Author’s response to reviews

Title: Genomic analysis of a spinal muscular atrophy (SMA) discordant family identifies a novel mutation in TLL2, an activator of growth differentiation factor 8 (myostatin): a case report

Authors:
Jianping Jiang (jianping.jiang@sjtu.edu.cn)
Jinwei Huang (jinwei1314w@aliyun.com)
Jianlei Gu (jianleigu@sjtu.edu.cn)
Xiaoshu Cai (cxs_123@163.com)
Hongyu Zhao (hongyu.zhao@yale.edu)
Hui Lu (huilu@sjtu.edu.cn)

Version: 4 Date: 30 Nov 2019

Author’s response to reviews:

MGTC-D-19-00496R3
Genomic analysis of a spinal muscular atrophy (SMA) discordant family identifies a novel mutation in TLL2, an activator of growth differentiation factor 8 (myostatin): a case report
Jianping Jiang; Jinwei Huang; Jianlei Gu; Xiaoshu Cai; Hongyu Zhao; Hui Lu
BMC Medical Genetics

Dear Dr. Lu,

Your manuscript "Genomic analysis of a spinal muscular atrophy (SMA) discordant family identifies a novel mutation in TLL2, an activator of growth differentiation factor 8 (myostatin): a case report" (MGTC-D-19-00496R3) has been assessed. Many thanks for your response to our revision requests. However, there are a few final editorial requests we would like you to address before we accept the manuscript, detailed below.

----

1. MANUSCRIPT format

We note that your manuscript is still organised into the following sections: Background, Case presentation, Methods, Results, Discussion and conclusion. As per our submission guidelines, we require all case reports to be organised into the following sections: Background, Case Presentation, Discussion and Conclusions. As such, please reformat your manuscript according to the sections:

- Background - the Background section should explain the background to the case report or study, its aims, a summary of the existing literature.
- Case presentation - this section should include a description of the patient’s relevant demographic
details, medical history, symptoms and signs, treatment or intervention, outcomes and any other significant details.
• Discussion and Conclusions - this should discuss the relevant existing literature and should state clearly the main conclusions, including an explanation of their relevance or importance to the field.

Response:
Reformatted.

2. ABSTRACT format

Please reformat your Abstract according to the sections:

• Background: why the case should be reported and its novelty
• Case presentation: a brief description of the patient’s clinical and demographic details, the diagnosis, any interventions and the outcomes
• Conclusions: a brief summary of the clinical impact or potential implications of the case report

Please also ensure that the Abstract entered into your manuscript is the same as that entered into the submission system.
Response:
Reformatted.

3. Table S2

As per our submission guidelines, we require that all additional files be cited in the MAIN text in a manuscript. Table S2 is currently not cited in your manuscript. Please ensure that it is appropriately cited within the main body of text in your manuscript. Any items which are not cited may be deleted by our production department upon publication.
Response:
Cited in the main text.

4. Availability of data and materials

Please note that BMC mandates that data types, listed below, be deposited and made publicly accessible. We are an open access journal and submission of a manuscript to a BMC journal implies that materials described in the manuscript, including all relevant raw data, will be freely available to any scientist wishing to use them for non-commercial purposes.

Seeing as this applies to your manuscript, please note that we cannot publish your manuscript if your data is not deposited. As such, we ask that you provide the accession numbers or web links to your datasets which allow for public access and include these in the Availability of data and materials section of your manuscript. Please ensure that all data is already released and that any accession numbers/web links are in the ‘Availability of Data and Materials’ in their final form, and that all links and accessions allow access to public data.
Response:
We have put all the detected variants in Tables S3. However, the raw sequence datasets generated during the current study are not publicly available because it is possible that individual privacy could be compromised. However, datasets are available from the corresponding author on reasonable request.
5. Consent to participate

In the section 'Ethics Approval and Consent to Participate', please clarify whether the parents/legal guardians of any minors provided written consent to participate.

Response:
Revised.

6. Consent for publication

Please note that in the case of minors, which refers to individuals younger than the age of 18, consent to publish must be obtained from their parents or legal guardians. As such, we ask that in your “Consent for publication” section, to clarify whether written consent to publish, was obtained from the parents/legal guardians of the minors (male patient and female patient) and clearly state this in your manuscript.

Response:
Revised.

7. 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.

----

Once you have made the necessary corrections, please submit a revised manuscript online at:

https://www.editorialmanager.com/mgtc/

If you have forgotten your password, please use the 'Send Login Details' link on the login page at https://www.editorialmanager.com/mgtc/. For security reasons, your password will be reset.

We request that a point-by-point response letter accompanies your revised manuscript. This letter must provide a detailed response to each reviewer/editorial point raised, describing what amendments have been made to the manuscript text and where these can be found (e.g. Methods section, line 12, page 5). If you disagree with any comments raised, please provide a detailed rebuttal to help explain and justify your decision.

Please also ensure that your revised manuscript conforms to the journal style, which can be found at the Submission Guidelines on the journal homepage.

A decision will be made once we have received your revised manuscript, which we expect by 01 Dec 2019.

Please note that you will not be able to add, remove, or change the order of authors once the editor has accepted your manuscript for publication. Any proposed changes to the authorship must be requested during peer-review, and adhere to our criteria for authorship as outlined in BioMed Central's policies.
To request a change in authorship, please download the 'Request for change in authorship form' which can be found here - http://www.biomedcentral.com/about/editorialpolicies#authorship. Please note that incomplete forms will be rejected. Your request will be taken into consideration by the editor, and you will be advised whether any changes will be permitted. Please be aware that we may investigate, or ask your institute to investigate, any unauthorized attempts to change authorship or discrepancies in authorship between the submitted and revised versions of your manuscript.

We look forward to receiving your revised manuscript and please do not hesitate to contact us if you have any questions.

Best wishes,

Arwa Tawfiq
BMC Medical Genetics
https://bmcmedgenet.biomedcentral.com/

Recipients of this email are registered users within the Editorial Manager database for this journal. We will keep your information on file to use in the process of submitting, evaluating and publishing a manuscript. For more information on how we use your personal details please see our privacy policy at https://www.springernature.com/production-privacy-policy. If you no longer wish to receive messages from this journal or you have questions regarding database management, please contact the Publication Office at the link below.

In compliance with data protection regulations, you may request that we remove your personal registration details at any time. (Use the following URL: https://www.editorialmanager.com/mgtc/login.asp?a=r). Please contact the publication office if you have any questions.