Supplementary Materials

Table S1. Summary of bioinformatics tools with data access details. Tabulated list of 7 bioinformatics tools with links to online data access (links correct and functional as of 15 November 2019). PMID, PubMed ID.

Tool	Purpose	Link	PMID	Citation
CADD	General-purpose pathogenicity scoring	Tabulated datasets: https://cadd.gs.washington.edu/download Online lookup: https://cadd.gs.washington.edu/snv	30371827, 24487276	[1,2]
TraP	Quantification of variant impact on transcripts	Request for access to tabulated scores: http://innovation.columbia.edu/technologies/cu17233_pathogenicity-database-for-identification-of-disease-causing-non-coding-genetic-variations Online lookup: http://trap-score.org/Search?version=v3	28794409	[3]
SPANR	Cassette exon skipping prediction	Online lookup: http://tools.genes.toronto.edu Link for ANNOVAR download: http://annovar.openbioinformatics.org/en/latest/user-guide/download/	25525159	[4]
CryptSplice	Effect of variants on existing splice sites and cryptic splice site prediction	Bitbucket source code download: https://bitbucket.org/jhucidr/cryptsplice/src/master/	28475858	[5]
MMSplice	Prediction of exon skipping, competitive interactions, changes in splicing efficiency and pathogenicity	GitHub source code download: https://github.com/gagneurlab/MMSplice	30823901	[6]
S-CAP	Variant pathogenicity scoring with the compartmentalization of genomic space	Tabulated datasets: http://bejerano.stanford.edu/scap/ Bitbucket source code download: https://bitbucket.org/bejerano/splicing_classifier/src/master/	30804562	[7]
SpliceAI	Prediction of variant impact on acceptor/donor loss or gain	GitHub source code download: https://github.com/Illumina/SpliceAI	30661751	[8]

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