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DOI- <https://doi.org/10.13107/ijpo.2025.v11.i01.220>
www.ijponline.com

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The Hip in Skeletal Dysplasia: Evaluation & Management

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Abstract

Skeletal dysplasias represent a heterogeneous group of over 500 genetic disorders affecting the growth, development, and structural integrity of bone and cartilage. Hip deformities in skeletal dysplasia (S.D) represent a significant challenge in pediatric orthopaedic practice. They significantly impact mobility and quality of life in children with S.D. Despite advances in surgical techniques and implant technology, patients with skeletal dysplasia continue to face higher complication rates and more challenging management decisions.

Early identification and intervention can prevent progressive deformity and secondary complications. A multidisciplinary approach combining careful preoperative planning, specialised surgical techniques, and dedicated rehabilitation protocols offers the best opportunity to optimise outcome.

Keywords: Skeletal Dysplasia, Hip deformities, Natural History, Management, Outcomes.

Introduction

Skeletal dysplasias represent a heterogeneous group of over 500 genetic disorders affecting the growth, development, and structural integrity of bone and cartilage [1]. With a prevalence of approximately 1 in 4,000 live births, most conditions are identifiable early in life and persist throughout the lifespan [1]. Common orthopaedic presentations include short stature, axial and appendicular deformities, joint surface irregularities, ligamentous laxity, and limb length discrepancies. These features frequently lead to pain, mobility limitations, and decreased quality of life. Understanding the natural history and surgical implications of these disorders is essential for tailoring interventions that optimize function and minimize long-term complications [2, 3].

Hip deformities in skeletal dysplasia (S.D) represent a significant challenge in pediatric orthopaedic practice. They significantly impact mobility and quality of life in children with S.D. These complex deformities arise from underlying genetic abnormalities that affect bone and cartilage development, thereby impacting the child's skeletal growth. Each skeletal dysplasia presents unique hip pathoanatomy requiring individualized management approaches. Early identification and intervention can prevent progressive deformity and secondary complications. A multidisciplinary team approach is ideal to manage these hip deformities in S.D. patients [4].

Epidemiology:

Hip problems are highly prevalent in individuals with skeletal dysplasia, though the exact rates vary depending on the specific condition and diagnostic criteria. (Table 1)

Age: Age of presentation varies with the skeletal dysplasia type & severity. (Table 2)

Submitted: 13/01/2025; Reviewed: 08/02/2025; Accepted: 15/03/2025; Published: 10/04/2025

Dysplasia Type	Hip Pathology	Prevalence / Risk	Clinical Notes
Achondroplasia (Figure 1)	Shallow acetabulum, limited abduction	50-80% ⁵ Moderate risk of DDH and early OA	Often presents with lumbar hyperlordosis and a waddling gait
Spondyloepiphyseal Dysplasia	Epiphyseal irregularities, acetabular dysplasia	90-95% ⁶ High risk of hip subluxation/dislocation	May require early surgical intervention; associated with short trunk dwarfism
Multiple Epiphyseal Dysplasia	Delayed ossification, joint incongruity	70-90% ⁷ , Moderate to high risk of early OA	Pain and stiffness often emerge in adolescence
Pseudoachondroplasia (Figure 2)	Coxa vara, acetabular dysplasia	More than 90% ⁹ , High risk of progressive hip degeneration,	Often misdiagnosed early, hip pain is a common presenting symptom
Diastrophic Dysplasia	Coxa vara, joint contractures	More than 80% ⁸ , High risk of hip dislocation	Associated with clubfoot and scoliosis; early bracing or surgery may be needed
Larsen Syndrome (Figure 3)	Congenital dislocations, ligamentous laxity	80% ¹⁹ Very high risk of DDH	Hip dislocation often presents at birth; surgical correction is frequently required
Osteogenesis Imperfecta (Figure 11)	Joint laxity, Coxa vara, acetabular dysplasia, Protrusio acetabuli	25-55% ²⁰ Variable; depends on type	Fracture risk complicates surgical management; careful imaging is essential
Campomelic Dysplasia	Hip dislocation, abnormal femoral shape	75% High risk; often bilateral	Associated with respiratory issues and sex reversal in some cases

