OSTEOGENESIS IMPERFECTA

C12

Objectives

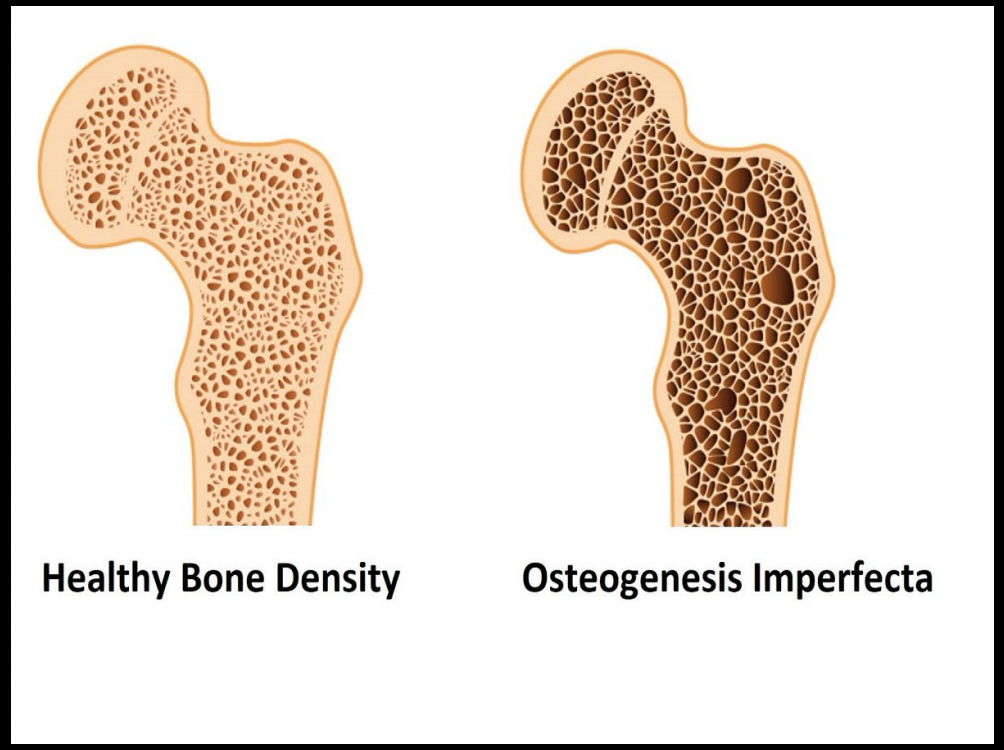
- Osteogenesis Imperfecta (OI) and its underlying genetic basis.
- The pathogenesis and classification of Osteogenesis Imperfecta.
- The common signs, symptoms, and possible complications associated with Osteogenesis Imperfecta.
- Explain the diagnostic process and factors influencing the prognosis of Osteogenesis Imperfecta.
- The role of medical imaging professionals in the evaluation of Osteogenesis Imperfecta and typical imaging manifestations.
- The various surgical, medical, and supportive treatment strategies used in the management of Osteogenesis Imperfecta.

https://www.britishmuseum.org/collection/object/Y_EA41603

Background on Osteogenesis Imperfecta

- Osteogenesis Imperfecta has been dated back to 1000 BC in an Egyptian Mummy.
- Osteogenesis Imperfecta has been identified as the medical condition that Ivan the Boneless had who lived in 9th century Denmark.
  - He was carried into battle on a shield because he could not walk on his soft legs.
- In the 1970s in Australia, Dr. David Sillence and his team created the system of categorization using the “types” that are currently being used today.

What is Osteogenesis Imperfecta?

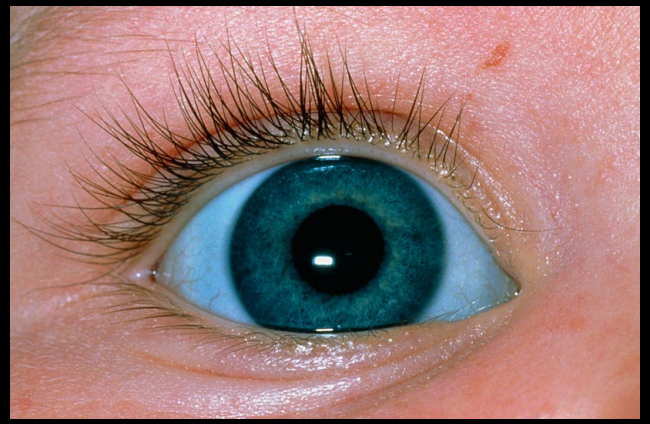


https://www.thehealthsite.com/diseases-conditions/osteogenesis-imperfecta/

- Also known as OI or brittle bone disease.
- It is a genetic bone disorder that causes fragile bones to break easily.
- It is a rare genetic condition that affects 1 in 10,000 to 20,000 births.
  - The range is wide because most people born with a mild form go undiagnosed.
- About 25% of children born with OI have no family history of the condition.
- There is no way to prevent the disease.
- There are at least eight different types of OI.
- OI is a spectrum and not everyone will experience the same symptoms even if they have the same type.

