Author's response to reviews

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

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Author's response to reviews: see over
Dear Editor

Please find included our revised manuscript entitled

**Mutations in FKB10 can cause a severe form of isolated Osteogenesis imperfecta**

that we would like to resubmit to BMC Medical Genetics.

We would like to thank the reviewers for their work and valuable comments that have helped us to improve the quality of our initial submission. According to their suggestions we made the following changes in our manuscript:

**Reviewer 1:**

1. Results of alkaline phosphatase testing have been added to the results section. However, it should be kept in mind that the three brothers are over 40 years old, with massively reduced bone mass. Therefore today’s lab results are probably not representative for those that would have been obtained in younger years. According to law, old records are mostly only kept for 10 years, thus they are no longer available.

2. A dental photo showing dentinogenesis imperfecta of the primary teeth of the youngest brother has been added to figure 2.

3. Following the reviewers’ suggestion we performed estimates of the relative relationship by identity-by-state allele sharing and discuss its conclusions in the first paragraph of Discussion and in the Conclusions paragraph. The detailed results are added as a supplementary file.

4. The OMIM number has been added to the background section.

5. The suggested change regarding the family history has been made.
6. Following the reviewer’s suggestion we added a short paragraph about the possible functional consequences of FKBP10 mutations to the discussion.

**Reviewer 2:**

The missing OI gene, SERPINF1, was added to the background section. The matching reference was put into the reference section.

**Additional changes:**

A statement about the compliance with the Helsinki Declaration and the approval by the ethics committee was added to the Method section.

Table 1 is now included in the main manuscript file, after the reference section.

Again, we would like to thank both reviewers for their constructive comments and suggestions.

With best regards,

Ortrud K. Steinlein, M.D., Ph.D.