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

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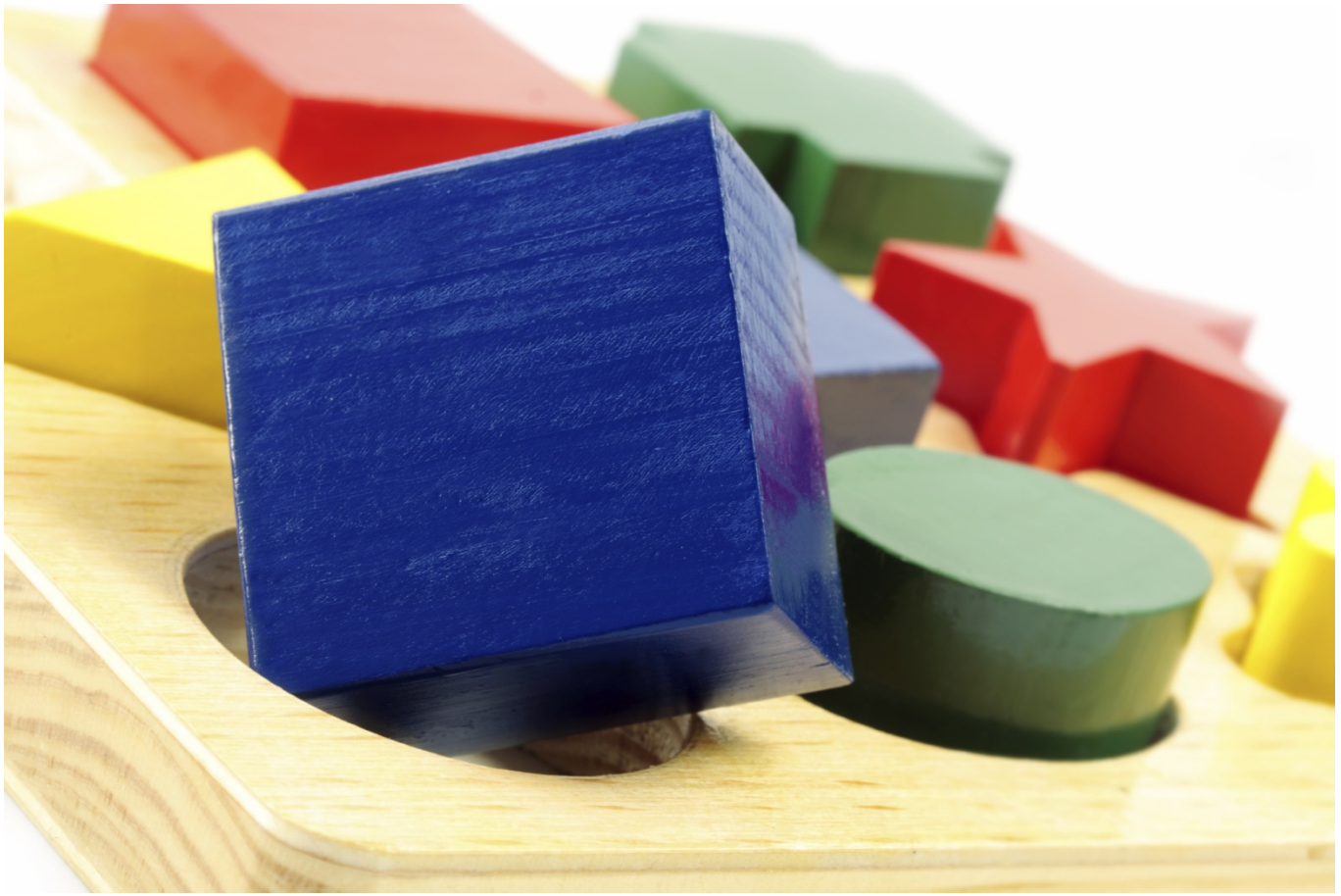
PUBLISHING SOFTWARE: SQUARE PEG, ROUND HOLE!



