Table 1

Clinical, genetic and mechanistic subtypes of osteogenesis imperfecta, adapted from Forlino & Marini[1]

Group	Subtype	Genes	Severity	Inheritance	Mechanism
Group A	Type I	*COL1A1* or *COL1A2*	Mild	AD	Defects in collagen synthesis, structure or processing
	Type II	*COL1A1* or *COL1A2*	Lethal	AD	
	Type III	*COL1A1* or *COL1A2*	Severe	AD	
	Type IV	COL1A1 or *COL1A2*	Moderate	AD	
	Type XIII	*BMP1*	Moderate	AR	
Group B	Type VII	*CRTAP*	Severe	AR	Post-translational modification of collagen
	Type VIII	*LEPRE1*	Severe	AR	
	Type IX	*PPB*	Severe	AR	
	Type XIV	*TMEM38B*	Severe	AR	
Group C	X	*SERPINH1*	Severe	AR	Defects with collagen folding or cross-linking
	XI	*FKBP10*	Severe	AR	
	-	*PLOD2*	Severe	AR	
	-	*P4HB*§	Mild	AD	
Group D	V	*IFTM5*	Moderate	AD	Defects in bone mineralisation
	VI	*SERPINF1*	Severe	AR	
Group E	XII	*SP7*	Severe	AR	Defects in osteoblast differentiation
	XV	*WNT1*	Severe	AR	
	XVI	*CREB3L1*	Severe	AR	

The groupings shown are based on the underlying mechanisms of disease.
AR = autosomal recessive AD = autosomal dominant
§ Mutations in *P4HB* have been associated with a Cole-Carpenter syndrome in which an osteogenesis imperfecta-like phenotype occurs as part of a wider syndrome, but recent reports indicate that *P4HB* mutations can also result in a mild osteogenesis imperfecta phenotype