Reviewer's report

Title: Identification of a deep intronic mutation in the COL6A2 gene by a novel custom oligonucleotide CGH array designed to explore allelic and genetic heterogeneity in Collagen VI-related myopathies.

Version: 2 Date: 16 December 2009

Reviewer: Lee-Jun Wong

Reviewer’s report:

There is no evidence that the absence of cDNA is linked the intronic deletion of 2 probes because this deletion cannot be confirmed by PCR. The absence of cDNA could be due to other unidentified mutations. This fact should be made clear in the manuscript.

The significance of V117A is not known. The software analyses yielded discordant results.

Level of interest: An article of limited interest

Quality of written English: Needs some language corrections before being published

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