Ji[1], Bin Liu[2], Christopher Amos[1]
[1]Dartmouth College, USA, [2]MD Anderson Cancer Center

Tue(3)-P-8

Clinicopathological factors of breast cancer in women under 35 years: A retrospective statistical analysis

Masahiro Kitada, Nana Takahashi, Shyunsuke Yasuda, Kei Ishibashi, Satoshi Hayashi
Department of Breast Disease Center, Asahikawa Medical University, Japan

Tue(3)-P-9

A competing endogenous RNA (ceRNA) network regulates *KRAS* gene expression in human colorectal cancer cells

Marian Abigaile N Manongdo, John Paul T Rigor, Liezel U Tamon, Joshua Reginald P Malapit, Robert Lorenz C Chua, Jose Paulo E Lorenzo, Reynaldo L Garcia
National Institute of Molecular Biology and Biotechnology, University of the Philippines Diliman, Philippines

Tue(3)-P-10

Enhancing *SHP-1* and *PRG2* expression by 5-Azacytidine may inhibit STAT3 activation and confer sensitivity in Lestaurtinib (CEP-701) resistant *FLT3*-ITD positive Acute Myeloid Leukemia

Muhammad Farid Johan[1], Hamid AN Al-Jamal[1], Siti Asmaa Mat Jusoh[1], Shaharum Shamsuddin[2]
[1]Department of Haematology, Universiti Sains Malaysia, Malaysia, [2]School of Health Sciences, Universiti Sains Malaysia

Tue(3)-P-11

Correlation between *MGMT* promoter methylation and *hMSH2* mRNA expression in primary frontal high grade anaplastic glioma (HGAG)

Jeru Manoj Manuel[1], Chetan Ghati K[1], Narasinga Rao K V L[2], Venkatesh H N[1]
[1]Human Genetics, National Institute of Mental Health and Neurosciences (NIMHANS), India, [2]Neurosurgery, National Institute of Mental Health and Neurosciences (NIMHANS)

Tue(3)-P-12

Cytotoxic effects of Palladium (II) Complex on Prostate Cancer Cells

Hale Samli[1], Murat Samli[2], Nazlihan Aztopal[1], Buse Vatansever[1], Ozlem Sigva[1], Deniz Dincel[1], Cumhur Gunduz[3]
[1]Department of Genetics, Uludag University, Turkey, [2]Department of Urology, Acibadem University, [3]Department of Medical Biology, Ege University

Tue(3)-P-13

Detection of p53 gene polymorphisms in patients with Hepatocellular Carcinoma in India

Subramaniam Mohana Devi[1], Vellingiri Balachandar[2], Keshavarao Sasikala[1]
[1]Human Molecular Genetics Laboratory, Department of Zoology, Bharathiar University, India, [2]Department of Human Genetics and Molecular Biology, Bharathiar University, India

Tue(3)-P-14

High Mutation Detection Rate and Novel Mutations Identified in Major and Minor- Risk Cancer Genes by Applying Multigene Panels in Hereditary Cancer Clinic

Guy Rosner[1,2], Sivan Aharon Caspi[1,2], Merav Ben-Yehoyada [1,2], Dani Bercovich[3,4], Zamir Halpern[1,2], Erwin Santo[1,2], Revital Kariv[1,2]
[1]Gastroenterology, Tel-Aviv Sourasky Medical Center, Israel, [2]Sackler School of Medicine, Tel-Aviv University, Tel-Aviv, Israel, [3]Human Molecular Genetics and Pharmacogenetics, Migal - Galilee Bio-Technology Center, Kiryat Shmona, Israel, [4]Tel-Hai Academic College, Kiryat Shmona, Israel

Tue(3)-P-15

The importance of multidisciplinary approach to HBOC patients ~the experience in a general hospital~

Daisuke Takabatake, Kazuyuki Oishi
Breast Oncology, Kochi Health Science Center, Japan

Tue(3)-P-16

Hedgehog signaling and genetic diseases

Yoshiro Nakano[1], Kazuma Noguchi[2], Hideaki Chiyo[3,4], Ritsuko Pooh[3], Hiromitsu Kishimoto[2], Tomoko Hashimoto-Tamaoki[1,4]
[1]Genetics, Hyogo College of Medicine, Japan, [2]Oral Maxillofacial Surgery, Hyogo College of Medicine, [3]CRIFM Clinical Research Institute of Fetal Medicine, [4]Clinical Genetics, Hyogo College of Medicine

Tue(3)-P-17

Genomic copy number changes in CML patients with the Philadelphia chromosome (Ph+): an update

Yuan Ren[1,2], Young Mi Kim[1], Xianfu Wang[1], Xianglan Lu[1], Yue Gu[1], Mingran Sun[1], Yunpeng Shi[1], Jianqin Zhang[1], Shibo Li[1], Lijun Zhang[2]
[1]Pediatrics, The university of Oklahoma Health Science Center, China, [2]Hematology, The First Affiliated Hospital of China Medical University

Tue(3)-P-18

Exploring clinicians' attitudes about using aspirin for risk reduction in people with Lynch Syndrome with no personal diagnosis of colorectal cancer

Yanni Chen[1,3], Bettina Meiser[2], Rajneesh Tim[2], Michelle Peate[4], Judy Kirk[5], Robyn Ward[4], Annabel Goodwin[6], Finlay Macrae[4], Janet Hiller[7], Alison Trainer[8], Gillian Mitchell[9]
[1]School of Medicine, University of Sydney, Singapore, [2]University of New South Wales, [3]National Cancer Centre Singapore, [4]University of Melbourne, [5]Westmead Millennium Institute, [6]Concord Cancer Centre, [7]Swinburne University of Technology, [8]Peter MacCallum Cancer Centre, [9]British Columbia Cancer Agency

Tue(3)-P-19

Genetic and epigenetic alterations of netrin-1 receptors in gastric cancer

Takeshi Nagasaka[1], Yoshiko Mori[1,2], Kunitoshi Shigeyasu[1], Shinichi Toyooka[2,3], Toshiyoshi Fujiwara[1]
[1]Gastroenterological Surgery, Okayama University Graduate School of Medicine, Dentistry and Pharmaceutical Sciences, Japan, [2]Clinical Genomic Medicine, Okayama University Graduate School of Medicine, Dentistry and Pharmaceutical Sciences, [3]General Thoracic Surgery, Okayama University Graduate School of Medicine, Dentistry and Pharmaceutical Sciences

Tue(3)-P-20

Functional characterization of RNAi-mediated regulation of the tumor suppressor gene Neurofibromin 2 (*NF2*)

Krizelle Mae M. Alcantara, Pixie Dale S. Alvarez, Reynaldo L. Garcia
Disease Molecular Biology and Epigenetics Laboratory, National Institute of Molecular Biology and Biotechnology, Philippines

Tue(3)-P-21

CpG island methylator phenotype is associated with the efficacy of chemotherapy for metastatic colorectal cancer

Hideki Shimodaira[1,2], Xiaofei Zhang[1], Keigo Komine[1,2], Shin Takahashi[1,2], Masanobu Takahashi[1,2], Chikashi Ishioka[1,2]
[1]Department of Medical Oncology, Institute of Development, Aging and Cancer, Tohoku University, Japan, [2]Department of Clinical Oncology, Tohoku University Hospital

Tue(3)-P-22

BRCA1/2 Mutation Dependent Effect on Survival of Advanced Stage Ovarian Cancer

Ramunas Janavicius[1,2], Vilius Rudaitis[3], Dovile Janulynaite[1], Laimonas Griskevicius[1]
[1]Hematology, Oncology and Transfusion Medicine Center, Vilnius University Santariskiu Hospital Clinics, Lithuania, [2]State Research Innovative Medicine Center, [3]Oncogynecology Unit, Vilnius university Santariskiu Hospital Clinics

Tue(3)-P-23

Expression of SYK and BTK in Different Breast Cancer Phenotypes of Brunei Patients

Mas Rina Wati Haji Abdul Hamid[1], Nur Izzyan Bungsu[1], Abdalla Mohamed Jama[2], Pg Bahrin Pg Haji Aliudin[3], Juniada Haji Jumat[3], P U Telisinghe[3]
[1]PAPRSB Institute of Health Sciences, Universiti Brunei Darussalam, Brunei Darussalam, [2]Faculty of Science, Universiti Brunei Darussalam, [3]Raja Isteri Pengiran Anak Saleha Hospital, Brunei Darussalam

Tue(3)-P-24

A case with pachydermoperiostosis with gastrointestinal malignancy

Mariko Kakudo[1], Hisatomo Ikehara[2], Hironori Niizeki[3], Kazuhiko Nakabayashi[4], Chika Sato[1], Hiroko Mimura[1], Tadayuki Oshima[2], Jiro Watari[2], Seiichi Hirota[5], Hiroto Miwa[2], Tomoko Hashimoto-Tamaoki[1,6]
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Tue(3)-P-25

Integrated analysis of whole-exome and transcriptome sequencing in signet ring cell carcinoma of colorectum

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Tue(3)-P-27

Revealing hidden complexity of the cancer genome with the aid of RNA sequencing

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Tue(3)-P-28

Targeted semiconductor sequencing of 409 cancer-related genes for somatic mutations and copy number variations in multiple myeloma

Hisayo Fukushima[1,2], Hiroshi Ikeda[3], Kazuya Ishiguro[3], Tetsuyuki Igarashi[3], Yuka Aoki[3], Tadao Ishida[3], Miyuki Tamura[2], Yasushi Sasaki[2], Akihiro Sakurai[1], Takashi Tokino[2]
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Tue(3)-P-29

Variants of uncertain significance in BRCA: Experience from the Japanese HBOC consortium trial survey

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Tue(3)-P-30

Reduction of physiologic endogenous DNA double strand breaks advances genomic instability in chronological aging yeast

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Tue(3)-P-31

ß-catenin controls the expression profiles of *KCNQ1OT1/LIT1 l*ong noncoding RNA

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Tue(3)-P-32

KRAS mutations and intratumoral heterogeneity in rectal adenomas and early carcinomas

