

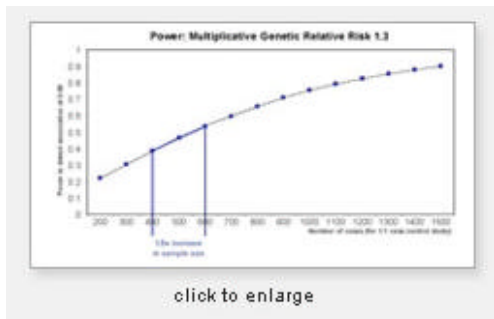
## Introducing GenomeWide SNP Array 6.0

### Pure performance & Genetic Power

[http://www.affymetrix.com/products/application/genome\\_wide\\_snp\\_6\\_ad.affx](http://www.affymetrix.com/products/application/genome_wide_snp_6_ad.affx)

The new Affymetrix® Genome-Wide Human SNP Array 6.0 features more than **1.8 million markers** for genetic variation, including more than 906,600 single nucleotide polymorphisms (SNPs) and more than 946,000 probes for the detection of copy number variation.

The most important study design factor affecting genetic power is the number of cases and controls for each phenotype (or sub-phenotype) under study. The number of samples processed is generally dictated by budget and cost effectiveness of the genotyping platform. **Affymetrix provides the greatest genetic power per dollar for your whole-genome association study by increasing the number of samples you can screen during your initial scan and replication phase.**



Another important study design factor is the quality of the data generated by the study. Random errors dilute the statistical signals or produce false positive associations. The SNP Array 6.0 demonstrates industry-leading performance, with **average call rates greater than 99 percent**. Average HapMap concordance exceeds 99.7%.

### A Whole-Genome Approach to Replication

The SNP Array 6.0 drives genetics forward by enabling a new study paradigm for researchers—a whole-genome approach to replication. This new approach increases the overall statistical genetic power to detect associations, and eliminates other disadvantages tied to selecting a limited subset of markers in the replication phase.

Traditional Study Design	Whole-genome Approach
Single Analysis Method	Meta Analysis from Multiple Data Sets
Lower-powered Scan	Maximally Powered Scan
Single Disease Hypothesis	Multiple Hypotheses
Delays in Workflow	Streamlined Workflow

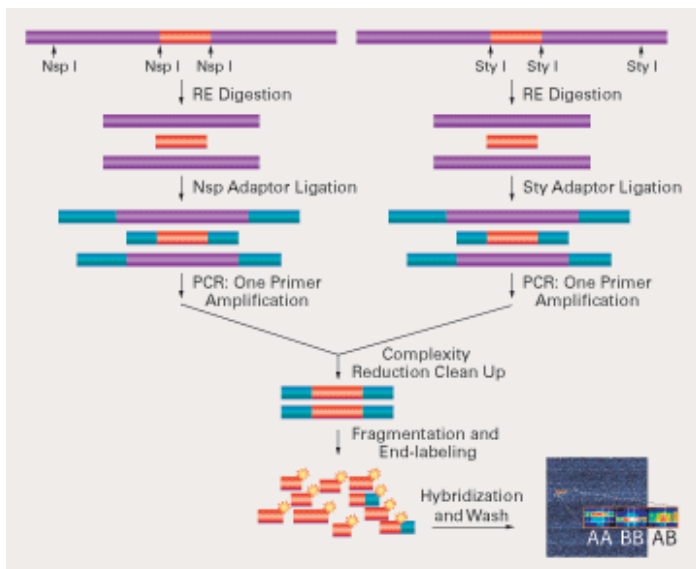
## Quick Facts

More than 906,600 single nucleotide polymorphisms

- Unbiased selection of 482,000 SNPs; historical SNPs from the SNP Array 5.0
- Selection of additional 424,000 SNPs
  - Tag SNPs
  - SNPs from chromosomes X and Y
  - Mitochondrial SNPs
  - New SNPs added to the dbSNP database
  - SNPs in recombination hotspots

More than 946,000 copy number probes

- 202,000 probes targeting 5,677 known CNV regions from the Toronto Database of Genomic Variants
- Regions resolve into 3,182 distinct, non-overlapping segments; on average 61 probes per region
- 744,000 probes, evenly spaced along the genome



Affymetrix Genome-Wide Human SNP Nsp/Sty Assay workflow

## New! Copy Number and LOH Analysis

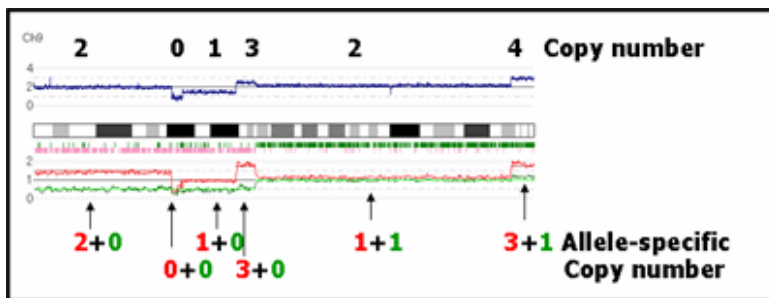
### Copy Number Analysis

High-resolution copy number and genotype information on a single proven platform for genetic analysis.

Affymetrix provides a portfolio of products for genetic research enabling scientists to analyze copy number, genotype, gene expression, and splice variant analysis on a single industry standard microarray platform.

#### **Copy number and allele-specific information enable detection of copy neutral events**

Researchers are finding that chromosomal abnormalities that do not alter copy number play an important role in diseases such as cancer and schizophrenia (Seal, et al., 2006). Genotype information in addition to the quantitative copy number information from a single assay allows researchers to identify uniparental disomy events and distinguish between different genetic mechanisms of loss of heterozygosity (LOH). Non-SNP arrays will only give copy number information, so you may be missing out on key determinants of your phenotype.



**Figure 1:** Genotype information in addition to the quantitative copy number information from a single assay allows researchers to identify uniparental disomy events and distinguish between different genetic mechanisms of loss of heterozygosity (LOH).

### **Applications for Copy Number Analysis**

- **Cancer** – Somatic changes in chromosomal copy number are hallmarks of tumor initiation and progression. Whole-genome copy number analysis identifies regions of amplification or deletion indicating the location of novel tumor suppressor and/or oncogenes.
- **Cytogenetics** – Historically, cytogenetics has focused on the detection of large regions of chromosomal loss or gain found in disorders such as Down Syndrome. Advances in SNP array technology enable the detection of smaller abnormalities that could not be detected by standard methodologies. Today, scientists are able to identify genetic causes in cases of mental retardation and schizophrenia that were previously unresolved by traditional testing
- **Association Studies** – Germline copy number polymorphisms are common in the general population and have been associated with specific diseases. Researchers now recognize the value of assessing copy number information in addition to SNP genotype data in association studies.