Author’s response to reviews

Title: Clinical Utility of Exome Sequencing in Individuals with Large Homozygous Regions Detected by Chromosomal Microarray Analysis

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September 27, 2017

Matteo Pasini, Ph.D.
Editor
BMC Medical Genetics

Dear Dr. Pasini,
Thanks for your review of our manuscript entitled “Clinical Utility of Exome Sequencing in Individuals with Large Homozygous Regions Detected by Chromosomal Microarray Analysis” (Manuscript ID: MGTC-D-17-00244).

We appreciate your comments on our manuscript. Please find below our point-by-point response:

1: Please clarify whether the consent to participate was written or verbal, and if verbal consent was given, was this approved by the ethics committee.

Please include this information in the Ethics approval and consent to participate section.

Author’s response: We obtained a waiver of consent from Western Institutional Review Board (IRB) Protocol No. 20162032, for this retrospective study. All patient data submitted to us for the purposes of clinical testing are covered under this IRB. Data was analysed in a coded fashion in compliance with our IRB protocol. Following editor’s suggestion, we have now added this information in the ‘Ethics approval and consent to participate’ section.

2: We have noted that you have stated Not applicable in the Consent to publish section, however, there are many indirect identifiers present in Additional File 1. Please clarify in the Consent to publish section whether the patients or their legal guardians consented to the publication of their medical data. We would also request that in order to anonymise the data that the ages be provided as ranges and the gender information is removed.

Author’s response: As mentioned above, we obtained a waiver of consent from Western Institutional Review Board (IRB) Protocol No. 20162032, for this retrospective study. This information has now been provided in the ‘Consent for publication’ section. According to
editor’s comments, we are happy to remove gender information and present ages of patients in ranges to further protect anonymity. An updated Additional File 1 is submitted for review.

3: Please clarify the role of the funding body with regards to this study. Please include this information in the Funding section.

Author’s response: This study was funded by Lineagen, Inc. Interpretation of the data and the conclusions of the study were derived from the work of the research and scientific team at Lineagen. These interpretations and conclusions were not influenced by the commercial interests of the company, nor did company management have any input into this work. We have now provided this detail in the ‘Funding’ section of the manuscript.

4: At this stage, we ask that you submit a clean version of your manuscript and do not include track changes or highlighting.

Author’s response: A revised manuscript is submitted after incorporating suggestions from the editor. No track changes or highlighting is done.

We sincerely hope you will find the revised version of the manuscript suitable for publication. Please contact us if you have further questions and concerns about our manuscript.

Thank you for your consideration and best regards,

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