Sprengel's Deformity – A Review Of The Literature

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Abstract

Sprengel's deformity is a congenital anomaly of the shoulder, often seen in children[1]. In normal fetal development, the scapula moves down the back to rest in its normal position. However, children with the deformity have a scapula that sits too high on one side. Invariably such children will have limitations in movement of the affected shoulder. Problems such as scoliosis often co-exist and affected children are at greater risk of developing renal disease and conditions such as Klippel-Feil syndrome. This article reviews the literature to date, covering: the definition, epidemiology, aetiology, anatomy, clinical features, associated abnormalities, classification, diagnosis and imaging, treatment –both medical and surgical and current controversies around the area.

INTRODUCTION

Sprengel's deformity is a congenital anomaly of the shoulder, often seen in children[1]. In normal fetal development, the scapula moves down the back to rest in its normal position. However, children with the deformity have a scapula that sits too high on one side. Invariably such children will have limitations in movement of the affected shoulder. Problems such as scoliosis often co-exist and affected children are at greater risk of developing renal disease and conditions such as Klippel-Feil syndrome. This article reviews the literature to date, covering: the definition, epidemiology, aetiology, anatomy, clinical features, associated abnormalities, classification, diagnosis and imaging, treatment –both medical and surgical and current controversies around the area.

METHODOLOGY

Pubmed and the Cochrane Database were searched for full-text articles using synonyms for Sprengel's deformity as search words, including Sprengel's shoulder, Sprengel's anomaly, congenital high scapula, congenital elevation of the scapula, failure of scapular descent, high-grade dislocation of the scapula, hypoplastic scapula and elevated scapula.

DEFINITION

Sprengel's deformity is a rare congenital anomaly which arises from the failure of normal descent of the scapula, during embryological development, from its position in the neck, to its normal position in the posterior thorax[2]. It is characterised by elevation and medial rotation of inferior part of the affected scapula, which is also smaller and more cephalad than the normal scapula[3]. There is usually also regional muscle hypoplasia or atrophy, which adds to the disfigurement and limitation of shoulder movement[4]. It can be unilateral or bilateral and can occur in isolation or as part of a syndrome[2]. In around a third of patients, the affected scapula is attached to the cervical spine by an omovertebral bone, cartilage or fibrous tissue, which when present, makes abduction of shoulder beyond 90 degrees virtually impossible[2,3]. Sprengel's shoulder was first probably first described by Moritz Michael Eulenburg in 1863, as "hochgradige dislocation der scapula" (ie a high-grade dislocation of the scapula)[5]. Twenty years later, Willet and Walsham reported two cases with anatomic descriptions of this anomaly, and were the first to describe the omovertebral bone and reported good results in excision of this bone[6]. Sprengel then extended the knowledge base, by describing four more cases of the condition in 1891[7]. Kolliker also described four cases in 1891, and it was he who gave the condition its eponym, the Sprengel deformity[8]. Multiple case reports and new surgical techniques followed in the years afterwards.

EPIDEMIOLOGY

Sprengel's deformity is the most common congenital malformation of the shoulder girdle[9]. It is more common in males, with a male to female ratio of 3:1[4]. The prevalence and incidence are unknown, because many patients are asymptomatic.

AETIOLOGY

The condition is sporadic. Rarely, it shows an autosomal dominant pattern of inheritance, which is known as Corno's disease[2,10,11].

ANATOMY/EMBRYOLOGY

The scapula is a cervical appendage that normally forms opposite the fourth, fifth, and sixth cervical vertebrae at around the fifth week of gestation[12]. It normally descends to the thorax by the end of the twelfth week of gestation; any impediment to its descent before it reaches its final position results in a hypoplastic, elevated scapula, known as Sprengel's deformity. Poor development of bone, cartilage and muscle also usually accompanies this. The left side is more commonly affected than the right side[4]. The condition may sometimes be bilateral, which is more cosmetically acceptable but functionally more disabling.

CLINICAL FEATURES

The scapula is located higher than normal, by around two to ten centimetres[4]. It is adducted and is smaller than normal in the vertical plane and appears larger than normal horizontally. The inferior angle is medially rotated, causing the glenoid to face inferiorly. The higher the scapula lies, the less it is rotated. A visible lump in the suprascapular region is characteristic due to the upwardly rotated superomedial angle of the scapula, which causes the ipsilateral side of the neck to appear fuller. Occasionally, some anterior bending of the supraspinous portion is present.