Table 1: Dysplasia type

Regional Variation:

Higher reported incidence of diastrophic dysplasia in Finland [21], Ellis-van Creveld syndrome in Amish populations [22]. Diagnostic capabilities and recognition patterns vary by region, affecting reported prevalence. Access to genetics testing and specialized care also influences both diagnosis and management outcomes across regions.

Classification of Hip Deformities in Skeletal Dysplasia

Hip deformities in skeletal dysplasia can be categorized as:

Acetabular dysplasia: Shallow, oblique, or malformed acetabulum. Common in SED, MED, and diastrophic dysplasia

Protrusio Acetabuli: Is a pathologic deepening of the

Age	Type of Skeletal Dysplasia
Congenital ¹⁰	Diastrophic dysplasia, Thanatophoric dysplasia, Severe forms of SED.
Early childhood (1-3years) ¹¹	Pseudoachondroplasia, MED, and moderate forms of SED
Mid-childhood (4-8 years) ⁵	Progressive hip contractures and femoral anteversion in Achondroplasia
Adolescence ⁴	Some milder forms of S.D.

Table 2: Age and type of Skeletal Dysplasia

Deformity	Skeletal dysplasia
Acetabular dysplasia	Diastrophic dysplasia MED Morquio syndrome
Coxa vara (Figure 5)	Cleidocranial dysplasia MED Pseudoachondroplasia SED congenita Schmid metaphyseal chondrodysplasia
Coxa Valga	Morquio syndrome
Hip degenerative	Pseudoachondroplasia, MED, SED

Table 3: Deformity and Skeletal Dysplasia

acetabulum, where the femoral head pushes inward into the pelvic cavity.

Femoral deformities Common in Osteogenesis Imperfecta:

Coxa vara (common in MED, chondrodysplasia punctata)

Coxa valga (seen in achondroplasia)

Femoral anteversion (prevalent in achondroplasia)

Femoral neck hypoplasia (characteristic SED)

Metaphyseal irregularities (seen in metaphyseal chondrodysplasia)



Figure 1: Anteroposterior radiograph of the pelvis with both hips in a 16-year-old male with achondroplasia showing broad, short femoral necks, horizontal acetabular roofs with a trident pelvis – flared iliac wings and narrow sciatic notches bilaterally.

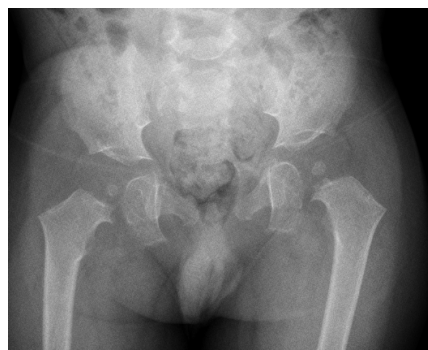


Figure 2: Pelvic X-ray of a child with Pseudoachondroplasia showing dysplastic acetabulum and proximal femur.



Figure 3: Pelvic X-ray of a child with Larsen Syndrome showing bilaterally dislocated proximal femur with small acetabular sockets.



Figure 4: Pelvic X-ray of a child with Metaphysio-Epiphyseal Dysplasia showing irregular femoral head and high riding greater trochanter.



Figure 5: Pelvic X-ray of a child with Spondylo-Metaphyseal Dysplasia showing severe Coxa Vara with high riding greater trochanters.



Figure 6: Pelvic X-ray of a child with SED Tarda showing arthritic Left hip joint.



Figure 7 a, b: Preoperative anteroposterior and lateral radiographs in a 6 yo patient with Spondylometaphyseal dysplasia showing bilateral ossified femoral heads with significant coxa vara and trochanteric overriding.

