Reviewer’s report

Title: Bilateral Giant Retinal Tears in Osteogenesis Imperfecta

Version: 0 Date: 01 May 2017

Reviewer: Pingping Zheng

Reviewer’s report:

Summary:

Osteogenesis Imperfecta is a heterogeneous genetic disorder commonly characterized by bones that break easily. The patients with OI genetic defect may also have blue sclera, dentinogenesis imperfect and otosclerosis. In this manuscript, the authors reported a 9 year old child with Osteogenesis Imperfecta (OI) subtype III and loss of vision and a retinal tear and detachment in right eye. After reviewing history of patients and clinical examination, the authors did the genetic screen of this patient and his parents to identify the OI subtype. After surgery, retinal detachment (RD) of the right eye was repaired and retinal stability was achieved in both eyes visual acuity was improved in both eyes. The authors found that a homozygous splice site mutation c.1914+1G>A (NM_001243246.1) of P3H1 gene in the patient, and his parents were carriers of this mutation.

Comments:

A few questions about genetic analysis: Page5 Line38-44."Genetic analysis revealed a rare autosomal recessive type of OI…"

The manuscript didn't describe the details of methods and introduction of this gene. It was not clear that:

1. What kind of screening methods were used in the manuscript? How did the authors screen the mutation?

2. Was P3H1 the only screened gene or locus? If yes, why this gene was selected/importance for this case? If not, which other candidate genes were screened? How did the authors select this mutation as the disease causing mutation?

3. How did the authors analyze the screening data?

4. How did the authors conclude that the P3H1 mutation (c.1914+1G>A) was the disease causing gene or mutation?

5. Functional prediction: the mutation located at intron and was a splice site mutation. Would the authors infer protein sequence changes from this mutation and discuss possible dysfunction of this gene caused by a splice site mutation?
Most of the readers have no ideas about OI subtypes and their genetic characterization. If the authors can provide a little bit more information about disease causing genes/mutations in OI and RD in the introduction, that would help the readers to understand the results better.

**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.

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.

I am able to assess the statistics

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

Acceptable

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