Reviewer’s report

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

Version: 1 Date: 30 Mar 2017

Reviewer: Marta Olszewska

Reviewer's report:

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:

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 look like native-correction - still some basic language mistakes are present, also commas).

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

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

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 interetsing genes is TEX11...' or something like that.
   - l 107-125 this paragraph should move 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 paragraph should be combined together with data starting in line 68 - that are the same data.
   - l 140-125 this paragraph should be 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).

5. 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.

6. l 176 what do you mean 'chromosome'? Karyotyping? It should be clearly written. And remember about ISCN.

7. Table I - 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.

8. 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.'

9. l 200-212 should be before l 192-199

10. l 192-199 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?

11. 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.

12. l 201 checking chromosomes cannot be done
on peripheral blood. Please, write this in a correct form!- 204 you forgot to cite ISCN (it is a book, it has to be in references, similarly as WHO guidelines)- 206, 210 SRY should be in italic; Company of the FISH probes? Anything more about FISH (1-2 short sentences)?- 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 1 170.- 215-217 what about temperature, time used in experiments? Or any reference, if it was done according to it?- 220-221 move to 170 'and their body...hospital'- 231 what mutation? Here should be data that are in 1 239-241; please, combine this part of text- 234 'two' change into 'both' - if you use 'two' it suggests that there were more brothers- 242-245 start Results from this paragraph. 'Chromosomal status' change into 'karyotype'. What was the frequency of gonadal mosaicism in mother?- 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?- 306 'the precise...mutations' it suggests that you would like to write something about mechanisms of mutations' creation- 312-313 move to 199- 314 something more? What investigations? Why authors have not done that?- 315-327 combine with 280-314 - the same, doubled data- 318 lack of reference- 324 examples of intronic alterations and silent mutations are missing- 338 add: 'and families of both donor testes'.5. You declared that you prepared also literature review concerning TEX11. I do not agree - you missed at least 3 papers (PMID: 26086992, 25975716, 24410912).

Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

Yes

Does the work include the necessary controls?
If not, please specify which controls are required in your comments to the authors.

Unable to assess

Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.

Yes

Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?
If an additional statistical review is recommended, please specify what aspects require further assessment in your comments to the editors.

Not relevant to this manuscript

Quality of written English
Please indicate the quality of language in the manuscript:

Needs some language corrections before being published
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