Reviewer's report

Title: Exon deletions and intragenic insertions as a frequent cause for ataxia oculomotor-apraxia 2

Version: 1 Date: 20 May 2009

Reviewer: craig bennett

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Major Compulsory Revisions
At the end of the Background section (basically the introduction) it is stated that the analysis was undertaken for ten patients with symptoms and signs characteristic of AOA2. However, this is the last mention of patients in which no SETX mutations were detected. Also what is meant by … “absence of ataxia in the family history”? Table 1 should be expanded to include the other six patients in which no SETX mutations were detected? Given that the general features of AOA2 in combination with the mildly elevated AFP is usually a good indicator of senataxin defects, one wonders if the other six patients did or did not show elevated AFP, for example?

Essential Revisions:
In the results and conclusion section of the Abstract – the wording could be clearer. For example, “Sequence analysis in four patients failed to reveal two mutations” - this should be rephrased.

Discretionary Revisions:
For patients that did show elevated AFP but were absent detectable SETX mutations; might expression level qPCR potentially indicate the presence of regulatory sequence mutations?

Level of interest: An article of importance in its field

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.