Reviewer’s report

Title: A possible role for miRNA silencing in disease phenotype variation in Swedish transthyretin V30M carriers

Version: 1 Date: 7 May 2010

Reviewer: Joel Buxbaum

Reviewer’s report:

With respect to the analysis the authors do not appear to have corrected the significance values for multiple comparisons.

While the speculation regarding microRNA binding sites is interesting they provide no evidence that it is the case. If they showed some correlation with serum TTR levels of individuals with each allele then the argument concerning the role of microRNA suppression of expression would have been more valid. In addition most TTR mutations in most populations are associated with lower TTR serum levels (although levels of abnormal protein have not been determined in all) yet the Swedes have reduced penetrance.

The SNP analysis was clearly worthwhile but they have not done appropriate follow up experiments to justify the hypotheses generated by the computational analyses.