

HudsonAlpha Using PacBio Sequencing for Childhood Developmental Disabilities Research

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NIH-funded Program Focuses on Challenging Pediatric Cases; SMRT Sequencing Expected to Increase Solve Rate for Genetic Diagnosis

MENLO PARK, Calif., April 11, 2018 (GLOBE NEWSWIRE) -- Pacific Biosciences of California, Inc. (Nasdaq:PACB), the leader in long-read, high-resolution sequencing, today announced that the HudsonAlpha Institute for Biotechnology is using the organization's newest Sequel® Sequencing System to support the goal of dramatically increasing the diagnostic success rate in challenging pediatric clinical cases.

Led by faculty investigator Greg Cooper, Ph.D., the institution's NIH-funded [Clinical Sequencing Exploratory Research](#) (CSER) project is applying whole genome sequencing to better understand the genetic basis of intellectual and developmental disabilities in children and provide diagnostic information to affected families. More than 500 children and their parents have been enrolled in the study.

The group's efforts to diagnose these children using short-read sequencing technology have achieved a success rate of about 30 percent, but it is widely known that these platforms are unable to detect certain types of genetic variation that contribute to disease. Structural variants such as repeat expansions and copy number variations are larger and more complex than short-read sequencers can typically resolve, and likely underlie some of the cases that have gone undiagnosed. Diseases already known to be caused by these larger structural variants include ALS, autism, Fragile X syndrome, and Huntington's disease, among others. HudsonAlpha is now applying PacBio® long-read sequencing technology for whole genome sequencing of affected children along with their parents. This approach could produce answers for many cases that have proven intractable with other technologies.

"By applying whole genome PacBio Sequencing in this study we hope to more sensitively identify all sizes of genetic variants, thereby increasing our solve rate for previously undiagnosed children," said Dr. Cooper. "In many cases, an accurate clinical diagnosis can improve our ability to manage the child's condition. We also anticipate that we will make novel discoveries through this work that may benefit many families beyond those directly tested here."

Scientists at HudsonAlpha have been using PacBio Single Molecule, Real-Time (SMRT®) Sequencing technology in their agricultural biology work. Based on success applying long-read SMRT Sequencing to challenging plant genomes, the organization has added a second Sequel System for clinical research use in addition to its existing PacBio instrumentation used for AgBio research -- a Sequel System as well as a PacBio RS II System.

"PacBio Sequencing has made it possible for us to generate high-quality reference genomes for most plants, and we now want to apply the benefits of this technology in the human disease space," said Jeremy Schmutz, another HudsonAlpha faculty investigator. "With further scale increases and cost decreases in the future, true individual *de novo* genome sequencing may become the tool of choice for addressing difficult medical cases."

Kevin Corcoran, Senior Vice President for Market Development at Pacific Biosciences commented: "We believe projects like HudsonAlpha's CSER program to help solve undiagnosed genetic disease in children are among the most important and rewarding uses for our technology. We are delighted to see that one of our most successful customers working in the plant sciences has decided to apply our long-read technology to their research in patient populations. We look forward to seeing how PacBio sequencing can both improve their clinical sequencing success rate as well as support new discoveries."

A number of other studies have been announced in the past year that are using, or plan to use, highly accurate PacBio whole-genome sequencing to with a goal of increasing the solve rate for genetic disease. These include [Stanford University School of Medicine](#); the [European Solve-RD Consortium](#), which is performing a 500-individual rare disease study; and [Novogene](#), which has announced plans to sequence 1,000 Chinese genomes.

About Pacific Biosciences

Pacific Biosciences of California, Inc. (NASDAQ:PACB) offers sequencing systems to help scientists resolve genetically complex problems. Based on its novel Single Molecule, Real-Time (SMRT®) technology, Pacific Biosciences' products enable: *de novo* genome assembly to finish genomes in order to more fully identify, annotate and decipher genomic structures; full-length transcript analysis to improve annotations in reference genomes, characterize alternatively spliced isoforms in important gene families, and find novel genes; targeted sequencing to more comprehensively characterize genetic variations; and real-time kinetic information for epigenome characterization. Pacific Biosciences' technology provides high accuracy, ultra-long reads, uniform coverage, and the ability to simultaneously detect epigenetic changes. PacBio® sequencing systems, including consumables and software, provide a simple, fast, end-to-end workflow for SMRT Sequencing. More information is available at www.pacb.com.

About HudsonAlpha

HudsonAlpha Institute for Biotechnology is a nonprofit institute dedicated to developing and applying scientific advances to health, agriculture, learning, and commercialization. Opened in 2008, HudsonAlpha's vision is to leverage the synergy between discovery, education, medicine, and economic development in genomic sciences to improve the human condition around the globe. The HudsonAlpha biotechnology campus consists of 152 acres nestled within Cummings Research Park, the nation's second largest research park. The state-of-the-art facilities co-locate nonprofit scientific researchers with entrepreneurs and educators. HudsonAlpha has become a national and international leader in genetics and genomics research and biotech education and includes more than 30 diverse biotech companies on campus. To learn more about HudsonAlpha, visit hudsonalpha.org.

Forward-Looking Statements

All statements in this press release that are not historical are forward-looking statements, including, among other things, statements relating to future availability, uses, accuracy, quality or performance of, or benefits of using, products or technologies, the suitability or utility of products or technologies for particular applications, studies or projects, including the CSER project at HudsonAlpha, and other future events. You should not place undue reliance on forward-looking statements because they involve known and unknown risks, uncertainties, changes in circumstances and other factors that are, in some cases, beyond Pacific Biosciences' control and could cause actual results to differ materially from the information expressed or implied by forward-looking statements made in this press release. Factors that could materially affect actual results can be found in Pacific Biosciences' most recent filings with the Securities and Exchange Commission, including Pacific Biosciences' most recent reports on Forms 8-K, 10-K and 10-Q, and include those listed under the caption "Risk Factors."

Pacific Biosciences undertakes no obligation to revise or update information in this press release to reflect events or circumstances in the future, even if new information becomes available.

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