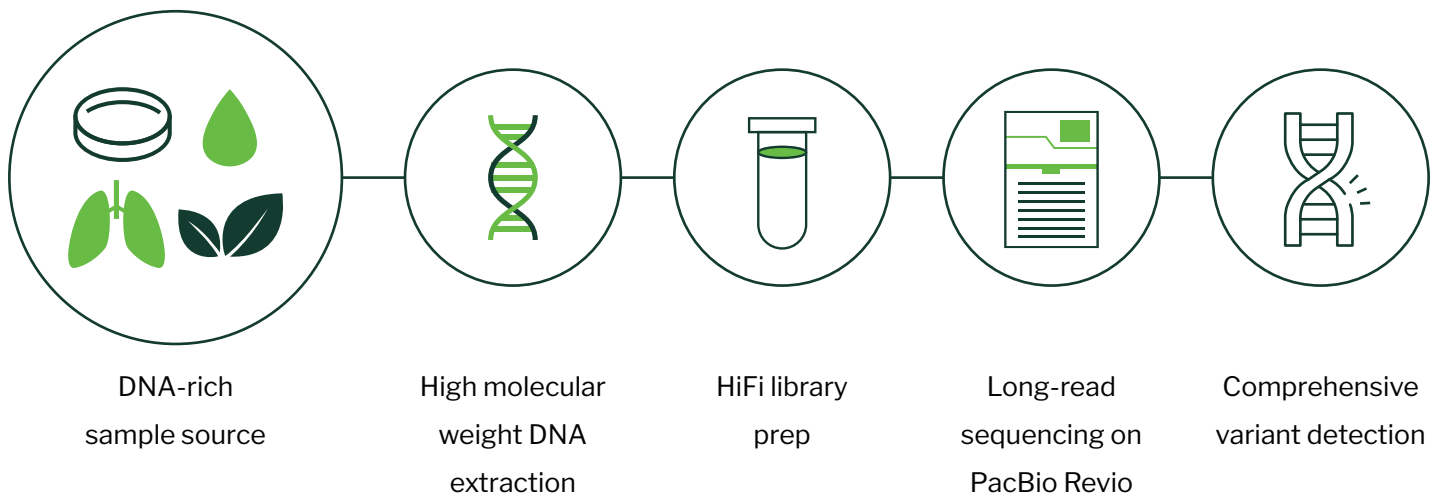


## Unmasking the full spectrum of human genetic variation with PacBio HiFi sequencing

Understanding the complete landscape of human genetic variation is crucial for advancing disease research, enabling precision medicine, and unraveling the complexities of human evolution. While short-read sequencing technologies have been instrumental in identifying single nucleotide variants (SNVs), they often fall short in detecting and characterizing complex structural variants (SVs) and variations in difficult-to-sequence regions. PacBio HiFi sequencing, with its long reads and high accuracy, overcomes these limitations, providing a comprehensive view of the human genome and its variations.

Signios Bio offers a powerful solution for comprehensive human variant detection using PacBio HiFi sequencing. Our integrated workflow combines cutting-edge technology with expert bioinformatics analysis, enabling researchers to uncover the full spectrum of genetic variation, from single base changes to complex genomic rearrangements.

### A seamless workflow for unbiased variant discovery



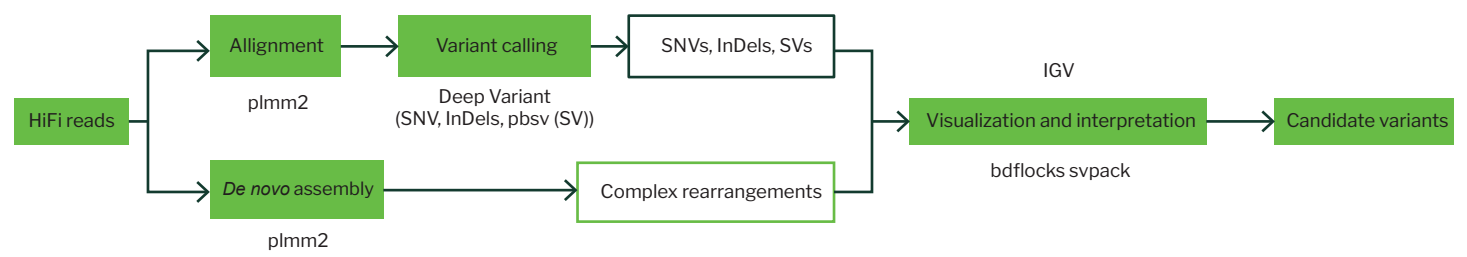
### Benefits of our integrated approach

- **Unparalleled variant detection:** Detect the full spectrum of genetic variation with high accuracy, including challenging SVs and variants in complex genomic regions often missed by short-read sequencing.
- **Comprehensive genomic coverage:** Achieve uniform coverage across the entire genome, minimizing biases and enabling confident variant calling even in GC-rich and repetitive regions.
- **Haplotype resolution:** Distinguish variants on homologous chromosomes, providing valuable insights into allele-specific expression and compound heterozygosity.
- **Streamlined workflow:** Benefit from a single, integrated solutions provider for all your variant detection needs, saving time and resources.
- **Expert support:** Our team of experienced scientists and bioinformaticians provides expert guidance and support throughout the entire project.

# Human whole genome variant detection analysis solutions

Our dedicated team of bioinformaticians utilizes a robust pipeline for:

- **Genome alignment and variant calling:** Accurate mapping of HiFi reads to the reference genome and identification of diverse variant types with high confidence.
- **Variant annotation and interpretation:** Comprehensive annotation of variants using multiple databases and prediction algorithms to assess their potential functional impact.
- **Visualization and reporting:** Clear and intuitive visualization of variants across the genome and generation of detailed reports summarizing variant findings.






## Specifications for human whole genome variant detection projects

Coverage		10-fold	15-fold	30-fold
Samples/SMRT Cell		3	2	1
Input DNA		≥5µg	≥5µg	≥5µg
Variant detection performance (% accuracy F1)	SNVs	99.0%	99.8%	99.9%
	InDels	92.6%	96.9%	97.9%
	SVs	92.6%	95.7%	95.7%

## Signios Bio: Your partner in comprehensive variant discovery

Signios Bio's comprehensive human variant detection service using PacBio HiFi sequencing empowers researchers with an unprecedented view of the human genome. Our integrated solution delivers highly accurate and comprehensive variant data, facilitating a deeper understanding of human health, disease, and evolution. Contact us today to discuss how we can help you unlock the full potential of your genomic data and advance your research or clinical goals.

Contact us today for pricing and to begin planning your customized solution.

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