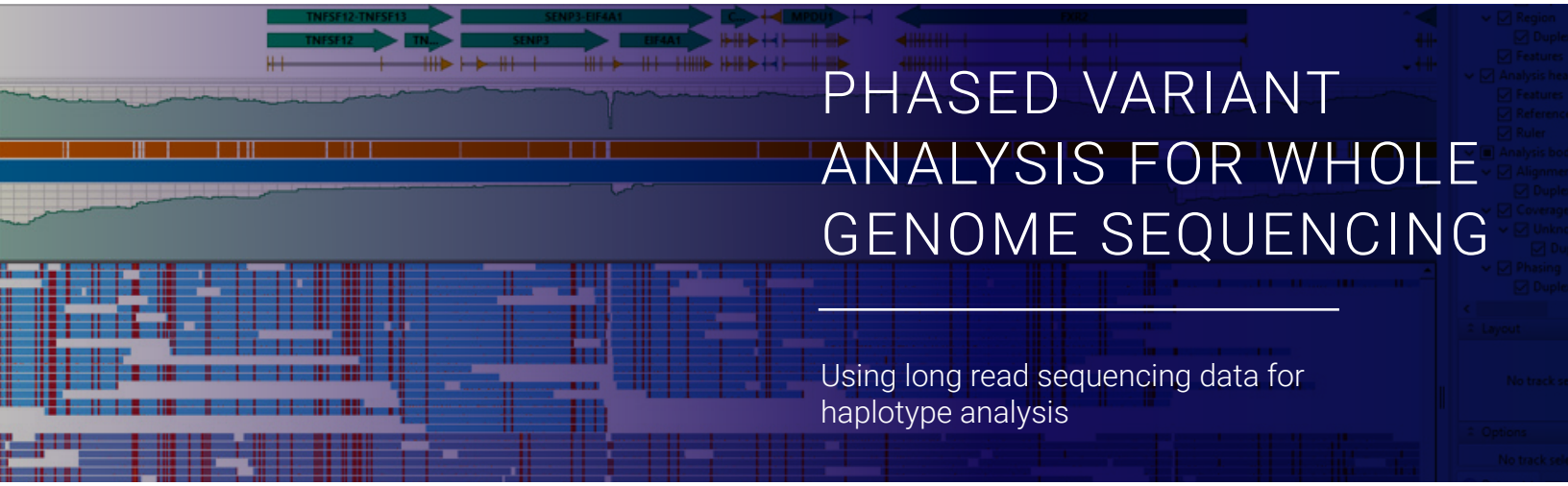
DNASTAR LASERGENE



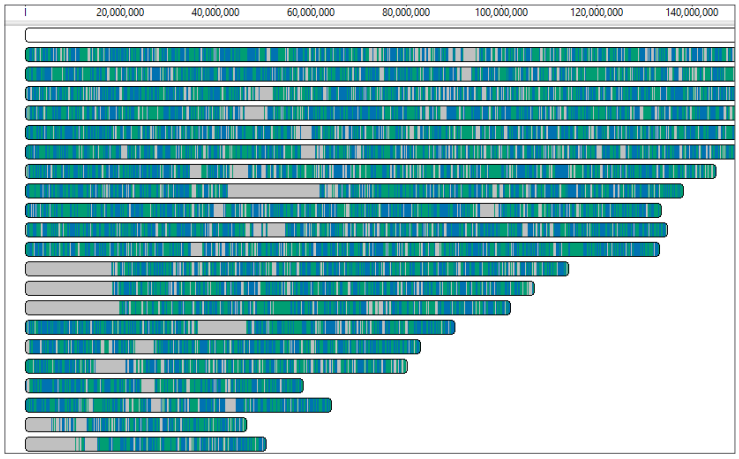
PHASED VARIANT ANALYSIS FOR WHOLE GENOME SEQUENCING

Using long read sequencing data for haplotype analysis

Next-generation sequencing (NGS) has played a pivotal role in identifying candidate genes and variants associated with diseases and traits across cohorts, populations, and individual clinical samples. Short-read sequencing (SRS) methods excel in accurately detecting individual SNP variants. However, SRS data is inadequate for haplotype phasing and for resolving larger structural variants. These limitations can be overcome with long read data from Pacific Biosciences (PacBio) or Oxford Nanopore Technologies (ONT), which provide the long-range connectivity needed for haplotype phasing and structural variant detection. But until now, assembly and analysis of this data has required users to string together multiple software applications.

To address these challenges, DNASTAR has introduced several new features in Lasergene 18 that enable end-to-end assembly and analysis of long-read sequencing data from human samples, including the identification of compound heterozygotes.

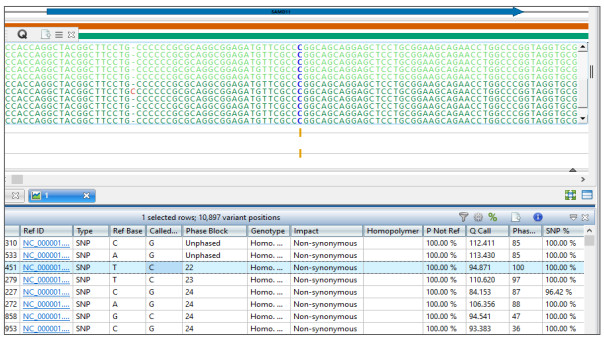
These flexible tools provide a comprehensive solution bridging the gap between SRS and LRS technologies, empowering clinical researchers with efficient, scalable, and intuitive tools for genomic analysis.



Genome View in GenVision Pro showing phasing across each chromosome.

