Osteogenesis Imperfecta

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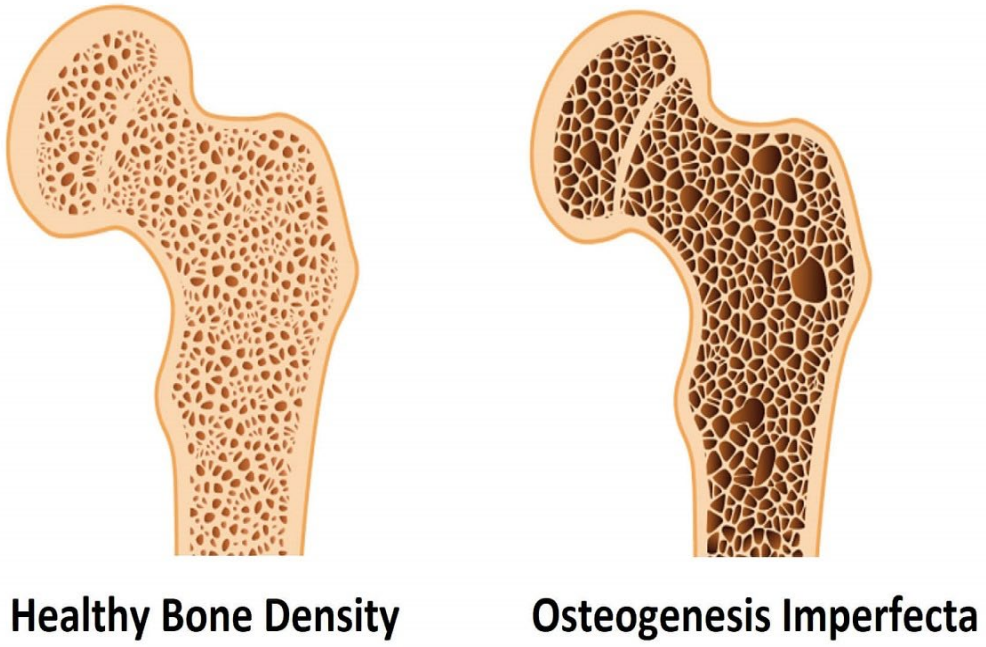
Overview

- What is Osteogenesis Imperfecta
- Epidemiology
- Etiology
- Impairments
- Patient Presentation
- Evaluative techniques
- Rehabilitation pathways



What is Osteogenesis Imperfecta

- Osteogenesis imperfecta (OI), also known as brittle bone disease, is a hereditary genetic connective tissue disorder that causes increased fragility in the bones.

Patient example

- 15-year-old male, reports having multiple fractures over the years. Low bone mineral density and repeated fractures led to an OI type I diagnosis. The patient began increasing calcium intake volume as well as vitamin D.
- Male neonate born with clinical features of OI type II. He was born with inability to cry at birth, multiple deformities with abnormal body posture.

Epidemiology

- The study of the distribution of causes of health outcomes and diseases in certain populations.

Epidemiology

- 1 in 15,000 to 20,000 births
    - Frequency ranges between OI variant

Etiology

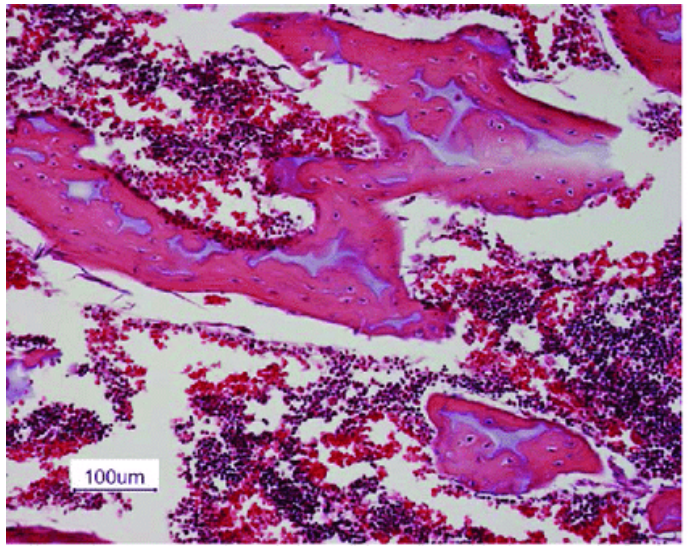
– The cause of a disease or condition.