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Tue(3)-P-33

Action of the hereditary breast cancer ovarian cancer syndrome practice cooperation in Kochi

Fuminori Aki[1,2], Sueyoshi Ito[1], Takeki Sugimoto[2], Syuin Taro[2], Mari Tashiro[2], Koki Hirano[3], Daisuke Takabatake[2,4], Kazuyuki Oisi[2,4], Ryuhei Nagai[2,4], Yusei Nakata[2,4], Ichiro Yamasaki[2], Nobuo Ikenoue[2], Chiaki Izumiya[2], Kenji Matsushita[2], Toru Kubo[2]
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Tue(3)-P-34

Genomic and epigenetic changes underlying retinoblastoma tumors

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Tue(3)-P-35

Identification of driver gene mutations and fusion events in patients with Sezary Syndrome

Aparna Prasad[1,2,3], Raquel Rabionet[1,2,3], Anna Puiggros[4], Luis Zapata[1,2,3], Carme Melero[4], Anna Puig[1,2,3], Yaris Sarria Trujillo[1,2,3], Blanca Espinet[4], Fernando Gallardo[5], Ramon Pujol[5], Xavier Estivill[1,2,3]
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Tue(3)-P-36

Early development of rare tumors in individuals with congenital malformation syndrome

Mari Minatogawa[1], Fuminori Iwasaki[2], Kunio Fukuda[2], Chihiro Hatano[1], Takayuki Yokoi[1], Yumi Enomoto[1], Kazumi Ida[1], Yoshinori Tsurusaki[1], Noriaki Harada[1], Toshiyuki Saitou[1], Junichi Nagai[1], Hiroaki Goto[2], Kenji Kurosawa[1]
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Tue(3)-P-37

The Role of SLURP-1 in melanoma promoting microenvironment

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Tue(3)-P-38

SNPs in MEG3 lncRNA could alter its tumor suppressive capacity

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Tue(3)-P-40

Preliminary Investigations on the Putative PIK3CA-ZNF148 ceRNA Network

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Tue(3)-P-41

Model comparison of cancer genetics practice in Japan: How to provide genetic counseling, genetic testing, cancer surveillance, and risk-reducing options for people who may have high cancer risks

Chieko Tamura[1,2,3], Yukiko Tsunematsu[2], Miki Kimura[2], Kohji Tanakaya[3], Keiko Matsuda[3,4], Chikako Nishida[3], Hiromi Sanai[3,5], Hiromi Arakawa[1], Yasushi Nakamura[1]
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Tue(3)-P-42

The new subcellular role of BRCA2 involved in FIP3-dependent endosome function

Miho Takaoka, Akira Nakanishi, Yoshio Miki
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Tue(3)-P-43

Can surgeon provide early BRCA gene counseling for advanced ovary cancer?

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Tue(3)-P-44

Overexpression of miR-127 and miR-375 in Medullary Thyroid Carcinoma Tumors

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Tue(3)-P-45

Germline Mutational Analysis of *RET* Proto Oncogene in Iranian Medullary Thyroid Carcinoma Patients: a 14-year Study

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Tue(3)-P-46

Genome-wide DNA copy number analysis of primary head and neck squamous cell carcinoma (HNSCC) and second primary esophageal squamous cell carcinoma (ESCC)

Meng-Shin Shiao[1], Sacarin Bunbanjerdsuk[2], Tanjitti Pongrujikorn[2], Tanadech Dechaphunkul[3], Somkiat Sunpaweravong[4], Natini Jinawath[1,2]
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Tue(3)-P-47

Evaluation of propolis effect on microRNA expression profiling in acute lymphoblastic leukemia cell lines

Ozgur Cogulu[1], Ugur Cem Yilmaz[2], Emin Karaca[1], Asude Durmaz[1], Burak Durmaz[1], Ayca Aykut[1], Cigir Biray Avci[3], Sunde Yilmaz Susluer[3], Bakiye Goker[3], Hüsniye Kayalar[4], Cumhur Gunduz[3]
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Tue(3)-P-48

Clinical utility of genomic SNP microarrays in hepatosplenic γδ T-cell lymphoma

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Tue(3)-P-49

Roles of Tyrosine Phosphorylation of histone H4 in Breast Cancer

Ruey-Hwang Chou[1], Ying-Nai Wang[2], Wei-Chao Chang[1], Ling-Chu Chang[3], Weiya Xia[2], Yung-Luen Yu[1], Mien-Chie Hung[1,2]
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Tue(3)-P-50

Targeted Single Cell Sequencing for Accurate Mutation Detection in Heterogeneous Cellular Populations

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Tue(3)-P-51

Global nuclear radial distribution of chromosome territories in various cancer cell lines

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Tue(3)-P-52

Nonclonal chromosome abnormalities in hematologic malignancies

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Complex Traits and Polygenic Disorders 2

Tue(3)-P-53

Replication of 49 type 2 diabetes associated risk variants in Punjabi Pakistani population

Asima Zia[1], Xingbin Wang[2], Attya Bhatti[1], F.Y Demirci[2], Wei Zhao[3], Asif Rasheed[4], Maria Samuel[4], Aysha K Kiani[1], Muhammad Ismail[5], Jamal Zafar[6], Peter John[1], Danish Saleheen[4,7], M I Kamboh[2]
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Tue(3)-P-54

Copy number variations play important roles in heredity of common diseases: a novel method to calculate heritability of a polymorphism

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Tue(3)-P-55

Determination of IFIT1 Gene Polymorphisms on Human Cerebral Malaria in Thai Population

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Tue(3)-P-56

Genome-wide association studies (GWAS) for adult height and body mass index in the Japanese population using the JPDSC database

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Tue(3)-P-57

Mutation identification of *ABCA1* gene in subjects with low level of high-density lipoprotein

Nani Maharani[1,2], Udin Bahrudin[1,3], Hesty Wahyuningsih[1], Isna R Fara[1], Ferdy K Cayami[1], Sodiqur Rifqi[3], Sultana MH Faradz[1], Ichiro Hisatome[1,2]
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Tue(3)-P-58

Leveraging Compartmental Modeling to Assess the Pathophysiologic Effect of Genetic Variation Underlying Risk for Type 2 Diabetes

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Tue(3)-P-59

Location, Loci or Lifestyle? Dissecting Health-Associated Regional Variation in Scotland

Carmen Amador[1], Charley Xia[1], Archie Campbell[1], David Porteous[1], Generation Scotland[3], Nick Hastie[1], Veronique Vitart[1], Caroline Hayward[1], Pau Navarro[1], Chris S Haley[1,2]
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Tue(3)-P-60

Effects of HLA-DPB1 genotypes on HBV-related diseases in Japanese population

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Tue(3)-P-61

Associations between the orexin (hypocretin) receptor 2 gene polymorphism Val308Ile and nicotine dependence in genome-wide and subsequent association studies

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Tue(3)-P-62

Meta-analysis of GWAS followed by replication identifies new susceptibility genes on X chromosome for SLE in cross-ethnic populations

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Tue(3)-P-63

The power of family: Linkage Analysis vs GWAS in family-based cohorts

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Tue(3)-P-64

Decreased Severity of Experimental Autoimmune Arthritis in Peptidylarginine Deiminase Type 4 Knockout Mice

Akari Suzuki[1], Yuta Kochi[1], Hirofumi Shoda[2], Keishi Fujio[2], Kazuhiko Yamamoto[1,2]

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Tue(3)-P-65

A replication study of four candidate loci for sex hormone levels previously identified by genome-wide association studies

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Tue(3)-P-66

Genome-wide Association Studies on Immunoglobulin G Glycosylation Patterns

Annika Laser[1,2], Lucija Klaric[3,4,5], Elisa Benedetti[6], Marija Pezer[4], Marian Beekman[7], Joris Deelen[7], Anton J.M. de Craen[8], Manfred Wuhrer[9], Rosina Plomp[9], Harald Grallert[1,2,10], Jan Krumsiek[6,10], Konstantin Strauch[11,12], Annette Peters[2], Thomas Meitinger[13], P. Eline Slagboom[9], Gordan Lauc[4,14], Christian Gieger[1,2]

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Tue(3)-P-67

The *crispld2* story: From gene function to new NSCLP candidate gene identification

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Combined genome-wide linkage and association analysis of depressive symptomes point to *CTNNA3* gene

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Tue(3)-P-69

Shared and unique genetic determinants between pediatric and adult celiac disease

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Tue(3)-P-70

The role of regulatory single-nucleotide genetic polymorphisms of placental tissue in the development of preeclampsia in different ethnic groups

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Tue(3)-P-71

Genetic Association of *HLA-C* Region with Kawasaki Disease in the Korean Population

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Tue(3)-P-72

Dissecting the unexpected role of *PAG1* in asthma

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Tue(3)-P-73

Gene-gene interaction for markers in 16p13.3 may contribute to the risk of non-syndromic cleft lip with or without palate in Chinese population

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Tue(3)-P-74

Association of serum biotin and total/specific IgE levels and a common locus for biotin and cedar pollen-specific IgE levels

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Tue(3)-P-75

Genome-wide association study identified new susceptible genetic variants in HLA class I region for hepatitis B virus-related hepatocellular carcinoma

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Metabolomic and transcriptomic fingerprints of menopausal hormone therapy in 3479 Finnish women

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Genome-wide association study on cephalic form in Japanese

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Tue(3)-P-78

Identification and Replication of Height Loci in African Ancestry Populations

Maggie C.Y. Ng[1], Mariaelisa Graff[2], Anne Justice[2], Ying Chang Lu[3], Poorva Mudgal[1], Ching-Ti Liu[4], Kristin Rand[5], Qing Duan[2], Brian E. Cade[6], Jennifer Brody[7], Mary K. Wojczynski[8], Mary F. Feitosa[8], Lisa R. Yanek[9], Michael A. Nalls[10], Leslie Lange[2], Sailaja Vedantam[11], Xiuqing Guo[12], Christopher A. Haiman[5], Ruth J.F. Loos[3], Kari E North[2], the African Ancestry Anthropometry Genetics Consortium
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Tue(3)-P-79

