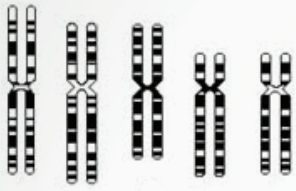
# A Comprehensive View of Genetic Diversity

Single Molecule, Real-Time (SMRT®) Sequencing combines long reads with unbiased coverage to generate high-quality, contiguous assemblies. High-quality genomes and evidence-based annotations enable improved genetic marker development, discovery of novel genes, and structural variation characterization.

## Generate High-Quality Reference Genomes



"..Superior to any previous *de novo* [reference genome] assemblies of these organisms."[1]

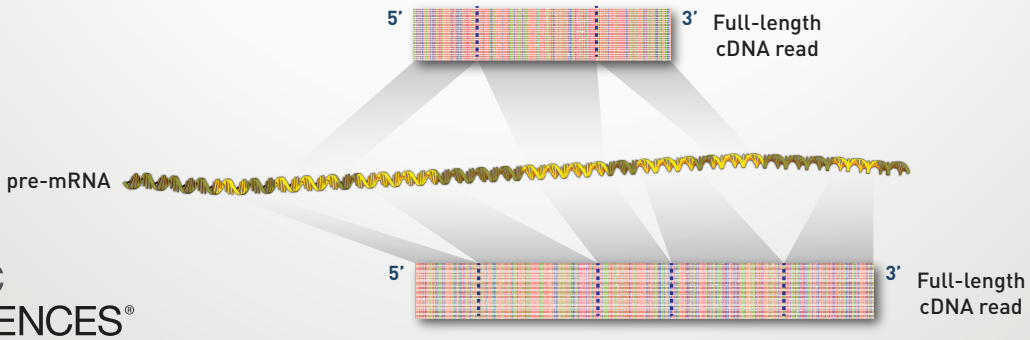
- Capture previously undetected SNPs, genes, and regulatory regions for improved marker development
- Reduce number of contigs for more complete mapping of short reads against the reference
- Access variation other than SNPs to resolve missing heritabilities

## Reveal the Complexity of the Transcriptome

- Discover novel genes and gene isoforms
- Uncover direct gene annotation evidence with full-length transcript reads
- Improve gene expression quantification with isoform resolution[2]
- Distinguish important stress response, developmental, and tissue-specific isoforms

"..We were able to identify hundreds of new genes – complete genes that had not been annotated previously."

– Alisha Holloway, The Gladstone Institutes[3]

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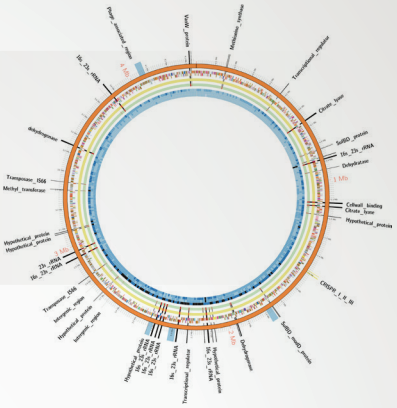
## Characterize Microbes and Microbial Communities

"Single molecule sequencing will be increasingly used to produce finished microbial genomes."

— Michael Koepke, LanzaTech[4]

- Completely characterize pathogen genomes, including antibiotic-resistance genes
- Improve capabilities for bioprospecting in environmental samples
- Enable complete reconstruction of metabolic networks



## Uncover the Diversity of Gene Families


- Study complex traits via targeted sequencing of multi-gene families, with flexible assay design
- Accurately characterize genes with repetitive sequences or structural variations
- Identify previously unrecognized gene diversity and allelic variation
- Differentiate polyploid gene duplications, paralogs, and orthologs

"...Long read lengths eliminated the necessity for downstream assembly of highly similar fragments..."

— Anne Yoder, Duke University[5]

## Key References


1. Berlin K, Koren S, Chin CS, Drake J, Landolin JM, Phillippy AM. (2014) Assembling large genomes with single-molecule sequencing and locality sensitive hashing. *bioRxiv.* doi: http://dx.doi.org/10.1101/008003.
2. Au KF, Sebastiano V, Afshar PT, Durruthy JD, Lee L, Williams BA, van Bakel H, Schadt EE, Reijo-Pera RA, Underwood JG, Wong WH. (2013) Characterization of the human ESC transcriptome by hybrid sequencing. *PNAS.* **110**(50):e4821-30.
3. Thomas S, Underwood JG, Tseng E, Holloway AK. (2014) Long-read sequencing of chicken transcripts and identification of new transcript isoforms. *PLoS ONE.* **9**(4): e94650.
4. Brown SD, et al. (2014) Comparison of single-molecule sequencing and hybrid approaches for finishing the genome of *Clostridium autoethanogenum* and analysis of CRISPR systems in industrial relevant Clostridia. Biotechnology for Biofuels. **7**:40.
5. Larsen PA, Heilman AM, Yoder AD. (2014) The utility of PacBio circular consensus sequencing for characterizing complex gene families in non-model organisms. *BMC Genomics.* **15**:720.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