Reviewer's report

Title: Mucopolysaccharidosis III B and Mild Skeletal Anomalies: coexistence of NAGLU and CYP26B1 missense variations in the same patient in a Chinese family

Version: 0 Date: 24 Jan 2018

Reviewer: Orazio Gabrielli

Reviewer's report:
The authors report a patient with biallelic variation both in NAGLU (MPS IIIB) and in CYP26B1 (skeletal dysplasia). These genetic variations, obtained by Whole exome sequencing, are novel; moreover, the clinical signs are a combination of the two different diseases. It is my opinion that this present report could contribute to a better understanding of these two diseases. The article is well written, exhaustive both the introduction and the discussion of the data. For this reason, I consider useful to accept this paper for the publication in BMC Medical Genetics.

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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