Reviewer's report

Title: Novel PTRF-CAVIN mutation in a child with mild myopathy and very mild congenital lipodystrophy

Version: 4 Date: 23 May 2013

Reviewer: Anurupa Maitra

Reviewer's report:

Major Compulsory Revisions:
1. Case Report Para 6: Authors claim that the novel mutation introduces a STOP codon 27 bases downstream of the correct STOP codon. This needs to be substantiated by the DNA sequence. Evidence from Fig 1 indicates a protein larger than the expected size. Needs clarification.

Minor Essential Revisions:
1. Discussion Para 3: Delete "therefore unsurprising". Insert "not surprising".

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, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:

I declare that I have no competing interests.