Reviewer’s report

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

Version: 4 Date: 27 May 2013

Reviewer: YASUHIRO TAKEHIMA

Reviewer’s report:

An article entitled "Novel PTRF-CAVIN mutation in a child with mild myopathy and very mild congenital lipodystrophy" is described about a congenital generalized lipodystrophy type 4 (CGL4) case with a novel mutation in the PTRF-CAVIN gene. Until now, few PTRF-CAVIN mutations have been reported in humans, and this article is the fifth reports.

This article is relevant to this journal, and mentions the original and interesting result. However some points should be reconsidered as below.

#1 One of the major findings of CGL is the decrease of serum leptin and adiponecint levels. These levels should be mentioned in this article

#2 In the case of heterozygous mutation of one nucleotide deletion, double waves should be appeared downstream from the deleted nucleotide. However, sequence results of parents in figure 3D, double waves are appeared upstream from the deleted nucleotide. The reason should be mentioned.

Level of interest: An article of limited interest

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.