An omovertebral connection exists in about one third of cases[9,13]. This is a rhomboid or trapezoid shaped structure that usually lies in a strong fascial sheath, which extends from the superomedial angle of the scapula to the spinous process, lamina or transverse process of the cervical vertebrae (most commonly the fourth to seventh cervical vertebrae). It may be fibrous, cartilaginous or bony. The omovertebral connection is usually unilateral. It is the primary cause of restricted shoulder motion in patients with Sprengel's deformity[14]. It is always associated with a fixed, elevated scapula[15] and has a major role in determining the shape and the malpositioning of the scapula[16].

The spinoscapular muscles are also adversely affected. The trapezius, rhomboid, serratus anterior, levator scapulae, pectoralis major, latissimus dorsi or the sternocleidomastoid muscle may be absent, hypoplastic or contain multiple fibrous adhesions. The trapezius muscle is the most commonly affected muscle[4]. If the serratus anterior muscle is weak, winging of the scapula may occur.

Generally, Sprengel's deformity tends to be painless and many patients are not diagnosed until adolescence[3]. On examination, passive movement of the glenohumeral joint, including initial abduction, external and internal rotation may be normal. However, scapulothoracic movements are usually limited. The most common limitations are abduction and forward flexion. In 40% of patients, abduction is limited to less than one hundred degrees[4]. This is more likely if an omovertebral bone is present[3].

ASSOCIATED ABNORMALITIES

Sprengel's deformity almost never occurs as an isolated malformation. It is usually accompanied by various other anomolies, especially in the cervicothoracic vertebrae or the thoracic rib cage. The most common anomalies are absent or fused ribs, chest wall asymmetry, cervical ribs, congenital scoliosis, and cervical spina bifida[4]. Syndromes associated with this condition include

- Klippel-Feil syndrome (characterised by the congenital fusion of any two of the seven cervical vertebrae)17, 18, 19, 20
- Greig syndrome (characterised by polydactyly, cutaneous syndactyly, ocular hypertelorism, macrocephaly and a high, prominent forehead)21
- Poland syndrome (characterised by hypoplasia or absence of the pectoralis on one side of the body and cutaneous syndactyly of the ipsilateral hand)22
- VATER association (characterised by vertebral defects, imperforate anus, tracheoesophageal fistula, radial dysplasia and renal dysplasia)23
- DiGeorge Syndrome (characterised by heart defects, cleft palate, autism, learning disabilities, recurrent infections and hypocalcemia)24
- Floating-harbor syndrome (characterised by short stature, delayed bone growth, delayed communication skills and distinct facial features)25
- Goldenhar syndrome (characterised by incomplete development of the ear, nose, soft palate, lip, and mandible)26

The most common syndrome is Klippel-Feil syndrome[4].

Other rarer syndromes include X-linked dominant hydrocephalus and mental disturbance syndrome[11]. A relationship between Sprengel's deformity and diastematomyelia (a condition in which a part of the spinal cord is split, usually at the level of the upper lumbar vertebra) has also been shown[4].

CLASSIFICATION

Sprengel's deformity can be classified by severity according to the Cavendish grades[27]. Grade 1 is the mildest, the shoulders are almost level and it cannot be noticed with clothes on. Grade 2 is also mild, but the superomedial portion of the scapula is visible as a lump. In grade 3, the deformity is moderate, visible and the affected shoulder is two to five centimetres higher than the normal shoulder. In grade 4, the deformity is severe, the scapula is very high, with the superomedial angle at the occiput, with neck webbing and brevicollis. One limitation of this classification is that it is difficult to apply in bilateral cases[4].

IMAGING

Sprengel's deformity is best seen on an anteroposterior radiograph of the chest and both shoulders[4]. The scapular displacement can be measured by a method described by Leibovic et al.[28], which uses three lines drawn on an anteroposterior radiograph, to calculate the superior scapular angle and the inferior scapular angle which give the viewer some idea about scapular rotation. CT scans may be performed to visualise the affected region, delineate the attachments of the omovertebral bone or to determine the presence of spina bifida occulta or an intraspinous lesion before surgery. Appropriate imaging studies should also be performed for any associated anomalies.

