

MassARRAY® iPLEX® Gold – SNP Genotyping



Finest in Fine Mapping

SEQUENOM®
www.sequenom.com

CCGATGATCGACCAGTATGCGCATGATGATCGAAGTATGCGCATTATGCGCAT
GCGCATTATGCGCATGATGATCGAAGCCGATGATCGACCAGTATGCGCATGA
GCGCATTATGCGCATGATGATCGAAGTATCATGATGATCGAAGCCGATGA

Overview

Unprecedented Levels of Accuracy

iPLEX® Gold 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 new iPLEX® Gold 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.

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 of the 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.

The New iPLEX® Gold Assay

Scalable Throughput

FROM MEDIUM TO HIGH THROUGHPUT

- Routine multiplexing at 36-plex level (PCR and MassEXTEND®)
- Broad throughput ranges from few hundred to 100,000 genotypes per day
- Ideally suited for fine mapping 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® Gold assay – or up to 384 different multiplexed assays with one sample
- 3.5¢ per genotype in a 36-plex reaction

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



■ Success from Assay Design to Result

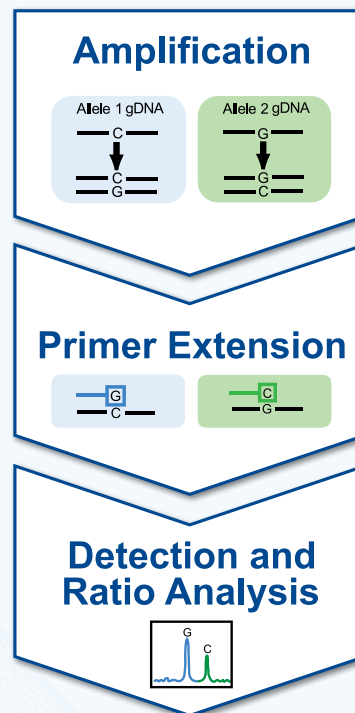
How it Works

Assay Design – The MassARRAY® Designer software can automatically design both PCR and MassEXTEND® 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. This software has a proven design efficiency of over 95% with a throughput of more than 10,000 assays per day.

MassEXTEND Reaction – MassEXTEND® is a primer extension process designed to detect sequence differences at the single nucleotide level. The iPLEX® Gold 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.

Instrumentation – Several system options are available for different throughput requirements. SEQUENOM developed the MassARRAY® system and its SpectroCHIP® arrays to specifically meet the requirements of moderate to high throughput genetic analysis.

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 384 position 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 sophisticated, yet 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

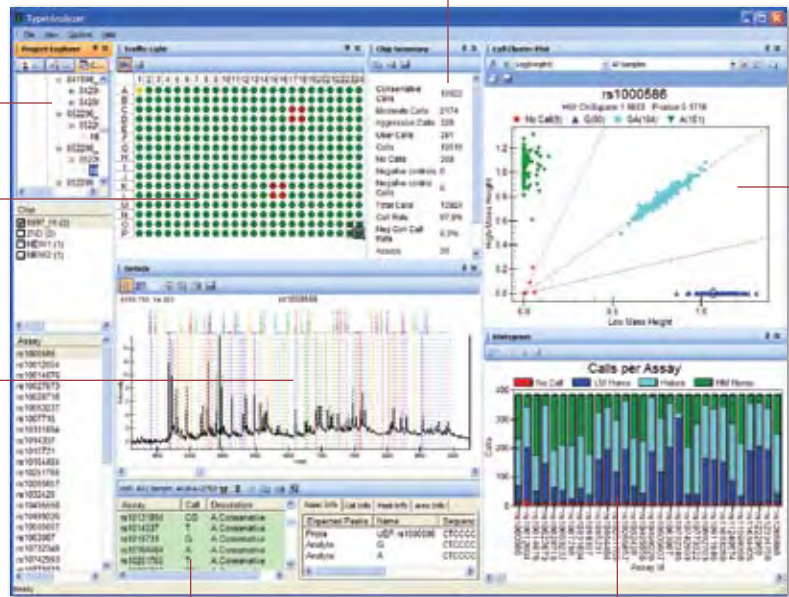
- Quick assessment tool for the percent successful call rate of an assay per well
- Four user-definable colors describe the percent of calls per well

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 TYPED 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

■ MassARRAY® System

MassARRAY® Compact System

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:

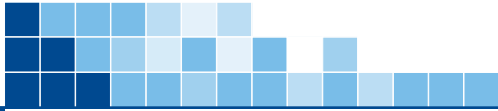
- SNP Genotyping
- Quantitative Gene Expression
- Methylation Analysis
- Comparative Sequence Analysis

MassARRAY® Advantage

MassARRAY Genotyping facilitates identification and prioritization of genetic targets within each stage of biomedical research. The Quantitative Gene Expression and Methylation applications allow functional analysis and target characterization.



Flexibility of Scale with
Versatility of Application



■ 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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