



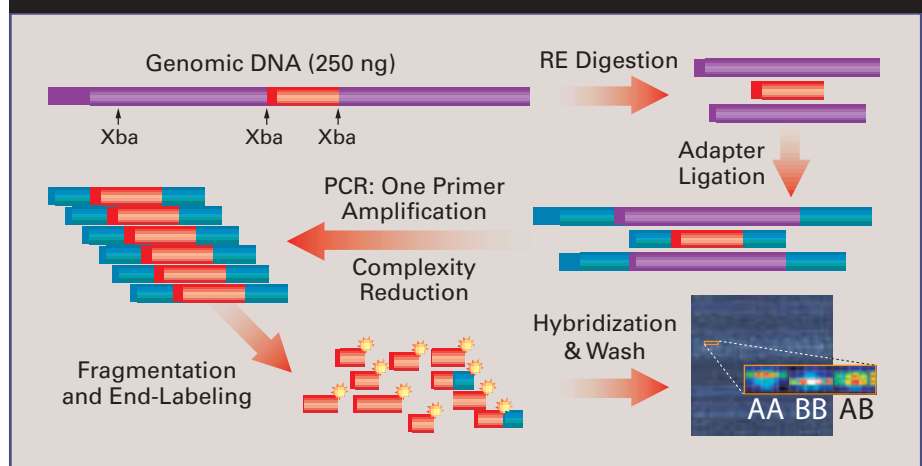
Data Sheet

GeneChip® Human Mapping 10K Array and Assay Kit

The GeneChip® Human Mapping 10K Array Xba 131 (GeneChip® Mapping 10K Array) is a new SNP genotyping tool for investigating the genetics of complex human disease.

More power, less PCR and less DNA:
10,000 SNPs, One Primer, One Array.

Figure 1: GeneChip® Mapping Assay Overview.



INNOVATIVE ASSAY ELIMINATES THE NEED FOR LOCUS-SPECIFIC PCR:

The GeneChip® Mapping Assay uses a simple approach for reducing complexity of the genome, allowing efficient genotyping with a microarray containing over 10,000 SNPs. The assay uses basic steps as outlined in Figure 1. Total genomic DNA (250ng) is digested with a restriction enzyme (XbaI) and ligated to adapters that recognize the cohesive four basepair (bp) overhangs. All fragments resulting from restriction enzyme digestion, regardless of size, are substrates for adapter ligation. A generic primer that recognizes the adapter sequence is used to amplify adapter ligated DNA fragments. PCR conditions have been optimized to preferentially amplify fragments in the 250 to 1,000 bp size range. The amplified DNA is then fragmented, labeled, and hybridized to the GeneChip Mapping 10K Array.

SNP SELECTION AND GENOME COVERAGE:

Approximately 11,500 SNPs from The SNP Consortium (TSC) database are tiled on a single array. Each SNP lies within one of the 250 to 1,000 base XbaI fragments amplified by the Mapping Assay. The median physical distance between SNPs is approximately 105 kb, and the average distance between SNPs is 210 kb (see Figure 2). The average heterozygosity of these SNPs is 0.37.

MORE POWER FROM 10,000 GENOTYPES:

Marker informativeness will vary from population to population and family to family. 11,500 SNPs ensure sufficient informative markers in your samples. Overall, this gives a significant increase in genetic power, which should enable higher experimental success rates, identification of additional loci, decreased linkage intervals, and higher LOD scores. Customers should expect call

rates of greater than 90%, giving in excess of 10,000 genotypes per sample. Internal validation studies have shown >95% call rates over more than 200 individuals from Caucasian, African American, and Asian populations.

USE LESS DNA STARTING MATERIAL:

The assay requires only 250 ng genomic DNA as starting material for the entire assay.

HIGHLY ACCURATE AND REPRODUCIBLE:

538 SNPs across 40 individuals were randomly selected; those genotype calls were compared to reference genotypes generated by single-base extension and capillary sequencing. Concordance with the reference data was >99.6% across >21,000 genotypes. Mendelian inheritance errors were measured at .05% over 30 CEPH trios. Reproducibility was 99.96% when measured over nine samples six times each.

AUTOMATED GENOTYPE CALLS IN SOFTWARE:

The GeneChip® DNA Analysis Software 2.0 (GDAS 2.0) uses an automated genotype calling algorithm that provides a quality score for each individual genotype. These calls can be easily exported for additional analysis, using third-party software. This algorithm was developed and validated as part of the entire Mapping 10K system.

