Author’s response to reviews

Title: Expectations and psychological issues before genetic counseling: analysis of distress determinant factors

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Author’s response to reviews:

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Point-by-point reply to reviewer’s comments.

Reviewer’ comments.

Reviewer #1: This is an interesting article that adds to our understanding of the importance of risk perception and support in determining the impact of distress related to genetic testing for a
hereditary cancer predisposition. Whilst the manuscript is well written, there are several places where a spelling or grammatical error should be corrected through careful reading and editing. Some specific comments are below:

1. Abstract:
   -the conclusion does not follow from the information presented in the results section

2. Background:
   -it may be worth noting that HNPCC is also known as Lynch Syndrome
   -check and make tense consistent

Authors’ response:
We thank the reviewer for the comment and we modified the text accordingly for these points.

3. Methods:
   -could you define what you classified as 'mental disease' as an exclusion criterion?

Authors’ response:
We thank the reviewer for the comment. We modified accordingly as your advice.

Page 2, line 3
“They were healthy subjects or cancer patients. Selection criteria were: age of 18 years or older, no previous genetic counseling/test, no diagnosis of major psychiatric disorders, ability to give informed consent.”

-questionnaire: you have listed reference 8 but I cannot see that the questionnaire used in that paper included questions regarding attitude towards and reasons for pursuing genetic testing.

Authors’ response:
We thank the reviewer for the comment and we modify the text since it’s a typesetting mistake.

Page 2, line 10
“Besides socio-demographic data, questionnaire was made of several multiple-choice items regarding attitudes towards and reasons for pursuing genetic test. Questions are described in Table 2”

-do you have a reference for the HADS being validated for use in Italy?
Authors’ response:
We thank the reviewer for the comment and we added a reference to support the use of HADS also in Italy

Page 2, line 17
“The HADS has been validated for use in Italy [21].”


Results:
-you have said that 227 at least partially completed the questionnaire? How many were determined eligible, what was the drop out rate and was there a reason given?
Authors’ response:
We thank the reviewer and we modified taking into account drop out rate.
The survey was proposed to a total of 250 subjects and of these, 227 partially or totally completed the questionnaire before the first genetic counseling interview. There were 23 dropout patients and the reason was the reported absence of interest.

Authors’ response:
We thank the reviewer for the comment and we correct accordingly. The authors are grateful to Marco Ballatore for reviewing the English of the manuscript.

4. Discussion:
-first paragraph, it reads as if you are discussing your own findings but I think you are referring to previous reports in the literature. May be helpful to make this clear. Are there any more recent references that could be included? Many are quite old.
I enjoyed reading this manuscript and thank you for your contribution to increasing our understanding of the psychosocial issues to be aware of when counseling patients for a possible hereditary cancer predisposition.
Authors’ response:
We thank the reviewer for the comment and we have tried to express the concept more clearly. Discussion of our studies, we considered the most relevant and relative data, trying wherever possible to use the most recent ones.

Page 3, line 4
“Furthermore, evidence from systematic reviews illustrates that a genetic counseling intervention does not appear to increase distress and therefore could improve the accuracy of individual’s perceptions of their personal risk [23, 24].”

Page 3, line 9
“However recent data also shows that patients having genetic testing shortly after a diagnosis develop a cancer-related distress due to a significantly different number of psycho-emotional symptoms, which decreased with time. [28, 29]”

Reviewer #2: Interesting paper looking at general anxiety/depression prior to a genetic counseling visit.
1. Needs to be edited much more closely for spelling errors and sentence structure. Some sentence structure problems make it difficult to understand meaning.
Authors’ response:
We thank you for the comment, we took better care of the text and to better review the grammatical and linguistic part. The authors are grateful to Marco Ballatore for reviewing the English of the manuscript.

2. The participant numbers seem to differ between those reported in the participants section and in the results. Please explain more clearly why the N varies in the text.
Authors’ response:
We thank the reviewer and we corrected mistakes.

3. Table 2- It is somewhat unclear to which comparison groups the P values are referring. Check this table for spelling and sentence structure.
Authors’ response:
We thank the reviewer and we modified accordingly.

4. Results- please include more statistical information in text as opposed to just p values
Authors’ response:
We thank the reviewer for the comment and we followed what was suggested.

5. Discussion makes a lot of broad assumptions. P 4 Line 7-16 the HAD does not specifically ask why patients have distress. This section seems to make a huge leap between a higher HAD score and assumptions as to why it may be higher in certain populations. I would recommend removing most of these statements. For example, results show that cancer patients have a higher level of distress than non cancer patients. However results do NOT show that they have a higher level of distress "about" cancer since the HAD didn't specifically ask this. P4 line 22 unsure what results warranted this conclusion. Claridy page 4 line 27-28.
Authors’ response:
We thank the reviewer for the comment and we modified accordingly.

6. In general, please review and revise the discussion for better flow and remove conclusions that do not specifically come from your data.
Authors’ response:
We thank the reviewer for the comment and we made corrections.

Page 4, line 17
“Moreover, our results showed an high perceived risk of hereditary predisposition “per se” both in cancer patients and in healthy individuals referred for a predictive test. Even if most subjects in both subgroups were not able to answer with certainty about the risk of inheritance, the majority of them reported they perceived it to be high. This result is probably due to the lack of information necessary to give a real perception of risk in the pre-counseling phase.”

7. P2 line 29-30- most studies show that patients pursue testing after providers recommend it and not necessarily from their own initiative.

Authors’ response:
We thank the reviewer for the comment. In our clinical experience there also probands who access genetic counseling on their own initiative or on the advice of family members.

Yours sincerely

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