Reviewer's report

Title: A Novel RUNX2 Missense Mutation Predicted to Disrupt DNA Binding Causes Cleidocranial Dysplasia in a Large Chinese Family with Hyperplastic Nails

Version: 2 Date: 26 September 2007

Reviewer: Filippo M. Santorelli

Reviewer's report:

General

This work reports a four-generation Chinese family with Cleidocranial dysplasia (CCD) and elevated bed nail. The clinical phenotype in the family was heterogeneous with different presentation of the CCD syndrome within relatives but all presented elevated bed nail, a feature that is reported rarely in CCD cases. To my knowledge, a single family has presented such a clinical feature.

All affected cases showed a novel p.Leu136Pro mutation in the RUNX2 gene. The mutation is predicted by computer modeling to affect CBF binding, but this was not directly tested. A functional study might have improved the pathogenic role of the mutation.

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

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

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Discretionary Revisions (which the author can choose to ignore)

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What next?: Reject because too small an advance to publish

Level of interest: An article of insufficient interest to warrant publication in a scientific/medical journal

Quality of written English: Acceptable

Statistical review: No, the manuscript does not need to be seen by a statistician.
Declaration of competing interests:

'I declare that I have no competing interests'