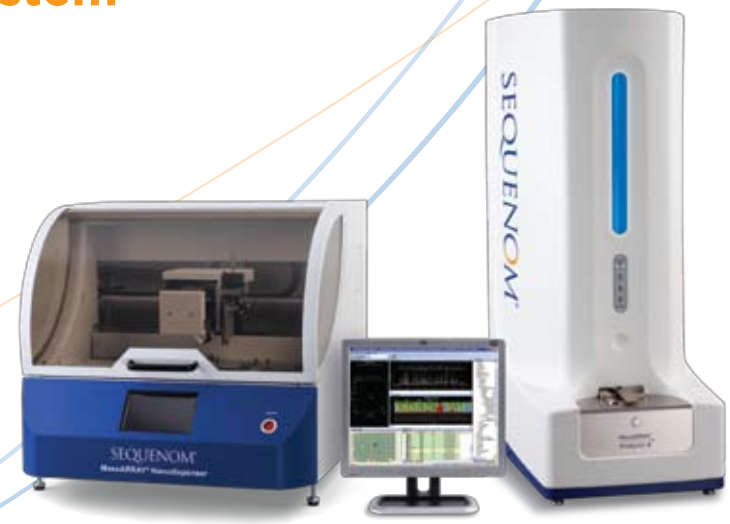
MassARRAY®

Genetic Analysis System



Genotyping

Methylation Analysis

Molecular Typing

Somatic Mutation Profiling

Quantitative Gene Expression (QGE)

SEQUENOM®

MassARRAY®

Genetic Analysis System*

Overview

Next-generation sequencing (NGS) and genome-wide association studies (GWAS) have revolutionized the genetic landscape, leading to the discovery of genetic markers with potential relevance for personalized medicine and pharmacogenomics. However, all genetic marker candidates – whether they are particular polymorphic loci, gene transcripts, or methylated regions – require further validation in order to advance to the next stage of utility and potentially lead to a molecular diagnostic test. An ideal method should be flexible and scalable in throughput to evaluate a varying number and type of markers. The MassARRAY system, available in 96- and 384-SpectroCHIP® formats, affords this capability.

MassARRAY System – an ideal genetic analysis system

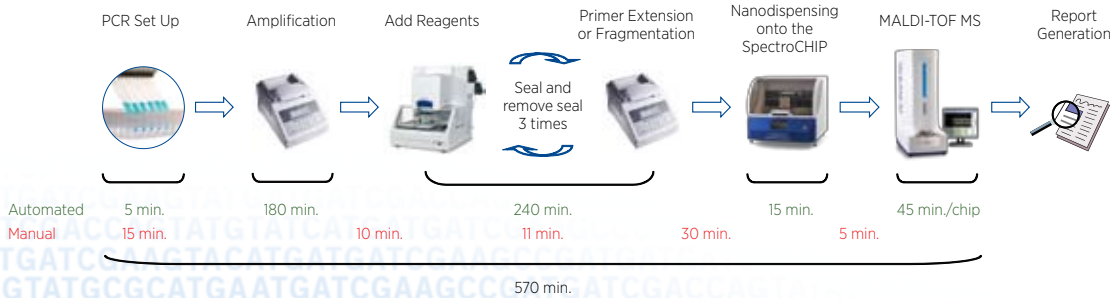
The MassARRAY system offers MALDI-TOF mass spectrometry (MS) for high accuracy and sensitivity, robust chemistry for reproducible results, and advanced data analysis software to meet the needs of any genomic laboratory. The MassARRAY system, a scalable platform with a suite of applications for quantitative and qualitative nucleic acid analysis, provides flexible assay design, fast time-to-results, and cost effective sample run from tens to thousands of samples daily.

These features make it the ideal genetic analysis system for validation and fine mapping studies in basic and translational research settings.

The Complete Solution for Genomic Analysis

- **High Performance** – Accurate MALDI-TOF MS detection provides unparalleled specificity and sensitivity for the most reliable results
- **Maximum Flexibility** – Analyze any combination of SNPs and samples to meet varying study requirements
- **Highly Scalable** – 96- and 384-well options for high and low-throughput applications
- **Easy-to-Use** – New data acquisition software streamlines your workflow, and robust assay design software automates primer design and optimization to maximize efficiency and minimize experimental variability
- **A Variety of Genomic Applications** – Software packages covering somatic mutations analysis, genotyping, methylation analysis and quantitative applications (gene expression and CNV) for easy and integrated workflow and short turnaround time

MassARRAY Workflow



Applications

The MassARRAY system is a flexible platform that allows researchers to perform a variety of DNA analysis applications. The following applications are currently available:

Somatic Mutation Profiling

Recent research studies suggest genetic variants play a significant role in tumorigenesis, pathogenesis, and disease progression[1,2]. Oncogene mutations are key to cancer research. A group at Dana Farber Cancer Institute developed a high-throughput method for genotyping based on the MassARRAY system with over 200 mutations and discovered that >30% of 1,000 tumor samples carried at least one mutation[3]. The group has since developed other panels[4].

