AFFYMETRIX®

Data Sheet

Affymetrix® GeneChip® Human Immune and Inflammation 9K SNP Kit

The Affymetrix GeneChip® Human Immune and Inflammation 9K SNP Kit contains over 9,000 SNPs in approximately 1,000 genes that have been implicated in, or are thought to be involved in, immune and inflammatory responses in humans. The 1,000 covered genes were selected in collaboration with leading researchers in the field. HapMap data from multiple populations were used to select SNPs to tag common (>5%) polymorphisms in these genes. In addition, the panel includes ~800 validated non-synonymous SNPs. The panel allows researchers engaged in studies of the immune system and inflammatory response to perform cost-effective genotyping. The Immune and Inflammation 9K SNP Kit is designed to work with the Affymetrix GeneChip® Scanner 3000 (with the Targeted Genotyping upgrade).

Features and Benefits

- A gene-focused approach to immune and inflammation studies has the potential to reduce genotyping and multiple testing costs.
- The use of tagging SNPs results in high genomic coverage for over 1,000 genes involved in immune and inflamation response.
- Tagging SNPs were selected using samples from multiple populations, resulting in a panel that is suitable for genotyping both Caucasian and non-Caucasian populations.
- The inclusion of potentially functional non-synonymous SNPs increases the possibility of directly detecting the causative variant.
- Significant associations can be linked directly to biological pathways and gene function.

Key Specifications

- Accuracy ≥99.25 percent
- Data Completeness ≥98 percent
- Repeatability ≥99.25 percent
- Quantity of genomic DNA required without amplification is 4.0 µg
- Throughput of 48 samples per day (~0.5 million genotypes/day)
- SNPs selected from HapMap #17. SNP and gene annotations from Ensembl build 34 (dbSNP build 124)

Panel Design

Using Molecular Inversion Probe (MIP) technology, Affymetrix has developed an application-specific SNP genotyping panel for studying the immune system and inflammatory response. The panel is designed to cover over 1,000 candidate genes related to immunity and inflammation that were selected in collaboration with leading researchers in the field. The panel is suitable for studies in multiple ethnic populations. These studies can exploit the MIP technology's ability to develop large multiplex panels in a single assay to conduct affordable, large-scale genetic studies in two important areas: identifying the genes responsible for autoimmune diseases such as lupus, multiple sclerosis and rheumatoid arthritis; and collecting information on individual susceptibility to infectious disease or patient response to immunization, for example, measuring the genetic basis of patient response to the flu vaccine.

The panel is designed to cover ~1,000 genes (coverage of certain selected GO categories is shown in Figure 1). Tagging SNPs were selected to result in an r^2 coverage of ≥0.8 for all HapMap SNPs with MAF greater than 5 percent. These tagging SNPs were selected based on genotypes in the HapMap CEPH and Yoruban samples and covered SNPs from 5 kb upstream and 5 kb downstream of the gene. Additionally, 773 non-synonymous (amino acid-changing) SNPs were selected in these genes from the 20K panel (see the Affymetrix GeneChip® Human 20K cSNP Kit). The

average number of all types of SNPs per gene is 10. Following design and manufacture of MIP probes, conversion to working assays was very high (>92 percent), resulting in a panel of approximately 9,200 working assays.

Panel Performance

In actual studies using this panel, 88 DNA samples from 76 unique individuals (CEPH Utah HapMap samples plus one anonymous sample) were genotyped for a total of 824,729 genotypes. Accuracy, measured using Mendelian inheritance across 25 trios, was 99.90 percent. In addition, reviewing the SNPs that overlapped with reference genotypes generated in the HapMap project, concordance was measured with these genotypes as 99.71 percent. Repeatability, measured across six different individuals who were each genotyped two to eight times, was 99.95 percent. Data completeness, the average call rate across passed samples for passed SNPs, was measured on genotypes across the panel at 99.57 percent.

Figure 1: Representation of certain Gene Ontology (GO) categories for the ~1,000 genes represented on the Affymetrix GeneChip® Human Immune and Inflammation 9K SNP Kit. Note that many genes appear in multiple GO categories.

