Reviewer's report

Title: A 9-year-old Korean girl with Fontaine progeroid syndrome: a case report with further phenotypical delineation and description of clinical course during long-term follow-up

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Reviewer: Tadashi Kaname

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Title: A 9-year-old Korean girl with Fontaine progeroid syndrome: a case report with further phenotypical delineation and clinical course during long-term follow-up.
Authors: Ryu et al.

The authors reported a 9-year-old Korean girl with skull deformities, a peculiar facial appearance with microphthalmia and midface hypoplasia, progeroid features with wrinkled skin, anomalies of digits and nails, a large umbilical hernia, and a normal developmental outcome. The patient harboured a de novo recurrent missense mutation in the SLC25A24 gene (c.650G>A, (p.Arg217His)).

Since there are only three papers of SLC25A24 mutations in patients with Fontaine progeroid syndrome, additional information would be helpful to understand the syndrome. However, it is not clear what is novel in this manuscript, because she has a recurrent variant in SLC25A24. The authors should emphasize what novel point(s) are in the text.

Platform and data (not pathogenic CNVs) of chromosomal microarray should be described as supplemental information.

Minor point

It might be better to add clinical diagnosis (sub-classified diagnosis) in table 1.
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?
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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