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Rare Case Report of Sprengel Deformity with Vertebral Fusion, Hemivertebra, Rib Fusion, and Spina Bifida Occulta

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Abstract **Case Report**

Sprengel deformity is a rare condition resulting from abnormal descent of the scapula during embryonic development. It is frequently misdiagnosed as scoliosis or overlooked when it occurs in conjunction with scoliosis. This deformity can be associated with several other conditions, including Klippel-Feil syndrome, omovertebra, spina bifida, rib anomalies, musculoskeletal dysfunctions, and tethered cord syndrome. Additionally, it may be linked to cardiac and renal anomalies, tracheoesophageal fistulas, anal atresia, absence of the pectoral muscles, and hand malformations. Our case of sprengel deformity with an omovertebral bar, scoliosis, hemivertebra, with concomitant fused vertebra, spina bifida occulta, fused and bifid ribs is an extremely rare combination. This further emphasizes the importance of a comprehensive evaluation for any patient with Sprengel shoulder to identify other potential anomalies. A thorough examination of all systems is essential.

Keywords: Sprengel; rare disease; shoulder; congenital undescended scapular syndrome; CT scan, Congenital high scapula, Sprengel's deformity.

INTRODUCTION

Although Sprengel deformity (SD) is the most common congenital shoulder abnormality, it is still a rare condition, with an incidence of 0.3 cases per 10,000 live births [1]. The severity of the disorder varies greatly, with some cases leading to significant functional limitations and cosmetic concerns [2, 3]. SD is frequently associated with other congenital anomalies, including Klippel–Feil syndrome, congenital scoliosis, renal abnormalities, and spina bifida, in a substantial percentage of cases. This association can complicate the physical examination at the time of presentation.

OBSERVATION

A 3 year old boy was referred for a consultation in pediatric orthopedic surgery for unsightly shoulder asymmetry and progressive reduction in the mobility of his left arm, Born to non-consanguineous parents; There were no systemic or constitutional symptoms. In birth history, the boy was first child of the family, born full-term via spontaneous vaginal delivery. Prenatal and postnatal history was unremarkable.

Physical examination showed asymmetry in both the shoulders. The left shoulder was higher than the right. On palpation, a hard swelling was noted over the left shoulder. Local signs of inflammation were absent. Both the active and passive movements were restricted in the left shoulder and crepitus was also presented. Abduction of the arm above 90 degrees elicited pain. Right shoulder examination was unremarkable.

A cervicothoracic CT scan in parenchymal and bone windows and 3d reconstructions showed elevation of the left scapula (Fig 1), an abnormal osseous connection between the medial border of the scapula and the left lamina of C7 vertebra was also seen (Fig 2). Imaging findings were consistent with an omovertebral bone with Sprengel deformity grade 2 of the Rigault classification.

And confirmed also the presence of cervical scoliosis, bifid and fused ribs (Fig 3), hemivertebra and fusion of the C2–C3 left posterior hemiarches (Fig 4), and revealed midline spinous process cleft of C6, D1, D2 and D3 (Fig 5). The diagnosis of Sprengel's deformity was made and a surgery is scheduled in the coming months, mainly for aesthetic reasons.