How does Osteogenesis Imperfecta develop?

- Usually, it is caused by autosomal dominant mutations in the type 1 collagen genes, COL1A1 and COL1A2.
  - 80-90% of OI cases are caused by these mutations.
- Can also be due to autosomal recessive mutations in any of six genes (SERPINF1, CRTAP, LEPRE1, PPIB, SERPINH1, and FKBP10).
- In most cases people with OI are born with it but sometimes the gene mutation is not inherited and happens after conception.
- This collagen is used to make:
  - bones strong
  - build tendons
  - build ligaments
  - make the whites of the eyes (sclera)

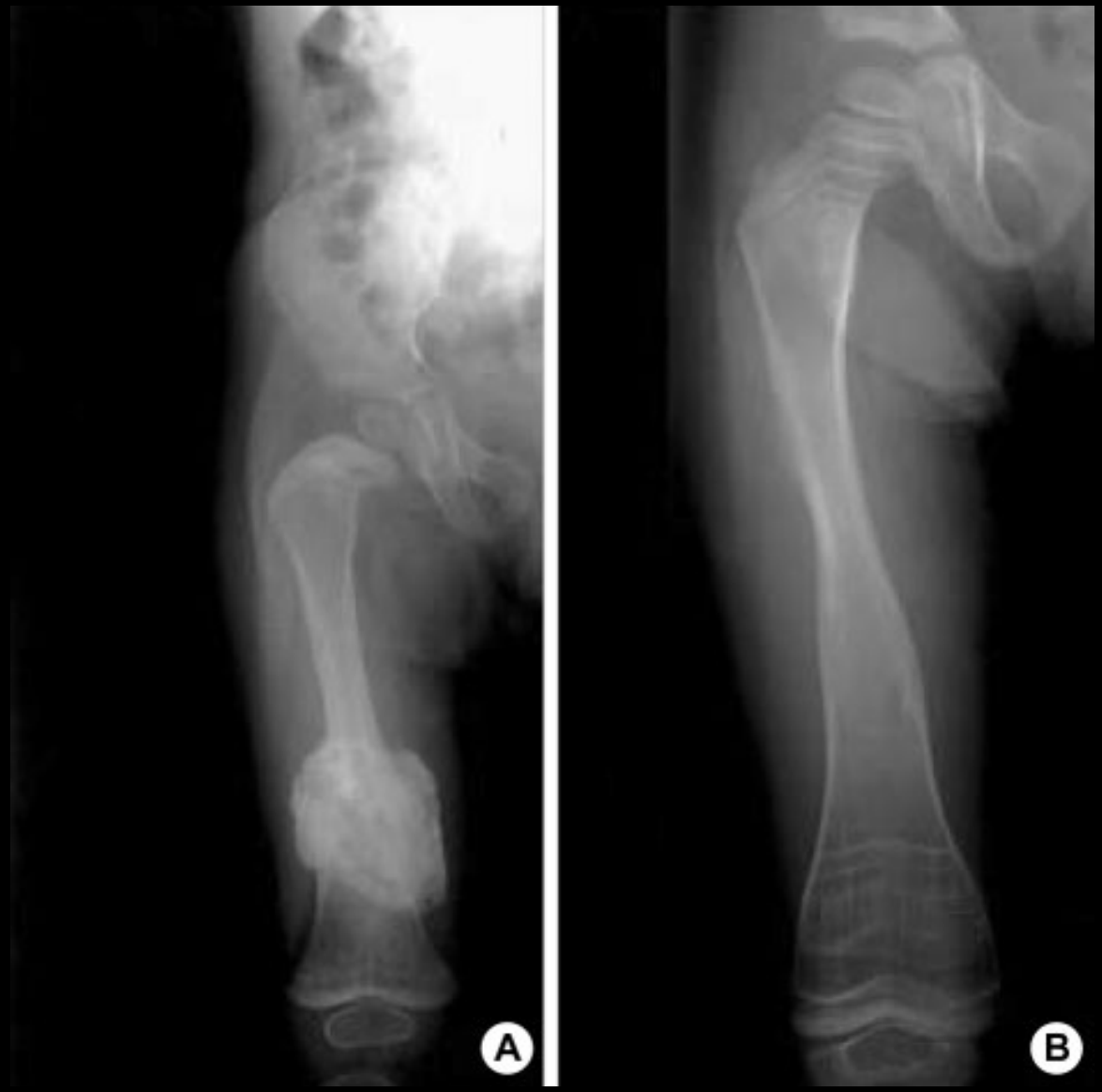


https://physicians.dukehealth.org/articles/diagnose-%E2%80%93-pediatrics-issue-92-intensely-blue-colored-sclera-seen-here-most-commonly

https://cbsupplements.com/cc/what-are-5-types-of-collagen/

What types of Osteogenesis Imperfecta is there?

