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

Version: 0 Date: 09 Aug 2019

Reviewer: Lionel Van Maldergem

Reviewer's report:

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The paper by Ryu et al. describes a single case report of a very rare progeroid syndrome named Gorlin-Chaudhry-Moss or Fontaine syndrome. Its molecular basis has been recently identified as a heterozygous SLC25A24 point mutation occurring de novo resulting in a specific residue change.

Although the report does not add significant knowledge on these allelic conditions, it is interesting for the long term follow-up data it provides. It is also worth publishing single case reports when so few cases are available in medical literature.

Interestingly, the authors were able to make a clinical diagnosis based on the singular phenotype of their patient and requested the specific SLC25A24 sequencing data analysis after ATP6V0A2 sequencing proved unsuccessful.

However, there are a number of weaknesses that hamper the message to be delivered properly:

- Language should be checked by a native English-speaker

- Genetics is poorly described. Nomenclature, NM number p.[..., ..., precise description of methods used rather than vague terms

- Clinical photographs require absence of eyes blurring for a condition affecting the eyes.

More importantly, an added value would be given by serial photographs at different ages. Should the authors be able to obtain a full consent to publish these photographs, it will undoubtedly improve the general quality of the paper

- More attention should be paid to the clinical description. Results of paraclinic investigations, SD, ...

- The discussion would benefit from being more structured by describing resemblances and differences between gcms and pp, then explaining why ps might belong to the same spectrum, avoiding a mix-up of speculations and established data, then explaining why their case supports a lumping of ffs and gcms
The discussion on catch-up growth and development and therapeutic issues with comparison to Hutchinspon-Gilford syndrome is highly speculative and does not seem relevant.

Minor comments

P2L28. Outcome should be described in a distinct sentence. Progeroid features deserves specification. Skin wrinkling would benefit from being detailed: face, body, limbs?
P2L33 Below 1st centile does not tell the reader the number of deviations to the mean. Please express growth parameters in SD.
P3L11 A reference concerning lumping of GCMS and FFS is required. It seems paradoxical to give a new name referring to the first author of a secondary paper and to neglect the first author of the seminal paper.
P3L33 'recent advances in genetic technology' does not reflect adequately the current state of the art of recent genetic advances.
P3L50 please use proper nomenclature.
P4L8 should read 31w+6days. Oligohydramnios from 26 w.
P4L15 specify SD.
P4L20 separate sentence with facial dysmorphism description and other clinical features. Provide a clinical photograph if available.
P4L37 specify SD / dilatation?
P6L40 needs reference.
P7L25 please define the syndrome by its clinical manifestations.
P7L29 Since molecular characterization of Petty's syndrome does not exist until now, it is premature to consider GCMS, FFS and PLWS together.
P7L33 should read differences between genders/ 'specific genetic etiology' Does the author means molecular basis?
P7L40 statistics are inappropriate for including Petty's syndrome. Similarly, translating therapeutic considerations valuable for HPGS to GCMS and FFS does not make much sense due to different pathomechanisms and courses.

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

Unable to assess

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.

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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