TruPath™ Genome

Accurate long-range insights—without long-read workflow trade-offs

TruPath Genome combines high-accuracy short-read sequencing with proximity-mapped read technology to support comprehensive human whole-genome analysis, including phased SNVs and indels, structural variants, CNVs, STRs, and high-homology regions, using the NovaSeq™ X Series.

Accurate. Comprehensive genome insights.

1

Long-range context: proximity signal supports phasing, SV characterization, and expanded STR size estimation

2

High-accuracy small variants: to deliver the most accurate small variant calls

3

Complex rearrangements: colocation-based filtering reduces calling errors in large translocations and inversions

4

High-homology regions: integrated haplotype visualization supports review of complex paralogous regions

TruPath Genome analytical accuracy, without trade-offs.

Higher confidence SV calls

Colocation-based filtering reduces calling errors in inter- and intra-chromosomal rearrangements (e.g., large translocations/ inversions), improving confidence in large structural variant calls

Consistency, not variability

Consistent performance across sample types and inputs—without dependence on specialized extraction or workflow optimization

Comprehensive variant detection

Phased small variants, SVs, CNV, and STRs

Coverage of difficult to map regions

Improved resolution of difficult-to-map and paralogous regions through proximity-enabled long-range context

Reduced need for orthogonal validation

Integrated signal improves confidence in variant calls, helping reduce reliance on orthogonal verification methods

TruPath™ Genome

Long-range insight.
Short-read accuracy.
Reveal more of the genome.

Small Variants

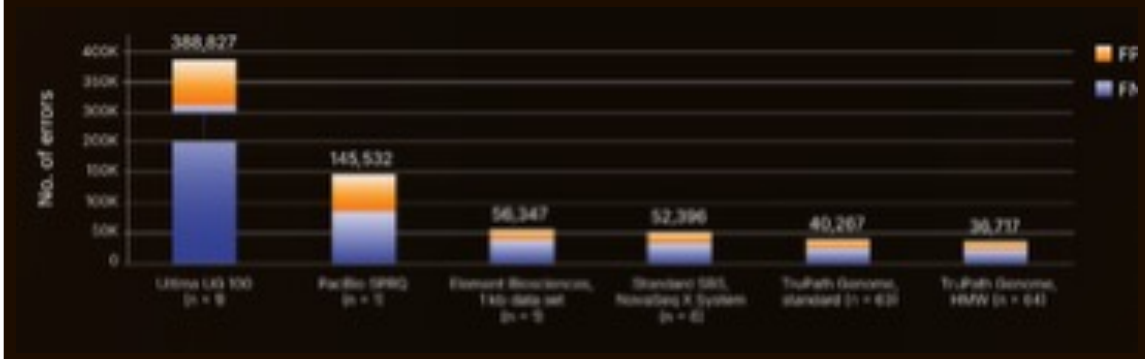
Fewer errors, focus on the variant signal that matters.

TruPath has best-in-class T2T-Q100 small variant calling accuracy: **F1 99.41% accuracy score.**

High small-variant accuracy is most compelling when it also reduces distracting incorrect data output and missed calls.

>3x fewer "dark genome coding regions" (low MapQ) than long-read technology (PacBio)

20k fewer errors than closest short-read technology (Element Biosciences)



Accuracy: FPs+FNs, SNPs+indels, HG002, T2T-Q100 v1.1

The most accurate genome

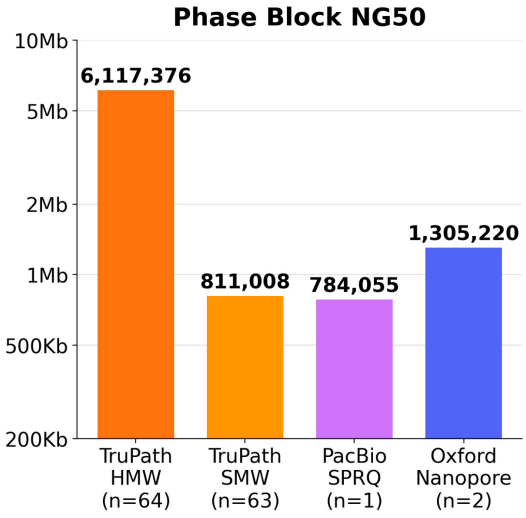
TruPath Genome delivers overall accuracy gains for 30%-90% compared to more error-prone on-market technologies, with > 3x fewer "dark genome coding regions"* than long-read technology (PacBio) and ~20k fewer errors than the closest short-ready technology (Element Biosciences). Benchmark: NIST Genome in a Bottle HG002 T2T-Q100 v1.1 small variant truth set, now known as v5.0q.

