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Radiological Diagnosis of Skeletal Dysplasias in Children

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Abstract

Radiological diagnosis of skeletal dysplasias in children relies on early recognition of disproportionate growth and characteristic imaging patterns across the skeleton. Skeletal dysplasias are a heterogenous group of genetic disorders of bone and cartilage with a combined birth prevalence of around 1 in 5,000 births. Early radiographic identification guides surveillance for spinal stenosis, craniovertebral anomalies, hip dysplasia and blood disorders. It also supports genetic counselling and, in some conditions, allows targeted therapies such as enzyme replacement or growth-modifying treatment. This chapter outlines key clinical clues that should alert paediatric orthopaedic surgeons to an underlying skeletal dysplasia and summarises indications and techniques for a dedicated dysplasia skeletal survey, including bone age assessment and targeted supplementary views. Furthermore, core radiological terminology and measurements used to describe disproportions and dysplastic change, are reviewed to support systematic pattern recognition. The chapter then describes the principal clinical and radiological hallmarks of common dysplasias relevant to paediatric orthopaedic practice, including achondroplasia and hypochondroplasia, pseudoachondroplasia and multiple epiphyseal dysplasia, spondyloepiphyseal dysplasia congenita, diastrophic dysplasia, osteogenesis imperfecta, sclerosing bone dysplasias, mucopolysaccharidoses and metaphyseal chondrodysplasias, highlighting key differential diagnoses and red flag complications. The role of advanced imaging, particularly at the craniovertebral junction, cervical spine and hips is emphasised where there is risk of cord compression, atlantoaxial instability or early degenerative change. Finally, the importance of multidisciplinary assessment, integrating clinical genetics, endocrinology, radiology, anesthesia and surgical specialties is stressed to minimise misdiagnosis and optimise long-term functional outcomes. Prenatal ultrasound and cross-sectional imaging features that raise suspicion of skeletal dysplasia are summarised, underscoring opportunities for early counselling, delivery planning and postnatal evaluation.

Keywords: Skeletal Dysplasia Radiology, Achondroplasia, Hypochondroplasia, MED, Storage disorders Radiology

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Introduction

Skeletal dysplasias, also known as osteochondrodysplasias, comprise a broad and heterogenous group of genetic conditions affecting bone and cartilage development. Although most dysplasias are individually rare, collectively they are common, with an overall prevalence of 1 in 5,000 births [1]. Early radiographic identification guides management (e.g. monitoring for spinal stenosis, cranio-vertebral anomalies, blood disorders, hip dysplasia), predicts complications, identifies associated systemic disorders, and enables genetic counseling. Radiological evaluation can also prompt targeted treatment, for example enzyme

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replacement in mucopolysaccharidoses (MPS) or newer therapies in achondroplasia [2].

Key clinical clues for suspecting skeletal dysplasias

Paediatric orthopaedic surgeons should look for disproportion and dysmorphism. A high index of suspicion is important, since many dysplasias have evolving radiographic features that may only become pronounced as the child grows. Common clinical clues to skeletal dysplasia include the following:

Head and face

- Macrocephaly
- Frontal bossing
- Mid-face hypoplasia
- Blue sclerae / corneal clouding

Neck and spine

- Short neck
- Cervical spine abnormalities (e.g. odontoid hypoplasia)
- Early / progressive scoliosis or kyphosis

Chest and trunk

- Small / narrow chest
- Organomegaly

Limbs

- Disproportionate short stature
- Limb shortening (rhizomelic, mesomelic, acromelic)
- Limb deformities
- Genu varum / valgum
- Joint laxity / contractures
- Congenital joint dislocations
- Recurrent fractures

Hands and feet

- Digital anomalies such as polydactyly, syndactyly and brachydactyly. Figure 1 shows a radiograph of a specific type of brachydactyly.

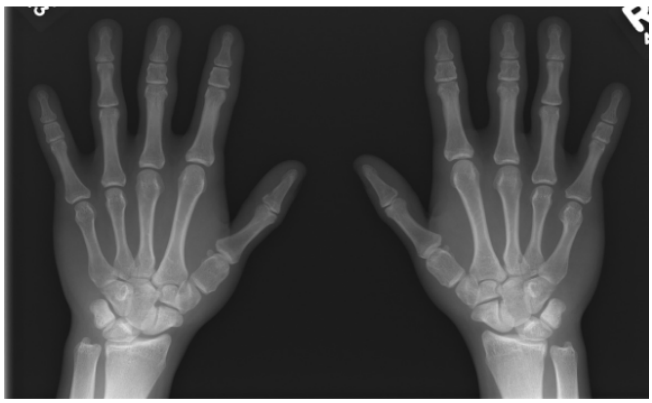

Figure 1: Radiograph of brachydactyly type C Shortening of middle phalanges of the index, ring and little fingers third digits. There is also shortening of the thumb metacarpal.

Any child with suspected skeletal dysplasia should undergo a complete skeletal survey [3]. This includes the following radiographs of:

- Skull (lateral and anteroposterior- AP)
- Lateral thoracolumbar spine
- Chest (AP): This includes the AP thoracic spine
- Pelvis (AP): This includes the AP lumbar spine
- One upper and one lower limb (AP) and one hand for bone age

This survey helps classify the pattern of involvement and pick up subtle findings. If asymmetrical limb involvement is found, views of both limbs should be obtained. If a condition associated with Wormian bones is suspected, consider obtaining a Towne's view of the skull. If a condition associated with odontoid hypoplasia is suspected, a dedicated lateral radiograph of the cervical spine should be obtained. Importantly, radiographs in early childhood are most diagnostic, since many dysplastic changes may disappear after skeletal maturity [4]. Bone age assessment is especially valuable for planning growth modulation procedures, evaluating suitability for growth hormone therapy, and determining the optimal timing of epiphysiodesis [5, 6].

