Reviewer’s report

Title: The coincidence of IgA nephropathy and Fabry disease

Version: 1 Date: 13 August 2012

Reviewer: Paula Rozenfeld

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Minor Essential Revisions

1) a-galactosidase A: correct a for alpha or the symbol #

2) In the introduction authors say “We present two patients with very rare coincidence of IgAN and FD.” And in discussion they say there are 6 reports of FD and IgAN. The coincidence is not “very rare”.

3) The actual nomenclature of the mutation should be p.Ile317Thr. The same values for the other mutation.

4) On enzyme replacement therapy myocardial hypertrophy has stabilised and other difficulties have disappeared

What do you mean with other difficulties?

5) Heterozygous females, due to different levels of alpha-galactosidase A activity, may have various clinical signs from asymptomatic to severe.

The reason why females display heterogeneous clinical manifestations is not due to different levels of enzymatic activity.

6) On the other hand, Fabry’s disease is rare in Japan, even though it’s specific incidence has not been assessed [22].

I would delete this phrase if there is no conclusive data.

Moreover, Fabry disease is a rare disease worldwide.

Major Compulsory Revisions

7) After the patient’s informed consent was obtained, DNA and RNA analyses (the standard restriction fragment length polymorphism method) were performed.

There is no standard RFLP analysis for Fabry disease. What was the genetic analysis performed?

8) Regarding case 1, authors wrote “electron microscopy was not performed because only sclerotic glomerulus was present in the sample”. Why did they say that? After that, in the next paragraphs, they wrote the result of electron microscopic study. Regarding EM study, what does “inclusive” mean in the sentence inclusive zebra bodies? I suggest including the pictures of
immunofluorescence, light microscopy and EM of this case.

9) Authors stated the patient had no other clinical symptoms apart of headache. However, after that they reported the patient suffered from febrile crisis and neuropathic pain in the extremities.

10) In the 2nd patient, fig 2b, the presence of foamy cells should be stated. The detection of foamy cells is useful for the suspicion of Fabry disease diagnosis.

Level of interest: An article of limited interest

Quality of written English: Not suitable for publication unless extensively edited

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:

I have received research grants and consultant fees from Shire HGT.