

Case Report

The sprengel deformity

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ABSTRACT

Sprengel shoulder is a rare congenital deformity of one or both scapulae that is usually detected at birth. It occurs due to failure of the scapula to descend during intrauterine development. Although the deformity appears randomly most of the time, familial cases have been reported. Sprengel shoulder is often associated with Klippel-Feil syndrome and other congenital skeletal deformities. Anteroposterior X-ray imaging can accurately diagnose Sprengel deformity. However, computed tomography and magnetic resonance scans with three-dimensional reconstruction are nowadays used in everyday practice in order to diagnose concomitant abnormalities, study in detail the anatomy of the affected shoulder(s), and plan appropriate management. We present here our imaging experience from one pediatric case with Sprengel shoulder and take the opportunity to discuss this rare entity, which is, nevertheless, the commonest congenital defect of the scapula.

Keywords: Birth defects, High scapula, shoulder, Sprengel deformity, Undescended scapula

INTRODUCTION

Sprengel deformity also known as congenital high scapula, undescended scapula, scapula elevate, is a rare congenital deformity of one or both scapulae that appears at birth¹ although Pellegrin et al., have published a single case in which the patient presented with Sprengel shoulder reported as appearing after trauma.² Undescended scapula results from failed migration of the scapula during early embryonic life,³ leading to a scapula that protrudes in the neck of the patient. However, the deformity is not simply an aesthetic concern, but more importantly it causes restricted mobility of the shoulder and cervical spine.⁴ Sprengel deformity appears either as a single defect or in association with other abnormalities, most commonly Klippel-Feil syndrome, scoliosis and rib defects.⁵ Conventional anteroposterior radiography of the chest including both shoulders is a simple and effective means of diagnosing Sprengel deformity, particularly when combined with appropriate clinical information. Computed tomography (CT) with three dimensional (3-

D) reconstruction and magnetic resonance imaging (MRI) are necessary nowadays for the diagnosis of coexisting abnormalities and treatment planning. Although the management of Sprengel deformity depends on the severity of the abnormality and the degree of motion restriction, the most common therapeutic choice for patients and orthopaedic surgeons is surgical intervention for cosmetic and functional recovery of the shoulder.⁵

CASE REPORT

Lonare Hindavi, a 3-years-old girl was referred to our department for imaging studies prior to surgical repair of her left undescended scapula. Anteroposterior X-ray scan of the shoulders were performed. X-ray examination showed left Sprengel deformity and the presence of an omovertebral bone. The CT scan, showed spina bifida of the spinous process of C-6 and C-7 vertebrae. Congenital fusion of the posterior elements and transverse processes of the C-5, C-6 and C-7 vertebrae. A fibro-osseous connection is seen arising from the tip of C-7 spinous

process on the left side reaching upto the left scapula – s/o omovertebral bone. CT scan was done in order to evaluate the omovertebral association and demonstrate the spectrum of abnormalities that was required for thorough planning of surgery (Figures 1 and 2).



Figure 1: Anteroposterior chest radiograph showing raised left scapula and formation of omovertebral bone.

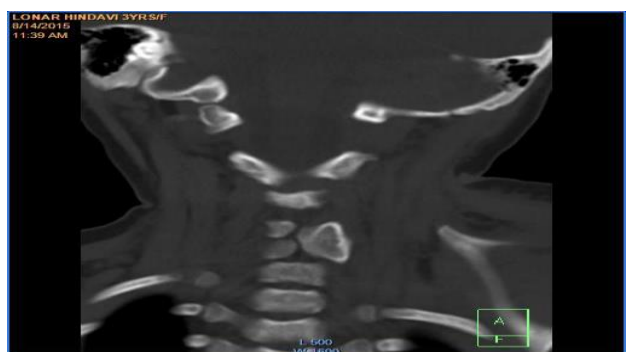


Figure 2: CT scan showing raised left scapula and formation of omovertebral bone.

DISCUSSION

Sprengel deformity was described for the first time in 1863 by Eulenberg, but it was not until 1891 that Sprengel was to give it its name by reporting four observations.⁶ The physiopathogenesis of this condition remains unknown. The scapula appears normally at the 5th week of intrauterine life in the form of a mesenchymal mass level with the 4th and 5th cervical vertebrae.⁷ From the 6th week, it migrates caudally to reach its definitive physiological position at the 12th week. The inferior angle of the scapula is then situated between the 6th and 8th thoracic vertebrae. During this caudal migration, the morphology of the scapula changes to become adapted to the prehensile function of the upper limb.⁷ From the 12th week of intrauterine life, the morphology of the scapula is similar to that of an adult scapula, higher than it is wide. Sprengel deformity results from this caudal migration being interrupted. The cause of this is still unknown but is possibly vascular.^{8,9} A few familial forms of Sprengel deformity have been described and suggest the possibility of genetic transmission.⁹

