Medicine Papers, 2015, 1 (1–4): 9–12

Sprengel deformity: a rare congenital anomaly of the scapula

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Summary
The authors report a case of a typical Sprengel deformity, a rare morphological anomaly which affects one or both scapula. The anomaly is more frequently a sporadic occurrence, but there are well-documented familial cases where inheritance has been reported.

Key Words
Orthopedics; Sprengel deformity; morphological anomalies; scapula.

Received 18.10.2015; accepted 11.11.2015; printed 30.11.2015

Introduction

Morphological anomalies of the scapula have been underrated in the literature due to the low incidence of this condition.

The most striking anomaly of the scapula is Sprengel deformity, which affects one or both scapula. Typically, the anomaly affects a single scapula, which appears located in a higher position and angles down towards the spine (Warkany, 1972). The anomaly is also known as scapula alata, undescended scapula, or scapula elevata, but the name "Sprengel deformity" is the most common.

The first report of the anomaly of the scapula is credited to Eulenburg (1863), who reported a single case. Twenty years later, Willet & Walsham (1883) reported an anatomical description of two patients, and Sprengel (1891) described a clinical report of four patients with this anomaly, which was later named after him.

The anomaly is rarely inherited and is more frequently a sporadic occurrence, but there are well-documented familial cases where a dominant pattern of inheritance has been reported (Wilson et al., 1997).

We report here a girl with a typical Sprengel deformity affecting the right scapula. The girl has been followed up for three years.

Case Report

The seven year old girl came to the Department of Orthopedics at the University Hospital "Vittorio Emanuele" in Catania, Italy, for a clinical work-up regarding her shoulder anomaly. She was born after a normal pregnancy and delivery. The parents are healthy and do not show clinical malformations; in particular, the location and symmetry of their scapula are normal.

The girl was born at term with a birth weight of 3,100 grams. Her height was 50 cm and her head circumference was 35 cm (all within normal limits).

The scapular anomaly was present since birth, but was initially unnoticed and only in the subsequent months was correct consideration to this anomaly given. Psychomotor developmental was normal. She attended the school with good performance. At physical examination, her weight was 18 kg and height 110 cm (within the third percentile). The neck was short. The right scapula was higher than

the left and was located within the superior border, just below the right clavicle. The right scapula was also shorter than the left, and sloped down at a lower angle that turns toward the spine (Figs. 1–3). She showed a mild cervico-thoracic scoliosis. The glenoid cavity is downward facing. No other malformation anomalies were seen. Examination of the chest and heart were normal, as were the genital organs. Neurological examination showed normal plantar reflexes, as well as good motor and language development. Right shoulder abduction was limited to less than 90°, with lack of motion of the scapula-thoracic junction and an inferiorly rotated glenoid.

Routine laboratory analysis was performed, including blood count, glucose, ketones, creatin-phosphokinases, and copper and thyroid markers. Audiometric examination, electro-cardiogram, and echocardiogram were all normal, as was a video-EEG conducted while asleep and awake. Skeletal X-rays showed the presence of a hypoplastic right scapula and scoliosis.

Surgical treatment was suggested,as the anomaly was moderate-severe, but the parents refused. Physical therapy was then carried out. We have followed the girl for three years, and have observed no clear modification of the scapular anomaly or of the scoliosis.

Discussion

The present patient shows a typical Sprengel deformity affecting the right scapula, which appears hypoplastic with reduced vertical length. The right scapula is superiorly placed when compared to its counterpart, and is medially rotated on the posterior thorax.

