



# Data Sheet

## Affymetrix® GeneChip® Human (Panels 1 & 2) 20K cSNP Kit

The Affymetrix GeneChip® Human (Panels 1 & 2) 20K cSNP Kit contains 20,000 amino acid-changing SNPs that are multiplexed in a single assay. The panel has been optimized to use the same protocol as all other application-specific and custom kits that employ the Molecular Inversion Probe (MIP) technology<sup>1,3</sup> from Affymetrix and is designed to work with the GeneChip® Scanner 3000 (with suitable Targeted Genotyping upgrade). It includes validated non-synonymous public SNPs that code for functional changes in more than 10,000 genes. It allows researchers to perform cost-effective studies for the direct detection of non-synonymous SNPs across the whole genome.

### Features and Benefits

- Since SNPs resulting in non-synonymous changes in proteins are the class of SNPs most likely to be disease causative<sup>4</sup>, large-scale association studies of complex diseases and traits using this panel result in a higher probability of a direct link to biological pathways and gene function.
- Largest panel of non-synonymous SNPs available for hypothesis-driven disease association studies.

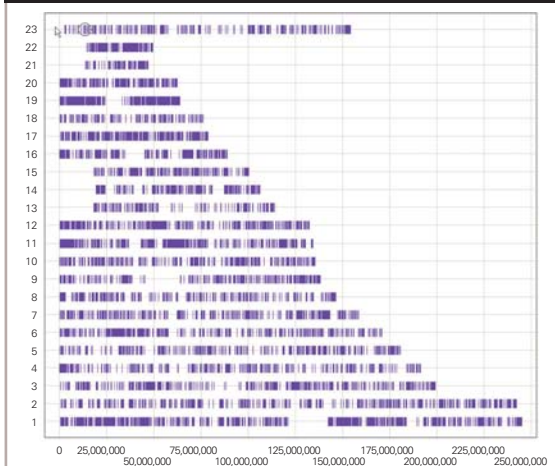
### Key Specifications

- Accuracy ≥ 99.25 percent
- Data completeness ≥ 98 percent
- Repeatability ≥ 99.25 percent
- Quantity of genomic DNA required without amplification 4.0 µg
- Throughput 48 samples per day (~1 million genotypes/day)
- Majority of SNP annotations from dbSNP build 124

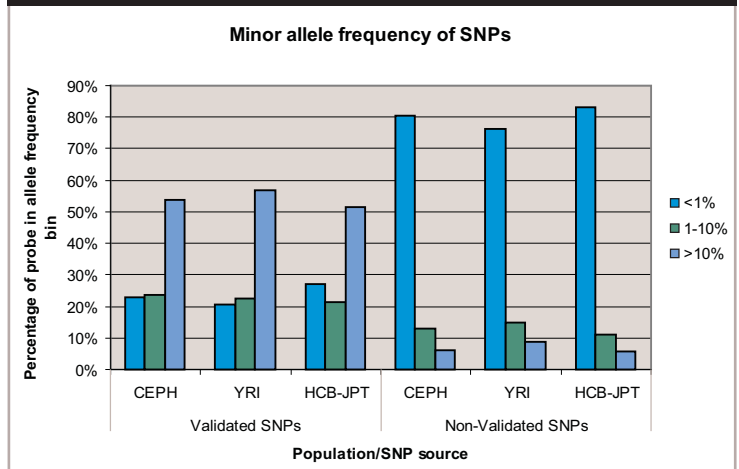
### Panel Design

Genotyping MIP probes were primarily designed using the SNP database, dbSNP124, containing ~60,000 non-synonymous SNPs. For the first panel of non-synonymous SNPs only validated, that is “double hit,” SNPs were selected for inclusion and these SNPs formed the basis for the Affymetrix GeneChip® Human (Panel 1) 10K cSNP Kit. Later, in order to expand this number, an additional ~23,000 SNPs were selected for possible inclusion. Validated SNPs were preferential; however, a large proportion of non-validated SNPs for which the reference human allele did not match the sequence of the chimp were also selected. (For human SNPs the allele found in chimps is typically assumed to be the ancestral allele and, hence, likely to be the major allele in humans.) Following the design and testing of probes to these SNPs in the molecular inversion probe assay, SNPs for which there was no evidence of

**Figure 1:** Distribution of 20K panel non-synonymous SNPs by chromosome.



**Figure 2:** Minor allele frequencies of 20K panel non-synonymous SNPs.



the minor allele (in any of the HapMap populations or in the chimp/bonobo/gorilla) were excluded. As a result of this process, an additional 10,000 non-synonymous SNPs were selected to create a panel of ~20,000 polymorphic SNPs.

The distribution of SNPs and minor allele frequency are shown in Figures 1 and 2. The panel has a median interval of less than 70 kb between SNPs and can be used in a single multiplexed reaction to genotype DNA samples.

#### THE USE OF NON-SYNONYMOUS SNPS IN ASSOCIATION STUDIES

Whole-genome association studies to identify loci involved in common disease have been advocated for years<sup>5</sup>. These studies

are particularly adaptable to the highly multiplexed genotyping technology that is afforded by the use of MIP technology. Approaches to detect genetic associations relying on linkage disequilibrium among SNPs can be complemented by direct assessment of SNPs that are most likely to have a functional consequence<sup>4</sup>. This product enables the task of large-scale characterization of non-synonymous SNPs, that is, the most obvious class of functional SNPs, in the study of human disease genetics. Using this panel during a validation study, a total of 533 DNA samples were genotyped—90 unique DNAs and 30 unique trios—which determined that the average full call repeatability for replicates was 99.86 percent and average full call trio concordance was 99.88 percent.

#### REFERENCES

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3. Jain, M., *et al.* Benchtop Genotyping. *Genetic Engineering News* **24**:18 (10/15/2004).
4. Botstein & Neil Risch, *et al.* Discovering Genotypes Underlying Human Phenotypes: Past Successes for Mendelian Disease, Future Approaches For Complex Disease. *Nature Genetics* **33**: 228-237 (2003).
5. Risch & Merikangas, *et al.* The Future of Genetic Studies of Complex Human Diseases. *Science* **273**:1516-1517 (1996).

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#### Ordering Information

Affymetrix GeneChip® Human  
(Panels 1 & 2) 20K cSNP Kit

**900827** *Kit contains enough reagents to process a total of 24 assays (including one control)*

Affymetrix GeneChip® Universal 25K  
Tag Array

**900857** *Arrays have approximately 25K features on each array that can detect 20K SNPs using the Affymetrix GeneChip® Scanner 3000 Targeted Genotyping System incorporating MIP technology*

**900858** *(96 pack)*

#### To Order

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