In Sprengel deformity, the scapula is in a high position and appears more or less hypoplastic and dysmorphic, wider than it is high, with a convex medial border and a concave lateral border, and associating scapular muscle hypoplasia. The scapula on the opposite side is in a normal position but sometimes slightly dysmorphic.¹⁰ Sprengel deformity is frequently associated with other bone abnormalities which can be costal (aplasia, hypoplasia, synostosis, bifidity, supernumerary ribs, etc.), clavicular (abnormalities in shape and/or position) and, above all, vertebral(Klippel-Feil syndrome, cervical and /or thoracic spina bifida occulta, hemivertebrae, secondary scoliosis, etc.). These vertebral malformations, which are frequently associated are explained by the common embryological origin of the scapula and the cervical spine, both derived from mesenchymal tissue known as the para-axial mesoderm. However the most common association is with an omovertebral bone, even though this is not always present. Its origin is still controversial: the omovertebral bone may develop from the posterior arch of vertebrae, from the scapula, or be an acquired ossification.¹¹

Table 1: Cavendish's clinical classification

Grade 1	Shoulders at the same level; deformity invisible when patient is dressed
Grade 2	Shoulders at the same level; deformity visible when patient is dressed (lump at the base of the neck)
Grade 3	Shoulders asymmetric: the affected shoulder is raised by 2 to 5 cm
Grade 4	The affected shoulder is raised by more than 5 cm (superior angle close to the occiput)

Sprengel deformity is usually unilateral (90% of cases), more frequent in girls than in boys (sex ratio of 1:3) with or without left or right predominance depending on the author.^{1,9,11} It is often diagnosed in early childhood. Parents consult because of unaesthetic disfigurement (scapular asymmetry, posterior thoracic swelling corresponding to the projection of the tip of the scapula) and / or limited active abduction of the shoulder. There may be multidirectional instability of the shoulder. The Cavendish classification (Table 1) differentiates four clinical grades of increasing severity, depending on the size of the swelling and how far the scapula is raised, from very slight malformation (grade 1) to severe malformation (grade 4).¹³ Diagnosis of Sprengel deformity is confirmed by X-ray images showing a high scapula. Rigault and Pouliquen's classification takes into account the amount by which the scapula is raised on standard images (Table 2): slightly raised (grade 1, omovertebral bone generally absent); raised to the usual degree (grade 2) or severely raised (grade 3).¹⁴ It is sometimes difficult to confirm the possible presence of an omovertebral bone because of the superposition of bones.

A CT scan and its 3D reconstructions is then very useful for detecting an omovertebral bone, giving its site and size, evaluating the position of the scapula in space (lateral tilt in the frontal plane, anterior tilt in the sagittal plan and medial tilt in the transverse plane) and the degree of scapular dysmorphia, detecting associated bone abnormalities, particularly of the vertebrae, and for planning the surgical procedure well.^{7,10} CT scans play a prominent role in preoperative evaluation.

Sprengel deformity may lead to discussion of neonatal paralysis of the brachial plexus, scapula alata (or winged scapula) due to paralysis of the serratus anterior muscle, scapula alata in facioscapulohumeral muscular dystrophy, or scapuloiliac dysostosis.⁹ It can usually be diagnosed from the medical history and imaging showing that only the scapula is involved.

Sprengel deformity is often treated surgically in childhood (between 3 and 7 years of age) for aesthetic reasons and to improve functioning of the upper limb.⁷ Resection of the omovertebral bone or its cartilaginous or fibrous equivalent is a prerequisite for any surgical procedure.

Table 2: Rigault and Pouliquen's radiological classification.

Grade	Position of the medial angle of the scapula
Grade 1	Between the transverse processes of T2 and T4
Grade 2	Between the transverse processes of C5 and T2
Grade 3	Above the transverse process of C5

CONCLUSION

Sprengel deformity is characterised to variable degrees by a high dysmorphic scapula. A supernumerary structure which may be ossified (an omovertebral bone) is often associated with it and occurs between the medial border of the scapula and the cervical spine. It is clinically diagnosed early because of unaesthetic disfigurement (shoulder asymmetry) and /or functional difficulties (limited abduction). X-ray images can confirm the diagnosis and objectify an omovertebral bone, which may be present, but they are sometimes difficult to interpret because of superimposed bones. When it has been decided to treat surgically, the supernumerary structure must be resected. Additional imaging relies principally on CT scans and there 3D reconstructions. The latter show both the site and dimensions of the omovertebral bone as well as often associated vertebral abnormalities.

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