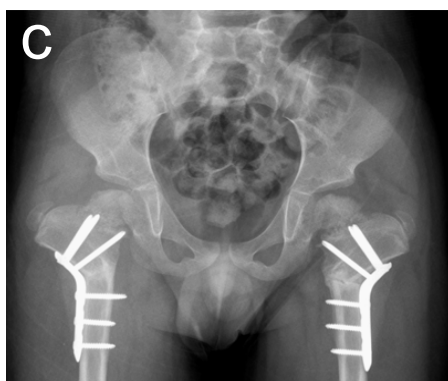


Figure 7 c, d: Postoperative anteroposterior and lateral radiographs showing a healed proximal femoral valgus, derotation osteotomy fixed with a proximal femoral locking plate and screws. Greater trochanteric overriding has been corrected. Note that the physis is now more horizontal compared to the preoperative position.

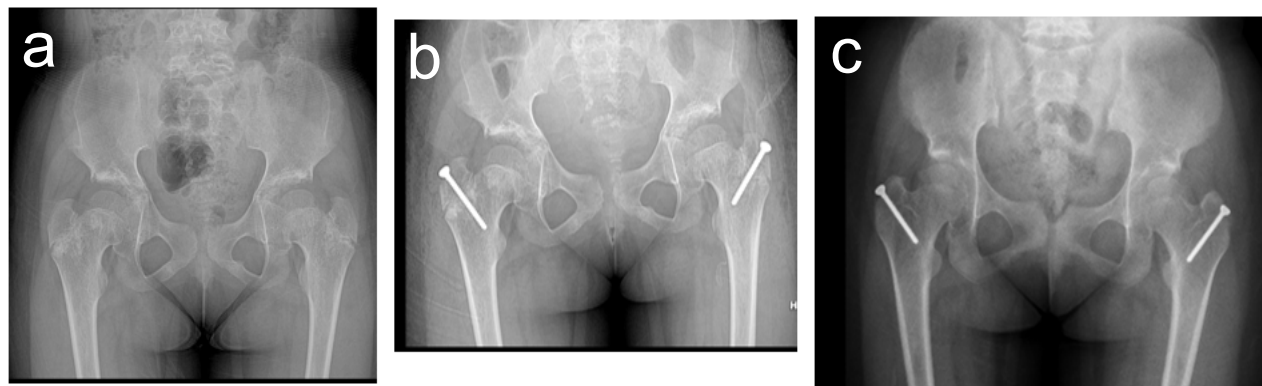


Figure 8 a, b, c: **Figure 8a:** Pelvic X ray of a 9 year old child with coxa vara secondary to SpondyloMetaphyseal Dysplasia. **Figure 8b:** Bilateral trochanter epiphysiodesis done with a percutaneous screw to treat coxa vara and high riding greater trochanter. **Figure 8c:** 5 year follow up of the same child showing well preserved proximal femoral anatomy.

