Case 14505
Osteogenesis imperfecta type II

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Section: Musculoskeletal System
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Patient: 1 day(s), female

Clinical History

Female neonate delivered with elective Caesarean section at 38 weeks of gestation, because of suspicion of severe skeletal dysplasia based on prenatal ultrasound. After delivery, the neonate deceased within four hours due to respiratory failure.

Imaging Findings

Prenatal ultrasound performed at 27 weeks of gestation showed severe skeletal dysplasia with a relatively small chest and multiple intrauterine fractures (Fig. 1 a-e). After interdisciplinary discussion and genetic counselling, elective Caesarean section was performed.
Post-mortem radiographic findings revealed osteopenia and markedly decreased ossification of the skull (Fig. 2 a), disproportionally small maxillofacial structures, flattening of the occipital bone (Fig. 2 b) and platyspondyly (Fig. 3 a-c). The thoracic cage was narrowed and the ribs showed a beaded appearance (Fig. 3 b). There were multiple fractures of the long bones with surrounding callus resulting in shortening, bowing and deformity (Fig. 4).

Discussion

Based on the antenatal findings on ultrasound and the postnatal radiographic findings, the diagnosis of osteogenesis imperfecta (OI) type 2 according to the Sillence and Glorieux classification was suggested. The diagnosis was confirmed postnatally by genetic testing consisting of analysis of COL1A1 and COL1A2 genes. OI is a genetic disorder characterized by bone fragility due to a defect of type I collagen. There are 7 types of OI, varying from mild to lethal (Fig. 5). The most common symptoms are bone fractures, skeletal deformity and short stature. Type 2 is a lethal form of OI incompatible with life. The cause of death is usually due to severe hypoxemia caused by pulmonary hypoplasia, leading to cardiac arrest or due to multiple fractures [1].

OI is a relatively uncommon disorder, affecting approximately 1 in 10, 000 to 20, 000 births [1]. Type 2 OI is rare, with an incidence of 1-2:100, 000 [2]. As a perinatal lethal condition it always presents as a de novo mutation [2].

Prenatal diagnosis can be made in severe forms of OI, such as type 2, by ultrasound during the second trimester. Ultrasound may show non-specific signs such as growth retardation and polyhydramnios. Other ultrasound findings include decreased echogenicity and increased plasticity of bone due to insufficient bone mineralization and micromelia. Intrauterine fractures with surrounding callus are readily detected. The role of other antenatal imaging modalities such as CT and MRI is limited [1].

Postnatal diagnosis of OI can be made solely on clinical findings only in severe cases. Conventional radiography is usually mandatory. Radiographic findings of type 2 OI consist of osteopenia of the skull with disturbance of intramembranous ossification, platyspondyly, small beaded ribs, severely deformed extremities and broad, crumpled, bent femurs [1-3].

Fig. 6. shows characteristic radiographic features of some other types of OI.

Confirmation of the diagnosis of type 2 OI can be obtained either with direct COL1A1/COL1A2 gene sequencing or biochemical analysis of type 1 collagen in cultured fibroblasts from a skin biopsy [2].

The prognosis of OI type 2 is poor, with some of the fetuses already dead in utero. Most infants will die in the perinatal period, 60% during the first day. Because of this infaust prognosis, the management is mainly supportive, with assisted ventilation and pain control [2].