Etiology

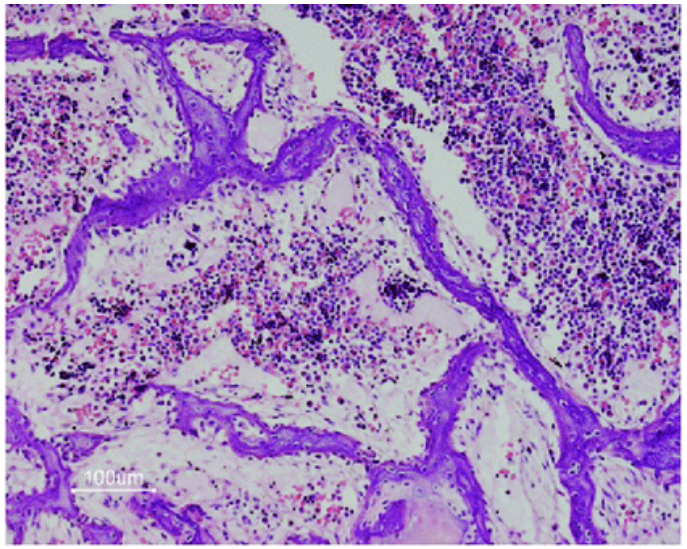
- Main cause is a genetic mutation of the COL1A1 and/or COL1A2 genes
    - About 80%-90% of mutations are caused by this autosomal mutation

Histology

- "Normal" bone can be characterized by:
  - thicker trabeculae (or supporting beams)
  - Less osteocytes
- Osteogenesis imperfecta bone can be characterized by:
  - Thinner trabeculae(or supporting beams)
  - More osteocytes