SNP ANNOTATION AVAILABLE THROUGH THE NETAFFX™ ANALYSIS CENTER:

Extensive annotation for each SNP is provided both within GDAS 2.0 and on the NetAffx™ Analysis Center. Affymetrix combines data from multiple sources within the public domain to provide the data in a single place. Annotation includes TSC ID, db SNP ID, nearest microsatellite markers, nearest gene, physical map location, cyto-band, genetic map location, and allele frequencies in multiple populations.

ARRAY SPECIFICATIONS:

Each array has more than 500,000 features. A feature consists of more than 1 million copies of a 25 bp oligonucleotide probe of defined sequence, synthesized in parallel by photolithographic manufacturing. For each

Critical Specifications

DNA Required	250 ng per sample
Number of SNPs on Array	11,555
Call Rate	>90%
Reproducibility	99.96%
Concordance	>99.6%
Genotype Calling	Automated with quality score
SNP Annotation	NetAffx™ Analysis Center
PCR Primers	1 per sample
PCR Reactions	4 per sample
Capital Equipment	Standard Affymetrix® Instrument Platform

SNP we tile 40 different 25 bp oligonucleotides, each with a slight variation in perfect matches, mismatches, and flanking sequence around the SNP.

NEW APPLICATIONS BUILT FOR THE STANDARD GENECHIP® INSTRUMENT SYSTEM:

The GeneChip Mapping 10K Array runs on the standard GeneChip instrument system, including the new GeneChip Scanner 3000 and Fluidics Station 450. The GeneChip Scanner 3000 is designed to enable high-resolution scanning of future high-density SNP arrays.

REAGENT KIT VALIDATED AS PART OF THE MAPPING 10K SYSTEM

GeneChip Mapping 10K Assay Kit contains validated and qualified reagents for the most critical steps in the GeneChip Mapping Assay. This includes the PCR primer and adapter necessary to selectively amplify a portion of the human genome, reagents to fragment and label the PCR products, as well as several control reagents. This kit was developed and validated as part of the entire Mapping 10K system.

GeneChip® Mapping 10K Xba Assay Kit Components

Adapter, Xba	Two annealed oligonucleotides specific for ligation to the XbaI restriction site.
PCR Primer, Xba	PCR primer to amplify ligated genomic DNA
Reference Genomic DNA, 103	Human genomic DNA control, with consensus genotypes
GeneChip® Fragmentation Reagent	DNaseI enzyme, formulated to fragment purified PCR amplicons
10X Fragmentation Buffer	Buffer for fragmentation reaction
GeneChip® DNA Labeling Reagent	Proprietary biotin-labeled reagent for end labeling fragmented PCR amplicons
Terminal Deoxynucleotidyl Transferase	Enzyme used to end label fragmented PCR amplicons with the GeneChip® DNA Labeling Reagent
5X Terminal Deoxynucleotidyl Transferase Buffer	Buffer for labeling reaction
Oligonucleotide Control Reagent	Mixture of five biotin-labeled oligonucleotides, which hybridize to control regions (gridding and array controls) on the GeneChip® Mapping 10K Array

REFERENCE GENOMIC DNA SERVES AS A PROCESS CONTROL

Each GeneChip Mapping 10K Xba Assay Kit contains a sample of human genomic DNA to serve as a control for the entire process from DNA to data, as well as for troubleshooting. Customers can expect a call rate that is >90% when using this DNA in their lab, resulting in more than 10,000 genotypes. In addition, Affymetrix provides the consensus genotypes for this sample from 10 independent replicates.

PROPRIETARY LABELING REAGENT ALLOWS TWO HOUR END LABELING

GeneChip DNA Labeling Reagent is a proprietary biotin-labeling molecule which allows reliable and efficient DNA end labeling in two hours. The entire course of the development of the Mapping 10K System, including SNP selection and assay optimization was done using the GeneChip DNA Labeling Reagent.

REAGENT KIT PACKAGING DESIGNED TO MINIMIZE DNA CROSS CONTAMINATION

As with all genotyping applications, DNA cross contamination is a concern. It can lead to genotyping errors, and, therefore, a reduction in genetic power. The GeneChip

Mapping 10K Xba Assay Kit is subdivided into three boxes to support a recommended workflow designed to minimize the possibility of DNA contamination. In addition, the GDAS 2.0 software contains a report to help identify samples that may have otherwise undetected DNA cross contamination.

