Pelvic radiograph in skeletal dysplasias: An approach

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Abstract

The bony pelvis is constituted by the ilium, ischium, pubis, and sacrum. The pelvic radiograph is an important component of the skeletal survey performed in suspected skeletal dysplasia. Most of the common skeletal dysplasias have either minor or major radiological abnormalities; hence, knowledge of the normal radiological appearance of bony pelvis is vital for recognizing the early signs of various skeletal dysplasias. This article discusses many common and some uncommon radiological findings on pelvic radiographs along with the specific dysplasia in which they are seen; common differential diagnostic considerations are also discussed.

Key words: Achondroplasia; pelvic radiograph; skeletal dysplasia

Introduction

The bony pelvis is constituted by ilium, ischium, pubis, and sacrum. The pelvic radiograph is commonly included in routine radiographic examinations, and consequently often serves as the first evidence of skeletal dysplasia. Knowledge of the normal pelvic embryological development is vital for recognizing the early signs of various skeletal dysplasias manifesting in pelvis. The pelvic radiograph often reveals characteristic abnormalities in pelvic shape, size, number of bones, density, timing of ossification, etc., which may provide vital clues in the recognition of various dysplasias. Caffey was the first to recognize the importance of the pelvis in the diagnosis of bone dysplasias when he conducted a detailed analysis of the skeletal anomalies encountered in achondroplasia.[1] Further down the line, Kaufmann[2] described many other patterns of malformation, which shed more light on the significance of pelvis in diagnosing constitutional diseases of bone. This article discusses many common and some uncommon radiological findings on pelvic radiographs along with the specific dysplasia in which they are seen; common differential diagnostic considerations are also discussed.

Ossification

For interpretation of radiographic abnormalities in bony pelvis, thorough knowledge about the ossification centres in and around the pelvis is essential. The time of appearance of the ossification centres is listed in Table 1.

In the full-term neonatal pelvic radiograph, ilium, superior pubic, and ischial rami, 1st to 5th sacral vertebrae, 1st to 4th neural arches, 1st and 2nd lateral centres, and the first coccygeal vertebra are usually seen to be ossified [Figure 1].[2]

Abnormalities in different skeletal dysplasias

Iliac bone and acetabulum

- **Flared iliac wing and flat acetabular roof** – Characteristic radiological signs seen in both achondroplasia and lethal dysplasias. The typical appearance in achondroplasia has been given numerous descriptions – “tombstone shape;” “mickey mouse ear pelvis;” “champagne glass pelvis.”

- **Square iliac wing** (vide supra) – Refers to small square-shaped iliac wings akin to a tombstone. This sign is seen in the most common skeletal dysplasia, i.e., achondroplasia. Incidentally, it is also seen in the second most common lethal skeletal dysplasia viz., thanatophoric dysplasia. Rhizomelic dwarfism is a common feature of both dysplasias; however, telephone handle shaped femur, short horizontal ribs, and cloverleaf skull are distinctive features of thanatophoric dysplasia.[4] Similarly, champagne glass shaped pelvic inlet, trident hand and pelvis (described below), metaphyseal flaring and horizontal acetabular roof are some of the unique features of achondroplasia that enable easy differentiation between the two entities [Figure 2]

- **Hypoplastic iliac wing** – Seen in several spondyloepiphyseal dysplasias, namely spondyloepiphyseal dysplasia tarda with mental retardation, spondylomegaepiphyseal dysplasia, and Dyggve–Melchior Clausen (DMC) syndrome. It should be differentiated from foreshortening of iliac bones in campomelic dysplasia (see below).

- **Champagne glass pelvis/mickey mouse ear pelvis** (also see above) – Refers to the flattening of iliac blades with increased acetabular angles and small sacrosacral notch giving rise to a champagne glass shaped pelvic inlet. This sign is seen in achondroplasia [Figure 2].

