Author's response to reviews

Title: PTPRF is disrupted in a patient with syndromic amastia

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Author's response to reviews: see over
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Anne M. Slavotinek, M.D., Ph.D.
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Dear Dr. Slavotinek:

Thank you very much for the encouraging response to our manuscript MS: 1456740163420130, “PTPRF is disrupted in a patient with bilateral amastia”. The paper has been revised according to the suggestions of the reviewers.

In response to the comments of reviewer 1:

Q1: 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 the 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.

As our studies revealed the PTPRF gene as a possible candidate for a syndromic form of amastia identified in our patient, the “syndromic amastia” will be used in this reported case throughout the manuscript to avoid confusion between candidate genes in the two different forms. We have also added “syndromic” to the title as “PTPRF is disrupted in a patient with syndromic amastia”. Please see the revised manuscript on page 1, the first line and page 3, the last paragraph.

Q2: The authors are invited to change the conclusions stressing the possible role instead of as a certainty.

As kindly suggested by the reviewer, to stress the possible role instead of as a certainty, some of the conclusions were changed to “We, for the first time, demonstrated the possible association of PTPRF with syndromic amastia, ……..” Please see the revised manuscript on page 3, last paragraph, under conclusions.

Q3: Greater clinical description of amastia, supported by pictures of the chest of the patient and the demonstration of instrumental tests performed may allow greater clinical case definition.

As there were no imaging studies of the chest performed before the total breast reconstruction was done on the patient, we were unfortunately unable to provide such documents as kindly suggested.

In response to the comments of reviewer 2:

Q1: Can they test the father to see if he has reduced expression of PTPRF compared to wild type?
As kindly suggested by the reviewer about testing the father, unfortunately the patient’s father was deceased. The sentence “Her father was deceased” was stated on page 5, line 14-15.

Q2: First two sentences of the abstract and background are identical.

As kindly pointed out by the reviewer, the first two sentences of the background were changed to “Excellent progress has been made in defining the signaling pathways responsible for mammary gland development in mice, current knowledge about human mammary gland development is however very restricted and requires further elucidation”. Please see the revised manuscript on page 4, lines 2-5.

Q3: P7: Transformation (was) occurred within 2 to 3 weeks after starting the culture.

The “was” was deleted from the text. Please see the revised manuscript on page 7, line 2.

Q4: How was the promoter sequence of PTPRF determined?

The PTPRF promoter was previously characterized (The allelic variant of LAR gene promoter -127 bp T-->A is associated with reduced risk of obesity and other features related to insulin resistance. J Mol Med 2004, 82(7):459-466). The PTPRF promoter was obtained by PCR and verified by direct sequencing. We added the position of the promoter used in this study and the reference to the “methods” section. Please see the revised manuscript on page 8, line 12.

Q5: Table 1: what are polywhorls?

The “polywhorls” in Table 1 are the polywhorls of scalp hair. The word “polywhorls” was previously used in the article (Nelson, M.M. and Cooper, C.K. S Afr Med J, 1982. 61(12): p. 434-6.). For better clarification, we added “of scalp hair” after polywhorls. Please see Table 1 of the revised supplementary information.

All the changes suggested by the reviewers were made in RED font. The author names and references are checked. Text, figures, tables are revised and correctly formatted.

We really appreciate reviewers’ helpful comments. Thank you very much for your kind consideration of this work. We hope that it meets all the requirements of the journal.

Sincerely,

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