Author’s response to reviews

Title: Molecular diagnosis in recessive pediatric neurogenetic disease can help reduce disease recurrence in families

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Response to Editorial Comments
Molecular diagnosis in recessive pediatric neurogenetic disease can help reduce disease recurrence in families

Editor Comments

1. Ethics approval
Please note that manuscripts reporting studies involving human participants, human data or human tissue must include a statement on ethics approval and consent (even where the need for approval was waived).
Please also note that ethics approval should:
• Be Independent from the investigators or sponsors
• Be Local - From the same institute, region or country in which the study took place
• Cover all countries – for studies performed in multiple countries, appropriate ethical committee approval must be included for each country.

We require a clear, unambiguous statement of ethics committee approval:
• Specific project approval, not simply general guideline adherence
• The full name and affiliation of each ethics committee
• Not simply “the local IRB approved the study”
• Reference numbers if applicable

Please confirm whether your study was submitted to and approved by a local ethics committee in Egypt and include a statement to this effect in the “Ethics approval and consent to participate” section of your Declarations. Please also ensure that the full name of the ethics committee is included in this statement.

If the need for ethics approval was waived by an Institutional Review Board (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.

Response.
We have included the required language in the Ethical assessment section on page 10 to address the journal requirements as instructed. We confirm that the study was submitted to and approved by the local ethics committee in Egypt.

2. Consent to Participate
In your “Ethical approval and consent to participate” section, please clarify whether informed consent to participate, written or verbal, was obtained from all of the participants in the study and clearly state this in your manuscript.
Please note that in the case of minors, which refers to individuals younger than the age of 16, consent to participate must be obtained from their parents or legal guardians. As such, we ask that in your “Ethics approval and consent to participate” section, to clarify whether informed consent to participate, written or verbal, was obtained from the parents or legal guardians of any participant under the age of 16 and clearly state this in your manuscript.
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.

Response.
We have included required information in the Ethical assessment section on page 10. We now state “All subjects or their parents or legal guardians signed a consent in their fluent language, and additionally an assent was signed by mentally capable underaged individuals in the form of a childhood assent if between the ages of 7-12, and an adolescent assent if between the ages of 13-18 years of age”.

3. Consent for publication
Please note that consent for publication refers to consent for the publication of identifying images or other personal or clinical details of participants that compromise anonymity. As such, we ask that in your “Consent for publication” section, to please clarify whether written informed consent for publication of clinical details and/or clinical images was obtained from all of the participants.
Please note that in the case of minors, which refers to individuals younger than the age of 18, consent for publication must be obtained from their parents or legal guardians. As such, we ask that in your “Consent for publication” section, to please clarify whether written informed consent for publication of clinical details and/or clinical images was obtained from the parents or legal guardians of any participant under the age of 18.

Response.
We have included the following text in the Consent for Publication section on p.22. “Consent for publication of identifying images or other personal or clinical details of participants that compromise anonymity was documented in the consent form. Consent to publish images of a minor was obtained from their parents or legal guardians.”

4. Availability of Data and Materials
Please ensure that you include the accession numbers or direct links and the full names of the data banks/repositories corresponding to any datasets obtained from web-based sources and subsequently analysed in your study in the Availability of Data and Materials section, including any reference datasets. If accession numbers or web links are listed in a table or additional file, please include an in-text reference to the file or table in the Availability of Data and Materials section.

Response. In the Availability of data and materials section on p.21, we have now included a weblink to the dbGaP submission as requested and verified that the link is active.

5. Authors’ contributions
a) We have noticed a discrepancy in the Authors’ contributions section of your manuscript. In regard to the individual authors’ contributions, you have not stated the individual contributions of Hasnaa M. Elbendary and Mohamed S. Abdel-Hamid. Please include the individual contributions of Hasnaa M. Elbendary and Mohamed S. Abdel-Hamid in the Authors’ contributions section of your manuscript.

Please also represent the author's names using their full initials, not their name, and without any titles. Guidance and criteria for authorship can be found here: http://www.biomedcentral.com/submissions/editorial-policies#authorship

Response. Thank you for noticing the discrepancy in the Author’s contributions section, which we now corrected, by clarifying the contributions of Hasnaa M. Elbendary and Mohamed S. Abdel-Hamid.

b) Please consider the list of authors as it currently stands with reference to our guidelines regarding qualification for authorship (http://www.biomedcentral.com/submissions/editorial-policies#authorship).
Currently, the contributions of author VS does not automatically qualify them for authorship. In the section “Authors’ contributions”, please provide further clarifications on their contributions, and see our guidelines for authorship below.

An 'author' is generally considered to be someone who has made substantive intellectual contributions to a published study. Authors are expected to fulfil the criteria below (adapted from McNutt et al., Proceedings of the National Academy of Sciences, Feb 2018, 201715374; DOI: 10.1073/pnas.1715374115; licensed under CC BY 4.0):

Each author is expected to have made substantial contributions to the conception OR design of the work; OR the acquisition, analysis, OR interpretation of data; OR the creation of new software used in the work; OR have drafted the work or substantively revised it

AND to have approved the submitted version (and any substantially modified version that involves the author's contribution to the study);

AND to have agreed both to be personally accountable for the author's own contributions and to ensure that questions related to the accuracy or integrity of any part of the work, even ones in which the author was not personally involved, are appropriately investigated, resolved, and the resolution documented in the literature.

Please note that acquisition of funding, collection of data or general supervision of the research group, alone, does not usually justify authorship. You may acknowledge individuals who assist in the acquisition of funding or of data or supervise the research group in the Acknowledgments section of your manuscript.

If you wish to amend the list of authors, please use the standardised form which you and your co-authors must complete. The authorship change form can be found at the following link: https://resource-cms.springernature.com/springer-cms/rest/v1/content/7454878/data/v5

All instructions can be found on the form, please treat the 'current authorship' section as the original authorship. Please return the form within 14 days by email to the Editorial Office with all author signatures (including those newly added/removed).

Response. We have reviewed criteria for authorship in the McNutt publication, and confirm that all listed authors fulfill criteria for authorship on this study. The study required the expertise of multiple researchers and clinicians, and thus the author list reflects this collaboration.

6. Section S2
We ask that in regard to ‘Section S2’, that you please include all abbreviations in a single section entitled ‘Abbreviations’ in the main manuscript, including this section after the Conclusions section in your manuscript.

Response. We have now moved the abbreviations from Section S2 into the main text following Conclusions.
7. Data S1
As per our submission guidelines, we require that all figures, tables and additional files be cited in the text in sequence in a manuscript. Data S1 is currently not cited in your manuscript. Please ensure that it is appropriately cited within the main body of text in your manuscript.

Response. Thank you for identifying this error. We have now cited Data S1 on p.13

8. 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.

Response. Done