Normal bone

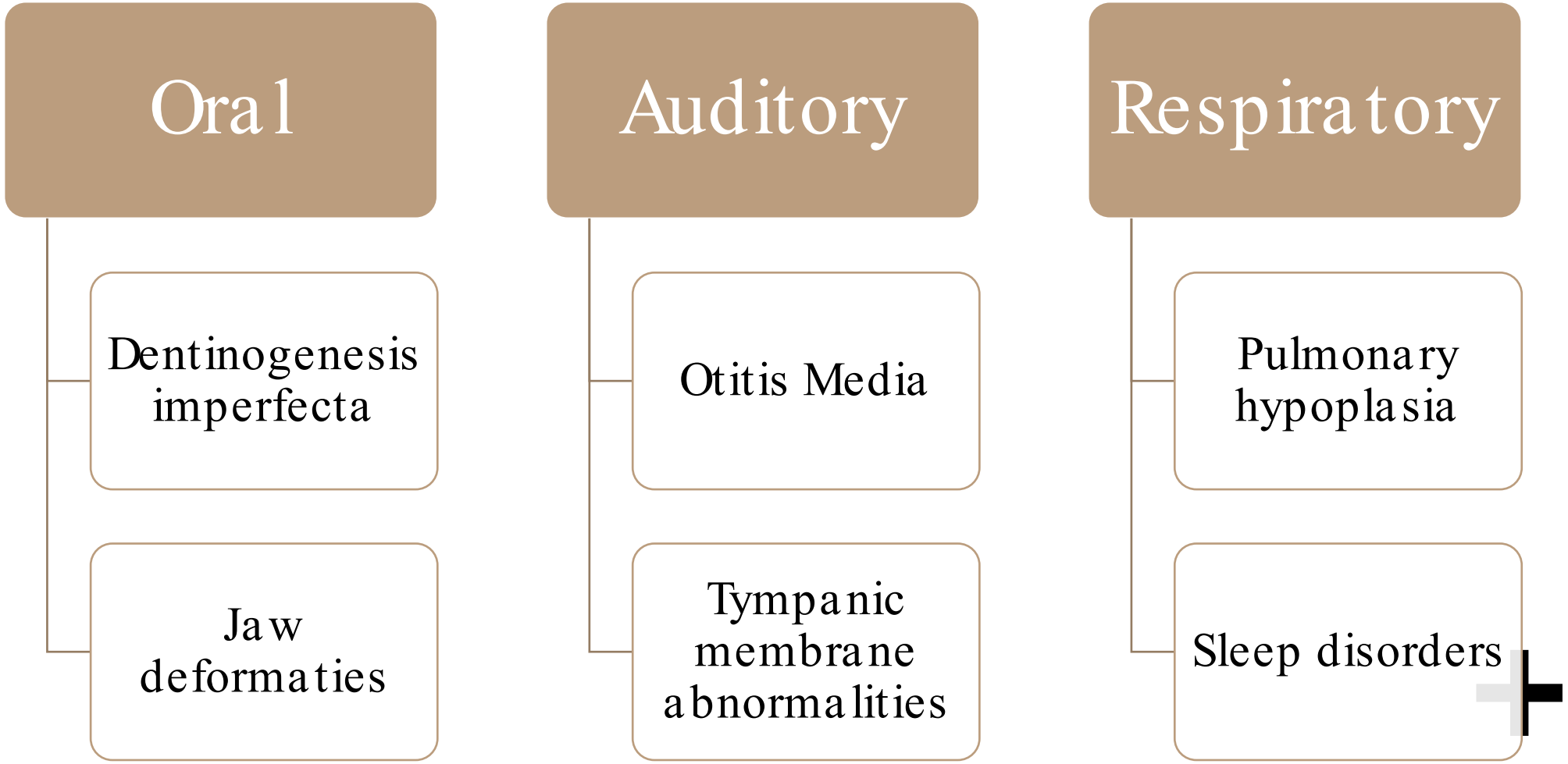


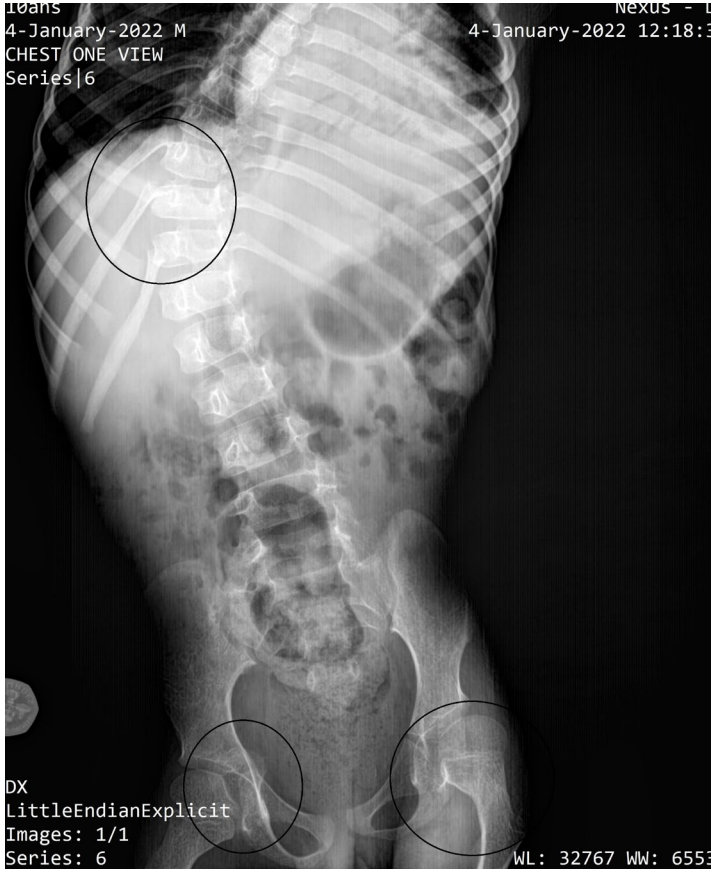
Osteogenesis imperfecta bone

Impairments

- Common Impairments
  - Musculoskeletal
    - Weak muscles
    - Bone deformity
    - Bone fragility
  - Motor delays