Final Diagnosis

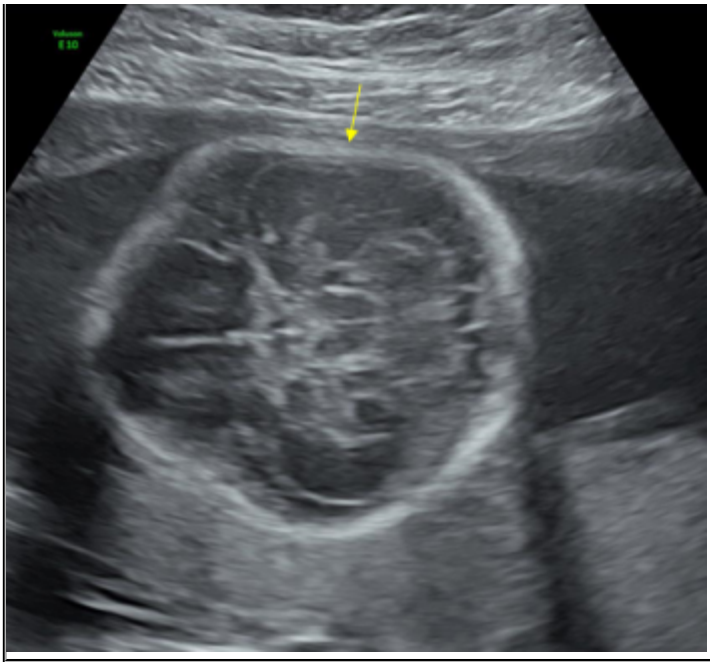
Osteogenesis imperfecta type 2 according to the Sillence and Glorieux classification.

Differential Diagnosis List

Other types of OI, Hypophosphatasia, Thanatophoric dysplasia, Campomelic dysplasia , Achondrogenesis.

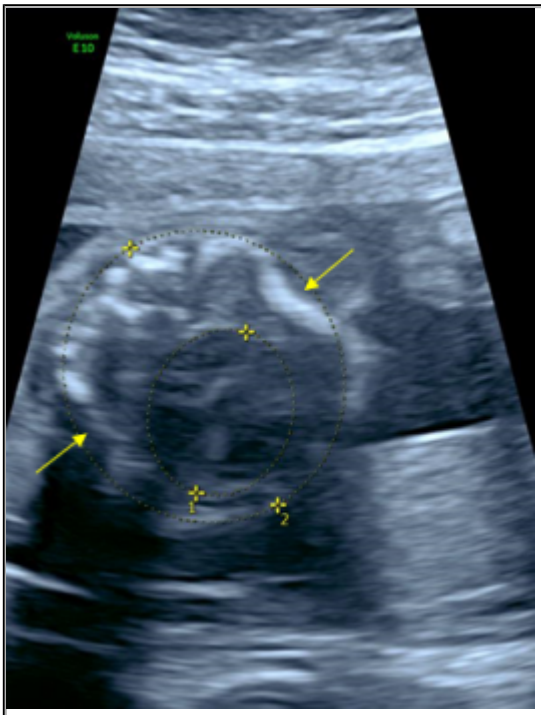
Figures

Figure 1 Prenatal ultrasound at 27 weeks of gestation.



Transverse image of the skull. Note soft, depressible skull, with flattening of the parietal bone on pressure with the ultrasound transducer (arrow).

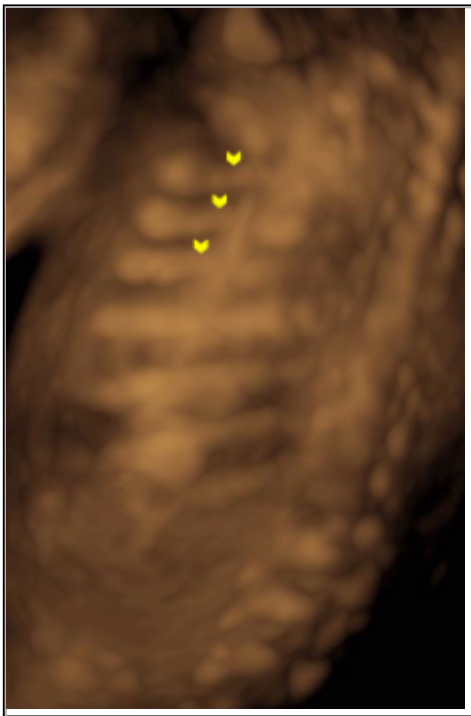
Area of Interest: Bones;
Imaging Technique: Ultrasound;
Procedure: Diagnostic procedure;
Special Focus: Dysplasias;



Axial image of the chest. The fetal cardio-thoracic circumference ratio is 0.55 (normal value < 0.5), indicating relative narrowing of the chest. Note deformed ribs (arrows) due to fractures.

