Reviewer's report

Title: An Atypical Variant of Anderson-Fabry Disease in a Female Patient with Renal and Cardiac Involvement and Transient Ischemic Attack: a Case Report

Version: 1 Date: 25 January 2012

Reviewer: Massimiliano Veroux

Reviewer's report:

Major Compulsory revisions

The Authors present an interesting case of atypical presentation of Fabry Disease in a young female with atypical mutation in the intronic regions of GLA gene.

The paper is well written and present many points of interest. However, I believe that some additional data should be included in the presentation of the case.

1) The introduction should be reduced, focusing on the description of cardiac symptoms and genetical alterations of Fabry Disease, which is a well known genetic disease that do not require long description in the introduction section.

2) Was an enzyme replacement therapy considered in this patients? If yes, which ERT?

3) Giving the clinical nature of the Journal, it could be useful to insert some data of the follow up of the patient, if available.

4) the authors should better specified the originality of this case report, focusing in the rarity or significance of the atypical genetic variation and how this could reflect in clinical practice.

Level of interest: An article of importance in its field

Quality of written English: Acceptable

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