Author’s response to reviews

Title: Two novel LHX3 mutations in patients with combined pituitary hormone deficiency including cervical rigidity and sensorineural hearing loss

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Manuscript Title: Two novel LHX3 mutations in patients with combined pituitary hormone deficiency associated with cervical rigidity and sensorineural hearing loss (BEND-D-16-00163)

Dear Zuleyha Karaca

Editor, BMC Endocrine Disorders

The authors would like to thank the reviewer for the comments. We have addressed the comment points and the corresponding changes have been incorporated in the revised manuscript. We hope this satisfies the editorial board to recommend publication.
Reviewer 2:

1). THE AUTHORS NEED TO ADD HOW THEIR ALLELIC VARIANTS ARE CLASSIFIED ACCORDING TO ACMG. YOU HAVE TO ADD AMONG THE 5 POSSIBILITIES (PATOGENIC, LIKELY PATOGENIC, VARIANT OF UNCERTAIN SIGNIFICANT, LIKELY BENIGN OR BENIGN) WHICH ONE IS THE RIGHT ONE FOR THE ALLELIC VARIANT YOU HAVE FOUND.

Thank you for asking to add this classification. We have added “Hence, the factors that p.Arg156Ter being a null variant and critical location of p.Cys146Phe in a well-established domain, their absence in population data/controls, computational evidence, segregation analysis and relevance to the patients phenotype, led us to classify these allelic variants; p.Arg156Ter and p.Cys146Phe as “pathogenic” and “likely pathogenic” respectively, according to the recommendations of ACMG guidelines [10] for the interpretation of sequence variants (see Table 1).” This information is also summarized and tabulated in Table 1.

2). MOUSE PROTEIN IS ALWAYS IN CAPITAL LETTER.

Sure, the corrections have been done throughout the manuscript.

A supplementary file is uploaded also