GeneChip®
HuSNP™ Mapping Assay

Support Data User's Guide

This document describes the two Excel data files provided on the compact disc entitled HuSNP™ Mapping Assay Support Data. These data may be used to facilitate applications of the GeneChip® HuSNP™ Mapping Assay.

Table 1 lists the Proficiency Panel derived from the HuSNP™ Mapping Assay marker genotypes that were obtained for nine commercially available CEPH DNA samples. The genotypes reported were obtained reproducibly in the assay and in some cases confirmed by ABI sequencing. These data may be used as a reference during analysis of HuSNP™ mapping experiments.

Table 2 displays marker allele frequencies and heterozygosities derived from results generated during the development of the assay. These results will vary with the populations used so the data provided may not apply directly to the user's experience.

Proficiency Panel of Validated HuSNP™ Mapping Assay Genotypes in Reference DNA Samples

Table 1 displays SNP genotypes generated using the GeneChip® HuSNP™ Mapping Assay in quadruplicate for 9 CEPH DNA samples (1331-01, 1408-01, 1408-02, 1332-01, 0002-01, 0002-02, 0102-01, 0102-02, 884-02). CEPH DNA 884-02 is the reference DNA (P/N 900221) included with the HuSNP™ Mapping Assay kit.

The CEPH DNA samples are available from Coriell. These data are provided as a reference for evaluating laboratory performance of the HuSNP™ Mapping Assay. Genotypes are not shown for markers for which there were any discordant data. Where genotypes are shown, definitive calls were obtained for that marker in at least three of four experiments. Thus the absence of a genotype call is denoted by a "-" and indicates insufficient or discordant data for that marker among the replicates. Some of the GeneChip® probe array results were confirmed by dideoxy sequencing in collaboration with deCode Genetics and are indicated by bold type and blue shading in the table.

Table 1, Sheet 1 lists the markers by WIAF number in the first column. The second column, entitled "Pool," lists the PCR primer pool from which that marker was amplified. The third column entitled SS# refers to the NCBI SNP database "assay id #" for these markers (www.ncbi.nlm.nih.gov/SNP/).

The additional column headings correspond to the CEPH DNA samples alternating with an index for that sample. GeneChip® HuSNP™ Mapping Assay genotype calls that meet the criteria described above are shown by sample and marker. All of the markers interrogated in the HuSNP™ Mapping Assay have two alleles; arbitrarily named A and B. Genotypes concordant with DNA sequence data obtained in collaboration with deCode Genetics (see *HuSNP™ Mapping Assay Technical Note #1*, P/N 700318) are shown in bold and blue. The column adjacent to the genotypes for each sample and labeled "' Sample Name' Index" contains a simple code that indicates whether the respective genotype has been confirmed by ABI genotyping: 1-yes, 0-no (the code may facilitate sorting of the data in Excel). These data are provided for use as a reference with which users may evaluate their experience and facility with the HuSNP™ Mapping Assay. Sheet 2 displays the same data in a more convenient format for database import.

Table 1

Marker	Pool	SS#	1331-01	1331-01Index	1408-01	1408-01Index
WIAF-1488	A15	3174	**AB**	1	**AB**	1
WIAF-1489	A17	826	AB	0	AA	0
WIAF-1490	A05	3175	AB	0	AB	0
WIAF-1500	A15	3184	**AA**	1	AA	0

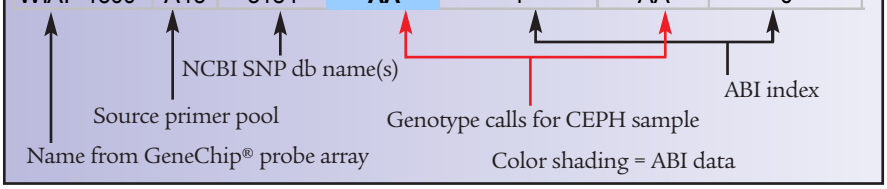

Table 2

Marker	Pool	SS#	#Definitive	A Freq	B Freq	Heterozygosity
WIAF-1000	A06	3099	93	39.78%	60.22%	47.91%
WIAF-1001	A02	3100	132	53.03%	46.97%	49.82%
WIAF-1002	A17	3101	102	54.90%	45.10%	49.52%
WIAF-1004	A06	3103	124	66.53%	33.47%	44.53%

Name from the GeneChip® probe array

Source primer pool

NCBI SNP db name(s)

A & B allele frequencies

Calculated heterozygosity
$H=1-(a^2+b^2)$

Allele Frequencies & Heterozygosities for the HuSNP™ Mapping Assay Marker Set

Using the GeneChip® HuSNP™ Mapping Assay, SNP genotypes were generated for DNA samples from 133 unrelated individuals. These samples derive predominantly from individuals of Western European descent but also include samples from ten African Americans and ten Asians. The definitive genotypes were used to calculate allele frequencies and heterozygosities that are displayed in Table 2. These values may vary considerably with samples derived from different populations.

Table 2 lists the markers by WIAF number in the first column. The second column, entitled "Pool," lists the PCR primer pool from which that marker was amplified. The third column entitled SS# refers to the NCBI SNP database "assay id #" for these markers (www.ncbi.nlm.nih.gov/SNP/). In the fourth column the number of definitive calls obtained from the 133 individuals is shown (i.e., the size of the data set available for allele frequency determination). The fifth and sixth columns contain the calculated frequencies of the A and B alleles respectively. The final column contains the calculated heterozygosity using the following equation: $H=1-(a^2+b^2)$, where a & b are the derived frequencies of the A & B alleles respectively. The median heterozygosity for these samples is 36%. The 25th percentile is 22.4% and the 75th percentile is 46.7%. Thus, 25% of the markers have relatively low heterozygosity, another 25% have nearly maximal heterozygosity, and the rest fall in the middle. (A biallelic marker cannot exceed 50% heterozygosity.)

Additional Information

GeneChip® probe array technology, products and services are marketed and sold internationally. For additional technical and pricing information about the HuSNP™ mapping assay and other GeneChip® probe array products, or to find the name and location of the distributor or sales office nearest you, please contact us at the following address:

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