SAMPLE THROUGHPUT

With the standard instrument configuration of one scanner and two fluidics stations, one person can process 96 samples per week from DNA to data. This modular system can easily be expanded to accommodate high-throughput needs to allow processing of tens of thousands of samples each year.

Additional reagents not supplied in the kit.

The GeneChip Mapping 10K Assay Kit supplies the most critical reagents needed for successful completion of the Mapping Assay. Several additional and widely available reagents are needed for completion of the assay, including XbaI Restriction Enzyme and Buffer, T4 DNA Ligase and Buffer, dNTPs and AmpliTaq Gold for PCR, and reagents for cleanup of the PCR products.

Ordering Information

GeneChip® Human Mapping 10K Array and Assay Kit

GeneChip® Human Mapping 10K Array Xba 131

900446 Contains 30 GeneChip® Mapping 10K Arrays

GeneChip® Mapping 10K Xba Assay Kit

900441 Sufficient for 30 reactions

GeneChip® Human Mapping 10K Array and Assay Kit Set

900459 Contains 30 GeneChip® Human Mapping 10K Arrays and 1 Mapping 10K Xba Assay Kit

Supporting Products

GeneChip® Operating Software (GCOS)

690031 GCOS is the core operating software for the GeneChip system providing instrument control, image analysis, workflow/ sample management, and expression analysis

GeneChip® DNA Analysis Software (GDAS)

690030 GDAS provides data analysis for Affymetrix GeneChip DNA arrays, including automated SNP and sequence analysis with quality scores

To Order

North America

888-DNA-CHIP 888-362-2447

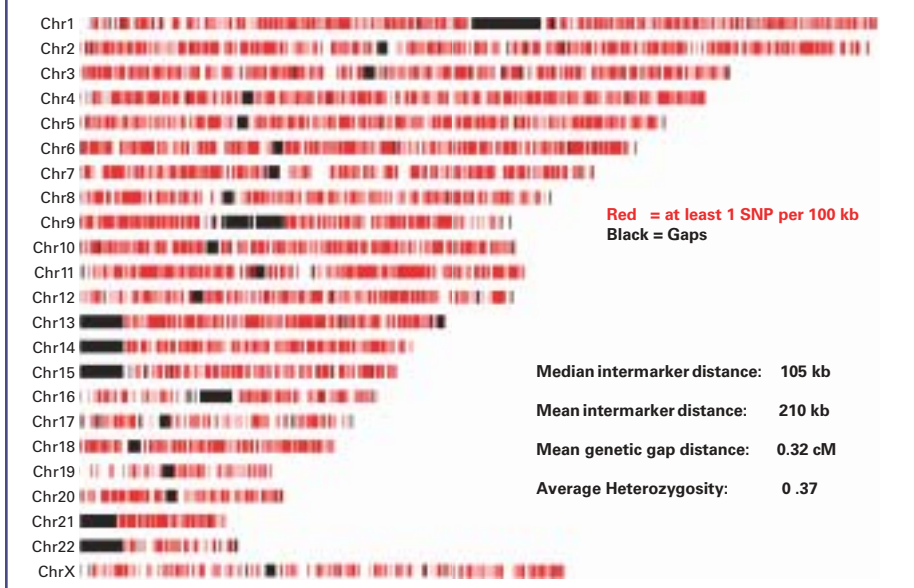
Europe

+44 (0) 1628 552550

Japan

+81-(0)3-5730-8200

Figure 2: Genome Coverage of Mapping 10K SNPs by Chromosome. Black areas represent gaps in the human genome sequence, primarily centromeres and telomeres.



AFFYMETRIX, INC.

3380 Central Expressway
Santa Clara, CA 95051 USA
Tel: 1-888-DNA-CHIP (1-888-362-2447)
Fax: 1-408-731-5441
sales@affymetrix.com
support@affymetrix.com

www.affymetrix.com

AFFYMETRIX UK Ltd




Voyager, Mercury Park,
Wycombe Lane, Wooburn Green,
High Wycombe HP10 0HH
United Kingdom
Tel: +44 (0) 1628 552550
Fax: +44 (0) 1628 552585
saleseurope@affymetrix.com
supporteurope@affymetrix.com

AFFYMETRIX JAPAN K.K.

Mita NN Bldg., 16 F
4-1-23 Shiba, Minato-ku,
Tokyo 108-0014 Japan
Tel: +81-(0)3-5730-8200
Fax: +81-(0)3-5730-8201
salesjapan@affymetrix.com
supportjapan@affymetrix.com

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