Quick and easy: 1-day-protocol for detection of normal alternative splicing events and aberrant spliceogenic transcripts with deep RNA sequencing

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Abstract

RNA sequencing is a promising technique for detecting normal and aberrant RNA isoforms. Here, we present a new single-gene, straightforward 1-day hands-on protocol for detection of splicing alterations with deep RNA sequencing from blood. We have validated our method's accuracy by detecting all normal splicing isoforms of STK11 gene that were previously published. Additionally, the same technique was used to provide the first comprehensive catalogue of naturally occurring alternative splicing events of the NBN gene in blood. Furthermore, we demonstrate that our approach can be used for detection of splicing impairment caused by genetics variants. Due to the simplicity of our approach it can be incorporated into any molecular diagnostics laboratory for determination of variant's impact on splicing.

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