Supplementary Table 1. Nosology of OI in Nosology of genetic skeletal disorders: 2023 revision (reference No. 56)

Group number/number of disorder	Name of group/name of disorder	Inheritance	Gene or locus
GROUP 26	**Osteogenesis imperfecta and bone fragility group**		
NOS 26-0010	Osteogenesis imperfecta, non‐deforming (Sillence type 1), COL1A1‐related	AD	*COL1A1*
NOS 26-0020	Osteogenesis imperfecta, non‐deforming (Sillence type 1), COL1A2‐related	AD	*COL1A2*
NOS 26-0030	Osteogenesis imperfecta, severe perinatal form (Sillence type 2) COL1A1‐related	AD	*COL1A1*
NOS 26-0040	Osteogenesis imperfecta, severe perinatal form (Sillence type 2), COL1A2‐related	AD	*COL1A2*
NOS 26-0050	Osteogenesis imperfecta, severe perinatal form (Sillence type 2), CRTAP‐related	AR	*CRTAP*
NOS 26-0060	Osteogenesis imperfecta, severe perinatal form (Sillence type 2), P3H1‐related	AR	*P3H1*
NOS 26-0070	Osteogenesis imperfecta, severe perinatal form (Sillence type 2), PPIB‐related	AR	*PPIB*
NOS 26-0080	Osteogenesis imperfecta, progressively deforming (Sillence type 3), COL1A1‐related	AD	*COL1A1*
NOS 26-0090	Osteogenesis imperfecta, progressively deforming (Sillence type 3), COL1A2‐related	AD	*COL1A2*
NOS 26-0100	Osteogenesis imperfecta, progressively deforming (Sillence type 3), IFITM5‐related	AD	*IFITM5*
NOS 26-0110	Osteogenesis imperfecta, progressively deforming (Sillence type 3), SERPINF1‐related	AR	*SERPINF1*
NOS 26–0120	Osteogenesis imperfecta, progressively deforming (Sillence type3), CRTAP-related	AR	*CRTAP*
NOS 26–0130	Osteogenesis imperfecta, progressively deforming (Sillence type 3), P3H1-related	AR	*P3H1*
NOS 26–0140	Osteogenesis imperfecta, progressively deforming (Sillence type 3), PPIB-related	AR	*PPIB*
NOS 26–0150	Osteogenesis imperfecta, progressively deforming (Sillence type 3), SERPINH1-related	AR	*SERPINH1*
NOS 26–0160	Osteogenesis imperfecta, progressively deforming (Sillence type 3), FKBP10-related	AR	*FKBP10*
NOS 26–0170	Osteogenesis imperfecta, progressively deforming (Sillence type 3), TMEM38B-related	AR	*TMEM38B*
NOS 26–0180	Osteogenesis imperfecta, progressively deforming (Sillence type 3), BMP1-related	AR	*BMP1*
NOS 26–0190	Osteogenesis imperfecta, progressively deforming (Sillence type 3), WNT1-related	AR	*WNT1*
NOS 26–0200	Osteogenesis imperfecta, progressively deforming (Sillence type 3), CREB3L1-related	AR	*CREB3L1*

NOS 26–0210	Osteogenesis imperfecta, progressively deforming (Sillence type 3), SPARC-related	AR	*SPARC*
NOS 26–0220	Osteogenesis imperfecta, progressively deforming (Sillence type 3), TENT5A-related	AR	*TENT5A*
NOS 26–0230	Osteogenesis imperfecta, progressively deforming (Sillence type 3), MBTPS2-related	XLR	*MBTPS2*
NOS 26–0240	Osteogenesis imperfecta, progressively deforming (Sillence type 3), MESD-related	AR	*MESD*
NOS 26–0250	Osteogenesis imperfecta, progressively deforming (Sillence type 3) with neurodevelopmental features, KDELR2-related	AR	*KDELR2*
NOS 26–0260	Osteogenesis imperfecta, progressively deforming (Sillence type 3), CCDC134-related	AR	*CCDC134*
NOS 26–0270	Osteogenesis imperfecta, moderate form (Sillence type 4), COL1A1-related	AD	*COL1A1*
NOS 26–0280	Osteogenesis imperfecta, moderate form (Sillence type 4), COL1A2-related	AD	*COL1A2*
NOS 26–0290	Osteogenesis imperfecta, moderate form (Sillence type 4), WNT1-related	AR	*WNT1*
NOS 26–0300	Osteogenesis imperfecta, moderate form (Sillence type 4), IFITM5-related	AD	*IFITM5*
NOS 26–0310	Osteogenesis imperfecta, moderate form (Sillence type 4), CRTAP-related	AR	*CRTAP*
NOS 26–0320	Osteogenesis imperfecta, moderate form (Sillence type 4), PPIB-related	AD	*PPIB*
NOS 26–0330	Osteogenesis imperfecta, moderate form (Sillence type 4), FKBP10-related	AR	*FKBP10*
NOS 26–0340	Osteogenesis imperfecta, moderate form (Sillence type 4), SP7-related	AR	*SP7*
NOS 26–0350	Osteogenesis imperfecta with calcification of interosseous membranes and/or hypertrophic callus (OI type 5), IFITM5-related	AD	*IFITM5*
NOS 26–0360	Osteogenesis imperfecta with craniosynostosis (Cole-Carpenter syndrome), P4HB-related	AD	*P4HB*
NOS 26–0370	Osteogenesis imperfecta with craniosynostosis (Cole-Carpenter syndrome), SEC24D-related	AR	*SEC24D*