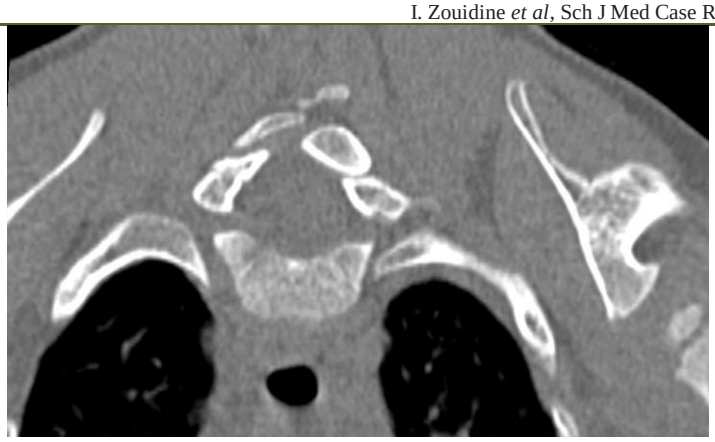

Fig 1: **Coronal CT scan image shows the elevation of the left scapula**

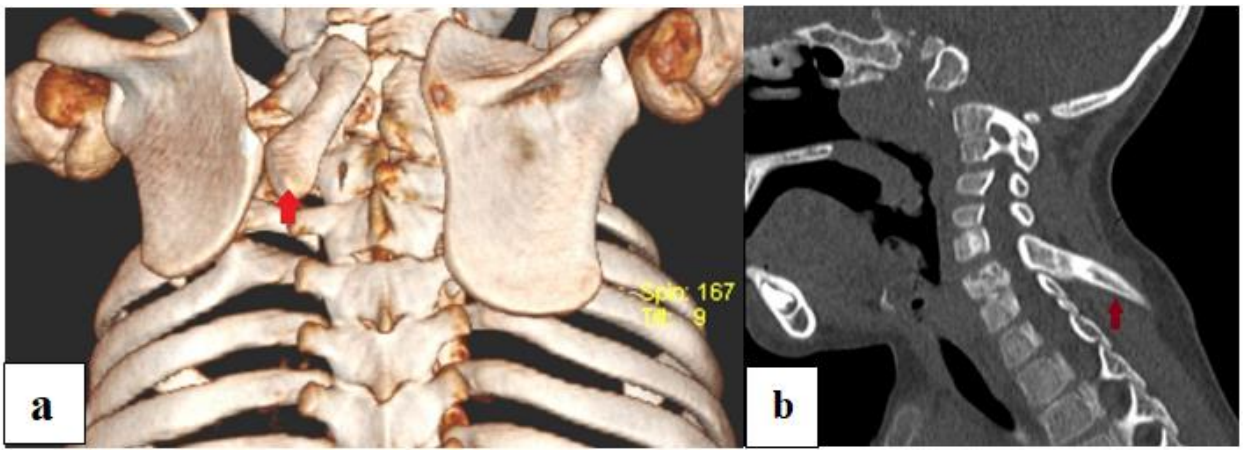

Fig 2: Posterior view of a 3-D volume-rendering reconstruction of the computed tomography (CT) image (a) and sagittal computed tomography (CT) image (b) show the omovertebral bone (arrow)

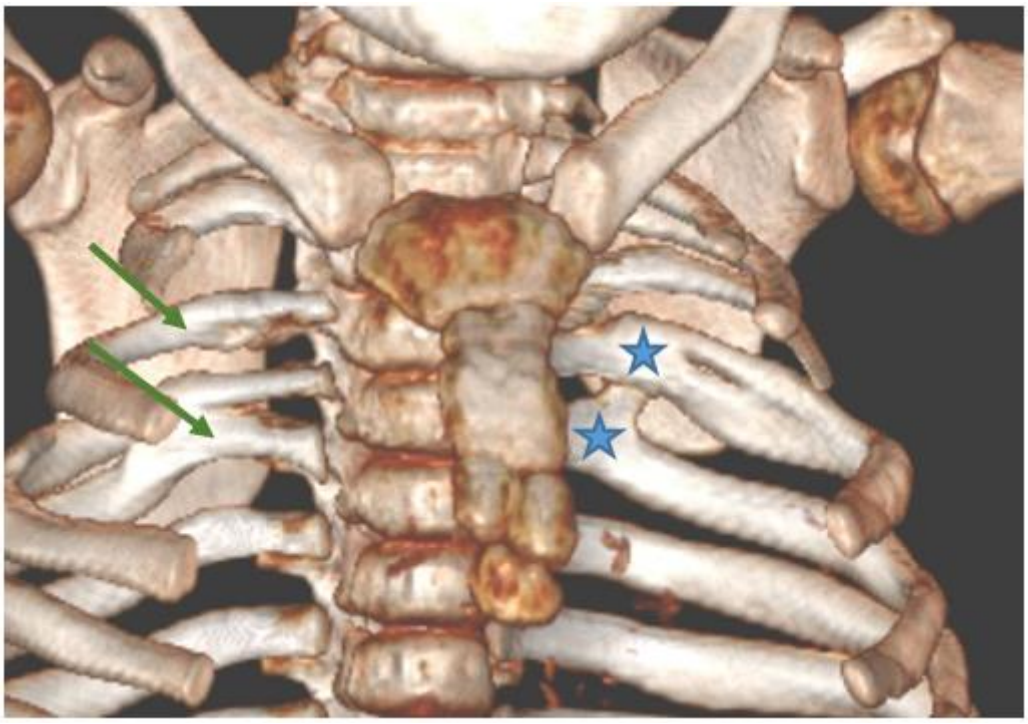

Fig 3: Anterior view of a 3-D volume-rendering reconstruction of the computed tomography (CT) image shows bifid (arrow) and fused ribs (star)

