Author’s response to reviews

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

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Version: 2 Date: 26 December 2014

Author’s response to reviews: see over
Cover letter

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We certify that the case report untitled “A rare homozygous AHI1 mutation in a Moroccan consanguineous family with Joubert Syndrome” that we submit for publication to Journal of Medical Case Reports.
- has not been published previously, that it is not under consideration for publication elsewhere, that its publication is approved by all authors.
- has been read and approved by all the authors.
- there is no financial or other conflict of interests

Otherwise, our article described a first case not described before of homozygous mutation (p.Thr304AsnfsX6) in AHI1 gene causing a JBTS in a Moroccan family with clinical specificity. This homozygous mutation would contribute to phenotype/genotype correlation in AHI1 mutated gene. Our cases showed that this syndrome is clinically and genetically heterogeneous.
More interestingly, this rare mutation was found only in heterozygous status, especially in a patient from Spain, which suggested a founder effect. The second hypothesis suggesting an hotspot mutation when considering the two patients mutated from Nethelands.
This result allows an accurate genetic counseling and may offer a molecular prenatal diagnosis to this family.

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