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Embracing Complexity in Personalized Medicine

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Dr. Lee's laboratory was the first to describe genome-wide structural genomic variants in the form of copy number variants (CNVs) among humans with the subsequent development of two human CNV maps that are now actively used in the diagnoses of array based genetic tests. He has published over 150 peer-reviewed articles in leading journals such as Cell, Nature and Science. Dr. Lee is the president of The Human Genome Organization (HUGO International). He is an elected fellow of the American Association for the Advancement of Science since 2012 and a Thompson Reuter Citation Laureate (2014).

Research focus

The research laboratory of Dr. Charles Lee at The Jackson Laboratory for Genomic Medicine uses state-of-the-art technologies to study structural genomic variation in human biology, evolution and disease. Ongoing studies in our group include:

- 1. accurate identification and annotation of structural variation (SV) in human and other vertebrate genomes;
- 2. identification and characterization of genetic aberrations associated with congenital birth defects:
- 3. the development of new diagnostic assays and platforms for genomic medicine:
- 4. understanding genome evolution in vertebrate species and its impact on genome stability