

# Make Illumina part of your DNA.

Integrated tools for DNA analysis.











# Integrated tools. Unprecedented research.

DNA. It's the foundation of life. A code to decipher. You ask questions, and every answer takes you closer. To answers that advance genetic knowledge. With the industry's most comprehensive set of DNA analysis tools, we're with you at every stage. From discovery to validation to screening. Providing unprecedented insights and helping you to see more. Publish faster. Be the first to discover.

Every research program is unique. You need to be able to look at DNA from different perspectives. That's why we work alongside the Illumina Community to constantly develop tools that help you do just that. From whole-genome discovery to identification of markers. Fully integrated for total discovery. Explore how we can help you analyze DNA like never before. With power, reproducibility, and ease.



# Multiple applications. Unlimited answers.

There are many ways to look at DNA. Combining approaches can make the difference between an interesting study and a bona fide breakthrough. Integrate our key platforms and discover more. Publish faster. Do it all with simple workflows and multiple assay options that are right for your study scope and focus.

With our integrated set of DNA analysis tools, you can take your research as far as you can imagine. We offer three distinct platforms for analyzing DNA:

- Sequencing technology on the Genome Analyzer and iScan Sequencing Module
- ▶ BeadArray<sup>™</sup> Technology on the iScan System
- ▶ VeraCode® Technology on the BeadXpress® Reader

Our solutions allow you to generate significant amounts of high-quality data faster, using fewer resources, at a lower cost. All connected using the GenomeStudio® data analysis software suite for true integration. All developed with the help of Illumina Community researchers.



Sequencing
GENOME ANALYZER
base-pair resolution



BeadArray Technology iSCAN SYSTEM 96 to > 1M markers



VeraCode Technology
BEADXPRESS READER
2- to 384-plex







## Robust data. Simple analysis.

Point and click. Graphic results without waiting. Our GenomeStudio data analysis software is a desktop application that behaves like a desktop application.

Fast and intuitive. It's a robust primary analysis package that consists of seven discrete application modules.

Conveniently compare and correlate data from different applications and platforms to obtain a complete picture of genomes and disease targets. Easily import data generated from the Genome Analyzer, iScan System, or BeadXpress Reader. Visualize and analyze using tables, plots, and genome browsing components. And, every module in GenomeStudio is designed with open architecture for integration with third-party applications, enabling ease of data parsing for advanced downstream analysis.

#### Integrated framework

Analyze data from numerous applications across all Illumina platforms using the same program.

#### User-friendly design

Learn one software tool, regardless of application; visualize data in multiple formats.

#### Powerful analytics

Filter data using several metrics, perform comparative analyses, and easily share projects with colleagues.

#### Open data access

Export data into third-party analysis tools for downstream analysis.

#### GenomeStudio Modules

#### **DNA Sequencing Module**

Analyze DNA sequencing data generated using the Genome Analyzer and Pipeline software.

#### Genotyping Module

Analyze genotyping and CNV data generated using the iScan System or BeadXpress Reader.

#### Methylation Module

Analyze methylation data from microarray images scanned by the iScan System or BeadXpress Reader.

#### ChIP Sequencing Module

Analyze data from whole-genome ChIP-Seq experiments performed using the Genome Analyzer.

#### Additional modules include:

RNA Sequencing Module Gene Expression Module Protein Analysis Module

#### **DNA SEQUENCING**

#### **Genome Sequencing**

Any genome. Every genetic variant.

- Analyze single-nucleotide variations, copy number variations, and chromosomal rearrangements across genomes.
- Generate *de novo* assembly of simple and complex genomes.



#### **SNP GENOTYPING**

#### Whole-Genome Genotyping

High data quality. Faster publication.

- Use tagSNPs to access the highest genomic and CNV coverage available, including content from the 1,000 Genomes Project.
- Obtain high-quality SNP data with > 99% average call rates and 99.9% reproducibility, making downstream data analysis easier.
- Process samples faster with multi-sample BeadChips.

#### **Custom and Focused Genotyping**

Any SNP. Any species.

- Design custom SNP genotyping panels from 1 to 200,000 SNPs.
- Select from a wide variety of fixed SNP genotyping panels.
- Unprecedented support for custom panel designs.



#### **COPY NUMBER VARIATION**

#### **CNV** Discovery

Any structural variant. Any genome.

- Access any genome at singlebase resolution.
- Discover CNVs without any a priori assumptions.
- Use unlimited dynamic range to discretely identify a wide range of copy number variations.