THE PUBLICATION MODEL

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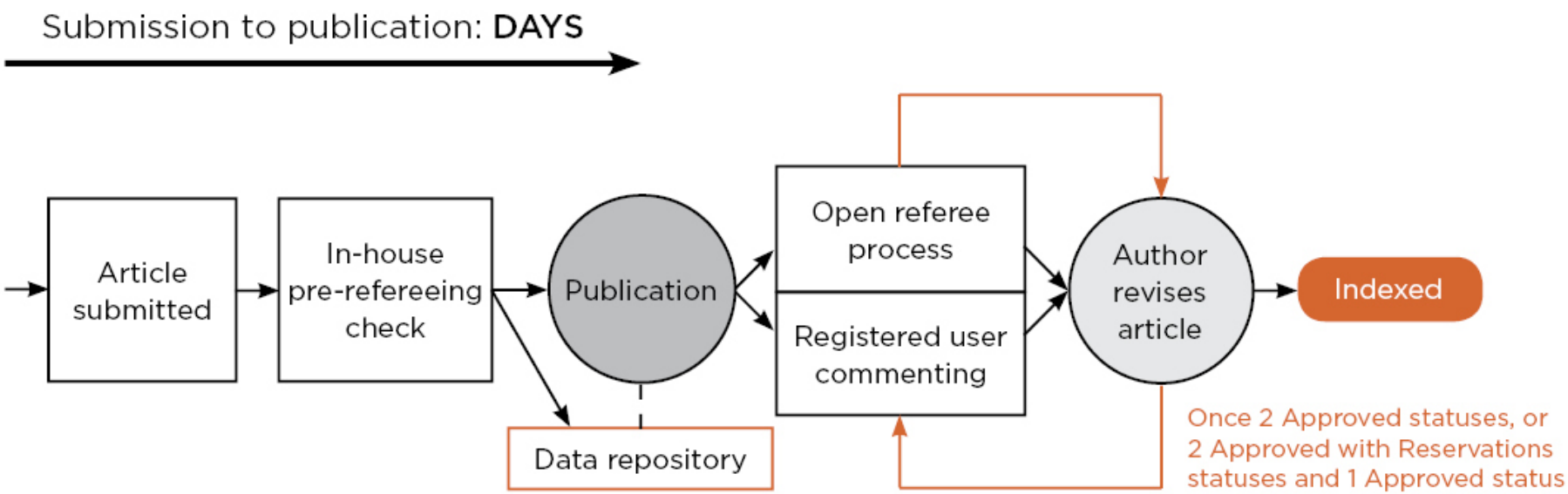

UPDATING DOCUMENTATION

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.

UPDATING DOCUMENTATION

F1000Research » Articles

SOFTWARE TOOL

UPDATE

CrossMark click for updates

Version 2 of 2

Validation of predicted mRNA splicing mutations using high-throughput transcriptome data [v2; ref status: indexed, http://f1000r.es/378]

Coby Viner[1], Stephanie N. Dorman[2], Ben C. Shirley[3], Peter K. Rogan[1-3]

Author affiliations

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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. Rogan

Open Peer Review

Invited Referee Responses

	1	2	3	4
version 1 published 13 Jan 2014	report	report	report	report
version 2 published 07 Apr 2014	report	report	report	

UPDATE

1 **Stefania Bortoluzzi**, University of Padova, Italy

2 **Francesc Xavier Roca**, Nanyang Technological University, Singapore

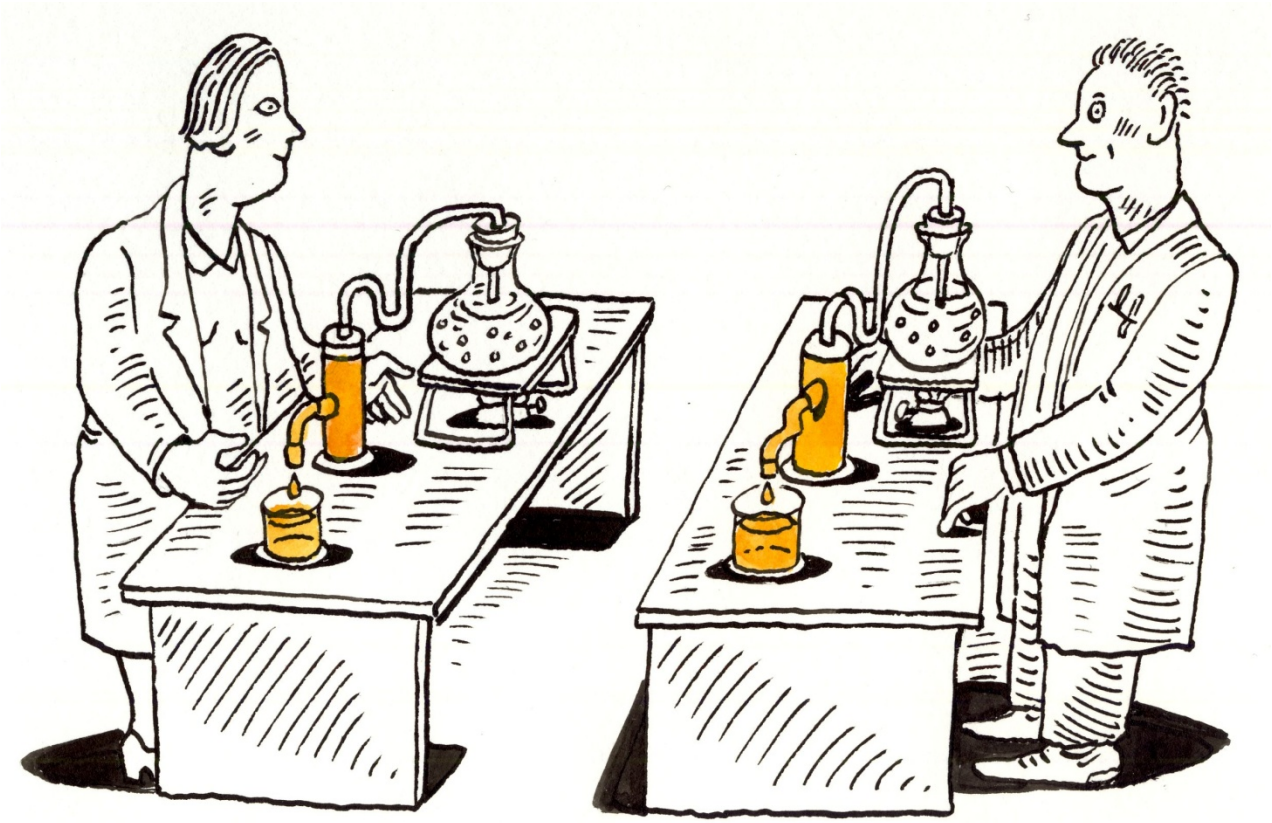
3 **Liliana Florea**, Johns Hopkins University, USA

