Osteogenesis imperfecta: clinical features and imaging overview

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Learning objectives

Review the classifications, genetic inheritance, clinical and imaging features of osteogenesis imperfecta.
**Background**

Osteogenesis imperfecta (OI) is a rare genetic disorder, autosomal recessive, affecting the musculoskeletal system, that leads to multiple bone deformities, increased bone fragility and fractures due to deficits in type I collagen, the main pathophysiologic effect of the disease. The bones break easily by simple trauma, or spontaneous fractures can occur without any apparent cause. Occurs every 1:60,000 births.

It has a wide clinical expression from mild to severe, including death during neonatal and fetal periods. It is most obvious in its effect on bones, but also involves the body's ligaments, tendons, fascia, eyes, skin, teeth and ears.

It is clinically characterized by frequent fractures sometimes with no history of trauma. Patients have significant osteopenia, capsular-ligamentous laxity, bluesclera and pre-senile onset hearing impairment.

The diagnosis is made from clinical, genetic and radiographic features.

The main consequence of osteogenesis imperfecta corresponds to an excessive fragility of the bones, which allows the emergence of fractures before minimal trauma or even spontaneously.

In more severe forms of osteogenesis imperfecta, these fractures begin to occur during fetal life and ultimately cause death of the baby even before birth or during their first weeks of life.

In milder forms, although the child can survive, is often affected by numerous fractures during the time of development, which leads to numerous defects to the skeleton and a global growth defect with a final size lower than normal.

In the simplest cases, less severe, sometimes only be detected in adolescence.

**Clinical features:**

- The clinical features vary according to each type, being the most prominent is bone fragility resulting in sagging and broken bones. It can still be seen, osteoporosis, blue sclera, abnormal dentition (Dentinogenesis Imperfecta)
and osteosclerosis early. Only two of these clinical manifestations must be present to suggest the diagnosis.

- Other clinical manifestations are described: laxity of joints, thin skin, diaphoresis with episodic abnormal regulation of temperature, light bruises with a tendency to bleeding, scarring hyperplastic and early vascular calcifications.

It is important to distinguish Osteogenesis imperfecta from child abuse in order to protect an abused child or to avoid an improper accusation of child abuse in a child with obvious OI.

There are 4 major types of OI, ranging from mild to severe. The various forms of Osteogenesis Imperfecta differ widely with respect to gravity.

- Type I is the mildest form of the disease and the most common, affecting about 80% of cases, without compromising the final height of the individual. Normal stature, blue sclerae permanent, relatively mild osteopenia, which determines the low frequency of fractures, deafness and around 30% of cases. It is subclassified into IA (normal teeth) or IB (imperfect dentition).
- Type II is the most severe form of the disease and is usually autosomal recessive, presenting with fractures and bone deformities intrauterine as well as undeveloped and curved limbs and severe bone fragility. The newborn, in general, is premature or small for gestational age and the affected individual, most often develops in the perinatal death or dies in the first days to weeks of life due to respiratory complications.
- Type III is the severe nonlethal. It is autosomal recessive. Recurrent fractures gradually lead to bone deformities in the limbs and spine, causing short stature, besides presenting with dental development imperfect. Despite occur deformity of long bones, joint laxity and scoliosis, ambulation is possible.
- Type IV is also compatible with life, with clinical characteristics similar to type I but with sclera normal. This type is associated with both gene interaction dominant and recessive.

Cardiovascular abnormalities and nephrolithiasis may be important extraskeletal manifestations of childhood OI.

Although there is great variability in the manifestations of Osteogenesis Imperfecta, confirmation of the diagnosis is mainly clinical history and physical examination.

Should be investigated family history, reports of fractures with trauma mechanism that would not be justified in people with musculoskeletal structure normal, and combination of physical characteristics such as blue sclera, dentinogenesis imperfecta, scoliosis, short stature and angular deformities, mainly lower limbs.
Although there are signs suggestive, it is very important to consider that there is still no clinical signs or complementary test pathognomonic to confirm the diagnosis, and thus should be warned that, especially in younger patients, suspected cases should be differential with other diseases bone or even with the syndrome child victim of physical abuse.
Osteogenesis imperfecta--whose name implies "imperfect birth of bone"--is one of these inherited fragile bone syndromes.

Radiographs, bone scans and other imaging tools are essential in the initial diagnosis, assessment of fracture risk, and planning and tracking of treatment.

Radiographically bone changes are found, which vary in amount and quality depending on the severity of the disease. There is usually a decrease in trabecular bone normal, a decrease of cortical thickness and angular deformities (table 1).

- In the radiographic evaluation of Type I Osteogenesis Imperfecta, there is generalized osteopenia and angulation deformities resulting from previous fractures at sites(Fig 2,3)
- With a careful radiographic study can distinguish three subtypes of the type II, being the II-lethal before birth or in the neonatal period. The radiographic observed: small chest with ribs shortened, thickened, with wavy contours (standard accounts); femur wide and rectangular with wavy margins, curved shaft, and tibia angled (FIGs. 3, 4, 5)
- Type III: In the radiographic evaluation was observed: Dentinogenesis Imperfecta, and progressive dysfunction and repair metaphysis, appearance similar to popcorn.
- Type IV: Can be divided into at least five subtypes, as the use of clinical and histomorphometric criteria. The radiographic observed: mild bone deformity and dentinogenesis imperfecta.

