Reviewer’s report

**Title:** Co-existence of Phenylketonuria and Fabry Disease on a 3 year-old boy: Case report

**Version:** 1  **Date:** 19 February 2010

**Reviewer:** Tauro M. Neri

**Reviewer’s report:**

This short report can be interesting for clinician working on rare metabolic diseases.

The clinical part is correctly reported.

I have some remarks about the genetic part of the work.

At the end of the first paragraph of page 4 (case presentation section) the authors wrote of a ‘family history’ that was not reported before (perhaps the maternal uncle?). A short family history should be added to explain.

In the sixth line of the first paragraph of page 4 the mutation in the protein is not correctly written (should be p.L48S and not p.[L48S]). The mutation at the nucleotide level should be reported as well and its effect of the genetic code explained.

The same should be done for the mutation of GLA gene.

In the second line of the second paragraph of page 4 the word GLA (name of the gene) should be erased, because what is tested is the enzymatic activity of a protein and not a gene coding for that protein.

End of the paragraph 2 of page 4: which members of the maternal side of the pedigree were tested for enzymatic activity?

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

**Quality of written English:** Acceptable

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

**Declaration of competing interests:**

I have not competing interests