Sprengel’s Deformity with situs inversus in a neonate
Ghulam Nabi, MD and Omar Nabi Siraj, MBBS

Introduction:
Sprengel deformity or congenital elevation of the scapula is a disorder of development that involves a high scapula and limited Scapulothoracic motion. Scapula is elevated on the affected side of the shoulder (1). The left side abnormality is more common than the right (2). There is asymmetry of the shoulder if the deformity is Unilateral (3). The deformity produces cosmetic and functional impairment. Sprengel’s deformity also known as Sprengel’s Shoulder, Sprengel’s anomaly, congenital high scapula, Hypoplastic Scapula, Elevated scapula, Sprengel’s phenotype and Sprengel’s syndrome. The scapula originates in early embryogenesis at a level Posterior to the fourth cervical vertebra but descends during development to below the seventh cervical vertebra. Failure of this decent either unilateral or bilateral is the Sprengel deformity (4). Scoliosis is frequently present and torticollis is occasionally associated. The abnormality may be unilateral or bilateral it usually happens in girls and may have genetic basis in some instances. A familial form of the Sprengel’s deformity is known as Cornos disease. Other associations with this disease are abnormalities of Cervical spine such as Klippel Feil syndrome, Congenital scoliosis, Cervical ribs, Rib fusion, Cervical Spina bifida, Congenital kyphosis, Syringomyelia, platybasia, Omovertebral bone, Greig syndrome, Poland syndrome, VATER association, Velocardiofacial syndrome, Floating harbor Syndrome, Goldenhar syndrome. X linked dominant Hydrocephalus, skeletal anomalies, mental disturbance syndrome, situs inversus, mandibulofascial dysostosis, Ipsilateral shortened Humerus, clavicular anomalies and rarely renal anomalies (5,6).

Eulenberg first described this anomaly what later became known as Sprengel deformity in the year 1863. In 1880 Willet and Walsham reported first case involving an omovertebral bone. Sprengel then described 4 cases of upward displacement of the scapula in 1891(7). Kolliker reported several such cases in 1891. Multiple case reports and surgical techniques followed in literature for Sprengel deformity. Very scanty reports have emphasized the cardiovascular involvement in this anomaly. In this report we describe a female baby born with Sprengel’s deformity with situs inversus. To our knowledge this is the first case report from the Middle East.

Keywords: Sprengel’s deformity, Dextrocardia, Situs inversus.

Case Report: A female, full term baby product of spontaneous normal vaginal delivery. Apgar score of 6 & 8 at 1 & 5 minutes respectively. Growth parameters were normal (weight 2.7 kg, length 49 cm and head circumference 34 centimeters). Non consanguineous parents. Mother 26 years old, gravida six, Para five. All other siblings were normal. The baby was admitted in the nursery for congenital anomalies. On examination full term baby was active, pink, vital signs were normal. She had a short neck, bilateral webbing of the neck, and a low hair line. Asymmetry of the shoulder due to high position of the right scapula was confirmed by the X-ray of neck and chest. Cervical and thoracic spine was normal. Side to side neck movements were restricted. Movements of the shoulder joint were within normal range except restriction of abduction of right shoulder. X-ray neck and chest revealed that the position of the right scapula was higher than the left scapula. There was no fusion of cervical vertebrae and no omovertebral bone. The heart shadow was on the right side of the chest (See Figure 1).
Heart sounds were best heard on the right side of the chest, first and second heart sounds were normal there was no murmur. Peripheral pulses were felt and there were no signs of congestive cardiac failure, blood pressure in all the four limbs was normal, oxygen saturation on room air was 92%. The diagnosis of dextrocardia with situs inversus was confirmed by pediatric cardiologist, by clinical and echocardiography. Examination of the abdomen revealed liver one Centimeter below costal margin on left side, other systems were Clinically normal orthopedic surgeon was Consulted and he agreed with the diagnosis. The baby was active and sucking well. She was discharged next day and was advised to attend neonatal, cardiology and orthopedic outpatient Clinic. The patient was followed up over a period of one year and was growing normally.

