Reviewer’s report

**Title:** A novel CHD7 variant disrupting acceptor splice site in a patient with mild features of CHARGE syndrome: a case report

**Version:** 0  **Date:** 17 Jun 2019

**Reviewer:** Frédéric Bilan

**Reviewer's report:**

SEMA3E should not be considered as a CHARGE syndrome causative gene. The authors should discuss the potential role of EFTUD2 (and maybe KMT2D) instead.

The patient described in the case report is a typical CHARGE syndrome patient according to Verloes updated criteria (or Hale). The authors should discuss why WES was performed rather than a targeted molecular analysis (many NGS panels containing CHD7 are available)

All the finding concerning GJB2 variant (non relevant for this case report) should be removed from the manuscript.

Figure 1 is useless and should be replaced by the RT-PCR gel electrophoresis.

As the CHD7 variation reported here should be classified "pathogenic" using the ACMG recommendation (no doubt about this !), the authors should explain why they perform a functional test.

**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:

Acceptable

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