The h^4 curse of genomic prediction

Xia Shen[1,2], Zheng Ning[1], Youngjo Lee[3], William G. Hill[2], Chris S. Haley[2], Yudi Pawitan[1]
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Tue(3)-P-80

Frequent Potential Mutations in 378 Candidate Genes of Glaucoma Identified by Whole Exome Sequencing of 257 Patients with Glaucoma

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Tue(3)-P-81

Functional annotation of chronic pancreatitis-associated intronic variants in the SPINK1 gene

Wen-Bin Zou[1,2,3,4], Arnaud Boulling[1,3], Emmanuelle Masson[1,5], David N. Cooper[6], Zhuan Liao[2,4], Zhao-Shen Li[2,4], Claude Ferec[1,3,5,7], Jian-Min Chen[1,3,7]
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Tue(3)-P-82

Multi-trait GWAS method comparison and application of summary statistic methods to publicly available data

Heather F Porter, Samir De Marchi, Paul F O'Reilly
SGDP Centre, IoPPN, King's College London, UK

Tue(3)-P-83

Systems Genetics of Plasma ^{1}H Nuclear Magnetic Resonance Metabotypes Associated with Cardiometabolic Diseases in a Lebanese Cohort

Andrea Rodriguez-Martinez[1], Michael Kyriakides[1], Nikita Gandhi[1], Jean-Baptiste Cazier[2], Joram M. Pasma[1], Jeremy.K Nicholson[1], Dominique Gauguier[3], Pierre Zalloua[4], Marc-Emmanuel Dumas[1]
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Tue(3)-P-84

Transcriptomic analysis to identify biological markers for antibody production introduced by seasonal influenza vaccination

Maiko Narahara[1], Toby Hocking[2], Guillaume Bourque[2], Mark Lathrop[2], Fumihiko Matsuda[1]
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Tue(3)-P-85

Characterizing the genetic architecture of ocular biometrics in an untested south Asian population: the Jirels of eastern Nepal (Jiri Eye Study)

Matthew P Johnson[1], Suman S Thapa[2], Kent L Anderson[3], Sandy Laston[1], Mohan K Shrestha[2], Bradford Towne[4], Janardan Subedi[5], John Blangero[1], Sarah Williams-Blangero[1]
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Tue(3)-P-86

Functional variants in a clinical setting: an example using *APOC3* R19X and extreme triglyceride levels extracted from electronic health records

Dana C. Crawford[1,2], Kirsten E. Diggins[3], Nicole A. Restrepo[1,2], Eric Farber-Eger[4], Quinn S. Wells[5,6]
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Tue(3)-P-87

Genetic variants of *FTO, LEPR, MC4R, PON1, SCL6A4, DRD2, MAOA* and *COMT*, associated to the genetic risk for overweight and obesity in children from Yucatan, Mexico

Lizbeth Gonzalez-Herrera[1], Maria Jose Lopez Gonzalez[1], Ruvy Alvarado-Vargas[1], Zenda Cardena-Carballo[1], Didier May-Hau[1], Gerardo Perez-Mendoza[1], Doris Pinto-Escalante[1], Guadalupe Garcia-Escalante[1], Fernando Herrera-Sanchez[2], Rodrigo Rubi-Castellanos[1]
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Tue(3)-P-88

Improved imputation accuracy using population-specific SNP array and haplotype reference panel

Yosuke Kawai, Takahiro Mimori, Kaname Kojima, Naoki Nariai, Kazuharu Misawa, Yumi Yamaguchi-Kabata, Yukuto Sato, Inaho Danjoh, Rumiko Saito, Fumiki Katsuoka, Jun Yasuda, Masayuki Yamamoto, Masao Nagasaki
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Tue(3)-P-89

Allelic imbalance in regulation of *ANRIL* through chromatin interaction at 9p21 endometriosis risk locus

Hirofumi Nakaoka, Aishwarya Gurumurthy, Takahide Hayano, Somayeh Ahmadloo, Waleed Omer, Kazuyoshi Hosomichi, Ituro Inoue
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Tue(3)-P-90

Stability profiling of HLA class II protein for disease association studies

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Tue(3)-P-91

SNP-set Kernel Association Tests (SKAT) of the association between interleukin (IL) polymorphisms and risk of *H. pylori* infection and related gastric atrophy

Asahi Hishida[1], Kenji Wakai[1], Mariko Naito[1], Hideo Tanaka[2]
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Tue(3)-P-92

Assessment of somatic DNA methylation profiling and copy number variations in atherosclerosis

Maria S. Nazarenko[1,2], Anton V. Markov[1,2], Aleksei A. Sleptcov[1,2], Igor N. Lebedev[1,2], Nikolay A. Skryabin[1,2], Aleksei V. Frolov[3], Olga L. Barbarash[3], Valery P. Puzyrev[1,2]
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Tue(3)-P-93

Predictive utility of a genetic risk score of common variants associated with type 2 diabetes in a black South African population

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Center of Excellence, North West University, South Africa

Tue(3)-P-94

Studies of Genetics & Environment interaction and health longevity in Bama population

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Tue(3)-P-95

Analysis of the regulatory role of fifty-two genetic loci influencing human electrically active myocardial mass on epigenetic human heart data

Daiane Hemerich[1,2], Vinicius Tragante[1], Jaiyi Pei[3], Jessica van Setten[4], Magdalena Harakalova[1], Michal Mokry[5], Pim van der Harst[6], Folkert W. Asselbergs[1,7,8]
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Clinical Genetics and Dysmorphology 2

Tue(3)-P-96

Waardenburg Syndrome Type IIE in a Japanese Patient Caused by a Novel Missense Mutation in the *SOX10* Gene

Tamio Suzuki[1], Ken Okamura[1], Naoki Oiso[2], Gen Tamiya[3], Satoshi Makino[3], Daishi Tsujioka[4], Yutaka Hozumi[1], Yoshikazu Shimomura[5], Yuko Abe[1]
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Tue(3)-P-98

A novel *HSF4* missense mutation in Iranian siblings causes autosomal recessive congenital cataract

Eri Imagawa[1], Mahdiyeh Behnam[2], Ahmad R Salehi[3], Firooze Ronasian[2], Mansoor Salehi[4], Noriko Miyake[1], Naomichi Matsumoto[1]
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Tue(3)-P-99

Tatton-Brown-Rahman syndrome due to 2p23 microdeletionTatton-Brown-Rahman syndrome due to 2p23 microdeletion

Kimiko Ueda[1], Nobuhiko Okamoto[1], Yasuhisa Toribe[2], Keiko Shimojima[3], Toshiyuki Yamamoto[3]
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Tue(3)-P-100

Mutational Analysis of *TSC1* and *TSC2* in Japanese Patients with Tuberous Sclerosis Complex Revealed Higher Incidence of TSC1 Patients than Previously Reported and unique *TSC1* mutational pool

Yo Niida[1], Mamoru Ozaki[1], Akiko Wakisaka[2], Takanori Tsuji[2], Mondo Kuroda[3], Yusuke Mitani[3], Ayano Yokoi[3]
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Tue(3)-P-101

***WDR62*/*MCPH2* mutations identified in patients with primary microcephaly by a combined approach of exome sequencing and genome editing technology**

Yoshinori Masatsuna[1], Silvia Natsuko Akustu[1], Kosuke Hosoba[1], Hiroyuki Morino[2], Hideki Kawakami[2], Takashi Yamamoto[3], Kenji Simizu[4], Hirofumi Oohashi[4], Tatsuo Miyamoto[1], Shinya Matsuura[1]
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Tue(3)-P-102

Dyschromatosis symmetrica hereditaria complicated by intracranial hemangiomas and Parry-Romberg syndrome

Kazuyoshi Fukai[1], Saki Yanagihara[1], Daisuke Tsuruta[1], Toshiyuki Seto[2], Taro Shimono[3], Ken Okamura[4], Yutaka Hozumi[4], Tamio Suzuki[4]
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Tue(3)-P-103

Clinical features and outcomes for infants with an antenatal/ perinatal diagnosis of Tuberous Sclerosis

Clara WT Chung[1], John A Lawson[2], Sean E Kennedy[3], Stephen Cooper[4], Vanessa Sarkozy[5], Orli Wargon[6], Jacqueline Robinson[1], Harry King[1], David R Mowat[1]
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Tue(3)-P-104

Clinical and genetic diagnosis of HPT-JT syndrome

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Tue(3)-P-105

De novo DNM1 mutations in two cases of epileptic encephalopathy

Mitsuko Nakashima[1], Takeshi Kouga[2,3], Charles Marques Lourenco[4], Masaaki Shiina[5], Tomohide Goto[3], Yoshinori Tsurusaki[1], Satoko Miyatake[1], Noriko Miyake[1], Hirotomo Saitsu[1], Kazuhiro Ogata[5], Hitoshi Osaka[2], Naomichi Matsumoto[1]
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Tue(3)-P-106

Renal tubular dysgenesis and intellectual disability with uniparental disomy of chromosome 1

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Tue(3)-P-107

Novel mutation in the *COL1A1* gene causes severe scoliosis and valvular heart disease in a Japanese family with osteogenesis imperfecta

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Tue(3)-P-108

Clinical utility of medical exome analysis in a tertiary pediatric referral center

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Tue(3)-P-109

Gross insertion in *FBN1* causes Marfan syndrome

Takayuki Yokoi[1], Chihiro Hatano[1], Yoshinori Tsurusaki[1], Yumi Enomoto[1], Takuya Naruto[2], Kenji Kurosawa[1]
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Tue(3)-P-110