Multidisciplinary assessment

A multidisciplinary approach is essential for the safe, timely, and accurate diagnosis of skeletal dysplasias. Given the complexity and overlap of many dysplasia phenotypes, it is vital to integrate clinical evaluation with targeted imaging, biochemical testing, and multidisciplinary input to ensure that both diagnosis and management are comprehensive and individualised. It is recommended for the following key reasons:

Accurate diagnosis

- Genetics input confirms specific mutations
- Endocrine review helps rule out metabolic bone disease or growth hormone disorders

Early detection of complications

- MRI/CT is essential for detecting cord compression (e.g. achondroplasia, Morquio)
- Ophthalmology/ audiology can detect early vision/ hearing loss in MPS, sclerosing bone dysplasias or collagenopathies
- Early referral to endocrinology is important in select dysplasias for timely initiation of growth hormone therapy to optimise final height

Avoiding misdiagnosis

- Blood tests (calcium, phosphate,alkaline phosphatase, vitamin D, parathyroid hormone) help differentiate dysplasia

from metabolic bone disease
- Urine GAGs may indicate MPS in ambiguous cases

Timely intervention
- Missed atlantoaxial instability can lead to neurological injury intra- or post-operatively
- Undiagnosed sclerosing dysplasias may cause cranial nerve palsies or complications due to marrow failure [7].

Planning long-term care
- Facilitates growth monitoring, surgical planning, and genetic counselling for families

Basic radiological terminology and measurements

The upper segment/lower segment (US/LS) ratio is used to assess body disproportion, with the upper segment measured from the pubis to the vertex of the head and the lower segment from the pubis to the floor. In healthy children, US/ LS decreases from ≈1.7 at birth to ~1.0 by age 10–11, US/ LS ratios decline from 1.05 at 4y to ~0.92 at 12y [8]. A markedly increased ratio (for example, >1.4 in a 5-year-old indicates relatively short lower limbs, as seen in achondroplasia, while a reduced ratio points to a short trunk, as in spondyloepiphyseal dysplasia. Below are some general radiographic descriptors:
- Stippling (punctate calcification): Tiny, spotty calcifications in cartilage/ epiphyses. Classically seen in chondrodysplasia puncta, where epiphyseal stippling is a characteristic finding in infancy [9]. This becomes less conspicuous with age. It can also be seen in foetal Warfarin syndrome.
- Overmodeled bones: Abnormally slender long bone shafts e.g. pycnodysostosis osteogenesis imperfecta, hypophosphatasia.
- Undermodeled bones: The long bones fail to show normal diaphyseal narrowing, instead demonstrating pronounced metaphyseal flaring with thin cortices, producing an Elenmeyer-flask configuration. Pyle's disease (metaphyseal dysplasia) is a classic example, characterised by wide, club-shaped metaphyses due to undermodelling [10].
- Epiphyseal fragmentation: Irregular or segmented appearance of epiphyseal ossification centers. Seen in conditions like pseudoachondroplasia or multiple epiphyseal dysplasia (MED), where epiphyses appear multiple or clefted on X-ray.
- Wormian bones: Accessory ossicles located within the cranial sutures, most commonly along the lambdoid suture. More than 10 Wormian bones suggest abnormal calvarial ossification (e.g. in osteogenesis imperfecta, pyknodysostosis) [11].
- Coronal/ sagittal clefts: Clefts in vertebral bodies on AP radiograph in the coronal or sagittal plane, respectively . e.g. chondrodysplasia punctata X-linked, Kniest dysplasia.
- Horizontal acetabula: Horizontal orientation of acetabular

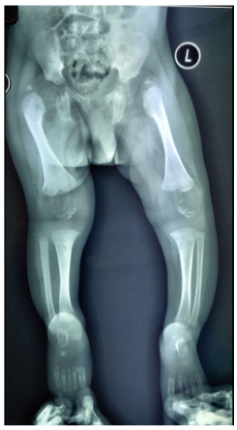

Figure 2: Radiograph of stippling seen in chondrodysplasia puncta Speckled calcification affecting the epiphyses of the acetabulum, proximal femora, and patellae, in keeping with stippled epiphyses.

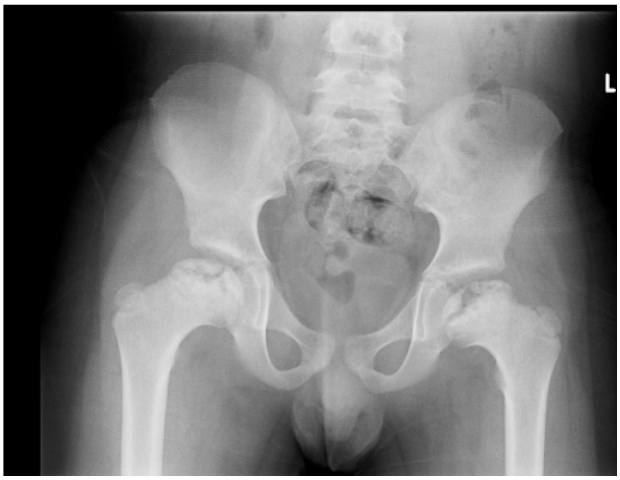

Figure 3: Radiograph of epiphyseal fragmentation seen in MED Coxa vara with flattening and fragmentation of bilateral proximal femoral epiphyses.

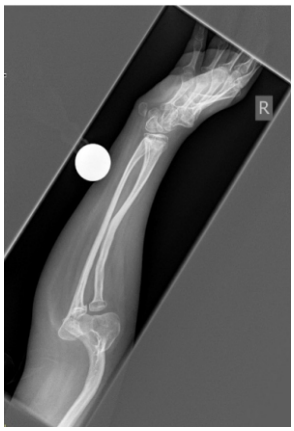

Figure 4: Radiograph of Cole Carpenter syndrome Gracile, overtubulated appearance to the radius, ulna and distal humerus. There is bowing of the distal humerus, radius and ulna. S-shaped appearance to the radius raises suspicion of previous fracture remodelling.

roofs is typical in achondroplasia.

● Short metacarpals/metatarsals: Apparent when a metacarpal/ metatarsal is significantly shorter than expected for age or relative to its neighbours. In Turner syndrome, the fourth metacarpal is classically short (the "metacarpal sign"); on X-ray a line drawn through the heads of metacarpals 4–5 fails to reach metacarpal 3 head [12]. Similar indices or age-norm charts can be used quantitatively (e.g. radiogrammetric metacarpal index). Brachymetatarsia (shortened 4th or 5th metatarsal) yields a toe that does not reach the level of adjacent metatarsals on standing radiographs. (Fig. 2, 3, 4)

Common skeletal dysplasias in paediatric orthopaedics

These are a few common dysplasias often encountered in paediatric orthopaedic practice, with their radiological hallmarks, distinctive clinical signs, and imaging recommendations.