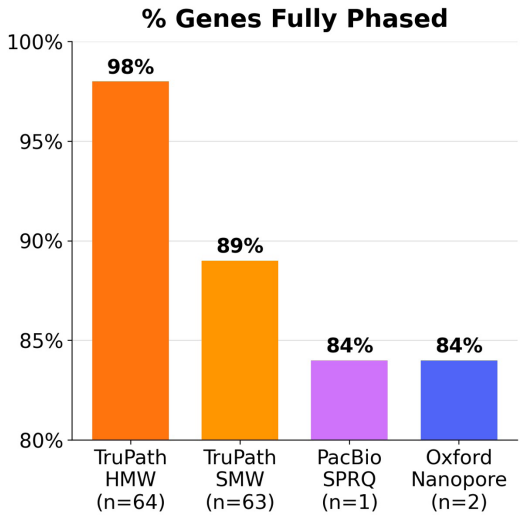
*Dark regions as defined by (Ebert et al. 2025)

Phasing

Longer phase blocks, more fully phased genes.

More genes phased without compromising accuracy. In publicly available datasets, TruPath Genome supports broader, longer-range phasing, with up to 98% fully phased genes using HMW input and 89% with standard input.





Note: performance in this slide shows only HG002. https://downloads.pacbcloud.com/public/revio/2024Q4/WGS/GIAB_trio/HG002_rep1/analysis/v3.0.2/

PacBio data. https://downloads.pacbcloud.com/public/revio/2024Q4/WGS/GIAB_trio/HG002_rep1/analysis/v3.0.2/
PacBio only provides HG002

ONT. https://42basepairs.com/browse/s3/ont-open-data/giab_2025.01/analysis/wf-human-variation/sup/ –median based on ONT provided results for HG001-7

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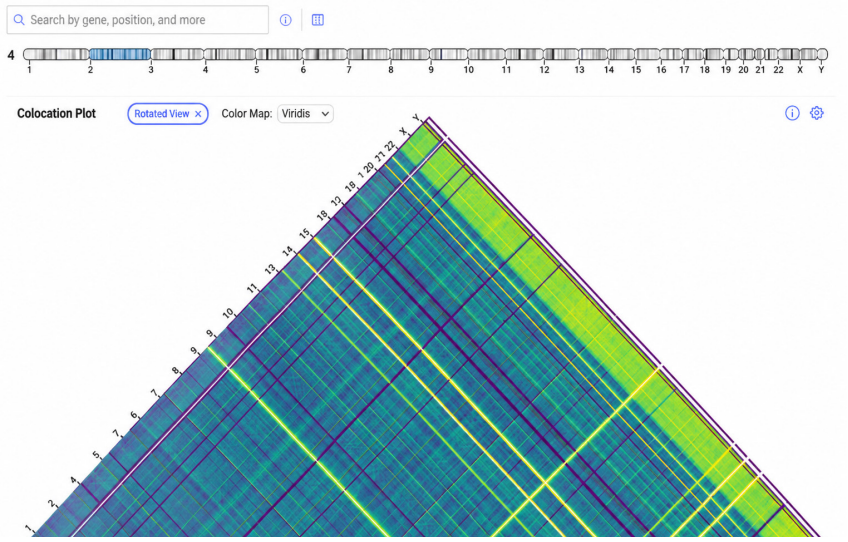
Complex SVs

High confidence calls, fewer incorrect data outputs.

TruPath's novel capability to visually **confirm large, complex events may reduce the need for orthogonal tests**—like no other WGS assay.



Example: ICA visualization

HIGH CONFIDENCE CALLS. REDUCED CALLING ERRORS.

Colocation maps

Depict long-distance relationships from 1 kbp to genome scale to help visualize large, complex events.

