Reviewer Report

Title: CNVpytor: a tool for CNV detection and analysis from read depth and allele imbalance in whole genome sequencing

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Reviewer Comments to Author:

Most of my comments are addressed.As authors mentioned the proposed tool is an extension of their previous tool (the main algorithm for CNV detection is the same). Since the novelty and scientific merit is not significant, using a new name for the tool rather than introducing a new version of the old one is confusing. It is better authors name the tool and introduce it as a new version, specially that the old tool is one of the commonly used CNV detection tool.In the response authors stated: "However, within scope of this paper we did not use BAF for detection on CNVs, only for visualization and genotyping. We are working on extension on CNVpytor that will use BAF as an additional information during calling step", Therefore using the title as "a tool for CNV/CNA detection and analysis from read depth and allele imbalance in whole genome sequencing" is misleading.Also authors added :" However, more accurate approach to cancer and mosaic cases are part of development in progress." Considering the mentioned statement, indicating "a tool for CNV/CNA detection" is not appropriate and "CNA" needs to be deleted.I recommend authors change the title. I am not convinced by the 2.3 response. All the approaches for CNV detection for single cell sequencing data are based on the read depth analysis since the coverage is very low. And they use a very large window size (more than 200K bp). But the default of the proposed tool is a window size of 100 bp which is not appropriate for single cell sequencing data. In real practice, the coverage of single cell DNA sequencing data is <1 and the statement :"Provided good amplification and high coverage detecting CNVs in single cell," is not a practical discussion about applicability of the proposed tool to the single cell sequencing data. In addition, BAF data used for visualization and genotyping in this tool (the new addition) cannot be obtained from very low coverage (<1) sequencing data. I recommend authors provide more practical discussion about the applicability of the tool for single cell sequencing data or do not mention it.

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