

MEETING REPORT

The ICG-V: advances in genomic profiling across the spectrum of biology and medicine

Jingde Zhu*

Abstract

A report of the 5th International Conference on Genomics (ICG-V), Shenzhen, China, 15-18 November 2010.

The 5th International Conference on Genomics (ICG-V) showcased the latest academic and technological advancements in genomic research from international groups and from the BGI (formerly known as the Beijing Genomics Institute; <http://www.genomics.cn/en/>). This 3-day conference enabled over 400 participants, more than 20% of them from outside mainland China, to witness scientific progress, share ideas, discuss strategies for better research, initiate collaborative efforts, and address urgent needs in genome research. The conference began with two plenary sessions on genomic perspectives at single or multiple levels of various biological systems. The plenary sessions were followed by eight concurrent sessions focusing on key issues in bioinformatics, genomic studies in Chinese hamster ovarian cells, human disorders, marine and aquaculture organisms, horticultural crops, cancer, and agricultural and ecological fields.

The motivation remains irresistible to sequence the genome of all species; the emphasis on new biological implication and mechanistic insights was evident in almost all the reports presented. Attempts to translate key genomic information from such large 'discovery or sequencing driven' projects into the daily routine, to control/prevent disease and improve economical crops and livestock, were more evident than in previous gatherings. Second generation sequencing technologies have offered monumental improvements in the production and accuracy of sequencing (in terms of read length, cost, and ease of operation), resulting in an avalanche of sequencing datasets. Nevertheless, matched bioinformatic

capability is urgently required to analyze these genomic datasets to obtain a better biological understanding of, and mechanistic insights into, disease-associated genetic variants and copy number/structural variations. It is imperative for genomicists to respond to this urgent need of the biological community by offering better access to the 'meaningful' information. Related to this, a concern was expressed about the lack of independent validation of the key conclusions that were drawn in most of the studies reported at the meeting.

Critical genetic variants that affect the initiation and progression of various major diseases, including cancer, autoimmune diseases, diabetes and immunodeficiency disorders, were specifically discussed at the conference. Whole-genome or -exome sequencing is now preferred over the array-based genome-wide association study (GWAS) platform for identification and validation of genetic variants of biological importance. This welcome move seems to be best attributed to the much wider availability of affordable and enhanced sequencing capabilities. The conceptual shift that emphasizes the role of rare genetic variants in the pathology of the major diseases remains to be widely accepted. Both the biological implications and mechanistic interpretations of the epigenetic (or epigenomic) makeup of mammals were also stressed, in view of the fact that the epigenetic framework is more amenable than DNA sequence to changing environments and directly dictates phenotypic diversity.

The complexity of biological systems is vividly reflected by the cellular heterogeneity in gene expression and therefore in phenotype. The merit of the conventional 'mean-of-the-sum' approaches to mRNA expression or genetic diversity has been challenged by the latest single cell assays. Using a SOLiD sequencing platform, Fuchou Tang (Peking University, Beijing, China) established a global mRNA expression profile at the single-cell level. Both coverage and depth of reads were impressive. Tang reported that the datasets, having 20 million to 100 million reads from each of 33 individual cells with approximately 60 to 80% meaningful, covered approximately 12,000 expressed genes (approximately 50% of the total annotated protein-coding genes). Luting Song (BGI,

*Correspondence: zhujingde@gmail.com
Shanghai Cancer Institute/Renjin Hospital, Shanghai Jiaotong University, Shanghai 200032, China

Shenzhen, China) reported their attempt to produce a draft single-cell genome. The present dataset consists of approximately 25-fold coverage of 80% of the genome. Both presentations highlighted the most recent technological refinements and the potential impact of such single-cell genomic approaches on today's biomedical research.

In addition, great advancements in the efforts aiming to create more robust and affordable platforms for genome research were witnessed. Stephen Turner (Pacific Biosciences, Foster City, USA) described the striking improvements in sequencing that they have achieved using nanopore or single-molecule sequencing apparatus. The aspiration of completing a human genome sequencing project with a budget of less than US\$1,000 should be achieved much sooner than previously anticipated. The construction of shorter reads from second generation sequencing machines to make a genome draft remains the biggest challenge. This need seems to be satisfied, at least partially, by the optical mapping platform with which the length of the fluorescence-labeled restriction fragments (a dozen kilobase pairs to several million base pairs in length) can be measured with great accuracy. David Schwartz (University of Wisconsin, Madison, USA) described their efforts to fill the gaps left in the

public genome databases and profile the disease-related gross structural genomic variations in breast cancer cells. Han Cao (Bionanomatrix, Philadelphia, USA) presented a new form of the nanochannel fluid array/optical mapping technology. The suggested utility of these two optical mapping technologies in future clinical practice for detection of diseases (such as cancer) that are notorious for prevalent gross structural changes in the genome was well received.

The International Conferences of Genomics under the BGI's organization follow Deng Xiaoping's philosophy, 'Developing is an unyielding principle', which has created the economic miracle in mainland China for more than three decades. The eagerness for new ideas, technologies and collaborations aired in this meeting, as in all the previous four annual events, has made it a unique opportunity to grasp new scientific concepts and initiate collaborative efforts in large-scale genomic research. There is no doubt that the ICG-VI in 2011 will bring more excitement from and to all the participants.

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