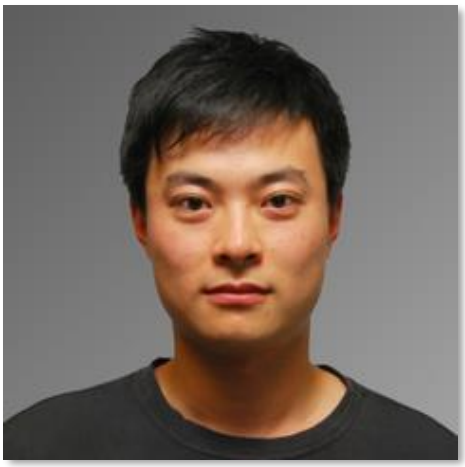
Hong Kong Genome Project

Biography

Dr Brian CHUNG Hon-Yin is a Clinical Associate Professor at Department of Paediatrics and Adolescent Medicine, LKS Faculty of Medicine, The University of Hong Kong. Specialized in clinical genetics and genomics, Dr Chung is interested in research fields including medical application of whole genome technologies, clinical genetics, genetic counselling, precision medicine and multi-omics. In recognition of his expertise in these areas, Dr Chung was awarded the Best Young Investigator Prize by the Hong Kong College of Paediatricians in 2017 and the Best Paper Award in the 2019 World Federation for Medical Education World Conference.

As a dedicated educator, Dr Chung was recognized for his efforts in teaching. He was awarded the 2018 Faculty Teaching Medal and the 2019 Outstanding Teaching Award by the University of Hong Kong. Apart from teaching, Dr Chung also takes up other responsibilities in the field of genetics and genomics. He is currently the President of Asia Pacific Society of Human Genetics. Since 2021, Dr Chung has been designated as Chief Scientific Officer at Hong Kong Genome Institute to promote the city's long-term development of genomic medicine by driving clinical application, advancing research, nurturing talents and enhancing public literacy.

Dr Becky MA graduated from the University of Hong Kong in 2014 with distinctions. She completed her training in Nephrology and Advanced Internal Medicine in Departments of Medicine of Queen Mary Hospital and Tung Wah Hospital, then postdoctoral fellow in Precision Medicine at Columbia University Centre for Precision Medicine and Genomics. She was awarded Gold Medal for the Best Thesis and highest score in Advancement Internal Medicine Assessment Award by the Hong Kong College of Physicians, the Hong Kong Academy of Medicine Distinguished Young Fellow. She has also been awarded the Health and Medical Research Fund Research Fellowship Grant on Unravelling the Genetic Causes of Chronic Kidney Disease in the Local Population. She is a member of the ClinGen Clinical Domain Working Group Curation Expert Panel and the International Society of Nephrology Emerging Leaders Program.



Dr Gene GAO is a specialist in neurology and is currently the associate consultant of Neurology in Queen Mary Hospital and honorary clinical assistant professor in the Department of Medicine, School of Clinical Medicine, The University of Hong Kong.

Dr Gao obtained his medical degree from Fudan University, Shanghai in 2004 and PhD in Biochemistry and Molecular Genetics from LKS Faculty of Medicine, The University of Hong Kong in 2011. He is interested and specialised in Neuromuscular and Neurogenetic Disorders, and he is one of the first fellows in Genetics and Genomics (Medicine) of The Hong Kong College of Physicians.

Dr Will CHAN is Clinical Assistant Professor at the Department of Medicine, The University of Hong Kong, and Honorary Associate Consultant in Cardiology at Queen Mary Hospital. He received overseas clinical training in Cambridge in the capacity of Sir David Todd Memorial Scholar of the Hong Kong College of Physicians, and is accredited for the Triple Fellowships in Cardiology, Advanced Internal Medicine, and Clinical Pharmacology & Therapeutics. He also received advanced Cardiology training as a Clinical Cardiac Electrophysiology Fellow at Royal Papworth Hospital, Cambridge.

Dr Chan's clinical and research interests entail the diverse spectrum of Cardiovascular Medicine. He has published more than 60 scientific papers in international peer-reviewed journals, including First-authored articles in JAMA, JAMA Cardiology, European Heart Journal, and Stroke. He has served as reviewer for reputable journals such as the European Heart Journal, and JAMA Network Open.

