

SEQUENOM®



MassARRAY® Genetic Analysis System

Translating Genetic Research Into Results That Make a Difference



MassARRAY Genetic Analysis System

The only genomics platform optimized for translational research

Closing the gap between discovery and clinical utilization

While whole genome studies have yielded a vast number of candidate biomarkers for diagnosis, toxicity, and efficacy, to date very few have been clinically validated. Closing this gap requires high-throughput tools that enable efficient and cost-effective validation of panels of biomarkers in population-based studies.

For translational research applications, Sequenom's MassARRAY Genetic Analysis System provides unmatched performance and versatility. Accurate quantitative assays, simple and flexible assay design, and a highly sensitive MS-based detection platform combine to enable efficient validation of broad classes of molecular biomarkers across sample types—at a price you can afford.

MassARRAY gives you the power and versatility to address your most pressing research challenges and accelerate the pace of discoveries reaching the clinic.

A ROBUST AND VERSATILE Platform for Translational Research

High-confidence biomarker validation

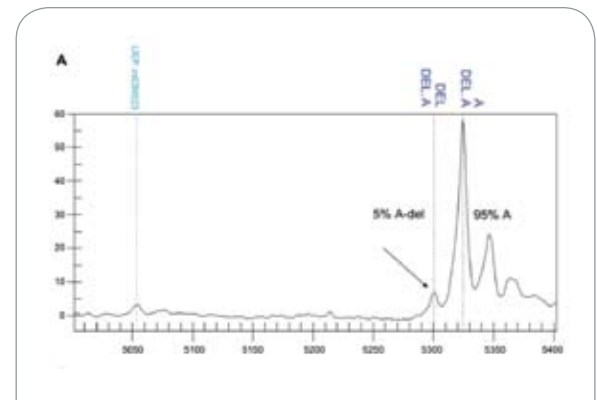
MassARRAY detects genetic targets with > 99% accuracy and attomolar sensitivity, giving you the confidence to validate biomarkers and to move your research forward. The system couples an efficient quantitative assay featuring a simple, robust, label-free primer extension chemistry iPLEX Gold with the sensitivity, accuracy, and precision of MALDI-TOF MS. Exceptional accuracy and sensitivity make MassARRAY ideally suited for the most demanding applications, such as the detection of low-frequency mutations that would otherwise be missed using conventional techniques, and the measurement of gene expression levels from FFPE tissue samples.

Flexible assays for multiple biomarker types

MassARRAY gives you the flexibility to choose from a large collection of pre-validated assays or to construct custom assays for a broad range of biomarkers. MassARRAY software automates the design and optimization of primer sets and assays based on user-defined sequences, allowing you to quickly and easily generate and refine assays throughout the validation process. The platform supports the analysis of multiple categories of molecular biomarkers, including SNPs, copy number variants, methylation patterns, and expression signatures, providing maximum flexibility and value.

Results in a fraction of the time and cost of other techniques

MassARRAY is scalable and can be tailored to run tens to thousands of samples per day, significantly minimizing costs per sample. With efficient multiplexed assays, flexible assay design, and fully automated data acquisition and analysis, you will get data in days, instead of weeks or months. Adopting the MassARRAY system helps you expedite the translation of research findings into validated biomarkers without compromising data quality.



MassARRAY technology can discriminate as little as 5% of SNP-containing mtDNA in a 95% background of mtDNA lacking the polymorphism.¹

MassARRAY—

Accelerating biomarker validation

As the only genetic analysis system optimized for translational research, MassARRAY is an ideal tool for a new era of medicine. With its exceptional accuracy and flexibility, researchers can move broad classes of molecular biomarkers towards clinical utility. The technology supports multiple applications on a single platform, enabling a comprehensive view into molecular events associated with disease and drug toxicity. MassARRAY provides the systems-level insight required to predict individual disease susceptibility and response to medication, advancing personalized healthcare.

