Computational and Machine Learning Methods to Study Genomics of Diseases*

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Abstract

Understanding structural variation, genome architecture, and liquid biopsy biomarkers is essential for advancing precision genomics and early disease detection.I will begin by first introducing the necessary background in genomics and biology, with a focus on the role of genetic variants, especially structural variants, in disease. In this talk, I will highlight three projects where we developed computational and AI-driven approaches to address key challenges in computational biology. First, I will introduce a mapping-free framework for comparative genome analysis using long-read sequencing data, which serves as the foundation for a structural variant detection method that combines multiple strategies to improve accuracy, particularly in repetitive regions. Second, I will present a method based on linear programming to predict the impact of structural variants on three-dimensional genome organization, and introducing the TAD fusion score, a metric that quantifies the effect of deletions on topological domain structure and their relevance to disease. Finally, I will describe Orion, a deep generative model using autoencoders for non-invasive cancer detection from circulating small RNAs, which shows strong performance in classifying early-stage lung cancer. Collectively, these studies demonstrate how novel computational frameworks, spanning structural variant discovery, genome comparison, and AI-driven biomarker analysis, are transforming both our understanding of genome biology and the clinical utility of genomics in disease detection.

Declaration on Generative AI

The author has not employed any Generative AI tools.

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