Case Report

DOI: 10.7860/JCDR/2025/77974.21261

Sprengel Shoulder with Omovertebral Bone and Left Renal Agenesis in a Paediatric Patient: A Rare Case Report

Paediatrics Section

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ABSTRACT

Sprengel shoulder is a rare congenital anomaly caused by disrupted scapular descent during development. It is associated with hypoplastic or absent muscles such as the trapezius, sternocleidomastoid, and serratus anterior, leading to scapular winging. Cavendish grading classifies the severity as very mild, mild, moderate, or severe. Mild cases may appear normal under clothing, while severe cases feature significant elevation, neck webbing, and brevicollis. One-third of cases have an omovertebral bone. Diagnosis is made through chest and shoulder radiography. Treatment is non-operative for mild cases, while moderate to severe cases require early surgical correction to prevent movement restriction. Sprengel shoulder is linked to conditions such as Klippel-Feil Syndrome (KFS), Poland syndrome, and VATER association, necessitating multidisciplinary evaluation. In this case report, a three-year-old girl presented with left shoulder elevation and restricted movement. Prenatal history revealed left renal agenesis. Examination showed an elevated left shoulder, restricted abduction, and apparent limb shortening. A typical Sprengel deformity with an omovertebral bone was diagnosed with the help of Magnetic Resonance Imaging (MRI). The Modified Excision and Reconstruction of Scapula (MEARS) procedure was planned, to improve mobility and quality of life for this patient. Additional evaluation was planned due to associated features such as a low hairline, vertebral fusion anomalies, and spina bifida, suggesting a possible genetic syndrome, most likely KFS. The rarity of Sprengel shoulder with an omovertebral bone and unilateral renal agenesis prompted this report.

Keywords: Congenital, Dysplasia, Paediatrics, Shoulder blade, Torticollis

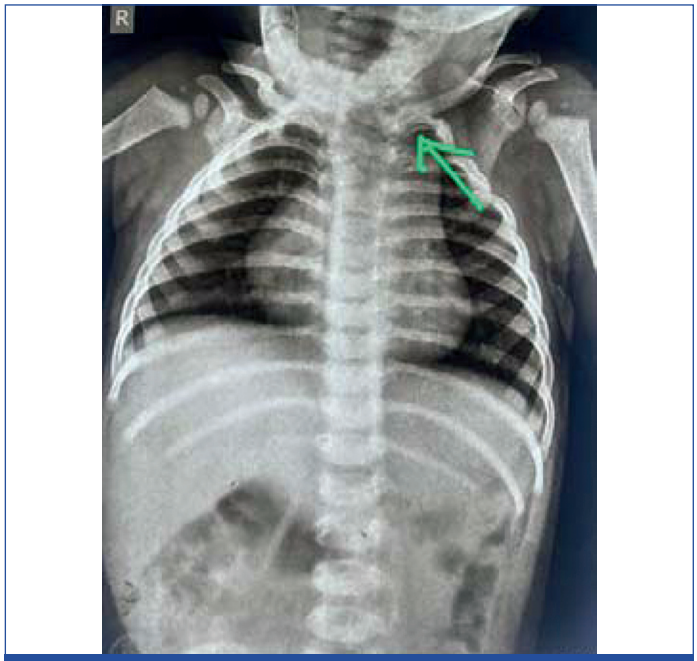
CASE REPORT

A three-year-old girl presented to the department of paediatrics with developmental delay and difficulty in neck and shoulder movements. Her parents first observed a visible elevation of her left shoulder at eight months of age. By the time of presentation, she exhibited difficulty moving her neck and raising her left arm above her head. At one year of age, the primary physician conducted initial investigations, including chest X-rays and an MRI of the spine, which revealed an elevated left shoulder with cervical fusion anomalies and a fibrous omovertebral bar connecting the scapula to the cervical spine, leading to the diagnosis of Sprengel shoulder. Antenatal scans at 16 weeks had also shown left renal agenesis. However, the family history did not indicate any associated anomalies that could have suggested inheritance. A follow-up review was scheduled for six months later to plan surgical intervention; however, the patient was lost to follow-up and did not return for the planned review.

