



HiFi Reads Enable Telomere-to-Telomere Consortium to Fill the Final Gaps in the Human Genome, Heralding a New Era of Research into Genetic Diseases and Human Biology

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Science magazine publication details how the human genome was finally completed with the help of PacBio long-read technology

MENLO PARK, Calif., March 31, 2022 (GLOBE NEWSWIRE) -- PacBio (NASDAQ: PACB), a leading provider of high-quality, highly accurate sequencing solutions, announced today PacBio's essential contribution to the completion of the first truly complete human reference genome. The [Telomere-to-Telomere \(T2T\) Consortium](#), an international team of scientists from the US National Human Genome Research Institute and dozens of other institutions worldwide, published "[The complete sequence of a human genome](#)" in the latest edition of *Science*, along with a series of companion papers highlighting novel aspects of the complete genome. As reported in the *Science* paper, researchers searching for the genetic keys to unlock complex regions of the genome to variational and functional analysis, including in connection with genetic disease, now have the first truly complete human reference genome.

The paper describes how scientists used HiFi sequencing to produce the first complete assembly of a human genome: all three billion base pairs across 23 chromosomes including the centromeres, telomeres, and other large repeats. The newly assembled regions cover a full eight percent of the human genome previously inaccessible with earlier technologies.

"The T2T Consortium's work could help scientists better understand human biology and evolution. Most importantly, it could one day ultimately change the lives of millions of people by helping researchers better understand the genomic basis associated with potentially dozens of genetic diseases, and eventually identify cures or therapeutic options for them," said Christian Henry, President and Chief Executive Officer of PacBio. "As reported in this exciting paper, by leveraging PacBio HiFi sequencing technology, the T2T Consortium has achieved a goal that scientists around the globe have been pursuing for more than 30 years – a truly complete human genome sequence, one that can potentially help create the basis for a complete medical genome to better human health. We look forward to seeing other scientists follow in their path helping the community further understand the full breadth of genetic diversity and its link to human health."

The complete, T2T assembly of a human genome marks a new era of genomics where no region of the genome is beyond reach. This milestone was made possible by HiFi sequencing, a cutting-edge technology capable of reading long fragments of DNA at higher accuracy than other known long-read sequencing technologies. Prior technologies provided either length or accuracy, but both length and accuracy are needed to elucidate the most difficult regions of the genome.

The paper explains that T2T's new reference genome known as T2T-CHM13 includes gapless assemblies for all 22 autosomes plus Chromosome X, corrects numerous errors, and introduces nearly 200 million base pairs of novel sequence representing the largest improvement to the human reference genome since its initial release.

Scientists in the T2T Consortium applied the new reference to look at genetic variation in 3,202 globally diverse individuals, identifying hundreds of thousands of new variants per genome. The improvements are particularly large for some critical duplicated genes such as *FRG1*, which is linked to facioscapulohumeral muscular dystrophy (FSHD). The T2T reference has 23 copies of *FRG1*, up from an incomplete set of nine copies in the prior reference. The result is a T2T reference that, in this regard, provides a better foundation for study of this disease.

The success of the T2T-CHM13 genome assembly has also inspired researchers in the Human Pangenome Reference Consortium (HPRC) to generate more than 300 human reference quality genomes from diverse backgrounds in the next three years. The individual genomes will be combined into a pangenome to provide a reference that better represents human diversity and will drive genomic discoveries for everyone. As recently presented in a [preprint](#) from the HPRC, PacBio HiFi sequencing technology is critical to producing complete, accurate genomes and pangenomes.

"The completion of the human genome sequence is a huge milestone in scientific history, the culmination of work started by the Human Genome Project in the 1990s. It shows that technology like HiFi sequencing that was developed after the initial Human Genome Project has brought not only cheaper genomes but also higher quality ones," said Jonas Koriach, Ph.D., Chief Scientific Officer at PacBio. "This milestone illustrates the power of HiFi sequencing to resolve difficult regions of the genome and its potential to revolutionize the way we understand genetic disease. In addition, a more complete reference genome may translate to more discovery, setting a new standard in genomics built on PacBio HiFi sequencing."

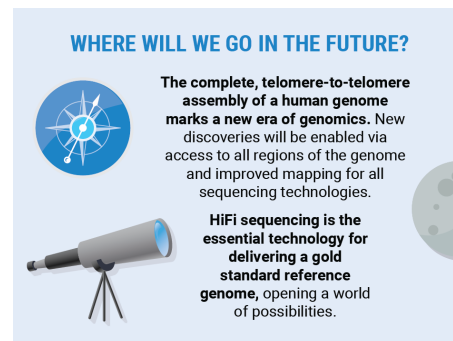
There is an infographic available here ([PDE](#)) and you can learn more about [whole-genome sequencing](#), the benefits of [HiFi Sequencing](#), and applications for [SMRT Sequencing](#) on the [PacBio website](#).

About PacBio

Pacific Biosciences of California, Inc. (NASDAQ: PACB) is empowering life scientists with highly accurate sequencing platforms. The company's innovative instruments are based on Single Molecule, Real-Time (SMRT®) Sequencing technology, which delivers a comprehensive view of genomes, transcriptomes, and epigenomes, enabling access to the full spectrum of genetic variation in any organism. Cited in thousands of peer-reviewed publications, PacBio® sequencing systems are in use by scientists around the world to drive discovery in human biomedical research, plant and animal sciences, and microbiology. For more information, please visit www.pacb.com and follow @PacBio.

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Where will we go in the future?



The complete, telomere-to-telomere assembly of a human genome marks a new era of genomics.

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, including statements relating to future availability, uses, accuracy, advantages, quality or performance of, or benefits or expected benefits of using, PacBio products or technologies; the suitability or utility of such products or technologies for particular applications or projects, including in connection with the T2T Consortium’s research; potential increases in variant detection and providing answers for rare disease research, in the T2T research in particular and rare disease and genetic diversity research in general, including in connection with potentially changing the lives of people by, among other things, better understanding the genomic basis associated with genetic diseases and diversity; statements regarding all regions of the genome being within reach for sequencing; the HPRC’s anticipated sequencing efforts over the next three to provide a reference genome that better represents human diversity; and other future events. Readers are cautioned not to place undue reliance on these forward-looking statements and any such forward-looking statements are qualified in their entirety by reference to the following cautionary statements. All forward-looking statements speak only as of the date of this press release and are based on current expectations and involve a number of assumptions, risks and uncertainties that could cause the actual results to differ materially from such forward-looking statements. Readers are strongly encouraged to read the full cautionary statements contained in the Company’s filings with the Securities and Exchange Commission, including the risks set forth in the company’s Forms 8-K, 10-K, and 10-Q. The Company disclaims any obligation to update or revise any forward-looking statements.

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A photo accompanying this announcement is available at <https://www.globenewswire.com/NewsRoom/AttachmentNg/363c6448-8c30-468e-b198-47d26f67a942>