Reviewer’s report

Title: Diagnosis of Noonan syndrome and related disorders using Target Next Generation Sequencing.

Version: Date: 26 November 2013

Reviewer: sajid rashid

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Major Compulsory Revisions

Authors characterized 11 mutations initially, however they were not described later in results and discussion.

The methods are not completely addressed. For example, page 1: How authors selected the gene list: they mentioned to apply NGS to 92% of region for the listed target genes, how threshold was selected, while the mutations in these genes were already reported?

Figures are mostly the screenshots of tools. There must be a detailed figure how the analysis was carried out especially describing the chromosomal locations of genes listed along with their individual contribution in disease. As in figure 2, only single mutation has been shown, more examples are required to show reproducibility.

Minor Essential Revisions:

Language: The language is not appropriate for publication. Many sentences are incomplete and/or incorrect. For example, page 7: TSCA approach reduced up to 12 the number of exons, page 9: while in the other case clinical evaluation was unable, page 9: with a significantly reduction of time to reach...

Typing errors: page 1: ass be after excluded, page 6: As a SNP database, inappropriate usage of adverbs (a, an, and the) at many places. Please ask a native English speaker for corrections.

All the gene names must be in italic.

Bibliography: Formatting no ok, For example: references 9 and 12: look year. References 2 and 3 : check author names formatting.

Level of interest: An article whose findings are important to those with closely related research interests

Quality of written English: Not suitable for publication unless extensively edited
Statistical review: No, the manuscript does not need to be seen by a statistician.