



# Osteogenesis Imperfecta

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# What is Osteogenesis Imperfecta?

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- A rare bone disease affecting approximately 20,000 to 50,000 people in the United States
- Brittle and weak
- Genes are mutated to form alterations in Type I collagen
- Quantitatively or qualitatively

# Classifications of OI

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- Type I, II, III, IV OI (autosomal dominant gene) affected by genes *COL1A1* and *COL1A2*
- Type V-XVIII OI (autosomal recessive gene)
- Bone fragility, teeth, the colors of sclera, hearing, the curvature of the spine

# The Effects of OI at a Molecular Level

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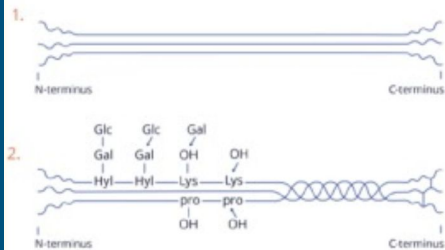
- Osteoblasts make alpha chains (units of Type I collagen)
- In OI, one alpha chain has an abnormally large amino acid
- Triple helix formation is slowed and hydroxylation & glycosylation is sped up
- The terminal ends of helix is cut off and released in the matrix as a collagen molecule

## (cont.)

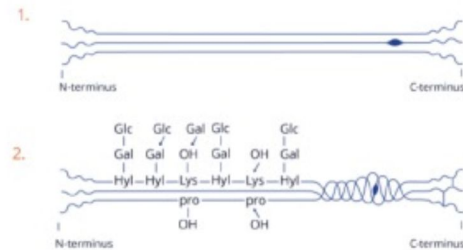
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- Multiple collagen molecules make an array that is wider than usual in OI conditions
- Osteoblasts make hydroxyapatite crystals that “fill in the gaps” between the array of collagen molecules
- More space=More mineralization

