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# Making SNP decisions

## The latest tools and technologies for SNP analysis

The study of SNPs (single nucleotide polymorphisms) is invaluable for biomedical research, as these small variations in DNA sequence can have a major impact on response to disease, environmental factors and therapies. Within the last 20 years, biomedical researchers have developed increasingly sophisticated techniques for detecting and analysing SNPs in the human genome. Here we take a look at some of the latest technologies introduced to the field.

### Microarrays and panels

Researchers frequently need to link common structural variation with a diverse range of phenotypes. The new **Infinium® HD Human660W-Quad** four-sample BeadChip from **Illumina** featuring 2.6 million genetic markers provides the ideal solution. The multi-sample format of the array dramatically increases sample throughput and reduces handling time in the lab, while industry-leading data quality reduces errors and data mining tasks. In addition to this, the HD BeadChips require 70% less DNA input per sample. “Recent studies confirm that copy number variation plays a vital role in the development of complex human diseases. The base content of the Human660W-Quad BeadChip has been successfully used in more than 30 studies linking various SNPs to a wide range of diseases and phenotypes,” comments Joel McComb, General Manager of Illumina’s Life Sciences Business Unit. The Infinium HD Human660W-Quad BeadChip builds on the content from Illumina’s HumanHap550+ BeadChip to target more than 5,000 regions in the human genome known to be associated with copy number variation (CNV).

**Affymetrix’ Genome-Wide Human SNP Array 6.0** allows researchers to perform powerful whole-genome association and copy number studies by genotyping more markers across more individuals at a lower cost per sample than previously possible. These higher-powered studies increase the probability of discovering genes associated with adverse drug response or complex diseases. Kevin King, president of Affymetrix, says: “With the SNP Array 6.0, researchers are now able to analyse more genetic variation on a single array than with any other product, providing maximum genetic power, all at a much more affordable cost per sample.” The products continue to accelerate genetic research and enable scientists to develop

diagnostics and tailor treatments for individual patients by identifying and measuring the genetic information associated with complex diseases.

SNPs and other gene variants associated with a variety of health conditions, including insulin sensitivity, inflammation, obesity, Alzheimer’s disease and hepatitis C, can be detected with the specific multiplexes in **Marligen Biosciences’ Signet™ Genotyping Panels**. These multiplexed assays are formatted on the xMAP® bead array and are compatible with all Luminex™ analysers. This high-throughput format allows hundreds of samples to be analysed in one day.

### Genotyping systems

**Fluidigm’s new EP1 system** with Fluidigm Dynamic Arrays offers superior data quality, a fast and easy workflow, and significant cost savings for SNP genotyping studies. The EP1 system delivers the highest quality SNP genotyping results on the market with better than 99% call rates and 99.75% or greater accuracy. All of this is achieved with an easy-to-use, high-throughput workflow that can provide up to 9,216 data points per integrated fluidic circuit (IFC) chip with results in just four hours. The EP1 system, which includes the IFC Controller, Stand Alone Thermal Cycler and End Point Reader, provides more than 27,000 genotypes a day. “Fluidigm’s EP1 system provides the power of IFC technology, the system flexibility to provide true, high-throughput genotyping, the ability to perform digital PCR and a cost-effective system price,” says Gajus Worthington, Fluidigm President and Chief Executive Officer. “Customers will benefit from the lowest running costs for TaqMan-based SNP genotyping because of the tiny reagent volumes required by integrated fluidic circuit technology and they can complete their work – from start to finish – in less than four hours.”

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The EP1 system from Fluidigm

Applied Biosystems has commercialised a new analysis system to enable researchers to perform high-sample-throughput genotyping studies that are faster, easier and more cost effective than other commercially available methods. The **TaqMan® OpenArray™ Genotyping System** provides PCR-based SNP analysis of thousands of samples and candidate SNPs. The system provides life scientists with the ease-of-use, accuracy, and reproducibility of Applied Biosystems' TaqMan SNP Genotyping Assays and TaqMan DME Assays in a flexible format. This system expands the potential uses of TaqMan technology across a wide range of genotyping applications, providing an extremely fast, high-sample-throughput validation and screening tool for researchers in agricultural, pharmaceutical, and other commercial industries, as well as academic institutions. Studies that associate genotypes with complex diseases, ethnic ancestry, drug-treatment response, and traits for breeding and quality control in agricultural studies are among the types of studies that should benefit from this system.

