Reviewer’s report

Title: APOA5 Q97X mutation identified through homozygosity mapping causes severe hypertriglyceridemia in a consanguineous family.

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Reviewer: Angelo Baldassare Cefalu

Reviewer’s report:

The manuscript by Dussaillant et al describes a family carriers of a mutation of the APOAV gene (Q97X) responsible of severe hypertriglyceridemia.

1) It is not properly explained why the authors used the genomewide linkage study approach as first line laboratory strategy. It is true that mutations in the known candidate genes explain a minority of cases with a severe hypetrygliceridemic phenotype but since the main aim of this study was to determine the genetic locus responsible for the severe HTG present in this consanguineous family the resequencing of the candidate genes in which pathogenic mutations have been previously described should have been considered as first line laboratory approach.

2) The discussion section needs extensive revision focusing the findings of this work and shortening the section describing the mechanisms of action of ApoAV.

Since a known mutation in one of the candidate genes was identified (Oliva et al., Charriere et al) it would be interesting to discuss the two different laboratory approach (gene resequencing vs genome wide) In terms of costs and time employed.

3) figure 3 and figure 4 may be moved to the supplemental data section since do not add major information

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