- **Sloping acetabular roof** – Refers to shallow acetabular fossae and steep slope of acetabular roof, which is a common feature in mucopolysaccharidosis. Similar finding may also be seen in acetabular dysplasia which may occur as a genetically determined condition or secondary to incomplete reduction of a congenital dislocation, damage to the lateral acetabular epiphysis, or femoral head maldevelopment [Figure 3].[5]

- **“Trident sign”** – Spurs at the medial and lateral acetabular margin and in the center of the acetabulum gives rise to shape resembling a three-pronged spear known as trident [Figure 2]. This sign is seen in achondroplasia, Jeune’s asphyxiating thoracic dystrophy (JATD), Ellis-van Creveld syndrome, and thanatophoric dysplasia.[6] Mesomelic pattern of dwarfism and bell shape of the chest are distinctive features of Ellis-van Creveld syndrome.

The imaging findings in achondroplasia include

- Trident pelvis/tombstone shape of the iliac bone
- Rhizomelic shortening of long bones with metaphyseal flaring
- Gradual narrowing of interpedicular distance in the lumbar spine
- Short pedicles of lumbar vertebrae
- Large calvarium and a relatively small base
- Trident hand (increased gap between third and fourth fingers)
JATD shows short ribs and a narrow thorax [Figure 4].

Thanatophoric dysplasia is a lethal dysplasia with FGFR3 mutation (allelic but much severe than achondroplasia and hypochondroplasia) with characteristic “cloverleaf skull,” extreme long bone shortening, bowed appearance of long bones (telephone handle shape of femora), more severe rib shortening, and increased platyspondyly with characteristic U and H-shaped vertebrae.\(^7\)

**Imaging clues towards Ellis-van Crevald syndrome** (clinical setting of post-axial polydactyly, midline cleft lip and cardiac anomalies) include

- Trident pelvis
- Acro-mesomelic limb shortening
- Outward bowing of humeri
- Bell-shaped thorax improving with age
- Laterally deficient upper tibial epiphyses
- Cone shaped epiphyses in hands, capito-hamate coalition

- **Foreshortened iliac bone** – In campomelic dysplasia, the iliac bone is small, oriented in the anterior-posterior direction, and hence appears foreshortened [Figure 5]. Campomelic dysplasia is a lethal dysplasia associated with XY sex reversal.

This appearance is also seen in chondroectodermal dysplasia and cleidocranial dysostosis. Bowed long bones, hypoplastic scapulae, and narrow chest seen in campomelic dysplasia allow differentiation from these entities.

**The imaging pointers towards campomelic dysplasia include**

- Bowing of long bones of extremities
- Angulated bowing of femur at the junction of upper third and middle third
- Angulated bowing of tibia at the middle and lower third junction
- Small/foreshortened iliac wing
- Hypoplastic angle of scapula

- **Lateral acetabular notch** – Refers to a notch at the superolateral margin of the acetabulum seen in acromesomelic dysplasia and in some cases of neonatal hip dysplasia

- **Lacy iliac wing** – This sign is ascribed to the iliac crest irregularity due to dystrophic ossification of the osteochondral junction giving an appearance of a lace border around the iliac crest. This radiological sign is a pathognomonic feature of Dyggve- Melchior Claussen (DMC) syndrome [Figure 6].\(^8\) Other pelvic findings in this syndrome include short iliac wings, wide sacroiliac joints, narrow sciatic notches, wide pubic and ischiai rami, dysplastic acetabular fossae, and laterally displaced femoral heads [Figure 6]. Similar findings are seen in enchondromatosis (Ollier’s disease) [Figure 7] as well as in Smith-McCort syndrome, a condition clinically and radiologically identical to DMC syndrome except for absence of mental retardation.\(^9\)

**The imaging pointers towards DMC and Smith-McCort syndrome include**

- Lacy iliac crest
- Dysplastic acetabulum with “laterally placed” femoral heads
- Wide sacroiliac joints
- Wide pubic and ischial rami
Central notching in the vertebrae
Lace-like appearance of scapular angle

Lacy appearance of iliac wings may also be seen in parastreptomac dwarfism (Greek word parastreptomac = twisted). Symmetrical bowing of the long bones, severe genu valgum, short neck, kyphoscoliosis, etc., permit easy differentiation of this entity from others.