Dr Chan was an awardee of the Hong Kong RGC Clinical Research Fellowship 2023/24, to carry out cardiovascular drug repositioning research. He also currently serves as a Member of the Drug Management Committee, and Drug Advisory Committee, of the Hong Kong Hospital Authority. He established the Genetics Research Program For Personalized Medicine in Cardio-Oncology at HKU with support by the Li Shu Pui Medical Foundation Fellowship, with an aim of shedding lights on aetiology and fight against the clinical burden of heart diseases in cancer patients from the genetic perspective.

Abstract

Hong Kong Genome Project: Promoting Genomics in Precision Medicine (Dr Brian CHUNG)

The "Strategic Development of Genomic Medicine in Hong Kong" published by the Health Bureau in 2019 recommended to launch the city-wide Hong Kong Genome Project (HKGP), implemented by the Hong Kong Genome Institute (HKGI), to drive genomic medicine in Hong Kong. Through HKGP, HKGI aims at establishing genome database of local population, building infrastructure and talent pool to advance research in genomic science and enhance public literacy. HKGI has set up Partnering Centres and Referring Networks including HKU/Hong Kong West Cluster to help recruit eligible participants to facilitate the incorporation of genomic medicine into mainstream clinical service in Hong Kong by improving genomic diagnosis, personalised treatment as well as personalised prediction and prevention of disease risks. Since the launch of HKGP, HKGI has worked closely with clinicians and established "genomics champions" in different clinical specialties. HKGI provided support to these genomics champions to raise awareness, disseminate information and support the mainstreaming of genomics in their specialty.

In this session, Dr Brian Chung will introduce the backgrounds and achievements of how HKGI and HKGP developed a robust whole-genome sequencing (WGS) platform from patient recruitment all the way to variant curation, discussion and release of results. The benefits of WGS will be highlighted by cases covering germline variant analysis in rare disease and hereditary cancer, and somatic variant analysis in cancer. After that, three genomics champions from nephrology, neurology and cardiology will share their experience with HKGP and explain how HKGP helped the management of their referred patients.

Clinical Utility of Genetic Testing in Patients with ADPKD (Dr Becky MA)

Autosomal dominant polycystic kidney disease (ADPKD) is the most common hereditary kidney disease. Monoallelic pathogenic variants on two major genes (*PKD1* and *PKD2*) account for the majority of genetically resolved cases. In recent years, next generation sequencing studies allowed identification of other cystogenes, including *GANAB*, *DNAJB11*, *ALG5*, *ALG8*, *ALG 9* and *IFT140*. Patients harboring pathogenic variants on these genes typically demonstrate atypical phenotypes and milder disease course compared to those harboring *PKD1* and *PKD2* variants. Genetic testing enables physicians to achieve an accurate molecular diagnosis and prognostication, prompts cascade testing and better informs family planning.

Overcoming Genetic Testing Challenges in Neuromuscular Disorders (Dr Gene GAO)

The application of modern genetic testing methodologies to clinical practice enables robust development of genetic neuromuscular disorders and ends many diagnostic odysseys. However, we are easy to be at a loss when encountering genetic testing-negative cases. In this session, I will present a case of rare genetic myopathy with initially negative genetic testing findings, and share with you the journey of how we hit the final molecular diagnosis with the help of the Hong Kong Genome Project. The take home message is that neurogenetics is not purely genetic testing for neurological disorders. It is of paramount significance that clinicians have in-depth understanding of the applications and limitations of different genetic testing methods, and molecular aetiology of various genetic neuromuscular disorders to avoid abusing the genetic services.

Contemporary Clinical Perspectives of Genetics as Applied to Cardiology (Dr Will CHAN)

With improved understandings of genetic regulatory mechanisms of body functions and development of state-of-the-art sequencing techniques, opportunities of deciphering prior enigma of cardiovascular diseases and altering their clinical courses are more exciting than ever. From the Cardiologist's perspective, targeted genetic sequencing allows for clinical confirmation of disorders of a suspected genetic basis, uncovering of a submerged clinical

iceberg in the proband as well as in the pedigree. Such a diagnosis may in some cases lead to disease-modifying definitive treatments. In some cases such as in familial hyperlipidemia, optimized clinical risk control may be made possible through adopting the use of novel therapeutics for intensification of treatment. Furthermore, current Mendelian randomization research presents unprecedented opportunities for causality analysis of putative cardiovascular risk factors through dissecting relevant molecular pathways, and provides a platform for novel therapeutic target prioritization and potentials for drug re-purposing. In this regard, the Hong Kong Genome Project presents an exciting opportunity for patients and physicians alike, to understand further the above endless opportunities for research and clinical practice advancement.