SPRENGEL'S DEFORMITY: A CASE REPORT AND REVIEW OF THE LITERATURE

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INTRODUCTION

Sprengel's deformity or congenital elevation of the scapula is a rare congenital disorder where the scapula is abnormally elevated and dysplastic (1). The condition was first described in 1863, and named after Otto Gerhard Karl Sprengel, who reported on four cases in 1891 (2). Although it is considered to be the most common congenital deformity of the shoulder, its incidence is unknown since a vast number of cases remain undiagnosed (3). The condition is more common in females, with a female-to-male ratio of 3:1 (4). Although the deformity typically affects one shoulder, it may also be bilateral, and occurs in approximately 10 to 30% of cases (5).

AETIOLOGY

The scapula is normally found between the 2nd and 7th thoracic vertebrae on the posterior thoracic wall. Sprengel's deformity occurs due to failure of the affected scapula to migrate caudally during early foetal development (6). The underlying cause for this to occur is currently unknown, and is often sporadic; however, an autosomal dominant inheritance pattern of the deformity may be present in rare cases, referred to as Corno's disease (7). It has been proposed that oligohydramnios or neural crest defects are the root cause of the deformity (2), causing a transient interruption in embryonic blood supply via the subclavian artery (4). During embryogenesis, the scapula normally forms at the level of the cervical spine, then moves caudally to its normal position during the third month of gestation (8). In Sprengel's deformity, this migration fails to occur (1).

Moreover, an omovertebral bone may be present in 19 – 47% of cases (9), extending between the affected scapula and the spinous processes; transverse processes; or laminae of vertebrae C4 to C7. This further impedes the normal caudal migration of the scapula, producing the abnormal elevation. The connection may be fibrous, osseous or cartilaginous (6,9).

CLASSIFICATION SYSTEMS

Sprengel's deformity is most commonly graded according to the clinical Cavendish classification, which groups the deformity based on the degree of elevation and associated deformity (1,3).

Table 1: Cavendish Classification

Grade	Description
Grade 1	Very mild deformity observed. When patient is dressed, the deformity is almost invisible.

Grade 2	The deformity is still mild but noticeable as a bump. The superomedial portion of the scapula is convex, forming the bump.
Grade 3	Moderate deformity, with 2 – 5 cm of visible elevation of the affected shoulder.
Grade 4	Severe deformity with > 5 cm elevation of the affected shoulder, accompanied by neck webbing.

In addition, the radiographic Rigault classification is utilised to grade the deformity on x-rays, based on the vertebral level of the scapula to indicate the grade (10).

Table 2: Rigault Classification

Grade	Description
Grade I	Superomedial scapular angle lower than T2 but above T4 transverse process.
Grade II	Superomedial angle positioned between C5 and T2 transverse process.
Grade III	Superomedial angle above C5 transverse process.

CLINICAL ASSOCIATIONS

In approximately 75% of cases, other abnormalities are present which may form part of a syndrome (11) and which must be considered when dealing with such patients. The following have all been reported in the context of this deformity (1,4,11):

- Klippel-Feil Syndrome (20 to 42% of patients (2);
- Congenital torticollis;
- Defects of the cervical vertebrae;
- Kyphoscoliosis or isolated scoliosis;
- Spina bifida;
- Clavicle hypoplasia.

Furthermore, atrophy or hypoplasia of the shoulder girdle musculature may be a feature of this deformity (1). With severe deformities, the inferior angle of the scapula is rotated medially, thereby directing the glenoid fossa downwards (6). This, together with the abnormal regional muscles, limits the range of motion of the affected shoulder, especially abduction (12).

DIAGNOSIS, IMAGING AND MANAGEMENT

Sprengel's deformity is frequently noted at infancy due to the cosmesis, especially if grade III or IV, as well as due to the restricted movement at the glenohumeral joint (11). A portion of cases are diagnosed later in life incidentally, during an unrelated physical examination or radiological study, as was the case with our patient.

A standard postero-anterior (PA) projection of the chest or an antero-posterior (AP) shoulder radiograph should be taken as a first-line modality to diagnose and assess the severity of the deformity (1,12). It may be necessary to obtain an x-ray of the cervical spine, in order to assess the omovertebral bone attachment and for any related bony anomalies (12). Further imaging may be done using computed tomography (CT) or magnetic resonance imaging (MRI) for pre-operative planning (6,13). Pre-natal diagnosis is possible using ultrasound and is recommended for screening in Corno's disease (14).

Mild cases are typically managed non-operatively, using physiotherapy (2). Surgical correction of Sprengel's deformity is the management of choice for severe deformities with cosmetic and functional limitations. The ideal age range for surgical intervention is between the ages of three to eight (2); however older individuals may still be operated without issue. Two surgical approaches are most commonly used, namely the modified Green and the Woodward procedures, which both transfer muscle origins and include an osteotomy (15). According to Gonen, 2010 and Borges, 1996 an improvement in range of motion by up to 50 degrees may be seen post-operatively with either procedure (16,17).

CASE PRESENTATION

A 60-year-old man presented to the emergency department complaining of new onset lower limb oedema, affecting the right leg and extending up to the right shin. Further examination revealed occasional shortness of breath.

