Reviewer’s report

Title: A novel study of Copy Number Variations in Hirschsprung disease using Multiple Ligation-dependent Probe Amplification (MLPA) technique.

Version: 2 Date: 31 August 2009

Reviewer: Sarah H Elsea

Reviewer’s report:

1. It is still not clear--were the 2 mutations reported in this study previously identified as variants in these cases or were they only discovered after MLPA gave spurious results?

2. Are the variants described commonly seen in this or any population? or are they private mutations? This could have a great importance on the effectiveness of the MLPA kit.

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