

# CLINICAL SEQUENCING MEETING & APSHG AUTUMN SCHOOL OF CLINICAL GENETICS 2018

31 Oct to 2 Nov 2018

Venue: National University of Singapore  
CeLS Auditorium

- Clinical Sequencing and Genomics Meeting
- Autumn School in Clinical Genetics
- Genetic Counselling, Case Presentations, Ethics, Discussions

3 to 4 Nov 2018

Venue: National University of Singapore  
MD11 (CRC) Symposium Rooms

- NGS Bioinformatics & Clinical Curation Workshop



Register/Enquiry at:  
[www.apshg.info](http://www.apshg.info)  
[secretariat.meeting2018@gmail.com](mailto:secretariat.meeting2018@gmail.com)

# 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 basics on clinical genetics, and 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 diagnostic interpretation, genetic counselling and clinical management in the practice of genomic medicine. The lectures from the invited Faculty will cover latest developments in implementation of genomic medicine and genetic 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 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 open. 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 for overseas delegates. Please make your plans early. You may contact the Secretariat at [secretariat.meeting2018@gmail.com](mailto:secretariat.meeting2018@gmail.com) for accommodation, registration or abstract submission enquiries.

## Some of the 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

## Programme Highlights:

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

Lectures on impact of NGS on clinical diagnosis & genetic counselling, classification of genomic variants, lessons from the Electronic Medical Records and Genomics (eMERGE) Network, databases and clinical applications in precision medicine, cloud genomics, super-computing and genomic data, diagnosis of rare diseases and syndromes, clinical dysmorphologies, adult genetics and inheritance of diseases of complex etiology, diabetes and precision medicine, novel disease gene identification, rare diseases beget common pathologies for therapeutics, etc.

Discussion on contemporary issues related to genomic medicine, genetic testing, counselling, community genetics, etc. Sharing of experiences and cautionary tales on ordering genomic tests and interpretation of reports, etc. Forum discussion will engage both local and overseas experts and local audience.

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

Face2Gene presentation: use of digital phenotyping apps to facilitate phenotyping analysis and genetic diagnosis

Genetic Counselling session: encompasses topics such as Psycho-Oncology: lesson learned from challenging cancer genetic counselling cases, bridging the gaps of uncertainty in genetic counselling, genetic counselling and testing approaches in the paediatric population, etc.

And MORE .....

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

Hands-on training on basic 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 fee apply for this training workshop.