A PUBLICATION MODEL THAT ALIGNS WITH THE KEY OPEN SOURCE SOFTWARE PRINCIPLES

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PUBLISHING SOFTWARE: SQUARE PEG, ROUND HOLE!
THE PUBLICATION MODEL

F1000Research

Submission to publication: DAYS

Article submitted → In-house pre-refereeing check → Publication

- Open referee process
- Registered user commenting

Author revises article → Indexed

Data repository

Once 2 Approved statuses, or 2 Approved with Reservations statuses and 1 Approved status
UPDATING DOCUMENTATION

**REvised**

Denotes an article that has been revised by the authors, usually following referee and/or reader feedback.

**UPDATE**

Denotes a small development to the study that is added by the authors.
Validation of predicted mRNA splicing mutations using high-throughput transcriptome data [v2; ref status: indexed, http://f1000r.es/378]

Coby Viner1, Stephanie N. Dorman2, Ben C. Shirley3, Peter K. Rogen1-3

Abstract

Interpretation of variants present in complete genomes or exomes reveals numerous sequence changes, only a fraction of which are likely to be pathogenic. Mutations have been traditionally inferred from allele frequencies and inheritance patterns in such data. Variants predicted to alter mRNA splicing can be validated by manual inspection of transcriptome sequencing data, however this approach is intractable for large datasets. These abnormal mRNA splicing patterns are characterized by reads demonstrating either exon skipping, cryptic splice site use, and high levels of intron inclusion, or combinations of these properties. We present, Veridical, an in silico method for the automatic validation of DNA sequencing variants that alter mRNA splicing. Veridical performs statistically valid comparisons of the normalized read counts of abnormal RNA species in mutant versus non-mutant tissues. This leverages large numbers of control samples to corroborate the consequences of predicted splicing variants in complete genomes and exomes.

Corresponding author: Peter K. Rogen

Open Peer Review

Invited Referee Responses

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1. Stefania Bortoluzzi, University of Padova, Italy
2. Francesco Xavier Roca, Nanyang Technological University, Singapore
3. Liliana Florea, Johns Hopkins University, USA
4. Peter Robinson, Universitätsklinikum Charité, Germany

Comments

No comments | Add Comment
REPRODUCIBILITY
## REPRODUCIBILITY

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Jim Procter  
Computational Biology, College of Life Sciences, University of Dundee, Dundee, UK  
Approved: 25 February 2014  
Referee Report: 25 Feb 2014  
doi: 10.5256/f1000research.3674.r3680  

Summary  
This F1000Research article describes an ensemble of JavaScript components designed for bioinformatics web developers that allow the retrieval, layout and display of positional annotation on a 1D coordinate system, such as a protein or nucleotide sequence. Special support is provided for the display of protein positional annotation retrieved from the Distributed Annotation System (requires some server-side configuration), and the system provides standard glyphs and shading styles to allow active sites and common types of post-translational modifications to be effectively displayed. Importantly, the components employ the BioJS framework, which allows them to exchange messages with other BioJS biological data visualization components (as well as any jQuery based module) to facilitate the creation of rich, interactive web interfaces.

Author Response  
L. Garcia, EMBL-EBI, UK  
Posted: 03 Apr 2014  
Dear James,  
Thanks for your review, it has been useful to improve our work. We have tried to addressed all you comments, however those related to the component itself, i.e., JavaScript code, will be taken into account for a new version of the software, and those related to BioJS in general will be sent to BioJS core developers. Please see our responses below.

Response to (1) and (2): We are currently working on an improved component to visualize protein sequence annotations. We have made notes about this suggestions and will take them into account for the new visualization. Unfortunately, such improvements are not yet ready to be integrated into the public BioJS GitHub repository.
ARTICLE COLLECTIONS