Imaging spectrum of osteogenesis imperfecta – Pictorial essay

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Abstract

Osteogenesis imperfecta (OI) is a rare genetic disorder caused by the disturbance in the synthesis of Type I Collagen and is divided into several types. Severe form of OI, mainly type II can be diagnosed in utero by ultrasonography during the second trimester of pregnancy. All the other types viz. Type I to Type VIII are diagnosed by conventional radiography. Many authors have classified these into several types. This essay is not meant for a comprehensive review of these types. The main features include fractures, gracile bones, kyphoscoliosis, deformities, and wormian bones in skull. These radiologic features are described pictorially.

Keywords: Osteogenesis Imperfecta; fragile bones; multiple fractures; osteopenia; pseudoarthrosis; wormian bones; hyperplastic callus

Introduction

Osteogenesis imperfecta (OI) is a genetic disorder and is also known as fragilitas ossium. Armand, in 1716, reported a case of brittle bones of a new born infant. This is said to be the first recorded observation of this condition [1]. It is familial with an autosomal dominant mode of inheritance in many cases. Very few cases are reported to have autosomal recessive mode of inheritance. The quality and quantity of bone is diminished. The skeletal manifestations include a spectrum of findings including osteopenia, multiple fractures, wormian bones in skull, gracile and thinning of bones, bending deformities which are all described with the aid of radiographic images in the following paragraphs. This article deals with 8 types of OI and some of these are quite rare. Great confusion exists in the past and continues to exist in contemporary literature with reference to the classification of brittle and soft bones.
Imaging features & discussion (Figures 1-13)
The literature is replete with genetic causes and clinical basis of the diagnosis. The estimated incidence is approximately 1 in every 12,000-15,000 births. Osteogenesis imperfecta is the result of a mutation in one of the two genes that carry instructions for making type 1 collagen. Mutations in the COL1A1 and COL1A2 genes, which encode the α2 and α2 polypeptide chains 7, are responsible for more than 90% of all cases. The patient is generally referred for fractures or suspected OI with Blue Sclera. The imaging features although are classical of OI, it is impossible to correlate them with the types that have been described in the literature [3, 15-17]. Biphosphonate therapy [19, 20] is the choice in the treatment of OI. Corrections of deformities are done by surgery.

The classical radiological features are listed in Table 1.

Table 1: Radiological findings.
- Osteopenia
- Multiple fractures
- Poorly mineralized skull (wormian bones) [4]
- Basilar impression [5]
- Otosclerosis
- Ribbon bones
- Hypercallosis
- Pseudoarthrosis
- Popcorn calcifications [12-14]
- Calcification of interosseous membrane - forearm
- Deformed pelvis – Protrusio acetabulum [8]
- Spondyloysis and spondylolisthesis [6, 7]

There are several synonyms for OI. These are listed in Table 2.

Table 2: Synonyms of OI.
- Van der Hoeve syndrome
- Trias fragilitas ossium
- Eddowe’s syndrome
- Osteopsathyrosis ideopathica of lobstein
- Ekman-Lobstein disease
- Osteogenesis imperfecta congenita
- Osteogenesis Type III lethalis
- Brittle bone disease

Classification by Silence and Glorieux [2] is listed in Table 3.

Table 3: Classification by Silence and Glorieux [2].
1. Type I (Mild)
   - Autosomal dominant, most common, mild to moderate
   - Fractures, minor deformities
   - Almost natural stature
   - Blue sclerae
   - “Dentinogenesis imperfecta” may be present
2. Type II (Lethal)
   - Autosomal recessive, lethal form.
   - Fractures in utero
   - Death before birth (respiratory deficiency)
3. Type III (Severe)
   - Autosomal recessive, progressively deforming, compatible
   - Fractures, kyphoscoliosis, major deformities
   - Very small stature
   - Triangular face; variable color of sclerae
   - “Dentinogenesis Imperfecta” is frequent
4. Type IV (Moderate)
   - Autosomal dominant, mildest, best prognosis
   - Fractures
   - Small stature
   - Variable color of sclerae
   - “Dentinogenesis imperfecta” may be present

Type V, VI, and VII have been added to the original classification system (no type I Collagen mutation, but abnormal bone on microscopy and similar phenotype). They are listed in Table 4.

Table 4: Original classification system (Type V, VI, and VII).
Type V [11]
- Fractures, Hyperplasticcallus[9,10], interosseous membrane ossification, metaphyseal dense lines
- Normal color of sclerae
- No “Dentinogenesis imperfecta”
Type VI
- Looser striations mimicking fractures
- No wormian bones
Type VII
- Fractures, coxa vara
- Normal color of sclerae

The imaging features are illustrated from head to foot.
Type I is mild and shows few features radiologically and is supported clinically by blue sclera (Picture 1a,b).

Picture 1a,b: Blue sclera

Figure 1: (a) Wormian bones [4], (b) basilar impression [5].

Figure 2: (a) Pseudoarthrosis clavicle, (b) Ribbon like thin ribs, (c) multiple fractures.

Figure 3: (a) Compression fractures of vertebrae and healing fractures of ribs at Costo vertebral junctions, (b) Compression fractures of vertebrae, (c) Spondylolisthesis L5-S1 [6, 7].
Figure 4: Deformed pelvis and protrusio acetabulae [8].

Figure 5a,b: Deformities and healing fractures of the bones of upper limb.

Figure 6a,b: Hypercalllosis.

Figure 7a,b: Deformities and healed fractures of bones of forearm.

Figure 8a,b: Hypercalllosis.

Figure 9a,b: Plastic bending of both femora.

Figure 10a,b: Pseudoarthrosis.
Bruck syndrome [18]

OI is associated with several syndromes including Bruck syndrome. It is a rare disorder characterized by association of OI and congenital joint contractures. They have white sclera. The fractures occur postnatally and the contractures are the primary abnormality and not a complication of the fractures (Figure 14). Also has been reported Osteogenesis Sarcoma in association with OI.

Biphosphonate therapy has been introduced and proved to be effective in the treatment of OI (Figure 15). The proof of this is Zebra lines at the metaphyseal ends of long bones [19, 20].
Conclusion

Osteogenesis imperfecta is a rare congenital genetic disorder and several types of classifications have been described in the literature. The classical findings of OI include osteopenia, multiple fractures, poorly mineralized skull (wormian bones), ribbon bones, hypercallosis, pseudoarthrosis, popcorn calcifications, and spondylolisthesis. It is extremely difficult to categorize them into various types without clinical correlation and genetic testing.

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Conflict of interest

Author declares no conflict of interest.

References