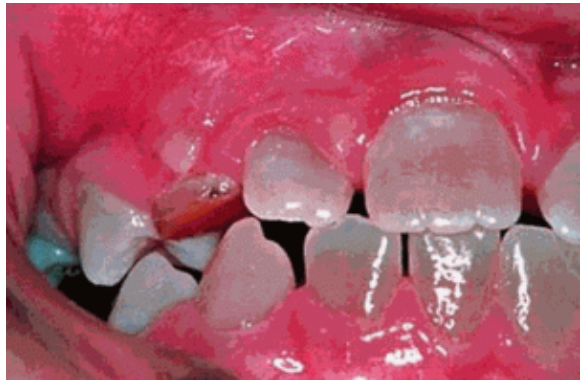
Impairments

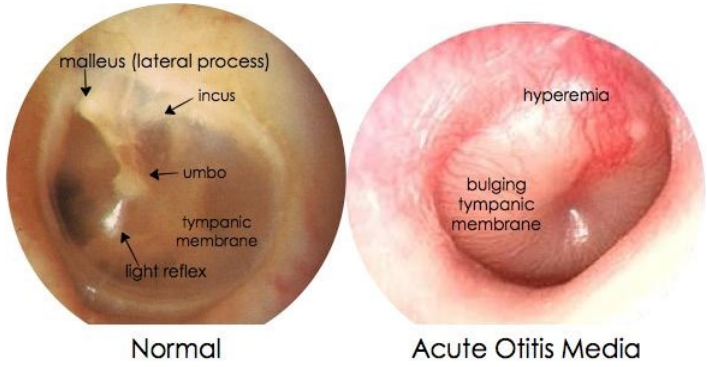




Impairments

- Left: Pulmonary hypoplasia
- Top right: Dentinogenesis imperfecta
- Bottom right: Otitis Media





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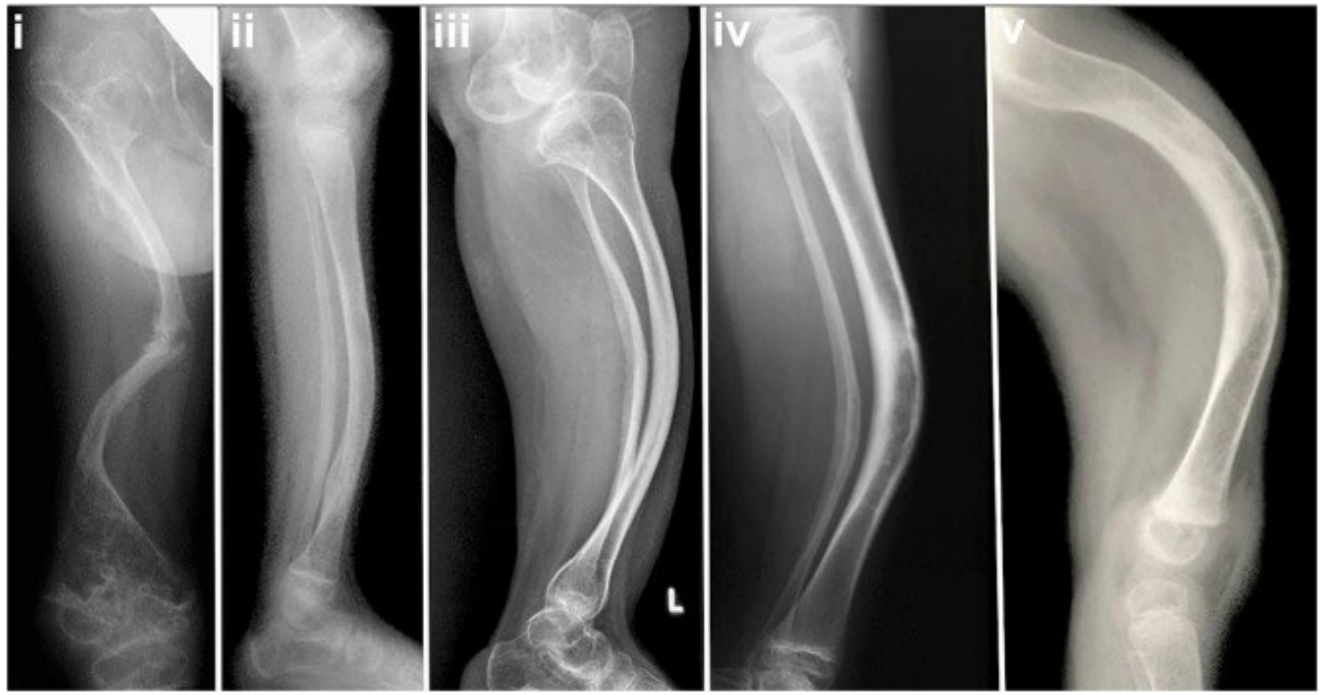
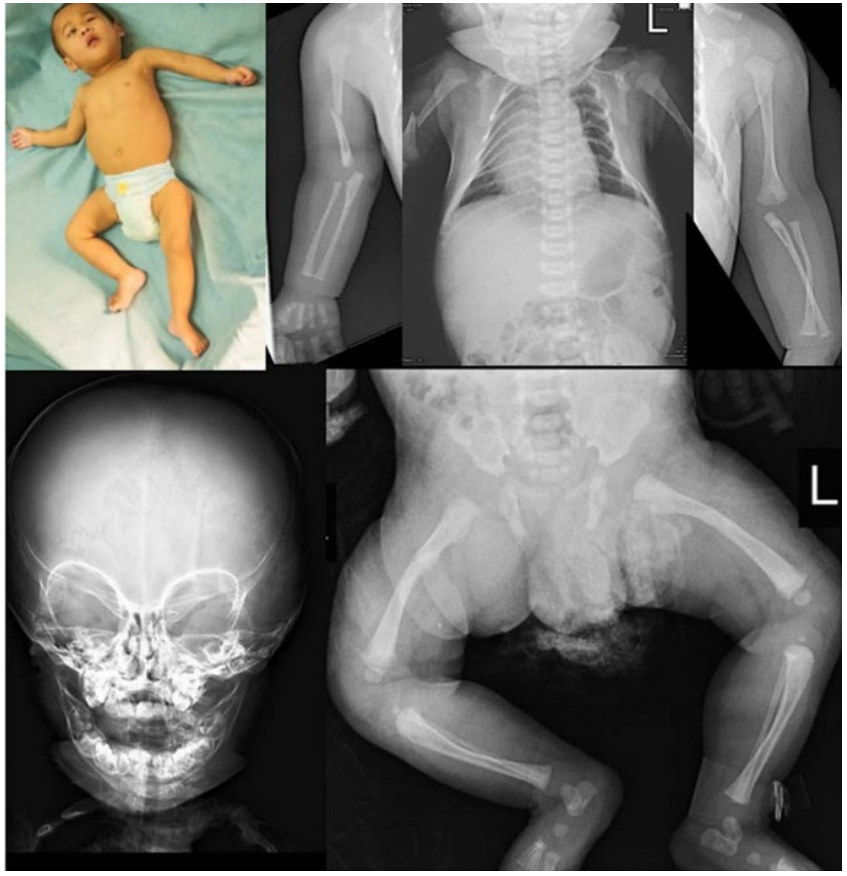


