SEVERE OSTEOGENESIS IMPERFECTA AND EPIDERMOLYSIS BULLOSA SIMPLEX CAUSED BY FKBP10 MUTATION: New Case

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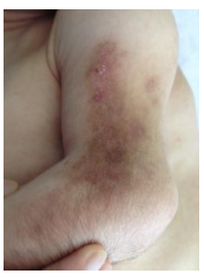
BACKGROUND

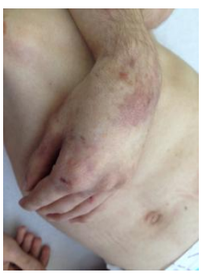
Mutations in genes encoding type 1 procollagen (T1PC) and proteins responsible for posttranslational modifications of the T1PC heterodimer may result in brittle bone disorder osteogenesis imperefecta (OI). FKBP65 is a known chaperone for type I procollagen and encoded by *FKBP10.* Autosomal-recessively inherited epidermolysis bullosa simplex and moderately severe OI caused by *FKBP10* mutation reported in consanguineous Turkish and Mexican families.

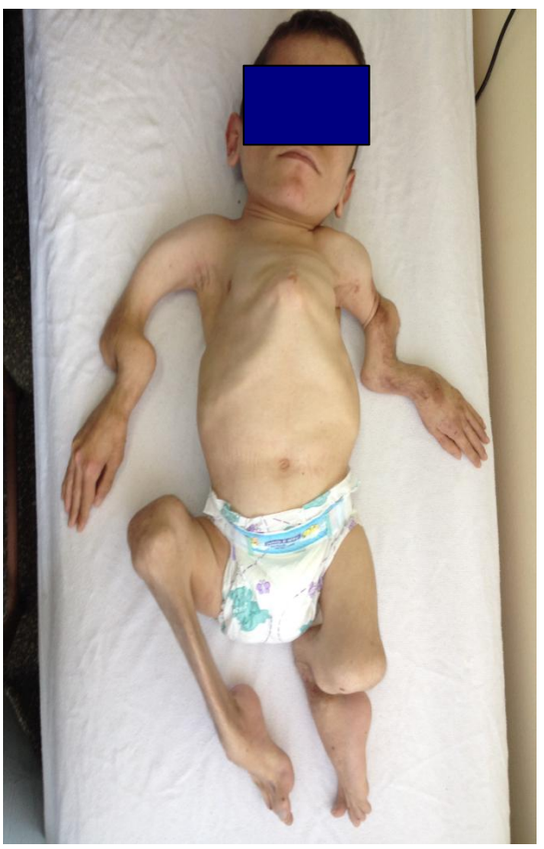
CASE REPORT

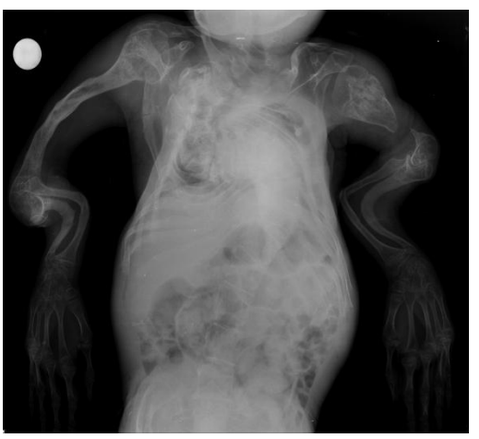
$19^{5/12}$ years-old male admitted with multiple skin lesions and recurrent bone fractures since birth. His parents was first cousin. He had normal birth length and weight but was born with skin bilsters on his body. His left arm was broken during delivery and until today, he had recurrent long bone fractures. He had also recurrent vertebral fractures prolonged progressive kyphoscoliosis.

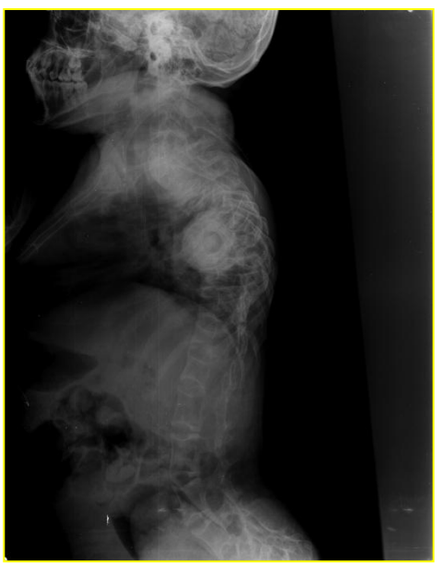
At 10 years-old, he diagnosed as OI and alendronat treatment started, however patient did not use drug. He had untreated for a period of nine years and had multiple fractures in long bones and vertebrae. On the admission his weight was15.9 kg , lenght was 87cm. He had greyish- white sclera and normal teeth. Diffuse bullous erythematous lesion was determined on extansor surface of both extremities and thorax. Severe kyphoscoliosis and multiple deformities in both extremities due to recurrent fractures was determined. He was early-pubertal stage.Skin biopsy consisting with bullous dermatitis. BMD Z score was -6.7 (0.340 gr/cm3) on lumbar vertebrae 1-4. Odiometric examination revealed mild mixt type sensorineural hearing lost. Pamidronat-sodium therapy was started as 1mg/kg/day for 3-days (three-monthly). *FKBP10* gene mutation analysis was performed in all family members. **Patient has homozygous p.Met107-Leu117del mutation in *FKBP10*.** Parents has heterozygous this mutation.

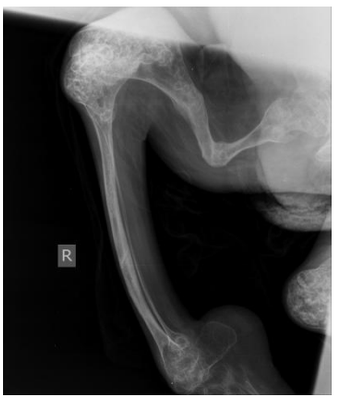












CONCLUSION

Homozygosity for a 33 base pair deletion (c.321_353del) in FKBP10 is resulted in deletion of 11 amino acids (p.Met107-Leu117del). Disrupted Type 1 collagen was synthesized and caused severe skin and bone disorders in patients.