Sequenom offers a family of pre-designed assays, OncoCarta™ Panels (v1.0, v2.0, v3.0), for comprehensive mutation screening of solid tumors, as well as a tissue specific melanoma panel, MelaCarta™ Panel v1.0. Other options include targeted and custom panels through Assays by Sequenom. Visit *www.sequenom.com/explore* to see a menu of assays for somatic mutation profiling, or request ones specific to your research interests.

Genotyping

iPLEX Gold is the leading technology for SNP genotyping. The MassARRAY system is widely used for fine mapping and validation of GWAS studies, as well as genetic testing of SNP panels of interest.

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

The iPLEX Gold assay has been used in hundreds of publications to design assays at a multiplexing level of up to 36-plex, offering a high level of flexibility and a low cost per genotype.

Methylation Analysis

MassARRAY EpiTYPER® for quantitative DNA methylation analysis combines base-specific enzymatic cleavage with MALDI-TOF mass spectrometry. This combination creates a highly accurate, sensitive, and high-throughput method for the quantitative analysis of DNA methylation. MassARRAY EpiTYPER is scalable and allows you to analyze multiple CpGs on a single amplicon without compromising accuracy, sensitivity, or reproducibility. The EpiTYPER software provides convenient solutions for data analysis and export.

Molecular Typing

The MassARRAY system with iSEQ™ software sets a new precedent for rapid, accurate molecular identification of microbes, viruses, and other haploid organisms. By combining the sensitivity of PCR and the accuracy of MALDI-TOF Mass Spectrometry, you can analyze one or more target regions on multiple samples in a convenient, homogeneous assay format.

Quantitative Gene Expression (QGE)

QGE combines competitive PCR with MALDI-TOF mass spectrometry enabling a highly accurate, sensitive, and high-throughput method for the quantitative analysis of gene expression. MassARRAY QGE is scalable, allowing you to perform assays without compromising accuracy, sensitivity, or reproducibility. In addition, the method does not use chemiluminescence, fluorescence or other secondary labeling approaches, making the approach cost-effective, in addition to enabling higher levels of multiplexing (up to 24-plex) end unparalleled precision. MassARRAY QGE is the method of choice for validation of microarray data, for the investigation of coding and noncoding transcripts – and whenever sensitivity and accuracy are required in transcription analysis.

Individual System Components

MassARRAY Analyzer 4

The MassARRAY Analyzer 4 is a benchtop MALDI-TOF mass spectrometer specifically designed for genomic research applications. With the ability to automatically process up to two multiplexed 96- and 384-well SpectroCHIP arrays per run, the MassARRAY Analyzer 4 can analyze from dozens to over 100,000 genotypes per day, and from tens to thousands of samples.

MassARRAY RS1000 Nanodispenser

The MassARRAY Nanodispenser is a benchtop dispensing instrument employing advanced robotics for the rapid transfer of nanoliter volumes of sample onto SpectroCHIP arrays. Its standard configuration holds up to two 96- and 384-well microtiter plates. 384 samples are transferred in less than 10 minutes.

MassARRAY Liquid Handler

The MassARRAY Liquid Handler is a small but versatile 96-channel automated pipetting workstation offering a cost-effective solution for higher-throughput Sequenom applications. Pre-programmed with optimized pipetting schemas, set-up and operation is greatly simplified. During microplate handling, disposable plastic tips prevent contamination, and optimized pipetting setups ensure cost-efficient reagent utilization.

MassARRAY®

Genetic Analysis System

Publications

1. Davies MA, Stemke-Hale K, Tellez C, et al. A novel AKT3 mutation in melanoma tumours and cell lines. *Br J Cancer.* 2008;99:1265-1268.

2. Dunlap J, Le C, Shukla A, et al. Phosphatidylinositol-3-kinase and AKT1 mutations occur early in breast carcinoma. *Breast Cancer Res Treat.* 2010 Apr;120(2):409-18.

3. Thomas RK, Baker AC, DeBiasi RM, et al. High-throughput oncogene mutation profiling in human cancer. *Nat Genetics.* 2007;39:283-294.

4. MacConaill LE, Campbell CD, Kehoe SM, et al. Profiling Critical Cancer Gene Mutations in Clinical Tumor Samples. *PLoS ONE.* 2009:4(11):e7887.

For more information please contact your area Sequenom office.

NORTH AMERICA: +1 (877) 443-6663
EUROPE: +49 4089 967640
ASIA-PACIFIC: +61 7 3845 369
www.sequenom.com

CHINA: +86 10 8048 07 37
sequenom.china@sequenom.com

JAPAN: +81- 3-6802-5590
sequenom.japan@sequenom.com

diatech pharmacogenetics

Diatech Pharmacogenetics srl
via P. Pellegrini, 3 - 60035 Jesi (AN) - ITALY
T + 39 0731 213243 - F + 39 0731 213239
info@diatechpharmacogenetics.com
www.diatechpharmacogenetics.com

SEQUENOM®

#sq177_sys_v7(0113-2011)