DNA Test Panel

The DNA Test Panel from Illumina® is a cost-effective and efficient tool for pre-screening DNA sample quality using the proven GoldenGate® Assay prior to conducting studies with larger numbers of loci.

DNA SAMPLE VALIDATION AND SAMPLE TRACKING

The DNA Test Panel can be used to quickly and inexpensively prescreen DNA samples before conducting studies with large numbers of loci. The DNA Test Panel includes 360 highly validated single nucleotide polymorphism (SNP) assays distributed across the genome with all chromosomes represented, including both X and Y for gender verification.

In addition, the DNA Test Panel can be used for DNA sample tracking. A large proportion of loci on the DNA Test Panel are included in the Linkage IVb, Human-1 and HumanHap300 Genotyping BeadChips and will be placed on whole-genome genotyping products in development. The DNA Test Panel can be used to 'barcode' samples for use with other Illumina standard products, minimizing sample tracking errors.

HIGHLIGHTS OF THE DNA TEST PANEL

- Save Time and Money: assess DNA quality using a panel of highly validated SNP loci with the proven GoldenGate® Assay
- Reduce Errors: track samples using overlapping loci within other Illumina Genotyping products
- Use as Genomic Controls: neutral loci selected outside of RefSeq* genes and evolutionarily conserved sequences

*Reference Sequence (RefSeq) genes from National Center for Biotechnology Information (NCBI)

GUIDELINES FOR ASSESSING DNA QUALITY

The DNA Test Panel can be used to save time and money by removing low performing DNA samples before embarking on larger studies. The following guidelines are recommended for obtaining the most meaningful and consistent results. *Figure 1* shows genoplots illustrating genotype clustering in both low-performing (*black data points; top panel*) and high-performing (*bottom panel*) DNA samples using the DNA Test Panel.

1. Accurate DNA sample concentration (50ng/µl) and assay input amount (250ng) into the GoldenGate® assay is recommended. Lower input DNA sample mass may perform well when used with the DNA Test Panel, but performance may not carry over consistently to other SNP assays. Superior and consistent DNA sample performance is achieved at the recommended DNA concentrations and assay input amounts.
2. When clustering DNA samples, clustering differences between sample subgroups may be observed (i.e., by DNA preparation method or ethnicity). A minimum of eight samples is recommended for each DNA subgroup for visualizing clustering patterns with a larger sample collection. Whole-genome amplified (WGA) DNA samples may require separate clustering. In this case, a minimum of sixteen samples is recommended.
3. For each group of DNA samples being tested, use at least four control DNA samples that are known to perform well and include samples that are ethnically diverse and representative of both genders. Excellent choices are samples NA17017, NA17116, NA17018, NA07034 available from the Coriell Cell Repository[1].
4. During data analysis, low-performing DNA samples can be identified by plotting both call rates and the 10% GenCall score (the GenCall score at the 10% rank if the scores are ranked for all loci) for all DNA samples. One should visually inspect the distribution for the 10% GC score. Alternatively, one may multiply 0.85 by the 90th percentile of the 10% GC score to identify low-performing DNA samples.

CONTENT DESIGN

SNP assays were selected from Illumina's highly validated and robust Linkage IVb Panel and are distributed across the genome, including both the X and Y chromosomes for gender verification *(Table 1)*. All chromosomes are represented with an average 8Mb spacing between loci *(Table 2)*.

