Reviewer’s report

Title: A homozygous AHI1 (Abelson Helper Integration site 1) gene mutation (p.Thr304AsnfsX6) in a consanguineous Moroccan family with Joubert Syndrome: a case report

Version: 6  Date: 29 September 2015

Reviewer: Michele Roberts

Which of the following best describes what type of case report this is?: Other

If other, please specify:

Genetic diagnosis.
Rapid detection of a disease-causing mutation in a family affected with a highly genetically heterogeneous disorder.

Do you believe the case report is authentic?: Yes.

Do you have any ethical concerns?: No ethical concerns. Diagnostic molecular genetics.

Is the Abstract representative of the case presented?:

Yes. The abstract describes the genetic disorder and the mapping/sequencing methods, summarizes the results, and mentions the implications.

Does the Introduction explain the relevance of the case to the medical literature?:

Yes. The introduction describes the clinical features of the disorder in the context of what has been described in the literature about this disorder and related disorders, explains the genetics, presents a family, and summarizes methods used and results obtained.

Does the article report relevant patient information?: Yes

Does the article report relevant physical examination findings?: Yes

Does the article report important dates and times in this case?: Yes
Does the article report the diagnostic assessments?: Yes

Does the article report the types of intervention?: No

Does the article report a summary of the clinical course of all follow-up visits?: No

If any information is missing from the reporting, please detail it here.:

This report was not about lifestyle issues, intervention, clinical course, or follow-up.
All relevant information was presented. Consanguinity, ethnicity, diagnostic techniques, and clinical features were adequately discussed.

Is the interpretation (discussion and conclusion) well balanced and supported by the case presented?:

Yes.

Does the case represent a useful contribution to the medical literature?:

Yes. Joubert syndrome is a genetically heterogeneous disorder with variable expressivity.

Was written informed consent to publish this case obtained?: Yes

Is the anonymity of the patient protected?:

No names, facial images, or identifying information other than age and ethnicity is/are presented.

Additional comments to authors:

I made a few additional recommendations in the text, mostly very minor stylistic and one small grammatical correction ("an 18-year-old").

Level of interest: An article of importance in its field

Quality of written English: Acceptable

Declaration of competing interests:

I declare that I have no competing interests.