- "Paraglider" or "crescent" shape of iliac wing – Deficient mineralization of the iliac wings in achondrogenesis gives rise to the characteristic "paraglider" shape of the iliac wings [Figure 8]. Achondrogenesis is a lethal skeletal dysplasia, with poor mineralization of the skeleton. Three types have been described, type IA, IB, and II. The imaging findings and the disease severity vary in these types. All the three types show a crescent/paraglider shape iliac bone. Type IA is a severe form of the disease, where the skull vault appears completely unmineralized. There is severe shortening of the long bones and the ribs are short and beaded. The vertebral bodies show poor mineralization (c.f: pedicles of thoracic vertebrae unmineralized in Campomelic dysplasia). In type IB, there is a typical expansion of the bony vertebral column at the lumbar level, giving rise to the "cobra head" appearance.

- Halberd pelvis – Metatropic dysplasia is a spondyloepimetaphyseal dysplasia with diagnostic radiographic findings comprising of marked platyspondyly, short tubular bones with broad metaphyses, and small epiphyses. The unique shape of the iliac wing – a notch between the shortened iliac body and prominent crescent-shaped iliac wing resembles a two-handed pole weapon known as halberd. This pathognomonic radiological sign distinguishes this entity from other common forms of spondyloepimetaphyseal and spondyloepimetafocal dysplasias.

- Snail-like appearance of iliac wing – Refers to hypoplastic iliac bones with a snail like configuration [Figure 9]. Seen in Scheneckebencken dysplasia (German for snail pelvis), a rare lethal neonatal chondrodysplasia with autosomal recessive inheritance. Other distinctive features seen in this disorder include flattened and hypoplastic vertebral bodies, short ribs, dumb bell-like appearance of long bones, and precocious ossification of the tarsal bones.

- Sunburst appearance of iliac wings – Dense bands of bone radiating from the acetalulum to iliac crest [Figure 10] in a fan-like pattern give rise to “sunburst” appearance which is a characteristic feature of osteopathia striata. Similar bands extending from metaphysis to diaphysis of long bones are the other important radiological feature of this dysplasia.

- Posterior iliac horn – Bilateral symmetrical posterior iliac horns [Figure 11] are a unique feature of Nail Patella syndrome (also known as Fong’s disease). The horns are formed as a result of exostoses from posterior aspect of iliac bones and are consider as a pathognomonic feature seen in as many as 80% of the patients with this syndrome.

- Bone within bone appearance – Increased density of pelvic bones with pelvic endobones (bone within bone)
appearance is a classical feature of type II autosomal dominant osteopetrosis [Figure 12].

- **Multiple scattered sclerotic foci in pelvic bones:**
  Seen in osteopoikilosis, a sclerosing bony dysplasia characterised by multiple bone islands. The pelvis is a common site of involvement with clustering of sclerotic foci around hip and sacroiliac joints [Figure 13].

**Sacrum and lower lumbar vertebrae**

- **Calcific Stippling of sacrum and lumbar vertebrae** – Seen in chondrodysplasia punctata, especially Sheffield type. This disorder is characterized by calcific stippling of cartilage, particularly epiphyses of long bones.[14] Stippling with rhizomelic shortening can suggest rhizomelic chondrodysplasia punctata [Figure 14].

The imaging findings include

- Stippling of sacral and lumbar vertebrae
- Rhizomelic shortening of long bones
- Tarsal bone stippling
- Coronal cleft vertebrae
- Scoliosis

- **Decreasing interpedicular distance in lumbar vertebrae** – seen in achondroplasia (see above)
Medially placed, more rounded pedicles of lumbar vertebrae – Seen in spondylometaphyseal dysplasia Kozlowski type.

