**Presentation 1:**

## How does genome sequencing help in medical research and enhance quality of healthcare

Professor Richard CHOY Kwong-wai
Department of Obstetrics and Gynaecology
The Chinese University of Hong Kong

**Summary:**

Advancements in genome sequencing technologies have revolutionized disease diagnosis. Genome sequencing detects single-nucleotide variants (SNVs), copy number variants (CNVs) and structural variants (SVs) across the human genome and its diagnostic application is an emerging field. We explored target-enrichment next-generation sequencing method for genetic screening and diagnosis of hereditary hearing loss patients. Soon after, our results demonstrated that the development and implementation of next-generation sequencing technologies enabled both research and diagnostic opportunities to investigate genetic variants across the genome even down to the single-nucleotide resolution. Subsequently by applying this state-of-the-arts technologies in a genetically undiagnosed prenatal cohorts, we discovered that causative genetic aberrations underlying many diseases are beyond the detection limit of conventional karyotyping and chromosomal microarray analysis. Our HMRF funded study shows genome sequencing provides additional genetic diagnoses in a significant portion of patients. Herein, we share our experiences on genome sequencing for prenatal diagnosis, discuss its clinical applications, and how it helps in research and enhance quality of healthcare in reproductive medicine.