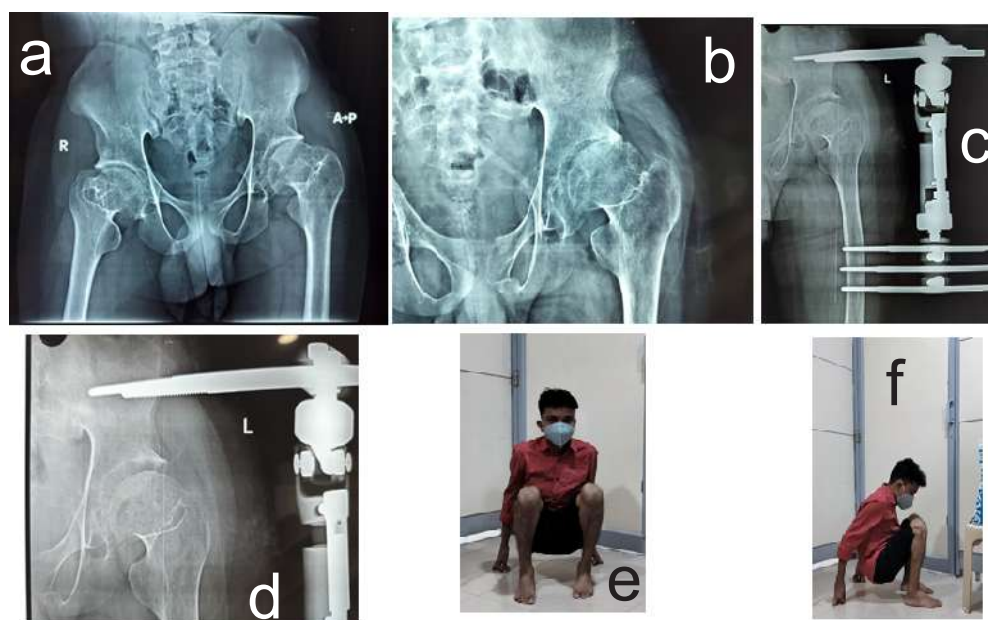


Figure 9: A 13 year old child with SED Tarda. **9a and 9b:** X ray showing arthritic Left hip with decreased left hip joint space. **9c and 9d:** X rays of the same patient showing distraction of the left hip joint using arthrodiastasis. **9e and 9f:** Clinical images of the same child after 1 year showing apin free squatting with support.

Cartilage and Labral abnormalities:

Abnormal cartilage composition and structure.
Labral hypertrophy or hypoplasia.

Progressive deformities:

Primary deformities present at birth.
Secondary deformities develop with growth and weight-bearing. (Table 3)

Diagnosis: A comprehensive clinical evaluation should include:

Detailed history:

Family history of skeletal dysplasia or disproportionate short stature
Developmental milestones, particularly regarding gross motor

skills

Onset and progression of symptoms (pain, limp, decreased mobility)

History of previous treatment or interventions

Associated symptoms in other organ systems

Physical examination:

Growth parameters (height, weight, head circumference)

Body proportions and limb-to-trunk ratio

Spine examination (kyphosis, lordosis, scoliosis)

Lower limb alignment

Hip range of motion assessment:

Presence of contractures

Trendelenburg test and gait analysis

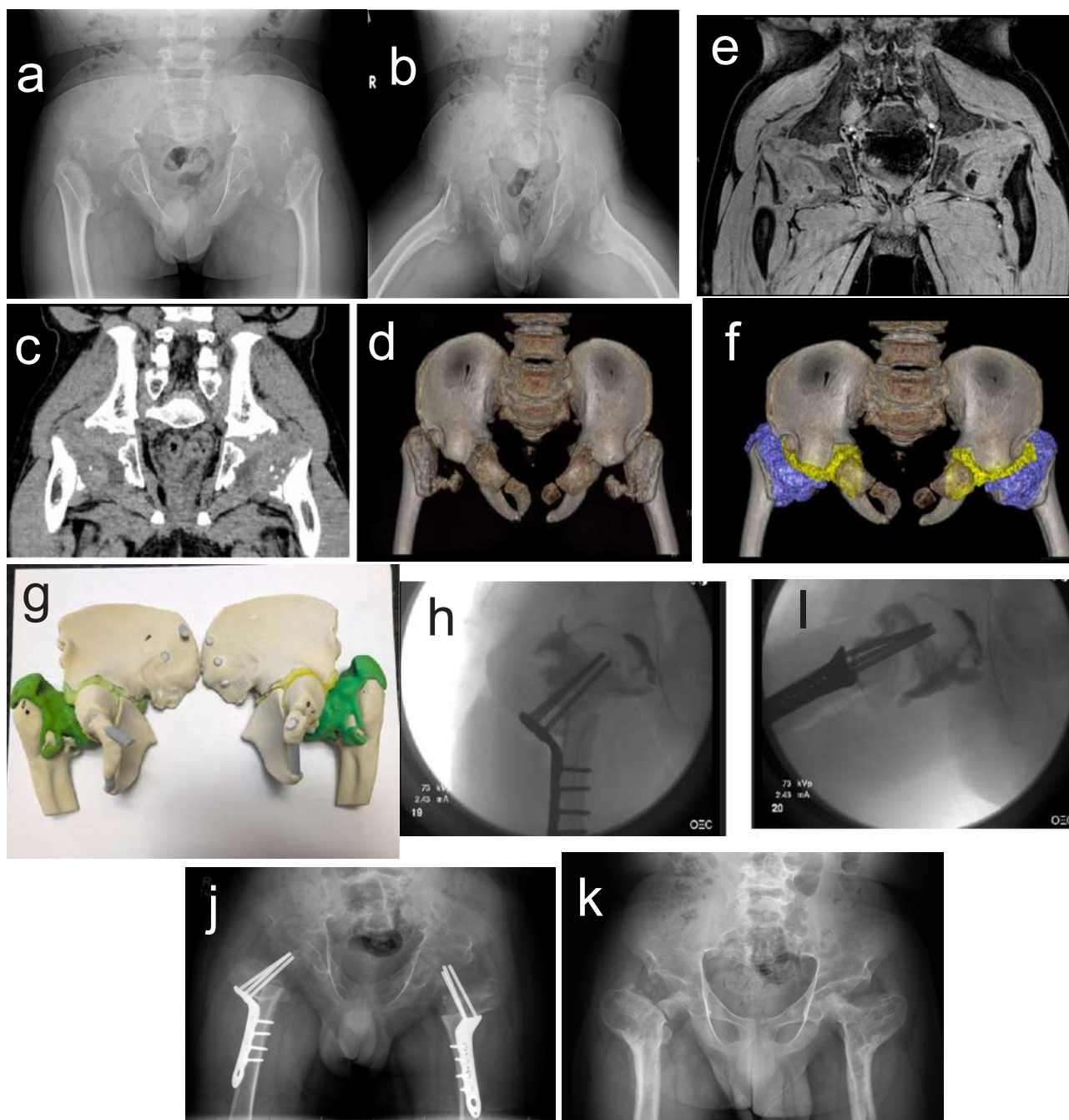


Figure 10: Spondyloepiphyseal Dysplasia (SED) **a, b:** Preoperative anteroposterior and lateral radiographs showing bilateral severe coxa vara with incompletely ossified and dysplastic femoral heads with greater trochanteric overriding. **c, d:** CT pelvis without contrast in the coronal plane (A) & CT with 3D reconstruction (B) showing markedly delayed ossification of the hips, and elevated greater trochanters are seen bilaterally. Dysplastic appearance of the acetabula with flattening of the acetabular roofs laterally. Abnormal cartilage is difficult to completely assess using the soft tissue window. **e:** Coronal MRI pelvis without contrast WATSC Protocol. There is a dysmorphic appearance of the femur, with high-riding metaphyses, due to coxa vara. Cartilage appears dysplastic and thickened using the WATS-c (water-selective cartilage protocol). **f:** Composite CT/MRI Rendering of pelvis with proximal femora, with 3D Reconstruction developed by fusing CT/MR images using Phillips Intellispace Portal and CT Viewer Software Program. **g:** Composite CT/MRI 3D Reconstructed Model of the patient's pelvis, which was used for patient education and pre-operative planning. **h, i:** Intraoperative anteroposterior and lateral fluoroscopy images (A, B) showing use of an arthrogram to delineate the femoral head during surgery with a proximal femoral valgus, extension, and derotation osteotomy fixed with a proximal femoral locking plate. **j:** Post-operative anteroposterior and lateral radiographs of the pelvis with both hips showing a healed proximal femoral valgus osteotomy fixed with a 3.5 mm locking proximal femoral plate bilaterally. Coxa vara and greater trochanteric overriding have been corrected. **k:** Postoperative 2-year anteroposterior radiograph showing healed proximal femoral osteotomies following hardware removal with incompletely ossified femoral heads.