- **Type I**: This is the most common form of OI. It has milder symptoms, and most of the broken bones occur before puberty and does not cause any bone deformities. It is also known as the classic non-deforming Osteogenesis Imperfect with the blue sclerae.
- **Type II**: This is the most serious form of OI. It has severe complications including underdeveloped lungs, bone deformities and broken bones before birth. Babies with this type die at birth or shortly after. It is also called the perinatally lethal Osteogenesis Imperfecta.
- **Type III**: This is the most severe form of OI that can survive birth. It causes severe bone deformities leading to physical disabilities. Bones are often broken at birth. It is also known as progressively deforming Osteogenesis Imperfecta.
- **Type IV**: more severe than type I but not as severe as type 3. It causes moderate bone deformities, causing bones to be more fragile than people without OI. It is also known as common variable Osteogenesis Imperfecta with normal sclerae.



https://www.researchgate.net/figure/Hypertrophic-callus-formation-six-weeks-after-femoral-shaft-fracture-at-age-9-month-A_fig4_6894888

What types of Osteogenesis Imperfecta is there?

Type V: Similar to type IV. It is common it see enlarged thickened areas where large bones are fractured/broken.

Type VI: Similar to type IV. Very rare form of Osteogenesis Imperfecta.

Type VII: Similar to type IV or II. It is common it see shorter than normal height, upper arms and thigh bones.

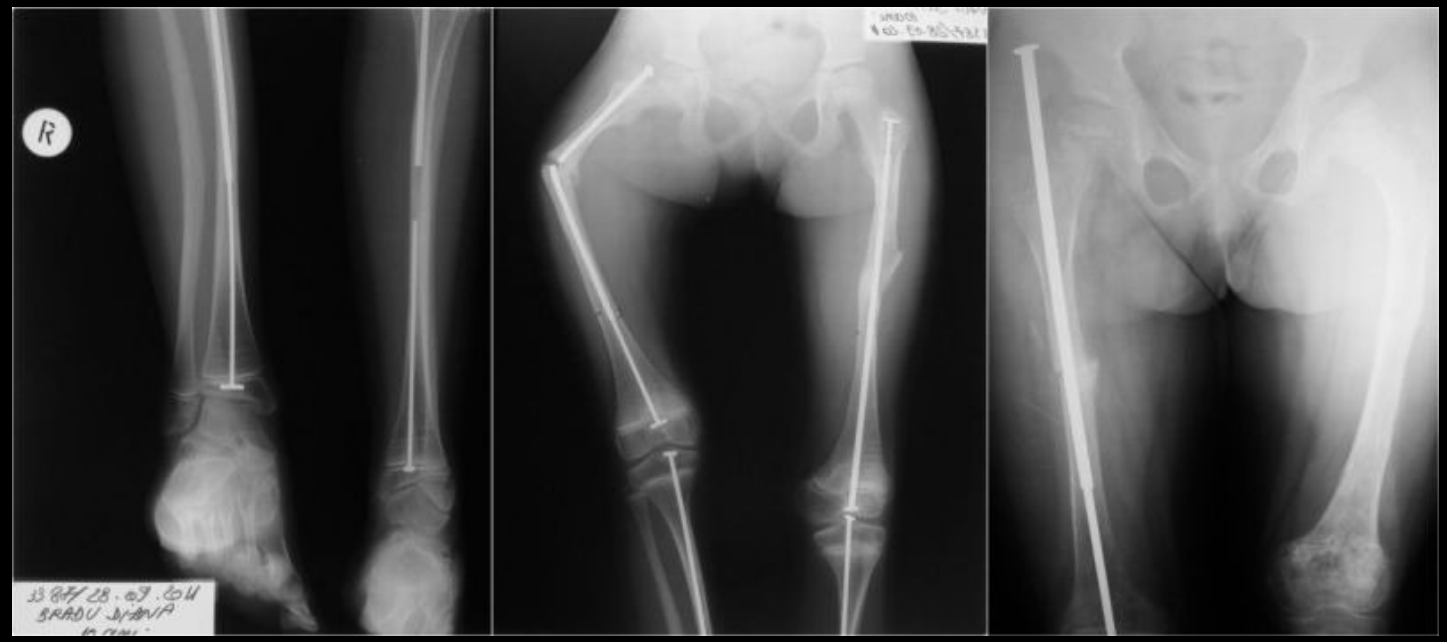
Type VIII: Similar to types II and III. It is common to see very soft bones and severe growth problems.

