Seminar 7

Saturday, 7 December 2019 • 11:30 – 12:30 • Lim Por Yen (G/F)

Genetics and Genomics in Clinical Practice

Dr. Ivan FM LO

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Dr. Ivan FM Lo is Head of the Clinical Genetic Service of the Department of Health, a tertiary referral centre of clinical genetic service in Hong Kong for over 30 years. Dr. Lo was trained in genetics and genomics in the Clinical Genetic Service since 1991, and received training in biochemical and molecular genetics at the University of British Columbia, Canada, in 1996. He also received Paediatric training in Hong Kong and became a Fellow of the Hong Kong College of Paediatricians in 1998. He is also Honorary Clinical Associate Professor of the Department of Paediatrics & Adolescent Medicine of both medical schools in Hong Kong, and Chairman of the Genetics & Genomics (Paediatrics) subspecialty board under the Hong Kong College of Paediatricians. Dr. Lo's special interests include dysmorphology, neurogenetics, molecular genetics and application of next generation sequencing technology. He has more than 100 publications in international peer reviewed medical/genetic/genomic journals.

A Genomic Approach to Paediatric Rare Disorders

The field of medical genetics has transitioned steadily from genetics to genomics over the last 10 years. It was also called a paradigm shift. From the laboratory perspective, a genetic approach is usually a targeted one that studies genes one by one, or even worse, exon by exon; while a genomic approach is one that studies multiple genes in one go, even without the need of a clear target. This is of particular relevance to a diagnostic laboratory for rare genetic disorders, because these disorders, though individually rare, are estimated to reach 5000-8000 in number. Furthermore, a lot of disorders are clinically non-specific, that means the clinical signs and symptoms cannot give a clue about the underlying genetic defect; and a lot of disorders are genetically heterogeneous, that means there are multiple genetic loci implicated in the same phenotype. The paradigm shift was most evident in the repertoire of genetic tests available in a genetic laboratory. In contrast to the "gene after gene" approach of conventional molecular genetics technology, NGS enables simultaneous analysis of hundreds or thousands of genes. Gene panel-based analyses can easily deal with the genetically heterogeneous disorders. The number of genes interrogated by these gene panels range from 2 to over 100. The diagnostic capacity is almost without limit. For ultra-rare genetic diseases, whole exome sequencing can be done and achieved a diagnostic rate of about 30%. In conclusion, the adoption of genomic technology in the genetic laboratory has highly increased the diagnostic capacity and efficiency.

Dr. Derrick KS AU

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Dr. Derrick AU received medical education at Brown University and specialist training in Hong Kong. He served in clinical service for two decades before taking up management positions in the Hospital Authority (HA), including Director of Quality & Safety, overseeing technology assessment among other duties. He joined CUHK in 2017. Presently he also serves as Chairman of the HA Clinical Ethics Committee through honorary appointment. Besides, Dr. Au is a writer on healthcare and humanities. A book newly published (《生命倫理的四季大廈》) looks at the intertwined journey of bioethics and biomedical technologies.

Genomic Medicine and Common Good: Promises and Tensions

Advances in genomics research and genomic medicine promise a new era of personalized medicine on the one hand, and on the other hand push for a change on traditional ethical framework in medicine and human research. While personalized medicine promises to provide the best case to individual patients, questions arise as to whether this promise can be truly delivered, and whether such paradigm is promoting a "Me Medicine" which, at its extreme, might be counterproductive for the common good. Paradoxically, the idea of "the common good" has also been used by advocates of genomic medicine to challenge the traditional framework of informed consent and data privacy. At heart of the tension between genomic medicine and the common good is the issue of equity. Will the idea of personalized medicine eventually prove too much to be sustainable and equitable in health care of the population? This presentation considers the promises of genomic medicine and the related ethical concerns.