Reviewer’s report

Title: Osteopoikilosis and multiple exostoses caused by novel mutations in LEMD3 and EXT1 genes respectively - coincidence within one family.

Version: 2 Date: 24 February 2010

Reviewer: YUN ZHANG

Reviewer’s report:

It is interesting to study the involvement of LEMD3 and EXT1 in a family with significant phenotypes of osteopoikilosis. The authors carried out straightforward experiments to confirm their hypothesis. It adds on additional information to the field.

Questions:
In the Results of the Abstract, mutation should be presented in standard format as (c.2203C>T) and (p.R735X), which later is corrected stated in the main text.
In the Methods section, if the authors mentioned all the details in the PCR mix, Mg++ should also be mentioned.
As cDNA sequence result was mentioned in the Results section, therefore cDNA sequencing should be mentioned in the Methods section.
Results part, page 9 line 17, the splice site mutation in exon as detected in DNA from the blood lymphocytes, and from cDNA. Please add which tissue cDNA was synthesised from though this has been mentioned in the Methods section.
Figure legend:
Should be legend for Figure 1 put in front of the one for Figure 2?
Figure 2. (A), please indicate clearly whether the sclerotic changes are in both hands, right or left feet, right or left side of the pelvis. Adding on arrows would be helpful to the readers. The same problems in (B).
Figure 3.(B) amino acid not aminoacid (also in Page 9 text).
Table 1. Indicating the patients’ age and sex would be helpful.

Level of interest: An article whose findings are important to those with closely related research interests

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.