

HIFI SEQUENCING WITH TWIST BIOSCIENCE TARGET ENRICHMENT

Targeted HiFi sequencing at scale with ready-made Twist Alliance Panels or custom designs

Combine **Twist Bioscience** target enrichment with **PacBio®** long and accurate HiFi reads to efficiently sequence your priority genomic regions at scale. Sequence enriched regions with a protocol optimized for HiFi reads to get comprehensive detection of single nucleotide variants, structural variants, and indels with haplotype resolution. HiFi target enrichment can deliver accurate alleles for complex gene families such as immune genes (e.g., *HLA*) and pharmacogenes (e.g., *CYP2D6*).



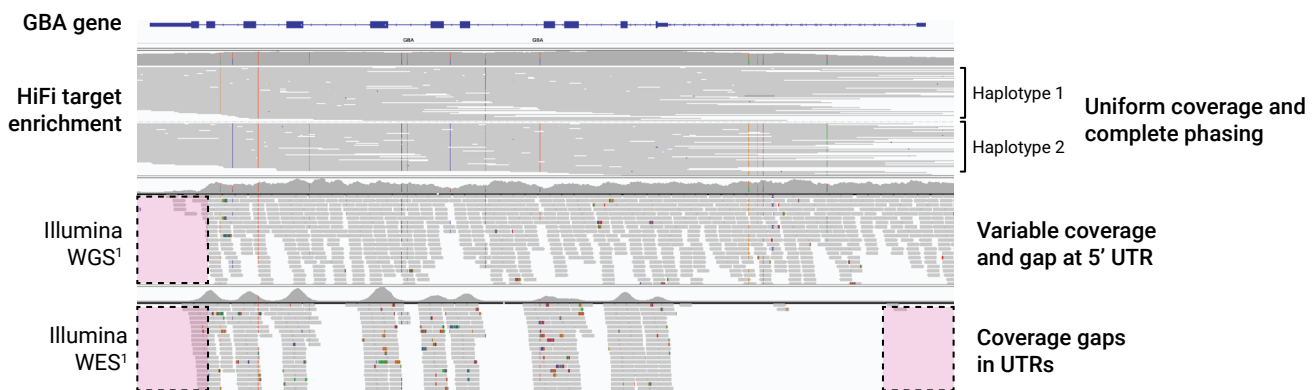
Why choose HiFi sequencing with Twist target enrichment?

- End-to-end workflow optimized for HiFi reads
- Custom design for any panel size
- Small and large cohorts on a single SMRT® Cell
- High-accuracy variant calls, including SVs
- Direct phasing, unambiguous haplotypes, ancestry-agnostic discovery

Pool multiple samples per SMRT Cell

Panel example	20 Mb	2 Mb	100 kb
Estimated gene number	400	50	< 10
Samples/Vega™ SMRT Cell	12	72	288
Samples/Revio® SPRQ™ SMRT Cell (run)	16 (64)	96 (384)	384 (1536)
Samples/Sequel® IIe SMRT Cell	4	24	96

Full gene coverage of medically relevant genes



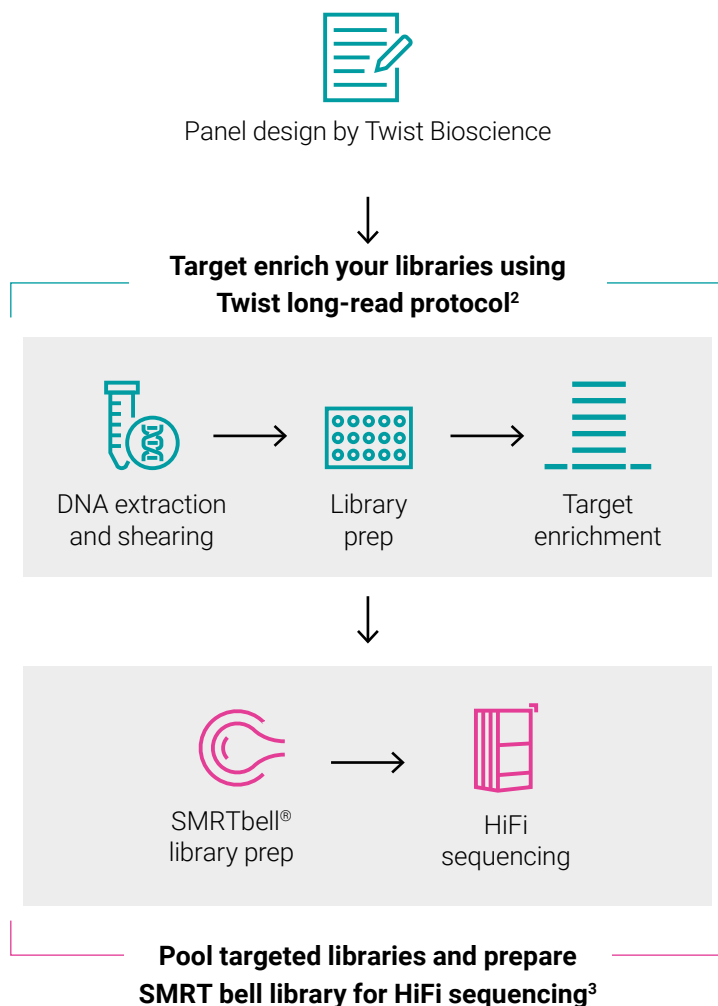
HG0001 GRCh38 chr1: 155,234,000–155,245,000 (11 kb)

