Reviewer's report

Title: Triple-A Syndrome with Prominent Ophthalmic Features and a Novel Mutation in the AAAS Gene: Case Report

Version: 3 Date: 14 May 2004

Reviewer: Elias I Traboulsi

Reviewer's report:

General

This is a very well written paper that presents a new case of AAA syndrome with novel mutations in the AAAS gene. Some issue needs to be resolved with respect to the general ocular findings and their interpretation.

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Major Compulsory Revisions (that the author must respond to before a decision on publication can be reached)

The authors interpret some of the ocular findings as evidence of autonomic dysfunction. While this is very possible for the strabismus and accommodation abnormalities, the reduced visual acuity in both eyes despite best refractive correction, the color vision abnormalities and the light sensitivity needs to be explained. The possibility of a retinal dystrophy involving the cone system has not been eliminated and the patient has not had an electroretinogram. A cone dystrophy can certainly explain the reduced vision, light sensitivity and color vision deficit. The fundus can be completely normal in such instances. An ERG has to be done to rule this possibility in or out.

Near vision also needs to be tested to determine whether the presumed dysfunction in accommodation has affected near vision.

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Minor Essential Revisions (such as missing labels on figures, or the wrong use of a term, which the author can be trusted to correct)

None

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Discretionary Revisions (which the author can choose to ignore)

None

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What next?: Unable to decide on acceptance or rejection until the authors have responded to the major compulsory revisions

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

Quality of written English: Acceptable
Statistical review: No