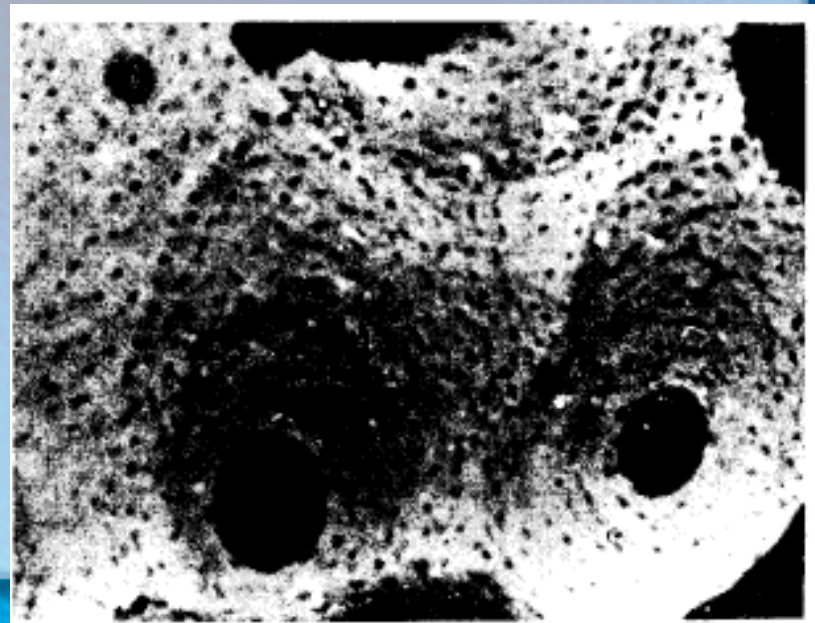
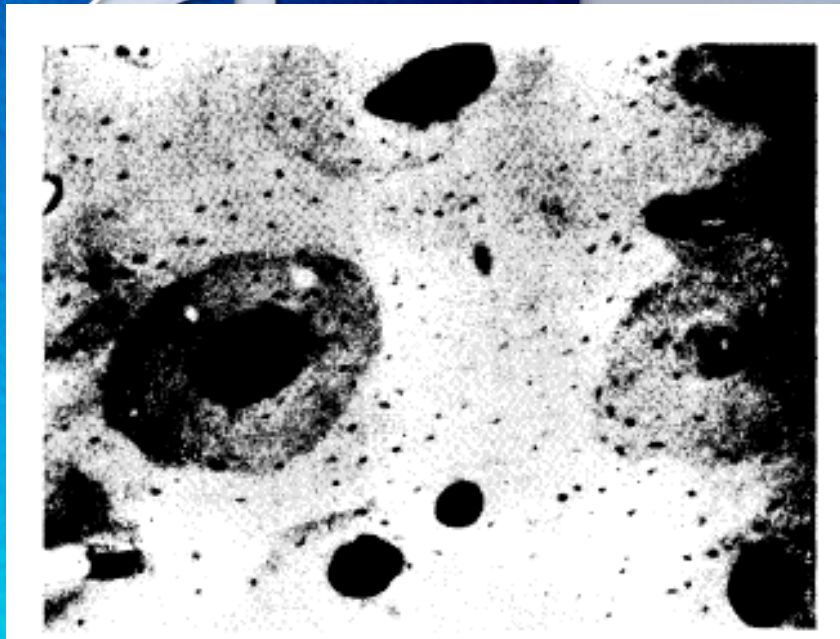




# Osteogenesis Imperfecta



# Way Back When...

- Ancient History
  - Egyptian, “Boneless”
- Ekman - 1788
  - Osteomalacia congenita
  - Lobstein
- Vrolik - 1849
- Sillence -1979
  - Types



Figure—Three examples of blue sclerae.



# When Genes Don't Fit...

- 90% - functional null allele of Col1A1 on Chromosome 17 or Col1A2 on Chromosome 7
- Autosomal Dominant (mostly)
  - 50%
- Collagen Biopsy Test
  - 87%, 98%
- DNA
  - ~100 Type I and II, less with III and IV



# Treatment - Then and Now

- Drug Therapy
  - Calcitonin?
- Surgery, Nonsurgical Management
  - Rodding
  - Physical therapy
- Proposed Gene Therapy?
  - Replacing cells
  - Silence mutant allele using antisense suppression therapy... put in an mRNA that complements sequence to prevent protein synthesis
- Biophosphanates

