Reviewer's report

Title: Germline mosaicism in X-linked periventricular nodular heterotopia

Version: 2 Date: 18 April 2014

Reviewer: Elena Parrini

Reviewer’s report:

This manuscript reports germline mosaicism in a family with two sisters with FLNA-associated PNH due to a loss of function mutation in FLNA and clinically unaffected parents. The association of FLNA germline mosaicism and PNH has never been reported before. These findings are important for genetic counselling.

I have only minor concerns:

- In the Mutation Analysis section, the authors should clarify how they verified parental relationships.
- In the Discussion section, the authors state that mutation detection in the parents was limited to peripheral lymphocytes. If possible, it would be highly relevant to test at least another type of cells (ex: saliva DNA) to exclude somatic mosaicism and strengthen the hypothesis of germline mosaicism.
- The FLNA mutation nomenclature reported in the Abstract differs from that reported in the Mutation Analysis section.

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