

# MassARRAY<sup>®</sup>

## iPLEX<sup>®</sup> Gold – SNP Genotyping

From target discovery to  
HTP validating



SEQUENOM<sup>®</sup>

# MassARRAY®

## iPLEX® Reagents – SNP Genotyping

### Overview

#### Unprecedented Levels of Accuracy

iPLEX® Reagents is the leading technology for SNP Genotyping. The MassARRAY system is widely used for fine mapping, linkage studies and routine genetic testing of SNP panels of interest.

MassARRAY combines the benefits of a simple, reproducible primer extension reaction chemistry with state of the art MALDI-TOF mass spectrometry to quickly and cost effectively characterize genotypes with the highest levels of accuracy.

The iPLEX assay allows you to routinely design assays at a multiplexing level of 36-plex, which gives a high level of flexibility and a low cost per genotype.

### The iPLEX Assay

#### Scalable Throughput

##### FROM MEDIUM TO HIGH THROUGHPUT

- Routine multiplexing at 36-plex level
- Broad throughput ranges from several hundred to 100,000 genotypes per day
- Ideally suited for fine mapping, GWAS and microarray follow-up studies

#### Accurate

##### HIGHEST AVAILABLE DATA QUALITY FOR YOUR RESEARCH

- Greater than 99.7% accuracy\*
- Direct mass detection of the molecule of interest – unexpected events such as failed PCR or tri-allelic SNPs can easily be discovered
- Highly reproducible

#### Cost Effective

##### LOWEST COST FOR ALL OF YOUR STUDIES

- Low cost per genotype even with low number of SNPs
- No fixed format. Load flexible SpectroCHIP with up to 384 samples with one multiplex iPLEX assay – or up to 384 different multiplexed assays with one sample
- New small kit formats available for the Analyzer 96 and 384 systems

#### Simple Workflow

##### RAPID TURN AROUND TIME – FROM ASSAY DESIGN TO RESULT

- Rapid and highly efficient assay design – working assays can be successfully designed for >95% of all confirmed SNPs
- Single termination mix and universal reaction conditions for all SNPs
- Convenient automated data analysis and reporting for unambiguous genotyping results

### Testimonial



“Sequenom’s iPLEX assay is a powerful tool for evaluating targeted sets of SNPs in a cost effective manner. Due to an efficient multiplexed assay design coupled with a robust chemistry, it is ideally suited for fine mapping and other genotyping studies using tens to hundreds of SNPs over hundreds to thousands of samples.”

#### Stacey Gabriel, Ph.D.

Director of the Genetic Analysis Platform  
Broad Institute at Harvard University and  
Massachusetts Institute of Technology

\*Gabriel, S.B., et al., *The Structure Of Haplotype Blocks in the Human Genome*. Science, 2002. 296(5576): p.2225-9.

## Success from Assay Design to Result

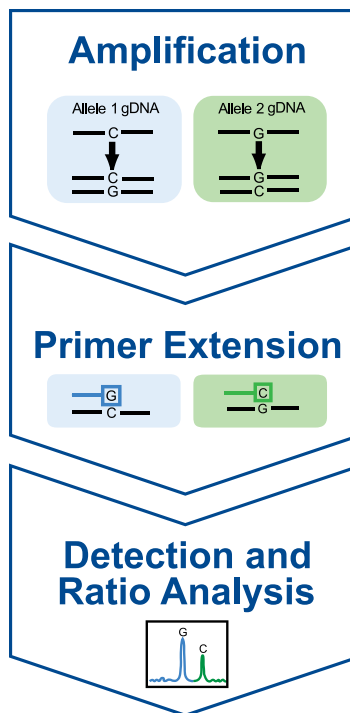
### How it Works

**Assay Design** - The MassARRAY® Designer software automatically designs both PCR and iPLEX single base extension primers for multiplexed assays. Sequence information can conveniently be imported from public databases and the primer design sequence will be automatically transferred into an oligonucleotide requisition form for efficient and rapid primer ordering. The software has a proven design efficiency of > 95% with a throughput of more than 10,000 assays per day.