It was described a classification based on radiographic severity of impairment of patients with osteogenesis imperfecta (Hanscom -1992):

- Type A: mild form of Osteogenesis Imperfecta, maintains vertebral contours;
- Type B: Deformity in the long bones with curving and cortical wide. The pelvis preserves the bony contours
- Type C: bowing of long bones with thin cortical and acetabular protrusion developed between five and ten years old;
- Type D: the same characteristics of type C associated with cysts in the metaphyseal areas of long bones around 5 years old. The epiphyses close early around the age of 15;
- Type E: deformities result in extreme disability, and kyphoscoliosis is early and very serious. In the long bones, cortical hardly visualize on radiographs
- Type F: complete collapse of the ribs, incompatible with survival.
• The largest number of fractures occur until the age of three, so we believe the hypothesis that this high rate is due to lack of education of health professionals and appropriate guidance to parents to deal with children with Osteogenesis Imperfecta. There is also difficulty and lack of preparation for handling the child, from birth in the delivery room during procedures, X-rays and other tests performed during hospitalization.

• There are cases where the diagnosis was made only when there was suspicion of physicians to the parents, referring mistreatment. The Osteogenesis Imperfecta was diagnosed only after several radiographs.

• The skeletal deformities are common, predominantly in the lower limbs and are due to fractures consolidated viciously. These deformities are common in the convexity of the long bones of the lower limbs, which may coexist with fractures in various stages of consolidation in the patient. In more severe forms are called calcifications in the "popcorn" in the metaphyseal region of long bones and rarely can we find more cases of acetabular protrusion (FIG 8, 9, 10)

• Bone density is decreased in patients with Osteogenesis Imperfecta, and this can be investigated by radiographic methods, by bone densitometry or tomography for lumbar spine (FIG 11, 12). Bone densitometry is a more effective method than radiography, with respect to objectively quantify the reduction of bone mass. Bone densitometry also lends itself to the monitoring of patients who are clinically treated with bisphosphonates or other drugs.

• The prenatal ultrasound plays a role in the diagnosis of osteogenesis imperfect. This condition is one of the most common skeletal dysplasia detected by prenatal ultrasonography. In the majority of cases are found incidentally on ultrasound examinations performed for other reasons. Typical incidental findings include fractures, decreased ossification or calvaria that are compressible to the transducer. In most cases of osteogenesis imperfecta that are recognized in this way are type II, and patients have no family history of disease.

This presentation was based on a retrospective study of 21 patients with confirmed diagnosis of osteogenesis imperfecta, treated and followed at the Fernandes Figueira Institute/Oswaldo Cruz Foundation (FIOCRUZ), Rio de Janeiro, Brazil.

The group was composed of 13 female patients (61.9%) and 08 males (38.1%), ranging in age from two months to sixteen years.

The radiographs were analyzed for the presence of deformities, femoral cortical thickness and density of the macroscopic observation of trabecular bone in the proximal femur medullary.
• The main radiographic findings were: bone deformity, osteopenia, wormian bones, fractures, growth arrest lines, platispondyia, exuberant callus, kyphoscoliosis, aspect biconcave vertebrae and acetabular dysplasia.
• At lower frequencies were found: flattening of the iliac wing, sella turcica in J, triangular face and brachycephaly.

• Osteogenesis imperfecta is known to have various spine lesions as complications.
• Basilar impression, atlantoaxial dislocation, and syringomyelia can be demonstrated by different imaging methods: plain radiography, tomography, three-dimensional computed tomography, and magnetic resonance imaging.
• Basilar impression assessed by either plain lateral skull radiograph or computerized tomography sagittal reconstruction of the craniocervical junction is a common finding occurring in 25% of subjects with osteogenesis imperfect. It appears to occur with highest frequency in a group of subjects with OI type IV B, i.e. patients with mild/moderate liability to fractures, normal sclerae but dentinogenesis imperfecta.
• Basilar impression and hydrocephalus complicating osteogenesis imperfecta is usually treated by anterior transoral decompression and posterior fixation.

• Fetal MRI proves to be superior to conventional fetography in the evaluation of the skeletal findings. The high soft tissue resolution, the large field of view and the multiplanar imaging make it possible to study the non-ossified fetal skeleton in detail. Compared to prenatal ultrasonography, fetal MRI gives important additional information about the expected lung functionality by estimating fetal lung volume and signal intensity and rules out possible additional abnormalities of major fetal organs.
**Fig. 1:** TABLE 1: Osteogenesis Imperfecta: Radiographic findings

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Fig. 2: Shortened upper limbs, with multiple fractures

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Fig. 3: Upper limb bones curved with fractures.

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Fig. 4: Chest radiograph demonstrating ribs with reduced thickness.

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**Fig. 5:** Radiography of the upper limbs with signs of osteoporosis, short and curved, with fractured left humerus.

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Fig. 6: Skull radiography in the anteroposterior, cortical bone with reduced thickness and the presence of wormian bones.

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**Fig. 7:** Radiography of the skull in lateral views, with the presence of wormian bones, tapered cortical and craniofacial disproportion

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Fig. 8: Radiograph of the pelvis showing signs of acetabular protrusion. Right femoral diaphyseal fracture

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**Fig. 9:** Radiography presenting with signs of osteoporosis, lower bones short and curved and enlargement of distal femoral and proximal tibial diaphysis.

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Fig. 10: Curved femur with fracture in the middle third and widening of the distal diaphysis.

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**Fig. 11:** Panoramic radiography of the spine showing vertebral body height reduced

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Fig. 12: Multiple collapses of the vertebral bodies with signs of osteoporosis.

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Conclusion

There are 4 major types of osteogenesis imperfecta, ranging from mild to severe. The diagnosis is made from clinical, genetic and radiographic features.

Radiographs and other imaging tools are essential in the initial diagnosis, assessment of fracture risk, and planning and tracking of treatment.

A broad general knowledge of the clinical and genetic aspects of the disease, as well as the imaging features of Osteogenesis imperfecta, is required for radiologists to knowledgeably provide the proper diagnosis and to participate responsibly in a team approach with geneticists, clinicians, lawyers and child protection services.
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