GENETICS

YOU GET WHAT YOU PAY FOR: GENOME SEQUENCING

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I always had the impression that the “standard” Illumina sequencing is a big business but not big science. A more recent paper of the Eichler group confirms that ““Multi-platform discovery of haplotype-resolved structural variation in human genomes”

The incomplete identification of structural variants (SVs) from whole-genome sequencing data limits studies of human genetic diversity and disease association. Here, we apply a suite of long-read, short-read, strand-specific sequencing technologies, optical mapping, and variant discovery algorithms to comprehensively analyze three trios to define the full spectrum of human genetic variation in a haplotype-resolved manner. We identify 818,054 indel variants (<50 bp) and 27,622 SVs (≥50 bp) per genome. We also discover 156 inversions per genome and 58 of the inversions intersect with the critical regions of recurrent microdeletion and microduplication syndromes. Taken together, our SV callsets represent a three to sevenfold increase in SV detection compared to most standard high-throughput sequencing studies, including those from the 1000 Genomes Project.