Reviewer's report

Title: Associations between speech features and phenotypic variability in Treacher Collins syndrome

Version: 1 Date: 13 December 2013

Reviewer: Anneli AY Yliherva

Reviewer's report:

Major Compulsory Revisions
none

Minor Essential Revisions
None

Discretionary Revisions
1. The introduction is well done but a little bit hard to follow for a reader with such a huge amount of information. One suggestion would be to make a table including all potential phenotypic expressions of TCS including structures in different areas of head and neck.

2. In background -section abbreviations are not used logically. For example Treacher Collins syndrome is sometimes written and sometimes abbreviated. This is the same even later in the manuscript. This is a small thing but careful reporting is important in scientific presentations.

3. The questions 1 and 2 are appropriate and well described, but the question number 3 is a little bit unclear because the speech is not defined. I have understood that the speech here means speech composite score including six different characteristics.

4. There is missing information concerning the areas from where the TCS cases came. Were they living nearby Oslo area or were they also from other areas? The prevalence of TCS is low (1 in 50 000 live births), so in Norway there should be in the whole country approximately 100 TCS cases? In that sense the number of 19 cases in the study sample is sufficient even though the age groups of children's and adolescents' are small including only 4 cases each.

5. When introducing the participants it would be interesting to know why the age groups were divided from 5 to 10 years, from 11 to 18 years and from 19 onwards. A professional knows that it is something to do with the growth but still justification for this would be useful.

6. One point which occupies is the use of parametric tests for testing normality (the Kolmogorov–Smirnov test (K–S test) which is a nonparametric test for the equality of continuous, one-dimensional probability distributions that can be used
to compare a sample with a reference probability distribution (one-sample K–S test), or to compare two samples (two-sample K–S test). But I suppose the authors could explain and justify this question.

6. I couldn’t find the titles of the figures, and also the abbreviations should be opened in all figures.

7. The discussion is quite extensive and also includes a lot of comments and suggestions on clinical applications. This is really a good thing but I would have put all the clinical suggestions in the end of the discussion or in the conclusion. In addition, there was a little bit repetition of results in some points. In conclusion the authors state that adults should be followed but what about children and adults? And how we could know what kind of phenotype especially is important to follow more carefully. What could be the signs of speech characteristics to screen in TCS cases etc. For a SLT all practical and concrete information is always very useful. What would be also useful is to summarize the information more concretely to help clinicians to follow TCS cases carefully.

8. I think it would have needed more discussion about the limitations concerning the children and adult groups (N=4 each). The authors put them in the same group but children between 5 to 10 and 11 to 18 years are different but how? They were first studied separately but I think they should have been discussing if this is the developmental group? The lack of information concerning the criteria how the groups were divided is essential here to report.

9. The title and abstract convey what has been found although the speech function/characteristics were also related not only to phenotypic variability/severity of TCS but also orofacial dysfunction measured by NOT-S. In that sense it should be: Association between speech features and phenotypic variability OR severity and orofacial function OR dysfunction in Treacher Collins syndrome. The title is not really wrong but a little bit imprecise (see also Background in Abstract). In abstract section and in the methods the authors express they have 19 cases between 5 to 74 years, but there is a wide gap between 19 and 29 years of age. Maybe it would have been useful to report 19 cases in three groups: 5-10, 11-19, 29-74 years of age? The last group really concerns the grown-up people. There were really adults > 29 years.

**Level of interest:** An article of importance in its field

**Quality of written English:** Acceptable

**Statistical review:** Yes, but I do not feel adequately qualified to assess the statistics.

**Declaration of competing interests:**

'I declare that I have no competing interests'