**iPLEX Reaction** - iPLEX assay is a primer extension process designed to detect sequence differences at the single nucleotide level. The iPLEX assay uses a single termination mix and universal reaction conditions for all SNPs. The primer is extended, dependent upon the template sequence, resulting in an allele-specific difference in mass between extension products. This mass difference allows the data analysis software to differentiate between SNP alleles. Two kits are now available - iPLEX Gold for routine genotyping, and iPLEX Pro for more demanding applications that require high performance and sensitivity such as somatic mutations.

**Instrumentation** - Several system options are available for moderate to high throughput genetic analysis. Choose from a 96 or 384 well analyzer system.

**Genotype Calling and Results** - The SpectroCHIP® arrays are placed into the MALDI-TOF mass spectrometer and the mass correlating genotype is determined in real time. A SpectroCHIP® is typically processed in 45-60 minutes. The results are automatically loaded into a database that allows convenient data analysis.



### Ideal for Analyzing Tens to Hundreds of SNPs in Hundreds to Thousands of Samples

- Scalable and cost effective for most SNP genotyping studies
- Simple workflow with convenient, universal reaction conditions
- Multiplexed assay design with efficient re-plexing function
- Rapid turn-around time from primer design to results
- Sensitivity and data accuracy through user friendly software

# MassARRAY® SNP Genotyping

## CHIP SUMMARY

- Overview of genotypes for 1 Chip

## PROJECT EXPLORER

- Allows you to select single or multiple chips or assays to obtain an overview of genotypes

## TRAFFIC LIGHT

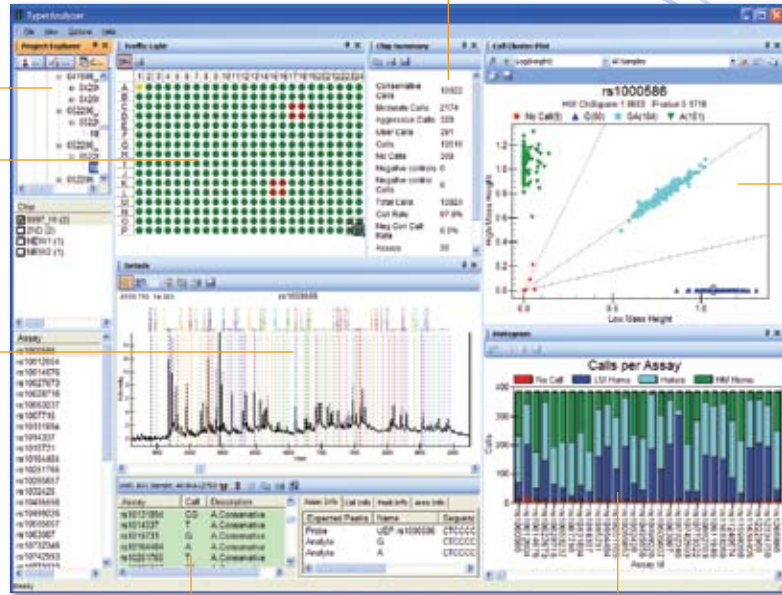
- Quick assessment tool for the percent successful call rate of an assay per well
- Four user-definable colors describe the percent of calls per well

## SPECTRUM

- Shows analyte signals, genotypes and mass range
- Annotations for all peaks
- Rough judgment of intensities, resolution, and signal-to-noise ratio

## WELL DATA

- Detailed information on and interaction with processed data stored in TYPER database
- Ability to manually interact with data.
- Well data tab allows the user to look at one particular well at a time for genotype call(s) and the confidence calculations of the call(s)
- Plate data tab gives information for the entire plate which can be sorted by the different headers. All data is contained here, such as area, resolution and primer primer peak scores



## HISTOGRAM

- Four categories: no calls, low mass homozygous, heterozygous, or high mass homozygous
- Quick analysis tool of the calls per assay over all samples within the plate
- Quickly monitor assay stability

