

Table 1: HumanCVD BeadChip Performance And Specifications

| Category | Specification | CEU | CHB+JPT | YRI | TOTal |
|---------------------------|---------------|---------|---------|--------|--------|
| Call Rate | > 99% | 99.84% | 99.85% | 99.87% | 99.79% |
| Reproducibility | > 99% | 99.99 % | 99.99% | 100% | 99.99% |
| Mendelian Inconsistencies | < 0.01% | 0.006% | 0.01% | N/A | 0.001% |

Results based on 284 HapMap samples including 59 trios and 15 replicates.

ture and ancestry informative SNP markers (AIMs) for African versus European ancestry. It also features 400 AIMs for regional European ancestral populations. See Table 2 for a description of HumanCVD BeadChip probe categories and coverage.

High Density

The HumanCVD BeadChip has an average density of 36.5 SNPs per locus across Group 1 loci, which are highly associated with CVD, providing more than twice the density of standard whole-genome genotyping arrays. This higher density offers greater resolution for detecting potentially causal variants, including variants with low odds ratios, without the need to increase sample size.

Validated Markers

The HumanCVD BeadChip was validated using 117 HapMap samples (Table 1). The Caucasian (CEU) and Han Chinese , Japanese (CHB+JPT), and Yoruban (YRI) populations contain 30,774, 28,640, and 35,600 SNPs, respectively, with MAFs ≥ 0.05. The HumanCVD BeadChip also offers coverage of rare variants with MAFs between 0.01 and 0.05 identified from HapMap allele frequencies (Figure 2). BeadChip markers target 4,815, 4,215, and 4,918 rare SNPs in

the CEU, CHB+JPT, and YRI populations, respectively. A significant portion of SNPs on the HumanCVD BeadChip are derived from SeattleSNPs⁶, the literature, and recent resequencing efforts⁷, offering coverage in populations beyond HapMap.

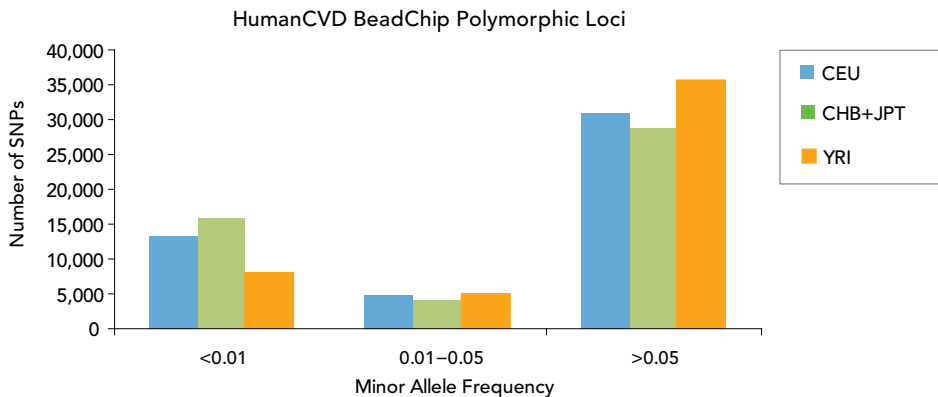
Analysis Simplified

Illumina’s BeadStudio analysis software is a powerful and user-friendly tool for analyzing data generated by the HumanCVD Genotyping BeadChip. The Genotyping Module provides automated genotype calling and sample clustering. Powerful visualization tools and quality-control features enable graphical output of data and reports on reproducibility and Mendelian consistency.

Automation

As with most of Illumina’s standard DNA Analysis products, an optional Laboratory Information Management System (LIMS) and robotic automation track samples to provide accurate and efficient workflow management and overall project management. This system, custom designed for Infinium Assay workflows, allows labs to maximize their throughput with a completely integrated microarray solution.

Figure 2: Human BeadChip Coverage Of Common And Rare SNPS



HumanCVD BeadChip content provides more than twice the density of standard genotyping arrays across loci associated with CVD.

