Reviewer's report

Title: Novel MLPA procedure using self-designed probes allows comprehensive analysis for CNVs of the genes involved in Hirschsprung disease

Version: 1 Date: 10 March 2010

Reviewer: Sarah H Elsea

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Major compulsory revisions:
1. The conclusion that an alternative peptide could be translated from the deleted allele seems to be purely speculation. Devoting a figure to this, seems unnecessary unless there is experimental evidence to support such a conclusion. For example, western analysis or expression studies to support creation of an alternative transcript. Just because a methionine exists does not mean mRNA is transcribed to create a protein.
2. Discussion is extremely long and wordy. It should be reduced.

Minor essential revisions:
1. Numerous spelling and grammatical mistakes. Suggest having a primary English speaking scientist review the text prior to final submission.
2. Table 1: change "pb" column heading to "bp".
3. Figure 1: not clear what the differences are between panels A and C or panels B and D. Are these the same samples, same assays? If so, one of each is sufficient. there is no indication of a patient number being used in these experiments. More details is the legend would be helpful. As well, adding arrows pointing to the affected exons would be most useful, as the traces are quite small and may be difficult to see by the reader.

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.