Reviewer’s report

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

Version: 0 Date: 12 Apr 2017

Reviewer: Jeffrey Botkin

Reviewer's report:

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.

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.

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?
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.

Yes

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

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Please indicate the quality of language in the manuscript:

Acceptable

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