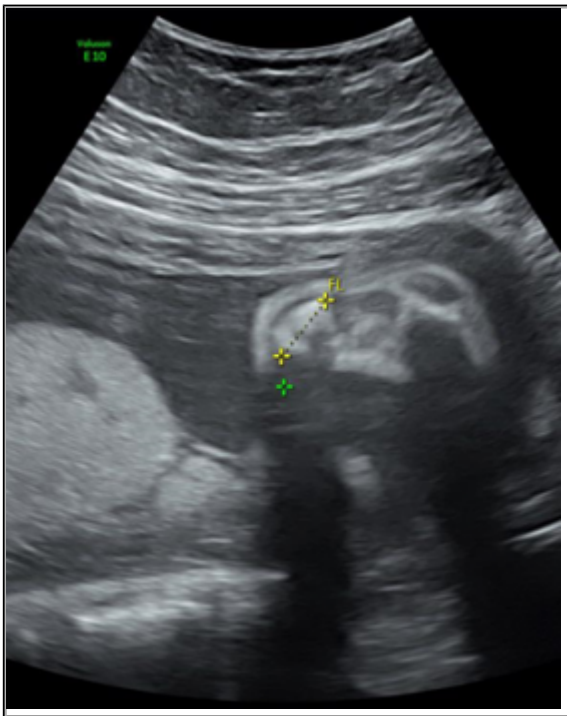
Area of Interest: Biliary Tract / Gallbladder;
Imaging Technique: Ultrasound;

Procedure: Diagnostic procedure;
Special Focus: Dysplasias;



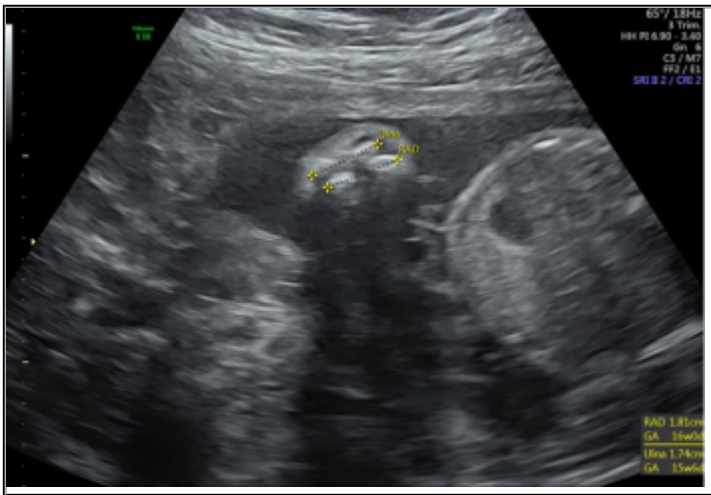
3D-ultrasound image of the chest depicting irregular delineation of multiple ribs in keeping with rib fractures (arrows).

Area of Interest: Bones;
Imaging Technique: Ultrasound;
Procedure: Diagnostic procedure;
Special Focus: Dysplasias;



Longitudinal image of the femur showing shortening of the diaphysis compared to gestational age, broadening and bowing.

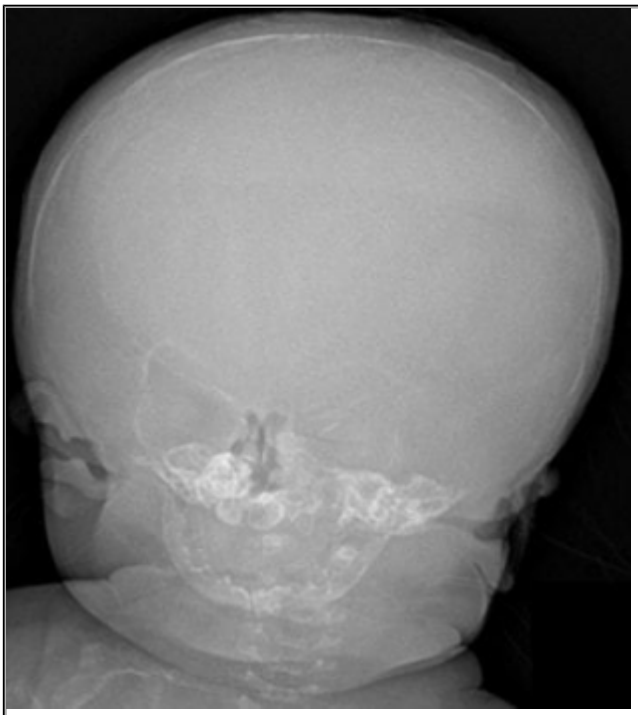
Area of Interest: Bones;
Imaging Technique: Ultrasound;
Procedure: Diagnostic procedure;
Special Focus: Dysplasias;



