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When Bones Tell the Story: Skeletal Manifestations of Mucopolysaccharidosis in Two Pediatric Patients

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ABSTRACT

Mucopolysaccharidoses (MPS) are inherited lysosomal storage disorders characterized by progressive skeletal abnormalities collectively known as dysostosis multiplex. Imaging plays a central role in recognizing these changes and guiding diagnosis.
We report two pediatric cases investigated for suspected MPS with characteristic skeletal involvement demonstrated on radiography and computed tomography.
The first case involved a 16-year-old patient presenting with congenital lower limb deformities and patellar instability. CT revealed femoral diaphyseal bowing, metaphyseal–epiphyseal enlargement, trochlear dysplasia, and lateral patellar dislocation.
The second case concerned 5-year-old child, in whom conventional imaging demonstrated paddle-shaped ribs, cervical vertebral body deformities, pelvic asymmetry, and superior dislocation of the right femoral head.
These findings were consistent with dysostosis multiplex and highlight the importance of imaging in supporting early diagnosis of mucopolysaccharidosis.

KEYWORDS

Mucopolysaccharidosis, Dysostosis multiplex, Pediatric skeletal deformities, Patellar instability

MAIN ARTICLE

INTRODUCTION

Mucopolysaccharidoses (MPS) are a group of rare inherited lysosomal storage diseases resulting from enzymatic deficiencies responsible for the degradation of glycosaminoglycans. The accumulation of these substances within tissues leads to progressive multisystem involvement, among which skeletal abnormalities represent one of the most characteristic and early detectable features.

The constellation of bone and joint abnormalities observed in MPS is referred to as dysostosis multiplex. These changes affect both axial and appendicular skeletons and may include vertebral deformities, hip dysplasia, long bone bowing, and joint instability. Imaging therefore plays a fundamental role not only in raising diagnostic suspicion but also in assessing disease severity and complications [1,2].

We report two pediatric cases illustrating typical skeletal manifestations of mucopolysaccharidosis identified through different imaging modalities.

CLINICAL INFORMATION

Case 1

A 16-year-old patient with suspected mucopolysaccharidosis was referred for imaging evaluation due to congenital lower limb deformities, including left genu valgum and right genu varum, associated with patellar instability.

Computed tomography of both knees was performed to assess joint alignment and bone morphology.

Case 2

A 5-year-old child with no significant medical history presented with a two-month history of spontaneous left ankle pain associated with limping and relative functional impairment. There was no history of trauma.

Plain radiography demonstrated a cortical bone abnormality of the left leg, prompting further evaluation with computed tomography for lesion characterization.

IMAGING FINDINGS

Case 1

CT examination demonstrated bilateral femoral diaphyseal bowing with cortical thickening and metaphyseal–epiphyseal enlargement of the distal femur. Trochlear dysplasia was identified in both knees.

On the left side, lateral patellar dislocation was noted, associated with increased lateral translation and tilt. The proximal tibial epiphysis appeared reduced in height and irregular (Figures 1A,1B).

On the right side, trochlear dysplasia was present without patellar dislocation.

Additional findings included mild joint effusion and bilateral Hoffa fat pad infiltration. These skeletal abnormalities were consistent with dysostosis multiplex in the context of mucopolysaccharidosis.

Case 2

Plain radiographs revealed characteristic skeletal abnormalities including paddle-shaped ribs, defined by proximal narrowing and distal widening (Figure 2A)

Cervical vertebral bodies showed anterior deformity with a hooked or “sabot-shaped” appearance (Figure 2B). Pelvic asymmetry was noted, with misalignment of the obturator foramina. Superior dislocation of the right femoral head was also identified (Figure 3).

These radiographic findings were highly suggestive of skeletal involvement associated with mucopolysaccharidosis.

DISCUSSION

Skeletal abnormalities in mucopolysaccharidosis arise from abnormal cartilage and bone development due to glycosaminoglycan accumulation. Dysostosis multiplex represents the radiological hallmark of these disorders and involves multiple components of the skeleton. Axial involvement commonly includes vertebral body deformities such as anterior beaking, while thoracic abnormalities may manifest as paddle-shaped ribs. Pelvic and hip abnormalities frequently include acetabular dysplasia and femoral head subluxation or dislocation. Long bones may demonstrate metaphyseal widening and diaphyseal bowing, contributing to progressive limb deformities [2,3].

Joint involvement is also common and may result in instability, as observed in our first case with patellar dislocation and trochlear dysplasia. Such findings reflect the progressive structural alterations affecting articular surfaces and supporting structures.