Performance for Twist Alliance Panels

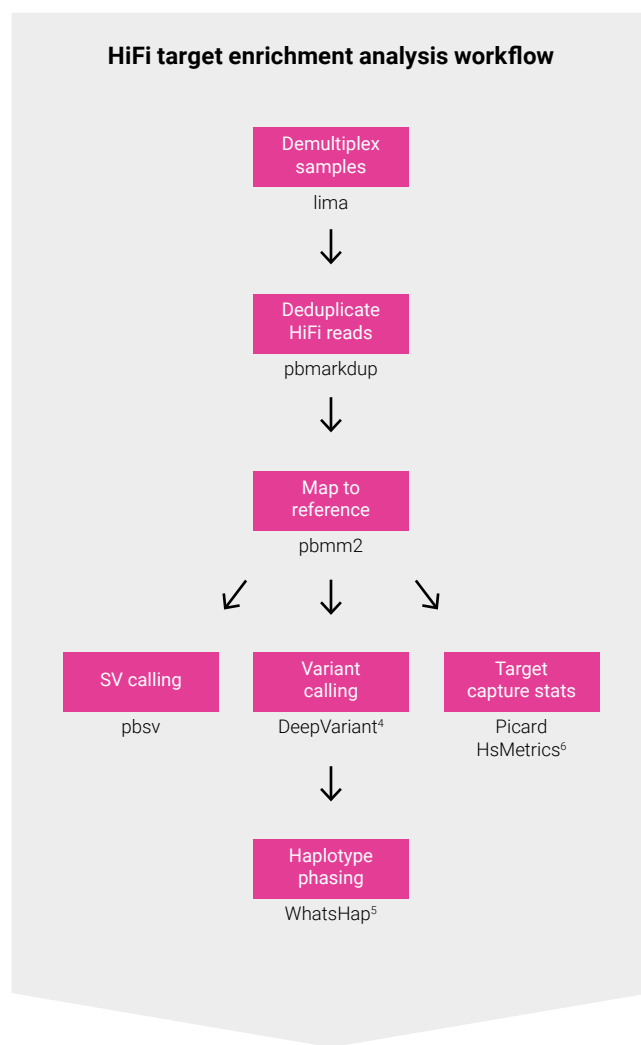
Twist Alliance Panel	Panel Size (Mb)	Number of genes	PacBio system	Samples per SMRT Cell	Mean target coverage	Fold enrichment	Target bases ≥10-fold read depth	Mean HiFi read length	Duplicate rate
Dark Genes	22	389	Revio	12	75-fold	65-fold	93%	5.5 kb	3%
Dark Genes	22	389	Sequel IIe	4	75-fold	54-fold	93%	5.2 kb	3%
Long Read PGx	2	49	Sequel IIe	24	190-fold	784-fold	99%	5.3 kb	2%

Latest datasets at <https://www.pacb.com/connect/datasets/#targeted-datasets>

From targets to HiFi reads



Supported analysis in SMRT[®] Link



KEY REFERENCES

- <https://www.biorxiv.org/content/10.1101/2020.12.11.422022v1.full> (HG002. novaseq.wes-agilent.50X)
- <https://www.twistbioscience.com/resources/protocol/long-read-library-preparation-and-standard-hyb-v2-enrichment>
- Procedure & checklist – Preparing-multiplexed-amplicon-libraries-using-SMRTbell-prep-kit-3.0** PacBio documentation.
- <https://github.com/google/deepvariant>
- <https://whatshap.readthedocs.io/en/latest/>
- <https://snakemake-wrappers.readthedocs.io/en/stable/wrappers/picard/collecthsmetrics.html>



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