Reviewer's report

Title: A novel mutation in the SH3BP2 gene causes cherubism

Version: 1 Date: 18 August 2006

Reviewer: Ernst Reichenberger

Reviewer's report:

General

Major Compulsory Revisions (that the author must respond to before a decision on publication can be reached)

Minor Essential Revisions (such as missing labels on figures, or the wrong use of a term, which the author can be trusted to correct)

The authors present a case study of 3 familial cases of cherubism. They performed mutation analysis in the known cherubism gene SH3BP2 and found a new mutation within the 6 amino acid region in which cherubism mutations have been described.

The description of this new mutation adds to the picture of cherubism. The discussion has review character, however, two items may cause misunderstanding for the reader.

1) in the discussion (page7) the authors write: “we confirmed the biochemical relevance of individual point mutation of 3BP2 in mast cells from patients with cherubism”. While it is expected that the mutation is present in all cell types, it is not shown that mast cells are relevant in any way in this manuscript. Please delete this statement.

2) the authors write on page 8: “The second and third molars also disappear in patients with cherubism.” While molars may not erupt in some patients, this is not a characteristic for cherubism. The authors may have been misled by the highly speculative remarks in Hyckel et al., 2005. Please delete this sentence or rephrase it in the correct context.

Discretionary Revisions (which the author can choose to ignore)

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:

we have a patent pending on the use of Sh3bp2 and the mutations that have been published in Ueki et al., 2001. The publication of this manuscript has no positive or negative impact on filed patent application.