Ambry Genetics and PacBio Announce Collaboration to Sequence Up to 7,000 Human Genomes Aimed at Providing Answers for Families Battling Rare Diseases

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Consortium Selects Leading Genomics Companies to Support Pediatric Mendelian Genomics Research Center Program

ALISO VIEJO, Calif. and MENLO PARK, Calif., May 15, 2024 /PRNewswire/ --Ambry Genetics, a prominent leader in clinical genomic testing and a subsidiary of REALM IDx, Inc., along with PacBio (NASDAQ: PACB), a leading developer of high-quality, highly accurate sequencing solutions, today announced their companies' selection by the University of California, Irvine (UCI) and the GREGoR Consortium (Genomics Research to Elucidate the Genetics of Rare diseases) to support the Pediatric Mendelian Genomics Research Center (MGRC) program to better understand the underlying biology of rare diseases.

The GREGoR Consortium is a National Institutes of Health-funded collaborative effort which aims to transform the landscape of Mendelian disease research by identifying the underlying genetic cause of rare disease in samples from individuals for whom prior genomic analysis did not yield answers. This ambitious research, among the largest programs of its kind, will use long-read sequencing technology to sequence and analyze up to 7,000 human whole genomes over three years, with a focus on developing new insights into rare disease etiology.

"There remain a multitude of rare diseases that are difficult to diagnose, and for which effective treatments remain elusive," said Eric Vilain M.D., Ph.D., director of the Institute for Clinical and Translational Science and the associate vice chancellor for Clinical and Translational Science at UCI. "Our research endeavors aim to shed light on these complexities, revealing insights that legacy technologies struggle to uncover. Collaborating with our partners at Ambry Genetics and PacBio, we are poised to enhance our comprehension of rare diseases and in the future revolutionize diagnostic capabilities. This collaborative effort is designed to offer hope not only to families in our study, but to all families looking to unlock answers for children facing rare diseases."

This pioneering initiative unites leading genomics researchers who will work collaboratively to incorporate innovative methods for understanding the biology of rare disease including phenotyping, variant identification, and functional analysis of both coding and non-coding sequence alterations. By using highly accurate 5-base, long-read sequencing technology, the researchers hope to discover new rare variants and to understand the role of epigenomics on disease manifestation. By building new analysis pipelines for these genomic and epigenomic data, the researchers hope to discover new Mendelian gene variations and to better categorize previously identified variants of unknown significance.

"Over the past few years, we've collaborated with leading genomics researchers around the world to advance the scientific community's understanding of the genomic basis of rare disease," said Christian Henry, President and Chief Executive Officer of PacBio. "This project with the GREGoR team represents a significant step forward for us. We hope that by partnering with scientists at U.C. Irvine and geneticists from Ambry Genetics, we will not only be able to help families better understand the underlying causes of rare disease, but also to identify new analysis pipelines that can speed this process for other labs."

"Through this collaboration, we will continue to advance the scientific community's understanding of rare disease and to support both patients enrolled in this study and others whose exomes were sequenced previously through our Patient for Life program," said Tom Schoenherr, CEO of Ambry Genetics. "This collaboration is an example of our steadfast commitment to excellence in genomics and relentless pursuit of innovation, which has been a driving force behind our work since we launched our first clinical offering for rare disease diagnosis more than 20 years ago."

For more information, visit the GREGoR Consortium.

About Ambry Genetics®
Ambry Genetics, a subsidiary of REALM IDx, Inc., translates scientific research into clinically actionable test results based upon a deep understanding of the human genome and the biology behind genetic disease. It is a leader in genetic testing that aims to improve health by understanding the relationship between genetics and disease. Its unparalleled track record of discoveries over 20 years, and growing database that continues to expand in collaboration with academic, corporate, and pharmaceutical partners, means Ambry Genetics is first to market with innovative products and comprehensive analysis that enable clinicians to confidently inform patient health decisions.

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 and follow @PacBio.

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

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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; anticipated number of whole human genomes to be sequenced in the collaboration and related discoveries in rare disease research; 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

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