Absent sacral vertebrae – Aplasia/dysplasia of the lumbosacral vertebral bodies is a common feature of caudal regression syndrome, sometimes associated with complete sacral agenesis [Figure 15].

Pubic bone and triradiate cartilage

Absence ossification of the pubic bone – Among the pathognomonic features of cleidocranial dysostosis is retarded/absent ossification of the pubic bones which gives rise to an apparent increase in interpubic distance (pseudo diastasis) [Figure 16]. This feature is also seen in achondrogenesis, hypochondrogenesis (short long bones), Sjogren Larssen syndrome, Wolf syndrome, and spondyloepiphyseal dysplasia congenital.\(^{[15]}\)

Imaging features of cleidocranial dysostosis include

- Widely spaced pubic bones, pubic diastasis like appearance
- Elongated femoral head (chef’s hat sign)
- Accessory epiphyses at the second metacarpal base
Wide open cranial sutures
Multiple wormian bones
Multiple unerupted teeth
Distal phalangeal tapering
Hypoplastic/absent clavicles

True dislocation of hip – Superolateral displacement of femoral head and shallow acetabulum is seen in the developmental dysplasia of hip. Other important pelvic radiographic findings include small capital femoral epiphysis, delayed femoral head ossification, and acetabular sclerosis. Delayed femoral head ossification renders plain film radiography unreliable in children less than 12 months of age with ultrasonography being the optimum imaging modality.

Apparent dislocation of hip – Delayed ossification of capital femoral epiphyses and horizontal acetabular roofs result in this radiographic appearance seen in spondyloepiphyseal dysplasia congenita (SEDC). This disease is a type II collagenopathy, having autosomal dominant transmission, and clinical presentations include hearing loss and retinal detachment. Other important pelvic findings in this disorder include metaphyseal flaring, triangular fragment on the inferior aspect of the broad femoral neck, and coxa vara [Figure 17]. Similar findings can also be seen in spondyloepimetaepiphysal dysplasia (Strudwick type) [Figure 18], but with more prominent metaphyseal irregularity.

SEDC has the imaging features as follows
• Absent ossification of pubic bones
• Apparent dislocation of hip joints
• Coxa vara

Figure 16: Cleidocranial dysostosis. Pelvic radiograph revealing retarded ossification of pubic bones resulting in widening of symphysis pubis (pseudo diastasis, asterisk). Also note the elongated shape of capital femoral epiphyses giving a “chef’s hat” sign, along with coxa vara (arrows)

Figure 17 (A and B): SEDC with pelvic radiograph (A) demonstrating delayed ossification of bilateral femoral heads and horizontal configuration of acetabular roofs (squiggly arrow). Also note bilateral femoral metaphyseal flaring (straight arrows) and coxa vara. Radiograph of lumbosacral spine (B) in the same patient depicts anisospondyly (L4 vertebral body larger than L5 vertebral body) (arrow)

• Poorly developed femoral head epiphyses
• Platyspondyly
• Anisospondyly (variable size of lumbar vertebrae; e.g., L1 bigger than L5)

Spondyloepiphyseal dysplasia tarda (SEDT), in comparison, has a typical morphology of the vertebral bodies; a “hump” at the posterior two-thirds of the vertebral bodies.

• Pubic diastasis – Refers to abnormally wide gap between the pubic bones of >10 mm. This radiological sign is seen in Bladder exostrophy, Prune Belly syndrome, and osteogenesis imperfecta.
• Wide triradiate cartilage – Widening and delayed fusion of triradiate cartilage is seen in pseudoachondroplasia [Figure 19].

