A diploid human genome (HG002) was sequenced on the Sequel II System to >25-fold coverage using a single SMRT Cell 8M in the CLR sequencing mode. Coverage was down sampled, and variants were detected using Structural Variant Calling analysis in SMRT Link and measured against the Genome in a Bottle v0.6 benchmark (HG002).
DATA ANALYSIS SOLUTIONS WITH SMRT ANALYSIS AND IGV

- Detect the broadest range of SV types including insertions, deletions, duplications, inversions, translocations, and copy number variants
- Discover SVs with the highest precision and recall
- Identify common SVs across multiple samples with joint calling
- Resolve breakpoints to sequence level
- Limit costly validation efforts with a low false discovery rate of <5%
- Output data in standard file formats—BAM and VCF—for seamless integration with downstream analysis tools
- Confirm SV calls visually with IGV<sup>8</sup> and GenomeRibbon<sup>11</sup>

SMRT SEQUENCING PROVIDES THE NECESSARY SENSITIVITY FOR SV DISCOVERY

![Visualization of long-read sequencing evidence in support of an insertion and deletion structural variant in IGV. Insertions are indicated with a purple box with the width of the box proportional to the size of the insertion, and the base pair size is written on the box. Deletions are indicated by a black line. The base pair size of the deletion is written on a white box at the center of the line.](image)

A comparison of recall for short-read sequencing and PacBio long-read sequencing for SV discovery in published human genomes<sup>2,12</sup>. PacBio long-read sequencing uncovers many SVs missed by short-read methods, providing up to five-fold higher recall<sup>8</sup>.

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KEY REFERENCES

4. SMRT Link user guide (v8.0). PacBio Documentation.
6. Data Set: Structural variants calls from one Sequel II System SMRT Cell 8M for HG002.