



## PacBio and International Research Consortium CoLoRS Announce Release of First-Ever HiFi Long-Read Variant Database

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*Consortium Delivers Publicly Available Datasets that Enrich Detection of Rare and Novel Genetic Variants for Advances in Human Disease Research*

MENLO PARK, Calif., June 10, 2024 /PRNewswire/ -- PacBio (NASDAQ: PACB), a leading developer of high-quality, highly accurate sequencing solutions, in collaboration with the international Consortium for Long-Read Sequencing (CoLoRS), today announced the launch of the first publicly available and free HiFi long-read variant frequency database with global representation. This innovative resource fills a critical void for rare disease researchers, providing access to genetic variants that are not detectable through short-read sequencing methods. By enabling the filtering of common structural variants from long-read sequencing data, this database allows for the identification of rare and novel variants in rare disease cases. The launch of this database better enables researchers to utilize long-read genomic data to advance human health research.

"Joining forces with PacBio has enabled us to leverage their HiFi sequencing capabilities, enhancing the depth and accuracy of the genomic data we can offer to the research community," said Michael Schatz, Bloomberg Distinguished Professor at Johns Hopkins University. "This effort complements existing databases such as gnomAD, as long reads enable much greater sensitivity and precision for complex variants (SVs) and tandem repeats. The CoLoRS database is a tremendous step forward in our collective effort to understand complex genetic variations and their implications in disease."

Studies have shown that long-read sequencing finds 15,000 more structural variants (SVs) and 300,000 more small variants than short reads. SVs are crucial as they account for much of the genetic diversity between humans and are associated with disease pathogenesis. The CoLoRS database integrates data from nearly 1,000 long-read genomes, providing detailed insights into human variation and improving the detection of complex variants and tandem repeats. This resource is developed by a global coalition from top clinical and academic institutions, pooling diverse long-read genome data from various research projects.

"We are thrilled to see the launch of the CoLoRS database, a pivotal development in genomics research," said Christian Henry, President, and Chief Executive Officer of PacBio. "This collaboration with the CoLoRS members exemplifies our commitment to supporting the scientific community with tools that enhance the accuracy and utility of genetic research. It also demonstrates the unique capabilities of our HiFi sequencing technology. By making this information accessible to the public, we are empowering researchers around the world to make significant advances in areas such as oncology, rare diseases, and genetic disorders."

Researchers are invited to access the CoLoRS database and explore its capabilities at [www.colorsdb.org](http://www.colorsdb.org). Supported by PacBio's HiFi sequencing technology, this platform provides researchers with new insights into human genomic variation, accelerating advancements in critical areas of medical research.

### About PacBio

PacBio (NASDAQ: PACB) is a premier life science technology company that designs, develops, and manufactures 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](http://www.pacb.com) and follow @PacBio.

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

### Forward-Looking Statements

This press release may contain "forward-looking statements" within the meaning of Section 21E of the Securities Exchange Act of 1934, as amended, and the U.S. Private Securities Litigation Reform Act of 1995. All statements other than statements of historical fact are forward-looking statements, including statements relating to the uses, coverage, advantages, quality or performance of, or benefits or expected benefits of using, PacBio products or technologies; enabling researchers to utilize long-read genomic data to advance human health research; empowering researchers to make significant advances in areas such as oncology, rare diseases, and genetic disorders; and other future events. You should not place undue reliance on forward-looking statements because they are subject to assumptions, risks, and uncertainties and could cause actual outcomes and results to differ materially from currently anticipated results, including, challenges inherent in sequencing a large number of whole human genomes, and the difficulty of generating discoveries across various areas of research; unanticipated increases in costs or expenses; interruptions or delays in the supply of components or materials for, or manufacturing of, PacBio products and products under development; potential product performance and quality issues; third-party claims alleging infringement of patents and proprietary rights or seeking to invalidate PacBio's patents or proprietary rights; and other risks associated with international operations. Additional factors that could materially affect actual results can be found in PacBio's most recent filings with the Securities and Exchange Commission, including PacBio's most recent reports on Forms 8-K, 10-K, and 10-Q, and include those listed under the caption "Risk Factors." These forward-looking statements are based on current expectations and speak only as of the date hereof; except as required by law, PacBio disclaims any obligation to revise or update these forward-looking statements to reflect events or circumstances in the future, even if new information becomes available.

### Contacts

Investors:

Todd Friedman

[ir@pacificbiosciences.com](mailto:ir@pacificbiosciences.com)

Media:

[pr@pacificbiosciences.com](mailto:pr@pacificbiosciences.com)

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