

PacBio Announces Complete Computational Workflow for Human Whole Genome Sequencing Data Analysis

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PacBio WGS Variant Pipeline Will Bring Standardization to PacBio HiFi Data Analysis

MENLO PARK, Calif., Oct. 11, 2023 /PRNewswire/ -- PacBio (NASDAQ: PACB), a leading developer of high-quality, highly accurate sequencing solutions, today announced the availability of PacBio WGS Variant Pipeline – a complete, standardized computational method for HiFi whole genome sequencing (WGS) data analysis. The new software pipeline will enable customers to resolve many different variant types, including single-nucleotide polymorphisms, insertions and deletions, structural variants, tandem repeats, segmental duplications, and copy number variants, in addition to providing methylation and phasing data, all in a single bioinformatic solution – making it the most complete human WGS secondary analysis pipeline available. The single computational workflow integrates PacBio and third-party tools, including TRGT, Paraphase, and Google DeepVariant, in an intuitive user interface, offering customers a best-practice for HiFi WGS analysis.



Previously, PacBio customers would need to use multiple bioinformatic tools to analyze HiFi data. With the new PacBio WGS Variant Pipeline, PacBio offers a streamlined and supported tool to access alignment, variant calling, joint calling, and genome annotation data across Amazon Web Services, Google Cloud, Azure, and on-premise high performance computing solutions.

"PacBio is focused on developing the most advanced sequencing technologies. This mission extends not only to providing our customers game-changing sequencers, but also delivering comprehensive analytical tools to interpret sequencing data," said Christian Henry, President and Chief Executive Officer of PacBio. "This is the first and most complete HiFi pipeline that integrates PacBio and third-party tools into a single, high-powered, bioinformatics solution, which will help our customers realize the full power of our Revio system and HiFi data, and allow them to spend more time on biological discovery."

"In our initial testing, we've found the PacBio WGS Variant Pipeline could be quickly adapted by running samples through the workflow to advance our rare disease-focused research," said Tomi Pastinen, MD, PhD., Director, Genomic Medicine Center, Children's Mercy Kansas City. "We also appreciate that the workflow is compatible with additional, optional tools to run in select samples, for example, personal assemblies."

These workflows are accessible through bioinformatics analysis platforms from DNAnexus, Form Bio, Terra, and DNAstack. These platforms have been vetted by both the PacBio and DNAstack computational biology teams and allow users to run bundled workflows in a turn-key manner. DNAstack, DNAnexus and Form Bio also enable users to access other PacBio bioinformatic workflows, including AAV QC, TRGT and Paraphase.

"PacBio making these streamlined and runnable workflows expands access to informatics methods through the community," said Andrew Carroll, Genomics Product Lead for Health AI at Google. "Google is honored for DeepVariant to be included, and that PacBio and ourselves share the value of democratizing access to genome analysis at high accuracy."

PacBio WGS Variant Pipeline will be available through GitHub. For more information about the PacBio WGS Variant Pipeline, visit https://bit.ly/3rlmiNO.

About PacBio

PacBio (NASDAQ: PACB) is a premier life science technology company that is designing, developing and manufacturing advanced sequencing solutions to help scientists and clinical researchers resolve genetically complex problems. Our products and technologies stem from two highly differentiated core technologies focused on accuracy, quality and completeness which include our HiFi long-read sequencing and our SBB® short-read sequencing technologies. Our products address solutions across a broad set of research applications including human germline sequencing, plant and animal sciences, infectious disease and microbiology, oncology, and other emerging applications. For more information, please visit www.pacb.com and follow @ PacBio.

PacBio products are provided for Research Use Only. Not for use in diagnostic procedures.

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