Sprengel deformity is a congenital, embryologic anomaly involving the development of the scapula and the surrounding structures of the upper extremity. The embryonic progenitor of the scapula appears during the fifth week of embryogenesis, and acquires its final morphology by the end of the eight week. The anomaly results from failure of the mesenchimal cells of the scapula to move from the cervical position to the normal thoracic location (Fealy et al., 2000). In this situation, descent does not happen, and the scapula tends to maintain the same shape as in the fetal form (i.e., longer in the transverse diameter than in the vertical). Clinically, the most evident presenting sign is an elevated scapula, which is also smaller, distorted, and with hypoplasia of the surrounding musculature. The anomaly is rarely limited to the scapula and the surrounding muscles: the nearest district may be shorter and the ribs deformed. The chest is frequently narrow and cylindroid (Fig. 3). The inferior section of the trapezius muscle is frankly hypoplastic, and the muscle hypoplasia may involve the Latissimus dorsi muscle and the rhomboids (Warkany, 1972). Klippel-Feil syndrome, Poland anomaly, Moebius syndrome, and diastematomyelia are some of the developmental defects that may be linked to Sprengel deformity in the anomalous embryonic development. In general, scoliosis is the most reported associated abnormality; in two large series of 112 and 77 patients, it was present in 35% and 55% of patients, respectively (Cavendish, 1972; Ross & Cruess, 1977). Other anomalies frequently associated with Sprengel deformity include Klippel-Feil syndrome (17–27%), rib anomalies (16-48%), omovertebral bone defects (20–50%), spina bifida (20–28%), and less frequently, clavicular abnormalities (1–16%), humeral shortening (6–13%), torticollis (4%), talipes equinovarus (1-3%), congenital dislocation of the hip (1–4%), pes planus (1–3%), femoral shortening (1%), and other conditions (1–3%) (Jeannapoulos, 1952; Cavendish, 1972; Ross & Cruess, 1977; Harvey et al., 2012).

The treatment of Sprengel deformity attempts to solve two main problems linked with this condition: cosmetic and functional impairments. The cosmetic abnormality is represented by the shape of the scapula, which is elevated and malrotated. The functional shoulder problem results in movement limitations, most notably reduced range of motor in shoulder abduction. Some classification systems have been proposed in an attempt to measure the different degree and severity of Sprengel deformity, with the aim of evaluating the effectiveness of surgical treatment and outcomes.

The Cavendish classification (Cavendish, 1972) is based on the degree of scapular elevation, which is separated into four grades. According to this classification, grade 1 (very mild) asymmetry cannot be observed if the patient is dressed; grade 2 (mild) refers to a shoulder that is almost in the correct position and deformity is visible as lump in the web of the neck, even when the patient is dressed; in grade 3 (moderate), the deformity has a shoulder elevation of 2–5 cm and can be easily seen; grade 4 (severe) refers to a shoulder that is markedly elevated,and the superior angle of the scapular lies at the level of the occiput (Cavendish, 1972).

Ross & Cruess (1977) suggested an evaluation system that measures the degree of shoulder elevation in relation to the top of the centre of the humeral head in relation to the trunk vertical axis. Radiographic classification has also been proposed by Rigault et al. (1976), which would distinguish between three grades of deformity on the basis of the projection of the

