ASpli: an integrative R package for the analysis of alternative splicing using RNA-Seq

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Background Alternative splicing (AS) is a prevalent mechanism of post transcriptional gene regulation in multicellular eukaryotes. It allows a single gene to increase functional and regulatory diversity, through the synthesis of multiple mRNA isoforms encoding structurally and functionally distinct proteins. AS occurs via 4 main events: intron retention (IR), exon skipping (ES) and alternative use of donor and acceptor sites (Alt5’Ss and Alt3’Ss). The development of novel high-throughput sequencing methods for RNA (RNA-Seq) provided a very powerful mean to study alternative splicing under multiple conditions at unprecedented depth. As long as new studies on post-transcriptional regulation arises, there are an increasing evidence than AS frequency is higher than expected. Despite It has became the new standard for studying gene and transcription expression, the use of RNA-seq for the study of transcripts repertoire in a given condition is not trivial [1].

Results Here we introduce a very flexible and easy-to-use R package named ASpli. We propose a count based integrative method taking into account gene expression, exon and intron differential usage and their relationship with junctions spanning those features.

Conclusion Using an annotated transcriptome we are able to classify subgenic features into alternative or not alternative regions. ASpli is intended to facilitate the analysis of RNAseq data for the quantification and discovery of AS events and it has been used in many recent publications from our lab. Results of the analysis are presented in a user friendly manner, including plots of the most relevant AS events discovered.

References