At three years of age, she presented again for further assessment and management, and relevant investigations were repeated, confirming the diagnosis. Physical examination revealed several key findings consistent with Sprengel deformity. The patient's neck appeared short and webbed, with a lower-than-expected posterior hairline. The superior border of the left scapula was positioned 2.5 cm higher than the right scapula, with medial rotation of the inferior angle, contributing to a visible "hump-like" deformity. Preoperative Range of Motion (ROM) assessment showed that left shoulder abduction was restricted to a maximum of 90 degrees, whereas the normal ROM exceeded 180 degrees [Table/Fig-1]. Shoulder flexion was also limited compared to the contralateral side.

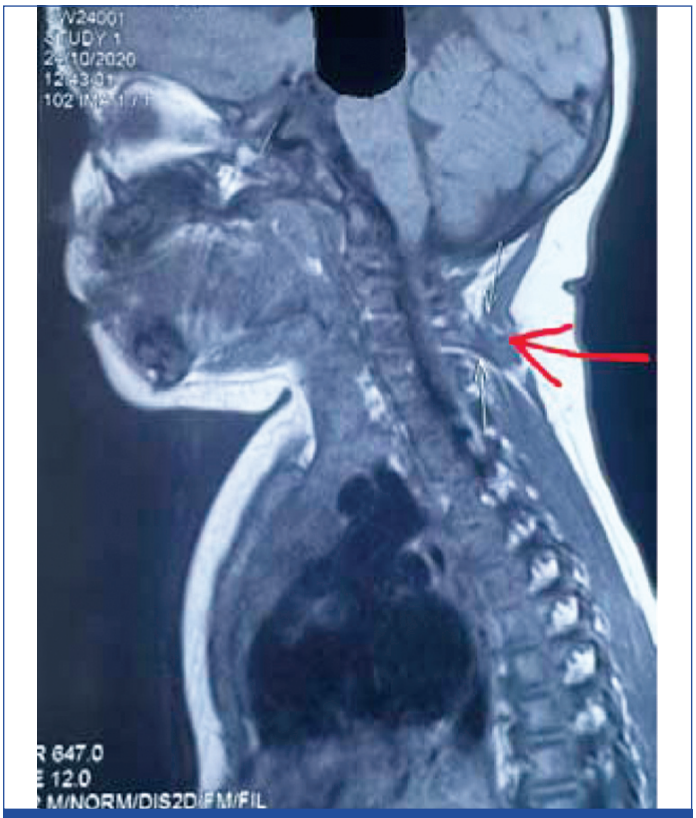
Imaging findings included a chest X-ray (anterior-posterior view), which showed an elevated and hypoplastic right scapula, a hallmark of this congenital condition caused by failure of scapular descent during embryonic development. The affected shoulder appeared abnormally high, with altered orientation of the scapular borders [Table/Fig-2]. MRI of the shoulder and spine demonstrated a left Sprengel shoulder, multiple vertebral segmentation-fusion anomalies

[Table/Fig-1]: Sprengel shoulder.

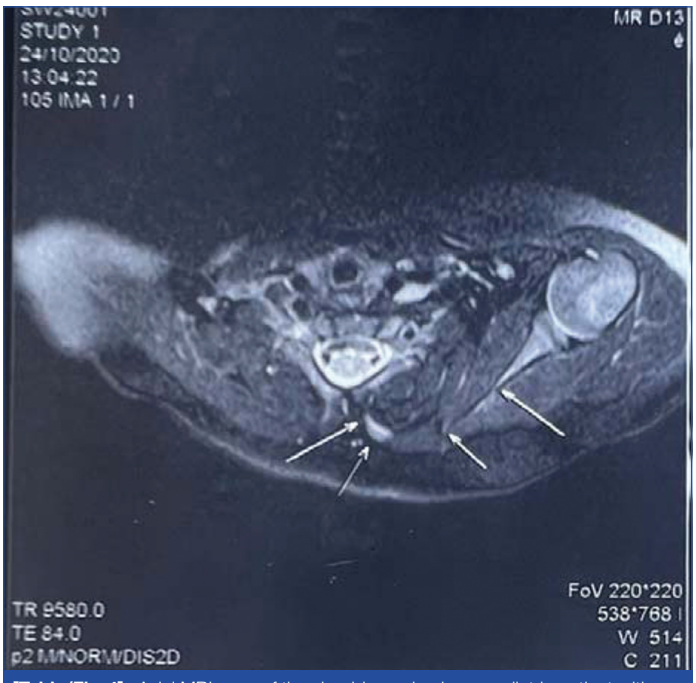


[Table/Fig-2]: Anteroposterior chest X-ray showing raised left scapula in paediatric patient with Sprengel shoulder.