The imaging findings in pseudoachondroplasia are
• Square iliac bones (similar to achondroplasia)
• Wide triradiate cartilage
• Delayed ossification of femoral head
• Metaphyseal flaring
• Irregular epiphyses
• Tongue like projection from anterior aspect of vertebrae
• Widened costo-chondral and costo-transverse junction.

Proximal femur and hip joints
• Coxa vara – Refers to characteristic hip deformity due to angle between femoral neck and shaft being <120 degrees. This sign is nonspecific and can be seen in many skeletal dysplasias such as spondyloepiphyseal dysplasia congenita, spondyloepimetaepiphysal dysplasia Strudwick type, congenital coxa vara with Fairbank’s triangle, and pseudoachondroplasia.
• Coxa valga – Refers to abnormally increased femoral neck shaft angle which may range normally up to 160 degrees in newborns and 145 degrees in children. This
sign is seen in mucopolysaccharidosis [Figure 3], Turner syndrome, and developmental dysplasia of hip.

- **Small capital femoral epiphyses/irregularity and fragmentation/snow cap epiphysis** – Seen in Perthes’ disease/avascular necrosis of head of femur. If bilateral, search should be made for any history of steroid intake, hypothyroidism, sickle cell disease, and other hemoglobinopathy. ARMED [Figure 20] is an autosomal recessive form of multiple epiphyseal dysplasia which can mimic the appearance of bilateral Perthes’ disease.

**The imaging findings in multiple epiphyseal dysplasia include**

- Flattened irregular capital femoral epiphyses
- Subluxation of femoral head/protrusio acetabulae
- Cone-shaped epiphyses
- Irregular carpal bones
- Delayed bone age

Double-layered patella, a characteristic finding seen in the lateral knee radiograph, is found in ARMED.

- **Chef’s hat sign** – Attributed to deformed capital femoral ossification centre with a rounded appearance and tapered edge directed towards femoral neck which gives the overall impression of a “Chef’s Hat.”[16] This sign is a characteristic feature of cleidocranial dysplasia with absent ossification of the pubic symphysis being the other important pelvic sign of this dysplasia [Figure 16].

- **Prominent medial beak at medial femoral neck** – Characteristic feature of pseudoachondroplasia, a rare rhizomelic short limbed skeletal dysplasia which shows radiographic similarity with achondroplasia in many aspects.[17] However, unlike achondroplasia, the skull is normal with normal interpedicular distance and marked platyspondyly [Figure 21].[17]

- **“Monkey-wrench femora”** – Seen in two rare osteochondrodysplasias, i.e., diastrophic dysplasia/desboquois dysplasia and Larsen’s syndrome of Reunion Island. In diastrophic dysplasia, the typical appearance of the femoral heads resembling a Swedish Key or monkey wrench [Figure 22], is a pathognomonic radiological sign with other important radiological features including advanced carpal ossification and radial deviation of the second phalanx with a supernumerary ossicle at base of second phalanx.[18] Larsen’s syndrome of Reunion Island shows similar clinical and radiological appearances including monkey wrench femora (inconstant feature), however, extra ossification centre is not seen in these patients enabling differentiation between the two.[18]

- **Irregular metaphyses of femur** – Seen in nutritional rickets, vitamin D dependent rickets, metaphyseal dysplasias, and spondylometaphyseal dysplasias. The different types of metaphyseal dysplasia are Schmidt type, McKusick type [Figure 23], and Jansen type (although several other rarer types exist). Spondylometaphyseal dysplasia group of diseases have additional changes in the spine.

