Mutations in the Gene Encoding the RER Protein FKBP65 Cause Autosomal-Recessive Osteogenesis Imperfecta

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(American Journal of Human Genetics, *86*, 551–559)

On page 555 under the section titled *Mutations in FKBP10 cause Recessive OI,* there are two errors in the nomenclature for the identified mutations. The *FKBP10* (NM_021939.3) mutation isolated in the Turkish cases (proband R06-113A) is c.321_353 del and is predicted to result in the deletion of eleven amino acids in the protein, p.Met107_Leu117 del. In the second paragraph of the subheading, the mutation in the Mexican-American family (proband R93-188) should be

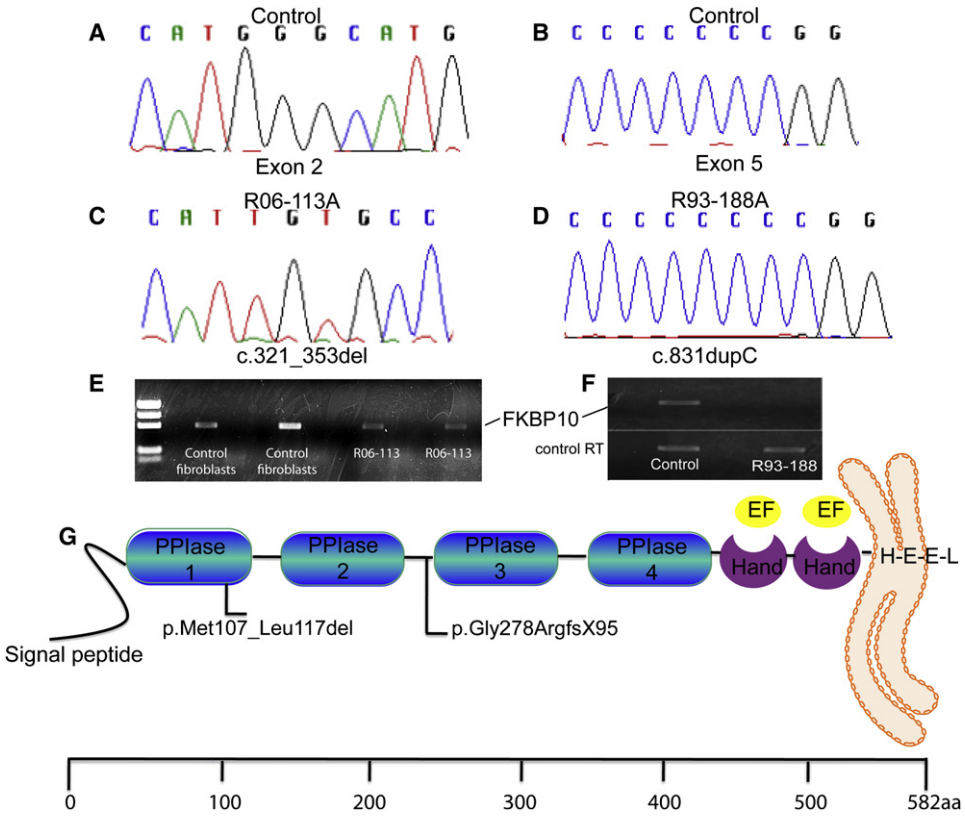

Figure 4. FKBP10 Mutation Detection
(A and B) Sequence analysis of exon 2 in control and a representative affected individual (R06-113A) with the mutation in the Turkish families.
(C and D) Exon 5 sequence analysis in control and a representative affected individual (R93-188A) with the mutation in the Mexican-American family (R93-188A).
(E) RT-PCR of FKBP10 cDNA from control and R06-113A fibroblasts showing that a FKBP10 cDNA is synthesized.
(F) RT-PCR of FKBP10 cDNA from control and R93-188A fibroblasts showing that a FKBP10 cDNA is not synthesized; lower band demonstrates control cDNA synthesis.
(G) Cartoon of the FKBP65 molecule with predicted protein consequences for each mutation. PPIase, peptidyl-prolyl cis-trans isomerase; EF/Hand domain; HEEL, putative ER-retention sequence.

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DOI 10.1016/j.ajhg.2010.09.002.

identified as c.831 dupC, which changes the glycine at position 278 to an arginine, and is predicted to result in a stop codon 94 amino acids downstream, p.Gly278ArgfsX95. The appropriate nomenclature modifications have been made to Figure 4. Note that the change in nomenclature also applies to the *Response to Shaheen et al.*[1] The authors regret the error.

Reference

1. Alanay, Y., and Krakow, D. (2010). Response to Shaheen et al. Am. J. Hum. Genet. *87*, 306–308.