Common signs and Symptoms

- Symptoms range from mild to severe and can vary from person to person
- Malformed or bowing of long bones
- Small Stature
- Skin that bruises easily
- Loose joints
- Weak muscles
- Whites of the eyes (sclera) that look blue, purple, or gray
- A curved spine
- A barrel shaped rib cage
- Compression of the vertebrae
- Brittle, misshaped, or discolored teeth
- Teeth that are not aligned properly (Malocclusion)
- Hearing Loss
- Difficulty breathing
- Deformed hip joint
- Joints that stay permanently bent or in a straightened position
- A triangle shaped face
- Scoliosis

Complications of Osteogenesis Imperfecta

- Heart Problems – common in type II
- Hearing loss – common in types I and III
- Frequent Pneumonia
- Respiratory Issues
- Spinal Cord Issues
- Brain Stem Problems
- Permanent Deformities
- Kidney Stones
- Eye Conditions like vision loss
- Bending, disengagement, fracture of the rod, or possible migration

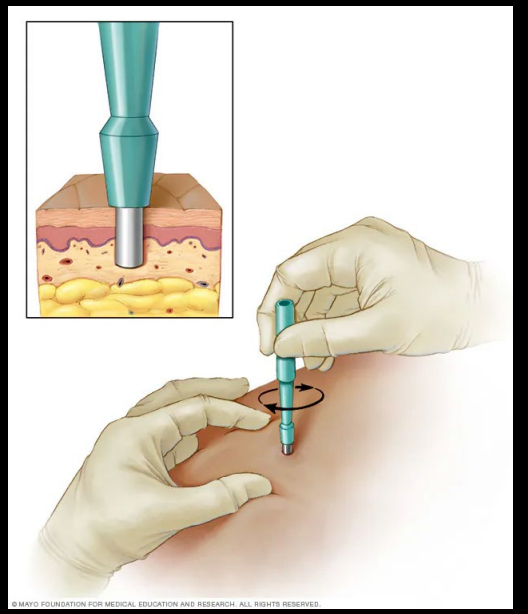


https://pmc.ncbi.nlm.nih.gov/articles/PMC3725451/

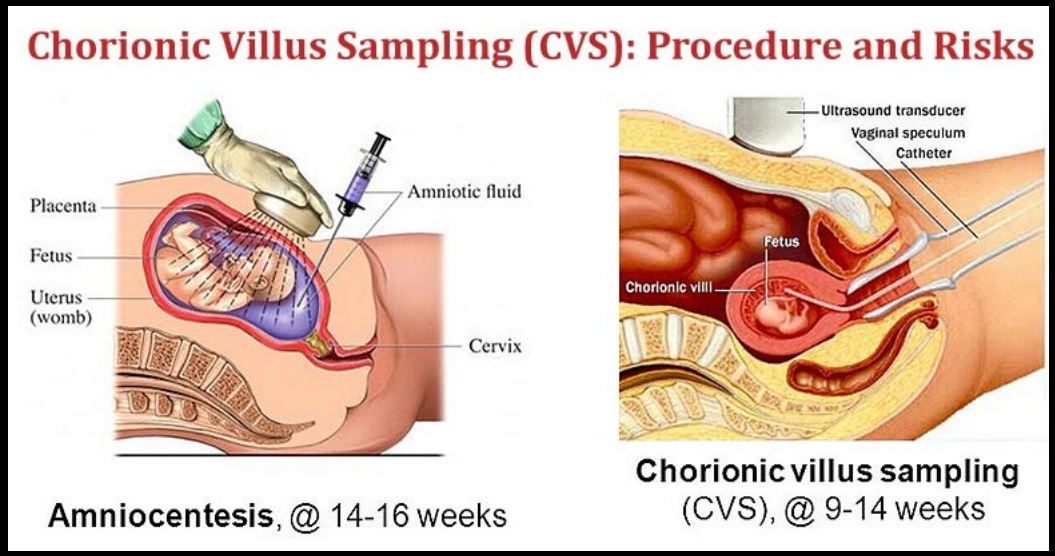
How is Osteogenesis Imperfecta diagnosed?

- The first sign of Osteogenesis Imperfecta are broken bones that occur with little or no force.
- Your doctor may ask about family and medical history.
- Your doctor may complete a physical exam.
- A blood test to identify the type of Osteogenesis Imperfecta and the change in the inherited gene.
- A negative genetic test does not rule out Osteogenesis Imperfecta.
- X-rays
- Ultrasound
- Amniocentesis or Chorionic Villus Sampling (CVS)
- Bone Density Test
- Bone Biopsy
- A definitive diagnosis may be made using a skin punch biopsy.

https://www.mayoclinic.org/tests-procedures/skin-biopsy/about/pac-20384634



https://www.londonspectra.com/2020/03/invasive-amniocentesis-cvs-prenatal.html



What is Osteogenesis Imperfecta prognosis?