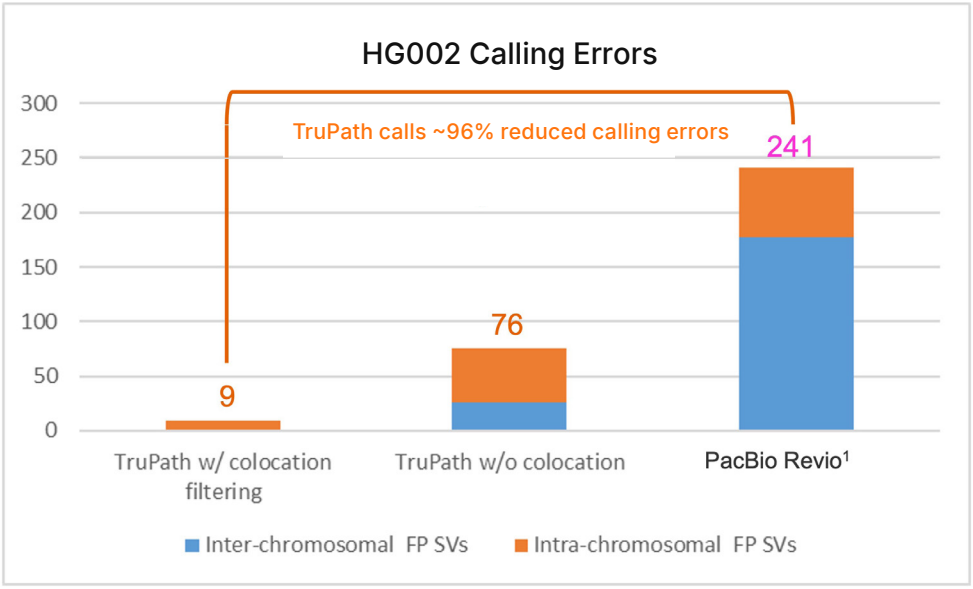
Colocation-based filtering

Uses long-distance proximity signal to reduce calling errors in large translocations and inversions.

Integrated visualization

Research interpretation of large, complex variants—no external tools required.

Genome in a Bottle NIST T2T-Q100 HG002 SV truth set



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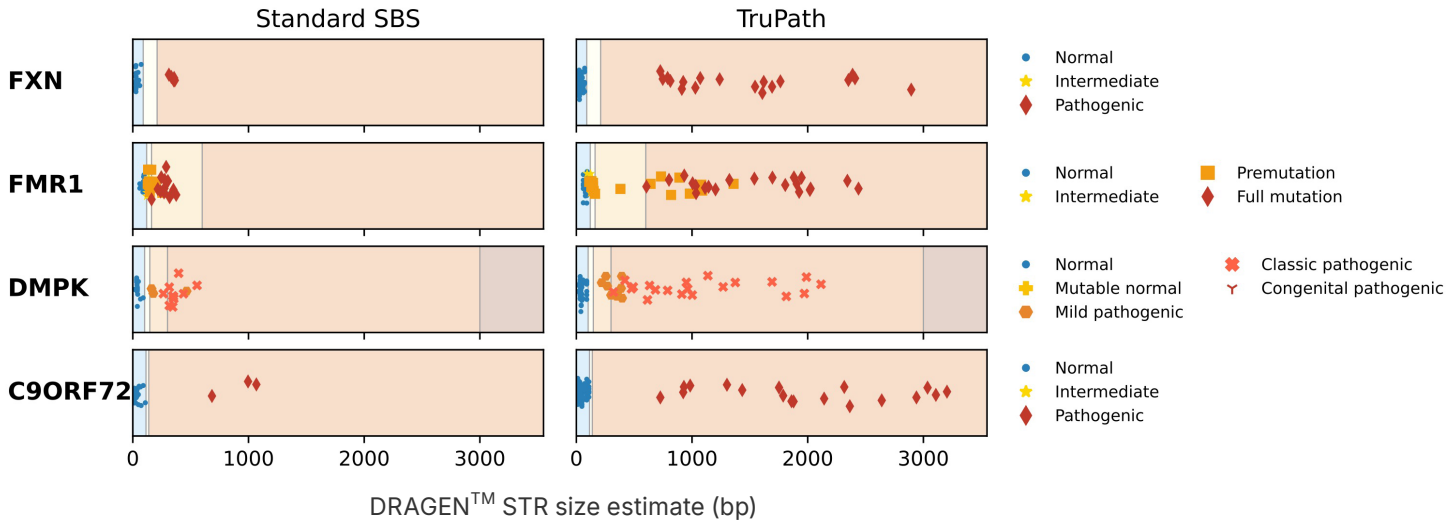
STRs

Proximity information supports long-range context enabling improved expanded STR size estimation. TruPath Genome extends STR size estimation into larger repeat ranges and can resolve expanded alleles independently.

