

Can Computational Biology and Deep Learning help to decode gene regulation?

Abstract

The human genome consists of approximately 3 billion base pairs, with each individual exhibiting unique genetic variations in their genome. These differences can directly cause diseases or predispose individuals to various conditions. Understanding the genome is critical to elucidate normal biological processes and disease mechanisms. However, its complexity grows exponentially when factoring in the diverse functions of the genome.

While changes in coding regions are well-studied, they make up just 2% of the genome, whilst the remaining 98% — non-coding regions — remain under-explored. Non-coding changes can have significant impacts, including structural alterations that affect multiple genes along a chromosome, disrupting the proteins they produce; splicing disruptions that interfere with RNA splicing and alter protein-coding sequences; and regulatory changes which directly influence gene expression. Modern research techniques, combining wet lab experiments with bioinformatics, can assess these variations. However, such approaches are both time-intensive and costly. This is where Computational Biology and Deep Learning (in particular in the genomic fields, with established Convolutional Neural Networks and now Transformers) have the ability to prioritise cell types, identify regulatory elements, and analyse variations in the genome. By reducing protracted and costly experimental processes, computational methods allow to hypothesise true bio-physic events, guiding experiments that simultaneously validate these hypotheses and elucidate genome-variant functions. This aids in better understanding the behaviour of the genome, effects of individual genetic variants, and paves the way for potential disease treatments.

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