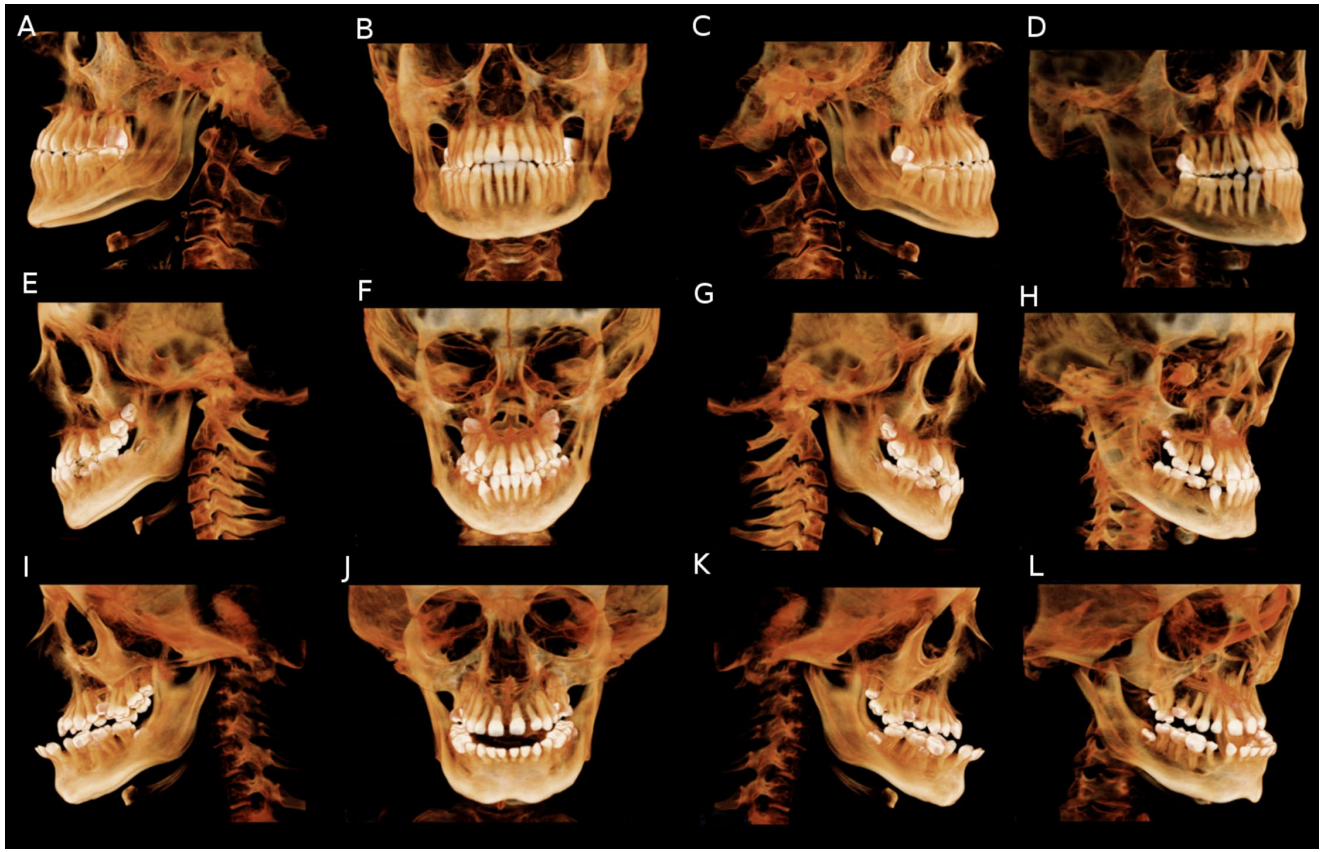
Patient Presentation

- Varies depending on OI type
  - Most common among all variants is bone fragility

Patient presentation

- Type I
  - Normal stature
  - No dentinogenesis imperfecta
  - Mild deformity
  - Vertebral fractures



