The MegAllele™ Genotyping Human 10K cSNP Panel 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 MegAllele Genotyping Human 10K cSNP Panel 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 technology (MIP™) 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.
- Significant associations can be directly linked to biological pathways and gene function since functional SNPs (amino acid-changing) have a higher probability of being causative.

Key Specifications
- Accuracy = 99.5%
- Data Completeness = 98%
- Repeatability = 99.5%
- Quantity of genomic DNA required without amplification is 4.0 µg
- Quantity of genomic DNA required with recommended amplification is less than 50 ng
- Throughput of 48 assays per day (~ 0.5 million genotypes/day)

Panel Design
MegAllele products utilize ParAllele’s next-generation genotyping technology to comprehensively and accurately score thousands of targeted SNPs for each DNA sample in a single assay. The unique chemistry exploits the innovative Molecular Inversion Probe (MIP™) technology 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).

The MegAllele Genotyping Human 10K cSNP Panel I is the first in a line of assay panels that enables direct whole-genome detection studies using functional SNPs. 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 1 percent. Most of the major drug target gene families are represented in this new assay panel.

Coverage and Performance
The MegAllele Genotyping Human 10K cSNP Panel I has a median interval of <100kb 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 successfully used as targets for drug development. To determine the performance of this 10K cSNP Panel, DNA samples across three populations (Caucasian, Asian, African) were genotyped (total of 2,900,476).

Accuracy measured using Mendelian Inheritance across 60 trios was 99.87 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 12 different individuals with 2 replicates each was >99.9 percent.

Data completeness (the average call rate across samples for passed SNPs) was measured on genotypes across the panel at 98.86 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


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**Figure 2.**

**Human 10K cSNP Panel I**

<table>
<thead>
<tr>
<th>Family name</th>
<th>Proportion of genes with at least 1 SNP</th>
</tr>
</thead>
<tbody>
<tr>
<td>Serine/Threonine protein kinase</td>
<td>0.9</td>
</tr>
<tr>
<td>Rhodopsin-like superfamily</td>
<td>0.8</td>
</tr>
<tr>
<td>GPCR</td>
<td>0.7</td>
</tr>
<tr>
<td>Tyrosine protein kinase</td>
<td>0.6</td>
</tr>
<tr>
<td>Neurotransmitter-gated ion-channel superfamily</td>
<td>0.5</td>
</tr>
<tr>
<td>Serine protease, trypsin family</td>
<td>0.4</td>
</tr>
<tr>
<td>Serine specific phosphatase</td>
<td>0.3</td>
</tr>
<tr>
<td>ABC transporter, transmembrane region</td>
<td>0.2</td>
</tr>
<tr>
<td>All validated SNPs in dbSNP 120</td>
<td>0.1</td>
</tr>
</tbody>
</table>

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**Ordering Information**

**MegAllele™ Genotyping Human 10K cSNP Panel Kit**

- **900630** Kit contains enough reagents to process a total of 24 assays (including 2 controls)

**GeneChip® ParAllele TrueTag™ 10K-A Array**

- **900604** ParAllele TrueTag Arrays have approximately 10K features on each array that can detect 10K SNPs using the MegAllele™ Genotyping System. (6 pack)
- **900580** (96 pack)

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