Type III Osteogenesis Imperfecta

Dana Wyman
September 30, 2010
BIOC 118Q

Introduction

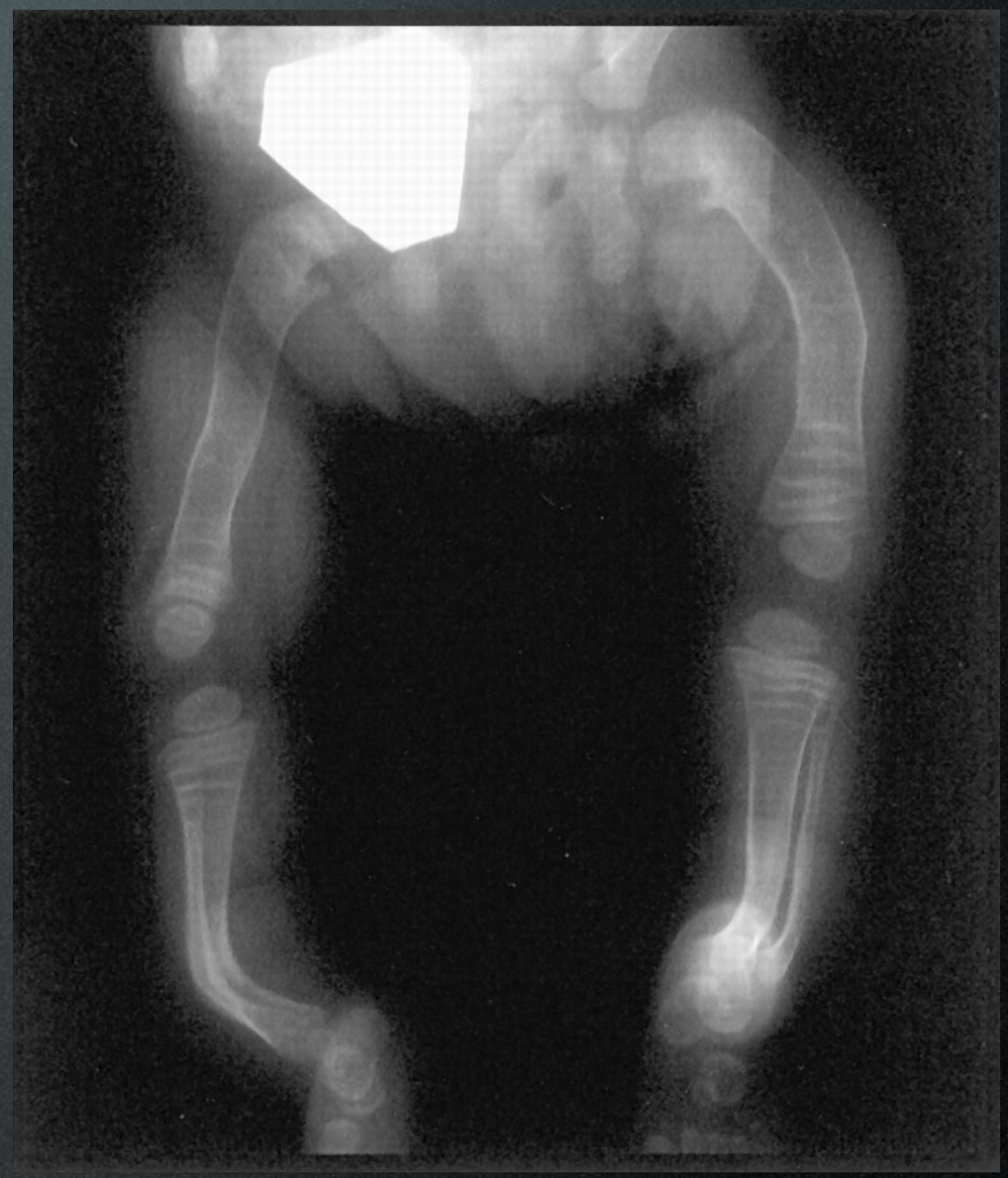
- Caused by a mutation to the COL1A1 or COL1A2 genes, which results in change to the structure of the pro alpha chains that make up the protein collagen
- Leads to deformed, easily fractured bones and severely restricts the mobility of the patient
- Prevalence of 1 to 2 for every 100,000 people

Characteristic Symptoms

- Frequent bone fractures caused by little or no trauma; may even occur before the baby is born
- Dentinogenesis imperfecta
- Hearing loss
- Micromelic Dwarfism
- Rib cage is barrel-shaped and other bones may be deformed
    - Kyphoscoliosis
- Blue Sclerae

Diagnostic Tools

- X-rays can reveal bone deformities, evidence of past fractures, and osteoporosis