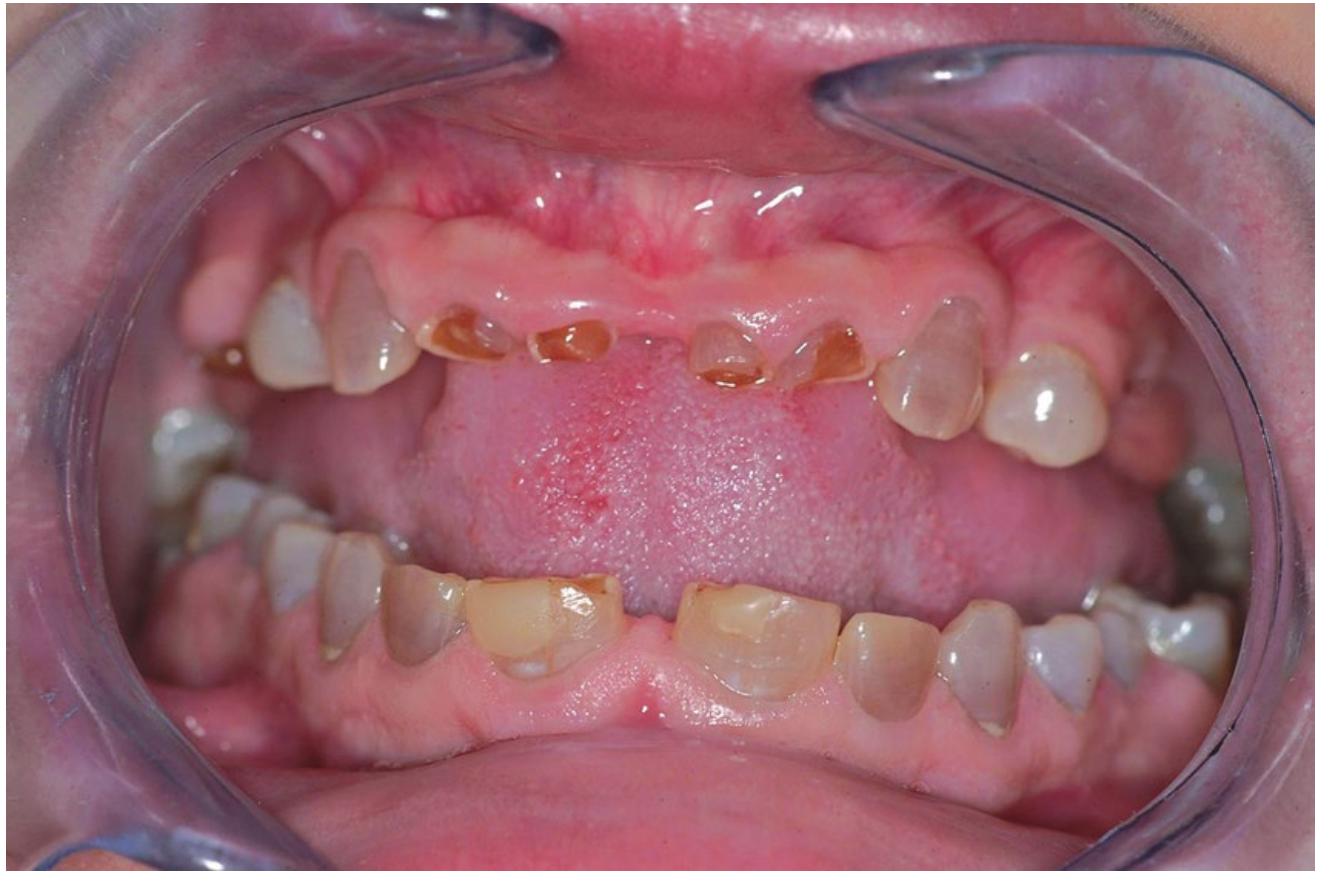


Patient Presentation

- Type II
  - Multiple rib and long bone fractures present at birth
  - Low skull bone density
  - Upper respiratory failure
  - Prenatally lethal

Patient Presentation

- Type III
  - Very short, triangular face
  - Severe bone deforming
  - Dentiogenesis imperfecta



Patient Presentation

- Type IV
  - Moderately short stature
  - Moderate bone deformity
  - Dentiogenesis imperfecta

Patient Presentation

- Type V
  - Moderately short stature
  - Moderate to severe bone deformity
  - Extreme calcification in long bones

Continuum of Osteogenesis Imperfecta