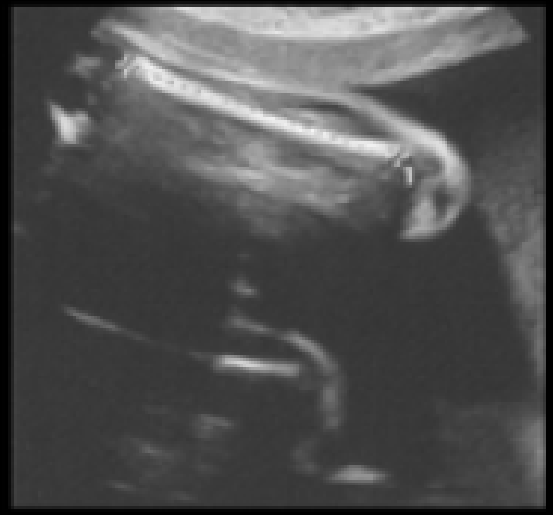
- The prognosis all depends on the type of Osteogenesis Imperfecta they have.
- In type I people typically live a normal lifespan.
- In type II it typically leads to death within the first year of life.
- In type III people have typically have lots of fractures and severe bone deformities. Many people are wheelchair bound and often have a shortened lifespan.
- In type IV people often need braces or crutches to walk, and they have a normal or near normal lifespan.
- The other types of Osteogenesis Imperfecta occur very rarely and are considered subtypes of type IV.

Role as a medical imaging professional

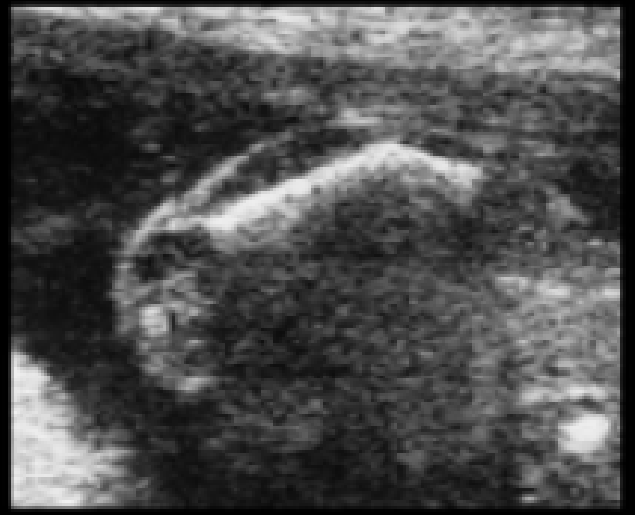
- Making sure to avoid making any further fractures.
- Never lift a child with Osteogenesis Imperfecta by the armpits.
- Do not be afraid to touch or hold a child with Osteogenesis Imperfecta.
- If they can not move, critically thinking to get the images needed.
- Making sure to use positioning aids like sponges to help sit or lie down.
- Decrease the technique on imaging exams.

How Osteogenesis Imperfecta manifests itself in Ultrasound

- Even though it is an established tool for the prenatal diagnosis it does not have a role postnatally.
  - Besides the exception of echocardiography to monitor cardiac abnormalities.
- Ultrasound is theoretically capable of diagnosing bone fractures.
- Ultrasound has been reported to have greater sensitivity than x-rays in the detection of rib fractures.



