Supplementary Table 1: Diagnostic criteria for patients with OI

OI type	Clinical features	Inheritance	Biochem.Abn	Mutation
I	Normal stature, little or no deformity, blue sclerae, hearing loss	AD	50% reduction in type I collagen synthesis	*COL1A1*
II	Lethal; minimal calvarial mineralization, beaded ribs, compressed femurs, long bone deformity	AD	Structural alterations of type I collagen chains - overmodification	*COL1A1* *COL1A2*
III	Progressively deforming bones, DI, hearing loss, very short stature	AD	Structural alterations of type I collagen chains – overmodification	*COL1A1* *COL1A2*
IV	Normal sclerae in adult, mild/moderate deformity, variable short stature, DI, some hearing loss	AD	Structural alterations of type I collagen chains – overmodification	*COL1A1* *COL1A2*
V	Similar to type IV plus calcification of interosseus membrane of forearm, hyperplastic callus formation	AD	None described	*IFITM5*
VI	Similar to type IV with vertebral compression; mineralization defect	AR	None described	*SERPINF1*
VII	Moderate to severe, with fractures at birth, early deformity and rhizomelia	AR	Overmodification	*CRTAP*

*Patients classied into as type III and IV OI on a clinical basis were reclassified into appropriate subtypes when genotype information was available.