Reviewer’s report

Title: Mutations in FKBP10 can cause a severe form of isolated Osteogenesis imperfecta

Version: 2 Date: 5 October 2011

Reviewer: Sajid Malik

Reviewer’s report:

Ref:

In this paper, Steinlein and colleagues report their clinical and molecular findings on brittle bone disorder, Osteogenesis imperfecta (OI). The manuscript is well written and the evidence of mutation causing the phenotype is convincing.

I have the following suggestions to improve the manuscript:

Major Compulsory Revisions:

- Results: Clinical findings: For a better clinical ascertainment, the authors should provide previous results of basic biochemical tests like alkaline phosphate level (ALP).

- ………. Permanent teeth are less affected. …..

As the authors propose to expand the phenotypic spectrum of the anomaly, they need to provide the dental photos of at least one affected subject.

- Two unrelated parents have three offspring affected with OI. This necessitates heterozygous status of both parents and independent origin of mutation in both paternal and maternal lineages. This information need to be interrogated. It would be worthwhile to check if the parents have common ancestry. This could be readily identified from the present genotyping data. This finding could have important implications in genetic counseling.

Minor Essential Revisions

- Background: Add OMIM number (610968)

- Clinical finding: The family history is empty of bone diseases or major malformations.

Suggested change: There is no family history of bone diseases or major malformations

- Discussion: A couple of sentences on the function of gene should be added. For instance, how a mutant protein might lead to the phenotype witnessed in the subjects.
- Discussion: Add full stop (.) at the end of third sentence.

**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.