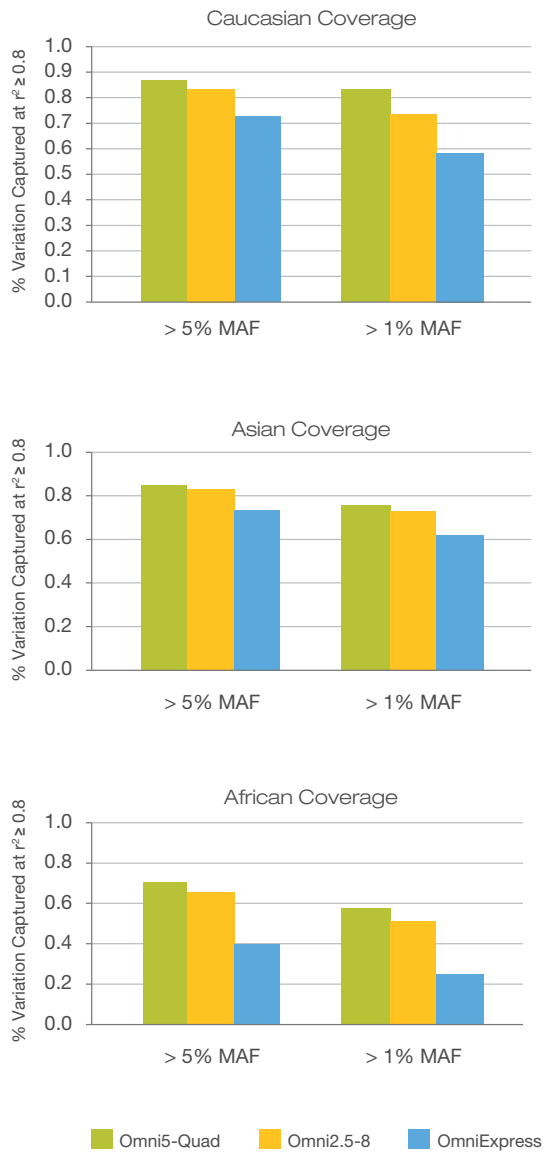


reference database, as we now know, only offered limited information about extent of genetic variation. By the end of the project, it contained ~3.5 million variants, targeting MAFs > 5%. In light of the more comprehensive data available from the 1kGP, the reference point for coverage statistics must be adjusted. As shown in Table 1 and Figure 2, Omni arrays offer greater than 80% coverage of variants with respect to 1kGP data (MAF > 1%).

Figure 2: Maximized Genomic Coverage



Omni whole-genome microarrays provide superior coverage of common and rare variants across Caucasian, Asian, and African populations. The Omni5 array provides the highest coverage of variants down to 1% MAF. With the option of selecting 500K additional custom variants on the Omni5, researchers can use tagSNPs to increase coverage by up to 10%.

Structural Variation Analysis

Structural variation is thought to be a significant contributor to the genetic basis of human disease. Dense genome-wide coverage on Omni microarrays, coupled with the sensitive Infinium assay, offer researchers a powerful tool for structural variation analysis. The assay delivers very high signal-to-noise ratios and low overall noise levels, which are ideal for precise structural variation analysis. Whether it's genotype calling, structural variation analysis, or both, Omni arrays provide a single solution for any course of genetic research.

Superior Data Quality

The Omni family of microarrays is powered by the Infinium assay, the industry's most trusted, proven DNA analysis platform for both genotyping and CNV studies. The assay is deployed using Illumina proprietary BeadArray™ technology, which allows Omni arrays to deliver a high degree of flexibility, enabling a number of sample formats and a wide multiplex range.

Infinium BeadChips have low DNA input requirements, expanding the range of sample sources that can be used for a study. Genetic researchers worldwide have embraced this technology to catalyze many revolutionary discoveries in disease research and have amassed a vast publication record. Infinium products deliver exceptionally high-quality data with respect to call rates (average > 99%), reproducibility (> 99.9%), and low sample redo rates (Table 1). With such high data quality, the assay minimizes the number of false positives, allowing researchers to avoid time-consuming and frustrating extra analysis and expensive follow-up studies on erroneous associations. High signal-to-noise ratios and low overall noise levels allow for precise, reliable copy number analysis.

Proven Technology

The combination of the Illumina well-proven BeadArray platform, assay technology, and proprietary algorithms present a powerful solution for genetic analysis, delivering the highest quality and most convenient user experience.

BeadArray Manufacturing

Illumina BeadArray technology is based on small silica beads that self assemble in microwells on planar silica slides. Each bead is covered with hundreds of thousands of copies of a specific oligonucleotide that act as the capture sequences in the Infinium assay. Once the beads have self assembled, a proprietary decoding process maps the location of every bead, ensuring that each one is individually quality controlled. The result of this manufacturing process is that every BeadChip undergoes rigorous testing to assure the highest possible quality standards.

Assay Chemistry

The Infinium assay can be scaled to unlimited multiplexing without compromising data quality, unlike many alternative PCR-dependent assays. The simple, streamlined workflow is common across all products, no matter how many SNPs are being interrogated. Likewise, the data acquisition process and analysis are the same. The Infinium assay protocol features single-tube sample preparation and whole-genome amplification without PCR or ligation steps, significantly reducing labor and sample handling errors. After hybridizing unlabeled