in the cervical spine, and an osseous fibrous omovertebral bone extending from the left-side lamina-spinous process of C4 to the scapula. Additionally, spina bifida was observed at all cervical levels [Table/Fig-3], where the MRI showed multiple cervical vertebral fusions characteristic of Klippel-Feil Syndrome (KFS), which commonly co-exists with Sprengel deformity. The presence of fused vertebrae resulted in a shortened neck with limited ROM. An axial MRI of the shoulder joint also confirmed the findings [Table/Fig-4], showing the omovertebral bone as an abnormal bony connection between the scapula and cervical spine, commonly seen in Sprengel deformity. This bony bridge contributed to restricted shoulder movement and deformity.



[Table/Fig-3]: Sagittal T2-weighted MRI of the cervical spine in a paediatric patient with Sprengel deformity. The image demonstrates multiple congenital cervical vertebral fusions (indicated by the red arrow), consistent with Klippel-Feil Syndrome (KFS), a condition frequently associated with Sprengel deformity.



[Table/Fig-4]: Axial MRI scan of the shoulder region in a paediatric patient with Sprengel shoulder, demonstrating the presence of an omovertebral bone (indicated by arrows).

Before confirming Sprengel deformity, other conditions were considered, including congenital muscular torticollis, which was ruled out due to the presence of scapular elevation and osseous abnormalities. KFS was considered because of overlapping features, but Sprengel deformity of grade II severity was determined to be the primary diagnosis due to the characteristic scapular elevation and omovertebral bone. Congenital scoliosis with scapular malposition was excluded as there was no primary spinal deformity affecting the thoracic region. The patient was treated with the Modified Excision and Reconstruction of Scapula (MEARS) procedure and supportive management in the form of immobilisation, as well as physiotherapy involving early Passive Range of Motion (PROM) and assisted active ROM. However, the patient was again lost to follow-up following the procedure.

DISCUSSION

The rare Sprengel deformity, also known as congenital high scapula, is the most frequent shoulder abnormality present at birth. This condition is mostly unilateral and more frequently affects the left side [1]. Between 10% and 30% of cases involve both shoulders (bilateral), and females are three times more likely to be affected than males [2]. This condition was first reported by Eulenburg in 1863 and later described by Sprengel in 1891, hence the name [3]. The deformity arises due to abnormal descent of the scapula during the 9th to 12th week of gestation and is primarily a cosmetic concern [3]. Cervical spine deformities can also be present in some patients [4].

Sprengel deformity may be associated with Klippel-Feil Syndrome (KFS), omovertebra, spina bifida, rib anomalies, musculoskeletal dysfunctions, and tethered cord syndrome. It may also be linked to cardiac anomalies, renal anomalies, tracheoesophageal fistulas, anal atresia, absence of pectoral muscles, and hand anomalies [5]. The severity of the deformity can vary significantly, ranging from being almost unnoticeable under clothing to having the shoulder elevated by over 5 cm, with associated neck webbing. The majority of cases appear spontaneously, with autosomal dominant inheritance being uncommon [6].

This report describes a case of Sprengel deformity affecting the left shoulder (unilateral involvement) in a three-year-old female patient. Significant clinical and radiographic findings are presented.

These findings led to a diagnosis of left Sprengel shoulder with an omovertebral bone and multiple vertebral segmentation-fusion anomalies of the cervical spine. The positioning of the scapula between the C5 and T2 vertebrae suggests a grade II deformity according to the Rigault classification [7]. This classification is based on the projection of the superomedial angle of the scapula in relation to the associated vertebral level. Grade I corresponds to the superomedial scapula angle being below T1, grade II lies between T1 and C5, and grade III is above C5 [8].

