

Case Report

A rare case of Sprengel deformity associated with spondylocostal dysostosis in a four-year child

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ABSTRACT

Sprengel deformity is a complex congenital anomaly affecting one or both scapulae with mispositioning and dysplasia of scapula. It occurs due to failure of descent of scapula during intrauterine development and is the commonest congenital skeletal deformity of the scapula, often associated with other skeletal deformities. spondylocostal dysostosis is a rare genetic disorder which is characterized by malformation of the bones of the spine and ribs. It occurs in approximately 1 in 2,00,000 people worldwide. In spondylocostal dysostosis, affected children have wedge shaped vertebrae along with ribs that are fused, forked or sometimes missing. Based on the severity of the defect's children may present with short stature, scoliosis or hypoplasia of lungs causing difficulty in breathing and recurrent respiratory tract infections. Management of both these skeletal deformities depends on their severity and the presence of associated deformities. Surgical intervention might be required for cosmetic and functional recovery of the shoulder. Authors had a rare and interesting case of a 4-year-old boy who had features of Sprengel deformity affecting left scapula which was associated with features of spondylocostal dysostosis.

Keywords: Birth defects, High scapula, Spondylocostal dysostosis (SCDO), Sprengel deformity

INTRODUCTION

Sprengel deformity or congenital elevation of scapula is a complex deformity of the pectoral girdle resulting in cosmetic and functional disabilities occurring due to abnormal descent with altered position of the scapula.¹ This condition is associated with regional muscle hypoplasia or atrophy leading to disfigurement as well as limitation of movement of the shoulder. It occurs equally in both sexes and left sided Sprengel deformity is commoner.² Sprengel deformity is often associated with other features such as fused or absent ribs, chest wall asymmetry, Klippel Feilsyndrome, cervical ribs, congenital scoliosis as well as cervical spina bifida.² Spondylocostal dysostosis (SCDO) is rare genetic disorder which can be identified radiologically with rib

abnormalities and multiple segmentation defects of the vertebrae.³ This autosomal recessive disorder has an incidence of approximately 1 in 2,00,000 people worldwide. This condition should be suspected with radiographic features of multiple segmentation defects of vertebrae, mild scoliosis, malalignment of ribs, variable rib fusions and absence of other congenital anomalies like cardiac or renal anomalies. Children with spondylocostal dysostosis have scoliosis due to malformation of the vertebra. Affected children also have short rigid neck and short trunk due to bony abnormalities causing short stature with short-trunk dwarfism.⁴ Some neonates may suffer from respiratory distress due to compromised thoracic function. Children may develop difficulty in breathing due to impaired lung growth and severe defects can lead to life threatening complication like respiratory

insufficiency.³ Some children with spondylocostal dystostosis have associated neural tube defects like diastematomyelia or meningocele.^{3,4} Spondylocostal dysostosis is a genetically inherited disorder, type 1 has autosomal recessive inheritance whereas other types can have autosomal dominant inheritance. There are six subtypes of autosomal recessive spondylocostal dystostosis.⁴ Here authors are presenting a rare case of a 4-year child with Sprengel deformity associated with features of Spondylocostal dysostosis.

CASE REPORT

A 4-year-old male child presented with history of deformity over the left shoulder since birth. The child had difficulty in abduction and elevation of left arm noticed by the parents in early infancy which was not progressive in nature. He was the second born child through normal vaginal delivery with no history of birth trauma to grade 2 consanguineous parents.



Figure 1: Clinical photograph showing elevated left shoulder with deformity of the spine and upper back.

His developmental milestones were normal and there was no history of neonatal respiratory problems or recurrent respiratory tract infection during early childhood. There was no history of similar complaints in other family members. On physical examination, his weight was 15kg (between 3rd-50th percentiles) and height was 96cm (between 3rd-50th percentiles). There were no dysmorphic facial features. Hernial orifices were normal. He had a short neck, deformed left shoulder, elevation of left scapula and scoliosis. The child had limited range of movement with 60-degree abduction of left shoulder. There was no associated neurovascular compromise. The right shoulder and right arm were normal. The boy had a visible deformity of the sternum and adjacent costochondral junction. Clinically the child appeared to be having Sprengel deformity of the shoulder with scoliosis. Systemic examination appeared to be normal.

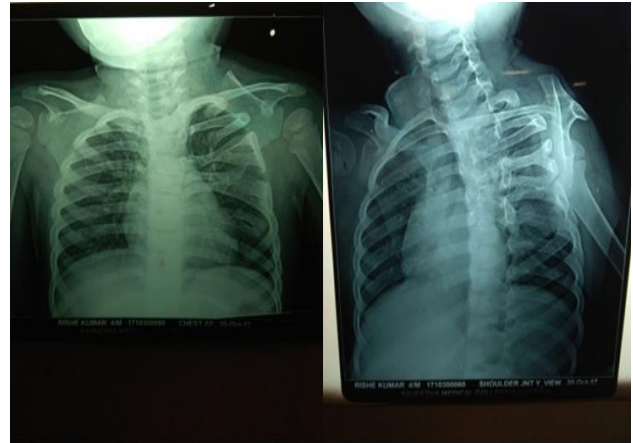


Figure 2: X-ray thorax AP view showing asymmetrical left scapula with high position of left scapular base, bifid ribs on left side, bifid vertebrae and scoliosis.

