Reviewer's report

Title: Variable expensiveness familial cherubism: woman transmitting cherubism without suffering the disease.

Version: 2 Date: 29 September 2013

Reviewer: Elif Bahar Tuna

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Major Compulsory Revisions
1- Methodology of mutation analysis should be explained better.
2- Results section might be add before discussion. Section names should be revised.
3- In the paper the authors should state that osteoclasts and mast cells express SH3BP2 and potential function of the protein coded for by this gene or how it may result in the cherubism phenotype. It also would be interesting to add how gene expression changes over time and the bone manifestations appear to dissolve with age.
4- Ethical committee decision should be added for this study.
5- Figures need to better resolution.

Minor Essential Revisions
1- There are a number of grammatical errors that require correction.
2- Cherubism appears both capitalized and lower case and it should be in lower case unless it occurs at the start of a sentence.
3- Differential diagnosis is already known well about cherubism and other diseases; this paragraph couldn’t reach the goal.
4- It seems necessary to include mutation analysis results since the molecular findings are already described in the text.
5- The pedigree is unusual and not necessary to use name’s on it.
6- Panoramic graphs might have better resolution especially in Figure 1.
7- Table 1 title and reference is missing.
8- Again, a well-thought out discussion of what is known about SH3BP2 in bone homeostasis would add significantly to the value of this paper for readers of the journal.

Discretionary Revisions
1- Make the paper more clear, it’s not usual to see names abbreviations in the description of cases in scientific papers.
2- Discussion part should revise carefully.
3- You can do figures all in one for per case, include patient’s face, panoramic graph and mutation analysis results. You can see similar figure designs in some good-prepared papers.

Level of interest: An article of importance in its field

Quality of written English: Needs some language corrections before being published

Declaration of competing interests:
I declare that I have no competing interests.