Conventional radiography remains the first-line imaging modality for detecting global skeletal changes, while computed tomography provides detailed evaluation of joint morphology and alignment. The complementary role of these modalities allows accurate characterization of skeletal abnormalities and helps differentiate MPS from other causes of pediatric skeletal dysplasia [4].

Early recognition of imaging features is essential in guiding further biochemical and genetic investigations and in preventing delayed diagnosis.

CONCLUSION

Skeletal manifestations are a central feature of mucopolysaccharidosis and may provide the first diagnostic clue. Recognition of characteristic imaging findings such as vertebral deformities, hip dislocation, long bone bowing, and joint dysplasia is crucial for early suspicion of the disease.

These two cases illustrate the spectrum of skeletal involvement detectable through radiography and CT, emphasizing the key role of imaging in supporting diagnosis and guiding multidisciplinary management.

FIGURES

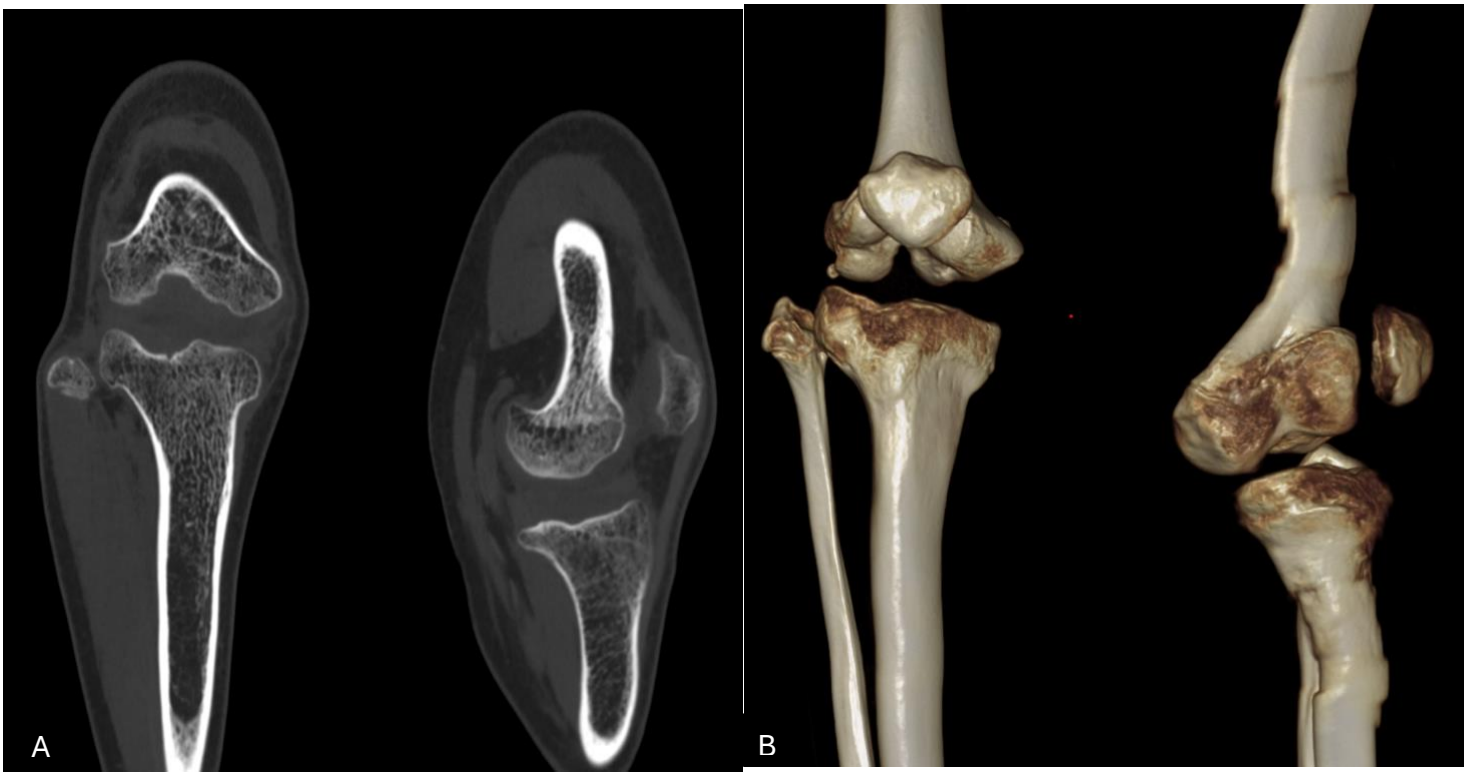

Figure 1: CT images (Case 1) demonstrating femoral diaphyseal bowing with metaphyseal enlargement and trochlear dysplasia.