Identified novel *FBN1* gene mutation in neonatal/ infantile Marfan syndrome

Yoo-MI Kim[1], Ji-Na Kim[1], Gil-Ho Ban[1], Young-Mi Han[1], Shin Youn Byun[1], Seung Kook Son[1], Yeon Joo Lee[1], Chong Kun Cheon[1], Gu-Hwan Kim[2], Han-Wook Yoo[2], Hyung-Doo Lee[1]
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Tue(3)-P-111

Oro-dental phenotypes associated with rare monogenic disorders

Emilia Severin[1], Octavian Dinca[1], Cristian Vladan[1], Dana Bodnar[1], Cristina Dragomir[2], Alexandru Bucur[1]
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Tue(3)-P-112

Haddad Syndrome due to PHOX2B Gene Mutation in a Filipino Infant

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Tue(3)-P-113

The Management of Pregnancy in Two Japanese Sisters who Developed Deep Vein Thrombosis with Congenital Antithrombin Deficiency

Yukiko Mikami[1], Sumiko Era[1], Yoshihisa Ono[1], Masahiro Saito[1], Yasushi Takai[1], Kazunori Baba[1], Hiroyuki Seki[1], Keiko Shinozawa[2], Katsuyuki Fukutake[2,3]
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Tue(3)-P-114

Natural history of motor function changes in childhood-onset spinal muscular atrophy

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Tue(3)-P-115

Homozygous 4-bp deletion in the *DDHD1* gene, resulting the complete deletion of DDHD domain, as a causative variant in a SPG28 patient

Takuya Morikawa[1], Shiroh Miura[1,2], Kohei Yamada[1], Gohsuke Hattori[3], Kengo Kosaka[1], Ryuta Fujioka[4], Manabu Motomura[5], Takayuki Taniwaki[2], Hiroki Shibata[1]
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Simultaneous detection of both single nucleotide variations and copy number alterations by next-generation sequencing in Gorlin syndrome

Kei-ichi Morita[1,2,3], Takuya Naruto[4], Kousuke Tanimoto[2,5,6], Chisato Yasukawa[1], Yu Oikawa[1], Kiyoshi Masuda[7], Issei Imoto[7], Johji Inazawa[2,3,6], Ken Omura[1,3,8], Hiroyuki Harada[1]
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Tue(3)-P-117

Mutation in the genes encoding eukaryotic translation initiation factor 2B in Japanese patients with vanishing white matter disease

Shino Shimada[1], Keiko Shimojima[2], Toshiyuki Yamamoto[2], Satoru Nagata[1]
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Tue(3)-P-119

Clinical courses and experiences of seven patients with Ehlers-Danlos syndrome caused by CHST14/D4ST1 deficiency

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Tue(3)-P-120

Next-generation sequencing identifies novel *ARID1B* mutations in patients with Coffin-Siris syndrome

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Tue(3)-P-121

The comprehensive genetic analysis of Rubinstein-Taybi syndrome (RSTS)

Yumi Enomoto[1], Takayuki Yokoi[2], Chihiro Hatano[2], Ikuko Ohashi[2], Yukiko Kuroda[2], Yoshinori Tsurusaki[1], Kazumi Ida[1], Takuya Naruto[1], Kenji Kurosawa[2]
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Tue(3)-P-122

A case of mandibulofacial dysostosis with microcephaly presenting with epilepsy

Mari Matsuo[1], Masako Sakauchi[2], Akemi Yamauchi[1], Yasushi Ito[2], Toshiyuki Yamamoto[3], Nobuhiko Okamoto[4], Yoshinori Tsurusaki[5], Noriko Miyake[5], Naomichi Matsumoto[5], Kayoko Saito[1]
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Tue(3)-P-123

Long-term clinical feature of West syndrome with a de novo mutation in *NR2F1*: A case report

Naomi Hino-Fukuyo[1,2], Atsuo Kikuchi[2], Natsuko Arai-Ichinoi[2], Tetsuya Niihori[3], Ryo Sato[2], Tasuku Suzuki[2], Hiroki Kudo[2], Ryo Funayama[4], Keiko Nakayama[4], Yoko Aoki[3], Shigeo Kure[2]
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Tue(3)-P-124

Two Japanese patients with two genes mutations, showing congenital sensorineural hearing loss

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Tue(3)-P-125

Brain morphology in children with nevoid basal cell carcinoma syndrome

Tadashi Shiohama[1], Katsunori Fujii[1], Toshiyuki Miyashita[2], Hideki Uchikawa[1,3], Hiromi Mizuochi[1,3], Hajime Ikehara[1], Tomoyuki Fukuhara[1], Naoki Shimojo[1]
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Tue(3)-P-126

Genetic evaluation of patients with intellectual disability (ID) using chromosomal microarray and targeted next-generation sequencing at the "ID clinic"

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A nationwide survey on genetically confirmed Danon disease in Japan

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Mental and physical development study of long-term survival patients of thanatophoric dysplasia

Hideaki Sawai, Mariko Ushioda, Study group of thanatophoric dysplasia by grant-in-aid of Ministry of Health, Labour and Welfare
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Microform holoprosencephaly with bilateral congenital elbow dislocation; a further case of Steinfeld syndrome related to a *CDON* mutation?

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Novel compound heterozygous mutations in *ISPD* gene from two cases of Japanese Walker-Warburg syndrome identified by whole-exome sequencing

Yonehiro Kanemura[1,2], Fuyuki Miya[3,4], Tomoko Shofuda[5], Ema Yoshioka[5], Daisuke Kanematsu[1], Kyoko Itoh[6], Shinji Fushiki[6], Takeshi Okinaga[7], Haruhiko Sago[8], Rika Kosaki[9], Kyoko Minagawa[10], Nobuhiko Okamoto[11], Tatsuhiko Tsunoda[3,4], Mitsuhiro Kato[12], Shinji Saitoh[13], Kenjiro Kosaki[14], Mami Yamasaki[15]

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The Myhre syndrome: report of a Japanese Female Case

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The minor nasopharyngeal anomaly in a family of Hypoparathyroidism, Deafness, and Renal dysplasia (HDR) syndrome

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SATB2-associated syndrome presenting with Rett like phenotype identified by whole exome sequencing

Jin Sook Lee[1], Yongjin Yoo[2], Jae So Cho[3], Hyewon Woo[3], Woojoong Kim[3], Byung Chan Lim[3], Ki Joong Kim[3], Murim Choi[2], Jong-Hee Chae[3]

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Mismatch repair cancer syndrome caused by homozygous deletion of exons 13-14 of *PMS2* gene

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Clinical and genetic characterization of patient with *SOX5* haploinsufficiency caused by *de novo* balanced chromosomal translocation

Nobuaki Wakamatsu[1], Daisuke Fukushi[1], Kaoru Suzuki[1], Noriko Nomura[1], Yasuyo Suzuki[1], Kenichiro Yamada[1], Mie Inaba[2], Seiji Mizuno[2]

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A Novel Mutation in the Flavin-Containing Monooxygenase 3 Gene (FMO3) of a Korean Family Causes Trimethylaminuria

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Delineation of the molecular basis of borderline hemoglobin A2 in Chinese individuals

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Monochorionic Diamniotic Twins With 45,X/46,XY Mosaic Who Showed Different External Genitals Due To Different Rates of Mosaicism: A Case Report

Taisuke Sato[1,3], Ken Takahashi[2,3], Yuki Ito[1,3], Aiko Sasaki[2], Aikou Okamoto[3], Kenichiro Hata[1], Haruhiko Sago[2]
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Multisystem involvement and progressive course in Woodhouse-Sakati syndrome: from detailed, comprehensive, and longitudinal observation of the first East Asian patient

Motoko Kamiya[1,2,3,4], Tomomi Yamaguchi[1], Kyoko Takano[1,2], Masanori Yamazaki[1,5], Masanori Yasuo[6], Maiko Miyagawa[7], Shin-ichi Usami[7], Akane Minagawa[8], Jun Takahashi[9], Masafumi Kanai[10], Kazuki Hirabayashi[11], Katsuya Nakamura[1,2], Masumi Ishikawa[1], Emiko Kise[1], Keiko Wakui[1,2], Yoshimitsu Fukushima[1,2], Tomoki Kosho[1,2]
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Caffey disease (CD) or infantile cortical hyperostosis: a novel mutation in *COL1A1* detected in a Chilean patient evidences the locus heterogeneity of the disease

Fanny Cortes[1,2], Karla Moenne[1,3], Ximena Ortega[1,3], Alejandro Baar[1,4], Alejandro Veloz[1,3,5], Takeshi Asahi[1,3,5]
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A novel RMRP mutation in a cartilage-hair dysplasia patient

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DLX6, *MSX1*, AND *EDN1*: DIFFERENTIAL EXPRESSION IN HUMAN MANDIBLES THAT POTENTIALLY REGULATE MANDIBULAR SIZE

Rachel BV Cooper[1], Donald R Oliver[1], Ki B Kim[1], Alexander Lin[2], Rolf G Behrents[1], Adriana M Montano[3]
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Neuroblastoma Amplified Sequence (NBAS) mutation in Recurrent Acute Liver Failure: confirmatory report in a sibship with very early onset, osteoporosis and developmental delay

Jose M Capo-chichi[1], Cybel Mehawej[2], Valerie Delague[3,4], Catherine Caillaud[5], Issam Khneisser[2], Fadi F Hamdan[1], Jacques L Michaud[1,6], Zoha Kibar[1], Andre Megabarne[2,7]
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A novel *TTN* mutation causing Tibial Muscular Dystrophy in a Turkish patient

Evren Gumus, Huseyin Aslan, Oguz Cilingir, Muhsin Ozdemir, Beyhan Durak Aras, Halime Onur Kucuk, Sevilhan Artan
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Tue(3)-P-145

Analysis of mitochondria-related gene from clinically suspected Charcot-Marie-Tooth patients by using whole exome sequencing

Yu Hiramatsu, Yuji Okamoto, Akiko Yoshimura, Junhui Yuan, Yujiro Higuchi, Akihiro Hashiguchi, Hiroshi Takashima
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Mulibrey Nanism in an Omani girl with Primary ovarian failure

