GORHAM disease: a paediatric case report
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Section: Paediatric radiology
Area of Interest: Spine Abdomen Bones
Procedure: Diagnostic procedure
Imaging Technique: Conventional radiography
Imaging Technique: CT
Imaging Technique: MR
Imaging Technique: Percutaneous
Special Focus: Haematologic diseases Demineralisation-Bone Cysts Case Type: Clinical Cases
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Patient: 3 years, male

Clinical History:

A 3-year-old boy had a 20 month history of spine deformity with no history of trauma or surgery. Physical examination showed a thoracic scoliosis without gait disorders. Standard blood laboratory tests were normal.

Imaging Findings:

Lateral and frontal spine X-ray views revealed patchy osteoporosis in thoracolumbar vertebrae and bone deformity associated with heterogeneous paraspinal opacity (Fig. 1). Thoraco-abdominal CT scan before and after contrast injection was first performed because of the unavailability of MRI and anaesthesia. This examination showed spine extensive osteolysis associated with a large non-enhancing soft tissue mass in the posterior mediastinum (Fig. 2 and 3). Bone involvement also concerned scapulae, ribs and pelvic girdle with soft tissue extension in the pelvic region (Fig. 4).

A body-MRI performed after CT revealed dissemination of the posterior mediastinum mass to paravertebral region (Fig. 5) and confirmed multifocal bony lesions (skull, scapulae, ribs, spine, left humerus, pelvic girdle (Fig. 6 to 8). Surgical biopsy in the left girdle region was performed. Anatomopathological and immunohistochemistry studies confirmed the diagnosis (Fig. 9 and 10).

Discussion:

Gorham’s disease is a rare entity that may affect any bone, particularly the scapular and pelvic girdles. Patient’s age has been reported to be from 1 month to 75 years of age with neither sex predilection nor hereditary predisposition [1, 2]. Although the exact aetiology of osteolysis is still unknown, it is considered to be due to nonmalignant, neoplastic proliferation of haemangiomatous or lymphangiomatous tissue [3, 4]. Unifocal involvement is usual, while multifocal involvement such as in our case is rarely observed. Clinical presentation depends on location, including sudden or insidious pain onset, pathological fractures, progressive functional limb disability and soft tissue atrophy. In our case and despite the multifocal involvement, the patient only had a painless spine deformity. Furthermore, the complications of this syndrome can be potentially fatal. For example, pleural effusion and chylothorax can dramatically influence the respiratory function. Chylothorax may occur due to the affected thoracic skeleton by the
extension of lymphangiectasia into the pleural cavity or by the invasion of the thoracic duct. Besides, haemangiomatous cutaneous lesions, bone infection, spinal cord involvement and paraplegia due to vertebral location, cerebrospinal fluid leakage and meningitis due to skull bone involvement, have been rarely reported [2]. Initial X-ray views reveal foci of intramedullary and subcortical lucency resembling patchy osteoporosis [4, 5]. At a later stage, bone deformity occurs with bone mass loss and concentric shrinkage in the long bones of upper and lower extremities. Near complete bone resorption can occur, resulting in the appearance of the so-called “vanishing bone” disease. CT is useful in the delineation of the soft-tissue extension and enables biopsy guidance. Three-dimensional CT reconstructions are valuable to the orthopaedic surgeon, planning a reconstruction attempt, better demonstrating the disease extent in complex structures [4, 5]. MRI is an adjuvant method with great variability in signal intensity and post-contrast enhancement, reflecting the spectrum of neovascular and fibrous progression [4, 5]. Despite the usefulness of the previously reported imaging modalities, the disease is confirmed by histopathological analysis of the lesions; biopsy shows nonmalignant hyperproliferation of small vessels. Differential diagnoses for Gorham’s disease include osteomyelitis, metastasis, osteolysis secondary to rheumatoid arthritis and hyperparathyroidism. Due to the rarity of this disease entity, there is no standard therapy available. The medical treatment for Gorham’s disease includes radiation therapy [6], anti-osteoclastic medication and alpha-2b interferon. The principal treatment modalities are surgery and radiation therapy. Prognosis for patients with Gorham’s disease is generally good unless vital structures are involved.

**Differential Diagnosis List:** Gorham’s disease, Osteomyelitis, Metastases, Hyperparathyroidism, Histiocytosis

**Final Diagnosis:** Gorham's disease

**References:**


Description: Frontal (1A) and lateral (1B) spine views demonstrating scoliosis (white arrows) with paravertebral opacity (black arrow). Origin: Ben Hassine L, Department of pediatric radiology, Tunis, Tunisia
Figure 2

Description: Axial thoraco-abdominal CT view with contrast showing a large non-enhancing soft tissue mass in the posterior mediastinum, presenting lobulated margins (white arrow). Origin: Ben Hassine L, Department of pediatric radiology, Tunis, Tunisia
Figure 3

Description: Sagittal CT views in soft tissue (3A) and bone windows (3B) showing extensive spine osteolysis responsible for thoracolumbar deformity (black arrows). Origin: Ben Hassine L, Department of pediatric radiology, Tunis, Tunisia
Figure 4

Description: Axial pelvic CT views in bone (4A) and soft tissue windows (4B) showing multiple lytic confluent lesions within the pelvic girdle (black arrow), muscular (black asterisk) and intracanalal extensions (white asterisk). Origin: Ben Hassine L, Department of pediatric radiology, Tunis, Tunisia
Figure 5

Description: Thoraco-lumbar T1 (5A), T2 weighted axial views (5B and 5C), demonstrating a liquid-like signal mass, white asterisk on T1, black asterisk on T2, posterior mediastinum and paravertebral region (black arrows), left pleural effusion (white arrow). Origin: Ben Hassine L, Department of pediatric radiology, Tunis, Tunisia.
Description: Sagittal T2 (6A), T1 before and after contrast injection (6B, 6C) views, showing spine deformity with vertebrae appearing in low signal on T1, heterogenous high signal on T2, visible enhancement after injection (white asterisk). Origin: Ben Hassine L, Department of pediatric radiology, Tunis, Tunisia
Description: Coronal T2 weighted image demonstrating multifocal bone dissemination with high signal appearance lesions of the scapulae (white arrow) and left humerus (white arrowhead). Origin: Ben Hassine L, Department of pediatric radiology, Tunis, Tunisia
Figure 8

Description: Axial T2 weighted views showing pelvic girdle hyperintense lesions (black asterisk, 8A) with intra-canalar extension (black arrowhead), acetabular involvement (8B, black arrow). 

Origin: Ben Hassine L, Department of pediatric radiology, Tunis, Tunisia
Figure 9

Description: (HE X 400) Vascular proliferation with anastomosing thin-walled lymphatic vessels between the remaining bony trabeculae. Origin: Abbassi I, Department of anatomopathology, Tunis, Tunisia
Description: Immunohistochemical positivity of the endothelial cells for the antibody D2-40. Origin: Abbassi I, Department of anatomopathology, Tunis, Tunisia