Marked shortening of the ulna and radius compared to gestational age.

Area of Interest: Musculoskeletal bone;
Imaging Technique: Ultrasound;
Procedure: Diagnostic procedure;
Special Focus: Dysplasias;

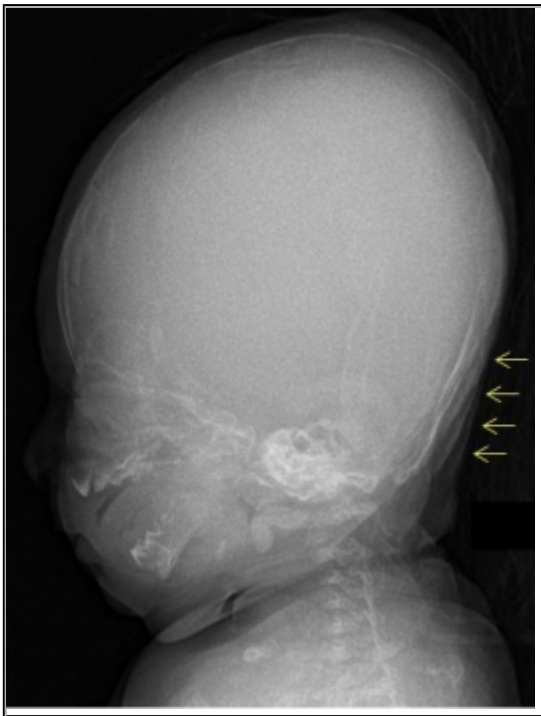
Figure 2 Post-mortem radiograph of the skull



Post-mortem anteroposterior radiograph of the skull shows decreased ossification of the calvaria.

Area of Interest: Musculoskeletal bone;

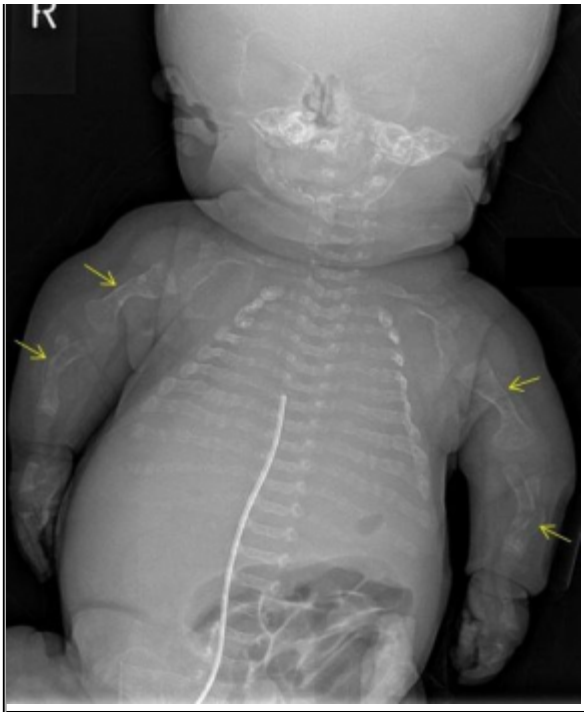
Imaging Technique: Plain radiographic studies;
Procedure: Diagnostic procedure;
Special Focus: Demineralisation-Bone;



Post-mortem lateral radiograph of the skull showing decreased ossification, flattening of the occipital bone (arrows) and disproportionally small maxillofacial structures.

