# Clinical Sequencing Meeting & APSHG Autumn School of Clinical Genetics 2018 SINGAPORE

**31 Oct to 2 Nov 2018**

- Clinical Sequencing and Genomics Meeting
- Autumn School of Clinical Genetics
- Face2Gene Workshop (phenotype 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 Variant Curation Workshop

**Venue: National University of Singapore**

A*STAR Genome Institute of Singapore

BRET Biomedical Research & Experimental Therapeutics Society of Singapore

NUS National University of Singapore

NUH kids National University Hospital

Asia Pacific Society of Human Genetics

# FIRST 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 genetic and genomic 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. Lectures from the invited Faculty will cover latest development 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 services and genetic testing companies.

**Target Audience:**

- Medical and clinical geneticists, genetic counsellors and other medical specialists involved in the diagnostic, management and counselling services of genetic conditions
- Laboratory professionals performing genetic testing
- Researchers , fellows, trainees and students in this field
- Industry professionals in next-gen sequencing and genetic testing

Registration and abstract submission opens in **June 2018**. Participants are advised to register early due to limited space. 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 rhe 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

**Programme Highlights:**

The three day meeting from Oct 31 to Nov 2 will cover:
Lectures on the impact of NGS on clinical diagnosis & genetic counselling, GWAS studies of multiple phenotypes and exomic analyses of Mendelian disorders, ACMG recommendation for reporting of variants, incidental findings, lessons from the Electronic Medical Records and Genomics (eMERGE) Network, clinical sequencing experience 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, etc.

Discussion on clinical testing; ethical, legal and social issues related 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, etc.

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

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 as 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. Only limited spaces are available and offered on a first-come first-serve basis. All participants are to bring their own lap-top computer and will be provided with training data-sets to work on. Separate registration fee apply for this training workshop.