Reviewer's report

Title: A novel mutation in the SH3BP2 gene causes cherubism

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Reviewer: Kiyonao Sada

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The manuscript by Cui-Ying Li & Shi-Feng Yu described a novel point mutation D419G in the SH3BP2 gene, which cause human inherited disease cherubism. The finding is very important and well described in the manuscript. However, there are some points that should be addressed before the publication.

1. Analysis of 3BP2/SH3BP2-deficient mice has been already published (de la Fuente, et al. Mol. Cell. Biol. 26(14):5214-5225, 2006) although the authors described that “3BP2-deficient mice are not available”. The author should mention about the phenotype of 3BP2/SH3BP2-deficient mice in this manuscript.
2. The author should unify the name of the molecule either “SH3BP2” or “3BP2”. In some sentences, the author used “3BP2” instead of “SH3BP2”.
3. In Discussion part, c-abl is NOT an oncogene.
4. Is it possible to compare the level of protein expression by immunohistochemistry and/or immunoblotting with the normal healthy control?

What next?: Accept after minor essential revisions

Level of interest: An article whose findings are important to those with closely related research interests

Quality of written English: Acceptable

Statistical review: No

Declaration of competing interests:

I declare that I have no competing interests.