Reviewer’s report

Title: Opportunities and challenges of integrating genetics education about human diversity into public health nurses’ responsibilities in Japan

Version: 1 Date: 10 Sep 2019

Reviewer: Angela Trepanier

Reviewer's report:

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I appreciate the opportunity to review this interesting manuscript. Public health nurses have the potential to facilitate the integration of evidence-based genomics applications into medical practice. As such, this manuscript investigates an important topic. However, some revision of the literature review (introduction), methods section, and discussion is needed as described below. In addition, further improvements in the English language translation of this manuscript are needed. There are numerous places throughout the manuscript where word choices/phrasing negatively impact the clarity of the reporting.

Abstract: In the background section, the term "awareness programs" is confusing. Do you mean that you are investigating opportunities and challenges of "integrating genetics education into their professional duties?" Or do you mean integrating genetics services into their professional duties? In the results section, the sentence, "Through the experience of duties, public health nurses needed specialized education, post-graduation studies, and mentoring about genetics," is also confusing. What do you mean by "Through the experience of duties? In the conclusion of the abstract, the first sentence is very clearly stated but the second seems to reach beyond the scope of the project. I do not think this study demonstrates that incorporating genetics "would solve health-related issues"- that is an overstatement. For the last part of the sentence, I think you mean, that it "would also provide an opportunity for clients to be informed about their latent genetic risks."
Background Section:

The Human Genome Project reference is really dated (2010). There are now more than 60,000 genetic tests for more than 18,000 different genes (https://www.ncbi.nlm.nih.gov/gtr/) or see "Genetic testing availability and spending. Where are we now? Where are we going? in Health Aff (Millwood). 2018 May; 37(5): 710-716, doi: 10.1377/hlthaff.2017.1427.

The sentence, "The increasing number of genetic tests has simultaneously allowed companies to have direct access to consumers (DTC), providing affordable and easy to use genetic tests by healthcare providers," is problematic. There is direct to consumer genetic testing through companies like 23andMe but some of their testing, such as tests for most multifactorial diseases, has not been clinically validated. See the Centers for Disease Control and Prevention Tier Table Database at https://phgkb.cdc.gov/PHGKB/topicFinder.action;jsessionid=926FE32EDC1A3AA7C3834C8AC3DD1148?Mysubmit=init&amp;query=tier+3 for a good review of what genetic tests have evidence to support implementation into public health programs and those that do not yet have sufficient evidence to recommend implementation. Your introduction could be strengthened by focusing on evidence-based genetic tests (e.g. CDC Tier 1) and moving away from discussions about DTC to focus on clinical testing.

On page 5, line 101, the meaning of the statement ‘...the understanding of human diversity could resolve the ethical issues associated with genetic and medical technologies" is unclear. Do you mean human genetic diversity?

Starting with line 110 on page 5, the sentence that starts with "In collaboration... is very long. Instead of semi-colons it would be clearer to break this long sentence into separate sentences. The part of the sentence that starts with "however,they do not have the proper knowledge on genetics to overcome the community demand through their duties (line 113, page 5) is confusing. Do you mean that nurses do not have sufficient knowledge to address the need for genetics services or that they do not have sufficient time or both?"
Similarly, the sentence (page 6, line 120) "In contrary (contrast), external to their professional duties...are facing challenges during awareness programs of health promotion especially (related to) genetic diseases. Does this mean that public health departments sponsor awareness programs on genetics topics? Are these programs meant to improve the awareness of the public or of the public health providers? Since your aims are investigating awareness programs I think a little more description of what they are would be beneficial and help provide more context to your study.

Methods:

Please reference the phenomenological research design.

Please justify your decision to conduct a single focus group with 6 participants. How did you determine this was a sufficient number of groups/group members?

Since you talk about maternal health and mental health public health nurses in your discussion, it would be helpful to know all the different areas your participants worked in.

Your interview guide does not ask any questions about awareness programs despite that being a focus according to your study aims. As mentioned above, please clarify what you mean by awareness programs.

Please reference your data analysis methods (qualitative content analysis).

Under the rigor section, please be more specific about how the data set was coded. Who developed the code book. How many investigators reviewed each coded sentence? Were all sentences coded by at least two authors? If not, what was the interrater reliability for coding?

Results: The results section is well-written and provides a thorough description of the results with clear categories and subcategories that align well with the provided quotes.
Discussion: Overall, the discussion thoroughly reviews the potential implications of your results. Attention to the English language translation would enhance clarity, though. Below are some additional recommendations.

It would be helpful to start the discussion by stating your study aims and a summary statement about the findings before you give specific examples.

On page 17, line 331, what do you mean when you say the role of public health nurses in promoting human diversity is very important? What do you mean by diversity? In gender, ethnicity, physical ability? How do nurses typically promote diversity?

Line 335 on page 17, as noted earlier in this review, please consider rephrasing the sentence about solving health-related issues and providing an opportunity for clients to notice latent genetic issues to improve clarity. This sentence appears a third time in the conclusion (so please consider modifying it there as well).

On page 17 starting with line 338, "For instance, the knowledge should be studied as part of the mandatory education system" - do you mean nursing education? What do you mean by "acquired as a specialized level?" When you say "Human diversity should be studies at a young age," as stated before, what do you mean by human diversity- in genetic makeup? other factors?

On page 18, when you say "In addition, lower level of genetic knowledge is related to age and education level" how is it related? Do younger people know more about genetics than older people or vice versa? Is higher education level associated with more genetics knowledge or is the opposite true?

The sentence that starts with "The reasons..." is awkwardly worded and hard to understand.

Do you have a reference to support your statement about "When children learn accurate information..." page 18, line 348?
In the conclusion you mention the development of organizations (this statement is also present in other parts of the paper). What types of organizations are you referring to? Professional organization for public health genetics? for public health nurses?

Thank you again for the opportunity to review this interesting study on a topic of relevance to the evolving practice of medical genetics.

**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

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

Not suitable for publication unless extensively edited
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