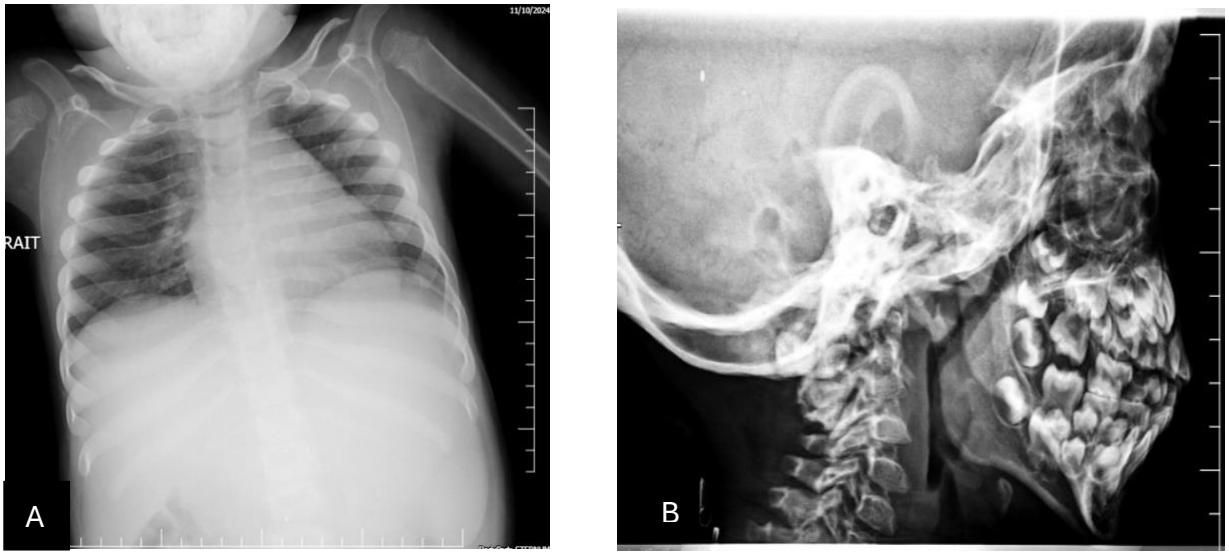

Figure 2: Radiograph (Case 2) showing paddle-shaped ribs and cervical vertebral body deformities.

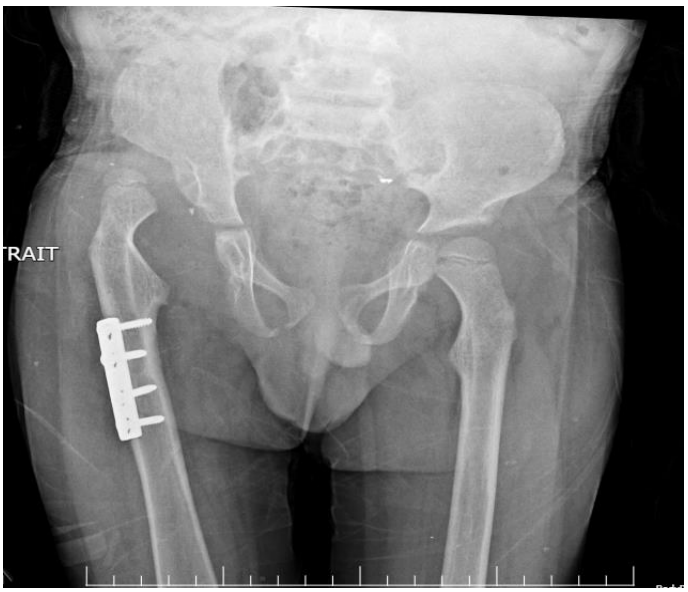

Figure 3: Pelvic radiograph (Case 2) illustrating asymmetry and superior dislocation of the right femoral head.

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REFERENCES

[1] White KK. Orthopaedic aspects of mucopolysaccharidoses. Rheumatology. 2020;59(Suppl 5):v49-v55. doi:10.1093/rheumatology/ker393.
https://doi.org/10.1093/rheumatology/ker393

[2] Muenzer J. Overview of the mucopolysaccharidoses. Rheumatology. 2019;58(Suppl 1):i4-i12. doi:10.1093/rheumatology/kez385.
https://doi.org/10.1093/rheumatology/kez385

[3] Hendriksz CJ, et al. Musculoskeletal manifestations of mucopolysaccharidoses. Journal of Inherited Metabolic Disease. 2019;42(5):1037-1048. doi:10.1007/s10545-019-00215-z.

[4] Ashworth JL, et al. Radiographic features of dysostosis multiplex. Pediatric Radiology. 2021;51(9):1592-1603. doi:10.1007/s00247-021-05000-0.