Reviewer’s report

Title: Congenital emphysematous lung disease associated with a novel Filamin A mutation. Case report and literature review

Version: 0 Date: 11 Nov 2018

Reviewer: Ralph Épaud

Reviewer's report:

I read with interest the clinical case of Pelizzo et al. The case is well presented and accompanied by a complete review of the literature.

I have 3 major concern:

1) In the abstract, it is mentioned the term of interstitial fibrosis, but I could not find any data in the manuscript to ascertain this feature (should be delated I guess)

2) The genetic should be more precise. What is the type and the frequency of the mutation of the mutation (nonsense, stof , frameshift, etc…) as well as the bioinformatic prediction of pathogenicity. Is it predicted to be benign probably damaging (please specify the software used such as SIFT, MutationTaster, Polyphen, etc..)

3) In the discussion part, I would have appreciated a little paragraph on lung emphysema clinical characteristics etiology (genetics?) and treatment (quiet surprising for your patient)

Minor concern:

1) Why waiting so much for the surgery of congenital emphysema?

2) Surgical procedure is not clear to me. A left lobectomy is mentioned?? But also something ("limited resection") that appears to me as segmentectomy?

3) It is likely that your genetic analysis was oriented by the phenotype. I don't think you should then mention CF of surfactant gene that do not correspond to the clinical or imaging presentation

4) What is Schinzel-Giedon syndrome? Is there any reason to mention that here?

5) Figure 1: B)b panel is a sagittal? Please use arrows to show the lesion (emphysema) C: explain the coloration and use arrows to show the different abnormalities D) again , use arrows!
6) In the discussion, maybe you should focus

**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.

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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