- **“Corner fracture” at upper femoral metaphysis** – Unique radiological feature seen in spondylometaphyseal dysplasia Sutcliffe type.[19] Spondylometaphyseal dysplasia Sutcliffe type (corner fracture type) is a rare disorder involving the spine and the long bone metaphyses. The clinical presentation is with waddling gait and coxa vara with imaging revealing distinctive “corner fractures” at the ends of long bone metaphyses [Figure 24]. Bilateral coxa vara is another important pelvic finding in this disorder. Additional imaging features include irregularity at vertebral endplates, odontoid hypoplasia, and ovoid shape of vertebral bodies. The differential diagnosis of “corner fractures” includes nonaccidental injury of childhood,
which needs to be carefully ruled out. Points favoring spondylometaphyseal dysplasia will include the presence of vertebral changes, generalized metaphyseal irregularity, and lack of any other fractures in the body.\[19\]

- **Delayed ossification and fusion of femoral head** – Feature of Meyer’s dysplasia and Perthes’ disease. Other differential diagnoses include SEDC, pseudoachondroplasia, multiple epiphyseal dysplasia, hypothyroidism [Figure 25], dyschondroplasia, arthritis, and infection.

**The imaging clues which can support a diagnosis of juvenile or congenital hypothyroidism are**

- Delayed bone age
- Small irregular epiphyses
- Irregular sclerosis of metaphyses
- Wide triradiate cartilage
- Dorsolumbar kyphosis, bullet-shaped L1 vertebra
- Enlarged sella

- **Delayed ossification of femoral head** – Seen in Mucopolysaccharidosis, pseudoachondroplasia, spondyloepiphyseal dysplasia, and developmental dysplasia of the hip. Irregular ossification of the femoral head in mucopolysaccharidosis along with presence of bilateral coxa vara and bulbous femoral metaphyses in pseudoachondroplasia\[20\] helps in differentiating between these entities based on pelvic radiographic findings.

**Abnormal soft tissue ossification**

Seen in fibrodysplasia ossificans progressiva [Figure 26], a rare hereditary mesodermal disorder with pelvic radiographs.

**Figure 20:** Small capital femoral epiphyses in multiple epiphyseal dysplasia. Radiograph of the pelvis shows small and irregular bilateral femoral head (marked); akin to bilateral Perthe’s disease. Radiograph of the knee (not shown here) showed irregular epiphyses around the knee joint, and a tibiotalar slant was apparent in ankle radiograph.

**Figure 21 (A and B):** Pseudoachondroplasia – Pelvic radiograph (A) demonstrating metaphyseal flaring (block arrows), squared iliac wings, and horizontal acetabular roofs (squiggly arrow) akin to achondroplasia. Note that the femoral head are small (straight thin arrow). Also, in lateral spine radiograph (B), the tongue-shaped projection from the lumbar vertebrae (arrows) are a specific imaging finding in pseudoachondroplasia.

**Figure 22:** Case of Desboquois dysplasia – Anteroposterior radiograph of the pelvis showing “monkey wrench” appearance of both femora, elevated greater trochanters (thin arrows), coxa vara and dysplastic femoral epiphyses (thick arrow).

**Figure 23:** Irregular upper femoral metaphyses in McKusick type metaphyseal chondrodysplasia.
revealing ectopic soft tissue ossification which may ultimately result in bony bridging between thorax and pelvis.[21]

**The imaging findings in this rare condition include**
- Multiple sheet-like ossification in soft tissues
- Bridging ossifications in soft tissues of neck and back
- Hallux valgus
- Microdactyly of the great toe

**Conclusion**

Radiograph of the pelvis is an important component of the skeletal survey in suspected skeletal dysplasia. Even in isolation, it can provide a host of information to guide the radiologist towards a possible group of disorders; thus, helping to perform a more specific/targeted search for additional findings to finally reach a diagnosis.

**Essentials**
- Knowledge of the ossification centres in the bony pelvis is very important [Table 1]

**Table 2: Differential diagnoses of various imaging findings in pelvis radiograph**

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<th>Imaging findings</th>
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Few named signs and specific morphology can help make a definitive diagnosis in some dysplasias [Table 2]

A combined interpretation of the entire skeletal survey is essential to reach a diagnosis [Table 3].

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Conflicts of interest
There are no conflicts of interest.

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