Reviewer's report

Title: Inherited FGFR2 mutation in a Chinese patient with Crouzon syndrome and luxation of bulbus oculi provoked by trauma: A case report

Version: 0 Date: 31 Jul 2019
Reviewer: Anastasia Pilat

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Submitted publication "Luxation of bulbus oculi in Crouzon syndrome: a case report" by Ji Yang et al. presents the case of the patient with the rare combination of trauma and Crouzon syndrome. The topic would be interesting to the readers, however currently there are over 5 similar reports in the literature presented already.

Moreiver, there are some details that need to be addressed:

1. Authors state that they "have attempted to report a rare symptom in CS". In the described circumstances this is not a symptom of the Crouzon syndrome, otherwise it would be present without triggering trauma. Authors need to rephrase this statement as the luxation was provoked by the trauma due to the shallow orbits present in patient with Crouzon syndrome.

2. In the case presentation section I would suggest authors to describe all findings related to the proband and particular trauma case: findings just after the trauma, what treatment has been done, ocular findings after treatment (including eye movements, presence of strabismus/diplopia; nerve function-RAPD, nerve appearance, intraocular pressure). All this information can be interesting for readers. Then it will be logical to describe family members and genetic testing results.

3. Can authors please clarify what was actual visual acuity at the time of presentation? "The visual acuity of his left was at stake …"

4. Can the intraocular pressure be clarified as well?

5. Figures need to be re-written.

What is the difference in the results between CT and MRI? Authors need to comment what they want us to see on the scans? If it is ocular proptosis of the proband why on CT the sizes of the eyeballs look different? Is it motion artefact? Authors state that the facial photographs
of the affected subjects are shown. Only eye/orbital appearance is seen. These are nor facial
photos.

6. The conclusion "In conclusion, we identify a mutation in FGFR2 causing Crouzon
syndrome." is very strong. There are other publications describing this mutation in patients
with Crouzon syndrome including data from the Chinese population. Please refer to:
Jiayan Fan et al., An inherited FGFR2 mutation increased osteogenesis gene expression and
result in Crouzon syndrome. //BMC Medical Genetics volume 19, Article number: 91 (2018)
Anh Lan Thi Luong et al., Detection of G338R FGFR2 mutation in a Vietnamese patient with

7. Can authors please explain how the results of this study "enhance the current
knowledge of CS phenotypic and genotypic heterogeneity, but also assist genetic diagnoses
of a rare symptom in patient"? How the genetic retesting affected the particular symptom in
the presented case?

8. Extensive revision of English is needed before the publication will be accepted.

**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.

Unable to assess

**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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