Normal fetal thigh bone

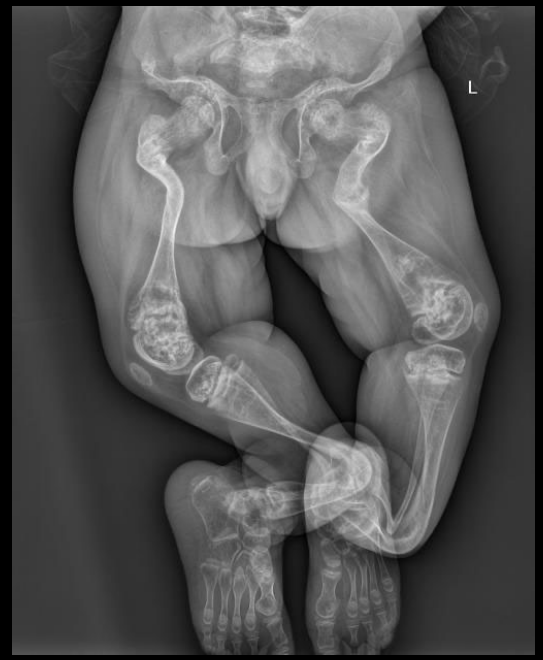


Thigh bone of a baby with osteogenesis imperfecta

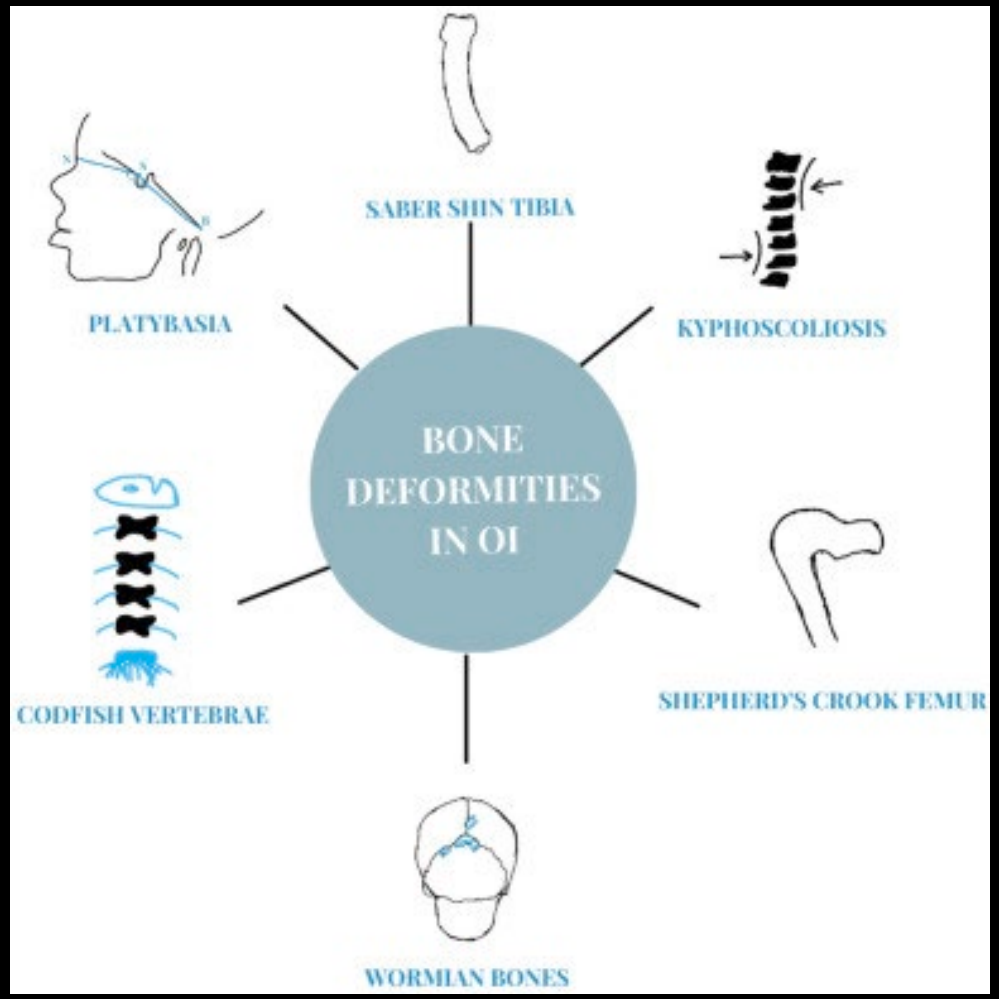
https://www.ucl.ac.uk/womens-health/utero-stem-cell-transplantation

How Osteogenesis Imperfecta manifests itself in diagnostic radiology

- A standard skeletal survey protocol is preferable for a precise diagnosis.
- Main features of OI include: osteopenia/osteoporosis, bone fractures, and bone deformities (bowing of long bones).
  - Not always present in mild forms and it may be hard to diagnosis based on radiographic features.



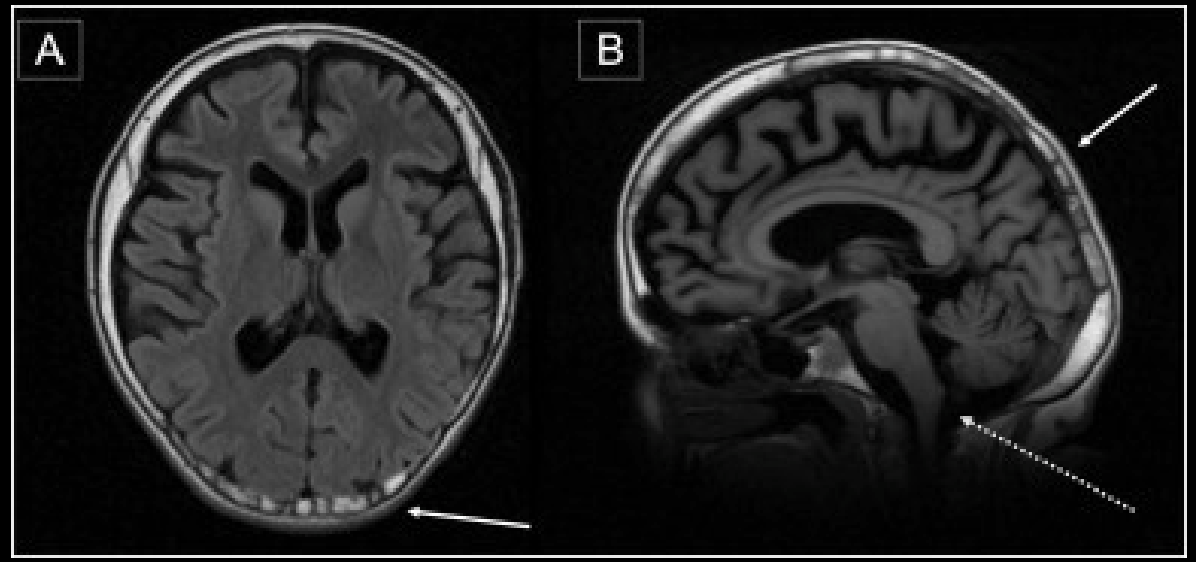
https://radiopaedia.org/articles/osteogenesis-imperfecta-1



https://www.sciencedirect.com/science/article/pii/S1769721224000181

How Osteogenesis Imperfecta manifests itself in CT and MRI

- CT and MRI and not routinely used for diagnosis but can help in the distinction of hyperplastic callus from osteosarcoma.
- CT and MRI can improve the characterization of basilar impression.
- CT and MRI of the temporal bones can assist in confirming the diagnosis.
- CT and MRI play a role in pre-op planning.
- CT is accurate in evaluating osseous structures.
- MRI is indicated in patients with abnormal lateral skull x-rays or neurological symptoms.



