Reviewer’s report

Title: Novel NOG (p.P42S) mutation causes proximal symphalangism in a four-generation Chinese family

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Reviewer: Tadashi Kaname

Reviewer's report:

Manuscript No: MGTC-D-19-00081

Title: Identification of a novel NOG mutation (P42S) in a four generation Chinese family with proximal symphalangism

Authors: Sha et al.

The authors reported a Chinese family with proximal symphalangism and no hearing loss. The patients harboured a novel missense variant in the NOG gene (c.124C>T, (p.P42S)).

This manuscript could provide additional data for molecular pathology of NOG. The manuscript, however, do not seem to be enough for publication in the BMC Medical Genetics.

At least new clinical or biological findings should be needed.

There are three reports of patients with variants at the same amino acid of P42 (Horm Res 69:221-6 (2008), Joint Bone Spine 81:533-6 (2013), J Plast Reconstr Aesthet Surg 66:e287-9(2014)). The authors should compare clinical findings among them.

The text has some non-logical explanations and wrong usage of technical terms. The authors should make an effort to describe logical expression and to add more data.

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

No

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

No
Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.

No

Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?
If an additional statistical review is recommended, please specify what aspects require further assessment in your comments to the editors.

Not relevant to this manuscript

Quality of written English
Please indicate the quality of language in the manuscript:

Needs some language corrections before being published

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