Author's response to reviews

Title: The effect of homozygous deletion of the BBOX1 and Fibin genes on carnitine level and acyl carnitine profile

Authors:

Ali Rashidi-Nezhad (ali_rashidinejad@yahoo.com)
Saeed Talebi (talebisaaid@yahoo.com)
Homeira Saebnouri (saebnoori@yahoo.com)
Seyed Mohammad Akrami (akramism@sina.tums.ac.ir)
Alexandre Reymond (Alexandre.Reymond@unil.ch)

Version: 4 Date: 25 June 2014

Author's response to reviews: see over
Manuscript submission:

Dear Editor,

Please find enclosed a revised version of our manuscript MS: 1013817221110522 entitled "the effect of homozygous deletion of the BBOX1 and Fibin genes on carnitine level and acyl carnitine profile" for consideration for publication in BMC Medical Genetics.

We have addressed all the reviewers’ comments and editorial requirements as specified in the answers-to-reviewers file. In particular we included the name of the ethics committee that approved the study as required.

We hope that you will find this revised version of our study interesting and suitable for publication.

Many thanks for your expert editorial assistance.

We are looking forward to hearing from you at your earliest convenience.

Sincerely,

Alexandre Reymond
Referee 1:

Reviewer: Ayman W El-Hattab

Reviewer's report:

The authors have responded appropriately to the previous comments and suggestions. I have some minor comments:

1. Please make sure to use the italic font for the gene names
   Answer: We have modified the text to comply with common usage of italic and normal font when referring to genes and proteins, respectively.
2. Please add some details about the FIBIN and its relation with the retinoic acid and wnt signaling.
   Answer: We modified the text to mention the relationship between fibin and the RA and wnt signaling.
3. The first paragraph of the ?Background?. The sentence ?A process that results??derivatives? is out of context and can be removed.
   Answer: We have modified the text as suggested.
4. Please add normal reference ranges for all the values in tables 2 and 3.
   Answer: We are puzzled by this comment as the same reviewer stated during the first round of review (his point 3) that: “there is no need to present normal values (e.g. tables 2 and 3)”. Complying with this earlier comment we deleted the columns reporting these values in the revised version of the manuscript.

Level of interest: An article whose findings are important to those with closely related research interests
Quality of written English: Acceptable
Statistical review: No, the manuscript does not need to be seen by a statistician.
Declaration of competing interests: I declare that I have no competing interests

******************************************************************************************

Referee 2:

Reviewer: Audrey Boutron

Reviewer's report:

The revised manuscript is in good agreement with asked revisions

Discretionary Revisions:
1. Title should be in adequation with the revised manuscript ie : A 241kb pair 11-chromosomal deletion covering the BBOX1 and FIBIN genes in a patient with microcephaly
   Answer: We will keep the proposed title (the effect of homozygous deletion of the BBOX1 and Fibin genes on carnitine level and acyl carnitine profile) as it better reflects the general scope of our report.
2-table 3 mean from two measurements at two different age (42 and 60 months) is quite irrelevant
Answer: We deleted the corresponding column in Table 3 as suggested

Level of interest: An article whose findings are important to those with closely related research interests
Quality of written English: Acceptable
Statistical review: No, the manuscript does not need to be seen by a statistician.