Reviewer’s report

Title: A missense founder mutation in VLDLR is associated with Dysequilibrium Syndrome without the quadrupedal locomotion

Version: 3 Date: 4 July 2012

Reviewer: Jillian Parboosingh

Reviewer’s report:

The authors have addressed most of the questions and concerns raised in the first review. The haplotype data supports the conclusion that a founder mutation exists in this population.

Major compulsory revisions.

None

Minor essential revisions.

1) abstract, results section: change “This is the first homozygous reported missense mutation” to “This is the first reported homozygous missense mutation”.

2) Page 5, paragraph 1. Missing opening brackets “heights (5-25%)”.

3) abstract, methods section: as written, readers would expect that massively parallel sequencing was performed in both cases. This should be edited to reflect the two approaches. It would be clearer to say something like “SNP mapping and candidate gene sequencing in one consanguineous Omani family from the United Arab Emirates with cerebellar hypoplasia, moderate mental retardation, delayed ambulation and truncal ataxia was used to identify the mutation. In a second unrelated consanguineous Omani family, massively parallel exonic sequencing was used”.

Discretionary revisions.

None

Level of interest: An article of importance in its field

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.