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Detection of alternative splicing: deep sequencing or deep learning

Alternative splicing enables the expression of a variety of isoforms coding for functionally diverse proteins from a single gene. RNA sequencing (RNAseq) has become the state-of-the-art tool for profiling the transcriptome, but still reliable detection of alternative splicing events in RNAseq from virus-infected cells with low number of reads is challenging. Few computational tools, such as Junction coverage compatibility (JCC) and Deep Splice, try to address this. We compare those tools' performance with sequence-based prediction tool SpliceAI on subsampled 50M read data from four dilated cardiomyopathy patients. The tools were found to show high precision but poor recall. Using these insights, we developed SpliceStat, leveraging both sequence and RNAseq data for improved accuracy in alternative splicing prediction.

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Keywords

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Autor: HACKL, Lena Maria

Co-Autoren: BAUMBACH, Jan (Universität Hamburg); TSOY, Olga (Universität Hamburg)