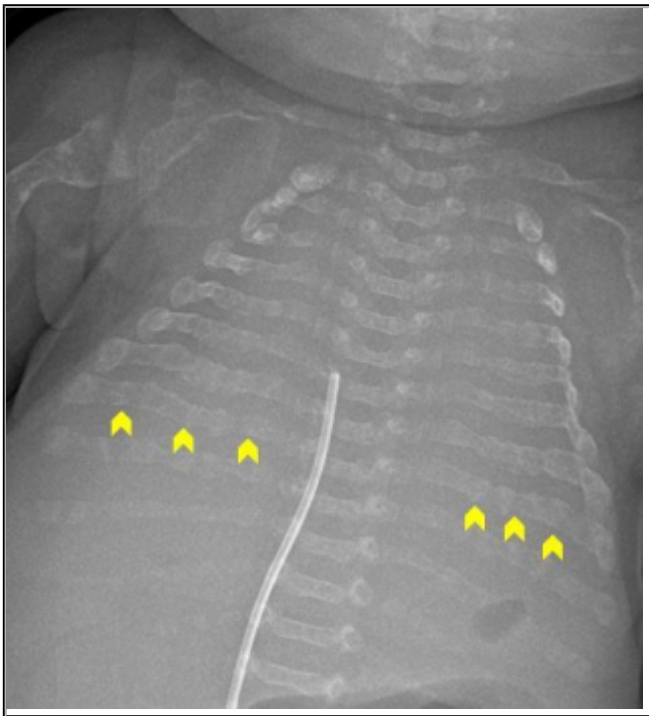
Area of Interest: Musculoskeletal bone;
Imaging Technique: Plain radiographic studies;
Procedure: Diagnostic procedure;
Special Focus: Demineralisation-Bone;

Figure 3 Post-mortem radiograph of the chest and abdomen



Post-mortem anteroposterior radiograph of the trunk showing marked demineralization of the skeleton and abnormal bowing of the humeri, radii and ulnae (arrows).

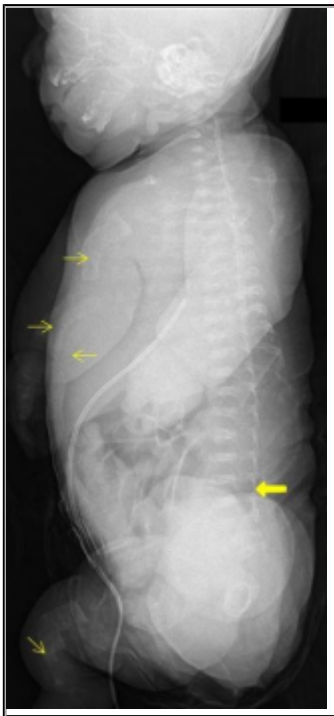
Area of Interest: Musculoskeletal bone;
Imaging Technique: Plain radiographic studies;
Procedure: Diagnostic procedure;
Special Focus: Dysplasias;



Enlarged view of the chest revealing beaded appearance of the multiple ribs (as demonstrated with arrowheads on the course of rib 8 R/L). Note also presence of an umbilical vein catheter.

Area of Interest: Musculoskeletal bone;

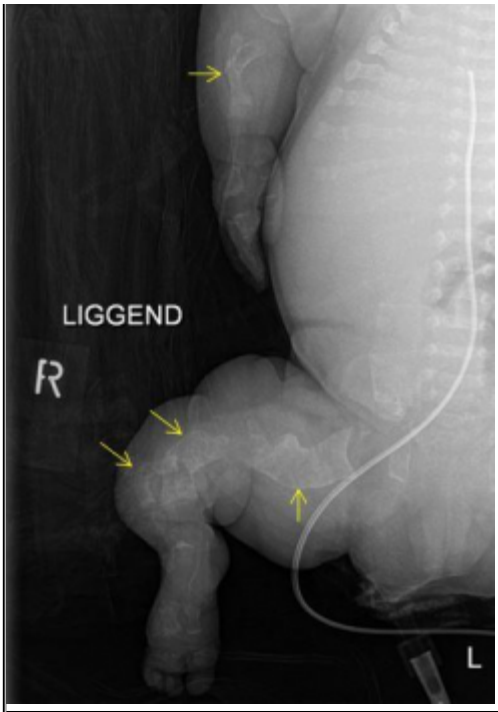
Imaging Technique: Plain radiographic studies;
Procedure: Diagnostic procedure;
Special Focus: Dysplasias;