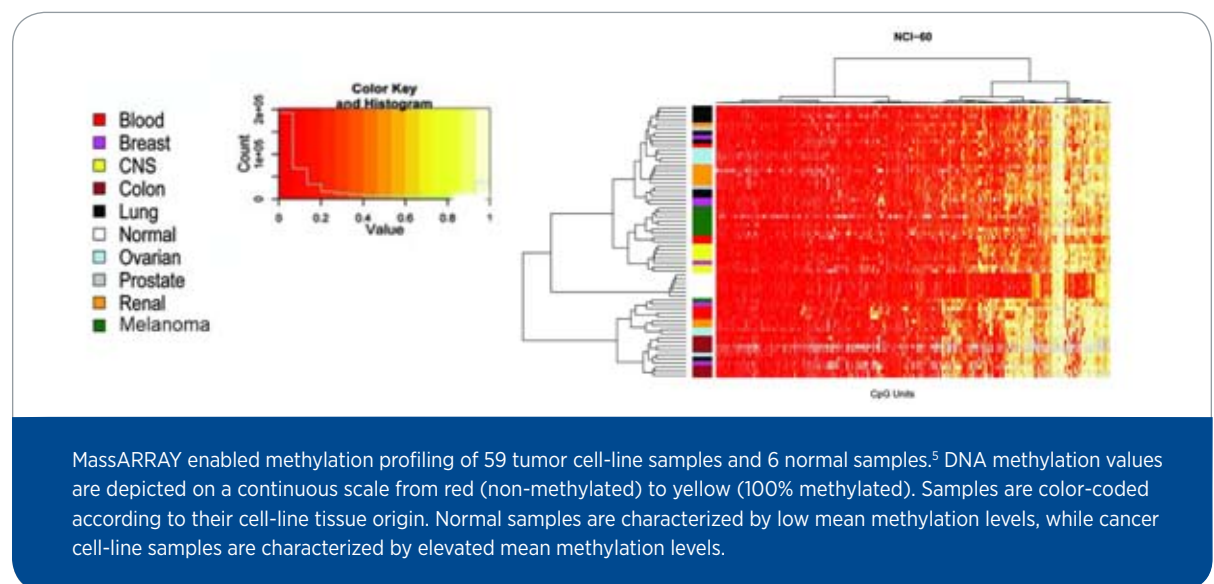
SNP Genotyping

The MassARRAY system is routinely used for evaluating targeted SNP panels. Sequenom's iPLEX Gold genotyping assay combines the robustness of single-base primer extension biochemistry with the sensitivity and accuracy of MALDI-TOF MS detection, so that even alleles represented at low frequencies in the DNA pool will be detected.

With MassARRAY, you can detect low-frequency mutations in heterogeneous samples with confidence. Researchers at Dana Farber Cancer Institute used MassARRAY technology to detect mutations contained in as few as 9% of the cells in lung cancer samples—findings which were missed by standard Sanger sequencing.² To further expedite validation efforts in oncology, Sequenom's OncoCarta™ Panel enables simultaneous detection of 238 mutations across 19 of the more common oncogenes, including EGFR, BRAF, KIT, and KRAS.

Gene Expression Profiling

Gene expression profile-based biomarker signatures have proliferated in recent years due to the broad adoption of microarray technology. MassARRAY is the ideal tool for precisely measuring gene expression levels for post-array validation. Sequenom's gene expression-based assays are able to differentiate changes in expression levels as little as 10%.^{3,4} In addition, you can look at events not possible



with other techniques, such as absolute transcript abundance and allele specific expression patterns.

Copy Number Variants

While the field of genomics has long been focused on SNPs as the key to understanding genetic variation and its role in disease, recent studies have shown CNVs to be equally important. With MassARRAY, researchers can accurately validate known copy number variants, events associated with clinical phenotypes, and perform highly accurate fine-mapping of genomic regions containing variants.

Methylation

Methylation patterns are another growing and important class of biomarkers, particularly in oncology. The Sequenom EpiTYPER assay allows you to quickly and accurately perform quantitative measurements of methylation ratios across multiple candidate genomic regions. Scientists have recently used MassARRAY technology to obtain DNA methylation profiles for more than 400 cancer-related genes over 59 cell lines (NCI60) and normal tissues derived from 9 different tissue types, which revealed distinct patterns and degrees of methylation unique to certain cancer types and samples.⁵

“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

Making personalized medicine a reality

Around the world, scientists are working on ways to turn today's research into tomorrow's clinical breakthroughs. Significant advances in treatments for disease and drug toxicity, as well as other key aspects of personalized medicine, promise to change the status of modern healthcare and to improve the quality of life for everyone. This critical effort requires not just dedicated scientists and researchers, but exceptional tools as well.



Sequenom's MassARRAY Genetic Analysis System offers unmatched performance, versatility and value, making it the ideal platform—and an indispensable tool—for today's translational researchers.

References

1. A rapid and accurate approach to identify single nucleotide polymorphisms of mitochondrial DNA using MALDI-TOF mass spectrometry. *Clin Chem Lab Med* 2008 46(3):299-305.
2. High-throughput oncogene mutation profiling in human cancer. *Nat Genet* 2007 39(3):347-351.
3. Quantitative analysis of nucleic acids—the last few years of progress. *J Biochem and Mol. Biol.* 2004 37(1):1-10.
4. Cytoplasmic BKCa channel intron-containing mRNAs contribute to the intrinsic excitability of hippocampal neurons. *Proc Natl Acad Sci* 2008 105(6):1901-1906.
5. Cytosine methylation profiling of cancer cell lines. *Proc Natl Acad Sci* 2008 105(12):4844-9.