Author’s response to reviews

Title: Pre- and post-testing counseling considerations for the provision of expanded carrier screening: exploration of European geneticists' views

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Author’s response to reviews:

Dear Dr. Partridge,

On behalf of all co-authors, I would like to thank you for your decision letter. In response to the comments and suggestions of the two reviewers, we revised the manuscript and implemented changes accordingly. Several other changes were also made, either to improve the structure/grammar of the paper, or to incorporate recent relevant information (e.g. new publications and authors’ institutional affiliations). All these changes are highlighted and can be easily identified in the text. Below you will find detailed responses to the comments and suggestions of the reviewers.

Kind regards,

Sandra Janssens
The manuscript "Pre- and post-testing counseling considerations for the provision of expanded carrier screening: exploration of European geneticists' views" investigates the attitudes of a group of European geneticists towards ECS, examining their suggestions and recommendations for the use of ECS in the clinical setting.

Suggestions:

- The manuscript is well written and clear, nevertheless I suggest to synthetize more schematically the findings.

Response: To address this comment, we have integrated a table on page 6, which presents the findings in a schematic way.

Method section:

- Describe more in detail the sample of geneticists recruited - Describe more in detail the semi-structured interview used and the method used to code the answers (e.g.: How many cross-validations of each interview have been done?)

Response: The Methods section has been expanded to incorporate the reviewer’s suggestions (pages 4-5). Unfortunately, we cannot provide more detailed information about our study sample, as this may allow some participants to be identified.

Limits and future direction section:

- The authors should include separate considerations for direct-to-consumer tests

- The authors should consider to deepen the issue about pre-test information/education.

(e.g.: the first medical practitioners that could probably inform the patients about the existence of ECS are family physicians or gynaecologists, are they really prepared to inform correctly individuals or couples?)

Response: Limitations of the study have been further elaborated in the paper (page 23) and now include considerations for direct-to-consumer providers of genetic testing and non-geneticist healthcare professionals.
(Reviewer 2): This manuscript reports a study of attitudes of European geneticists regarding the use of expanded carrier screening for reproductive purposes. The work is nicely written and addresses a timely and important set of issues.

The study engaged a relatively small number of geneticists (16), not all of whom are clinicians. This small number and the focus on geneticists is a limitation of the study but the study has value none-the-less as these respondents have relevant expertise and this is an early stage of implementation of the technology.

I have only a few relatively minor suggestions for revisions.

1) Page 3, para 2: Readers may benefit from a somewhat more complete description of the current landscape of expanded carrier screening in Europe. Reference #5 and #6 are in US populations. Data may be anecdotal but it would be interesting to know whether ECS has become routine in Europe on a broad scale or is primarily used in certain clinical centers, etc.

Response: To address this comment, we have briefly described the current state of affairs of ECS in Europe.

2) Page 6: The section on "Acceptability of screening individuals" would benefit from a description of what this means in this context. Specifically, clinicians may pursue cascade screening, that is, offer carrier screening to the pregnant woman and then screen the father if the mother is a carrier for one or more conditions. Does this approach constitute individual screening or screening of the couple? Further, as you note in the Discussion section, individual screening of the pregnant woman makes sense when the condition is X-linked. That issue should be briefly noted in this section.

Response: We thank the reviewer for this comment pertaining to a highly complex issue of screening individuals. We agree that the paper would benefit from further clarifications and have made changes to the section, as well as renamed it to “Issues with screening of individuals.

In the context of screening individuals, there were two closely related scenarios discussed by our participants. The first was performing carrier screening in individuals who request the test in the absence of a reproductive partner. Clearly, the cascade or sequential screening procedure of a pregnant couple, as described by the reviewer, would not apply in this case.

The second issue referred to communicating individual test results where both members of the couple are available and provide samples for carrier screening. In this case, some participants felt that individual carrier status should not be communicated and reporting should be limited to couple-based results. This matter is the focus of the section “Subcategory 2.1 Communication of
individual carrier status”, which has been modified (Pages 14). Even though individual test results are not disclosed in this approach, screening is performed in couples (either simultaneously for all the conditions on the ECS panel, or sequentially, where the sample from the father is screened for those conditions for which the mother is found to be a carrier).

3) Page 17, line 387: The authors recommend the development of educational tools to support decision-making about ECS. Many of the current educational tools in the US are produced by the commercial vendors of the tests. An additional sentence or two here would be welcome about who or what organization should develop educational tools. Should this be the work of professional societies or independent patient advocacy groups or others?

Response: We have added a commented to address this suggestion (lines 458–462)