Reviewer’s report

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

Version: 0 Date: 23 Dec 2018

Reviewer: Russell J. Ferland

Reviewer's report:

This is a case report that details a new pathogenic FLNA gene mosaic variant (c.7391_7403del;p.Val2464AlafsTer5) in a male who developed pulmonary emphysematous lesions and perivascular and interstitial fibrosis. The case is straightforward and I only have minor issues which would aid in assessing the manuscript. Were there other mutations identified in other genes with the genomic sequencing and how were these excluded as not being implicated. A table of such information would be useful. The letters in the actual figures are very hard to see and there appears to be mislabeled letters in the text (figure legends). It would also be helpful to have leaders pointing to the imaging defects in the figure. How was it determined that there were mosaic variants (were the parents sequenced)? Was the lung biopsy ever assessed for sequencing of FLNA or cDNA? Lastly, human gene names should be capitalized and italicized, and protein names being only capitalized.

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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