

**Abstract Title:** Complete human genome sequencing and complex genetic variation

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**Abstract:** The discovery and resolution of genetic variation is critical to understanding disease and disease susceptibility. I will present our most recent work sequencing diverse human genomes telomere-to-telomere (T2T) using both ultra-long and high-fidelity long-read sequencing technologies. The approach allows us to sequence, assemble, and phase all forms of human genetic variation, including complex structural and copy-number variants irrespective of size—the vast majority of which are not routinely characterized by short-read sequencing. Advances in this area have made possible the first T2T assemblies of both human and non-human primate chromosomes providing new biological insights into regions typically excluded from human genetic and comparative studies. This is leading to new insights into chromosomal biology, human genetic diversity, and recurrent copy-number changes underlying neurodevelopmental disease. Assembly-based variant discovery has the potential to provide a complete understanding of human genetic variation at every level and, we predict, will be the future of genetic and clinical-based research.

**Area of expertise:** segmental duplication, structural variation, neurodevelopmental disease, copy number variation, long-read sequencing, T2T genomes, autism research