Reviewer’s report

Title: Clinical and Molecular Characterization of Ataxia with Oculomotor Apraxia (AOA) Patients In Saudi Arabia

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Reviewer: Vinodh Narayanan

Reviewer’s report:

This is a study relating genotype to phenotype in patients diagnosed with ataxia with oculomotor apraxia (AOA). The authors describe nine patients from four consanguineous families, summarize their clinical findings, and were able to define the gene mutation in two of these families. In one family, they identify a novel mutation in the SETX gene, and in another family, they demonstrate (by linkage and sequencing) a mutation in the MRE11 gene (associated with ATLD). In two of the families, no causative mutation was discovered in the SETX or APTX gene, but linkage analysis to localize the gene defect apparently has not been done.

This report emphasizes the phenotypic heterogeneity in this group of disorders, and demonstrates the importance of genetic screening of a group of genes implicated in the AOAs.

Minor essential revisions:

The authors might consider describing the phenotypes of the patients in some more detail - for instance, were oculocutaneous telangiectasias present in any of them? Were there white matter T2 hyperintensities on MRI scanning? Did they perform a colony sensitivity assay (radiation sensitivity) in lymphoblasts from any of these patients?

A number of minor typographical/spelling errors should be corrected.

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

Quality of written English: Needs some language corrections before being published

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.