Author's response to reviews

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

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Author's response to reviews: see over
Ponta Delgada, July 22\textsuperscript{nd}, 2015

Dear Doctor Nawsheen Boodhun,

Executive Editor of \textit{BMC Pediatrics}

It is with great pleasure that I am resubmitting to \textit{BMC Pediatrics} the re-revised manuscript “A unique phenotype in a patient with a rare triplication of the 22q11.2 region and new clinical insights of the 22q11.2 microduplication syndrome: a report of two cases” (MS: 2042602960148469) by Sara O. Vaz, Renato Pires, Luís M. Pires, Isabel M. Carreira, Rui Anjos, Paula Maciel and myself.

Please note that:
\begin{itemize}
  \item one of the Reviewers (Dr. Shi-Hui Yu) proposed that the manuscript was modified appropriately and did not make any additional comments
  \item the comments from the other Reviewer (Dr. Heather McDermid) were very helpful and added value to our manuscript
  \item we addressed all of Dr. McDermid’s comments
  \item all changes made from the revised manuscript are highlighted in blue in the re-revised version.
\end{itemize}

I appreciated the Reviewers’ work and hope that this manuscript is now suitable for publication in \textit{BMC Pediatrics}.

Kind regards,

Luisa Mota-Vieira

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

Answers to Reviewer HM (Dr. Heather McDermid)

Comments  Author’s corrections

Minor Essential Revisions

☐ The Reviewer mentions “please indicate for each patient the corresponding name in Pires et al. For instance: Patient 1 (corresponding to Patient C in Pires et al.) is a 23 year-old female...”. We agree and indicated for patient 1 and 2 the corresponding names in Pires et al., which are patient C and B, respectively (please see Case Presentation section, page 6 line 105, and page 7 line 136).

☐ The Reviewer asks why “In the abstract and line 103 Case 1 is referred to as a 23 year-old. In Pires et al. she is referred to as a 24 year-old”. We updated the age of patient 1 to 24 year-old, which is her age at the second clinical evaluation. There were no clinical differences comparing with the first evaluation performed at 23 year-old (please see Abstract section, page 3 line 50; Case Presentation section, page 6 line 105; and Table 1, page 16).

☐ The Reviewer points out that “When referring to the new feature that case 1 illustrates, you need to say what that feature is”. We agree and proceeded accordingly, referring this new clinical feature as hyperdontia (please see Abstract section, page 3 line 61; Background section, page 6 line 98; Conclusions section, page 8 line 165, and page 10 line 202).

☐ The Reviewer advises that is better to refer to a 23 year-old female as “the woman” or “the patient”, instead of “the girl”. We agree and changed the term “girl” by “patient” (please see Cases Presentation section, page 7 line 125).

☐ The Reviewer asks for clarification regarding the person “who has mild intellectual difficulties...”. We added information in order to refer that this person is case 2’s father (please see Cases Presentation section, page 7 lines 137-138).

☐ The Reviewer suggests that is not appropriate to refer to a 20 year-old female as “the girl”. We agree and proceeded accordingly, changing “girl” by “patient” (please see Cases Presentation section, page 7 line 139).

☐ The Reviewer requests the addition of “differences in genetic background” as a reason that triplications may vary in severity. We agree and added this information (please see Conclusions section, page 10 lines 213-214).

☐ The Reviewer refers that “There are still minor grammatical errors to fix in editing, especially in the new sections [from the revised version of the manuscript]”. We appreciated this comment and corrected these errors, after a thorough reading of the manuscript (please see lines 62-64, 126, 170, 181-182, 203-204, and 210-211).

Quality of written English

☐ The Reviewer states that the manuscript “needs some language corrections before being published”. We also appreciated this comment and proceeded accordingly (please see the previous response to the Reviewer).