Post-mortem lateral radiograph showing flattening of the vertebral bodies (as demonstrated on a lumbar vertebra (thick arrow); Note also bowing fractures of the long bones (small arrows). Presence of an umbilical vein catheter.

Area of Interest: Musculoskeletal bone;
Imaging Technique: Plain radiographic studies;
Procedure: Diagnostic procedure;
Special Focus: Dysplasias;

Figure 4 Post-mortem radiograph of the right upper and lower limb



Post-mortem radiograph of the right limbs showing shortening, bowing and callus formation of the long bones due to fractures (arrows). This results in a typical accordion-like appearance of the tubular bones.

Area of Interest: Musculoskeletal bone;
Imaging Technique: Plain radiographic studies;
Procedure: Diagnostic procedure;
Special Focus: Dysplasias;

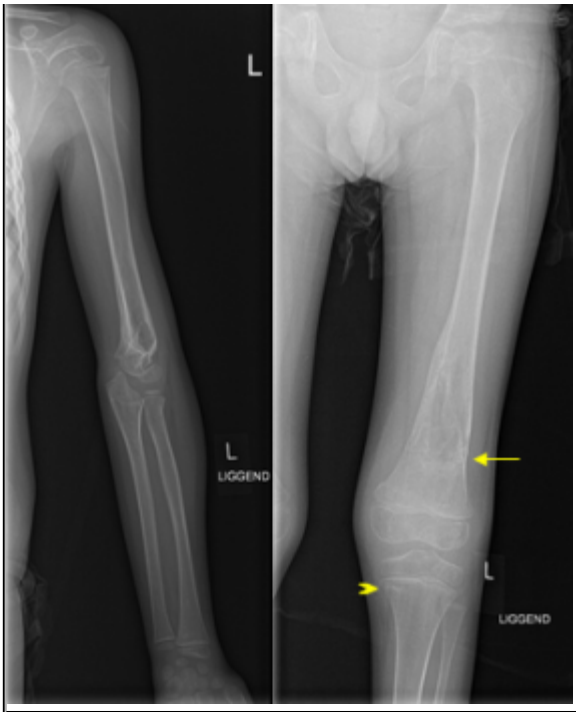
Figure 5 Table showing different types of OI with characteristic (imaging) features.

Type I (mild)	Fractures, minor deformities, Wormian bones, codfish vertebrae, osteopenia
Type II (perinatally lethal)	Fractures in utero, severe deformities, beaded ribs, small thorax, platyspondyly
Type III (severe)	Fractures, major deformities, popcorn calcifications (metaphyses), Wormian bones, codfish vertebrae, kyphoscoliosis, severe osteopenia
Type IV (moderate)	Fractures, Wormian bones, codfish vertebrae
Types V, VI, VII and VIII have recently been added. (These types do not have a characteristic type I collagen mutation)	
Type V	Fractures, hyperplastic callus formation, metaphyseal dense lines, interosseous membrane ossification
Type VI	Looser striations mimicking fractures
Type VII	Fractures, coxa vara
Type VIII	Fractures at birth, severe osteopenia

Table showing the different types of OI according to the Sillence and Glorieux classification and most characteristic (imaging) features.