Achondroplasia (FGFR3-related rhizomelic dwarfism)

Clinical signs: Disproportionate short stature from birth; large head with frontal bossing; depressed nasal bridge; lumbar lordosis with exaggerated kyphosis when sitting. Ligaments are lax (leading to genu varum or spinal stenosis). Trident hand: the 2nd and 3rd digits are separated creating a trident configuration.

Radiographic findings:

● Skull: Small posterior fossa (narrow foramen magnum), frontal bossing (thickened cranial vault) and midface hypoplasia.

● Spine: Hypoplastic odontoid peg, increased lumbar lordosis with exaggerated thoracic kyphosis. Hypoplastic first lumbar (L1) vertebra. Lumbar pedicles converge caudally – the interpedicular distance narrows from L1 to L5. The vertebral bodies may demonstrate posterior scalloping or assume a bullet-shaped configuration.

● Thorax: Short ribs with flattening of AP diameter

● Hand: Short metacarpals, bullet shaped phalanges, trident hand, seen in 45-50% of cases [13].

● Pelvis: The pelvis is narrow and tall at the base with flared iliac wings, giving an '18th century' champagne glass appearance. Square iliac wings, horizontally oriented acetabular roofs, and narrowed sacrosciatic notches. Trident acetabula at younger age due to medial and lateral spurs.

● Long bones: Marked rhizomelic shortening of all long bones (humerus more than femur and humerus/femur more than radius/ ulna); metaphyseal flaring and splaying with relatively normal looking epiphyses. Prominent trochanters. Genu varum and overriding fibulae. Varus ankle. The distal femur and proximal humerus show a "chevron" physis as the metaphyseal 'V' embraces the epiphysis [14].

Special imaging: Foramen magnum compression causing cervical cord signal change may occur in 14% of cases in the first two years (5-10%) [15]. MRI of the cervicomedullary junction is recommended during infancy to evaluate for foramen magnum stenosis and associated spinal cord compression [16]. These infants are predisposed to sudden death as a result of cervicomedullary compression secondary to a narrow foramen magnum, a hypoplastic odontoid process and ligament laxity and this risk can be exacerbated by neck extension during anaesthesia or surgical positioning. Imaging is thus recommended before any procedure under general anaesthesia. MRI would be more beneficial in infants. Lateral flexion-extension films are more reliable after 2-3 years of age, when ossification of the first (C1) and second (C2) cervical vertebrae is complete. Serial lateral spine films are recommended to assess for thoracolumbar kyphosis.

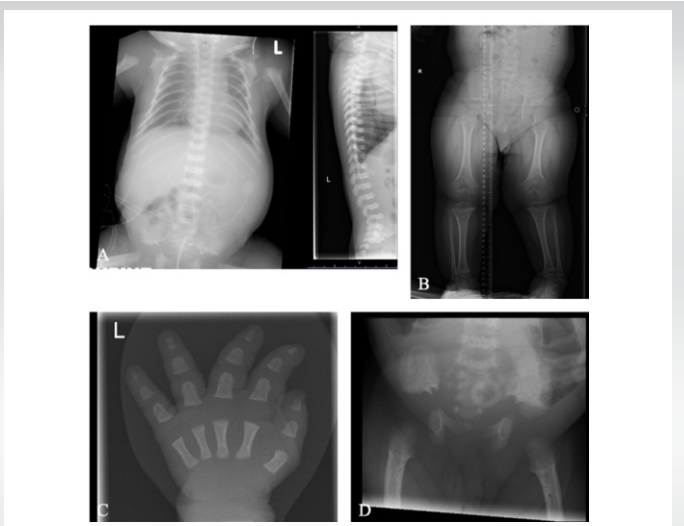

Figure 5: Radiographic features of Achondroplasia (A-D) A: AP neonatal chest and abdomen (babygram) radiograph and a lateral spine radiograph demonstrating multiple short, flat, bullet shaped vertebral bodies. B: Leg length xray in a preschooler demonstrating notched (v-shaped) growth plates, at the knees and ankles but most pronounced at the distal femurs. The other features include narrowing of the interpedicular distance, squared iliac wings and flat acetabular roofs. C: DP view of the left hand in a neonate demonstrating multiple broad and relatively short metacarpals and phalanges. D: AP pelvis xray of a neonate demonstrating champagne glass appearance of the pelvis, squared iliac wings, short narrow sciatic notch and flat acetabular roofs

Differentials: Hypochondroplasia (a milder FGFR3 mutation) has similar but subtler findings and often normal skull base (genetic testing distinguishes them). Thanatophoric dysplasia (lethal) shows extreme limb shortening (telephone receiver femora) and cloverleaf skull, and can be ruled out by genetic testing and postnatal survival [17]. Spondyloepiphyseal dysplasia congenita (SEDC) can also present with short stature; however, trunk involvement is more pronounced, with marked platyspondyly and coxa vara, and FGFR3 mutation testing is negative. (Fig. 5)

Hypochondroplasia (mild FGFR3 short stature)

Clinical signs: Short stature typically becomes apparent in late infancy or childhood (often normal at birth); disproportion is