Radiological Assessment

Plain radiographs:

AP pelvis: Provides assessment of acetabular development, femoral head position and morphology.

Frog-leg lateral: Evaluates femoral head coverage and femoral neck-shaft angle.

Standing full-length Radiograph of the lower extremities: Assesses overall limb alignment, and limb length discrepancy.

Advanced imaging:

CT-Scan: (Computer Tomography)

- . Provides detailed bony anatomy.
- . Helpful for surgical planning.
- . 3D reconstruction and 3D printed models aid understanding of complex deformities, family education, preoperative planning of corrective osteotomies and to create patient specific implants [12].

MRI: (Magnetic Resonance Imaging)

- . Evaluates cartilaginous structure not visible on radiographs.
- . Assesses labral integrity.
- . Evaluates articular cartilage thickness and quality.
- . Identifies early degenerative changes.
- . Evaluates soft tissue abnormalities around the hip.

Genetic Testing

Genetics testing has become essential in the diagnose and classification of skeletal dysplasias [13]:

- . Next -generation sequencing panels targeting known skeletal dysplasia genes
- . Whole exome sequencing for cases without identified mutations on targeted panels
- . Specific gene testing guided by clinical and radiographic feature:
 - . FGFR3 testing for suspected achondroplasia
 - . COL2A1 testing for suspected SED

- . COMP testing for suspected pseudoachondroplasia or MED

Goals of treatment

Treatment goals must be individualized based on the specific dysplasia type, severity of deformity, age of the patient, and functional limitations:

- . To maintain or improve hip joint congruity and stability
- . To prevent progressive deformity and secondary complications
- . To alleviate pain and improve function
- . To optimize mobility and independence
- . To delay the onset of degenerative changes
- . To preserve long-term hip function
- . To accommodate for the altered biomechanics of the associated skeletal dysplasia

Other considerations

- . Consider impact of interventions on future growth
- . Balance between corrective procedures and functional needs
- . Address pain and functional limitations affecting education and socialization
- . Preparation for transition to adult care
- . Vocational and quality of life considerations

Individualized approaches

Treatment strategies must be tailored to the specific dysplasia type and associated deformities:

Achondroplasia: Focus on managing flexion contractures and addressing functional limitations [23].

SED: Early intervention for Coxa vara, progressive hip dysplasia and subluxation to delay onset of osteoarthritis [24]. (Fig. 6, 9)

MED: Monitoring and intervention for coxa vara and early degenerative changes [25]. (Fig. 4)

Diastrophic dysplasia: Early management of hip dislocation to optimize function [26].



Figure 11 a: Anteroposterior radiograph in a 10-year-old girl with Type 3 Osteogenesis imperfecta showing bilateral coxa vara with retained Fassier-Duval rods.



Figure 11 b: Postoperative anteroposterior radiograph of the pelvis with both femora showing healed bilateral proximal femoral valgus osteotomies fixed with Steinmann pins and cerclage wires around revised Fassier-Duval rods.

Non-Surgical Management

Physical Therapy and Rehabilitation

It plays a critical role in managing hip deformities [14, 15]. It includes:

- . Range of motion exercise to prevent contracture
- . Strengthening exercise targeting hip stabilizers (abductors and extensors)
- . Gait training and mobility optimization
- . Functional training for activities of daily living

Orthotic Management

Orthotic interventions can provide support and prevent progression of deformity. It includes:

- . Abduction bracing for hip instability in young children
 - . Night positioning devices to manage contractures
 - . HKAFO (Hip-Knee-Ankle-Foot Orthosis) for severe instability
 - . Custom seating systems to accommodate hip deformities
 - . Gait assistive devices tailored to specific biomechanical needs
- Pain Management

Comprehensive pain management should include:

- . Activity modification strategies
- . Non-steroidal anti-inflammatory medications
- . Physical modalities (heat, cold therapy)
- . Weight management to reduce joint stress
- . Judicious use of intra-articular injections in selected cases.