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Development

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Genetic analysis of 30 families with Joubert syndrome and related disorders

Toshifumi Suzuki[1,2], Noriko Miyake[1], Mitsuko Nakashima[1], Hirotomo Saitsu[1], Satoru Takeda[2], Naomichi Matsumoto[1]
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The first Japanese patients with genetically definite Bardet-Biedl syndrome

Makito Hirano[1,2], Wataru Satake[3], Kenji Ihara[4], Ikuya Tsuge[5], Yutaka Suzuki[6], Yusaku Nakamura[1], Susumu Kusunoki[2], Tatsushi Toda[3]
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Placental epigenome vary in relation to inadequate gestational weight gain

Tomoko Kawai[1], Takahiro Yamada[4], Kosei Abe[1], Kohji Okamura[2], Hiromi Kamura[1], Rina Akaishi[4], Hisanori Minakami[4], Kazuhiko Nakabayashi[3], Kenichiro Hata[1]
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Differentiation of iPS cells into cranial neural crest cells to model congenital disorder that arises from defects in the development of the embryonic cranial neural crest cell lineage

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Nutrigenomic aspects of adaptive responses to maternal high-sucrose feeding in rat models of metabolic syndrome

Lucie Sedova[1], Elena Skolnikova[1], Frantisek Liska[1], Ludmila Kazdova[2], Drahomira Krenova[1], Vladimir Kren[1], Pavel Hamet[3], Johanne Tremblay[3], Ondrej Seda[1]
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Assessment of EGF gene and EGF-R expression following verification of 2-cell and blastocysts mouse embryos

Saima Abbaspour, Parvaneh Keshavarz, Mojhgan Eskandari, Alireza Sharafshah Rostami
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Lgr4 plays as an anti-testis gene of fetal gonads in mice

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VA10, an immortalized broncho-epithelial cell line, as a tool for *in vitro* lung developmental studies

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A Boolean network model of normal gonadal sex determination in human and related disorders of sex development

Leda Torres[1], Osiris Rios[1,2], Alfredo Rodriguez[1,4], Susana Kofman[5], Horacio Merchant[3], Luis Mendoza[3,6], Sara Frias[1,3]
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Identification and characterization of non-coding RNAs associated with chromatin in pluripotency

Alessandro Bonetti, K Kashi, Kosuke Hashimoto, Alexandre Fort, Piero Carninci
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Evolutionary and Population Genetics

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Fingerprint Pattern and Blood Groups in Twins - A Genetic Perspective

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Oral Health Related Habits and Oral Hygiene Practices among Identical and Non-Identical-A Genetic Perspective

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Genetic Analysis Provides Evidence for Increased Disease Prevalence of Systemic Lupus Erythematosus in Chinese Populations compared to Europeans

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First Report of Hemophilia-A Point Mutation Detection in Egypt: AMean for Providing Proper Genetic and Prenatal Counseling

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Tue(3)-P-161

Signatures of geographically restricted adaptation in the Sea Island Gullah African Americans

Paula S Ramos[1], Satria Sajuthi[2], Jasmin Jasmin Divers[2], Uma Nayak[3], Wei-Min Chen[3], Kelly J Hunt[1], Diane L Kamen[1], Gary S Gilkeson[1], Jyotika K Fernandes[1], Ida J Spruill[1], W T Garvey[4], Michele M Sale[3], Carl D Langefeld[2]
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Molecular Characterization of G6PD Deficient Variants in Karen and Lao populations in Thailand

Chalisa L. Cheepsunthorn[1], Kanjanawadee Prasittisa[2], Arparkorn Kanchanavithayakul[2], Petcharat Kittiwatanasarn[3], Issarang Nuchprayoon[4]
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Allelic imbalance of mRNA transcription in α2-HS glycoprotein (fetuin-A) gene

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Mitochondrial DNA variation at the Sindh population of Pakistan

Shahzad Bhatti[1], Muhammad Aslamkhan[1], Sana Abbas[1], Marcella Attimonelli[2], Hakan Aydin[3]
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Detection of population specific signals of positive election in Mongolians

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Adaptive patterns of genetic diversity in native Siberian populations

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Analysis of polymorphisms associated with skin pigmentation in Oceanic populations

Izumi Naka[1], Nao Nishida[2], Ryosuke Kimura[3], Kyoko Yamaguchi[4], Takuro Furusawa[5], Taro Yamauchi[6], Kazumi Natsuhara[7], Yuji Ataka[8], Takafumi Ishida[1], Tsukasa Inaoka[9], Yasuhiro Matsumura[10], Ryutaro Ohtsuka[11], Jun Ohashi[1]
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GENETIC TESTING OF HERODOTUS' THEORY ON THE ORIGIN OF ARMENIANS

Anahit Hovhannisyan, Ashot Margaryan, Hrant Hovhannisyan, Zaruhi Khachatryan, Armine Khudoyan, Levon Yepiskoposyan
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Colour-blindness: Impact of Consanguinity and Environment

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***BCL11A* erythroid-specific enhancer and fetal hemoglobin levels among Sickle Cell Disease Patients in Cameroon: Implications for future therapeutic interventions**

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Tue(3)-P-171

Population genetics analysis of Negrito groups in Southeast Asia

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Geographical and Cultural Influences on Genetic Diversity: Patterns of the Y-Chromosomal Variation in Populations with Patronymic Tradition

Maxat Zhabagin[1,2], Yuldash Yusupov[3], Zhaxylyk Sabitov[6], Roza Shalyakho[2,4], Anastasiya Agdzhoyan[2,4], Zhaxybay Zhumadilov[5], Elena Balanovska[2,4], Oleg Balanovsky[4]
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Prognostic role of Interleukin-1 α and β gene polymorphisms in preterm birth

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An update to the distribution of allele frequencies and forensic parameters for 15 autosomal STRs in the Mestizo population from Península of Yucatán, Mexico

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Whole genome sequence analysis of fetal hemoglobin in a sickle cell disease cohort

Evadnie Rampersaud, Guolian Kang, Yasui Yutaka, Jiao Yunnian, Wang Shuoguo, Palmer Lance, Feng Ruopeng, Estepp Jeremie, Zhang Jinghui, Hankins Jane, Wu Gang, Weiss J Mitchell
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Molecular-genetic evaluation of G2956A (rs112287730) alteration of FBN1 gene in Russian Marfan patients

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Mutational complexity of a classic founder disease: tyrosinaemia in Quebec

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AIMs and Ascertainment Bias in Genomic Datasets: Considerations for Personalized Medicine

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Where is Brazil located at? A study on 100 Alu insertion polymorphisms

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A frequent mutation in the *DYSF* gene in the Avar's population from northern Caucasus

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Research Centre of Medical Genetics, Russia

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Y chromosome haplogrouping using the next generation sequencing system

Eriko Ochiai, Keiko Miyashita, Kiyoshi Minaguchi, Yu Kakimoto, Fumiko Satoh, Motoki Osawa
Department of Forensic Medicine, Tokai University School of Medicine, Japan

Tue(3)-P-183

The genetic landscape of Dagestan from Y-chromosome markers: phylogeny and phylogeography of J1 haplogroup and territorial subdivision of native populations

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Genomic relationships using DNA fractals rather than sequence alignments

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Whole-genome sequencing of the Tibetan population

Lynn B Jorde[1], Hao Hu[2], Tatum Simonson[3], Jonathan Downie[1], Gustavo Glusman[4], Jared Roach[4], Josef Prchal[1], Peter Robbins[5], Gianpiero Cavalleri[6], Alan Rogers[7], Ryan Bohlender[7], Felipe Lorenzo[1], Donald McClain[1], Chad Huff[2]
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Population Structure, Divergence and Admixture of Han Chinese, Japanese and Korean Populations

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MITOCHONDRIAL GENETIC DIVERSITY OF THE PRE-HISPANIC MEXICAN MAYA POPULATIONS FROM PALENQUE AND EL REY

Mirna Isabel I.O. Ochoa Lugo[1], Gerardo G.P. Pérez Ramírez[1], Javiera J.C. Cervini-Silva[2], Miguel M.M. Moreno Galeana[1], Eduardo E.R. Ramos[3], Arturo A.R. Romano-Pacheco[4,5], María de Lourdes d.L. M. Muñoz[1,6]
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Molecular Basis of Mendelian Disorders 1

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Japanese male twins with Leber congenital amaurosis possibly caused by the *GUCY2D* gene mutation

Katsuhiro Hosono[1], Shinsei Minoshima[2], Yuko Harada[1], Kentaro Kurata[1], Akiko Hikoya[1], Miho Sato[1], Yoshihiro Hotta[1]
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Low-prevalence somatic *TSC2* mutations in sporadic lymphangioleiomyomatosis identified by deep-sequencing

Atsushi Fujita[1], Katsutoshi Ando[2], Etsuko Kobayashi[2], Keiko Mitani[3], Koji Okudera[4], Mitsuko Nakashima[1], Satoko Miyatake[1], Yoshinori Tsurusaki[1], Hirotomo Saitsu[1], Kuniaki Seyama[2], Noriko Miyake[1], Naomichi Matsumoto[1]
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First report of compound heterozygous mutations in the *TRAPPC9* gene showing clinical features of autism and intellectual disability in a Thai family

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Novel *GDAP1* mutations in Japanese patients with Charcot-Marie-Tooth disease

Akiko Yoshimura[1], Junhui Yuan[1], Yujiro Higuchi[1], Akihiro Hashiguchi[1], Yuji Okamoto[1], Shoji Tsuji[2], Hiroshi Takashima[1]
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Whole Exome Sequencing revealed a nonsense mutation in STAB2 Gene associated with intellectual disability and oligodontia

Huoru Zhang[1], Yong-Fei Wang[1], Houyan Xia[3], Tony MF Tong[2], Ho Ming Luk[2], Jing Yang[1], Yu Lung Lau[1], Wanling Yang[1]
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Whole Exome Sequencing helps characterize the mysterious skeletal disorder of a village in Jammu and Kashmir, India