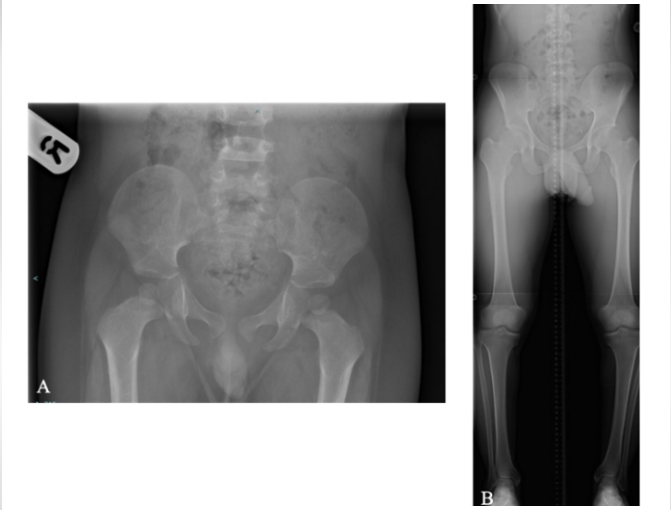

Figure 6: Radiographic features of hypochondroplasia (A-B) A: Pelvic radiograph of a grade schooler demonstrating short iliac bones with flat acetabular roofs and wide-appearing proximal femoral diaphysis. B: Leg length radiograph demonstrating subtle decrease in the interpedicular distance from first to fifth lumbar vertebrae, flat acetabular roof, mild flaring of metaphyseal–epiphyseal junction of the femurs and tibiae, short and broad femoral necks and rectangular proximal tibial epiphyses.

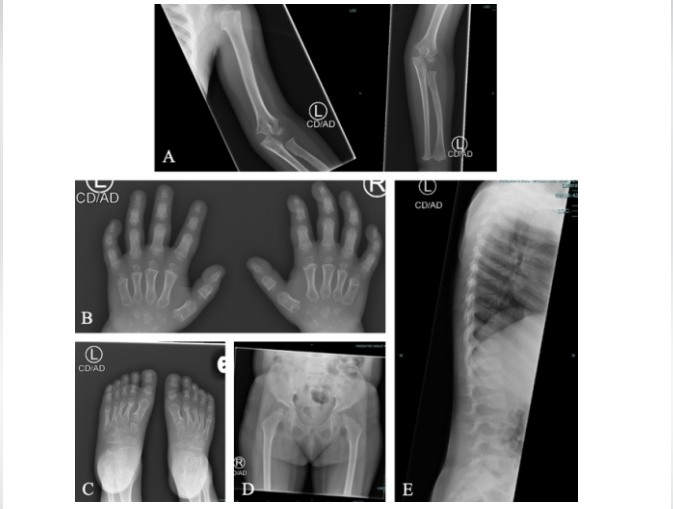

Figure 7: Radiographic features of Pseudoachondroplasia (A-E) A: Chest and upper limb radiographs demonstrating fragmentation and irregularities of the developing epiphyses. B and C: Both hands and feet radiographs demonstrating small epiphyses, irregular metaphyses; hypoplastic, abnormally contoured carpal and tarsal bones. There is also rounding at the proximal ends of metacarpals on the hand xray. D: Pelvic radiograph demonstrating dysplastic acetabuli with rounded ilia and small proximal femoral epiphyses. E: Lateral spine radiograph demonstrating irregularity and exaggeration of (superior and inferior) epiphyseal grooves of the vertebral end plates

milder. Facial features are largely unremarkable, with no obvious frontal bossing. Intelligence is normal.

Radiographic findings: Essentially a milder form of achondroplasia. There is mild rhizomelic shortening of the long bones with subtle metaphyseal flaring. Skull base changes and pedicle narrowing are less pronounced or absent compared to achondroplasia. Radiographs may appear nearly normal or only subtly dysplastic.

Special imaging: Asymptomatic infants do not require routine MRI (unlike achondroplasia). Standard dysplasia skeletal surveys may show only borderline findings. Confirmation of the diagnosis often depends on molecular analysis of the FGR3 gene, most commonly identifying the N540K mutation [18].

Differential: Achondroplasia (distinguished by severity and skull base involvement). Other causes of familial short stature (Leri-Weill with Madelung deformity, Turner syndrome) can mimic mild hypochondroplasia, but these have different limb proportions and genetic tests. (Fig. 6)

Pseudoachondroplasia (COMP-related spondylo-epiphyseal dysplasia)

Clinical signs: Normal facial appearance and head size. Short stature becomes apparent by age 2–4. Joint laxity leads to early onset osteoarthritis (knees, hips, ankles) and gait disturbances [19]. Ligament laxity may cause genu varum or valgum, "windswept" knees. Intelligence is normal.

Radiographic findings: Radiographic changes typically become evident after 2-3 years of age, with disproportionate limb shortening emerging in early childhood; affected children are often normal at birth.

- Skull: Normal
- Spine: Changes are common: vertebral bodies can be oval ("codfish" vertebrae) or have anterior beaking or platyspondyly and odontoid hypoplasia.
- Hand: Proximal pointing of metacarpals
- Pelvis: Flattened femoral heads and dysplastic acetabula in nearly all cases.19 Strikingly wide triradiate cartilage. Pelvis is relatively preserved (no champagne-glass pelvis).
- Long bones: The epiphyses of the long bones, particularly at the hips and knees, are small, irregular and fragmented. Metaphyses are flared or "trumpeted," and long bones may be mild-moderately shortened. Genu varum/ valgum/ windswept knees may be present due to ligament laxity. Osteoarthritis changes early in life [19].

Special imaging: Spine films are important due to risk of kyphosis/ scoliosis. MRI or CT is warranted when spinal cord compression is suspected, as anterior vertebral beaking may reduce the spinal canal diameter.

Differential: MED also causes small epiphyses, but MED generally has milder short stature, symmetric hip involvement, and no spinal deformities, whereas pseudoachondroplasia often has platyspondyly/ kyphosis. Achondroplasia can be excluded by the presence of normal skull and pelvic anatomy in pseudoachondroplasia. Genetic testing of COMP can confirm pseudoachondroplasia (vs. MATN3/ COL9 in MED) [20]. (Fig. 7)

MED (autosomal dominant and autosomal recessive types)

Clinical signs: Milder short stature than in pseudo-achondroplasia; short trunk and short limbs may be present. Patients typically have a waddling gait, with joint pain and early

onset osteoarthritis often developing during adolescence [21]. Hands and feet are usually of normal length, although joint contractures may restrict mobility. Facial features and intelligence are normal.

Radiographic findings: Bone age is usually delayed by a maximum of 6 to 8 years, regardless of measurement method [22].

