Seminar 3

Saturday, 7 December 2019 • 09:00 – 11:00 • Lim Por Yen (G/F)

Clinical Application of Genetics and Genomics

Prof. LEUNG Tak Yeung

MBChB(CUHK), MD(CUHK), FHKCOG, FHKAM(O&G), FRCOG

Assistant Dean (Mainland Affairs)
Chairman, Department of Obstetrics and Gynaecology, Faculty of Medicine,
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Prof. Tak-yeung Leung is the Chairman of the Department of Obstetrics and Gynaecology of The Chinese University of Hong Kong, as well as the Assistant Dean (Mainland Affairs) of the Medical Faculty of CUHK.

Prof. Leung is also the Director of Maternal Fetal Medicine of the same unit, and leading the research and development in fetal genetic screening, diagnosis and therapy, such as carrier screening, chromosomal microarray, low-pass sequencing, and fetoscopic surgery. Professor Leung has published more than 290 papers in international peer reviewed journals, with an H-index of 38.

Prenatal Genetic Diagnosis: from Karyotyping to Whole Exome Sequencing

Prof. Rossa CHIU

MBBS, PhD, FHKCPath, FHKAM, FRCPA

Choh-Ming Li Professor of Chemical Pathology, Associate Dean (Development), Faculty of Medicine, The Chinese University of Hong Kong and Honorary Chief of Service of the Department of Chemical Pathology, Prince of Wales Hospital, Hong Kong

Prof. Rossa Chiu's research interests lie in the analysis of circulating nucleic acids found in human plasma. She has made significant contributions to the development of non-invasive prenatal diagnosis which led to the worldwide introduction of non-invasive plasma DNA tests for Down syndrome screening. She continues to develop applications of non-invasive diagnostics for prenatal and cancer assessments through cell-free nucleic acid analyses. Prof Chiu has published over 170 peer-reviewed research articles and has over 250 granted patents. She is a contributor of the key reference textbook in Clinical Chemistry, the Tietz textbook. Prof Chiu has received a number of international research awards and serves as Secretary to the Board of Directors of the International Society for Prenatal Diagnosis.

Genetics and Genomics in Current Clinical Practice

Diseases caused by genetic or genomic abnormalities were once managed by the rare individuals with highly specialised medical expertise. With the rapid expansion in knowledge in human genomics and the availability of powerful investigational tools, the use of genetic and genomic tests are now infiltrating many branches of medicine. For example, the risks of Steven-Johnson syndrome and toxic epidermal necrolysis could be mitigated by avoiding carbamazepine prescription in Asian individuals with HLA-B*1502 allele. Warfarin doses could be titrated according to the person's CYP2C9 and VKORC1 genotypes. Tyrosine kinase inhibitors are the first line drugs for patients with non-small cell lung cancer harbouring sensitising epidermal growth factor receptor mutations. Carrier screening, preimplantation genetic diagnosis and prenatal diagnosis of suspected single gene diseases are a part of pregnancy management. Maternal blood DNA analysis allows for sensitive and specific, yet non-invasive, screening of fetal chromosomal aneuploidies. Evidently, the incorporation of genetic and genomic information in medical care, aka. genomic medicine, is becoming a daily routine in many areas of medical practice now. How may we, as healthcare professionals, better equip ourselves to embrace this new paradigm? (Supported by the Research Grants Council of the Hong Kong SAR Government under the Theme-based research scheme (T12-401/16-W) and (T12-403/15-N)).

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Prof. SHAM Pak Chung

BA (Cambridge), BM BCh (Oxford), MSc (London), PhD (Cambridge), MRPsych

Suen Chi-Sun Professor in Clinical Science and Chair Professor of Psychiatric Genomics, Li Ka Shing Faculty of Medicine, The University of Hong Kong

Prof. Pak-chung Sham studied Medicine at Cambridge and Oxford Universities, and subsequently trained in Psychiatry at the Bethlem Royal and Maudsley Hospitals in the UK. In 2000, He was appointed Professor of Psychiatric and Statistical Genetics at the MRC Social, Genetic and Development Psychiatry Research Centre at King's College London. He was the Head of Department of Psychiatry, The University of Hong Kong from 2007 to 2011, and served as the Director of the Centre for Genomic Sciences from 2011 to 2019. Professor Sham has developed new statistical methods for the analysis of genetic data, and applied such methods to study the etiology of psychiatric disorders and other complex diseases.

Genetic Prediction of Common Diseases

All common diseases are caused by a combination of multiple genetic and environmental influences. The greater the contribution of genetic influences (as measured by the heritability), the greater is the clinical relevance of family history, which provides an indirect measure of genetic predisposition. Recent advances in genomic technologies and the meta-analysis of results from multiple studies have now enabled the identification of numerous genetic variants which confer increased risk of disease, and the possibility of directly measuring genetic predisposition. However, because of the small effect size of any single genetic variant, prediction requires multiple genetic variants to be aggregated into a single "polygenic score" in order to achieve sufficient prediction accuracy to have clinical impact. For many diseases, the predictive power of polygenic scores are now comparable to, or have exceeded, that of a positive family history. This may have implications for the prevention or early detection and intervention of common diseases.

Dr. Henry CK SZE

MBBS (HK), FRCR, FHKCR, FHKAM (Radiology), PDip Epidemiology and Biostatistics

Specialist in Clinical Oncology

Dr. Henry Sze obtained his medical degree from the University of Hong Kong and started his career in the Department of Clinical Oncology, Pamela Youde Nethersole Eastern Hospital. He obtained fellowship from the Royal College of Radiologists and was awarded the Rohan Williams Medal. From 2013 to 2015, he was Clinical Assistant Professor in the Department of Clinical Oncology, Faculty of Medicine, The University of Hong Kong. In 2016, he joined Pamela Youde Nethersole Eastern Hospital as Associate Consultant. He started private practice since 2019. He has multiple publications in peer-reviewed journals and is the author of several book chapters.

Cancer Genomics and Personalised Oncology Care

Cancer is caused by the accumulation of genetic and epigenetic alterations in DNA of normal somatic cells. Early cancer research focused on genetic and epigenetic mechanisms of carcinogenesis have led to the discovery of crucial genetic events for many of particular malignancies. The greatest discoveries in the field have been driven by advancements in technology. In the 1990s, the human genome project and the use of polymerase chain reaction (PCR)-based target sequencing led to some of the most important genomic discoveries and corresponding pathways to date. They have not only helped to elucidate the mechanism of carcinogenesis but also opened up a brand new avenue in new cancer therapies by creation of drugs targeting all the specific genes and proteins that allow the cancer cells to grow and survive. Targeted therapy has revolutionized cancer treatment and has largely replaced chemotherapy in many oncological diseases. As we are now entering the era of personalized medicine in oncology, more precise decision tools based on genomic profiling are developed to allow oncologist to help patients based on a genetic understanding of their disease.