Special Issue

Detecting and Interpreting Structural Variation in the Human Genome

Message from the Guest Editor

Structural variation (SV) in the human genome can come in several forms including duplications, deletions, inversions and translocations. More complex variations can include several SVs that encompass a single complex genomic rearrangement (CGR) within an individual's genome. Whether simple or complex, this type of variation can lead to a pathogenic consequence when it disrupts either gene dosage, creates to a gene fusion event or a direct interruption of a genes-coding sequence. Originally detected through traditional karyotyping, the identification of structural variation has vastly improved with the advent of high-resolution array comparative genomic hybridization (aCGH), short- and long-read sequencing methodologies, optical genome mapping (OGM) and bioinformatic tools specifically designed to find this type of variation within sequencing data. The fact that structural variation is frequently mediated by repetitive elements within the genome makes it particularly difficult to detect and characterize its final architecture. As large-scale sequencing projects continue to increase, the detection and need for interpretation of structural variation will continue to increase.

Guest Editor

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