- Skull: Normal
- Spine: Radiographs are typically normal (unlike spondylo-epi dysplasias), but may have irregularities of the end plates.
- Hands: Cone-shaped pseudoepiphysis in metacarpals and phalanges
- Pelvis: femoral head epiphysis are always involved with Perthes like changes- fragmentation and flattening, coxa magna and wide necks [21]. By late childhood, secondary osteoarthritis (especially at hips) may appear [21].
- Long bones: Delayed and irregular ossification of epiphyses in many joints, classically hips and knees. Distal femoral and proximal tibial epiphysis may be small, fragmented or flattened. Metaphyses are usually normal (distinguishing MED from metaphyseal dysplasias). Leg-length equality is usually maintained.

Radiographic differences among MED subtypes:

- MED1 (COMP): Both knees and hips are equally affected
- MED2 (COL9A2): Deformities are more around the knees with lateral femoral epiphysis fragmented. MED 2 is less affected than MEDtype 1.
- MED3 (COL9A3): Knees are the most severely involved, and capital femoral epiphysis shows greater lateral flattening.
- MED4 (SLC26A2): Flattening of femoral head is more pronounced medially than laterally

Differential: Pseudoachondroplasia – MED has no spinal deformity and lacks ligament laxity, whereas pseudoachondroplasia has spinal changes (codfish vertebrae) and more severe short stature. Acetabular and femoral shape: MED often has coxa vara, while pseudoachondroplasia has severe acetabular dysplasia. Genetic panels can distinguish. (Fig. 8)

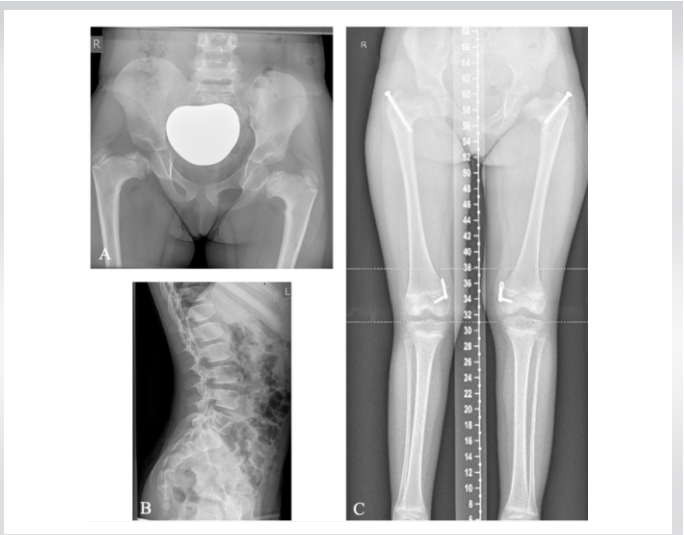

Figure 8: Radiographic features of MED (A-C) A: Coxa vara with flattened, fragmented and sclerosis proximal femoral epiphyses. B: Mild vertebral endplate irregularity. No anterior wedging. C: Greater trochanter and distal femoral epiphysiodesis. Coxa vara with irregular capital femoral epiphysis

Spondyloepiphyseal dysplasia congenita (SEDC, type II collagenopathy)

Clinical signs: Short-trunk dwarfism with very short stature and disproportionately longer limbs. Characteristic short neck and trunk. cleft palate, myopia, hearing loss (midline collagen defect). Atlantoaxial instability from odontoid hypoplasia is common. Cleft palate is present.

Radiographic findings:

- Skull: Mid-face hypoplasia
- Spine: Odontoid hypoplasia with possibility of Atlanto-axial dislocation, vertebral bodies may appear bulbous or flat (platyspondyly) in infancy (anisospondyly- irregularly shaped vertebrae with varying heights).
- Hands: Delayed ossification of carpals
- Long bones: Delayed ossification of capital femoral with a "truncated" (sliced off) appearance is characteristic; they may not appear until 4-5 years; absent capital epiphysis with severe coxa vara and trochanteric over-riding may give the false appearance of dislocated hips [23]. Metaphyses are usually normal, but epiphyses (femoral head, proximal femur) are small.
- Pelvis: Horizontal acetabular roofs, short broad iliac wings, and frequently absent pubic rami. Radiologically, the proximal segments of the limbs are markedly affected, with a gradual decrease in severity distally. Consequently, the humeri and femora are most markedly affected, whereas the hands and feet are comparatively spared.

Special imaging: Early lateral cervical spine films and/ or MRI are mandatory because of odontoid hypoplasia and atlantoaxial instability [24]. Eye and ear evaluation should be obtained. Orthopaedic surveillance focuses on spine (kyphoscoliosis) and hip development (risk of early osteoarthritis).

Differential: In contrast to achondroplasia, which is characterised by short limbs and macrocephaly, SEDC presents with short trunk and small thorax while head size is typically normal. Radiologically, achondroplasia has pelvic and skull changes absent in SEDC. Morquio syndrome (MPS IV) can mimic SEDC radiographically (both have platyspondyly), but MPS has other features (corneal clouding, characteristic radiologic "central beaking" of vertebrae, and lab findings) [25].

Diastrophic dysplasia (SLC26A2-related, autosomal recessive)

Clinical signs: Onset at birth. Cleft palate, clubfoot deformities, and cauliflower ears are frequently observed. Pectus carinatum,