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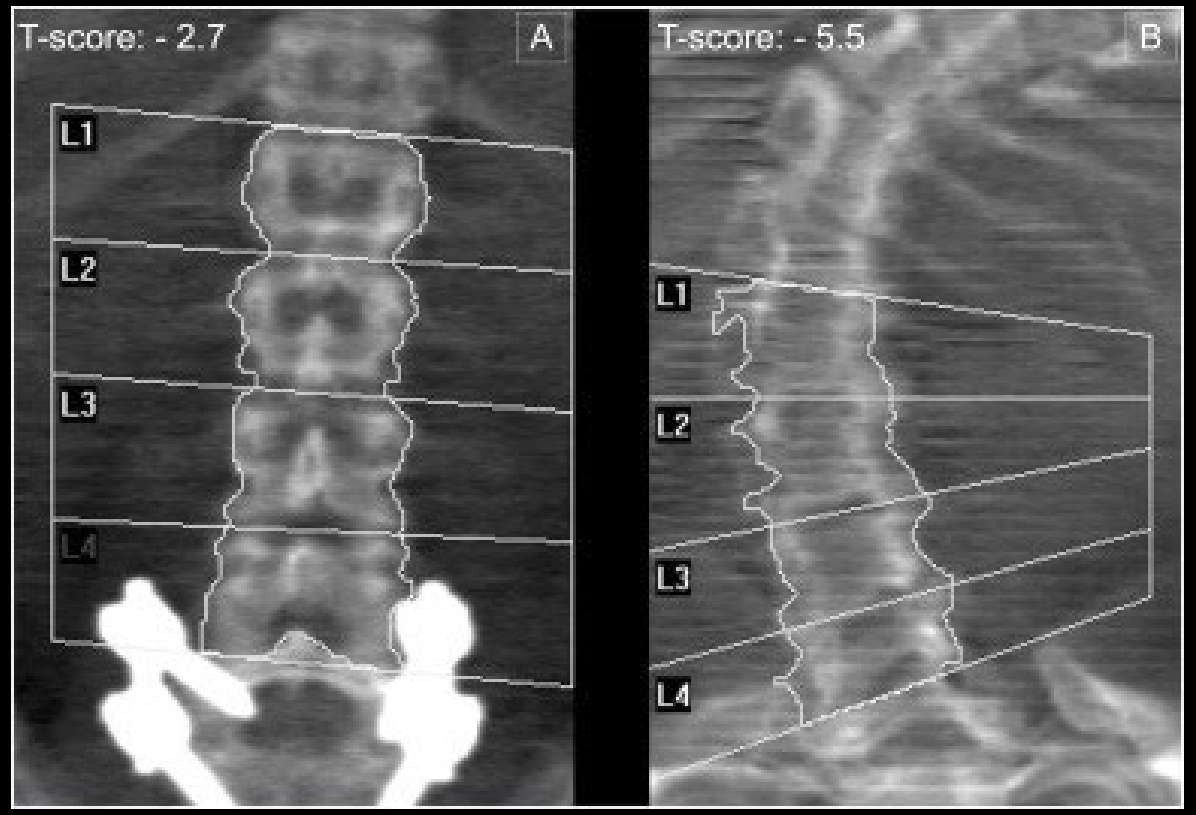
How Osteogenesis Imperfecta manifests itself in DXA

- Dual-Energy X-ray Absorptiometry
- DXA is the preferred method to evaluated bone mineral density.
  - Since a main symptom is osteoporosis/osteopenia
- The usual sites for adults are the PA lumbar spine and hip.
- Most centers for adults perform DXA scans every 2 years but this interval can be shortened.
- DXA should be performed before starting therapy to make sure the changes are real and not machine errors.



