Reviewer’s report

Title: Two monogenic disorders masquerading as one: Severe congenital neutropenia with monocytosis and non-syndromic sensorineural hearing loss

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Reviewer: Annalaura Torella

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In the present study Venugopal et al reported a large family with four successive generation with complex phenotype of severe congenital neutropenia, partially penetrant monocytosis and mild to severe hearing loss. The authors performed whole exome sequencing to detect the causative variants and Sanger sequencing whose performed for segregation analysis.

They reported the identification of a pathogenic GFI1 variant an a potentially pathogenic variant in MYO6 with generate the hypothesis that this variant combination underlay the complex disease phenotype within the screened family.

The study is well performed and up to date for the techniques employed. The manuscript is well written and clear. The findings are not novel in their separate value but generate the hypothesis that causative variant in the two identified genes underlie the pathogenesis of polygenic complex disease therefore highlighting clinical benefits of broad screening approach for complex disease.

Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

Yes

Does the work include the necessary controls?
If not, please specify which controls are required in your comments to the authors.

Yes

Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.

Yes

Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?
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Not relevant to this manuscript
Quality of written English
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Acceptable

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