10x Genomics' GemCode Technology Utilized for de novo Assembly and Phasing of Korean Genome

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New research published in Nature provides insights into new human reference genomes

PLEASANTON, Calif. — October 6, 2016 – 10x Genomics, a company focused on improving and broadening the application of genomic information, today announced the use of the GemCode Technology in a new publication in the journal *Nature* demonstrating the utility of Linked-Reads for diploid *de novo* assembly and haplotype phasing of the Korean genome, AK1.

This work presents the most contiguous diploid human genome assembly thus far, with extensive investigation of unreported and Asian-specific structural variants, as well as high-quality haplotyping of clinically relevant alleles having potential to improve precision medicine. This is a significant improvement over current methods for genome analysis, which are biased towards European and Caucasian populations. In addition, this method improves issues in situations where short-read sequences from an individual are aligned to a haploid consensus reference genome, biasing the resulting analyses and often failing to capture sequences novel to a given individual genome or ethnic population.

To more fully investigate the range of structural variation and better understand the phased diploid genome architecture of AK1, investigators performed *de novo* assembly for each "haplotig" representing the assembly of each of the two haplotypes from each homologous chromosome pair. Compared to other methods, integrating 10x Genomics' Linked-Read data with PacBio long-reads, Illumina short-reads, and reads from BAC libraries, drives the assembly of considerably larger phase blocks than other integrated methods, with an N50 length of 5.7 Mb.

"10x Genomics' GemCode platform and targeted-sequencing of BAC clones were indispensable for the successful *de novo* haplotype phasing in this study," said Jeong-Sun Seo, M.D., Ph.D., lead author of the study and Professor and Director of the Department of Biochemistry and Molecular Biology ILCHUN Genomic Medicine Institute at Seoul National University College of Medicine. "I look forward to the further advances and application of 10x Genomics technology and their potential applications towards precision medicine."

The results of the study demonstrate the power of *de novo* genome assembly and "true" diploid phasing for detection of the full range of genetic variation, and for understanding the haplotype structure in clinically relevant genes. These genes include the human leukocyte antigen (HLA) genes of the hypervariable major histocompatability complex (MHC), which are genes important for pharmacogenomics such as *CYP2D6*, as well as genes thought to be involved in rare inherited autosomal recessive disorders like familial Mediterranean fever and Upshaw–Shalman syndrome.

"*de novo* assembly and phasing are in the cross-hairs of our mission, and our technology is well-suited for clinical and population-scale sequencing," said Serge Saxonov, co-founder and chief executive officer at 10x Genomics. "This study clearly demonstrates the potential for *de novo* assembly methods which resolve haplotypes and reveal novel genetic variation that was previously undetected. Streamlined methods that provide *de novo* assembly and haplotyping will usher in a new standard in human genome sequencing."

The paper titled, "*de novo* assembly and phasing of a Korean human genome" was published yesterday in *Nature* (doi:10.1038/nature20098) and can be accessed online at:
http://www.nature.com/nature/journal/vaop/ncurrent/full/nature20098.html

About 10x Genomics

10x Genomics is changing the definition of sequencing by providing an innovative genomics platform that dramatically upgrades the capabilities of existing sequencing technologies. This is achieved through a combination of new microfluidic science, chemistry and bioinformatics. By implementing GemCode Technology within the Chromium System, researchers can now, for the first time, find new structural variants, haplotypes, and other valuable genomic information with comprehensive workflows for Single Cell, Genome and Exome applications that incorporate their pre-existing sequencing technologies.

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