A chest x-ray was requested, and no cardiovascular or pulmonary abnormalities were shown (Figure 1). Sprengel's deformity of the left shoulder was noted incidentally, with presence of an omovertebral bone, extending from the medial angle of the scapula to C6 (marked by a red arrow on figures 1 and 2). Interestingly, the patient reported a history of spina bifida, which is commonly associated with this deformity, as discussed prior (1,11). The left shoulder was significantly raised with some restriction of movement present. Based on the radiographs available, this case would classify as a grade II deformity using the Rigault classification (10). An x-ray of the cervical spine (Figure 2) and left shoulder had been acquired two years prior, which clearly demonstrated the omovertebral bone as well as a mild cervical scoliotic curvature (4), which is also often found in individuals with Sprengel's deformity. No relevant further testing was performed and the patient was sent home later that day, considering that the deformity was asymptomatic and not pertinent to the presenting complaint.

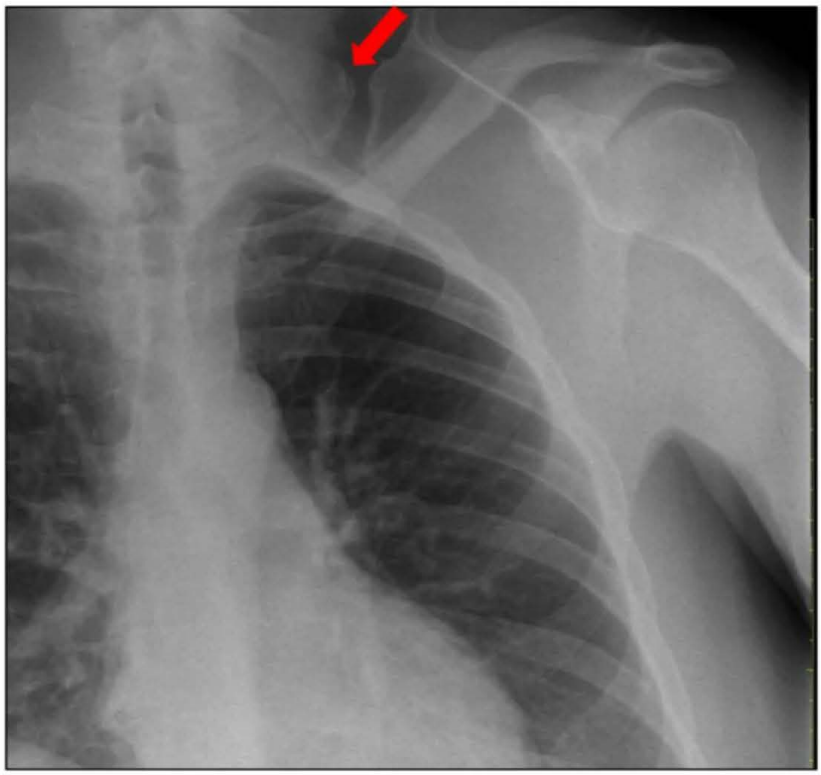

Figure 1: Cropped PA chest x-ray showing the deformity (arrow marking omovertebral bone)

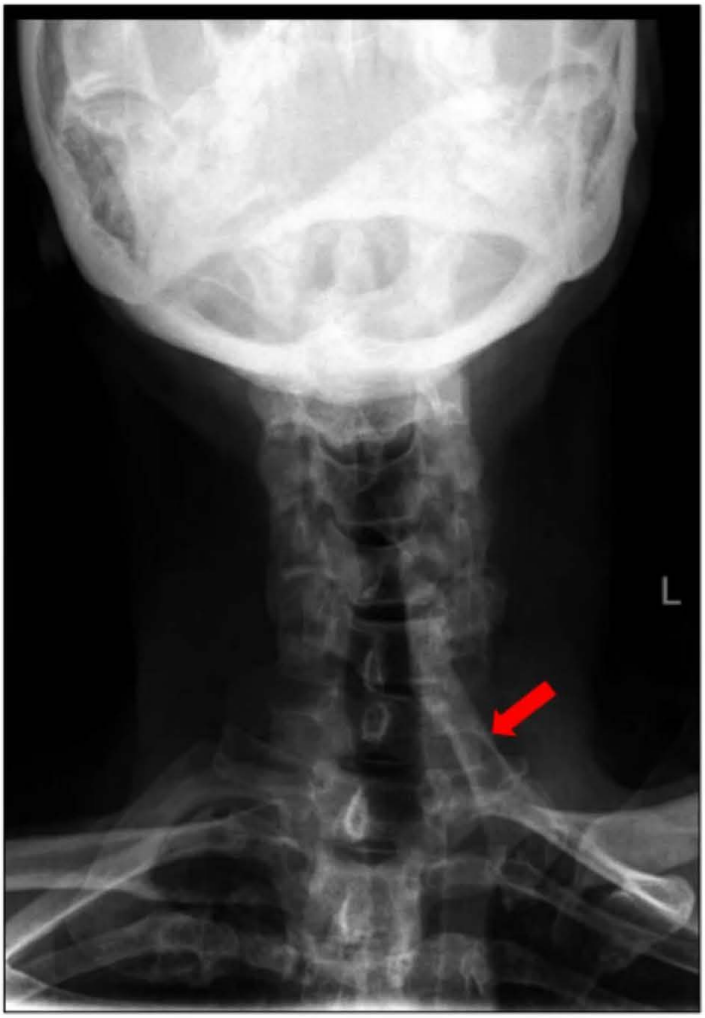

Figure 2: AP cervical spine x-ray showing the omovertebral bone (arrow)

CONCLUSION

This case report and brief review of the relevant literature outlines the nature of Sprengel's deformity: a relatively rare musculoskeletal disorder with varying degrees of severity. This particular case demonstrated that most cases of Sprengel's deformity are in fact detected incidentally and do not necessitate any intervention or follow-ups. With severe cases, detection often occurs at infancy and may require correction.

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