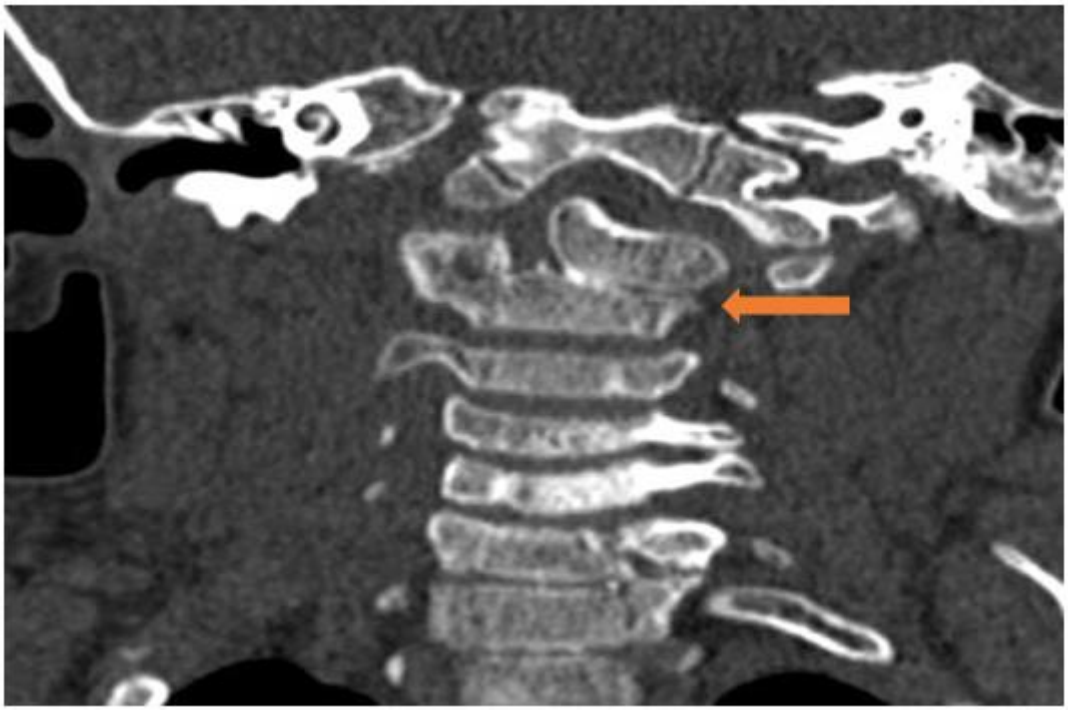

Fig 4: Coronal computed tomography (CT) image shows the cervical scoliosis, hemivertebra and fusion of the C2–C3 (arrow)

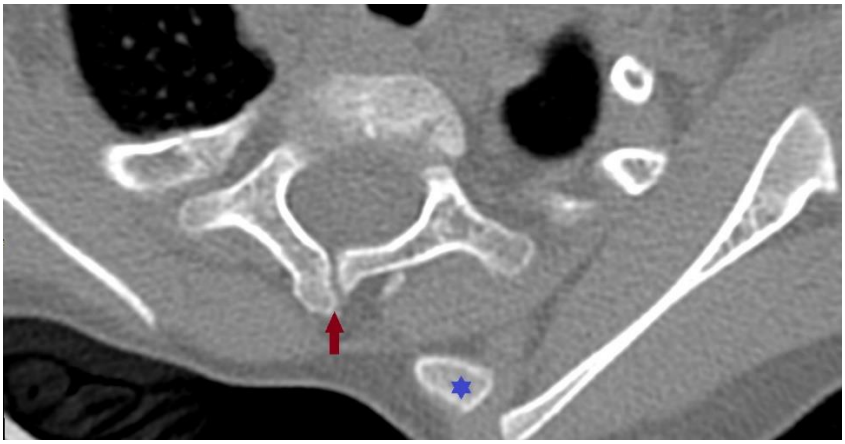

Fig 5: Axial computed tomography (CT) image shows bifid spinous process of C6, indicating spina bifida occulta (arrow) and the omovertebral bone extending to the medial border of the left scapula (Star)

DISCUSSION

Sprengel's deformity (SD), also known as congenital elevation of the scapula, is a rare congenital condition affecting the shoulder girdle. It results from the incomplete descent of the scapula during development, leading to an abnormally positioned, hypoplastic, and rotated scapula [2, 3]. SD is the most frequently observed congenital shoulder abnormality in children [2, 4, 5].

