Author's response to reviews

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

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Dear Editor

Please find included a manuscript entitled

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

that we would like to submit to BMC Medical Genetics.

Osteogenesis imperfecta (OI) is a brittle bone disorder that, in the majority of patients, is caused by autosomal dominant mutations in procollagen genes. Recently, some genes have been identified that cause recessive forms of OI. One of these genes is *FKBP10*, a putative procollagen chaperon. Most of the previously identified patients with *FKBP10* mutations had Bruck syndrome, a genetic disorder characterized by arthrogryposis congenita and OI. Only a few of these patients were described as having isolated OI type III, a classification that was questioned in follow-up reports. The patients presented in our report demonstrate that *FKBP10* mutations are indeed able to cause isolated OI. Furthermore they show that the phenotypic spectrum is much broader than previously believed and that the most severe form of OI type IV as well as Dentinogenesis imperfecta have to be added to the list of possible outcomes.

Far less is known about recessive forms of OI than about their autosomal dominant counterpart parts. We therefore think that our data will help to understand the clinical variation that can be associated with these rare disorders.

We sincerely hope that you will find our work suitable for publication and would very much like to thank you for your time and effort in advance

With best regards,

Sincerely,

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