Advantages to Phased Variant Analysis

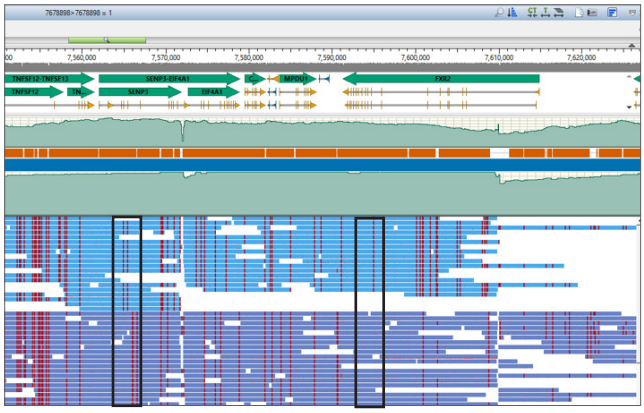
- Identify genes that are inherited together (linked) because they are located close to each other on the same chromosome, enabling a deeper understanding of the genetic basis of complex diseases.
- Trace the ancestry of individuals and populations through haplotype analysis.
- Study the genetic diversity of populations and track the evolution of genetic variations over time.
- Determine whether or not there is at least one functional allele in cases of compound heterozygosity.
- Predict how a patient will respond to certain medications, leading to more personalized and effective treatments.



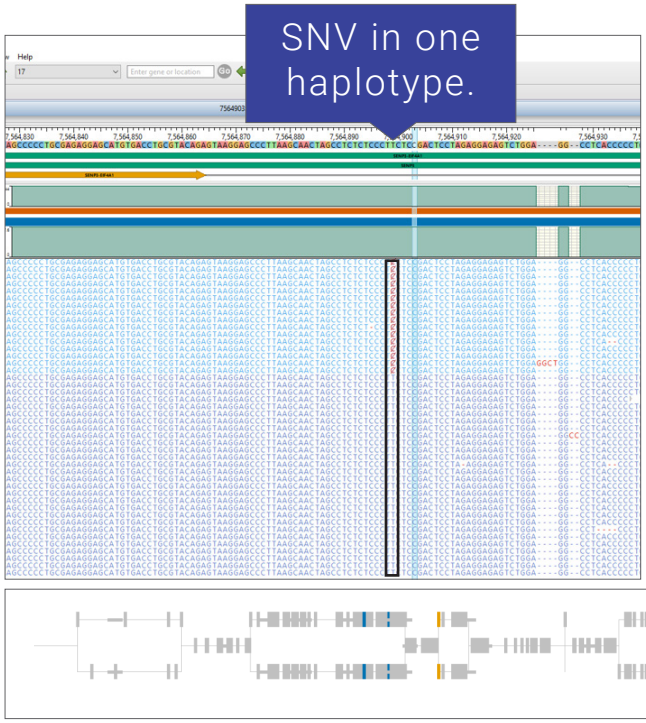
Integrated variant table and alignment view for phased genome assembly. This variant table can be used to apply filters and locate variants in the alignment view.

Results

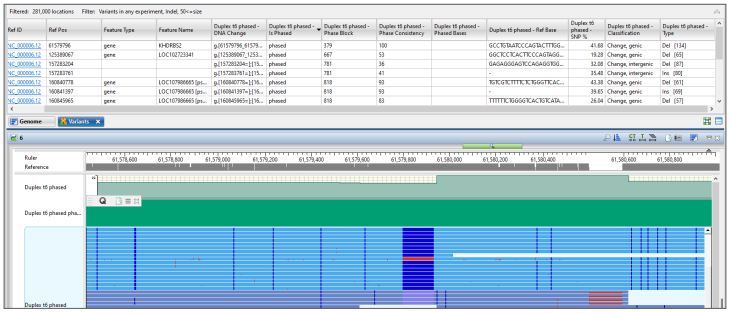
GenVision Pro uses computational algorithms to separate the parent chromosomes and analyze the genetic variants on each. Phase blocks are then correlated to one or more genes of interest to identify compound heterozygous variants. The Analysis View allows you to view the alignment of reads phased into the two sister chromosomes, one in light blue, the other in dark blue. SNVs are indicated as red tick marks. The two black boxes contain examples of heterozygous SNVs that have been phased.



You can zoom in to see a more granular view of the alignment. Here, you can see a variant in one of the haplotypes.



At certain zoom levels, the Variants track is bifurcated at each phase block to show the variants in each of the two alleles (parents).



The Variants table can also be used to locate larger variants and structural variants.

Workflow

All the steps for the haplotype phasing workflow can be completed on a Windows or Mac computer, quickly taking you from raw sequencing data to results.

01

Assemble Sequencing Data

Use SeqMan NGen to assemble sequence reads to the human genome template. These assemblies contain annotated small variant calls, SV characterization, and haplotype phasing information.

02

View and Filter Variants

Access small variant and structural variant tables in GenVision Pro. Use the integrated filtering and statistical tools to identify variants and genes within individual samples or experimental sets.

03

Visualize Results

Use the compact variant track, phased read alignment track, and phasing consistency plots alongside the integrated, customizable genome browser to dive deeper into genes and variants of interest.

04

Import and Export

Optionally, import and export BAM alignments and VCF files for straightforward comparison of sequence data processed in other assembly and analysis pipelines.

Free Trial

Obtain a fully functional, free trial version of Lasergene to try this workflow on your laptop or desktop computer.

www.dnastar.com/freetrial

Contact Us

1.608.258.7420 tel
1.866.511.5090 U.S., toll free
0.808.271.1041 U.K.
0.800.182.4747 Germany

www.dnastar.com
info@dnastar.

1202 Ann Street
Madison, WI