The condition exhibits a female predominance, typically presents unilaterally, and is more commonly found on the left side. Sprengel deformity was first described by Eulenberg in 1863, and later, Willet and Walsham [6] documented two cases, providing a detailed anatomical description of the deformity and recognizing a possible association with spinal and rib anomalies. In 1891, Sprengel [7] reported four cases, and the condition was subsequently named after him.

Sprengel's deformity almost always occurs alongside other abnormalities and is frequently associated with deformities of the thoracic rib cage as well as the cervical and thoracic vertebrae. Commonly observed associated conditions include congenital scoliosis, fused or absent ribs, chest wall asymmetry, cervical ribs, and cervical spina bifida.

As noted earlier, our patient exhibited skeletal deformities, including scoliosis, bifid ribs, rib fusion, hemivertebra, spina bifida occulta, and fused vertebrae. Several syndromes have been associated with Sprengel's deformity, with the most common being Klippel-Feil syndrome, which occurs in approximately 19–27% of

cases. The severity of the disorder ranges from mild limitations in shoulder range of motion (ROM) and subtle cosmetic deformities to more significant functional impairments and severe clinical abnormalities [8].

Imaging:

Radiographic evaluation is essential during the initial assessment of any patient suspected of having Sprengel deformity. Plain radiographs should be taken first to evaluate the position of the scapula relative to the vertebrae and the contralateral side. These images are also useful in identifying associated abnormalities, such as scoliosis, rib defects, or the presence of an omovertebral bone.

CT imaging is employed to identify associated abnormalities, such as congenital scoliosis and cervicoscapular connections. It is particularly useful in severe cases that require surgical intervention. CT scans can help define the presence of an omovertebral bone, especially in relation to the spine. Additionally, three-dimensional CT reconstructions are essential for further assessing the complex pathoanatomy, which aids in formulating an appropriate surgical plan [9].

CT scans are essential in preoperative evaluation but have limitations in assessing non-ossified structures or identifying fibrous or cartilaginous components. In contrast, MRI is useful for evaluating these soft tissue elements and detecting associated spinal abnormalities, such as diastematomyelia. However, MRI can be challenging to perform in young children and may require sedation or general anesthesia [10].

Previous studies have highlighted the importance of 3D-CT in preoperative planning and postoperative evaluation [11]. However, due to the relatively high radiation exposure, routine follow-up using 3D-CT is not recommended. Advances in low-dose CT protocols could enhance the utility of this imaging technique in future research, enabling its safe use in clinical practice for routine follow-up.

Classification:

The Cavendish classification is commonly used for clinical assessment of Sprengel's deformity (SD) and divides the condition into four grades:

- **Grade 1**: The shoulders are level, with a very mild deformity that is almost invisible when covered by clothing.
- **Grade 2**: The deformity remains mild, presenting as a small bump, but the glenohumeral joints remain level.
- **Grade 3**: A moderate deformity with a visible elevation of 2–5 cm in the affected shoulder.
- **Grade 4**: A severe deformity with more than 5 cm of elevation in the affected shoulder, often accompanied by neck webbing.

The radiographic classification introduced by Rigault *et al*., [12] is based on the projection of the superomedial scapular angle in relation to the corresponding vertebral level. According to this system:

- **Grade 1**: The superomedial scapular angle is located below the T1 vertebra.
- **Grade 2**: The angle lies between T1 and C5.
- **Grade 3**: The angle is positioned above C5.

Therapeutic management:

An early and accurate diagnosis is crucial, with surgery indicated for severe cases. Various surgical techniques have been documented in the literature, typically with favorable outcomes reported [13].

CONCLUSION

Sprengel deformity is the most prevalent congenital abnormality of the shoulder girdle and can result in considerable cosmetic and functional impairments. Accurate diagnosis is crucial, as the condition may be accompanied by various associated abnormalities and, if not properly managed, can lead to significant morbidity. X-ray images can help confirm the diagnosis and identify the presence of an omovertebral bone, though interpretation can be challenging due to overlapping bones. Further imaging typically involves CT scans and their 3D reconstructions, which provide more detailed information. CT scans with 3D reconstructions clearly display both the location and size of the omovertebral bone, as well as any associated rib and vertebral abnormalities. When surgical intervention is indicated, the supernumerary structure must be resected.

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