Author's response to reviews

Title: Knowledge and Attitudes to Personal Genomic Testing for Complex Diseases among Nigerians

Authors:

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Author's response to reviews: see over
Reviewer's report
Title: Knowledge and Attitudes to Personal Genomic Testing for Complex Diseases among Nigerians
Version: 2 Date: 27 October 2013
Reviewer: Ruth Chadwick

Reviewer's report:
(1) General comments. This is an interesting paper outlining and discussing the results of empirical research concerning attitudes to genomic testing in 2 districts, rural and urban, of Abuja in Nigeria. The methods are clearly explained and the limitations of the study are acknowledged.
(2) The paper makes a significant contribution to knowledge in this area and raises questions for further work. Appropriate comparison is made with other studies.
(3) There is one major compulsory revision. there seems to be a quotation missing in section C on page 12. inserted
(4) Although the level of writing is high there are some minor typographical errors, e.g., on page 15 line 2, 'familial' should be 'familiar'. On page 7 section B, 'Majority' in first line should say 'The majority...' noted and corrected.
(5) Discretionary revisions. Some of the discussion could be enlarged. For example, there is discussion about the ethics of disclosing test results to children, but there is no acknowledgement in the discussion of the possible consequences for their upbringing of other people knowing a result which they do not. Even if no one raised this, that is interesting in itself. Second, the sections on the results concerning, and discussion of, the effects of religion are quite thin and perhaps could be expanded.

Reviewer's report
Title: Knowledge and Attitudes to Personal Genomic Testing for Complex Diseases among Nigerians
Version: 2 Date: 10 September 2013
Reviewer: Lei-Shih Chen

Reviewer's report:
Major Compulsory Revisions
This manuscript addresses important questions about the Nigerians’ knowledge and attitudes towards personal genomic testing. However, it seems that several concerns detracted from the manuscript as a whole. I have listed those concerns as follows:
1. Overall, it seems that the authors have covered too many topics in this manuscript (especially in the Results section), prohibiting an in-depth discussion of any one area.
2. Introduction section: It seems that the introduction is too “thin”, and therefore, does not provide sufficient information to the readers to understand the background of the study Expanded. For example, the authors might want to provide more information about the H3Africa Project H3 Africa i.e.Human Heredity and Health in Africa (H3Africa) Initiative aims to facilitate a contemporary research approach to the study of genomics and environmental determinants of common diseases with the goal of improving the health of African populations.I can not immediately figure how to bring it into this work. (it seems to be a very interesting project)
and genomic testing issues in Nigeria (Nigeria does not represent the whole of Africa). Sure and the research title clearly showed that the work was in Nigeria and the Data were purely Nigeria data. The work was not broad enough to be extrapolated as the whole of Africa data. Mentions were made of Africa because many of the issues that affect Nigeria affect Africa. Nigeria being the most populous African country. Another example is that the authors mention that education, poverty, culture, religion, and health behavioral practices differ between Africa and the rest of the world. The authors might want to provide some data and also explain education, poverty, culture, religion, and health behavioral practices in “Nigeria.” The background is expanded with more focus on the cultural variance of Africans and how this may affect Africans view of genomic tests.

3. Some of the research questions and terms (e.g., affect) used in this manuscript are more appropriate for quantitative than qualitative data (e.g., "Culture and religion were found to affect perspectives of respondents on genomics test… [Abstract]; and “Most respondents in the FGDs claimed that their perception of genomic testing was affected by religion or culture… [page 14]"). This could be interpreted as showing a statistically significant difference. The word affect was changed to a synonym.

4. Why were some data collected by focus group discussion and some by key informant interviews? The KII was to elicit more in-depth responses after the FGDs, using follow-up questions, prompts and probes.

5. It is unclear how the authors assessed participants’ knowledge about the use of genomic tests. In general, it is better to assess “knowledge” by giving participants a number of multiple choices or true/false test questions. We used open ended questions and not True/False nor multiple choice questions.

6. Discussion section: It is unclear why the authors compared their research findings with the findings of Chen et al. (2007). The purpose of Chen’s article is to conduct a systematic review to assess the factors affecting individuals’ decision to accept or decline cystic fibrosis carrier testing/screening. Given the research questions, samples, methods, and variables between the two studies are different, I would suggest the authors to find other references which are more relevant to their study. This reference has been replaced.