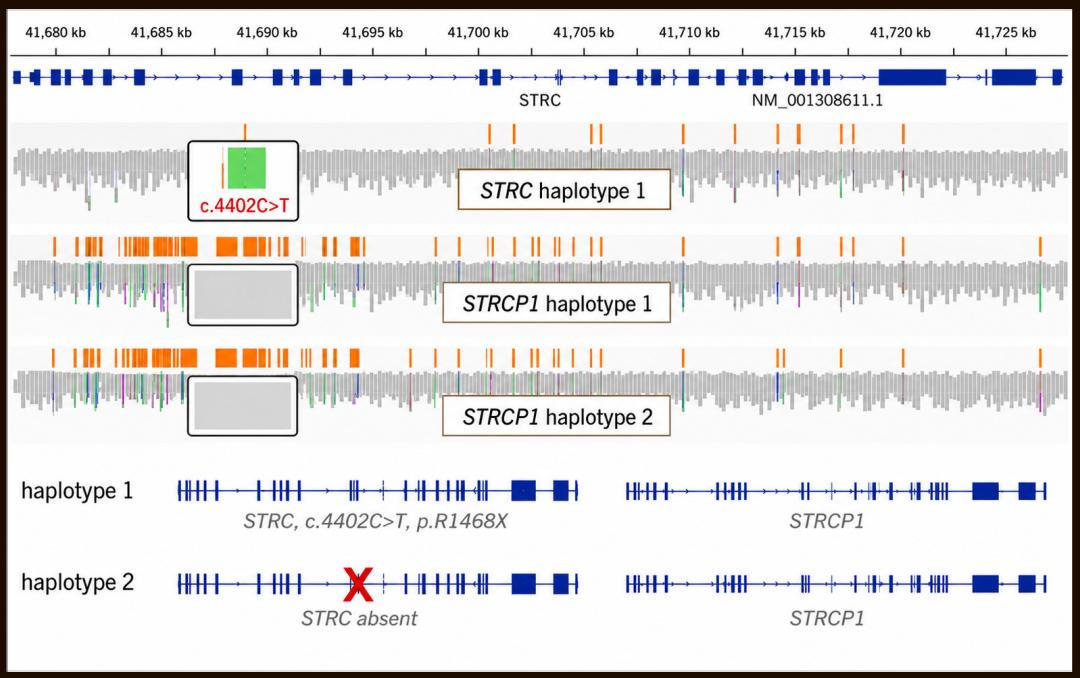
Long-range context enables accurate resolution of challenging regions and STRs—without the complexity of long-read workflows.



Paralogous Genes

Resolve the region, interpret the haplotype.

Integrated haplotype visualization powered by Multi-Region Joint Detection (MRJD) makes complex paralogous regions easier to review, including identifying full-gene deletions and resolving small variant haplotypes.



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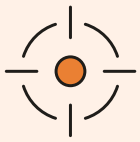
TruPath™ Genome

Accurate long-range insights.
One workflow. No trade-offs.

TruPath Genome redefines whole-genome sequencing by combining high-accuracy short-read performance with long-range genomic insight in a single, integrated workflow.

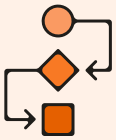
By delivering consistent accuracy across variant classes—without the complexity, variability, or trade offs of long-read approaches—TruPath enables researchers to scale comprehensive genome analysis with confidence.

Fewer incorrect data outputs. Greater confidence.



Accurate across all variant classes

Small variants, SVs, CNvs, STRs, and phasing—together.



Simplified and consistent

Streamlined process, reliable performance.

Built for comprehensive research

More genomic context. More complete genome insights.

Results you can trust

Comprehensive analysis. Fewer incorrect data outputs. Greater confidence.

Learn more about TruPath™ Genome

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