

TruPath™ Genome

Remove uncertainty in structural variants with spatial support

TruPath Genome colocation adds an independent, flow-cell-based signal to validate complex structural variants (SV) that cannot be confidently resolved by sequence information alone, including cases where long-read data may be limited or impractical at scale.

Colocation

Colocation is a powerful Illumina TruPath Genome capability that captures how DNA fragments physically appear together on the flow cell during sequencing. By adding spatial context beyond sequence data alone, colocation reveals long-range relationships that sequence-only approaches, including short- and long-read methods, may not fully capture.

This spatial context enables:

- Long-range insight within standard WGS workflows
- More confident interpretation of complex genomic patterns
- Easier identification and validation of SVs



Improve confidence in large structural variant calls



Visualize complex and large structural variants

Improving SV call confidence without loss of sensitivity

Confirming whether SV calls are real can be challenging when relying on sequence data alone. By combining DRAGEN™ SV calls with colocation signals, researchers can determine whether reads supporting a variant appear together spatially on the flow cell. This added spatial context helps distinguish true structural variants from false positives. With TruPath Genome, colocation eliminates approximately 80% of false positive translocations and inversions without impacting sensitivity, delivering higher confidence than long-read approaches (see Figure 1).

HG002 Putative False Positives

	w/ colocation (in release)	w/o colocation	PacBio
Inter-chromosomal	0	26	177
Intra-chromosomal	9	50	64

TruPath Coriell Samples PASS Event Counts (median, n=45)

	w/ colocation (in release)	w/o colocation	% Change
Inter-chromosomal	8	44	-81.8%
Intra-chromosomal	28	59	-52.5%

	w/ colocation (in release)	w/o colocation	% Change
True Positive Events	40	40	0%

Figure 1: Colocation-based filtering increases confidence in SV calls
TruPath Genome's colocation performance is shown compared to the lack of colocation and a competitor using the HG002 benchmark and TruPath Coriell samples.

Visualization of structural variation through colocation maps

DRAGEN generates a colocation matrix that visualizes long-range (>200 kb) proximity between reads, producing intuitive patterns that reveal inversions, translocations, and other complex structural rearrangements. This visualization provides structural insight beyond standard linear alignments, allowing researchers to see where reads co-occur on the flow cell and confirm true SVs.

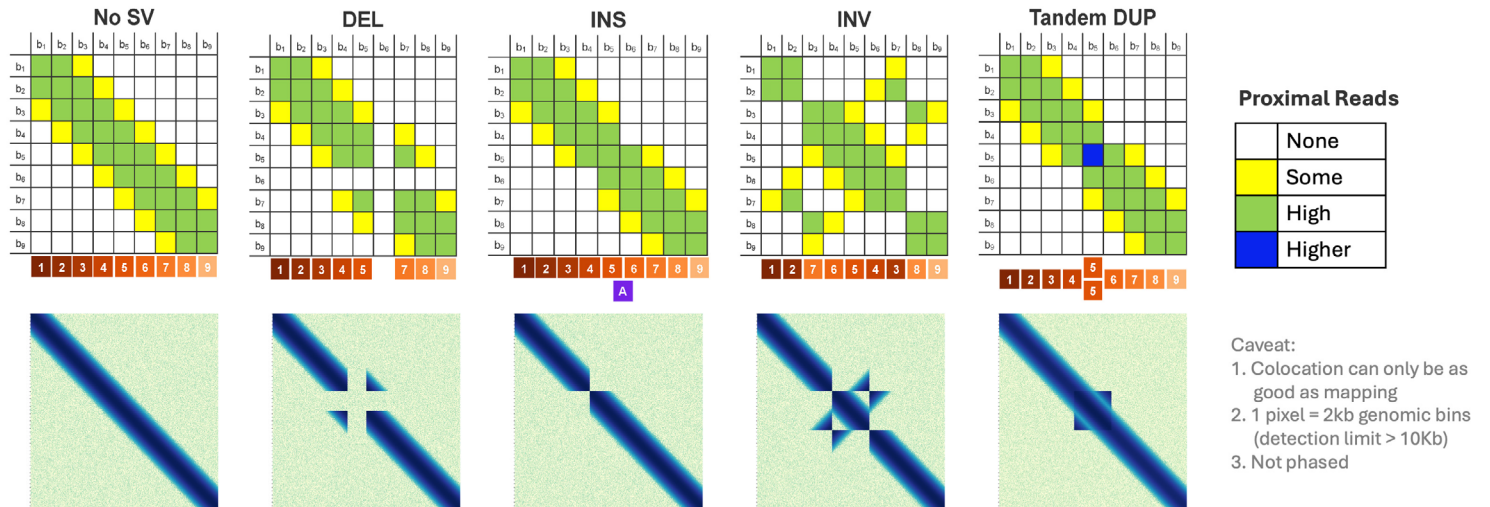


Figure 2: Easy-to-view SV patterns in colocation maps

This graphic demonstrates how different types of SVs — including deletions (DEL), insertions (INS), inversions (INV), and tandem duplications (Tandem DUP) — produce distinct, diagnostic patterns in a colocation map, which plots the frequency of proximity-linked reads between genomic bins.



Learn more about TruPath Genome

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