Author’s response to reviews

Title: A novel TEX11 mutation induces azoospermia: a case report of infertile brothers and literature review

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Dear Dr. Pasini, Thank you very much for your decision on our manuscript entitled “A novel TEX11 mutation induces azoospermia: a case report of infertile brothers and literature review” (Manuscript ID MGTC-D-16-00052R1). We also thank the reviewers for the constructive and positive comments and suggestions. According to reviewers' comments, we have revised the manuscript accordingly. All amendments are highlighted in red in the revised manuscript. In addition, point-by-point responses to the comments are listed below this letter. The revised manuscript has been edited and proofread by a native English editor from Medjaden Bioscience Limited. We hope that the revision is acceptable for the publication in your journal. If you have any question for the revision, please don’t hesitate to contact us. Look forward to hearing from you soon. With best wishes, First of all, we would like to express our sincere gratitude to the reviewers for their constructive and positive comments. Replies to Reviewer 1: Reviewers' comments: Marta Olszewska, Ph.D., M.Sc. Eng. (Reviewer 1): Dear Authors of the manuscript, you performed some updates - good. However, I have some minor points for you, that should be done before further processing: Response: Thanks for your valuable comments and nice suggestions. We have edited the revision carefully according to your comments and suggestions. We hope you will satisfy with the revision. 1. Paper still needs English correction. However, only some small improvements are needed to give better flow to your text. Please, do that with native speaker (previous correction
was done by some English editor, but it does not looks like native-correction - still some basic language mistakes are present, also commas).

Response: Thanks for your comment. The revised manuscript has been edited and proofread by a native English editor from Medjaden Bioscience Limited.

2. italic text for genes: please, do that for SRY gene too.

Response: We have changed it.

3. WHO and ISCN guidelines still are not cited - please, fix that.

Response: We have fixed it.

4. Some reorganization in the text is needed:- l 45 (in two...) is the same as in l 47-48- l 49 word 'two' change into 'both'; word 'because' is wrong: you did wrong cause-effect sense and it needs native-speaker correction - l 55-57 something is unclear in this sentence. Please, add something more or rewrite it.- l 63-64 'male infertility is characterized by decreased semen parameters leading to azoospermia' - no! decreased semen parameters are not a cause - it is a phenotype of azoo and oligo; causes of azoo are mostly genetic! Please, change this sentence.- l 72 there should be some link between those two sentences, i.e. 'One of teh most interesting genes is TEX11...' or something like that.- l 107-125 this paragraph should me moved into line 66 where genetic causes are described. It should be also shortened (AZF section is no needed to be so detailed - it is not a topic of this paper)- l 126-139 this aragraph should be combined togetehr with data starting in line 68 - that are the same data.- l 140-161 again: similar facts that in paragraph started in line 82 - please, rearrange that to not double facts.- l 162 nonobstructive azoo? Please, include this info also in a purpose of the study. This firts sentence of the study is in fact a result, not a purpose! Please, rewrite this paragraph to proper 'purpose' form.- l 170 add info from lines 220-221; then, information a about the type of biological material is missing - please, add if it was DNA, blood, semen, biopsies or so.- l 176 what do you mean 'chromosome'? Karyotyping? It should be clearly written. And remember about ISCN. Table I - where is it? I could not find it.- l 177 shorter - delete 'were healthy' - it is clear.- l 178-181 it is not necessary, it is known that all was OK. Write only about hormones etc. (181-184 is OK and enough). Again Table I - ??- l 186 change 'infertility' into 'azoospermia' - it better suits here in the whole context- l 187 delete 'two brothers'- l 188 WHO - you missed to cite this position in references! Last sentence should be rewritten, i.e.: 'No sperm was found in each round of analyses.'- l 200-212 should be before l 192-199- l 192 you cannot write that you are determining genetic causes of azoospermia. You can write 'possible mutations causing azoospermia'- 195 coverage of the samples? how many variants in now many genes totally were found? How many after filtering? There should be any criteria of filtering added. What about mother? Was her DNA analyzed too?- l 200 'chromosomal status' menas that you have done something more that karyotyping. If not, then please, delete this, and leave only karyotyping. It was done for who? Brother 1? Brother 2? Mother? Please, add this info.- l 201 checking chromosomes cannot be done on peripheral blood. Please, write this in a correct form!- l 204 you forgot to cite ISCN (it is a book, it has to be in references, similarly as WHO guidelines)- l 206, 210 SRY should be in italic; Company of the FISH probes? Anything more about FISH (1-2 short sentences)?- l 214 'to determine...and azoospermia' you have not done that! I know what do you mean and this is why your text needs corrections of native speaker. Simply, please delete this part of the sentence. Then, 'testicular...approval' should be moved into l 170.- l 215-217 what about temperature, time used in experiments? Or any refernece, if it was done according to it?- l 220-221 move to 170 'and their body...hospital'- 231 what mutation? Here should be data tahta are in l 239-241; please, combine this part of text- l 234 'two' change into 'both' - if you use 'two' it suggests that there were more brothers- l 242-245 start Results from this paragraph. 'Chromosomal status' change into 'karyotype'. What was the frequency of gonadal mosaicism in mother?- l 280 it should be 'mutations' not 'mutants'- 286 where is Table 2? I have not seen it. What is (14) at the end of the line?- l 306 'the precise...mutations' it suggests that you would like to write something