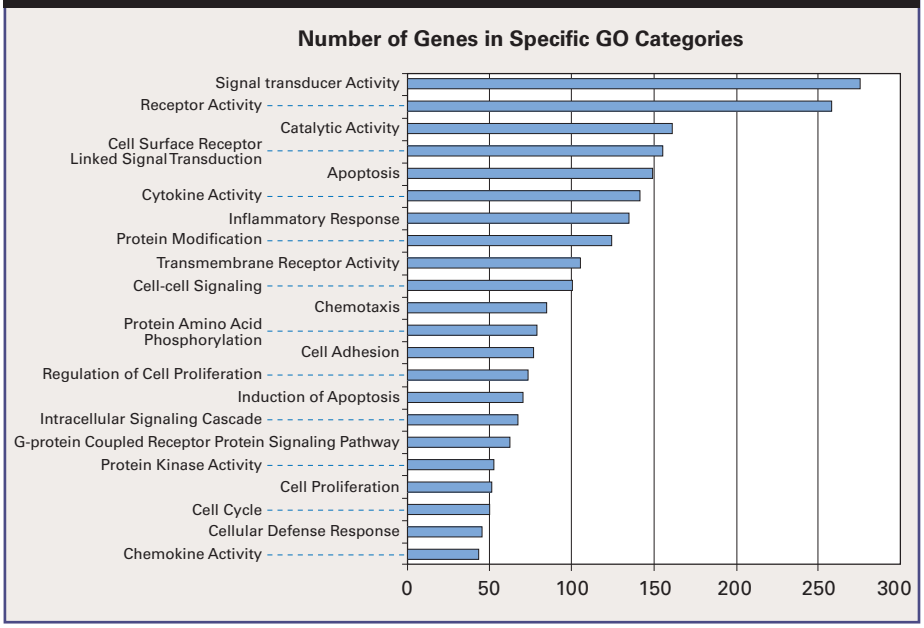
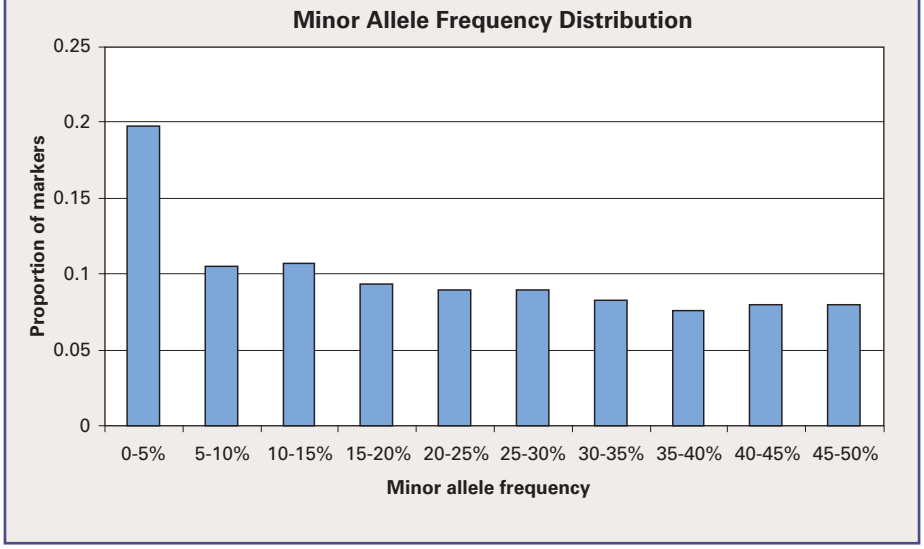


Figure 2: CEPH HapMap sample. Minor allele frequencies of the 9,200 SNPs that are present in the Affymetrix GeneChip® Human Immune and Inflammation 9K SNP Kit. SNPs with low minor allele frequencies in CEPH were either picked as tagging SNPs in the Yoruban HapMap samples (and hence were above 5% in that population) or are non-synonymous SNPs (the panel includes only nsSNPs validated as polymorphic in at least one HapMap population).



Notes:

Ordering Information

Affymetrix GeneChip® Human Immune and Inflammation 9K SNP Kit

900868	*Contains enough reagents to process a total of 24 samples (including one control)*

Affymetrix GeneChip® Universal 10K Tag Array

900604	*(6 pack) Arrays have approximately 10K features on each array that can detect 10K SNPs using the Affymetrix GeneChip® DNA Analysis System incorporating MIP technology*
900580	*(96 pack)*

To Order

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