Author's response to reviews

Title: An Intronic Alteration of the Fibroblast Growth Factor 10 Gene Causing ALSG- (aplasia of lacrimal and salivary glands) Syndrome

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Version: 4 Date: 20 August 2008

Author's response to reviews: see over
Reviewer's report
Title: An Intronic Alteration of the Fibroblast Growth Factor 10 Gene Causing ALSG- (aplasia of lacrimal and salivary glands) Syndrome
Version: 3 Date: 26 June 2008
Reviewer: Bernd Wollnik

Your comment:
The revision improved the quality of the manuscript. Especially the identification of the mutant transcript using an alternative splice site is an important new finding. The quantitative data are still not convincing and I would suggest to take abolish this part including figure 5. Afterwards, I highly recommend to accept and publish this paper.

Our response:
Using Real-time quantitative RT-PCR as well as sensitive, semi-quantitative RT-PCR analysis by silver-stained polyacrylamide gels, we were able to reproduce the finding of substantially diminished FGF10 transcript in the patient’s fibroblasts. However, as included in the discussion and in the previous reviewer’s response, we did not identify the underlying mechanism for transcript levels less than expected for heterozygous cells. As previously mentioned, one explanation could be that further single nucleotide variations in the promoter of the FGF10 gene, which was not subjected to sequence analysis, may result in diminished transcription. Therefore we would like to include the quantitative data.

Level of interest: An article of importance in its field
Quality of written English: Acceptable
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

Reviewer's report
Title: An Intronic Alteration of the Fibroblast Growth Factor 10 Gene Causing ALSG- (aplasia of lacrimal and salivary glands) Syndrome
Version: 3 Date: 4 July 2008
Reviewer: Hideyo Ohuchi

Your comment:
The manuscript has been well revised. Further minor modifications and comments are as follows:
1. Setting subtitles may help to read the results and “Analysis of the Fgf10 transcript" in Material and Methods, more smoothly.

Our response:
Subtitles are included now.

Your comment:
2. There is no description on the necessity and purpose of this study in Introduction, but there is in Discussion, such as “A combined aplasia, …..is rarely diagnosed.” and “These studies prompted us to perform genetic analysis to
identify...."

**Our response:**
The introduction has been reworked accordingly.

**Level of interest:** An article whose findings are important to those with closely related research interests

**Quality of written English:** Acceptable

**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.