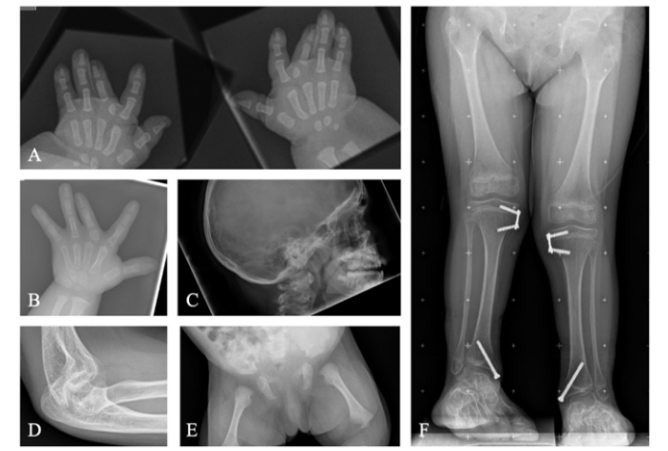

Figure 9: Radiographic features of Diastrophic Dysplasia (A-F) A: Bilateral delta phalanx of the middle finger proximal phalanx. B: Shortened, ovoid thumb metacarpal with a proximally located thumb. Longitudinal bracketed epiphysis of the middle finger proximal phalanx. Supernumerary ossicle at the proximal portion of the second metacarpal proximal phalanx. C: Platyspondyly and hypoplasia of the mid to lower cervical spine, C3-6. D: Dysplastic elbow with radial head dislocation. E: Monkey wrench appearance of the femora with absence of the proximal femoral epiphyses, horizontally dysplastic acetabulae and a tongue-like projection of the lesser trochanters. F: Leg length discrepancy with bilateral proximal and distal tibial epiphysiodesis. Coxa magna with bilateral flattened, sclerotic proximal femoral epiphyses.

spinal deformity (scoliosis, kyphosis) develops, as does cervical kyphosis with risk of cord compression. Intelligence is normal.

Radiographic findings:

- Skull: Normal
- Spine: Dysplastic odontoid process, cervical kyphosis, abnormal shaped vertebrae, progressive scoliosis, posterior scalloping of vertebrae, absent vertebrae.
- Hands: Short and irregular lengths of metacarpals, short first metacarpal - hitchhiker's thumb [26].
- Long bones: Undermodelled short bones, chevron shaped distal femora, irregular epiphysis and metaphysis, double-layered patellae, and dislocated radial heads.
- Pelvis: Flat acetabulum and flared iliac wings, coxa vara or valga and hypoplastic pubis.

Special imaging: Newborn screening spine films should include a lateral cervical view for early kyphosis. MRI of the cervical spine is indicated to evaluate the cervicomedullary junction, as atlantoaxial instability can occur.

Differential: Pseudoachondroplasia can be differentiated by the presence of hitchhiker thumbs and associated cleft palate or ear anomalies. Achondroplasia is ruled out by the characteristic hands and spinal changes. (Fig. 9)

Osteogenesis imperfecta (type I collagen defect)

Clinical signs: Varying presentation based on the sub-types. Frequent long-bone and rib fractures (sometimes leading to misdiagnosis of non-accidental trauma). Joint laxity, scoliosis, hernia and easy bruising. Blue sclerae and dentinogenesis imperfecta are clues. Hearing loss.

Radiographic findings: Profound osteopenia with multiple fractures in different stages of healing. Generalised osteosclerosis may occur following bisphosphonate therapy

- Skull: Thin cortex, wide anterior fontanelle and Wormian bones
- Spine: Scoliosis, platyspondyly, multiple wedge fractures, biconcave (codfish) vertebrae due to central end-plate depression
- Chest: Slender ribs with fractures
- Long bones: Slender bones with thin cortices, multiple fractures which are more common in lower limbs, bowing, angular deformities at shaft level rather than at joint level, limb length discrepancy. Bulb like widening of metaphysis with pop-corn appearance in the metaphysis and epiphysis (seen in type III and IV OI) [27]. Hyperplasticity callus and interosseous ossification seen with fractures in type V. Zebra lines in metaphysis: dense metaphyseal bands following bisphosphonate therapy.
- Pelvis: Narrow pelvic inlet, triradiate pelvis due to protrusio acetabuli and coxa vara or valga.

Special imaging: Follow-up radiographs to monitor healing fractures. DEXA scan is generally not needed for diagnosis but is a valuable tool as a baseline measure before treatment initiation and for monitoring response to treatment. It is also used as a surrogate end-point in clinical trials of novel modalities in OI [28].

Differentials: an inconsistent and/ or concealed history and a poor or withdrawn child should raise the suspicion of non

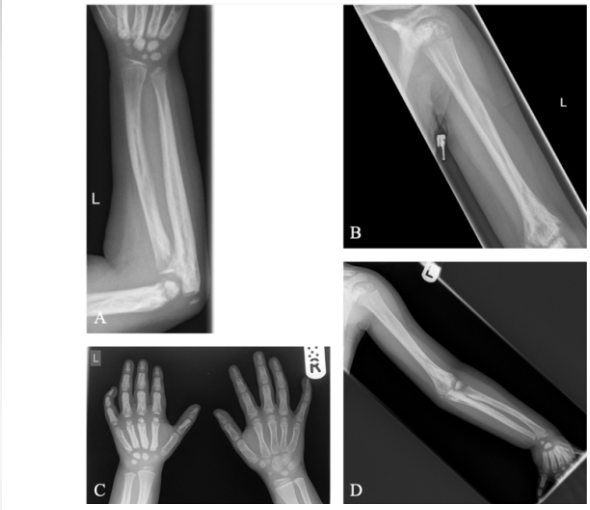

Figure 10: Radiographic features of sclerosing bone dysplasia (A-D) A: Cortical thickening to the left radial and ulnar diaphyses, sparing the metaphyses. Partial sclerosis of the humeral head and elbow ossification centres, little finger, thumb and trapezium. B: Cortical thickening to the humerus, left radial and ulnar diaphyses, sparing the metaphyses. Partial sclerosis of the humeral head and elbow ossification centres. C: Asymmetrical distribution, only affecting the left side. Dense, thickly striated appearance to nearly all phalanges and metacarpals of the index to little fingers and carpal bones. Partial radial sided sclerosis to the little finger metacarpal, partial ulnar sided sclerosis to the thumb digit and distal radial epiphysis. Sparing of the trapezium. Cortical thickening to the left radial and ulnar diaphyses, sparing the metaphyses. D: Cortical thickening to the humerus, left radial and ulnar diaphyses, sparing the metaphyses. Partial sclerosis of the humeral head and elbow ossification centres, little finger, thumb and trapezium.

accidental injury. Cole-Carpenter Syndrome has more marked cranio-facial features with marked proptosis, mid-face hypoplasia and macrocephaly. Genetic testing (COL1A1 or COL1A2) or family history confirms OI [29].

