# Osteogenesis imperfecta en gerelateerde aandoeningen v3.0

## (41 genen, incl. 7 core-NL genen)

Amsterdam UMC
Universitair Medische Centra

| Gene | % covered > 30x | Associated phenotype description and OMIM ID |
|---|---|---|
| **ALPL** | >99 | Hypophosphatasia adult, 146300<br>Hypophosphatasia childhood, 241510<br>Hypophosphatasia infantile, 241500<br>Odontohypophosphatasia, 146300 |
| ANO5 | >99 | Gnathodiaphyseal dysplasia, 166260<br>Miyoshi muscular dystrophy 3, 613319<br>Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307 |
| B3GALT6 | >80 | Ehlers-Danlos syndrome, progeroid type, 2, 615349<br>Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 |
| B3GAT3 | >99 | Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600 |
| B4GALT7 | >99 | Ehlers-Danlos syndrome with short stature and limb anomalies, 130070 |
| BMP1 | >99 | Osteogenesis imperfecta, type XIII, 614856 |
| CCDC134 | >99 | no OMIM phenotype |
| CHST3 | >99 | Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095 |
| **COL1A1** | >99 | Caffey disease, 114000<br>Ehlers-Danlos syndrome, classic, 130000<br>Ehlers-Danlos syndrome, type VIIA, 130060<br>Osteogenesis imperfecta, type I, 166200<br>Osteogenesis imperfecta, type II, 166210<br>Osteogenesis imperfecta, type III, 259420<br>Osteogenesis imperfecta, type IV, 166220<br>{Bone mineral density variation QTL, osteoporosis}, 166710 |
| **COL1A2** | >99 | Ehlers-Danlos syndrome, cardiac valvular form, 225320<br>Ehlers-Danlos syndrome, type VIIB, 130060<br>Osteogenesis imperfecta, type II, 166210<br>Osteogenesis imperfecta, type III, 259420<br>Osteogenesis imperfecta, type IV, 166220<br>{Osteoporosis, postmenopausal}, 166710 |
| CREB3L1 | >99 | Osteogenesis imperfecta, type XVI, 616229: not associated with the gene in OMIM, but with a contiguous gene deletion including CREB3L1 |
| CRTAP | >99 | Osteogenesis imperfecta, type VII, 610682 |
| FKBP10 | >99 | Bruck syndrome 1, 259450<br>Osteogenesis imperfecta, type XI, 610968 |
| GORAB | >99 | Geroderma osteodysplasticum, 231070 |
| **IFITM5** | >99 | Osteogenesis imperfecta, type V, 610967 |
| KDELR2 | >99 | Osteogenesis imperfecta 21, 619131 |
| LIFR | >99 | Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559 |
| **LRP5** | >99 | Exudative vitreoretinopathy 4, 601813<br>Hyperostosis, endosteal, 144750<br>Osteopetrosis, autosomal dominant 1, 607634<br>Osteoporosis-pseudoglioma syndrome, 259770<br>Osteosclerosis, 144750<br>van Buchem disease, type 2, 607636<br>[Bone mineral density variability 1], 601884<br>{Osteoporosis}, 166710 |
| MBTPS2 | >99 | ?Olmsted syndrome, X-linked, 300918<br>IFAP syndrome with or without BRESHECK syndrome, 308205<br>Keratosis follicularis spinulosa decalvans, X-linked, 308800<br>Osteogenesis imperfecta, type XIX, 301014 |
| MESD | >99 | Osteogenesis imperfecta, type XX, 618644 |
| NBAS | >99 | Infantile liver failure syndrome 2, 616483<br>Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 |
| P3H1 | >99 | Osteogenesis imperfecta, type VIII, 610915 |

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| P4HA1 | >99 | no OMIM phenotype |
| P4HB | >99 | Cole-Carpenter syndrome 1, 112240 |
| PLOD2 | >99 | Bruck syndrome 2, 609220 |
| PLOD3 | >99 | Lysyl hydroxylase 3 deficiency, 612394 |
| **PLS3** | >99 | Bone mineral density QTL18, osteoporosis, 300910 |
| PPIB | >99 | Osteogenesis imperfecta, type IX, 259440 |
| SEC24D | >99 | Cole-Carpenter syndrome 2, 616294 |
| SERPINF1 | >99 | Osteogenesis imperfecta, type VI, 613982 |
| SERPINH1 | >99 | ?Osteogenesis imperfecta, type X, 613848<br>{Preterm premature rupture of the membranes, susceptibility to}, 610504 |
| SGMS2 | >99 | Calvarial doughnut lesions with bone fragility with or without spondylometaphyseal dysplasia, 126550 |
| SLC10A7 | >99 | Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363 |
| SP7 | >99 | ?Osteogenesis imperfecta, type XII, 613849 |
| SPARC | >99 | Osteogenesis imperfecta, type XVII, 616507 |
| TAPT1 | >99 | Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897 |
| TENT5A (=FAM46A) | >99 | Osteogenesis imperfecta, type XVIII, 617952 |
| TMEM38B | >99 | Osteogenesis imperfecta, type XIV, 615066 |
| **WNT1** | >99 | Osteogenesis imperfecta, type XV, 615220<br>{Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221 |
| XYLT1 | >99 | Desbuquois dysplasia 2, 615777 |
| XYLT2 | >99 | Spondyloocular syndrome, 605822 |

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014
OMIM release used for OMIM disease identifiers and descriptions : June 30th, 2015
“No OMIM phenotype” signifies a gene without a current OMIM association
OMIM phenotype descriptions between {} signify risk factors