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: 17 September 2007

Reviewer: Florian Otto

Reviewer's report:

General

1) The manuscript describes two novel findings: the identification of a novel mutation in the RUNX2 gene in a family with cleidocranial dysplasia and a novel clinical phenotype (hyperplastic tails) that co-segregates with the typical CCD features in this family. It remains unclear whether this additional phenotypic feature is caused by the RUNX2 mutation or another genetic alteration in the vicinity of the RUNX2 gene.

-----------------------------------------------------------------------------------------------

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)

2) There seems to be some misspelling throughout the manuscript (e.g. hypoplastic --> hyperplastic; recruited --> recruited; hypotrophic --> hypertrophic; latter --> lateral; informal --> informed; etc.). The expression "polydramnia" is unclear to me. Maybe a native English speaker (N.S.?) could go over the manuscript again. The sentence "There is such a sequence variant at that position in unaffected individuals." seems should probably read "no such variant". Since some of these typographical errors disrupt the meaning they must be corrected.

-----------------------------------------------------------------------------------------------

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: Needs some language corrections before being published

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.