Researchers Create New Way To Locate Big Genetic Variants

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(PhysOrg.com) -- Yale University researchers, analyzing hundreds of billions of bits of genetic information, have collated and standardized 2,000 signposts that mark the boundaries of large blocks of human genomic variants.

This library of genomic “breakpoints” was published in the Dec. 27th edition of Nature Biotechnology.

Genome sequencing, or the ordering of the billions of nucleotides that make up the genomes of living organisms, has been a key tool in modern biological research. The quest for variants that change the function of genes began by first focusing on changes, called single nucleotide polymorphisms (SNPs) in a single genetic letter (or nucleotide). Advances in sequencing technology now enable scientists to decode the genome more rapidly and efficiently, and have paved the way for identifying large block variants, called structural variants or SVs.

These variants cause more nucleotide differences between individuals than SNPs. Some SVs, in fact, involve thousands of base pairs and can wipe out whole genes or create additional copies of other genes that can have major effects on an organism. SVs are sometimes associated with diseases such as cancer and HIV and also with developmental disorders.

Graduate students Hugo Lam and Jasmine Mu and their colleagues in the research team led by Mark Gerstein, professor of molecular biophysics and biochemistry, computer science, computational biology and

bioinformatics, analyzed data from recent personal genomic studies, such as the 1000 Genomes Project, to identify precise “breakpoint” locations of SVs.

They have shown how this library of breakpoints can help researchers rapidly scan for and characterize SVs in a newly sequenced personal genome. In fact, the sequences in the library can even be put on a commercial SNP chip, which can then be used to assess SVs quickly in population studies.

Provided by Yale University