## CLUSTER PLOT

- Plot of low mass allele vs. high mass allele for chosen assay and data type over all unique samples
- Hardy-Weinberg values calculated for each genotyped population per assay
- Click on any data point in cluster plot to review spectra and determine quality of individual assay
- Provides whole population assessment of assay behavior and quality

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## Genetic Analysis System

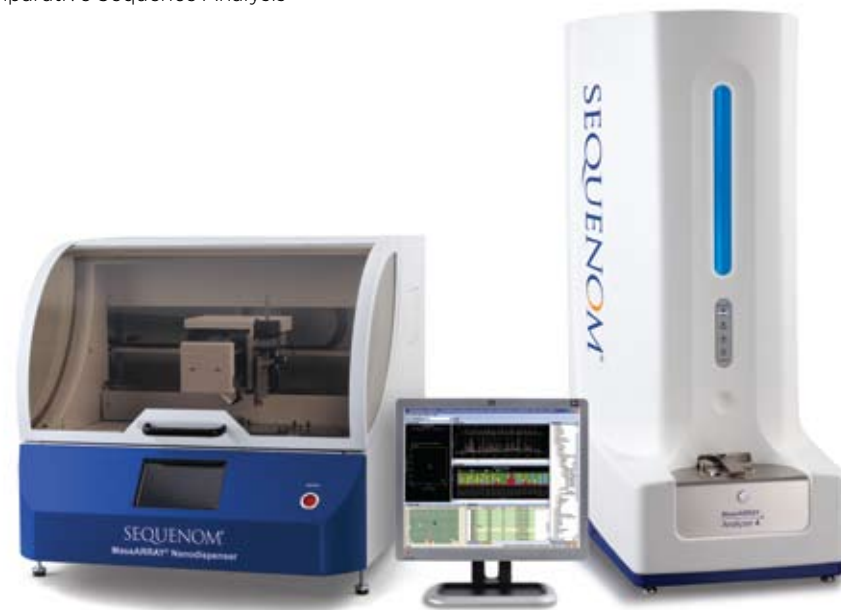
### MassARRAY<sup>®</sup> Analyzer 4 System<sup>®</sup>

The MassARRAY technology is trusted by the leading genetics institutions worldwide. The bench top MassARRAY Compact System is a multi-application platform that addresses the following applications:

- Quantitative Methylation Analysis
- SNP Genotyping
- Somatic Mutations
- Quantitative Gene Expression
- Comparative Sequence Analysis

### MassARRAY<sup>®</sup> Advantage

MassARRAY Genotyping facilitates identification and prioritization of genetic targets within each stage of biomedical research. From targeted discovery utilizing 10s to 100s of multiplexed assays to validation of select markers against 100s to 1000s of samples, the MassARRAY system powers a variety of genomic studies.



**Flexibility of Scale with  
Versatility of Application**

ATGATGATCGAAGCCGATGATCGACCAGTATGTATCATGATGATCGAAGC  
ATGATCGAAGCCGATGATGATGATCGAAGTACATGATGATCGAAGCCGATGATG  
TCGAAGCCGATGATCGACCAGTATGCGCATGAATGATCGAAGCCGATGATCGACCAGT  
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GTATCATGATGATCGAAGCCGATGATCGACCAGTATGGCGCATTATGCGCGCATGATGATCGAAGTATCATGATGATCGA



## Publications

Zheng, S.L. et al. (2008)

"Cumulative association of five genetic variants with prostate cancer."  
N Eng J Med 358 (9): 910-919.

Söderlund-Strand A., et al. (2008)

"High-throughput genotyping of oncogenic human papilloma viruses with MALDI-TOF mass spectrometry"  
Clin Chem. 54 (1): 86-92.

Sladek, R. et al. (2007)

"A genome-wide association study identifies novel risk loci for type 2 diabetes."  
Nature 445 (7130): 881-5.

Rioux J.D. et al. (2007)

"Genome-wide association study identifies new susceptibility loci for Crohn disease and implicates autophagy in disease pathogenesis."  
Nat Genet 39 (5): 596-604.

Cox, A. et al. (2007)

"A common coding variant in CASP8 is associated with breast cancer risk."  
Nat Genet 39 (3): 352-8.

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