



Data Sheet

Affymetrix GeneChip® Human Panel 1 10K cSNP Kit

The Affymetrix GeneChip® Human Panel 1 10K cSNP Kit contains 10,000 amino acid-changing SNPs that are multiplexed in a single assay. The panel includes SNPs chosen to represent more than 8,000 genes that have validated (double-hit) non-synonymous public SNPs that code for functional changes. It allows researchers to perform cost-effective studies for the direct detection of non-synonymous SNPs in the whole genome. The Human Panel 1 10K cSNP Kit is designed to work with the Affymetrix GeneChip® Scanner 3000 (with the appropriate upgrade.)

Features and Benefits

- An innovative, highly multiplexed assay that exploits the Molecular Inversion Probe (MIP) technology using the highest level of multiplexing to enable affordable whole-genome association studies.
- Direct detection of disease alleles can be obtained without reliance on linkage disequilibrium (LD) studies.
- Since functional SNPs (amino acid-changing) have a higher probability of being causative, significant associations can be directly linked to biological pathways and gene function.

Key Specifications

- Accuracy ≥ 99.3 percent
- Data Completeness ≥ 98 percent
- Repeatability ≥ 99.25 percent
- Quantity of genomic DNA required without amplification is 4.0 μg
- Quantity of genomic DNA required with recommended amplification is 50 $\pm 10\text{ng}$
- Throughput of 48 samples per day (~ 0.5 million genotypes/day)

Panel Design

The GeneChip® Human Panel 1 10K cSNP Kit exploits the innovative Molecular Inversion Probe (MIP) technology¹⁻³ resulting in a highly multiplexed assay that includes 10,000 SNPs that can be

used in human genotyping studies. It has been proven in large-scale applications such as the International Haplotype Mapping project (www.hapmap.org/).

This panel, representing >8,000 human genes, consists of 10,000 non-synonymous validated SNPs from the public database that occur at a frequency of greater than one percent. Most of the major drug target gene families are represented in this SNP panel.

Coverage and Performance

The Human Panel 1 10K cSNP Kit has a median interval of <100 kb between SNPs. The genome coverage is shown in Figure 1. Reduced density is expected in gene-poor regions, such as telomeres and centromeres. Figure 2 shows the typical coverage of gene families that have been used successfully as targets for drug development. To determine the performance of this 10K cSNP

Figure 1.

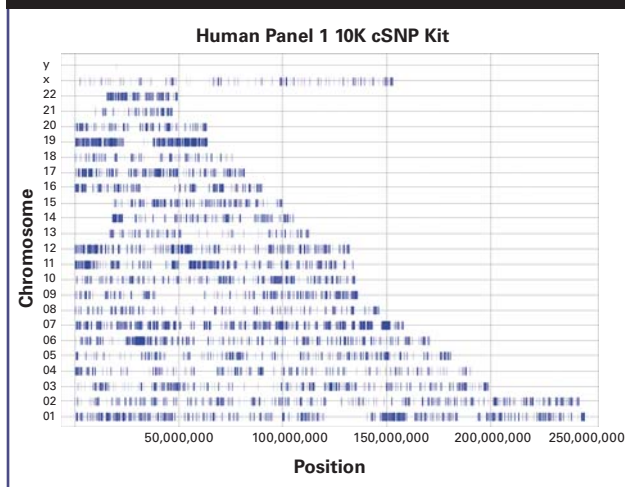
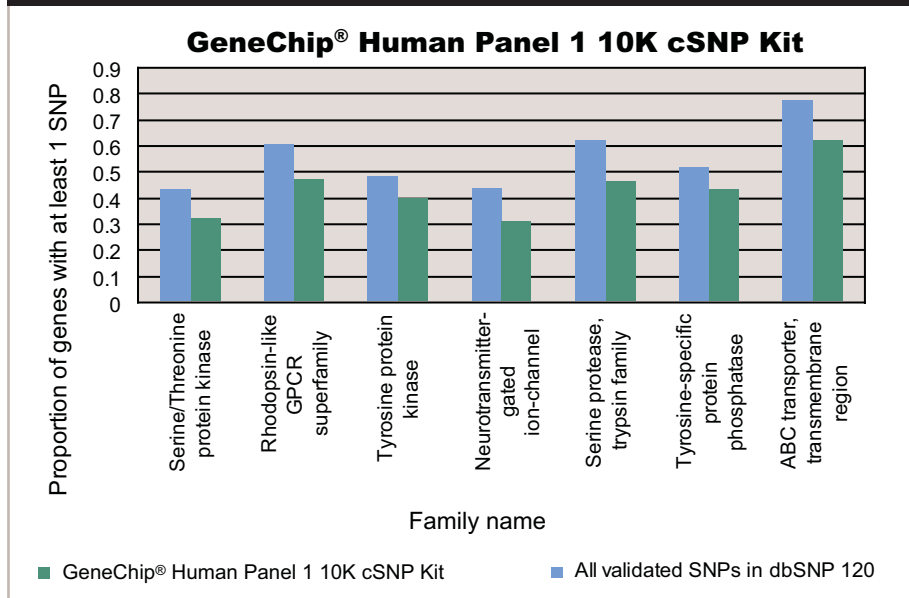


Figure 2.



Panel, DNA samples across three populations (Caucasian, Asian, African) were genotyped (total of 2,900,476 genotypes).

Accuracy measured using Mendelian inheritance across 60 trios was 99.81 percent. In addition 2,700 cSNPs in the panel overlapped with reference genotypes generated in the HapMap project. The concordance with these genotypes was 99.6 percent.

Repeatability measured across 28 different individuals who were each genotyped two to three times was 99.89 percent.

Data completeness (the average call rate across passed samples for passed SNPs) was measured on genotypes across the panel at 99.06 percent.

Minor allele frequency was measured at 21 percent in the Caucasian population, 20 percent in the Asian population, and 21 percent in the African population.

REFERENCES

1. Hardenbol, P, *et al.* Highly multiplexed molecular inversion probe genotyping: Over 10,000 targeted SNPs genotyped in a single tube assay. *Genome Research* **15**:269-275 (2005).
2. Hardenbol, P, *et al.* Multiplexing Genotyping with Sequenced Tagged Molecular Inversion Probes. *Nature Biotechnology* **21**:6 (June 2003).
3. Jain, M., *et al.* Benchtop Genotyping. *Genetic Engineering News* **24**:18 (10/15/2004).

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Ordering Information

Affymetrix GeneChip® Human Panel 1
10K cSNP Kit, Version 1.5

900866 Kit contains enough reagents to process a total of 24 assays (including one control)

Affymetrix GeneChip® Universal 10K
Tag Array

900604 Arrays have approximately 10K features on each array that can detect 10K SNPs using the Affymetrix GeneChip DNA Analysis System incorporating MIP technology (six pack)

900580 (96 pack)

To Order

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