- Type 1: Most Common, least severe
- Type 2: Lethal
- Type 3: Most severe form compatible with life
- Type 4:Mild severity, comparable to type 3
- Type 5: Moderate severity, rare form

Evaluative techniques

History and development review

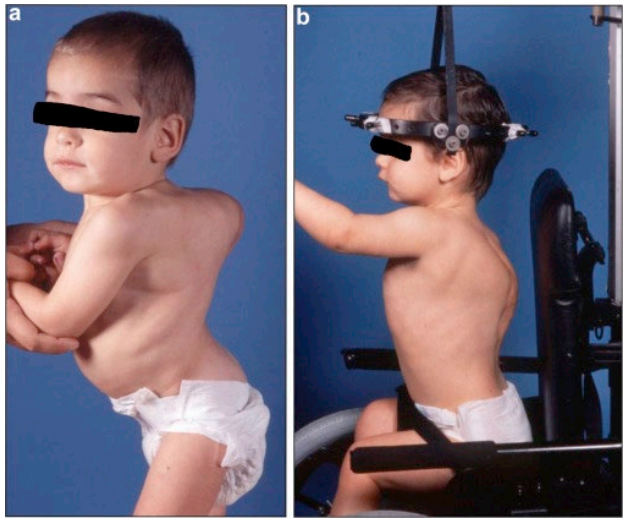
Home lifestyle review

Observe and Interact

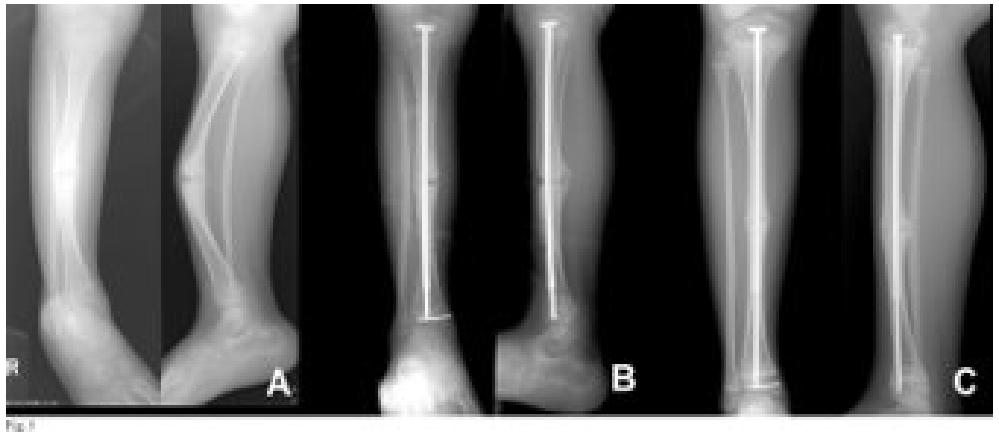
Develop a plan

Surgical Interventions

Halo Gravity Traction



Intramedullary Elongating Rods



Rehabilitation program

- Strengthen postural muscles
- Strengthen biceps, deltoid, and rotator cuff muscles
- Strengthen glute muscles
- Increasing gross and fine motor skills

