Reviewer's report

Title: Rare recessive loss-of-function methionyl-tRNA synthetase mutations presenting as a multi-organ phenotype

Version: 2 Date: 26 June 2013

Reviewer: Bernard Brais

Reviewer's report:

This case report contributes to the growing number of AAR related disorders. Though it is a single case report of mutations uncovered by exome sequencing, the authors have done what they could to confirm the functional impact of the mutations. I have no major corrections to propose. It is a well written paper that could be shorten and the biopsy images could be placed in the supplementals.

My only minor correction is: “The lack of symptoms in her heterozygous parents indicates that a PREDICTED ~40% reduction of MARS activity is well tolerated.”

Level of interest: An article whose findings are important to those with closely related research interests

Quality of written English: Acceptable

Statistical review: Yes, and I have assessed the statistics in my report.