Reviewer’s report

Title: Patients affected with Fabry disease have an increase incidence of progressive hearing loss and sudden deafness

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Reviewer: Dr Gregory Pastores

Level of interest: A paper whose findings are important to those with closely related research interests

Advice on publication: Accept after discretionary revisions

The paper by Germain and colleagues, based on investigations performed on 22 males with Fabry disease (FD), documents the common occurrence of progressive hearing loss as a complication. There are several reports documenting the occurrence of this problem in patients with FD, including the recent paper by McDermott et al (J Med Genet 2001 Nov;38(11):750-60) which noted high frequency sensorineural deafness in hemizygous males (N=98), which was confirmed in 78% of audiograms. However, Germain's paper focuses on this problem in particular and does provide a more detailed characterization of the problem based on the systematic assessment of patients and proposes a basis for its development. Furthermore, they correlate the findings with other FD-related complications (resulting from renal, cardiac and neurologic involvement). The authors site the probable role of vascular occlusion, which may have a contributory role and the observed correlation with cerebrovascular involvement is suggested as supportive of this hypothesis. The toxicity associated with certain medication explains the co-occurrence of renal dysfunction and hearing impairment exposed to these drugs. A similar pattern of morbidity encountered in FD is used by the authors to allude to the possibility that the glycolipid storage may lead to cytotoxicity along the same lines.

Suggestions:

It may be useful to the readers for the authors to provide their recommendation for assessment of hearing impairment in FD patients in light of the fact that all patients had sensorineural loss.

Rather than providing a table which details the actual test results, it may be more helpful to list the patients' clinical findings in tabular, including a column on hearing status (the final column of the paper's Table 1), and noting the presence or absence of renal, cardiac and cerebrovascular disease with + or - signs and giving the value for GFR in parenthesis for instance, for the patients with renal insufficiency or failure and other notations accordingly. At listing of age at onset if known may also be of interest, in relation to that for renal dysfunction.

Competing interests:
None declared.