Sclerosing bone dysplasias (Fig.10)

Osteopetrosis (marble bone disease)

Clinical signs: lethal in severe types, short stature in severe types, developmental delay, pancytopenia and anemia, hepatosplenomegaly, thickened cranial base can lead to cranial nerve palsies, poor dentition, fracture of teeth and small teeth, fractures from trivial injuries.

Radiographic findings: radiographs show diffuse sclerotic bones.

- Skull: Skull base is thickened and fontanelle closed
- Spine: "Rugger jersey" spine due to sclerotic band at the superior and inferior end plates
- Thorax: Thick and sclerotic ribs
- Hands: "Bone-within-bone" or endobone appearance
- Long bones: Club shaped undermodelled metaphysis, obliterated medullary canal, fractures with delayed or non-union, loss of distinction between cortical and cancellous bones
- Pelvis: Arcuate sclerotic bands in the iliac wings

Pycnodysostosis

Clinical signs: slightly short stature with mild limb shortening, pathological fractures, stubby finger tips, prominent frontal and occipital skull, small mid-face, micrognathia, delayed tooth eruption and mandibular osteomyelitis.

Radiographic findings: diffuse osteosclerosis with characteristic acro-osteolysis which is resorption of distal phalanges of fingers and toes.

- Skull: Wormian bones
- Spine: Segmentation defects, spondylolysis
- Thorax: Short clavicles
- Hands: Acro-osteolysis
- Long bones: Overmodelled and slender diaphyses
- Pelvis: Supra-acetabular constriction

Differentials: Pycnodysostosis must be differentiated from osteopetrosis by its acroosteolysis and open sutures [30].

Melorheostosis

It is a segmental sclerosing dysplasia. Radiographs show a flowing pattern of hyperostosis along the cortex ("dripping candle wax" appearance) in one limb or sclerotome distribution. It is typically unilateral, affecting long bones or adjacent joints with severe cortical thickening. Unlike osteopetrosis or pycnodysostosis, melorheostosis produces exuberant cortical hyperostosis and may cause deformity or limb-length discrepancy. It is usually diagnosed clinically by the classical radiographic pattern.

Morquio syndrome (MPS IV – lysosomal storage disorder)

Clinical signs: Disproportionate short stature with short trunk, corneal clouding, hearing loss, and normal intelligence, pigeon-chest (pectus carinatum), tracheal abnormalities, hepatosplenomegaly, joint laxity and genu valgum.

Radiographic findings: Delayed ossification of several bones

- Skull: Relatively normal
- Spine: Hypoplastic odontoid peg is universal and associated with instability, platyspondyly with anterior beaking- usually in the centre of the body, kyphosis/ gibbus.
- Thorax: Acute manubrio-sternal angle, oar-shaped ribs
- Hands: Bullet-shaped phalanges, metacarpals are pointed proximally
- Long bones: Flared and irregular metaphysis, delayed ossification of lateral proximal tibial epiphysis and metaphysis.
- Pelvis: Flared iliac wings with shallow sciatic notch and inferior ilium is a hallmark, coxa valga

Special imaging: MRI/ CT of the cervical spine is essential to evaluate for cord compression from odontoid hypoplasia. Serial spine films to assess for kyphosis. Echocardiogram may be needed for valve disease.

Differential: SEDC can appear similar (both have short trunk and platyspondyly) but SEDC has near-normal intelligence and facial structure, whereas Morquio has the storage phenotype. Genetically they differ (GALNS enzyme deficiency in Morquio, COL2A1 mutation in SEDC) [25,31]. Other MPS disorders such as Hurler and Hunter, typical present with coarse facies and mental impairment, not seen in Morquio and can be differentiated radiologically and biochemically. (Fig. 11)

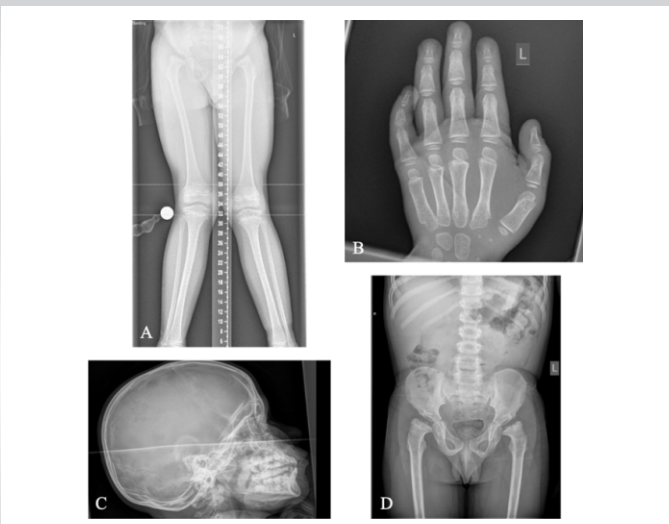

Figure 11: Radiographic features of Morquio syndrome (A-F) A: Coxa valga. Constricted iliac wings, steeply oblique acetabular roofs with absence of the ossified femoral heads. Genu valgus. B: Proximal pointing of the second through fifth metacarpals. C: Mildly dolichocephalic skull (longer than wide). D: Coxa valga. Constricted iliac wings, steeply oblique acetabular roofs with small fragmented proximal femoral epiphyses, irregularity and flaring of the metaphyses.

Hurler syndrome (MPS I) – MPS with dysostosis multiplex

Clinical findings: Coarse facies with macroglossia and frontal bossing with hearing loss and corneal clouding. Eventually develop short stature, joint contractures, hepatosplenomegaly and hernia. Cardiopulmonary complications later in life, hip dislocations, thoracolumbar kyphosis. It is a progressive disease with death occurring from cardio-pulmonary complications, if untreated.

Radiographic findings: Generalised osteopenia

- Skull: Sagittal synostosis- large, scaphocephalic skull with thick calvarium, J-shaped and elongated sella turcica, short mandible
- Spine: Biconvex or flat vertebrae with anterior-inferior beaking of vertebral body, kyphosis/ gibbus with apex usually at 2nd lumbar vertebra
- Thorax: Paddle-shaped ribs, hypoplastic clavicles
- Hands: Short and stubby, bullet-shaped phalanges, metacarpals are pointed proximally
- Long bones: Undermodelled bones with shafts as wide as or wider than the metaphysis
- Pelvis: Small pelvic inlet with flared iliac wings with shallow sciatic notch and inferior ilium is a hallmark, coxa valga and sloping acetabulae.