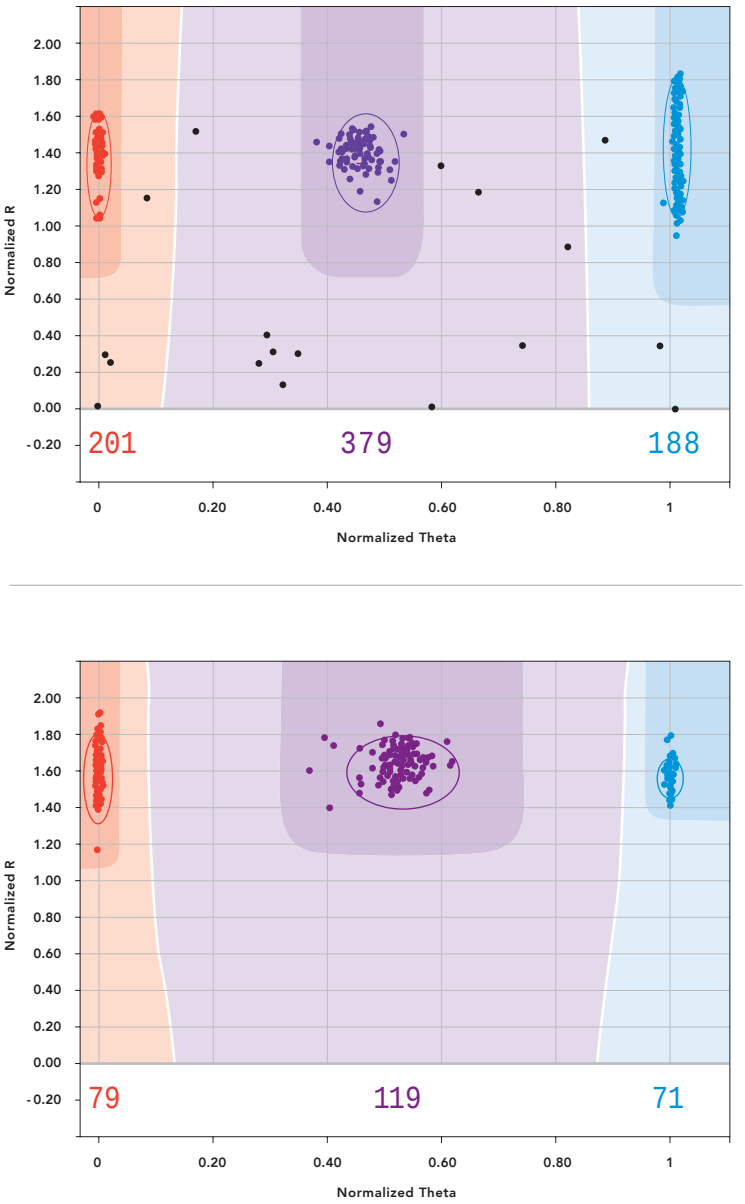
DNA Test Panel loci were designed as genomic controls having similar allele frequencies between disease and non-diseased populations and are not likely to be associated with disease. Assays for SNP loci were only included if they were located > 10kb from a RefSeq transcript or did not reside in an Evolutionarily Conserved Region (ECR). ECRs were defined from multiple species alignments of human, chimp, mouse, rat, dog, chicken, fungus and zebrafish. PHAST Conservation scores[2] and the respective genomic regions demonstrating conservation across species were obtained from the UCSC Genome Browser[3]. Assays for SNP loci were excluded from the DNA Test Panel if the SNP mapped to one of the phastCons elements with a PHAST score ≥ 50.

Pairwise allele frequency comparisons were carried out between the following populations to identify SNPs with the largest allele frequency differences:

- CEU and CHB+JPT
- CEU and YRI
- CHB+JPT and YRI

The identified SNPs, called ancestry informative markers (AIMs), may provide population stratification information in a study population.

FIGURE 1: EXAMPLE GENOTYPE CLUSTERING PATTERS USING THE DNA TEST PANEL



Two genoplots illustrating examples of genotype clustering containing some low-performing (*top panel*) and all high-performing (*bottom panel*) DNA samples using the DNA Test Panel.

ILLUMINA SOLUTIONS FOR GENOTYPING

The high-quality data and low cost per-genotype of the DNA Test Panel are made possible by powerful Illumina technologies that include the GoldenGate Assay with multi-sample Sentrix® Array Matrix and BeadChip formats. Illumina's genotyping solutions enable linkage and association mapping, as well as high-resolution, genome-wide scans. Whether using standard or custom content, Illumina genotyping panels can be accessed via Fast-Track Genotyping Services or with an Illumina System. Illumina solutions provide industry leading levels of accuracy, flexibility and affordability.

TABLE 1: DISTRIBUTION OF LOCI (*n*)

No. Loci	360
Autosomal	313
X Chromosome	40
Y Chromosome	7

TABLE 2: SPACING BETWEEN LOCI (MB)

Mean	7.9
Median	4.8
Minimum	0.01
Maximum	71.2

ORDERING INFORMATION

CATALOG NO.	PRODUCT	DESCRIPTION
GT-17-221	DNA Test Panel	One oligo pool (OPA) for 360 SNP loci. Sufficient for 96 DNA samples.
GT-95-201	Single-Use Activation Kit (576 Samples)	Used in combination with the GoldenGate Assay Kit. Contains reagents for six, 96-well plates.
GT-95-205	GoldenGate Assay Kit II with UDG (96 Samples)	Sufficient reagent for preparing genotyping reactions for 96 DNA samples. Contains UDG enzyme for contamination control.
FA-12-107	Sentrix Universal-96 Array Matrix	One Sentrix Universal-96 Array Matrix can process 96 samples and up to 384 assays/sample.

REFERENCES

(1) http://locus.umdnj.edu/ccr/
(2) PHylogenetic Analysis with Space/Time models (PHAST)
(3) http://genome.ucsc.edu/cgi-bin/hgGateway

ADDITIONAL INFORMATION

Contact us for more information about Illumina's DNA Test Panel or other Illumina products or services.

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