

# Go Beyond the Genome You Know

## More Understanding, Not Just More Data

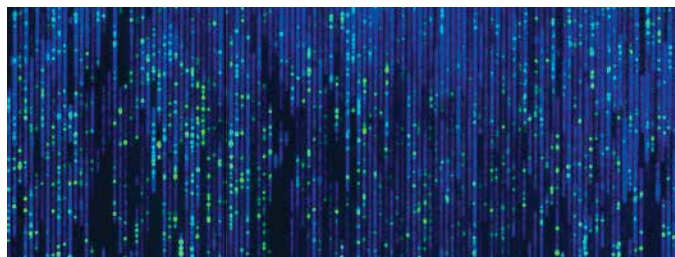
Despite remarkable access to massive amounts of relatively inexpensive short reads of sequence data, biologists still don't have sufficient tools to easily view the full context and architecture of the expansive genomic landscape. It is becoming increasingly clear that this long-range view of contiguity is essential for understanding structural variation.

## Long DNA Molecules

At the heart of the Irys System is a patented chip technology with micro- and nanostructures that unravel randomly coiled DNA and feed it into nano-scale channels. These NanoChannels prevent semi-flexible molecules from folding back or tangling so they can be directly imaged in a massively parallel array (Figure 1). IrysPrep™ reagent kits label long molecules of DNA at specific sequence motifs to generate signature patterns that uniquely identify the genomic region and surrounding context.

## The Genome in Context

Visualizing whole genomes at the single-molecule level eliminates bias induced by amplification and shearing, while overcoming the ambiguities and time investment that hinders large-scale assembly. A whole-genome physical map provides scaffolding to make sequencing experiments more efficient and complete. Long-range genome architecture is preserved in the maps to identify biologically significant structural variants.



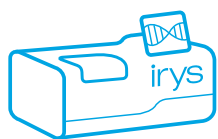
(Figure 1) Single-molecule imaging of DNA up to megabase-length

**Long Molecules:** Intact DNA molecules, hundreds of kb long, span repeats and variants

**High-Quality Data:** Single-molecule, high-resolution imaging of uniformly stretched, linearized DNA

**Scalable Open Platform:** Automated multicolor IrysChip™ imager for use with IrysPrep kits or user-developed labeling schemes

## Irys System—The Complete Solution



Irys Instrument

Effortlessly linearize DNA and analyze genome data with automated, single-molecule imaging of extremely long DNA.



IrysChip

The heart of the Irys System, proprietary NanoChannels uniformly stretch DNA for high-throughput analysis of genomic architecture.



IrysReagents

Extract and label long DNA for use in Irys *de novo* assembly and structural variation detection.



IrysView Software

Powerful analysis software for *de novo* assembly, structural variation detection, visualizing and exporting run results.



IrysSolve  
Computational Solutions

IrysSolve is our streamlined data analysis pipeline implemented on a variety of computational solutions.



# Applications

Irys empowers a host of applications that were previously too expensive, time consuming or limited by short-read technologies. IrysPrep kits and IrysView™ analysis software support structural variation detection, assembly validation and scaffolding of NGS data and enable virtually unlimited user-developed applications utilizing long-read single-molecule data.

## Structural Variation Analysis

Irys is particularly well suited for analyzing challenging structural variation, including inversions, that can have functional significance in essentially any species (Figure 2). NGS suffers when short reads collapse CNVs in addition to complications inherent to *de novo* assembly. Older methods, such as array-CGH, lack positional information and depend on probes limited only to model species reference sequence. Irys overcomes these limitations by creating, *de novo*, a genome-wide view of structural variation with read lengths long enough to capture the wide range of variant sizes. By visualizing the genome in its native state, positional information, such as balanced translocations and the localization of duplication insertion points, is readily apparent (Figure 3). Furthermore, using single-molecule detection, rather than aggregate measures in NGS and CGH, means subpopulations can be detected directly.

## NGS Anchoring and Scaffolding

Irys facilitates *de novo* assembly to higher levels of completion, in less time, by providing high-resolution genome maps to scaffold NGS data. Genome maps orient contigs and size gaps with long-range connectivity bridging across repeats and other complex elements that break NGS assemblies (Figure 4).

Other restriction-based mapping methods do not use molecules long enough to capture complex repeats, whereas the proprietary Irys nicking and labeling chemistry retains very long molecules intact. These long molecules allow us to assemble contiguous maps and visualize inheritance of genes in a heterozygous individual who receives alleles from each parent with different copy number variations (Figure 5). Without the ability to capture very long molecules, repeats may collapse upon assembly just as in any short-read method.

## Assembly Validation

Irys genome maps represent a powerful orthogonal validation method for assembled genomes. In IrysView, discrepancies from imported NGS assemblies are readily visible in a graphical browser and listed in tabular form for deeper exploration to individual molecule images, allowing one to quickly design additional confirmation or sequencing experiments.

## System Performance

Parameter	Value
Flowcells per IrysChip	2
Cycles per Flowcell	up to 26
Runtime per Cycle	55 min
Filtered Throughput per IrysChip (24 hrs)*	96 Gbp
DNA labeling Input Amount*	300 ng
DNA Labeling Time (total   hands-on)*	1.5 day   3 hr
Average Labeled Molecule Size (post filter)*	225,000 bp
Consensus Label Error Rate (FP/FN)**	~1%
Consensus Label Precision**	150 bp

\*for supported samples

\*\*at 50X coverage, for consensus segments 25 kbp or less

## Learn More

For more information about Irys or to place an order, please contact [info@bionanogenomics.com](mailto:info@bionanogenomics.com) or visit [www.bionanogenomics.com](http://www.bionanogenomics.com).

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