FKBP10 and Bruck Syndrome: Phenotypic Heterogeneity or Call for Reclassification?

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We mistakenly designated the mutation in FKBP10 (NM_021939.3) in our two patients as c.1023insGGAGAATT and p.T342GfsX367. The proper designation is c.1016_1023dup and p.Thr342GlyfsX26. This error appears both in the text and in Figure 1E, in which we also showed the two mutations mistakenly described by Alanay et al as p.Gly107 Leu117del (the correct designation is p.Met107_Leu117del) and p.Gly278ArgfsX295 (the correct designation is p.Gly278ArgfsX95).¹ A revised version of Figure 1 with the three mutations properly named is shown below. Additionally, "protrusion" now reads "protrusio," and "inserted" has been replaced with "duplicated" in the legend. The authors regret this oversight and appreciate the opportunity to amend the record.

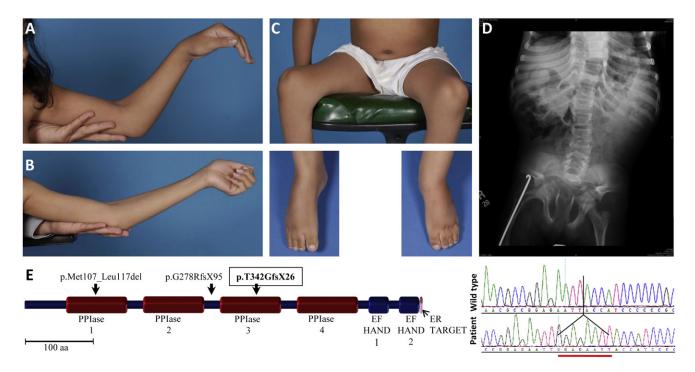


Figure 1. A Novel FKBP10 Mutation in Two Siblings with Bruck Syndrome

(A) Clinical photographs of the index patient and (B) his brother showing fixed flexion deformity of the elbows. (C) Note the severe flexion deformity of the knees in the index patient; the ankles are less severely involved. (D) Thoracolumar spine X-ray showing scoliosis and osteopenia. Severe protrusio acetabuli and intrmedullary rod fixation of the right femur fracture can also be seen. (E) Schematic representation of FKBP65 with the location of mutations indicated by arrows. Our mutation is boxed and shown next to the sequence chromatogram; the duplicated 8 bp are indicated by a red line.

Reference

1. Alanay, Y., Avaygan, H., Camacho, N., Utine, G.E., Boduroglu, K., Aktas, D., Alikasifoglu, M., Tuncbilek, E., Orhan, D., Bakar, F.T., et al. (2010). Mutations in the gene encoding the RER protein FKBP65 cause autosomal-recessive osteogenesis imperfecta. Am. J. Hum. Genet. 86, 551-559.

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