Reviewer's report

Title: Cryptic FMR1 mosaic deletion in a phenotypically normal mother of a boy with Fragile X Syndrome: case report

Version: 1 Date: 4 August 2014

Reviewer: Giovanni Neri

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Minor essential revisions

On p. 9 and again in the Conclusions section the expression "focused prenatal diagnosis is strongly recommended for normal conception" should be changed to "focused prenatal diagnosis is strongly recommended to verify the normality of the fetus".

The authors should not overemphasize the need to do the kind of careful study they did, just for the sake of prenatal diagnosis. In a case like this, even if the mother had been found normal in all tissues, prenatal diagnosis would still be indicated, given the possibility of a germline-confined mosaicism.

In Fig. 2C there is a symbol for the proband, which however is not visible in the bar graph.

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: no competing interests