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"Integrative computational models for functional interpretation of human genomic sequence variants"

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Bahen Centre for Information Technology
40, St. George Street, Room 1180

Abstract:

The decreasing cost of sequencing is leading to a growing repertoire of personal genomes. However, we are lagging behind in understanding the functional consequences of the millions of variants obtained from sequencing. For cancer genomes, an understanding of the functional impact of mutations can help identification of drivers that lead to tumorigenesis. A large proportion of genomic variants are noncoding and their interpretation is especially challenging. In my talk, I will present integrative computational models to understand the global system-wide effects of mutations. These models combine population-scale sequencing data (from 1000 Genomes consortium), noncoding functional annotations (from ENCODE consortium) and gene interactions from biological networks (including protein-protein, regulatory and metabolic interactions). I will discuss how contrasting patterns of natural polymorphisms and somatic variants can lead to identification of cancer driver mutations. Finally, I will discuss computational tools that I have developed to prioritize variants (both coding and noncoding) in disease studies.

Host: Michael Brudno