

Reviewer Report

Title: **SV-plaudit: A cloud-based framework for manually curating thousands of structural variants**

Version: **Original Submission** Date: 4/9/2018

Reviewer name: **Fritz Sedlazeck**

Reviewer Comments to Author:

The authors of the manuscript "SV-plaudit: A cloud-based framework for manually curating thousands of structural variants" propose a framework to easily manually assess if SVs are potentially false or true. This is enabled based on a cloud based pipeline, which allows to look at multiple thousand sites for a larger community.

Overall I think that this is an important contribution for multiple projects such as GiaB or other where scientist need to assess the quality of their discovered SVs.

In the following some concerns and questions:

1. I am wondering if you could comment what had a deeper impact in the evaluation: a) the visualization or b) the ability to look at the trio
2. I would encourage to include the mappability track of some kind (e.g. 36bp) to give the users more control and insight of the variability observed at the breakpoints. I know you stated that this needs to be part of a future research, but I think that is easy to obtain (UCSC) and integrate. Another maybe very useful feature would be the frequency of the reads that support the event.
3. I would encourage you to provide also figures for the other types of SVs not just Deletions. E.g. how do you visualize BND or other events?
4. I think your demonstration is really nice over the 1000 genomes data. What I would liked to see further is for some validated SVs if the figures are consistently clear. I know this is maybe out of the scope of this study, but maybe showing a few examples of the pass vs. non pass SVs from GiaB call set 0.5.0, which hopefully are close to the truth might give further insights on the reliability of the method. This is especially interesting since you mention false discovery and sensitivity issues over computational genotyping SVs.
5. I found Figure 1 A rather confusing since I only see the coverage. Is this due to the size of the region and thus the points on the bottom are the read pairs? In that case there should be some pairs that span the deletion, right? Could you maybe sort the reads better that support the SV, or more general show abnormal distances?

Methods

Are the methods appropriate to the aims of the study, are they well described, and are necessary controls included? Yes

Conclusions

Are the conclusions adequately supported by the data shown? Yes

Reporting Standards

Does the manuscript adhere to the journal's guidelines on [minimum standards of reporting?](#) Yes

Choose an item.

Statistics

Are you able to assess all statistics in the manuscript, including the appropriateness of statistical tests used? There are no statistics in the manuscript.

Quality of Written English

Please indicate the quality of language in the manuscript: Acceptable

Declaration of Competing Interests

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