The orthopaedic team was involved and is planning a surgical intervention using the MEARS procedure, followed by range-of-motion physiotherapy. The MEARS procedure involves a triphasic approach that includes scapular osteotomy to reposition the scapula definitively. Partial scapular excision may be performed to create additional space for improved mobility. The release of the long head of the triceps, a potential movement-limiting muscle attaching to the scapula, may also be necessary [9]. In some cases, a concurrent clavicular osteotomy can be performed to protect the brachial plexus during the MEARS procedure. Postoperatively, patients typically undergo a period of immobilisation with a shoulder sling, followed by the early initiation of range-of-motion exercises to promote the restoration of joint function [9].

Sprengel's shoulder has a connection to KFS. Mutations in the GDF6, GDF3, and MEOX1 genes can lead to KFS being inherited, but most cases occur without a family history [10]. A comprehensive neonatal physical examination is crucial for identifying KFS due

to the potential presence of various associated anomalies. This examination should include a craniofacial evaluation for facial dysmorphism, microcephaly, or a low posterior hairline. Additionally, auscultation can detect murmurs that may indicate an underlying cardiac abnormality. This thorough examination approach enhances the chance of early KFS diagnosis and identification of any coexisting conditions [10]. Similar studies by Lotfian G et al., report a paediatric patient presenting with KFS, Sprengel deformity accompanied by an omovertebral bone, and congenital unilateral renal agenesis. The report emphasises the importance of recognising the constellation of anomalies for accurate diagnosis and management [11].

A comprehensive multidisciplinary approach, incorporating surgical intervention and postoperative rehabilitation, is essential for enhancing long-term function and quality of life in individuals with Sprengel deformity [12,13]. Another study by Kariminasab MH et al., discusses a patient with unilateral Sprengel's deformity linked to multiple musculoskeletal abnormalities and renal agenesis. It highlights the necessity for comprehensive evaluation to identify associated anomalies that may influence treatment decisions [14]. A similar study by Stelzer JW et al., involves a three-year-old girl exhibiting KFS, Sprengel deformity, and additional skeletal anomalies. The report states the complexity of such cases and the importance of multidisciplinary management approaches [15].

The widely adopted MEARS procedure follows a triphasic technique: it includes scapular osteotomy to achieve definitive repositioning of the scapula, partial scapular excision to create more space for improved mobility, and release of the long head of the triceps to enhance shoulder function [16]. In some instances, a clavicular osteotomy may also be performed concurrently to safeguard the brachial plexus during the procedure.

Postoperative rehabilitation typically begins with a brief period of immobilisation, followed by the early initiation of range-of-motion exercises aimed at restoring joint function [17]. Surgical intervention performed at an early age, preferably before six to eight years, has been shown to significantly improve long-term outcomes by reducing functional limitations and minimising the risk of neurological complications [12,18].

A notable limitation in this case was the patient's loss to follow-up, which delayed the planned surgical management and restricted the evaluation of early treatment outcomes. Such disruptions in follow-up care underscore the importance of establishing a structured patient monitoring system to ensure timely intervention and better prognostic outcomes.

Future studies should investigate the long-term effects of treatment in patients with Sprengel deformity, especially those presenting with additional congenital anomalies, to refine surgical techniques and optimise rehabilitation protocols further [13]. This case adds to the expanding literature on congenital skeletal anomalies and highlights the significance of early diagnosis, comprehensive genetic evaluation, and prompt surgical correction [13,19].

CONCLUSION(S)

Sprengel deformity is a rare congenital condition affecting movement, appearance, and function, with significant physical and emotional impacts. Early diagnosis and multidisciplinary intervention are crucial to prevent complications, such as nerve compression, and to ensure optimal long-term outcomes.

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PLAGIARISM CHECKING METHODS: [Jain H et al.]
- Plagiarism X-checker: Jan 16, 2025
- Manual Googling: May 22, 2025
- iThenticate Software: May 24, 2025 (10%)

ETYMOLOGY: Author Origin

EMENDATIONS: 7

AUTHOR DECLARATION:
- Financial or Other Competing Interests: None
- Was informed consent obtained from the subjects involved in the study? Yes
- For any images presented appropriate consent has been obtained from the subjects. Yes

Date of Submission: **Jan 13, 2025**
Date of Peer Review: **Mar 12, 2025**
Date of Acceptance: **May 27, 2025**
Date of Publishing: **Jul 01, 2025**