Surgical Management

Timing of surgical Intervention

The optimal timing for surgical intervention varies based on:

- . Skeletal dysplasia type and natural history.
- . Severity and progressive rate of deformity.
- . Presence of symptoms affecting function and quality of life.

Early intervention is generally indicated for:

- . Progressive subluxation or dislocation
- . Severe acetabular dysplasia
- . Progressive coxa vara/valga affecting joint stability
- . Significant functional limitations or pain

Types of Surgical Interventions

Soft Tissue Procedures

- . Adductor and iliopsoas tenotomies for contracture management
- . Open reduction for dislocated hips with soft tissue interposition
- . Capsular plication for instability
- . Arthroscopic labral repairs or debridement for symptomatic labral tears

Acetabular Procedures

. Pelvic osteotomies:

- . Salter innominate osteotomy for younger children with acetabular deficiency
- . Pemberton or Dega osteotomy for deficient acetabular roof
- . Triple innominate osteotomy for older children with global acetabular deficiency
- . Periacetabular osteotomy (PAO) for adolescents and young adults
- . Shelf procedures for lateral coverage

Femoral Procedure

- . Proximal femoral osteotomies
- . Varus osteotomy for hip instability or excessive coxa valga
- . Valgus osteotomy for severe coxa vara
- . Derotational osteotomy for excessive femoral anteversion
- . Shortening osteotomy to reduce joint pressure in containment procedure

Combined Procedures

Many hip deformities in skeletal dysplasia require combined approaches

- . Combined femoral and acetabular procedure for complex deformities
- . Staged approaches for severe deformities in young children
- . Concurrent addressing of extra-articular deformities affecting hip biomechanics

Salvage Procedures

For severe deformities or failed previous surgeries

- . Hip arthrodesis (rarely used now but historically important)
- . Excision arthroplasty and Total hip arthroplasty for end-stage disease
- . Hip resurfacing in selected adolescent or young adult cases.
- . Pelvic support osteotomy.

Considerations specific to skeletal dysplasia:

- . Thinner and often more brittle bone requiring modified fixation strategies
- . Altered pelvic anatomy requiring careful preoperative planning
- . Potential for increased blood loss due to abnormal vascularity
- . Modified techniques to accommodate the unique anatomical variations
- . Smaller femoral canals affecting implant selection
- . Altered metaphyseal bone quality affecting fixation
- . Modified entry points and angles for fixation devices
- . Need for custom implants in severe deformities

Preoperative planning

Detailed preoperative planning is crucial in skeletal dysplasia cases:

- . Comprehensive radiographic assessment including 3D imaging when indicated
- . Template-based planning for implant sizing and positioning [16].
- . Computer-assisted planning for complex osteotomies
- . Consideration of future growth and remodeling potential
- . Anticipation of anatomical challenges specific to the dysplasia type
- . Assessment of bone quality

Anaesthesia Considerations

. Airways concerns [17]:

- . Midface hypoplasia and mandibular abnormalities affecting intubation
- . Potential for cervical spine instability requiring careful positioning
- . Tracheal stenosis in specific dysplasia

. Respiratory considerations:

- . Restrictive lung disease due to thoracic deformities
- . Sleep apnea requiring perioperative management
- . Increased risk of postoperative respiratory complications

. Cardiovascular issue:

- . Associated cardiac anomalies in certain dysplasia types
- . Altered cardiovascular response to positioning
- . Careful fluid management and blood loss estimation

. Neurological considerations:

- . Potential for spinal cord compression requiring neuromonitoring
- . Modified positioning to protect vulnerable neurological structures
- . Consideration of regional anaesthesia techniques