Swarkar Sharma[1], Ekta Rai[1], Ankit Mahajan[2], Parvinder Kumar[3], Arshia Angural[1], Manoj K Dhar[2], Sushil Razdan[4], Kumarasamy Thangaraj[5], Carol Wise[6], Shiro Ikegawa[7], Kamal K Pandita[8]
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Exome sequencing identifies pathogenic mutations in the patient with severe combined immunodeficiency

Hui-Wen Yu, Cheng-Yu Liao, Chi-Chang Shieh, Peng-Chieh Chen
National Cheng Kung University, Institute of Clinical Medicine, College of Medicine, Taiwan

Tue(3)-P-196

Novel zebrafish models of neuromuscular diseases

Genri Kawahara, Yukiko K Hayashi
Department of Pathophysiology, Tokyo Medical School, Japan

Tue(3)-P-197

Identification of novel mutations and molecular modelling of novel missense mutations in Pakistani patients of congenital afibrinogenemia

Tehmina Nafees Sonia Khan[1], Arshi Naz[1], Arijit Biswas[2], Tahir Sultan Shamsi[1]
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Tue(3)-P-198

Refinement of an Autosomal Dominant Hereditary Gingival Fibromatosis Locus on chromosome 2p23

Miao Sun[1], Bin Wei[1], Zili Ge[1], Yingying Zhang[1], Xingshun Xu[2], Xue Zhang[3]
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Tue(3)-P-199

Rare beta-thalassaemia mutation detected in South East Asian population - A dilemma in calling carrier status

Hai-Yang Law[1,2], Guek-Peng Tan[2], Angeline HM Lai[1], Ivy ISL Ng[1,2]
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Tue(3)-P-200

Novel hearing loss-causative point mutations and copy number variation identified by exon sequencing

Aki Sakata[1,2], Akinori Kashio[2], Shotaro Karino[2], Yu Matsumoto[2], Akinobu Kakigi[2], Tatsuya Yamasoba[2], Hiroki Ueda[1], Shogo Yamamoto[1], Kenji Tatsuno[1], Hiroyuki Aburatani[1]
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Tue(3)-P-201

Novel mutation in WWOX gene is associated with Intellectual disablity in a consanguineous Arab family

Asem Alkhateeb[1,2], Samah Aburahma[2], Wesal Habbab[1], Richard Thompson[1]
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Tue(3)-P-202

Utility of a multigene next-generation sequencing panel for molecular diagnosis of Noonan syndrome and other RASopathies

Maggie Brett[1], Eileen Lim[1], Siew Peng Lee[1], Angeline Lai[2], Ee Shien Tan[2], Saumya Jamuar[2], Ivy Ng[2], Breana Cham[2], Jiin Ying Lim[2], Ene Choo Tan[1]
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Whole-exome sequencing identifies a novel mutation in primary ciliary dyskinesia from a Chinese consanguinity family

Hong Luo, Ting Guo
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Tue(3)-P-204

Identification and Functional Analysis for Novel Gene Mutation Responsible for Autosomal Dominant Macular Dystrophy involved Dysfunction of ON-type Bipolar Cells

Yuichi Kawamura[1,2], Takuro Fujimaki[2], Kazutoshi Yoshitake[1,3], Kazushige Tsunoda[1], Yukihiro Baba[4], Akiko Suga[1], Hiroshi Kuribayashi[4], Sumiko Watanabe[4], Kazuho Ikeo[5], Akira Murakami[2], Takeshi Iwata[1]
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Tue(3)-P-205

Knockout the ceramide kinase-like gene causes retinal degeneration in zebrafish

Mugen Liu, Shanshan Yu, Zhaohui Tang, Fei Liu, Jiaxiang Chen
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Tue(3)-P-206

Molecular genetics analysis of Von Willebrand disease type III: studies in Cohort Pakistani patients

Shariq Ahmed[1], Arshi Naz[1], Hamideh Yadegari[2], Julia Driesen[2], Johannes Oldenburg[2], Nisar Ahmed[3], Shehla Tariq[6], Samina Amanat[4], Fazale Raziq[5], Muhammad Nadeem[1], Tahir Sultan Shamsi[1]
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Tue(3)-P-207

A loss-of-function mutation in *JAK1* is associated with epidermodysplasia verruciformis

Rongrong Wang[1], Jiawei Liu[2], Lili Zhang[1], Donglai Ma[2], Xue Zhang[1]
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Tue(3)-P-208

Molecular genetic study of 12 Pakistani families with autosomal recessive sensorineural hearing loss

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Tue(3)-P-209

Gene-based association analysis of familial pulmonary arterial hypertension

Koichiro Higasa[1], Aiko Ogawa[2], Chikashi Terao[1], Masakazu Shimizu[1], Shinji Kosugi[3], Ryo Yamada[1], Hiroshi Date[4], Hiromi Matsubara[2], Fumihiko Matsuda[1]
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Tue(3)-P-210

A *de novo* mutation of the *MYH7* gene in a large Chinese family with autosomal dominant myopathy

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Tue(3)-P-211

Cystic Fibrosis in Chinese: Frequent and Novel Mutations in *CFTR*

Yaping Liu[1], Xinlun Tian[2], Jun Yang[1], Han Wang[1], Tao Liu[2], Wenbin Xu[2], Kai-Feng Xu[2], Xue Zhang[1]
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Tue(3)-P-212

CHCHD2 is novel gene for autosomal dominant Parkinson's disease

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Tue(3)-P-213

Mutations in the patients with Nakajo Nishimura Syndrome - like autoinflammatory diseases

Akira Kinoshita[1,2], Nobuo Kanazawa[3], Noriko Kinjo[4], Hiroyuki Mishima[1], Ko-Ichiro Yoshiura[1]
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Tue(3)-P-214

Expanding the clinical phenotype of the novel connective tissue disorder due to variants in the *PLOD3* gene

Lisa J Ewans[1,2], Alison Colley[3], Mark J Cowley[1,2], Ying Zhu[4], Velimir Gayevskiy[2], Kevin Ying[2], Corrina Walsh[5], Eric Lee[5], Edwin Kirk[5,6], Michael Field[4], David Miller[2], Paula Morris[2], Michael Buckley[5], Marcel Dinger[1,2], Tony Roscioli[1,2,7]
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Genomics in the genetic clinic: An Australian perspective

Lisa J Ewans[1,2], Mark J Cowley[1,2], Ying Zhu[3], Velimir Gayevskiy[2], Kevin Ying[2], Corrina Walsh[4], Eric Lee[4], Edwin Kirk[4,5,6], Alison Colley[7], Anne Turner[5,6], David Mowat[5,6], Lisa Worgan[7], Mary-Louise Freckmann[5,6], Rani Sachdev[5,6], Michael Field[3], David Miller[2], Paula Morris[2], Michael Buckley[4], Marcel E Dinger[1,2], Tony Roscioli[1,2,5]
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Tue(3)-P-216

Clinical application of next generation sequencing in a family with undiagnosed genetic conditions

Erina Ozaki[1,2], Minenori Eguchi-Ishimae[3], Yuko Tezuka[3,4], Keiro Kagata[4], Takuya Naruto[5], Issei Imoto[5], Mariko Eguchi[2,3], Eiichi Ishi[2,3]
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Tue(3)-P-217

Mutational analysis of Usher syndrome in Taiwan

Liang-Hsuan Hu Hu, Chia-Ying Chien, Tzu-Yen Hsu, Wun-Ying Lin, Shun-Ping Huang, Yung-Hao Ching
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Tue(3)-P-218

Haplotype analysis combined with whole exome sequencing for the identification of the causative mutation of a X-linked Retinitis Pigmentosa family

Yung-Hao Ching[1], Lian-Hsuan Hu[1], Chi-Jia Huang[1], Chi-hsuan Chung[1], Wun-Ying Lin[1], Jia-Ling Jiang[2], Shun-Ping Huang[1]
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Tue(3)-P-219

Genomics & Social Justice - Diagnosing Cystic Fibrosis in South Africa

Cheryl S Stewart[1], Jeanne van Rensburg[1], Refiloe Masekela[2], Marco Zampoli[3], Tamara Kerbelker[3], Robin J Green[2], Michael S Pepper[1]
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Tue(3)-P-220

Therapeutic research in a mouse model of cardio-facio-cutaneous syndrome

Daiju Oba[1], Shin-ichi Inoue[1], Mitsuji Moriya[1,2], Yusuke Watanabe[3], Tetsuya Niihori[1], Sachiko Miyagawa-Tomita[4], Shigeo Kure[2], Toshihiko Ogura[3], Yoichi Matsubara[1,5], Yoko Aoki[1]
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Tue(3)-P-221

Expression profile of inflammatory genes in placenta from sickle cell disease patients

Monica B Melo[1], Leticia C Baptista[1], Regiane Ferreira[2], Fernanda GC Surita[3], Dulcineia M Albuquerque[2], Mary A Parpinelli[3], Kleber Y Fertrin[2], Carolina Lanaro[2], Fernando F Costa[2], Maria Laura Costa[3]
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Tue(3)-P-222

A novel locus for autosomal dominant high hyperopia mapped to chromosomal 11

Xueshan Xiao, Shiqiang Li, Xiaoyun Jia, Xiangming Guo, Qingjiong Zhang
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Novel gene discovery across a large cohort of patients with syndromic craniofacial anomalies

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Tue(3)-P-224

Next Generation Sequencing in Spinal muscular atrophy syndromes: involvement beyond the anterior horn cell

Tony Roscioli[1,2,3], Hooi LIng Teoh[4,5], Ying Zhu[6], Hugo Sampaio[4,5], David Mowat[3,5], MIchael F Buckley[7], Michelle Farrar[4,5]
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Tue(3)-P-225

The mechanism study of proximal symphalangism induced by p.Leu373Arg variant in the GDF5 proregion

Yang Luo, Xinxin Zhang, Xuesha Xing, Lihua Cao, Shusen Wang, Xue Zhang
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Tue(3)-P-226