**Discussion**

Sprengel’s Shoulder is the most common congenital malformation of the shoulder girdle. The male to female ratio is 3;1.(8,9). The condition is sporadic. Rarely it may run in families (autosomal dominant pattern of inheritance). The scapula is a cervical appendage that normally differentiates opposite the fourth, fifth and sixth cervical vertebrae at about five weeks gestation. It normally descends to the thorax by the end of the third month of intrauterine life. Any impairment to its descent results in a hypoplastic, elevated scapula (Sprengel deformity). The usual post migrational location of the scapula is between the levels of the second and eighth posterior ribs (10). Engel et al report no satisfactory explanation exists regarding the pathogenesis of the deformity. The hallmarks of this condition are shoulder asymmetry and restriction of shoulder abduction (11). Clinically the affected scapula is usually elevated 2-10 centimeter the deformity is characterized by elevation and medial rotation of inferior scapula. Involved scapula is both smaller and more cephalic than normal. In 30% of patients the scapula is attached to the cervical spine by an omovertebral bone, cartilage or fibrous tissue which when present can severely limit scapulothoracic motion. Deformity tends to be painless and many patients are not diagnosed until adolescence due to scapular asymmetry, some patients are mistakenly thought to have scoliosis. Minor asymmetries commonly seen between right and left scapula should not be designated Sprengel's shoulder, look for loss of shoulder abduction and forward flexion. If an omovertebral bone is present, abduction of the shoulder is commonly limited to less than 90 degree. The scapula is adducted and its inferior pole is medially rotated. Due to this rotation the glenoid...
faces inferiorly. Prominence in the suprascapular region is characteristic due to the upwardly rotated superomedial angle of the scapula. Passive moment of the glenohumeral joint including abduction, external and internal rotation may be normal. In 40% of patients with Sprengel’s deformity combined abduction is limited to less than 100 degree. The omo vertebral bone may also limit abduction by affecting Scapular mobility. The left side is more commonly affected than the right side. The condition may sometimes be bilateral in which case it is cosmetically much more acceptable but functionally it is more disabling. Based on the severity of the condition the most comprehensive information has so far been provided by a multi centre study published by Cavendish which involved 110 patients (12). Omo-vertebral bone is present in these cases between 18% and 60%. It’s complete removal is a part of the surgical treatment (3,14).

Sprengel’s deformity can be classified as follows (Cavendish grades).
Grade I: The deformity is very mild. The shoulders are almost at level and the deformity cannot be noticed with the clothes on.
Grade II: The deformity is mild; the shoulders are almost at level but the super medial portion of the high scapula is visible as a lump.
Grade III: The deformity is moderate; it is visible and the affected shoulder is elevated 2-5 cm higher than the opposite shoulder.
Grade IV: The deformity is severe; the scapula is very high with the super medial angle at occipital with neck webbing and brevicollis.

Imaging studies: Sprengel deformity is best visualized on the anterior posterior (AP) view of the chest and shoulders. Lateral view of the cervical and thoracic spine must also be obtained to rule out associated spinal anomalies (15). CT scan with 3-dimensional reconstruction may be performed to visualize the patho anatomy and the omovertebral bar. CT scan may also help in planning the surgery if indicated. Chinn reported that Sprengel’s deformity can be diagnosed ante-nataly by Ultrasonography (16,17). Physical therapy and exercises are used to maintain the range of motion and to strengthen the weak periscapular muscles. Many patients with Sprengel’s deformity do not require operative intervention. For those who do require surgery, the aims of correction of the deformity are two fold, first the cause of the scapular binding must be released and the scapula must be relocated. For functional and cosmetic reasons the ideal age of the surgery is between 3-8 years. Patients more than 8 years of age are not good candidates for this procedure.

Conclusion:
In mild cases treatment is generally unnecessary although a prominent and unsightly super medial corner of the scapula can be excised. Surgical procedures to remove the prominent superiomedial tip of the scapula are cosmetic and do not improve shoulder function. Procedures to bring about descent of the whole scapula are associated with significant potential complications and long sometimes ugly scarring (18). Clinicians caring for pediatric population should include this entity in the differential diagnosis of webbing of the neck.

References:

Author Information: Ghulam Nabi, MD is Pediatric consultant and Neonatologist, Department of Pediatrics, Bugshan Hospital, Jeddah, Kingdom of Saudi Arabia. Omar Nabi Siraj, MBBS is Registrar Department of Internal Medicine, Gulf Medical University Hospital and Research Center, Ajman, UAE. E-mail: omarsiraj@gmail.com

Corresponding Author: Ghulam Nabi, MD. Bugshan Hospital P.O.Box 5860. Jeddah 21432. Saudi Arabia. Telefax: 00966 02 6727284; Email: drgnabi2@gmail.com