4 Peter Robinson, Universitätsklinikum Charité, Germany

Comments

No comments | Add Comment

REPRODUCIBILITY



REPRODUCIBILITY

1	**SOFTWARE ACCESS** URL link to where the software/ program can be downloaded and/or used.
2	**LATEST SOURCE CODE** URL link to the authors most up-to-date source code on a Version Control System (VCS) such as GitHub, BitBucket and SourceForge.
3	**SOURCE CODE AS AT THE TIME OF PUBLICATION** URL link to a copy of the author's source code forked from the author's VCS repository at the time of publication of this paper.
4	**ARCHIVED SOURCE CODE AS AT THE TIME OF PUBLICATION** Persistent DOI link to F1000Research's archival space within Zenodo. The source code should be cited using this DOI.
5	**SOFTWARE LICENCE** The licence under which the software has been published.

OPEN PEER REVIEW AND OPEN COMMUNITIES

Jim Procter
Computational Biology, College of Life Sciences, University of Dundee, Dundee, UK

Approved: 25 February 2014

Referee Report: 25 Feb 2014 36

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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

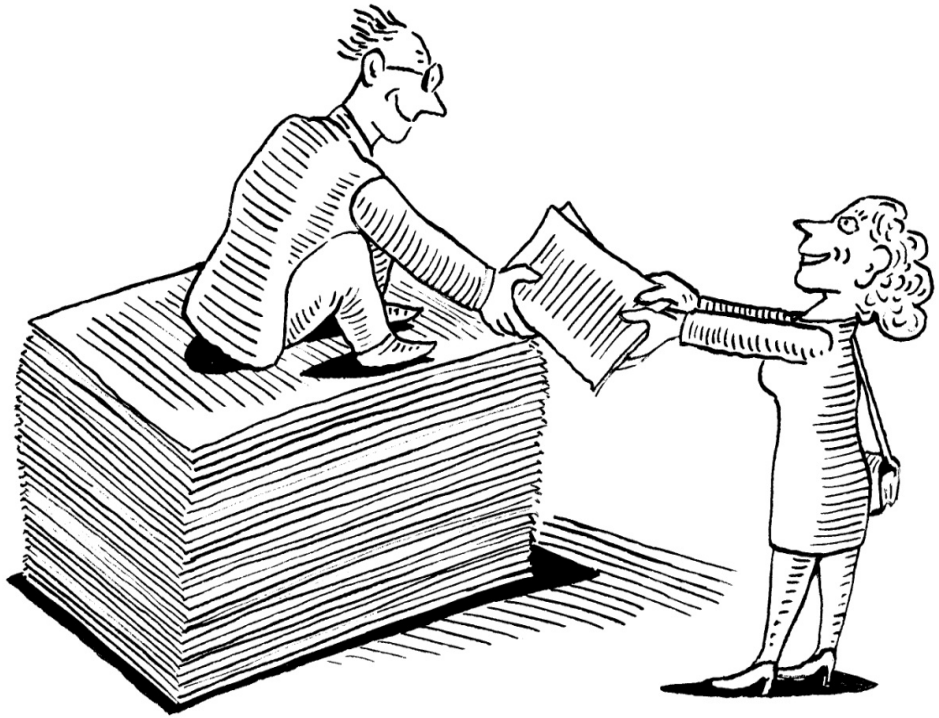
L. Garcia, EMBL-EBI, UK
Posted: 03 Apr 2014

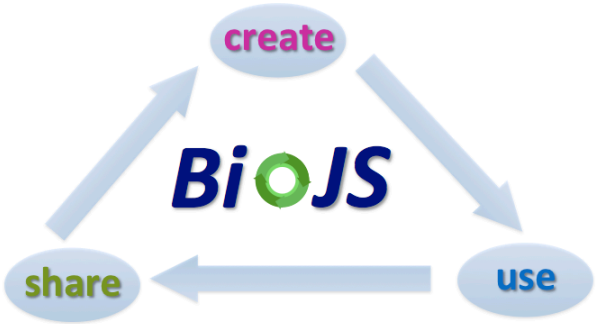
Dear James,

Thanks for your review, it has have 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





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