Reviewer's report

Title: Type I Gaucher disease with exophthalmos and pulmonary arteriovenous malformation

Version: 2 Date: 22 February 2005

Reviewer: Deborah Elstein

Reviewer's report:

Major Compulsory Revisions
1. The authors have adhered to nearly all my recommendations of major importance for the validity of their presentation. However, in the spirit of my original suggestion that the fact of dissimilar phenotypes among the three sisters with the same genotype is awkward to say the least if the authors wish to make sweeping statements such as "This case should give important implications for the mechanism of Gaucher disease.", I would require these attempts at universality /commonality be eliminated (see the Abstract and the Conclusions).
2. The Discussion of AVM abnormality and hepatopulmonary syndrome as versus pulmonary hypertension because of ERT is overly long, primarily since we see no overt evidence of severe hepatic disease other than a mildly enlarged liver on palpation, apparently. Whereas I accept that these authors are committed to continue ERT in this patient, the Discussion should not be skewed to hepatopulmonary if there is no hepatic component in this patient.
3. While the syntax, spelling and grammar are greatly improved, the text needs some help from a native speaker and or "spellcheck".

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: Needs some language corrections before being published

Statistical review: No

Declaration of competing interests:

I declare that I have no competing interests.