MEDICAL TREATMENT

Exercises can be used to maintain an individual's range of motion and to strengthen the weak periscapular muscles[4]. However, they are not always successful[3].

SURGICAL TREATMENT

Many patients with mild deformity do not require operative intervention. However, in patients with more severe disease, surgery is considered as first line treatment[4]. The main objectives of surgery are to improve the cosmetic appearance and to improve the scapular function[4]. The basic surgical technique is to first release the cause of the scapular binding and secondly to relocate the scapula[4].

CONTRAINDICATIONS

Contraindications to surgery include very mild deformity with a minimal restriction of movement or the presence of associated syndromes that may affect the final functional outcome[4].

METHODS

Surgical options include subperiosteal resection of part of the scapula, extraperiosteal release, transplantation of the muscular origins of the scapula, excision of the superomedial portion of the scapula, and vertical scapular osteotomy[29, 30, 31, 32]. Clavicle resection and excision of the omovertebral bone have also been described. Mears described a technique with partial resection of the scapula and a release of the long head of triceps, with good results[33]. A report by McMurtry et al, showed the results of vertical scapular osteotomy (with a mean follow-up of 10.4 years) to be favourable, with an increase in abduction of the shoulder of 53° and an improvement in cosmetic appearance by a mean of 1.5 levels on the Cavendish scale[34]. The ability to increase shoulder abduction with surgery is unfortunately limited and most of these procedures leave unsightly scars[4].

Although multiple surgical procedures have been described, the Green and the Woodward procedures remain the gold standards for correction of Sprengel's deformity[4].

The green procedure detaches the muscles from the scapula whereas the woodward procedure detaches the origins of the trapezius and rhomboids from the spinous processes[35].

The green procedure involves making a supraclavicular incision, in the supine position, and then gently producing a greenstick fracture of the clavicle. Then, in the prone position, through a midline incision, the trapezius and supraspinatus are sectioned. The omovertebral bar is excised. The insertions of the levator scapulae and rhomboid muscles are dissected and the supraspinous portion of the scapula is excised. The scapular attachments of the latissimus dorsi muscle are divided, the scapula is displaced distally and is fixed to the adjacent ribs. The muscles are then reattached[4].

The woodward technique involves resection of the omovertebral bone and division of vertebral spinous processes attachments of trapezius, rhomboids, and levator scapula, through a extended midline incision, with the patient in the prone position. The scapula is rotated and translated caudally. Detached muscles are then sutured back onto more inferior spinous processes. Postoperatively, the

arm is held in a sling for 3 weeks[4]. Overall, it has a 79% satisfactory functional and cosmetic result[36]. Increased shoulder abduction ranges from 34-60 degrees, depending on the child's age at operation and differing methods of measurement[3].

After surgery, the patient can be followed up in clinic at regular intervals to assess the surgical scar, scapular symmetry, presence and degree of winging, scapular range of motion, muscle bulk and strength[4].

COMPLICATIONS

The dorsal scapular nerve, spinal accessory nerve and suprascapular nerve can be injured during surgery[37]. The dorsal scapular nerve runs close to the superomedial border of the scapula and can be injured during dissection of the periscapular muscles at the superomedial angle of the scapula. The spinal accessory nerve is located between the trapezius and rhomboid muscles and is, therefore, at risk theoretically; however injury is uncommon when these muscles operate as a single unit. The suprascapular nerve runs in the suprascapular notch of the scapula and may be injured if the dissection is carried too far laterally when the superior portion of the scapula is resected. By staying at least one centimetre medial to the notch, the risk of injury can be reduced.

Winging of the scapula resulting from incomplete reattachment of the serratus anterior muscle and brachial plexus injury are other possible complications of surgery[4].

PROGNOSIS

Prognostic factors include the severity of the deformity, the age at surgery (generally, results of surgery in children older than age six years are not as good), type of procedure (relocation surgeries have better functional outcomes) and associated anomalies (anomalies such as Klippel-Feil syndrome compromise the eventual result)[4].

CONTROVERSIES

Surgery is indicated for children between three to eight years of age with significant deformities, both functional and cosmetic. Patients older than eight years of age are generally not good candidates for scapular displacement procedures[3]. However, in a study by Doita et al, the authors had good results after surgical correction in two adults, and they recommended surgery even in older patients[14]. The optimal age for operative intervention is still under debate[4].

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