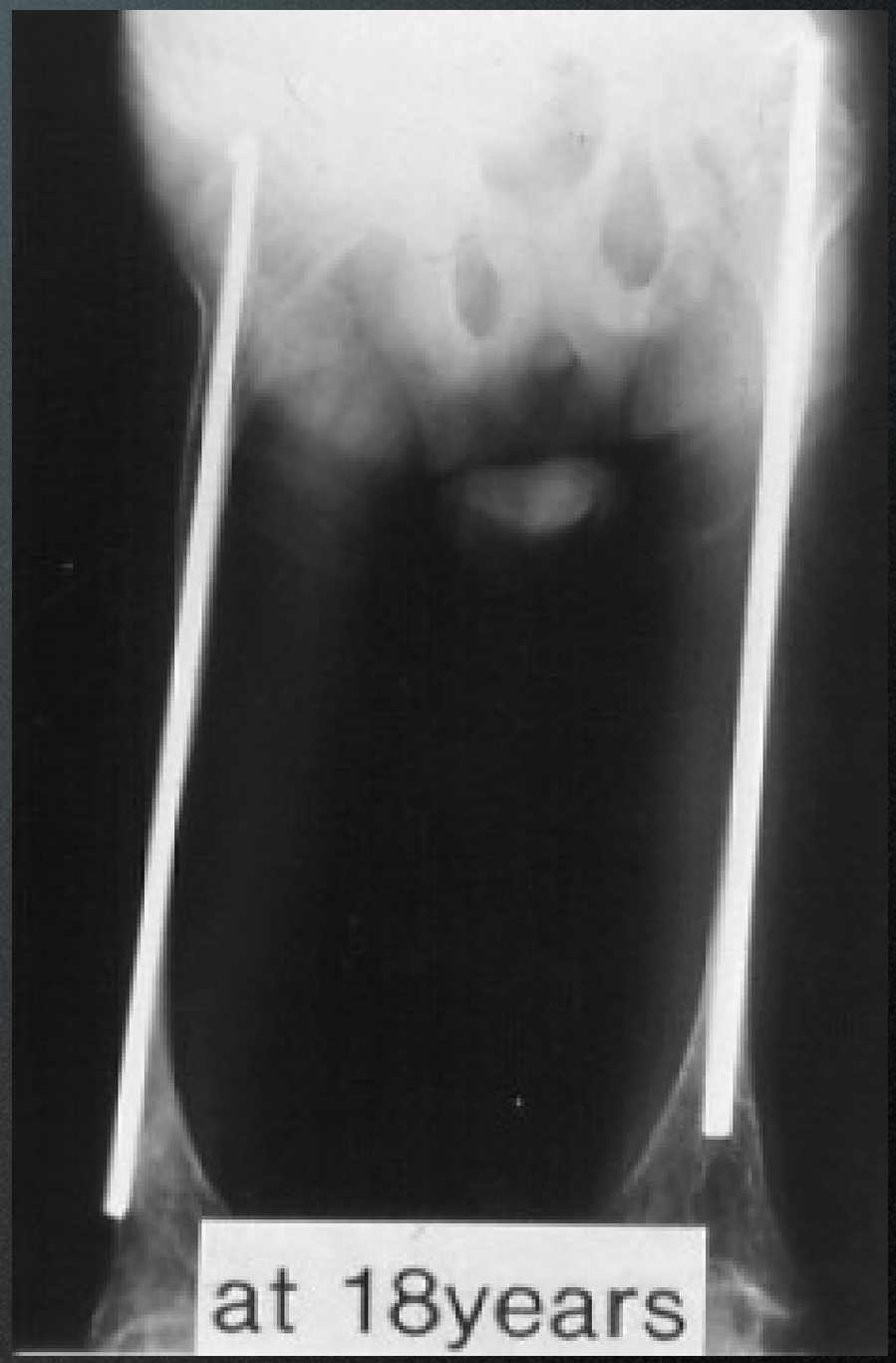


- Collagen analysis from a biopsy examines the types and quantities of collagen present

http://www.gfmer.ch/genetic_diseases_v2/gendis_detail_list.php?cat3=2147

Classical Treatments

- Non-Surgical
  - Physical Therapy
  - Bracing and Splinting
- Surgical
  - Intramedullary Rod Positioning
  - Bone Marrow Replacement (Being studied as a possibility)
- Medications
  - Pamidronate (Biphosphonate Drug)



http://gait.aidi.udel.edu/res695/homepage/pd_ortho/educate/clincase/clcsimge/homaoi3.jpg

Inheritance

- Autosomal Dominant, in many cases the result of a sporadic mutation
  - Chromosome 17, Location 17q21.33
- Sometimes autosomal recessive, but in those cases genes other than COL1A1 or COL1A2 tend to be involved
- Other types of Osteogenesis Imperfecta are autosomal dominant as well
- Full penetrance

The Role of Genetics in Diagnosis and Treatment

- DNA sequencing can identify close to 100% of mutations to the COL1A1 and COL1A2 genes
- Gene therapy is difficult due to the variety of genes involved

Sources

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