Mutations spectrum of *COL1A1/COL1A2* in Chinese with osteogenesis imperfecta

Xiuli Zhao[1], Jifang Xiao[1], Yiyang Wu[1], Jingsong Gao[2], Xiuzhi Ren[3], Chaoxia Lu[1], Han Wang[1], Yue Sun[2], Xue Zhang[1]
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Genetic testing of inherited cardiomyopathy by next generation semiconductor sequencing technologies

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Tue(3)-P-228

WHOLE EXOME SEQUENCING IDENTIFIED A NOVEL MISSENSE CHRNG MUTATION IN A TURKISH CHILD WITH AN ATYPICAL PRESENTATION OF ESCOBAR SYNDROME

Ayca Aykut[1], Fillippo Beleggia[2], Huseyin Onay[1], Emin Karaca[1], Bernd Wollnik[2], Ferda Ozkinay[1]
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Tue(3)-P-229

ISOLATED OPTIC NERVE HYPOPLASIA IN 5 FAMILY TRIOS - A CLINICAL AND EXOME STUDY

Pierre Bitoun[1,3], Anne Boland-Auge[2], Delphine Bacq-Daian[2], Eva Pipiras[3], Brigitte Benzacken[3], Austin Alexander[4], Suzanne Kuzbari[5], Jean-Francois Deleuze[2]
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Tue(3)-P-230

New mutations in *PLOD1* and *COL3A1* in two cases with Ehlers-Danlos syndrome

Hakan Ulucan[1], Ugur Gumus[1], Emre Kirat[1], Alper Gezdirici[2], Asuman Koparir[1], Adnan Yuksel[3], Bert Callewaert[4], Anne De Paepe[4], Mehmet Seven[1]
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Tue(3)-P-231

Decreased performance in IDUA knockout mouse mimic limitations of joint function and locomotion in patients with Hurler syndrome

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Epigenetics

Tue(3)-P-232

Aberrant methylation at imprinted DMRs is associated with placental mesenchymal dysplasia

Saori Aoki[1,4], Ken Higashimoto[1], Hidenori Hidaka[1], Hidetaka Watanabe[1], Yasufumi Ohtsuka[2], Hiroyuki Mishima[3], Koh-ichiro Yoshiura[3], Hitomi Yatsuki[1], Kenichi Nishioka[1], Keiichiro Joh[1], Takashi Ohba[4], Hidetaka Katabuchi[4], Hidenobu Soejima[1]
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H2A.Z genetically interacts with DRG2 which physically associates with RWDD1 and Nup107

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Tue(3)-P-234

MicroRNA promotes the decidualization of eutopic and ectopic endometrium

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Tue(3)-P-235

Epigenetic regulator, Uhrf1, is a positive regulator in chondrocyte differentiation

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Tue(3)-P-236

Altered levels of epigenetic marks/factors on regulatory regions of sperm chromatin condensing genes in testicular biopsies infertile men

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Is the association between sweet and bitter perception due to genetics?

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Clinical and molecular findings in a patient with 46,XX/47,XX,+14 mosaicism caused by postzygotic duplication of a paternally derived chromosome 14

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Assessment of the oral health status of monozygotic and dizygotic twins - a comparative study

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Association of epigenetic role of *BRDT* in spermatogenesis and male infertility

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Differential histone modification of marker genes involved in stemness and differentiation in human pluripotent and differentiated cells

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Discovering site-specific changes in 5-hydroxymethylcytosine in suicide completers through next-generation sequencing

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DNA Methylation Reflects Early Life Chronic Stress Environment: A Biomarker for Childhood Cortisol

Evan Gatev[1,2], Mina Park[2], Rachel Edgar[2], Lisa McEwen[2], Julia MacIsaac[2], Sarah Goodman[2], Nicole Bush[5], W. Thomas Boyce[5], Michael Kobor[1,2,3,4]
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Epigenetic role of the nuclear factor NF-Y on *ID* family genes in endometrial tissues of women with endometriosis

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Monozygotic twins concordant for ICR2 hypomethylation in different tissues but discordant for Beckwith-Wiedemann syndrome phenotype

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Exploration of hydroxymethylation in Kagami-Ogata syndrome caused by hypermethylation of imprinting control regions

Kazuki Yamazawa[1,2], Keiko Matsubara[2], Masayo Kagami[2], Kazuhiko Nakabayashi[3], Kenichiro Hata[3], Maki Fukami[2], Tsutomu Ogata[2,4]
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ANALYSIS OF CHROMOSOMAL ABNORMALITIES AND DNA METHYLATION AT *SNRPN* GENE IN PRADER WILLI SYNDROME - COIMBATORE POPULATION

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Meta-Analysis of Epigenome-wide Association Studies on Serum Urate Levels including 7600 individuals

Christian Gieger[1,2], Annika Laser[1,2], Benjamin C Lehne[3], Stefan Gustafsson[4], Tao Zhang[5], Sonja Kunze[1,2], Dianjianyi Sun[5], Shengxu Li[5], Gerald Berenson[5], Fabian Theis[6], Annette Peters[2], Gabi Kastenmueller[7], Erik Ingelsson[4], Wei Chen[5], Lars Lind[4], John Chambers[3], Melanie Waldenberger[1,2]
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Effect of butylated hydroxytoluene (BHT) on BDNF gene methylation, learning and memory in male wistar rats

Parvaneh Keshavarz, Mahsan Maleki, Parvin Babaei, Alireza Sharafshah Rostami, Ali Albonaim, Vahid Omarmeli
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Multilocus methylation defects in a patient presenting with both clinical phenotype of pseudohypoparathyroidism type Ib and Beckwith-Wiedemann syndrome

Shinichiro Sano[1], Keiko Matsubara[1], Keisuke Nagasaki[3], Akie Nakamura[1], Kazuhiro Nakabayashi[2], Kenichiro Hata[2], Maki Fukami[1], Tsutomu Ogata[4], Masayo Kagami[1]
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The Transcriptome and DNA Methylome Landscapes of Human Primordial Germ Cells

Fan Guo, Liying Yan, Hongshan Guo, Lin Li, Fuchou Tang, Jie Qiao
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Investigation of maternal effects, maternal-fetal interactions and parent-of-origin effects (imprinting), using mothers and their offspring with schizophrenia

Byung Dae Lee
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Tue(3)-P-253

Telomere Length and Epigenetics, TERRA Transcript Level and Telomerase Expression as Dynamic Genetic Parameters in Poly Cystic Ovary Syndrome

Narges Ghobadi[1], Maryam Shahhoseini[1], Poopak EftekhariYazdi[2], Raha Favaedi[1], Fatemeh Hassani[2], Bahar Movaghar[2], Leyli Karimian[2]
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LRRC37A2 and *SNORD42B* methylation analysis in gastric cancer tissues using next-generation sequencing

Fernanda Wisnieski[1], Leonardo Caires Santos[1], Mariana Ferreira Leal[1], Jaqueline Geraldis Cruz[1], Ana Carolina Anauate Pereira[1], Danielle Queiroz Calcagno[2], Carolina Oliveira Gigek[1], Elizabeth Suchi Chen[1], Samia Demachki[2], Ricardo Artigiani[3], Paulo Pimentel Assumpção[2], Laércio Gomes Lourenço[4], Rommel Rodríguez Burbano[5], Marília Cardoso Smith[1]
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A fluorescence polarization-based method with methyl-sensitive one-label extension for quantification of single CpG dinucleotide methylation

Cunyou Zhao, Shufen Li, Zhongju Wang, Lin Zhou, Fu Luo
Department of Medical Genetics, Southern Medical University, China

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Comprehensive methylation microarray analysis for placental genomic DNA in abruption cases

Jun Konno, Akizawa Yoshika, Ogawa Masaki, Matsui Hideo
Obstetrics and Gynecology, Tokyo Women's Medical University, Japan

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Maternal undernutrition alters DNA methylation profiles in rat embryonic kidney

Mariko Hida
Neonatology, Yokohama Rosai Hospital, Japan

Tue(3)-P-258

The epigenetic impact of a 6 month lifestyle intervention programme on women aged 18-40

Michelle C Thunders, Victoria Chinn, Rachel Page
College of Health, Massey University, New Zealand

Tue(3)-P-259

Skewed pattern of X chromosome inactivation in Brazilian women without familial history of X-linked intellectual disability

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Functional analysis of *Xist* long noncoding RNA using mouse artificial chromosome (MAC)

Daigo Inaoka[1], Naohiro Sunamura[1], Miki Kataoka[1], Yuji Nakayama[2], Mitsuo Oshimura[3], Hiroyuki Kugoh[1,3]
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Health Services Research

Tue(3)-P-261

Breast cancer, genetics or bad karma: Meanings and experiences of Thai women living with breast cancer in southern Thailand

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Breast Cancer and Genetical Belief: Barriers to Screening Programmes amongst Thai Migrant Women in Australia

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Tue(3)-P-263

The relationship between the social competence of children and adults with Down's syndrome and caregivers' burden

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Tue(3)-P-264

CD44, ALDHI, E-cadherin and Snail gene protein analysis in ameloblastoma

Chong Huat Siar[1], Zainal Ariff Bin Abdul Rahman[1], Nurharnani Binti Harun[2], Hidetsugu Tsujigiwa[3], Hitoshi Nagatsuka[4], Kok Han Ng[5]
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Community Genetics in Cuba

Beatriz Marcheco-Teruel
National Center of Medical Genetics, Cuba

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Using Quality Improvement Methods and Time-Driven Activity Based Costing to Improve Value-Based Cancer Care Delivery at a Cancer Genetics Clinic

Ryan Tan[1], Marie Met-Domestici[1], Ke Zhou[2], Alexis B Guzman[3], Soon Thye Lim[2], Khee Chee Soo[2], Thomas W Feeley[3,4], Joanne Ngeow[1,2]
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Cost is a barrier to accept germline mutation testing for known cancer syndrome in Japan

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Fulfilling the promise of personalised medicine - prioritising our investment

