Author's response to reviews

Title: A novel mutation in the SH3BP2 gene causes cherubism

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Answers to the Reviewers’ comments

Reviewer 1
We have added a couple of sentences in page 2: "The dysplasia histologically consists of a mononuclear fibroblastic stroma with large numbers of multinucleated giant cells and cyst formation (Southgate, 1998), but the characteristic perivascular collagen cuffing is sometimes present."
We have made some mistakes with Figure 1. We have corrected them. In fact, the family consists of 19 biologically related members, 3 of whom were female patients with cherubism. In this study, we just recruited 17 members for the genetic analysis as two male members, II-8 and II-11, were missing. The 2 missing male subjects may not affect the estimate of penetrance in females.
We have added the panoramic X-ray films and facial photos of the other two patients with cherubism (Figures 3 and 4). Their expressivity varies based on their facial appearances and panoramic radiographic findings.
We have shortened the Discussion section and added one paragraph to discuss penetrance and expressivity (pages 8-9).
We have removed the old Figure 3 (electropherograms of partial sequences of exon 9 of the SH3BP2 gene).

Reviewer 2
We have deleted the two sentences as suggested by the reviewer.

Reviewer 3
We have deleted the sentences regarding the 3BP2-deficient mice to shorten the discussion as suggested by reviewer 1. It may not be necessary to mention the animal model in this study.
We have unified the name of the genes. We use the SH3BP2 throughout the text.
We have deleted the sentences regarding the c-abl gene in the Discussion section to shorten it as suggested by reviewer 1.
We did not have results of the levels of protein expression by immunohistochemistry and/or immunoblotting with the normal healthy control.