ABERRANT SPLICING DETECTION USING CONVOLUTIONAL NEURAL NETWORKS (CNNS)

ABSTRACT

The technology disclosed relates to constructing a convolutional neural network-based classifier for variant classification. In particular, it relates to training a convolutional neural network-based classifier on training data using a backpropagation-based gradient update technique that progressively match outputs of the convolutional neural network-based classifier with corresponding ground truth labels. The convolutional neural network-based classifier comprises groups of residual blocks, each group of residual blocks is parameterized by a number of convolution filters in the residual blocks, a convolution window size of the residual blocks, and an atrous convolution rate of the residual blocks, the size of convolution window varies between groups of residual blocks, the atrous convolution rate varies between groups of residual blocks. The training data includes benign training examples and pathogenic training examples of translated sequence pairs generated from benign variants and pathogenic variants.

Figure 31