References

- *Current concepts in osteogenesis imperfecta: Bone Structure ...* (n.d.). Retrieved February 1, 2023, from https://www.researchgate.net/publication/330146748_Current_concepts_in_osteogenesis_imperfecta_Bone_structure_biomechanics_and_medical_management
- Edelu, B., Ndu, I., Asinobi, I., Obu, H., & Adimora, G. (2014, March). *Osteogenesis imperfecta: A case report and review of literature*. Annals of medical and health sciences research. Retrieved November 17, 2022, from https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4083720/
- Elsevier. (2020, December 1). *Histopathology of osteogenesis imperfecta bone. Supramolecular assessment of cells and matrices in the context of woven and lamellar bone formation using light, polarization and ultrastructural microscopy*. Bone Reports. Retrieved January 31, 2023, from https://www.sciencedirect.com/science/article/pii/S2352187220304940
- *The four parts of a pediatric therapy evaluation*. Fleming Therapy Services. (2016, May 8). Retrieved November 12, 2022, from https://flemingtherapy.com/success-story/four-parts-pediatric-therapy-evaluation/
- Leotard, A., Taytard, J., Aouate, M., Boule, M., Forin, V., & Lallemant-Dudek, P. (2018, September 15). *Diagnosis, follow-up and management of sleep disordered breathing in children with osteogenesis imperfecta*. European Respiratory Society. Retrieved November 12, 2022, from https://erj.ersjournals.com/content/52/suppl_62/PA2552
- Najirad, M., Ma, M. S., Rauch, F., Sutton, V. R., Lee, B., Retrouvey, J.-M., & Esfandiari, S. (2018, October 25). *Oral health-related quality of life in children and adolescents with osteogenesis imperfecta: Cross-sectional Study - Orphanet Journal of rare diseases*. BioMed Central. Retrieved November 12, 2022, from https://ojrd.biomedcentral.com/articles/10.1186/s13023-018-0935-y
- *Osteogenesis imperfecta - statpearls - NCBI bookshelf*. (n.d.). Retrieved November 13, 2022, from https://www.ncbi.nlm.nih.gov/books/NBK536957/
- Rauch, F., & Glorieux, F. H. (2004, April 23). *Osteogenesis imperfecta*. The Lancet. Retrieved November 12, 2022, from https://www.sciencedirect.com/science/article/pii/S0140673604160510?via%3Dihub#cesec30
- Ren, J., Xu, X., Jian, X., & Wang, J. (2014, June). *Osteogenesis imperfecta type I: A case report*. Experimental and therapeutic medicine. Retrieved November 17, 2022, from https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4043568/
- Saunders, F. R., Gregory, J. S., Pavlova, A. V., Muthuri, S. G., Hardy, R. J., Martin, K. R., Barr, R. J., Adams, J. E., Kuh, D., Aspden, R. M., Cooper, R., & Ireland, A. (2020, December). *Motor development in infancy and spine shape in early old age: Findings from a British Birth Cohort Study*. Journal of orthopaedic research : official publication of the Orthopaedic Research Society. Retrieved November 12, 2022, from https://www.ncbi.nlm.nih.gov/pmc/articles/PMC8641380/
- Swinnen, F. K. R., De Leenheer, E. M. R., Coucke, P. J., Cremers, C. W., & Dhooge, I. J. (2009). Audiometric, surgical, and genetic findings in 15 ears of patients with osteogenesis imperfecta. *The Laryngoscope*, *119*(6), 1171–1179. https://doi.org/10.1002/lary.20155
- U.S. Department of Health and Human Services. (n.d.). *Osteogenesis imperfecta (OI)*. Eunice Kennedy Shriver National Institute of Child Health and Human Development. Retrieved November 12, 2022, from https://www.nichd.nih.gov/health/topics/osteogenesisimp
- U.S. Department of Health and Human Services. (n.d.). *What are the treatments for osteogenesis imperfecta (OI)?* Eunice Kennedy Shriver National Institute of Child Health and Human Development. Retrieved November 12, 2022, from https://www.nichd.nih.gov/health/topics/osteogenesisimp/conditioninfo/treatments#therapy
- U.S. Department of Health and Human Services. (n.d.). *What causes osteogenesis imperfecta (OI)?* Eunice Kennedy Shriver National Institute of Child Health and Human Development. Retrieved November 12, 2022, from https://www.nichd.nih.gov/health/topics/osteogenesisimp/conditioninfo/causes