The PacBio® Systems combine single-molecule resolution, long reads, and uniform coverage, to fully characterize genetic complexity – including structural variants, indels, copy number variants, microsatellites, somatic variants, haplotypes, and phasing. Move beyond simply cataloging SNPs and target all types of variation across relevant genomic regions, including low complexity regions like repeat expansions, promoters, and flanking regions of transposable elements.

- Phase variants with allele specificity
- Sequence through low-complexity and highly repetitive regions
- Deconvolute haplotypes of complex populations with linear quantitation and minor variant detection below 0.1%
- Resolve structural variation breakpoints
- Accurately determine heterozygosity through de novo consensus generation
- Validate sequence variants cost-effectively and with high accuracy

RESOLUTION OF REPEAT EXPANSIONS

1. SMRT® Sequencing reads of the Fragile X region containing (CGG)n trinucleotide repeats. An AGG repeat interruption (boxed) is unambiguously recognized in all reads.
2. A full Fragile X mutation allele of 750 CGG repeats.

SINGLE-BASE RESOLUTION OF COMPLEX MIXTURES OF FULL-LENGTH GENOMES

Estimated haplotypes and relative abundances of five nearly identical full-length HIV-1 genomes (9.6 kb) from SMRT® Sequencing of a synthetic mixture of HIV-1 clones differing by only 1–2 bases.
Sequencing with PacBio Systems

- Take advantage of the Sequel™ System to reduce project costs and generate 7X more reads compared with the PacBio RS II
- Achieve ~10 kb average read lengths, with some reads as long as 60 kb
- Adjust run times (0.5 to 6 hours) to maximize sample throughput and turn-around time
- Attain consensus accuracies > 99.999% by avoiding mapping and systematic errors
- Produce high intra-molecular accuracies with multiple observations of single circularized templates for complex population resolution

Data analysis with SMRT Analysis or PacBio DevNet

- Long Amplicon Analysis: generate reference-agnostic phased consensus assemblies from heterogeneous pools of amplicons
- Minor Variant Analysis: detect and quantify SNPs in a heterogeneous data set against a reference
- ClusterConsensus: reference-free deconvolution of genomes in a complex mixture
- HGAP: create high-quality de novo assemblies of plasmids, BACs or fosmids

Library Preparation

- Prepare SMRTbell™ libraries for a variety of insert sizes (250 bp to 40 kb)
- Utilize standard target-enrichment methods
- Multiplex 384 barcoded samples
- Enrich for longest inserts with size-selection options
- Support available for library automation

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KEY REFERENCES


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