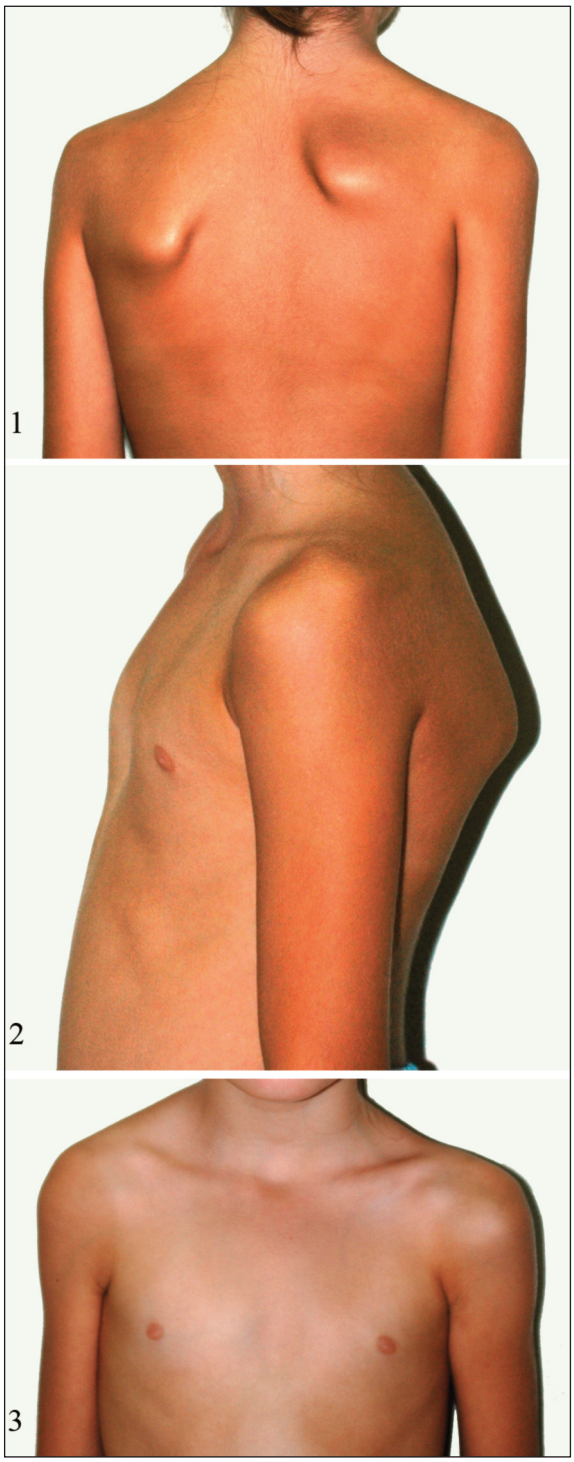

Figure 1. The girl, age 7, shows a right scapular Sprengel deformity; the scapula is just under the right clavicle.
Figure 2. The girl, age 7, shows cervico-thoracic scoliosis.
Figure 3. The girl, age 7, shows a profound cavity under the clavicle in the anterior part of the trunk.

supero-medial angles of the scapula. In addition, a great contribution came from the Magnetic Resonance Imaging with three dimension reconstruction, in order to diagnose concomitant abnormalities to reveal the anomaly in detail and thus create a better plan for treatment (Cavendish, 1972; Harvey et al., 2012).

The treatment of Sprengel deformity may be surgical or non-surgical, depending on the severity of the anomaly (Harvey et al., 2012). Non-surgical treatment is advised for children with little cosmetic deformity and mild abnormal motion, where physical therapy can allow the patient to achieve a good range of shoulder motion and prevent torticollis. Many surgical techniques have been developed to treat severe cases, and are generally divided into three categories (Rigault et al., 1976): 1) capsulectomy or scapulotomy, 2) scapula translation by replacing the insertion of the shoulder girdle muscles on the scapula, and 3) replacement of the shoulder girdle muscle from its origin to the vertebrae.

Surgical procedures have been proposed by Green, Leibovic (modified Green technique), and Woodward (Rigault et al., 1976; Harvey et al., 2012; Walstra et al., 2013; Barth et al., 2014). The latter is the most widely used procedure, and consists of the release and relocation of the origin of the muscle attached to the medial border of the scapula as well as the resection of any omovertebral bone. This technique may be associated with clavicular osteotomy. Recently, the Woodward procedure has been modified to ease the positioning of the glenoid cavity. Other surgical techniques are indicated in the most severe cases. They consist in splitting the lower portion of the trapezius muscle or the fibrous bands when present and extend/resect bone and muscle, with the aim of improved mobility. In the present patient, surgical treatment was not performed due to parent refusal, but it would have been useful, since the anomaly presented by the patient measures as a grade three under the Cavendish scoring system.

Finally, we want to emphasize that no clear modification of the clinical features (i.e., both cosmetic and functional) have been observed in three years of follow-up.

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