Author’s response to reviews

Title: Reinterpretation, reclassification, and its downstream effects: Challenges for clinical laboratory geneticists

Authors:

Julia el Mecky (j.el-mecky@soton.ac.uk)
Lennart Johansson (l.johansson@umcg.nl)
Mirjam Plantinga (m.plantinga@umcg.nl)
Angela Fenwick (a.j.fenwick@soton.ac.uk)
Anneke Lucassen (a.m.lucassen@soton.ac.uk)
Trijnie Dijkhuizen (t.dijkhuizen@umcg.nl)
Annemieke van der Hout (a.h.van.der.hout@umcg.nl)
Kate Lyle (k.lyle@soton.ac.uk)
Irene van Langen (i.m.van.langen@umcg.nl)

Version: 2 Date: 18 Oct 2019

Author’s response to reviews:

Department of Genetics
University of Groningen, University Medical Center Groningen,
The Netherlands

Clinical Ethics and Law Southampton
University of Southampton,
United Kingdom

18 October 2019

Dear Dr Tawfiq,

Thank you for providing us with editorial review on how to format our manuscript, as well as statements that need to be further included.
Please see below our response to your requested revisions.

Kind regards,

Julia El Mecky and Lennart Johansson (joint first authors of the manuscript)

On behalf of all authors,

Mirjam Plantinga
Angela Fenwick
Anneke Lucassen
Trijnie Dijkhuizen
Annemieke van der Hout
Kate Lyle
Irene van Langen

1. Corresponding author email

We note that the corresponding authors emails on the title page of your manuscript (j.elmecky@soton.ac.uk and l.johansson@umcg.nl) are different to the one entered into the submission system (j.el-mecky@soton.ac.uk). Please amend one of these entries and ensure that the corresponding authors email entered into the submission system and into the title page of your manuscript are the same. Please only list one email on the title page of your manuscript.

The entry has been amended to correspond to the one entered into the submission system (j.el-mecky@soton.ac.uk)

2. Declarations

Please move the Declarations section to after the Conclusions section.

The Declarations section has been moved to after the Conclusions section.

3. Results and Discussion

As per our submission guidelines, we require that the Results and Discussion be separated under the headings ‘Results’ and ‘Discussion’, respectively.
For more information, please see https://bmcmedgenomics.biomedcentral.com/submission-guidelines/preparing-your-manuscript/research-article

In an effort to be of most use to broader academic, policy, guideline, and practice discussions regarding reinterpretation and reclassification, we combined the results and discussion sections. This allowed for direct reflection on the findings in the context of current literature available on reinterpretation and recategorization.
We felt the most relevant way to present the data that ensued from our focus group discussion was by identifying and reflecting on challenging questions in clinical practice. In other words, we felt that describing the data in one section and subsequently reflecting on it in a separate section would undercut the usefulness, relevance and clarity of the manuscript. In making this decision, we were inspired by publications, such as Vears et al. (2018) “Points to consider for laboratories reporting results from diagnostic genomic sequencing,” a research paper that utilizes a combined results and discussion format to similarly pinpoint broad challenges, supported by their research data, that merit further focus and discussion in the field of genomics.

We assumed that the format of our manuscript was acceptable to the editorial office as it was sent to peer reviewers and subsequently considered potentially acceptable for publication in BMC. If we would now restructure the manuscript by including separate results and discussion sections, it would result in an entirely different manuscript that would need to be peer reviewed anew.

For the above reasons, we ask the editorial office to consider publishing our manuscript with a combined results and discussion section or to advise other possible solutions, such as publishing the manuscript as a debate, which does not require separate results and discussion sections, rather than a research paper.

4. Limitations

Please note that the required format of a research article is: Background, Methods, Results, Discussion, Conclusions. As such, we ask that you incorporate and combine the section “Limitations” with one of the aforementioned sections.

The Limitations section has been included at the end of the Conclusion (line 438).

5. Remove attachments

Please remove the additional file entitled ‘Tracked Changes on Revised Manuscript_El Mecky_Johansson.docx’ and ‘Re-submission letter_el Mecky_Johansson.docx’ from your manuscript and from the file inventory they are no longer needed at this stage.

This attachment has now been removed.

6. Role of funding body

Please provide more details regarding the specific role of the funding body in your study. If the funding body only provided the financial means to allow the authors to carry out the study, please state “The funding body played no role in the design of the study and collection, analysis, and interpretation of data and in writing the manuscript” and include this statement in the Funding section of your manuscript.

However, if the funding body took part in the design of the study and collection, analysis, and interpretation of data, and the writing of the manuscript, this must be stated in the Competing Interests section of your manuscript.
The statement “The funding body played no role in the design of the study and collection, analysis, and interpretation of data and in writing the manuscript” has been added to the Funding section of the manuscript.

7. Form of consent to participate

While you have stated that “Participants consented to taking part in the research” this is insufficient. In the section 'Ethics Approval and Consent to Participate', please state whether the informed consent obtained was written or verbal. If consent was verbal, please state the reason and whether the ethics committee approved this procedure. If the need for consent was waived by an IRB or is deemed unnecessary according to national regulations, please clearly state this, including the name of the IRB or a reference to the relevant legislation.

Electronic consent was obtained from participants on the online platform on which the focus group was held. Prior to the research, participants were informed what participating in the research would entail and how its data would be used by means of an information sheet. Subsequently, on the online platform that hosted our focus group discussion, participants were asked to consent to participating in the research by proceeding to the discussion. This explanation on how consent was obtained has been added to the section Ethics approval and consent to participate.

8. Consent for publication

Currently, the statement in your “Consent for publication” section of your declarations is incorrect. Consent for publication refers to consent for the publication of identifying images or other personal or clinical details of participants that compromise anonymity. Seeing as this is not applicable to your manuscript please state “Not Applicable” in this section.

We have replaced our previous statement with “not applicable” in this section.

9. Abbreviations

Please provide a list of all the abbreviations used in the manuscript. This list should be placed just before the Declarations section. All abbreviations should still be defined in the text at first use.

We have now included a list of all abbreviations before the Declarations section.

10. At this stage, please upload your manuscript as a single, final, clean version that does not contain any tracked changes, comments, highlights, strikethroughs or text in different colours. All relevant tables/figures/additional files should also be clean versions. Should you wish to respond to these revision requests, please put your responses to the reviewers'/editors’ comments in the Response to Reviewers box in Editorial Manager. Please do not upload a separate letter.
We have now uploaded our manuscript as a single, final, clean version and removed other files containing tracked changes and highlights.