

MGI Workshop

Part 1:



Dr. Kwong Wai Choy (Richard)

**Associate Professor, Dept of Obstetrics & Gynaecology,
Deputy Director, Prenatal Genetic Diagnosis Centre,
The Chinese University of Hong Kong**

Bio:

Dr. Choy obtained his BSc degree in Biomedical Sciences from the University of Bradford and M(Med)Sc in Immunology at University of Birmingham, UK. He received postgraduate training in genetics and was awarded Doctor of Philosophy by The Chinese University of Hong Kong in 1999. Dr. Choy's main research interests include the development of novel molecular diagnostic strategies and innovative applications of molecular analysis tools in prenatal diagnosis. His current research focuses on (1) structural genomic variation (including copy number variants); (2) development and application of the microarray-based Comparative Genomic Hybridization (array CGH) and massively parallel sequencing technology for prenatal diagnosis. In 2007 he founded the Agilent microarray core facilities in CUHK, which is the first centre of its kind in Asia dedicated to prenatal diagnosis with a focus on molecular basis of normal and abnormal fetal development. Dr. Choy is an Honorary Scientific Officer (medical) at the New Territories East Cluster of Hospitals, Hong Kong, where he is involved in the delivery of the laboratory diagnostic service. He is also the board member of the Hong Kong Society of Medical Genetics since 2009.

Title: Low-Pass Genome Sequencing versus Chromosomal Microarray Analysis: Implementation in Prenatal Diagnosis

Abstract:

Emerging studies suggest that low-pass genome sequencing (GS) provides additional diagnostic yield of clinically significant copy-number variants (CNVs) compared with chromosomal microarray analysis (CMA). However, a prospective back-to-back comparison evaluating accuracy, efficacy, and incremental yield of low-pass GS compared with CMA is warranted. It is concluded that in the context of prenatal diagnosis, low-pass GS identified additional and clinically significant information with enhanced resolution and increased sensitivity of detecting mosaicism as compared with the CMA platform used. This study provides strong evidence for applying low-pass GS as an alternative prenatal diagnostic test.

Details subject to change.

Part 2:



Dr. Matthew Callow
Senior Director of Biochemistry, Complete Genomics

Bio:

Dr. Matthew Callow is Senior Director of Biochemistry at Complete Genomics Inc. in San Jose, California. He received his PhD from the University of Western Australia in 1992 before embarking on post-doc research in the laboratory of Dr. Eddy Rubin at the Lawrence Berkeley National Laboratory. His early research focused on exploring lipoprotein metabolism utilizing transgenic mice and expression microarrays. He then joined Callida Genomics Inc. and Complete Genomics Inc., developing sequencing technologies which included early work on DNA nanoballs and sequencing-by-ligation strategies. With the acquisition of Complete Genomics Inc. by BGI Group, his research progressed into the development of antibody-based sequencing-by-synthesis

technologies including CoolMPS™.

Title: CoolMPS™: Advanced Massively Parallel Sequencing Using Base-Specific Antibodies

Abstract:

Massively parallel sequencing (MPS) on DNA nanoarrays allows for billions of reads at relatively low cost and enables a multitude of genomic applications. Improvements in read length, sequence quality and further cost reduction will enable more affordable and accurate health monitoring tests. Currently the most efficient MPS uses dye-labeled reversibly-terminated nucleotides (RTs) that can be expensive to make and challenging to incorporate by polymerases. Furthermore, a part of the dye-linker ("scar") remains on the nucleobase after cleavage of the fluorophore and can interfere with subsequent sequencing cycles. Here, we present the development of a novel MPS chemistry (CoolMPS™) utilizing unlabeled RTs and four natural nucleobase-specific fluorescently labeled antibodies with fast detection. We implemented CoolMPS™ on MGI's PCR-free DNBSEQ MPS platform using arrays of 200nm DNA nanoballs (DNBs) generated by rolling circle replication and demonstrate a three-fold improvement in signal intensity and elimination of scar interference. Up to 400-base single-end reads, and 2x150-base pair-end reads, with high quality were readily generated and with low out-of-phase incorporation. Furthermore, DNBs with less than 50 template copies were successfully sequenced by high intensity CoolMPS™ with three-times higher accuracy than in standard MPS. CoolMPS™ chemistry, based on natural nucleobases, has the potential to provide longer, more accurate and less expensive MPS reads, including highly accurate "four-color sequencing" on dye-crosstalk-free two-color imagers.

Details subject to change.

Part 3:



Dr. Zhouchun Shang (Shane)
Project Leader, BGI Research

Bio:

Dr. Zhouchun Shang received her PhD in Biomedical Engineering from Tongji University, China, where she studied the molecular regulation of cell fate decision in human early neural differentiation by single-cell transcriptome approach. She joined BGI-Shenzhen in 2010 and became a group leader since 2012. She is also Associate Professor of Northwest University in Xi'an, China. Combined with the trans-omics platform, presently her group focuses on embryonic development as well as stem cell research e.g., chromatin regulatory landscape during human pre- and post-implantation development; integrating the single-cell omics technology to study heterogeneity and functionally-distinct subsets e.g., single-cell transcriptome profiling of human placenta et al. She has published several papers in major international journals including Nature, Nature

Communications, Cell Research, Gigascience, Cell Death & Disease, Nature Cell Biology etc., and obtained over eight patents.

Title: Dissecting Cell Heterogeneity Using Single-cell Omics Powered by DNBSEQ

Abstract:

The rapid proliferation of single-cell sequencing technologies has greatly improved our understanding of heterogeneity in terms of genetic, epigenetic, and transcriptional regulation within cell populations. DNA nanoball sequencing (DNBSEQ) adopts rolling circle amplification (RCA) strategy and combinatorial probe-anchor synthesis (cPAS) based method. The advanced DNBSEQ facilitates a low rate of index hopping and template amplification error, enabling highly accurate sequencing data especially for single cell research. While combined with single-tube long fragment read (stLFR) technology, it will offer a strategy for single-cell full-length RNA profiling. This talk will introduce the advantages and applications of DNBSEQ and stLFR in the large-scale single-cell sequencing.

Details subject to change.