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The social and economic impacts of childhood syndromes of suspected genetic origin

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Cost effectiveness of whole exome sequencing compared with standard diagnostic care

Zornitza Stark[1], Deborah Schofield[1,2,3], Khurshid Alam[1], William Wilson[4], Nessie Mupfeki[1,6], Ivan Macciocca[1], Rupendra Shrestha[2], Susan M White[1,5], Clara Gaff[1,5,6], Melbourne Genomcis Health Alliance
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How do people feel on knowing their disease risks by genetic testing? -Attitudinal Study for 4,000 Japanese Respondents: Behavior Change after knowing their disease Risks and Impact of Communication Ways of Disease Risk -

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A prospective evaluation of whole exome sequencing as a first-tier molecular test in infants with suspected monogenic disorders

Susan M White[1,2], Zornitza Stark[1], Tiong Y Tan[1,2], Belinda Chong[1], Gemma Brett[1,5], Patrick Yap[1], Maie Walsh[1], Alison Yeung[1], Shannon Cowie[1], George McGillivray[1], Heidi Peters[1,2,4], Paul G Ekert[1,2], Christiane Theda[1,2,3], Ivan Macciocca[1], Katrina Bell[1], Alicia Oshlack[1,2], Simon Sadedin[2], Peter Georgeson[2], Charlotte Anderson[2], Natalie Thorne[1,2,5], Clara Gaff[2,5], Melbourne Genomics Health Alliance
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Resolving barriers to the use of genomic sequencing in clinical practice: evaluation of a whole-of-system approach

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Initiatives on Rare and Undiagnosed Diseases (IRUD) for adults: a national network deciphering rare and undiagnosed diseases

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Family history taking in pediatrics: it's much more than just a checklist

June C Carroll[1], Laure Tessier[2,3], Jamie C Brehaut[2], Beth K Potter[2], Pranesh Chakraborty[3], Brenda J Wilson[2], CIHR Emerging Team in Genomics in Screening
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APPROACH TO PATIENTS WITH GENETIC DISEASES IN EMERGENCY SERVİCE

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INVESTİGATION TO PATIENTS WITH GENETIC DISEASES IN EMERGENCY SERVİCE

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Patients and their Families and Friends as Developers of Medical Treatments/Devices

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Gender Differences in Genetic Contribution to Longevity

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Mainstreaming genomics - A theory-informed systematic review of clinicians' genetic testing practices

Jean L Paul[1], Hanna Leslie[2], Alison H Trainer[3,4], Clara L Gaff[4,5,6]
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MÉXICO´S NATIONAL BIOBANKING SERVICE LABORATORY

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Genetic Counseling

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The free software "f-tree" for drawing a pedigree in genetic counseling

Koji Kumagai, Masahiro Sakai, Takayoshi Maeda
Department of Gynecology, Osaka Railway Hospital, Japan

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Factors affecting the decision to undertake non-invasive prenatal testing

Masahiro Murakami, Kaori Mori, Akane Kondo, Tsuyako Iwai, Kazuhisa Maeda
Clinical Genetics, Shikoku Medical Center for Children and Adults, Japan

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The examination about wish of prenatal testing and mental background factorof pregnant women by assited reproductive technology

Miwa Sakamoto, Nahoko Shirato, Tatsuko Hirose, Keiko Miyagami, Akihiko Sekizawa
Obstetrics and Gynecology, Showa University, Japan

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Effect of the mental background factor of after childbirth women who done prenatal testings

Nahoko Shirato, Miwa Sakamoto, Keiko Miyagami, Junko Yotsumoto, Atshko Saito, Tatuko Hirose, Mikiko Izumi, Ryu Matsuoka, Kiyotake Ichizuka, Akihiko Sekizawa
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Genetic counseling in pregnant women whose fetus had Robertson translocation

Tatsuko Hirose, Keiko Miyagami, Nahoko Shirato, Mikiko Izumi, Shoko Hamada, Keiko Koide, Tetsuro Kondo, Junko Yotsumoto, Ryu Matsuoka, Kiyotake Ichizuka, Akihiko Sekizawa
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Role of Genetic Counseling in Pediatric Transplantation of Genetic Disorders: A Report from Children's Medical Center in Japan

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Uptake of gene test among family members with *BRCA 1/2* mutation in Japanese population Uptake of gene test among family members with *BRCA 1/2* mutation in Japanese population

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Charcot-Marie-Tooth disease (CMT) Patient Registry in Japan

Masanori Nakagawa[1], Kensuke Shiga[3], Yu-ichi Noto[2], Yukiko Tsuji[2], Toshiki Mizuno[2], The research group of clinical evidence to improve Charcot-Marie-Tooth Disease patient care
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Current Status of Social Issues for People with Down Syndrome in Japan: From Nationwide Survey

Hidehiko Miyake[1], Shigehito Yamada[1,2], Yosuke Fujii[3], Mariko Taniguchi-Ikeda[4], Mari Urano[5], Yuka Ozasa[6], Makoto Kanai[7], Akimune Fukushima[8], Yoichi Matsubara[9], Kayoko Saito[5], Ikuo Konishi[2]
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Genetic counseling for clinical sequencing using the next-generation sequencincer panel analysis

Tetsuya Okazaki[1,2], Megumi Murata[3], Masachika Kai[4], Kaori Adachi[3], Naoko Nakagawa[5], Noriko Kasagi[6], Wataru Matsumura[1,2], Yoshihiro Maegaki[1], Eiji Nanba[2,3,5]
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Characteristics of the Genetic Counseling in Kyoto University Hospital to Figure Out the Genetic Counseling Needs in Japan

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A case of osteogenesis imperfecta (OI) diagnosed during pregnancy whose mother's feeling for fetus changed from denial to acceptance and whose genetic diagnosis was planned after genetic counseling

Masahiro Shiba, Yasuhiro Matsumoto, Minako Shimizu, Takako Higa, Shigenari Namai, Hideo Kamata, Koichi Umezawa, Akinori Taguchi, Yukifumi Sasamori, Koichiro Kido, Eiji Ryo, Takuya Ayabe
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Genetic counsering of 46,XY DSD for eight years -a case report-

Nobuko Nishioka, Tomohito Ishiguro, Shihori Nishizawa, Atsuko Yamada
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Mental background of pregnant women in the view point of delivering facilties

Keiko Miyagami[1], Nahoko Shirato[1], Miwa Sakamoto[1], Junko Yotsumto[1], Atsuko Saito[1], Tatsuko Hirose[1], Mikiko Izumi[1], Taro Morimoto[2], Shuichi Kitamura[3], Ryu Matsuoka[1], Akihiko Sekizawa[1]
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Genetic diagnosis, counseling and management of androgen insensitivity syndrome : a case report

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Genetic Counseling for Patients and Family Members with Endocrine Disease: Experience of Specialized Genetic Counselor

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Families who were suspected to be HBOC families but didn't show pathogenic mutations in both *BRCA1* and *BRCA2* in genetic testing

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Factors influencing the decision not to choose prenatal aneuploidy screening in pregnant women receiving genetic counseling for advanced maternal ages

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Genetic counseling of a woman with malignant pheochromocytoma caused by multiple endocrine neoplasia type 2A

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The positive test result`s effects on condition of health, ability to function and mental health as evaluated by Finnish male BRCA1/2 mutation carriers

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Genetic counseling to couples having noninvasive prenatal genetic testing

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Genetic Counseling for Hereditary Breast and Ovarian Cancer Syndrome in our hospital

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Eleven-year summary of genetic counseling in Kyoto Prefectural University of Medicine

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Genetics/Genomics Education

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Genetic Counselling in Practice: an international course for clinical geneticists and genetic counsellors

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A proposal for clinical genetics (genetics in medicine) education for medical technologists and other health professionals in Japan

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Problems and their solutions in genetic counseling education in Japan

Hidetsugu Kohzaki
Yamato Univ./Kyoto Univ., Japan

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Exploring education models of genomic medicine for general publics in informal learning settings

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Develop multimedia genetic instruction according to the cognitive theory of multimedia learning and cognitive load theory

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Genetic Counseling Education at Kindai University

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Knowledge and attitudes of Gastroenterology fellows working in various hospitals of United States of America, on genetic testing for disease specific biomarkers and knowledge of Precision Medicine

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Tue(3)-P-312

Analysis of the education guidelines and textbooks to investigate the feasibility of educating human genetic in primary and secondary education system in Japan

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Tue(3)-P-313

The Current Landscape of the After-Sales Services of Direct-To-Consumer (DTC) Genetic Testing in Japan

Eriko Takamine[1], Hidehiko Miyake[1,2], Manami Matsukawa[1], Akira Inaba[1], Ayumi Yonei[1], Yumie Hiraoka[1], Sayaka Honda[1], Hitomi Nishio[1], Takahito Wada[1,2], Shinji Kosugi[1,2]
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Education tools to teach children about genetics, variation, and evolution

Tomoko Kobayashi[1,2], Aizawa Yayoi[2], Sugawara Michiko[5], Sakurai Yageta Mika[1], Danjoh Inaho[3], Yamaguchi Kabata Yumi[3], Kuriki Miho[4], Kuriyama Shinichi[5,6], Nagami Fuji[4], Yasuda Jun[3], Kawame Hiroshi[2], Yamamoto Masayuki[3], Suzuki Yoichi[1,2]
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The survey of recognition of medical students about recent topics related to genetic medicine

Satomi Aihara, Tomoko Yamamoto, Keisuke Tsumura, Satoshi Nisiyama, Yoshifumi Nakao, Masatoshi Yokoyama
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Developing genome science literacy at school: exploring opportunities in the Australian secondary school science curriculum

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Genetics Objective Structured Clinical Exam (Genetics OSCE) A tool for assessing and improving medical genetics communication skills and knowledge

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Innovative approaches to workforce transformation: Preparing England's National Health Service to deliver a genomic medicine service

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Developing a MANGA cartoon medium that can promote Family Health History and Human Genetics to the public

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