Deep neural networks and DNA variant effect predictions in humans

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Abstract

In this applied session we will discuss emerging approaches that rely on the generalizability and explainability of deep neural networks for understanding genomic variants and DNA regulatory activity in the non-coding regions of the human genome. Currently, these approaches rely on the use of convolutions or transformers and variational autoencoders and are achieving state-of-the-art performance comparable to experimental results in some use cases. During the talk we will detail the network structures and explore the recent history of the approaches.

Keywords: Deep neural networks, convolutions, transformers, multi head attention, autoencoders, DNA, genome, variant classification.