Reviewer's report

Title: Homozygosity for a mis-sense mutation in the 67kDa isoform of glutamate decarboxylase in a family with autosomal recessive spastic cerebral palsy: parallels with Stiff-Person Syndrome and other movement disorders.

Version: 2 Date: 4 November 2004

Reviewer: Lisbeth Tranebjaerg

Reviewer's report:

The re-submitted paper has incorporated some modifications following the reviewers’ comments.

The causal relationship between the identified missense mutation and the disease has not been clarified compared to the first version of the MS. In this regard, a search for mutations in this gene in a large number of CP patients would be highly relevant for determining the pathogenic role of this gene in the etiology of CP. Negative results from screening a large series of CP patients might not disprove the possibility of a causal relationship since the genetic fraction of CP is estimated to be as low as 1-2 %, but a positive result would be a strong support.

Similarly, it would add considerable strength to the present MS if the results from performed studies of transfection assays were actually reported within the MS.

Minor point: the four tables were not marked with table number.