

CLINICAL SEQUENCING MEETING & APSHG AUTUMN SCHOOL OF CLINICAL GENETICS 2018

31 Oct to 2 Nov 2018

- Clinical Sequencing and Genomics Meeting
- Autumn School in Clinical Genetics
- Face2Gene Workshop (phenotyping analysis, deep learning and artificial intelligence to transform big data into actionable genomic insights)
- Genetic Counselling, Case Presentations, Ethics, Discussions

3 to 4 Nov 2018

- NGS Bioinformatics & Clinical Curation Workshop

Register/Enquire at:

www.aphsg.info

secretariat.meeting2018@gmail.com

Venue: National University of Singapore



Biomedical Research & Experimental
Therapeutics Society of Singapore



Genome Institute
of Singapore



Asia Pacific Society
of Human Genetics



KK Women's and
Children's Hospital

SingHealth



SECOND ANNOUNCEMENT:CLINICAL SEQUENCING MEETING & APSHG AUTUMN SCHOOL OF CLINICAL GENETICS 2018

Following the first Summer School of the Asia Pacific Society of Human Genetics in Hong Kong in 2016, the second School will be held in Singapore from **Oct 31 (Wed) to Nov 2 (Fri)** this year.

The aims of the **Clinical Sequencing Meeting and APSHG Autumn School of Clinical Genetics 2018** are to: (1) share on the applications of next-gen sequencing in clinical settings, (2) discuss current approaches, technological advances and challenges in genetic testing, (3) promote genetics and genomics education, and (4) facilitate discussions on challenges faced in the types of tests to order, diagnostic interpretation, genetic counselling and clinical care in the practice of genomic medicine. The lectures from the invited Faculty will cover latest developments in genetic testing and screening, delivery of genetic services and other practical aspects of medical genetics and genomics. Participants will also learn of the latest products, technology solutions and services from sequencing and genetic testing companies.

Target Audience:

- Medical and clinical geneticists, genetic counsellors and other medical specialists providing diagnostic, management and counselling services for patients with genetic disorders
- Laboratory professionals conducting genetic testing
- Researchers, fellows, trainees and students in this field
- Industry professionals in next-gen sequencing and genetic testing

Registration and abstract submission has opened! Participants are encouraged to register early due to limited seats. All invited to present diagnostic dilemma cases or clinical/research posters. All sessions will be held at the National University of Singapore and limited accommodation available at service apartments on campus. Please make your plans early. You may contact the Secretariat at apshg2018@gmail.com for accommodation, registration or abstract submission enquiries.

Featured International Faculty Speakers include:

Dr Leslie BIESECKER, MD FACMG

Chief & Senior Investigator, Medical Genomics & Metabolic Genetics Branch

Head, Clinical Genomics Section, NIH and President-elect, American Society of Human Genetics (ASHG) – 2019

Dr Maximilian MUENKE, MD FACMG

Chief & Senior Investigator, Medical Genetics Branch, NIH & Head of Human Development. National Human Genome Research Institute; Editor-in-Chief, American Journal of Medical Genetics

Dr John C CAREY, MD MPH FACMG

Professor and Vice Chair of Academic Affairs, Department of Pediatrics, at the University of Utah

Former Editor-in-Chief, American Journal of Medical Genetics

Dr Gail P JARVIK, MD PhD FACMG

Arno G. Motulsky Endowed Chair in Medicine, Joint Professor of Medicine and Genome Sciences, and Head of the

Division of Medical Genetics; PI of the Clinical Sequencing Exploratory Research Consortia of NHGRI and NCI

GWAS studies of multiple phenotypes and exomic analyses of Mendelian disorders

Dr Kathleen A LEPPIG, MD FACMG

Chief, Genetic Services, Washington Permanente Medical Group; Clinical Professor, Dept. of Pathology, University of

Washington & PI on the Electronic Medical Records and Genomics (eMERGE) Network

Dr Han BRUNNER, MD FACMG

Chair and Professor, Radboud University Medical Center; Head, Clinical Genetics, Maastricht UMC+

Past President of European Society of Human Genetics

Dr Christian GILLISEN, PhD

Head of Bioinformatics, Genome Diagnostics Nijmegen

Dr Lisenka Vissers, PhD

Head of Translational Genomics, Genome Diagnostics Nijmegen

Dr Mercy LAURINO, PhD

Certified Genetic Counsellor, Seattle Cancer Care Alliance, Breast and Ovarian Cancer Prevention Program

Dr Brian CHUNG, MBBS FCCMG

Department of Paediatrics and Adolescent Medicine, LKS Faculty of Medicine, The University of Hong Kong

Dr Eva Maria Cutiongco-de la Paz, MD

Vice Chancellor for Research, University of the Philippines Manila, & Executive Director, National Institutes of Health

Research Professor, National Institutes of Health & Head -Genetics, Department of Pediatrics, Philippine General Hospital

Dr Anne Chun-Hui TSAI, MD MSc FAAP FACMG

Professor, Department of Pediatrics & Genetics, Section of Clinical Genetics & Metabolism, University of Colorado School of Medicine and Children's Hospital Colorado, USA

And more expert local speakers to be announced!

Programme Highlights:

The three day meeting from **Oct 31 to Nov 2** will include:

Lectures on the impact of NGS on clinical diagnosis & genetic counselling, GWAS studies of multiple phenotypes and exomic analyses of Mendelian disorders, ACMG recommendations of reporting of variants, incidental findings, lessons from the Electronic Medical Records and Genomics (eMERGE) Network, clinical sequencing experiences in North America, Europe and Asia, diagnosis of rare diseases and syndromes, clinical dysmorphologies, adult genetics and inheritance of diseases of complex etiology, novel disease gene identification, rare diseases beget common pathologies for therapeutics, precision medicine, population 'omics ,etc.

Discussion on ethical, legal and social issues related to genomic medicine, dealing with results of variants of uncertain significance, reporting of secondary findings, etc.

Case consults and discussions: diagnostic dilemmas, solved and unresolved cases, unknowns

Face2Gene session: phenotyping analysis, and use of deep learning and artificial intelligence to transform big data into actionable genomic insights

Special Genetic Counselling PSGCA Break-out session: covering topics such as Psycho-Oncology: Implications of Multigene Panel Testing on Cancer Patients, Genetic counselling and testing approaches in the paediatric population, Next Generation Genetic Counselling: How to be innovative when the genetic counsellor resource gap continues to widen, etc.

And MORE

Post -Autumn School Workshop (Nov 3 to Nov 4)

Hands-on training on NGS data analysis, filtration pipelines, variant analysis and curation, application of ACMG classification, assertions of pathogenicity and clinical interpretation. Participants will learn how to perform bioinformatics analysis and filter variants, use ACMG Standards and Guidelines for classifying sequence variants, perform clinical curation and interpretation. This workshop is suitable for diagnosticians and laboratorians: clinicians, genetic counsellors, molecular laboratory professionals, fellows, trainees, etc. Limited spaces available and extended on a first-come first-serve basis. All participants are expected to bring their own lap-top computer and will be provided with training data-sets to work on. Separate registration fees apply for this training workshop.