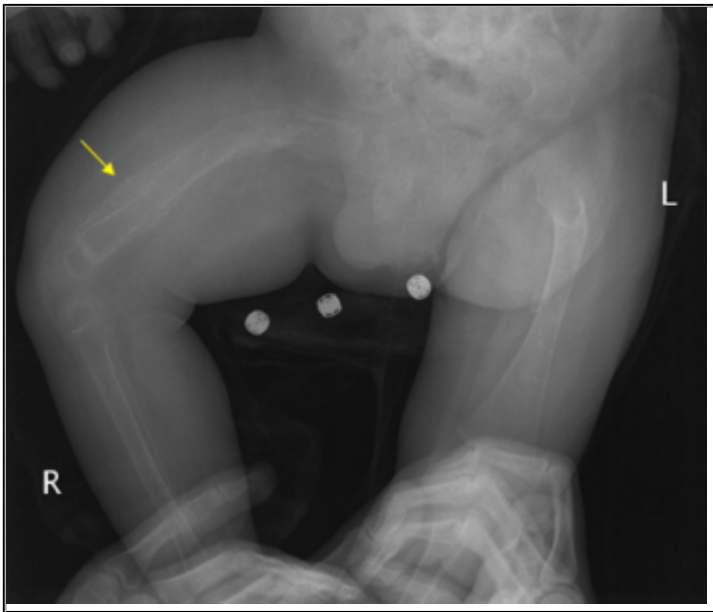
Area of Interest: Bones;
Imaging Technique: Plain radiographic studies;
Procedure: Diagnostic procedure;
Special Focus: Dysplasias;

Figure 6 Typical examples of different types of OI on radiographs



Type I OI. Note osteoporosis, mild overmodelling of the long bones and fractures sequelae of a different date at the left distal femoral diaphysis (arrow) and medial proximal tibia diaphysis (arrowhead).

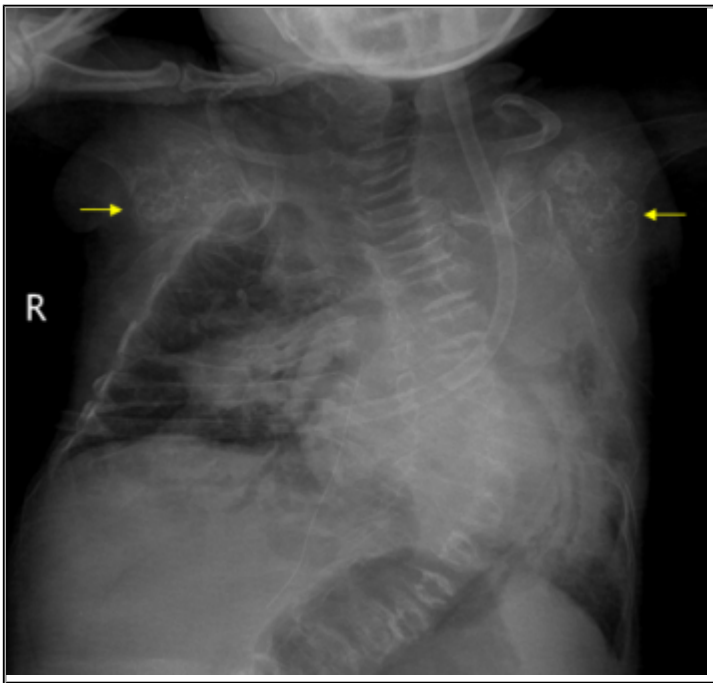
Area of Interest: Musculoskeletal bone;
Imaging Technique: Plain radiographic studies;
Procedure: Diagnostic procedure;
Special Focus: Dysplasias;



Type III OI in a 1 year-old male. Note osteoporosis, marked deformities of the femora with bilateral fracture sequelae of the femoral diaphyses. There is hypertrophic callus at the right femoral diaphysis (arrow).

