Reviewer's report

Title: A unique phenotype in a patient with a rare triplication of the 22q11.2 region, and new clinical insights of the 22q11.2 deletion and microduplication syndromes: a report of two cases

Version: 4 Date: 21 June 2015

Reviewer: Heather McDermid

Reviewer's report:

The authors have made almost all my requested changes. The changes to the Table are particularly useful. The manuscript now describes two interesting patients with a duplication and triplication of the 22q11.2 region, and information that will add to the literature for this syndrome.

A few minor essential revisions:

Naming the cases: I understand that the naming of the cases (case 1, case2, etc) must follow the journal convention. However, please indicate for each patient the corresponding name in Pires et al.

For instance: Patient 1 (corresponding to Patient C in Lires et al) is a 23 year-old female …

In the abstract and line103 Case 1 is referred to as a 23 year-old. In Lires et al, she is referred to as a 24 year-old. Which is it?

When referring to the new feature that case 1 illustrates, you need to say what that feature is. Are you referring to hyperdontia, the discordant twins, or both.

- line 122, please do not refer to a 23 year old female as “the girl”. The woman? The patient?

- line 134, please clarify who has mild intellectual difficulties …

- line 135, please do not refer to a 20 year old female as “the girl”

- line 208-211. Please add “differences in genetic background” as a reason that triplications may vary in severity. This is probably the most likely reason.

There are still minor grammatical errors to fix in editing, especially in the new sections.

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.