Reviewer's report

Title: De novo frameshift mutation in ASXL3 in a patient with global developmental delay, microcephaly, and craniofacial anomalies

Version: 1 Date: 29 July 2013

Reviewer: EC Tan

Reviewer's report:

• Major Compulsory Revisions
1. This paper describes the identification of a novel de novo 2-bp deletion in ASXL3 in a child with HHF1, developmental delay, microcephaly, autism and dysmorphism. The work adds to the current knowledge and the spectrum of mutations for the gene in a clinical population, it is of interest and there is no major error.

• Minor Essential Revisions
2. Instead of citations 18 and 19 which were not so appropriate, it would be more useful for the authors to provide a couple of sentences about the major features of the database.
3. Suggest revising “Conclusions” which is very lengthy. This section includes discussion on the position of the 2-bp mutation and comparison with another previously reported variant. It goes on for 2 full pages and should more appropriately be called “Discussion”.

• Discretionary Revisions
4. Can the authors specify whether CPGM contain data from healthy individuals such as family members?
5. A figure with the gene structure showing where the deletion occurs and the hotspots, if any (according to S Table 1), would be useful.

Level of interest: An article whose findings are important to those with closely related research interests

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