#### **CNV** Analysis

Premier coverage. Unsurpassed flexibility.

- Screen and discover CNVs with reliable detection of deletions, inversions, rearrangements, and amplifications.
- Leverage optimal signal-to-noise ratios through SNP markers and high feature redundancy.
- Access the industry's best coverage of common and rare CNV region.

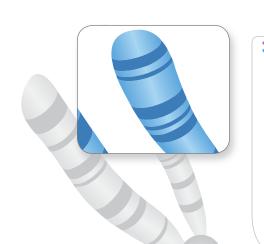
Increase the power of your association study and decrease your overall project cost. Our online controls database, iControlDB, is a repository for genotyping control data.

Access free control samples at: www.illumina.com/icontroldb

Genome Analyzer

iScan System

BeadXpress Reader



#### **CYTOGENETICS**

#### **Digital Karyotyping**

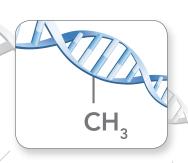
Advanced karyotyping tools. The ultimate molecular karyotype.

- Combine short-insert and longinsert paired-end sequencing to catalog any size genomic rearrangement.
- Discover insertions, deletions, tandem duplications, and combinations of aberrations with balanced translocations and inversions.

#### **Cytogenetic Abnormalities**

Rapid identification and discovery. Proven SNP technology.

- Detect known cytogenetic abnormalities and discover new regions.
- Detect intercellular mosaicism and copy-neutral events undetectable with array-CGH.
- Generate reports and analyze data with KaryoStudio software.



#### DNA METHYLATION ANALYSIS

#### DNA Methylation Discovery & Analysis

Any organism. Site-specific resolution.

 Generate base-by-base or site-specific maps of the epigenome, following bisulfite conversion or anti-methyl cytosine antibody-based selection.

#### **DNA Methylation Analysis**

High-throughput processing. Cost-effective analysis.

Interrogate over 27,000 rationally selected CpG loci covering more than 14,000 genes at singlenucleotide resolution.

#### **DNA Methylation Validation**

Custom biomarker validation. Simultaneous interrogation.

Interrogate 48–384 custom DNA methylation sites simultaneously.

#### **DNA-PROTEIN INTERACTIONS**

#### **DNA-Protein Interactions**

Rapid discovery. Minimal sample requirement.

- Use ultra-low 10 ng input, significantly less than is required for ChIP-chip.
- Study any immunoprecipitate from any organism with a sequenced genome.
- Obtain digital data that allow precise positional mapping of binding sites better than 50 bps.



### Dynamic community. Breakthrough discovery.

"At this point we haven't found anything that we can't sequence with the Genome Analyzer.
Wherever the biology leads us, that's where we'll go."

Brian D. Gregory, Ph.D.
 Damon Runyon Postdoctoral Fellow
 Salk Institute, Plant Biology Laboratory

"For us, using Infinium HD arrays is really a no-brainer. The data quality is just unbelievable. We demand extremely high accuracy since even a small percentage of false positives or negatives creates a huge amount of downstream work. High-quality data helps us find the answers faster."

Kevin Shianna, Ph.D.

Director, Genotyping Facility
Institute for Genome Sciences & Policy
Duke University

## Partnering with Illumina.

At Illumina, we aim to be your partner in science; your success is our goal. To help you meet your research objectives, we offer a range of collaborative services that provide expertise to drive your discoveries. Whether you decide to place a system in your own lab, collaborate with one of our CSPro® Certified Service Providers, or work with our internal FastTrack Services experts, we can provide a solution that is optimal for your lab and scale of research. A variety of options make Illumina's industry-leading technology convenient and accessible.

- ► FastTrack Sequencing Services

  Collaborative study design: Illumina sequencing experts

  work with you during study design and sample preparations
  for broad range of applications.
- FastTrack Genotyping Services
  Industry-leading performance: Illumina scientists help design genotyping experiments that produce superior results, with average call rates > 99.7% and high assay conversion rates.
- CSPro Program Quality and Convenience: CSPro service providers deliver the highest-quality data available for genetic analysis applications from locations around the world.







1.800.809.4566 toll-free 1.858.202.4566 tel



#### **WORLDWIDE HEADQUARTERS**

#### Illumina, Inc.

9885 Towne Centre Drive San Diego, CA 92121 USA 1.800.809.4566 toll-free 1.858.202.4566 tel 1.858.202.4766 fax

www.illumina.com

For local offices and international distributors, visit www.illumina.com/locations