On investigation, the X-ray of the thorax demonstrated asymmetrical and high positioned left scapula with bifid ribs involving 3rd to 6th ribs as well as thoracic scoliosis. CT scan thorax showed features suggestive of Grade 2 sprengel deformity (rigault classification). He also had bifid ribs in posterolateral ends affecting left 3rd - 6th ribs with partial fusion of posterolateral ends of left 4th and 5th ribs along hemivertebra and bifid spinous process of D1, D2, D4, D6, and D8 vertebrae. The X- ray and CT scan findings were suggestive of Sprengel deformity associated with Spondylocostal dysostosis. The other investigations like ultrasound abdomen, echocardiogram, thyroid function test, serum calcium, serum phosphorus and serum alkaline phosphatase were within normal limits. Orthopedics and neurosurgeon opinion were obtained.

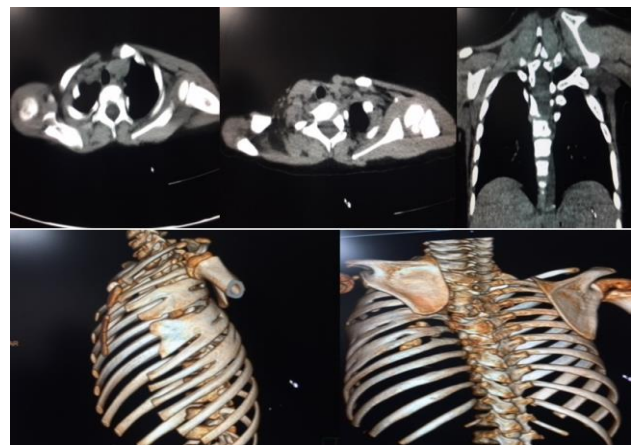


Figure 3: Axial CT showing left sided Sprengel deformity, bifid ribs in posterolateral ends of left 3rd and 6th ribs, partial fusion of posterolateral ends of left 4th and 5th ribs, hemivertebra with bifid spinous process of D1, D2, D4, D7 and D8 vertebrae.

After detailed assessment and consultations, conservative line of management was planned. The child was advised to undergo regular physiotherapy for improving range of movements for abduction of left shoulder and is under follow up.

DISCUSSION

Sprengel deformity is the commonest congenital defect of the scapula and usually recognized at birth or in early childhood.¹ Otto Sprengel was the first to publish many cases of upward dislocation of scapula in 1891. Sprengel deformity occurs due to failure of normal descent of scapula from initial mid-cervical position to final thoracic position. This scapular descent occurs during sixth and eighth weeks of gestation. Sprengel deformity may be associated with other defects such as congenital scoliosis, upper extremity anomalies and diastematomyelia and kidney disease and also associated with syndromes like Klippel Feil syndrome.⁵

Spondylocostal dysostosis has often been grouped with another similar condition called spondylothoracic dysostosis and both disorders are sometimes called Jarcho-Levin syndrome however, they are now considered distinct conditions.^{3,6} The term Jarcho-Levin syndrome is used to describe many conditions that have spinal and rib abnormalities. Spondylocostal dysostosis is a rare disorder, the exact incidence being unknown. Because it is rare disorder some people may go undiagnosed, making it difficult to determine the true frequency in the general population. The differential diagnosis for conditions having multiple segmentation defects of vertebra are Alagille syndrome, Campomelic dysplasia, charge syndrome and Goldenhar syndrome.^{7,8}

X ray of thorax, CT thorax with 3D Reconstruction and MRI are helpful for making a diagnosis of Sprengel deformity and spondylocostal dysostosis. Estimation of the severity of spondylocostal dysostosis by determination of associated osseous and muscular deformities is essential for choosing and planning appropriate line of management. Our patient had a combination of clinical and radiological features of Sprengel deformity as well as Spondylocostal dysostosis. It is important to confirm the diagnosis by molecular genetic testing. Genetic counselling should be done along with screening of other family members.

Management of individual case depends on the severity of the deformity. Children require monitoring of growth and development, respiratory function and spinal curvature. Surgery in Sprengel deformity is considered for children between 3 to 8 years who have moderate to severe disability.⁹

Children with severe scoliosis may require orthopaedic intervention. Surgical intervention may be also required for improving cosmetic and functional recovery of the shoulder. Considering the young age of the child and

presence of multiple anomalies, physiotherapy was initiated. The parents were counselled about the condition and child is under follow-up.¹⁰

CONCLUSION

Spondylocostal dysostosis in association with Sprengel deformity is a rare condition in children presenting with distinctive malformations of the spinal column, ribs and scapula. CT scan with 3D reconstruction and MRI are particularly useful for the accurate diagnosis and calculation of severity of the condition.

Determination of associated osseous and muscular deformities are essential for choosing and planning treatment. This case is being reported because of its rarity. It is important for paediatricians to be aware of this clinical condition. Screening of children with Sprengel deformity is important to identify associated bony abnormalities affecting the ribs and spine.

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