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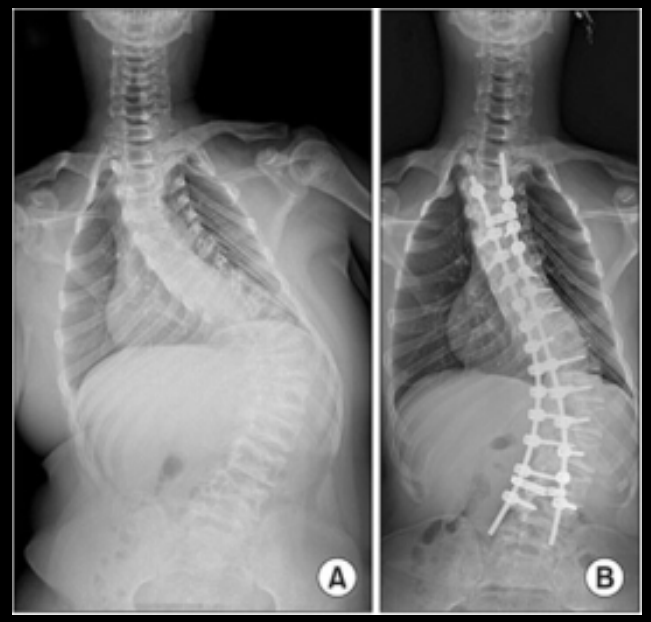
Various treatments for Osteogenesis Imperfecta

- There is no cure yet for this disease, but certain treatments can help reduce the pain and complications from OI.
- There are both surgical and non-surgical treatment options for people with OI.
- Bisphosphonate medicines – which help reduce loss of bone mass; by slowing down the loss of existing bone.
- Vitamin D supplements – necessary for the body to absorb calcium
- Fracture care
- Physical and Occupational Therapy
- Bracing
- Surgical procedures such as rodding in the long bones or to help with hearing loss.
- Hearing aids
- Dental procedures such as crowns, capping teeth, or braces

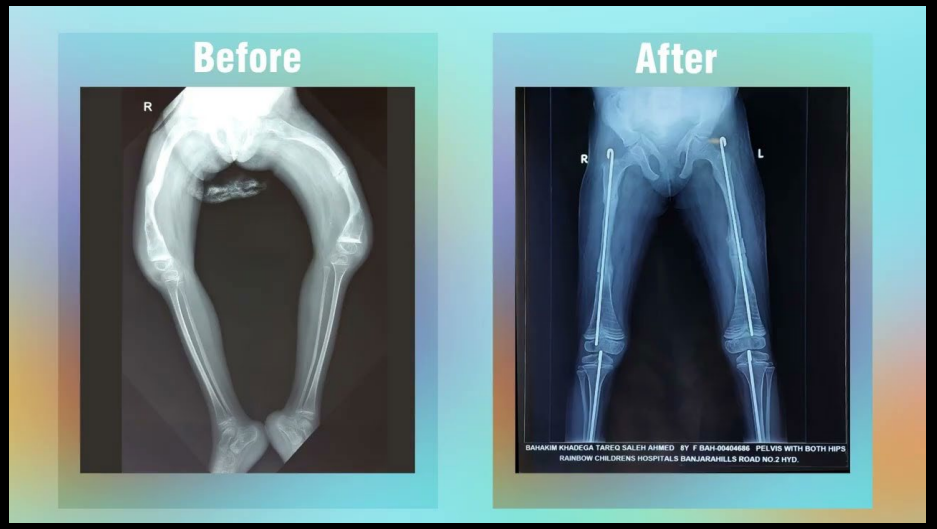
Surgery options for Osteogenesis Imperfecta

- Based on the Sofield-Millar technique
- Rodding surgery to prevent repeated fractures in the long bones of the arms and legs.
- Two main types of intramedullary rods:
  - Telescopic or non-telescopic
- Correct the abnormal curvature of the spine caused by scoliosis or kyphosis.
- Bone transplant
- Osteotomies



https://ecios.org/DOIx.php?id=10.4055/cios20060

https://www.youtube.com/watch?v=3LJXyPDZ_3c



Improving bone health with Osteogenesis Imperfecta

- Eat foods that are rich in calcium and vitamin D.
- Exercise such as swimming and walking to help build muscle and bone strength.
- Limiting alcohol and caffeine intake.
- Avoid smoking and avoiding secondhand smoke.
- Avoid drugs that have steroids in them.
- Weight management
- Avoiding infections

Conclusion

- Osteogenesis Imperfecta is a genetic bone disorder that has been dated back to 1000 BC and is known as OI and brittle bone disease.
- It can be a dominant or recessive mutation in type 1 collagen that causes bones to be fragile and break easily, it can affect 1 in 10,000 to 20,000 births.
- Dr. David Sillenece and his team created the “types” that are being used today. There are at least 8 different types being used today.
- Type I is the most common, Type II is the most serious, and Type III is the most severe. Each of the different types have different prognosis and variable symptoms.
- Some symptoms include weak muscles, loose joints, small stature, scoliosis, and the sclera that looks blue, purple, or even gray.
- Through the eyes of medical imaging we can see how the different modalities can compliment each other.
- There is not a cure for OI, however there are treatments to help a patient diagnosed with OI. Treatment options include a range of things from supplements all the way to corrective surgery.
- If you have a patient that has OI make sure to not look scared to touch or hold the patient, use positioning aids to make sure to get the best diagnostic images, and decreasing the technique.

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