Minor Essential Revisions

1. There are a number of places with formatting/spacing/typo errors and some of the sentences are too long (e.g., the third paragraph in the Background section has only one sentence – which is a long [five lines] sentence). To improve the clarity of this manuscript, I would strongly encourage the authors to carefully go through the whole manuscript for editing. The entire manuscript has been carefully reviewed.

2. There are a number of places where there are formatting errors and typos in the citations and references. For example, the citation style of references 1, 2, 3, 11, 12, 13, 14, 16 are different from the other references. Also, there are some errors in the reference list. For example, the issue of reference 5 (ref 5 is an encyclopedia obtained online with no issue but the year) is incorrect, and, the issue and page numbers of reference 6 are incorrect. Ref 6 has been replaced.

3. On page 18, it seems that the authors quote one sentence (“parents viewed the benefits of pediatric testing….”) from reference 12, but they have not stated
Reviewer's report
Title: Knowledge and Attitudes to Personal Genomic Testing for Complex Diseases among Nigerians
Version: 2
Date: 29 October 2013
Reviewer: Jantina De Vries

Reviewer's report:
This is a paper that discusses perspectives of Nigerian people living in an urban and peri-urban area on ‘genomic tests for complex disease’. The novelty of the paper lies in its focus on an African country – no published work yet considers the views of African people on DTC genomic tests. However, I have some concerns about the paper, which I outline below:

1. The paper concludes, in the discussion, that “It is notable that the participants showed positive attitudes towards genomic tests and potential benefits despite knowing almost nothing about the test”. As a social scientist, this signals to me a fundamental problem in the research design, namely: how can one seek to solicit people’s perspectives on an issue that they know nothing about? When participants give you their opinions or views, are they really pertinent to the topic under discussion? And how do you verify that? In other words, I would be concerned about the validity of the data thus collected, and I think the authors need to say much more in their paper about how they have dealt with this challenge. This needs to come out in the methods section, in the section on ‘limitations’, and it also needs to be expanded in the sections “Knowledge of Genomic Tests”. Exactly what did participants understand, how many people understood anything, what did they think these tests were all about, and how does all of that inform the validity of the data? (reading the quotes used in the paper, I have a strong suspicion that many of the informants confused ‘DTC genomic tests’ with other medical tests – e.g. quotes stating that participants can ‘test themselves’ could be taken to resonate with pregnancy tests that can be done at home, and comments about confidentiality could draw experiences w.r.t. HIV test results and other medical tests. How did the researchers rule out this possibility?). The statement has been rephrased and the paragraph more explicit. This has also reflected in the limitation section. Most respondents’ knowledge of genomic tests was limited to paternity and sickle cell genotype tests during initial assessment but the concept was clearer to participants after explanation what genomic test is. The education was done before assessing their feelings on the test.

2. The paper introduces some very controversial potential uses of DTC tests (for instance, use in children and as a prenatal screen) which are not engaged with or discussed. The use of genetic diagnostic tests in children is hugely controversial, for instance, and a lively literature has discussed various ethical considerations that may go into deciding whether to offer/use genetic tests for children. The paper does not engage with this literature yet it aims to use its data to inform that debate. Similarly, to my knowledge, DTC personal genomic tests are not used anywhere in the world for prenatal diagnosis and such use may even be illegal in many countries. Again, the authors should give much more credit to the myriad of ethical considerations that such use raises (and also satisfy point 1 above). This work did not evaluate the use of DTC in prenatal screening. A mention was made of DTC under the regulation of genomic test. Here participants views were obtained on
restricting genomic testing to health facilities or allowing consumers to have direct access to it without the need of a third party to help do the test.

3. The paper needs to critically revise language used, and also check for completeness (for instance, Section C on pg. 12 reads ‘A respondent said’ and is not followed by anything). Corrected.

5. On pg. 15 Section “Willingness to do genomic tests’ – I wonder how the Native American example is relevant to the Nigerian example? The Native American population are, in some ways, an exceptional case (in terms of historical marginalisation in the country and more recent recognition of their rights). Does the same apply to all the Nigerian informants and their population groups? The reference is expunged.

Required revisions:
1. expand the methods section, the section ‘knowledge of genomic tests’ and the ‘limitation’ section to reflect methodological challenges in the study; Expanded
2. tie the debates around the use of DTC in children and foetuses to relevant ethics literature and signal that the prenatal use of DTC genomic tests may in fact be illegal in many countries. This work did not evaluate DT in prenatal screening.