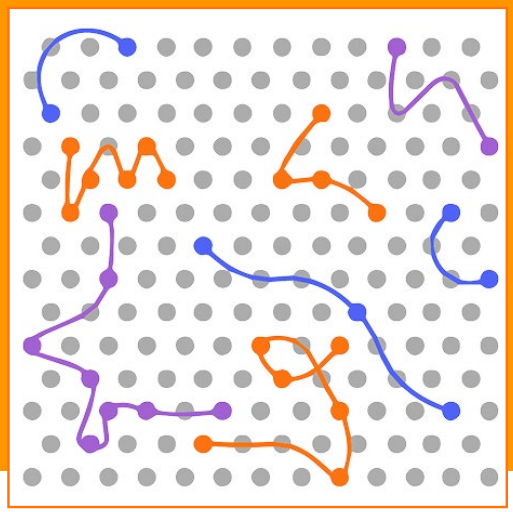
TruPath™ Genome

Accurate. Comprehensive. Effortlessly Simple.

Whole-genome sequencing with long-range insights— powered by short-read simplicity



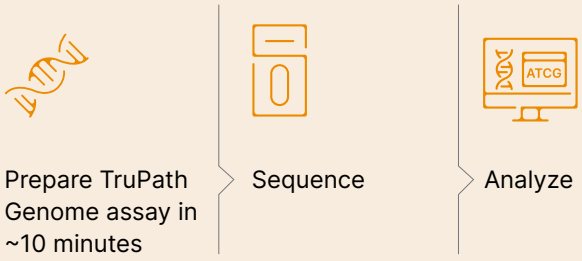
Illumina TruPath Genome transforms whole-genome sequencing (WGS) by combining the accuracy and accessibility of short-read sequencing with long-range insights traditionally limited to long-read platforms. Powered by proximity-enhanced mapped read technology, TruPath Genome delivers an ultra-simple workflow—with **~10 minutes hands-on time**—unlocking challenging genomic regions, structural variants, and phased variant calls across the human genome.

The result is a **comprehensive, high-accuracy human genome** made remarkably simple.

Simplest sample-to-sequencer WGS workflow

Ultra-simple library prep.
High-impact genomic insight.

- Proximity information enables high-confidence mapping of highly homologous, repetitive, and segmentally duplicated regions
- Demonstrated recovery of coverage in medically relevant genes such as *STRC* and *PMS2*, which contain > 99% identical pseudogenes that confound standard WGS
- Enables confident variant detection in “dark” or ambiguous genomic regions where short-reads typically fail

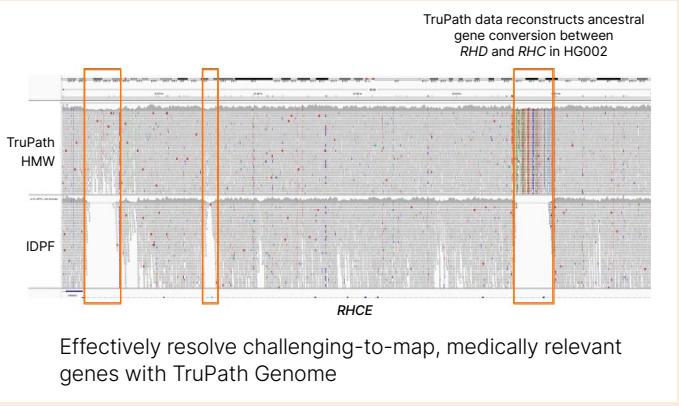


High-accuracy variant calling in difficult-to-map regions

Improved precision for SNVs, indels, and SVs*

- Benchmarking with HG002 shows **substantial reductions in false positives and false negatives** compared to all other technologies
- Proximity-enhanced mapping improves SNV and indel accuracy in difficult-to-map regions
- Structural variant recall improves dramatically compared to standard sequencing using DRAGEN™ analysis and T2T-Q100 truth sets

* SNV, single nucleotide variant; Indel, insertion-deletion; SV, structural variant.



Effectively resolve challenging-to-map, medically relevant genes with TruPath Genome

illumina®

1.800.809.4566 toll-free (US) | +1.858.202.4566 tel
techsupport@illumina.com | www.illumina.com

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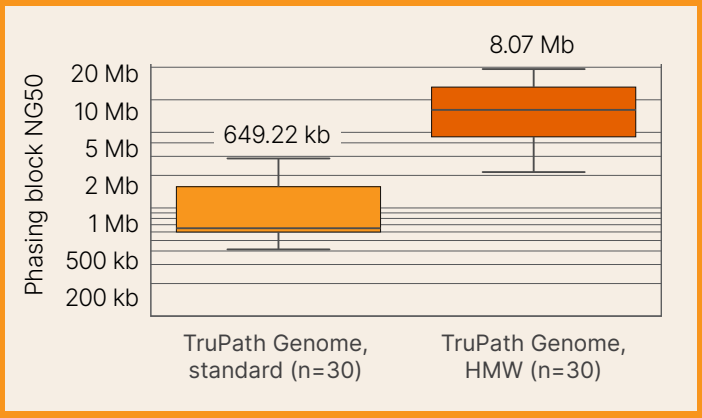
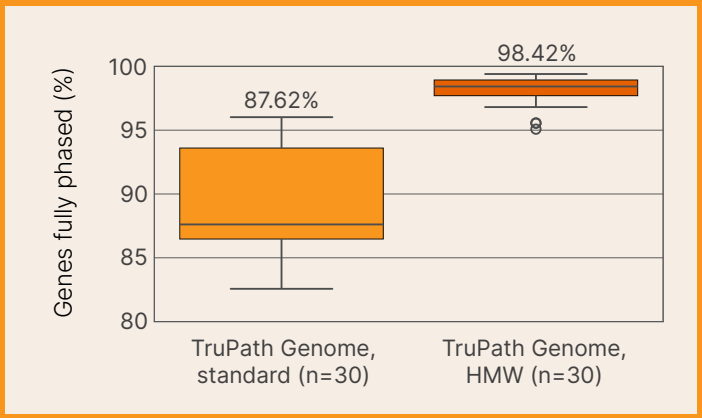
Long-range genomic insights with ultra-long phasing and SV visualization

Reveal haplotype-resolved genomes and complex rearrangements.

- Proximity mapped read technology produces phase blocks extending **hundreds of kilobases to several megabases,** enabling haplotype-resolved variant interpretation
- TruPath Genome phases **~98% of heterozygous SNVs** across high-molecular-weight (HMW) DNA extractions
- Multi-region joint detection (MRJD) provides accurate variant calls across paralogous genes, reducing the need for reflex assays
- Enables colocation plots—high-resolution structural rearrangement maps to reveal long-distance insights





Ultralong phasing with TruPath Genome on 30 matched HMW and standard extraction cell lines

Comprehensive WGS—without complexity

TruPath Genome delivers the **simplest WGS workflow** on the NovaSeq™ X Series while dramatically expanding what's possible with short-read sequencing. By preserving long-distance molecular relationships without complex library prep, **TruPath Genome delivers these capabilities in a single, streamlined WGS assay:**

- High-accuracy variant detection
- Enhanced structural variant resolution
- Ultra-long phasing
- Reliable mapping in challenging genomic regions

TruPath Genome represents a new standard for comprehensive human WGS—easy, fast, powerful, and ready for high-throughput labs.

Discover how TruPath Genome can simplify your workflow and elevate your insights

- **Visit** illumina.com/trupath
- **Contact** your Illumina representative for evaluation options
- **Learn** more about mapped read technology and NovaSeq™ X systems

Explore TruPath Genome and mapped read technology

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