Reviewer's report

Title: PTPRF is disrupted in a patient with bilateral amastia

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Reviewer: Anne Slavotinecraft

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PTPRF is disrupted in a patient with bilateral amastia

The authors describe an interesting constellation of features in a woman with bilateral amastia, ectodermal dysplasia and unilateral renal agenesis who had a balanced chromosome translocation, 46,XX,t(1;20)(p34.1;q13.13). The breakpoint on chromosome 1 was found to interrupt the PTPRF gene and RNA and protein from the patient were found to be significantly less than those of her unaffected family members. The proband had inherited a different PTPRF allele from her father compared to her sisters, and the authors hypothesize that a second mutation on this allele is responsible for the phenotype in the patient, although they could not detect it.

This is a well-written paper and the authors have done their best to implicate PTPRF in the pathogenesis of the amastia but they did not find a second mutation. However, can they test the father to see if he has reduced expression of PTPRF compared to wildtype? This is assumed from the proband’s results.

Minor points:
First two sentences of abstract and background are identical
P7: Transformation (was) occurred within 2 to 3 weeks after starting the culture.
How was the promoter sequence of PTPRF determined?
Table 1: what are polywhorls?

Quality of written English: Acceptable

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

Declaration of competing interests:
no conflict of interest