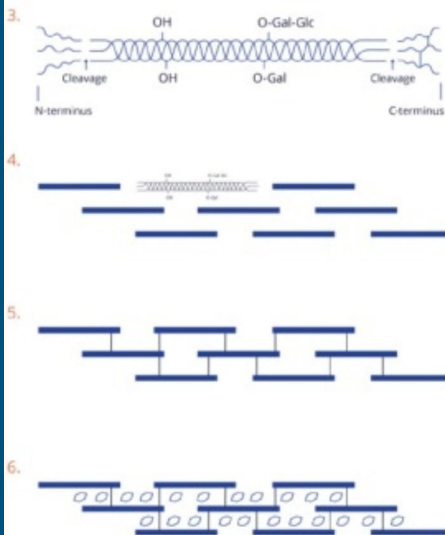
### Intracellular



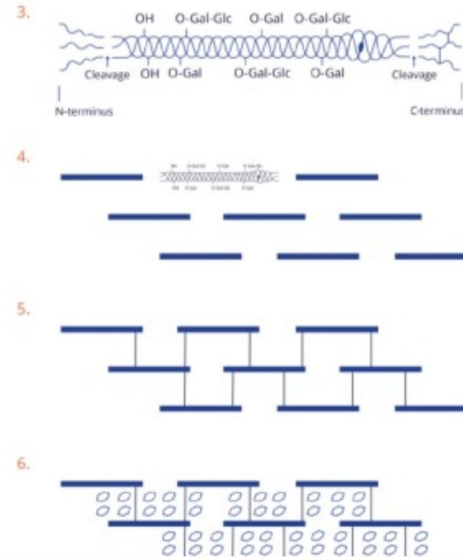
### Intracellular



### Extracellular



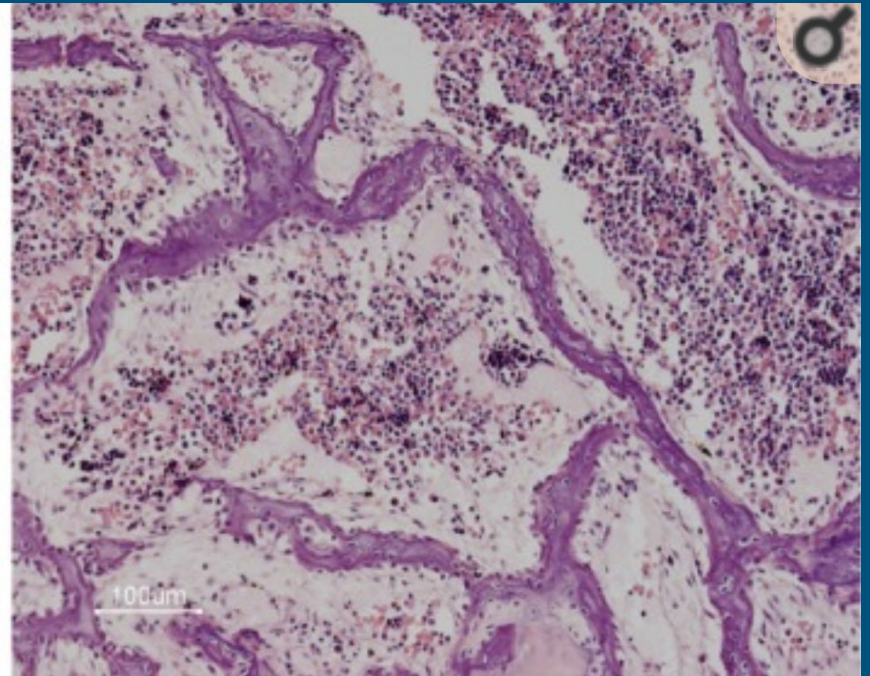
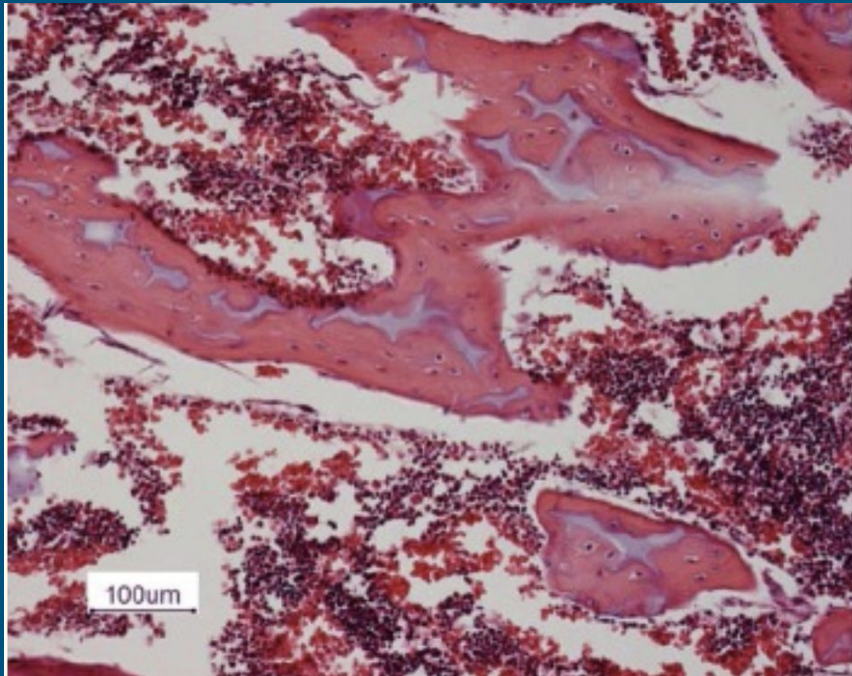
### Extracellular



# OI Effects on Bone Tissues

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- Increased mineralization in collagen molecules ultimately affects tissues
- Porosity for vascular vessels increases
- Lacunae (space containing osteocytes) widens
- Trabeculae are significantly thinner than how it should be





# What now?

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- Further research is being done to find solutions to OI
- Vitamin D monitoring and regulation
- Introduction of bisphosphonates used for Osteoporosis
- Single-sperm-based single nucleotide polymorphism haplotyping used for predictions
- OI is still very unknown to us as it is very rare and complex

# References

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Thank you