To understand the genetic factors underlying health and disease and to address hidden heritability, scientists require a more comprehensive view of all the variations in the human genome. Single Molecule, Real-Time (SMRT®) Sequencing delivers the read lengths, uniform coverage, and accuracy needed for accessing the complete size spectrum of sequence variant types – from single nucleotides to complex structural variants. PacBio’s long single-molecule reads also provide direct variant phasing information across full-length genes and chromosome haplotype blocks. With SMRT Sequencing, scientists gain new insight into the genetic basis of health and disease.

**CREATE GOLD-STANDARD POPULATION-BASED REFERENCES**
- Increase power by matching references to the genetic background of study populations
- Access novel types of genetic variation and difficult-to-characterize regions
- Improve variant calling with a more comprehensive reference genome
- Cost-effectively validate novel variants with a highly accurate orthogonal platform

**RESOLVE STRUCTURAL VARIATION**
- Uncover the hidden heritability linked to structural variation
- Identify sequence-level breakpoints to unravel the genetic etiology of disease
- See the unbiased range of structural variation of all sizes, types, and GC content in the genome

“Non-SNP variants, ranging from small indels to large CNVs and inversions, accounted for 74% of the total number of variant bases.”

“TO GET A MEDICAL-GRADE GENOME ... WE NEED TO HAVE THE MOST ACCURATE AND COMPLETE GENOME FOR EACH INDIVIDUAL. WE BELIEVE THE PACBIO SMRT MACHINES WILL HELP US REACH THIS GOAL.”
—J. CRAIG VENTER, HLI CO-FOUNDER

“WE NOW HAVE ACCESS TO A WHOLE NEW REALM OF GENETIC VARIATION THAT WAS OPAQUE TO US BEFORE.”
—PROFESSOR EVAN EICHLER, UNIVERSITY OF WASHINGTON

pacb.com/human
Resolution of two allelic copies of the MUC5AC gene

CHARACTERIZE COMPLEX REGIONS UNDERLYING GENETIC DISEASE

- Sequence previously unsequenceable loci associated with genetic disease
- Accurately and definitively phase polymorphisms across entire genes, such as HLA
- Multiplex samples to cost-effectively scale research

“WE BASICALLY CAN SEE THE ENTIRE PICTURE. WE’RE NOT LOOKING UNDER A LAMPPOST FOR THE KEYS. IT’S DAYLIGHT, AND WE CAN SEE THE WHOLE NEIGHBORHOOD. SO WE’RE GONNA FIND THE KEYS.”

—DAN GERAGHTY, FRED HUTCHINSON CANCER CENTER

PROFILE THE COMPLEXITY OF THE TRANSCRIPTOME WITH ISO-SEQ™ SEQUENCING

- Discover novel genes and gene isoforms
- Directly sequence full-length transcripts to eliminate the need for transcript reconstruction
- Differentiate isoform expression between cells, tissues, and disease states

“THE POWER OF LONG-READ SEQUENCING IS REALLY TO BE ABLE TO CAPTURE ALL OF THE INFORMATION IN ITS INTACT FORM WITHOUT TRYING TO SOLVE A JIGSAW PUZZLE THAT YOU MAY HAVE PUT TOGETHER WRONG.”

—MIKE SNYDER, STANFORD UNIVERSITY

KEY REFERENCES


14. Au, KF et al. (2013) Alternative splicing can have important implications for protein structure and function


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