Metaphyseal chondrodysplasia, Schmid Type (COL10A1 mutation)

Clinical signs: Can be mild to severe based on the number of metaphysis involvement. Mild short stature due to short lower limbs, genu varum (more common) or valgum, and waddling gait. Usually presents in early childhood with progressive knee deformity and lumbar lordosis.

Radiographic findings: Abnormal metaphyses with splaying and flaring

- Skull: Normal
- Spine: Normal, may be oval shaped
- Thorax: Normal
- Long bones: Normal diaphysis and epiphysis and wide and splayed or cupped metaphysis, genu varum.
- Pelvis: Coxa vara, medial beaking of proximal femoral metaphysis; Fairbank's triangle may be visible in more severe cases. In McKusick dysplasia, the metaphyseal changes are most prominent after 2 years at the knees, sparing the hips [32].

Differentials: X-linked metaphyseal dysplasia (XX-MED) – distinguished by X-linked inheritance and more severe limb shortening. Nutritional rickets – has similar metaphyseal cupping, but rickets shows abnormal lab values (low calcium/ phosphate, elevated alkaline phosphatase) and generalized bone demineralization, whereas Schmid type has normal labs and otherwise normal bone density. SEDC has involvement of capital femoral epiphysis and more severe coxa vara. (Fig. 12)

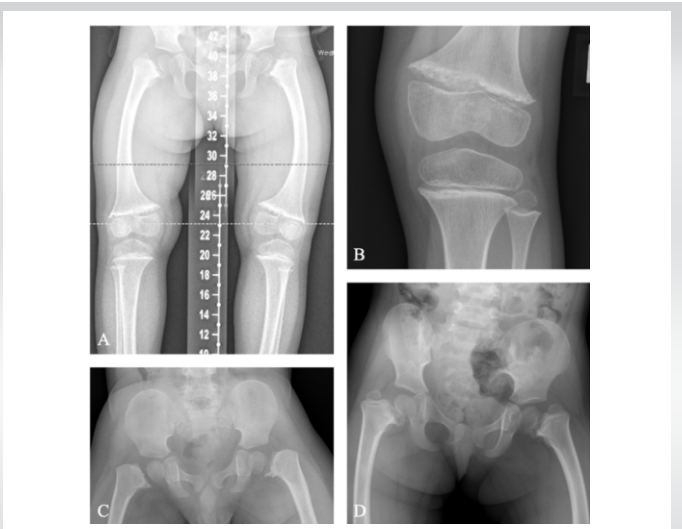

Figure 12: Radiographic features of metaphyseal chondrodysplasia, Schmid Type (A-D) A: Diffuse metaphyseal flaring, irregularity and physeal widening at the hips, knees and ankles. Coxa vara with bowing of the femora. B: Diffuse metaphyseal flaring, irregularity and physeal widening involving the distal femur, proximal tibia and fibula but most prominent at the distal femur C: Diffuse metaphyseal flaring, irregularity and physeal widening at the hips. Coxa vara. D: Diffuse metaphyseal flaring, irregularity and physeal widening at the hips. Coxa vara.

Imaging guidelines for specific dysplasias

While plain radiography is the mainstay, certain dysplasias require advanced imaging or special views.

- Achondroplasia: In addition to routine survey, obtain cranial MRI (or CT) in infancy to screen for foramen magnum stenosis and cervicomedullary compression. MRI spine or craniovertebral junction is also recommended if neurologic symptoms arise. Regular lateral spine films (seated vs standing) monitor kyphosis.
- SEDC: Always image the upper cervical spine (odontoid) with MRI or CT before surgical intervention, given the risk of atlantoaxial instability. Hip surveillance is necessary.
- Pseudoachondroplasia and MED: Radiographs usually suffice for diagnosis and follow-up. MRI of symptomatic knees or spine can assess early cartilage damage.
- Diastrophic Dysplasia: As noted, lateral cervical films and/ or MRI are critical to detect early cervical kyphosis or cord compression. Hand/ wrist films document the fixed thumb and carpal bone anomalies.
- OI: In addition to standard radiographic evaluation, genetic counselling plays a crucial role. Imaging is generally for fractures; no routine MRI/ CT is needed. Whole-body bone density scans are occasionally used for monitoring of treatment following bisphosphonate therapy.
- Morquio Syndrome (MPS IV): Perform regular MRI of the cervical spine (flexion-extension) to assess for atlantoaxial subluxation due to odontoid hypoplasia. Lateral spine X-rays to assess kyphosis. Hip/ knee MRI may show cartilage loss.
- Metaphyseal Dysplasia (Schmid): Periodic knee and hip radiographs to guide deformity management.
- Bone Age & Growth Monitoring: A left hand/wrist

radiograph should be part of the initial workup to assess skeletal maturity and growth potential. This is particularly important for planning interventions (growth modulation or epiphysiodesis).

Prenatal ultrasound features of skeletal dysplasia

Foetal ultrasound can strongly suggest dysplasia. Key clues include extremely short long-bone lengths (e.g. femur or humerus <2 SD for gestational age and <4 SD for lethal dysplasias [33], poor mineralisation, bowed or fractured long bones, narrow thorax, abnormal skull shape and presence of polydactyly, syndactyly or brachydactyly. Femoral or humeral length below the 5th percentile on a routine 18–22 week scan warrants referral for targeted dysplasia evaluation. Early MDT involvement and prenatal counselling for prognostication and management should be provided. If suspicion is high, dedicated high-resolution fetal sonography (often with 3D) should assess all bones and joints. Next steps include foetal MRI (useful to assess lung volume in lethal forms) and invasive genetic testing (such as targeted gene panels or exome sequencing by amniocytes) for confirmation. Fetal CT (low-dose) may also be performed in specialized centers after 26 weeks for definitive bone detail. Importantly, prenatal diagnosis allows planning for respiratory support or delivery in a tertiary care center when lethal dysplasias are identified.

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