

Disease

COL1A1 associated osteogenesis imperfecta^{1, 2}

Gene

COL1A1

Inheritance	AD
Current Treatments	Safe handling techniques, bisphosphonates
Evidence for Treatment	guideline ³
Non-Molecular Testing Availability	yes
Non-Molecular Test	skeletal survey ¹
Disease Frequency (per 100,000)	7.5 ²
Genes Included in Disease Frequency	COL1A1, COL1A2
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Osteogenesis+Imperfecta&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/books/NBK1295/>

² <https://pubmed.ncbi.nlm.nih.gov/25905334/>

³ <https://pubmed.ncbi.nlm.nih.gov/32117044/>