ASHBi SEMINAR

GENCODE Reference Gene Annotation: Past, Present and Future

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Abstract

The GENCODE project has worked to produce accurate and comprehensive reference gene annotation for the human and mouse genomes for more than 20 years. This period has seen a massive expansion of our knowledge of the genome and equally huge developments in the technologies and analysis methods that support it. I will describe how GENCODE has worked to incorporate new knowledge and data into its annotation with recent examples from our work with the Long-read RNA-Seq Genome Annotation Assessment Project (LRGASP), formed to evaluate the effectiveness of long-read approaches for transcriptome analysis and the global Ribo-seq community to capture and describe genomic regions that are transcribed and translated but not annotated as protein-coding genes.

Looking forward, I will present some of our current work on the annotation of the human reference pangenome where expert human annotators are identifying regions that are challenging to current automated annotation methods. The annotation produced from this work, the processes developed and the knowledge gained will be used to inform the automated annotation methods of the future. I will also discuss our own use of AI/ML in GENCODE annotation and our plans to make all the datasets created by the GENCODE consortium are optimised for machine learning applications.

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