Author’s response to reviews

Title: Identification of a novel nonsense mutation in SH2D1A gene from a patient with X-linked lymphoproliferative syndrom type 1: a case report

Authors:

Xiaodong Lyu (xiaodonglv2007@sina.com)
Zhen Guo (gzhenc2008@163.com)
Yangwei Li (liyangweimry@163.com)
Ruihua Fan (13653866010@163.com)
Yongping Song (ypsong2017@163.com)

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Identification of a novel nonsense mutation in SH2D1A gene from a patient with X-linked lymphoproliferative syndrom type 1: a case report

Xiaodong Lyu; Zhen Guo; Yangwei Li; Ruihua Fan; Yongping Song, Ph.D.

BMC Medical Genetics

Dear Editor,

We would like to thank you for your interest in our manuscript and for your valuable suggestions. As requested, we revised our manuscript in response to the comments. Our point-by-point responses can be found below.

We hope that you will now find our revised manuscript suitable for further process.
Thank you in advance for your kind consideration of this paper.

Sincerely,

Yongping Song.

POINT-BY-POINT RESPONSE

1) the manuscript needs to be revised by a native English person;

Reply: the manuscript had been revised by a specialized agency.

2) sequence variants should be described according to nomenclature guidelines (e.g. http://www.hgvs.org/mutnomen/); care must be taken in zygosity description;

Reply: the sequence variants had been revised according to HGVS guidelines. Zygosity description had been checked and revised.

3) regarding "likely pathogenic" a clear description of the novel variant impact including the method(s)/software(s) used in the assessment must be included;

Reply: A result of RT-PCR was included to assessment the impact of the novel nonsense mutation. The method was described and a supplementary table was attached.

4) the immediate impact of this case report is not clearly explored, and a genotype-phenotype correlation must be included.

Reply: We slightly revised the Discussion and Conclusions section to clarify the immediate impact. We believed that a genotype-phenotype correlation was demonstrated clearly by the result of RT-PCR.