The **JMP Genomics** desktop genomics package from SAS includes capabilities for candidate gene and whole-genome SNP analysis as well as streamlined analysis workflows for expression, exon, copy number and genotype data. The package provides researchers with several options for SNP assessment and analysis, including filtering of markers and individuals by various criteria, case-control analysis, PCA for population stratification (an implementation of the EIGENSTRAT method) and sophisticated linear modelling options. Building on the powerful analytic and visualisation capabilities of the SAS and JMP platforms, JMP Genomics also simplifies integration with partner technologies. Users can easily import from Illumina BeadStudio output formats, or import Affymetrix SNP CHP files using an interactive design wizard. JMP Genomics 3.2 has been optimised to handle whole-genome analysis for data sets as large as one million SNPs for 10,000 subjects.

#### Accessories

**Qiagen's** new **Type-it** product line for PCR-based genotyping experiments is available for genotyping of SNPs, microsatellites and



Marligen's Signet™ Genotyping Panels

mutations. The **Type-it Microsatellite PCR Kit** is a multiplex PCR-based kit for reliable detection of microsatellite loci in humans, animals, plants and bacteria using any detection platform. It allows high-yield co-amplification of all fragments without the need for optimisation of PCR parameters and can be used in challenging settings such as capillary sequencers. The **Type-it Mutation Detect PCR Kit** detects mutations such as deletions and translocations or for pre-amplification of SNPs using multiplex PCR technology. The **Type-it Fast SNP Probe PCR Kit** enables accurate SNP genotyping using 5'-nuclease assays. The kit ensures high call rates even when targeting difficult SNP loci or when using low template amounts.

Screening DNA from groups of disease cases and disease-free controls is the most common method to identify genetic markers of disease and to identify novel gene targets for treatment. **BioServe** has developed **SNPlates™** to help researchers cost-effectively validate these markers. SNPlates are pre-aliquotted 180:180 data-rich patient samples to perform case-control studies. These case control studies on a plate were designed by genetic epidemiologists to identify and validate genetic markers of disease and to validate previously identified genetic markers in specific disease states. SNPlates for diabetes, obesity, and breast, colon, lung and prostate cancer are now available, and will significantly reduce both the cost and complexity of how disease case-control studies are currently conducted.

#### SNP Creation

The **GENESIS™** gene-engineering platform from **Horizon Discovery**, which recently received a Medical Futures Innovation Award (MFLA), enables rapid and precise engineering of specific genetic variations, such as SNPs or disease-causing point-mutations, into any endogenous gene loci of normal or cancerous human cells. Horizon has used the GENESIS platform to develop more than 100 accurate disease models of human lung, breast, colorectal and other forms of cancer, called **X-MAN™** (Mutant And Normal) cell lines. These X-MAN cell lines are being deployed by global pharmaceutical companies to speed up and rationalise every step of the drug discovery process.

“Recent studies confirm that copy number variation plays a vital role in the development of complex human diseases.”

Joel McComb, General Manager,  
Life Sciences Business Unit, Illumina

Companies mentioned in this Product Focus:

**Affymetrix** – [www.affymetrix.com](http://www.affymetrix.com)  
**Applied Biosystems** – [www.appliedbiosystems.com](http://www.appliedbiosystems.com)  
**BioServe** – [www.bioserve.com](http://www.bioserve.com)  
**Fluidigm** – [www.fluidigm.com](http://www.fluidigm.com)  
**Horizon Discovery** – [www.horizondiscovery.com](http://www.horizondiscovery.com)  
**Illumina** – [www.illumina.com](http://www.illumina.com)  
**JMP (SAS)** – [www.jmp.com/genomics](http://www.jmp.com/genomics)  
**Marligen Biosciences** – [www.marligen.com](http://www.marligen.com)  
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