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ORIGINAL ARTICLE

The congenital undescended scapula syndrome: Sprengel and the cleithrum: a case series and hypothesis

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Background: Sprengel deformity is a rare congenital shoulder girdle anomaly characterized by scapula malposition, periscapular muscle atrophy, and limited shoulder movement. Traditionally, it has been managed by omovertebral bar excision and muscle transplantation procedures guided by age and Cavendish grade. We present a unique observation in humans: a case series with Sprengel deformity possessing a cleithrum, an ancestral remnant of shoulder girdle development found in archaic bony fish.

Methods: Nine patients presented with so-called Sprengel deformity to a tertiary referral shoulder clinic. All were assessed clinically and radiologically with scapular radiographs and computed tomography or magnetic resonance imaging scans. The clinical and radiologic features were classified according to Cavendish and Rigault systems, respectively, and the scapular ratio was assessed.

Results: All patients were assigned grade 4 on the Cavendish scale. Six were grade 2 and 3 were grade 3 on the Rigault scale. The distinguishing pathoanatomic feature was partial endomuscular ossification of medial scapular suspension muscles, analogous to the cleithrum of bony fish. Five cases were treated operatively and 4 nonoperatively. Mean elevation and abduction significantly improved in surgical cases.

Discussion and conclusion: This finding not only challenges classic management of these rare patients but offers insight into scapular embryology and development. The association of scapular developmental and urogenital anomalies suggests that screening of the renal tract and genetic investigation in those with undescended scapula syndrome be considered. We suggest, to emphasize the nature of incomplete scapular descent and associated congenital anomalies and to clarify imprecise common use of the term Sprengel deformity, that this condition be called the congenital undescended scapula syndrome.

Level of evidence: Level IV; Case Series; Treatment Study

Keywords: Cleithrum; Sprengel; scapula; endomuscular ossification; developmental; congenital undescended scapular syndrome; embryology

Sprengel deformity is a rare congenital anomaly of the shoulder girdle characterized by fixed elevation and lateral rotation of the scapula, with hypoplasia or atrophy of periscapular muscles causing disfigurement and limited

Institutional Review Board approval is not required for this case series.

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shoulder movement.[3] Eulenberg (1863)[6] first reported 3 cases of congenital undescended scapula, and Willett and Walsham in 1880[21] and 1883[20] later reported 2 more cases with anatomic descriptions of the condition. Sprengel and Kolliker reported cases in 1891; Sprengel suggested an etiology, and the condition has subsequently been referred to as Sprengel deformity.[2,10]

We describe 9 patients treated for an extreme form of Sprengel deformity. The genetic basis for and understanding of scapular embryology[14] suggest that the so-called elevated scapula represents a failure of somite differentiation as well as failure of descent relative to the neuraxis. The omovertebral tether described in our series takes the form of a sheet of bone replacing the medial scapular suspension muscles rather than interposed between them and the spine. This is equivalent to the cleithrum of archaic bony fish,[12] also present in modern amphibia. We postulate that in common with other congenital anomalies of failure of segmentation within the neuraxis and associated somitic ectodermal anlagen, the condition labeled Sprengel deformity is a spectrum of failure of somitic ectodermal differentiation and migration. In this paper, we describe the characteristics and management of a cohort of patients with this extreme variety of failure of descent of the scapula associated with a complete bony scapulospinal tether. We recommend that the Sprengel deformity be called the congenital undescended scapular syndrome to emphasize the embryologic basis for and the spectrum of anomalies present in this condition.

Materials and methods

This is a retrospective case-control series of 9 patients with 11 affected shoulders presenting with a Sprengel deformity to our tertiary referral center from 2008-2013. This represents the entire cohort of such cases referred to this institution during the period and, as such, cannot be used to determine the frequency with which this more extreme type of the so-called Sprengel deformity might be present in the community. The mean age at presentation was 15.3 years (median, 17 years; range, 3-32 years). We assessed patients with scapular radiographs and computed tomography or magnetic resonance imaging in all cases. The cervical spine was imaged if symptoms or deformity was present. The range of movement of the affected and nonaffected limb girdles was documented before and after intervention. All patients operated on were followed up for a mean of 24 months (range, 6-36 months). Patients who were not operated on were followed up for a comparable mean time of 22 months (range, 6-34 months).

Indications for operation included periscapular pain and prominence of the superior pole causing discomfort (in all cases) and intrusive restriction of motion (in 4 cases). The influence of scapular tethering on cervical or cervicothoracic spine growth was considered in the juvenile cases but was not used as an absolute indication for intervention. We considered the patient's preference for operative and nonoperative intervention in the decision-making. The range of motion was estimated with a hand-held goniometer using the neutral-zero method.[19] The preoperative and postoperative ranges of motion were compared using Student t-test with the significance level set at $P = .05$. Nonparametric data (Cavendish and Rigault scores) were analyzed using median, mean, mode, and Fisher exact test.

Surgical technique

A dorsal longitudinal incision was made parallel to the medial border of the "true" scapula, judged by a line joining the superior-medial pole with the inferior pole. The trapezius was identified and either elevated to expose the cleithrum, if it arose from the medial border of the scapula caudal to the spine of the scapula, or detached from the spine of scapula for later repair, if the cleithrum arose from the medial border proximal to the spine of scapula. The scapulae were rhomboidal, with a pseudarthrosis between the cleithrum and true scapular body in all cases. The cleithrum was identified, and the anatomy of the medial scapular suspension muscles was established as far as could be determined (Fig. 1). In all cases, the rhomboid major and minor were completely or nearly completely replaced by bone. The cleithrum was excised subperiosteally (there was always a well-formed periosteum) (Fig. 2). Osteovertebral pseudarthroses were present in all cases (Figs. 3-5). The cleithrum resembled a rudimentary rib in one case; the remainder were triangular.

The deep surface of the scapula was inspected for further anomalies (none were found in this series). The remnants of the medial muscles were sutured together if possible, and the trapezius was

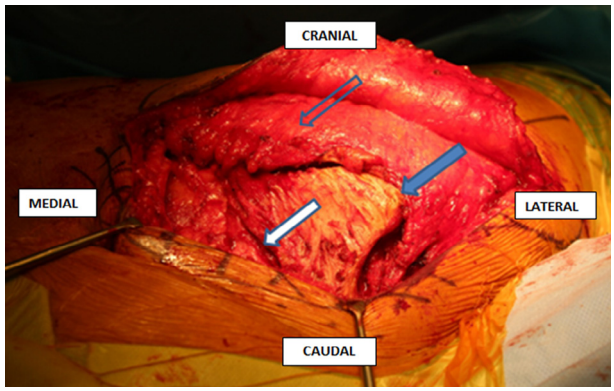

Figure 1 Cleithrum exposed by subperiosteal dissection. The patient's head is to the left. The trapezius is indicated with the *open arrow* and the medial border of the scapula with the *closed arrow*. The pseudarthrosis between the cleithrum and body of scapula is indicated with the *white arrow*.

Figure 2 Excised cleithrum.