Nosology (Classifications) of Osteogenesis Imperfecta

OI TYPE	INHERITANCE	PHENOTYPE	GENE MUTATION
DEFECTS IN COLLAGEN SYNTHESIS, STRUCTURE, OR PROCESSING			
I	AD	Mild	Null COL1A1 Allele
II	AD	Lethal	COL1A1 or COL1A2
III	AD	Progressive Deforming	COL1A1 or COL1A2
IV	AD	Moderate	COL1A1 or COL1A2
XIII	AR	Mild/Severe	BMP1
DEFECTS IN BONE MINERALIZATION			
V	AR	Variable, Distinctive Histology	IFITM5
VI	AR	Moderate/Severe	SERPINF1
DEFECTS IN COLLAGEN MODIFICATION			
VII	AR	Severe (Hypomorphic) Severe/Lethal (Null)	CRTAP
VIII	AR	Severe/Lethal	LEPRE1
IX	AR	Moderate/Lethal	PPIB
XIV	AR	Severe	TMEM38B
DEFECTS IN COLLAGEN FOLDING AND CROSS-LINKING			
X	AR	Severe/Lethal	SERPINH1
XI/BRKS1	AR	Mild/Severe	FKBP10
BRKS2	AR	Moderate/Severe	PLOD2
DEFECTS IN OSTEOBLAST DEVELOPMENT WITH COLLAGEN INSUFFICIENCY			
XII	AR	Severe	SP7
XV	AR	Severe	WNT1
XVI	AR	Severe	CREB3L1
XVII	AR	Progressive Severe	SPARC
XVIII	XR	Moderate/Severe	MBTPS2
XIX	AR	Severe	FAM46A/TENT5A
XX	AR	Progressive Severe/Lethal	MESD
XXI	AR	Severe + Neurodevelopmental	KDELR2
XXII	AR	Severe	CCDC134

Abbreviations:

AD = autosomal dominant; the mutation is inherited in a dominant manner
AR = autosomal recessive; the mutation is inherited in a recessive manner