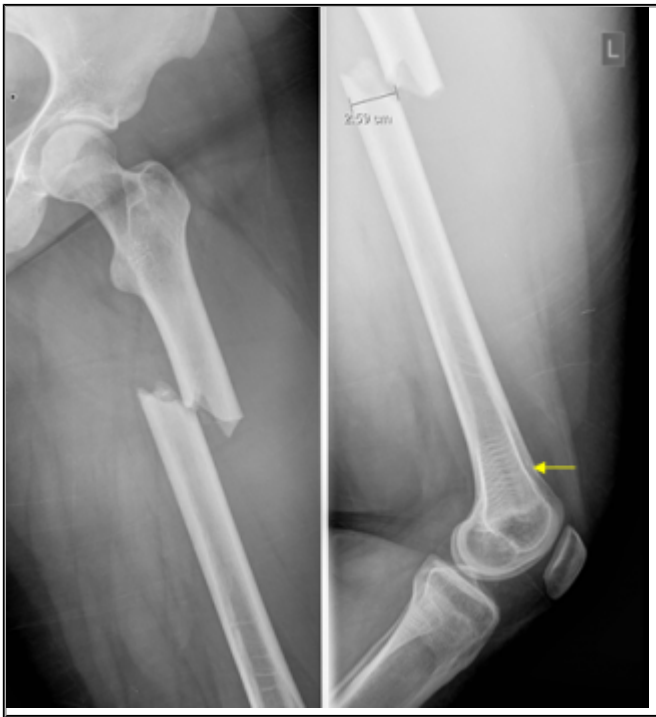
Area of Interest: Musculoskeletal bone;
Imaging Technique: Plain radiographic studies;
Procedure: Diagnostic procedure;

Special Focus: Dysplasias;



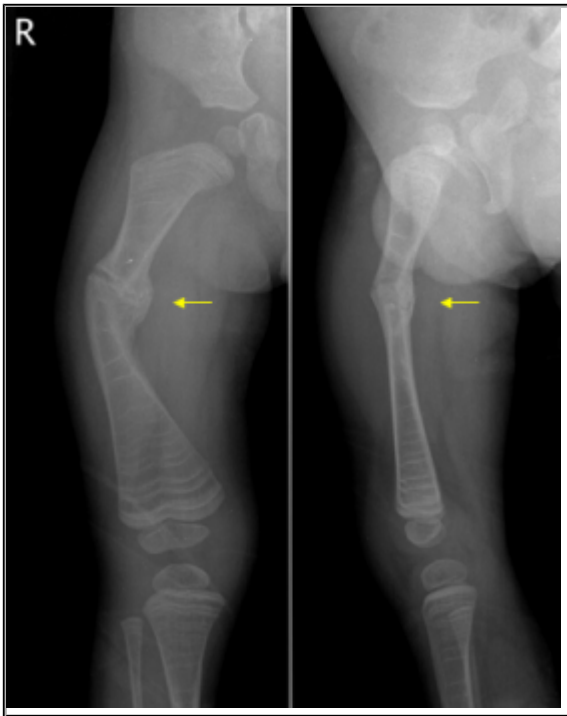
Type III OI. Note popcorn calcifications of the proximal humeri (arrows) and severe kyphoscoliosis.

Area of Interest: Musculoskeletal bone;
Imaging Technique: Plain radiographic studies;
Procedure: Diagnostic procedure;
Special Focus: Dysplasias;



Type IV OI in an adult patient Note spontaneous fracture at the left femoral diaphysis and dense metaphyseal bands in the distal femur (arrow) and proximal tibia due to bisphosphonate treatment.

Area of Interest: Musculoskeletal bone;
Imaging Technique: Plain radiographic studies;
Procedure: Diagnostic procedure;
Special Focus: Dysplasias;



Type V OI. There is an incompletely healed fracture at the middiaphysis of the right femur with hyperplastic callus formation (arrow). Note dense metaphyseal lines within the distal femur and proximal tibia.

Area of Interest: Musculoskeletal bone;
Imaging Technique: Plain radiographic studies;
Procedure: Diagnostic procedure;
Special Focus: Dysplasias;

References

[1] Renaud A, Aucourt J, Weill J, et al (2013) Radiographic features of osteogenesis imperfecta. Insights Imaging 4:417-429

[2] Steiner RD, Adsit J, Basel D (1993) COL1A1/2-related osteogenesis imperfecta In: Pagon RA, Adam MP, Ardinger HH et al, (eds) GeneReviews(R), Seattle (WA)

[3] Khandanpour N, Connolly DJ, Raghavan A, Griffiths PD, Hoggard N (2012) Craniospinal abnormalities and neurologic complications of osteogenesis imperfecta: imaging overview Radiographics 32:2101-2112

Citation

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