Osteogenesis Imperfecta
Way Back When…

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