Reviewer's report

Title: PTPRF is disrupted in a patient with bilateral amastia

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Reviewer: Sebastiano Bianca

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The manuscript is interesting and carefully prepared showed a detailed molecular studies. From what has emerged from studies is possible to consider the PTPRF gene as a candidate gene for amastia even if the reported case is a syndromic case and not an isolated amastia case. The difference between the possible association of PTPRF gene in cases of isolated and syndromic amastia should be pointed out by the authors to avoid confusion between candidate genes in the two different forms. The conclusions raised by the authors seem to place with certainty the possibility that the PTPRF gene should be considered the amastia candidate gene but against the molecular data obtained do not provide conclusive results. It therefore invited the authors to change the conclusions stressing the possible role instead of as a certainty.

Greater clinical description of amastia, supported by pictures of the chest of the patient and the demonstration of instrumental tests performed (ultrasound, x-ray, MRI, etc.) may allow greater clinical case definition.

Level of interest: An article of importance in its field

Quality of written English: Acceptable

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:

I declare that I have no competing interests