Reviewer's report

Title: Germline mosaicism in X-linked periventricular nodular heterotopia

Version: 2 Date: 13 May 2014

Reviewer: chantal C Depondt

Reviewer's report:

This is the first report of an apparent germline mutation of the FLNA gene resulting in BPNH in 2 sisters with unaffected parents. It is relevant to the field and important for genetic counseling.

The article is clearly written.

Minor revisions:
1. Abstract: line 1, "disorder" instead of "malformation"
2. Abstract, line 5: better "partial loss of function" instead of "partial or residual function"
3. Description of the second patient: it is not surprising that the EEG "did not reveal any electrographic seizures". Do the authors mean instead that there were no interictal abnormalities? Please clarify.
4. Mutation analysis: the coding of the protein mutation differs from that given in the abstract. Please correct.

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 have no competing interests