Complications

Complication rates are generally higher in skeletal dysplasia patients compared to those with isolated hip dysplasia:

General complications:

- . Increased blood loss (30-45% higher than in non-dysplastic cases)
- . Higher infection rates (3-7%)
- . Wound healing problems (5-10%)
- . Neurovascular injuries (2-5%)

Procedure-specific complications [18]:

- . Fixation failure (8-15%)
- . Delayed union or non-union (7-12%)
- . Implant-related complications (10-12%)
- . Heterotopic ossification (5-15%)

- . Avascular necrosis (5-10%)
- . Recurrent deformity (15-40%)

Long-term consideration:

- . Accelerated degenerative changes
- . Need for revision procedures
- . Conversion to total hip arthroplasty at younger ages
- . Functional limitations despite surgical intervention

Hips Deformities in Common Skeletal Dysplasias

Achondroplasia [23]

- . Focus on managing flexion contracture rather than joint instability
- . Careful assessment of lumbar lordosis contributing to apparent hip flexion
- . Consideration of limb lengthening and its effect on hip biomechanics
- . Managing obesity to reduce joint stress and improve function

Spondyloepiphyseal Dysplasia [24] (Fig. 7, 8)

- . Early intervention for progressive hip dysplasia
- . Consideration of physeal growth disturbance in surgical planning
- . Custom implants often required due to small femoral canals
- . High risk of recurrence requiring staged procedures

Multiple Epiphyseal Dysplasia [25]

- . Monitoring for progressive coxa vara
- . Addressing acetabular dysplasia when present
- . Earlier degenerative changes requiring consideration of joint preservation strategies
- . Variable progression rates between genetics subtypes

Diastrophic Dysplasia [26]

- . Severe acetabular dysplasia requiring comprehensive reconstruction
- . Multiple joint involvement affecting rehabilitation potential
- . Custom approaches for the distinctive hip anatomy
- . Higher complication rates requiring careful risk-benefit assessment

Multidisciplinary Approach

Comprehensive care requires coordination between:

- . Paediatric orthopaedic surgeons
- . Medical geneticists
- . Development paediatricians
- . Physical and occupational therapists
- . Orthotists and prosthetists
- . Pulmonologists for respiratory optimization
- . Anaesthesiologists familiar with skeletal dysplasia
- . Social workers and educational specialists

. Pain management specialists

Psychosocial Considerations

- . Body image concerns related to short stature and deformity
- . Peer relationships and social integration
- . Educational accommodation and vocational planning
- . Family adaptation and support systems
- . Transition to independent living
- . Long-term care planning and social support systems

Outcomes:

Future Directions

Emerging approaches include:

- . Improved genetics diagnose enabling targeted molecular interventions
- . Growth modulation techniques for progressive deformities
- . Advances in custom implant design using 3D printing technology [27]. (Fig. 10)
- . Biological approaches to enhance cartilage repair and regeneration

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Conclusion

The management of hip deformities in skeletal dysplasia requires a comprehensive understanding of the unique pathoanatomy and natural history of each dysplasia type. Treatment strategies must be individualised, considering the specific deformity pattern. Functional limitation, and long-term prognosis. A multidisciplinary approach combining careful preoperative planning, specialised surgical techniques, and dedicated rehabilitation protocols offers the best opportunity to optimise outcome. Despite advances in surgical techniques and implant technology, patients with skeletal dysplasia continue to face higher complication rates and more challenging management decisions.

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Declaration of patient consent : The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given the consent for his/ her images and other clinical information to be reported in the journal. The patient understands that his/ her names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Conflict of interest: Nil **Source of support:** None

How to Cite this Article

Gupta G, TR Easwar, Shah H, Belthur MV | The Hip in Skeletal Dysplasia: Evaluation & Management | *International Journal of Paediatric Orthopaedics* | January-April 2025; 11(1): 29-38