Journal of Research and Practice on the Musculoskeletal System
JRPMS

Review Article

Congenital Disorders of the Shoulder Girdle: A Review of the Literature

Stefania Kokkineli[1], Christos Zafeiris[2], Efstathios Chronopoulos[2]

[1] *8th Orthopedic Department, Shoulder Unit, General Hospital Asklepieion Voulas, Athens, Greece;*
[2] *Laboratory of Research of the Musculoskeletal System, National and Kapodistrian University of Athens, KAT Hospital, Athens, Greece*

Abstract

The shoulder girdle, a complex anatomical structure, plays a pivotal role in upper limb mobility and function. Congenital disorders of the shoulder girdle encompass a spectrum of rare developmental anomalies that affect the structure and function of the clavicle, scapula, and surrounding musculature, ranging from mild deformities to severe structural impairments. These conditions can lead to deformities, impaired mobility, and functional limitations, often impacting patients from early childhood. This narrative review explores etiology, clinical presentation, diagnostic strategies, and treatment options for congenital disorders such as Sprengel's deformity, cleidocranial dysplasia, glenoid hypoplasia, os acromiale, humerus varus, congenital deltoid fibrosis and congenital clavicle pseudarthrosis. By providing a comprehensive overview, this review aims to improve early recognition and management of these disorders.

Keywords: Shoulder girdle, Sprengel deformity, Cleidocranial dysplasia, Os acromiale, Clavicle pseudarthrosis

Introduction

The shoulder girdle, a complex anatomical structure, plays a pivotal role in upper limb mobility and function. Congenital disorders of the shoulder girdle are a rare but important group of musculoskeletal anomalies that result in notable cosmetic deformity of the shoulder complex. These disorders can result in varying degrees of deformity, restricted mobility, and functional impairment, significantly impacting patients' quality of life. Understanding embryological development, clinical presentation, and genetic factors underlying these conditions is crucial for accurate diagnosis and effective management. This narrative review aims to provide a comprehensive overview of congenital shoulder girdle disorders, such as Sprengel's deformity, cleidocranial dysplasia, glenoid hypoplasia, os acromiale, humerus varus, congenital deltoid fibrosis and congenital clavicle pseudarthrosis highlighting their etiology, clinical manifestations, diagnostic approaches, and treatment options.

Sprengel' s deformity

Sprengel's deformity or congenital high scapula is the most common congenital deformity of the shoulder girdle[1]. The frequency rate of the deformity is three times higher in males than females. It is unilateral in 80% of cases or bilateral because of abnormal descend during the embryonic period[2]. Eulenberg was the first who described this rare deformity, but in 1891 it was associated with the name of Otto Sprengel who further presented four clinical cases and proposed an etiology of this rare disorder[1,3].

The authors have no conflict of interest.

Corresponding author: *Stefania Kokkineli, MD, 8th Orthopaedic Department, Shoulder Unit, General Hospital Asklepieion Voulas, Athens, Greece. Dardanellion 97, 16562, Athens, Greece*

E-mail: *stephaniekokkineli@gmail.com*

Edited by: *Konstantinos Stathopoulos*

Accepted *30 December 2024*

The origin of the word "scapulae" is Greek from the word "scaptein" which means dig. Many authors tried to "dig into" the pathogenic mechanism of Sprengel deformity. The clinical spectrum of Sprengel deformity is the result of failed ectodermal differentiation of the somites and scapula descent in relation with the neuroaxis[3]. The absence of signaling molecules or cellular receptors is responsible for phenotypic mutations affecting not only normal development of the scapula but also the surrounding structures[1]. Disruption of cell signaling negatively affects the differentiation of mesenchymal cells to skeletal tissues and leads to malposition of the scapula[1,4]. EMX2, which is the responsible gene for scapula development, also regulates somite differentiation and the development of a bone, which connects the scapula with the cervical spine[5].

Sprengel deformity may be inherited in an autosomal dominant way or the result of subclavian artery lesions[1]. The theory that has been proposed is interruption of the blood supply during the embryonic period[6,7]. The autosomal dominant fashion characterizes the Hodgson-Chiu syndrome, which includes cervical anomalies, Sprengel deformity and cleft palate[8]. The embryological link between the neck and the shoulder region explains the associated anomalies in patients with Sprengel deformity[8]. In general, Sprengel anomaly is observed in 1 to 2% of patients with deletion of 22q11.2 chromosome. Both Sprengel deformity and deletion 22q11.2 syndrome are the genetic result of neural crest anomalies[8]. Pargas et al first reported Sprengel' s deformity in 2 biological sisters, who were both treated surgically[6].

Sprengel deformity is generally diagnosed in children younger than 8 years. The clinical spectrum ranges from mild deformity to severe clinical dysfunction of shoulder joint and cervical spine[1,9,10]. Patients may present with elevated rotated, dysplastic, rhomboid- shaped scapula, with anteroinferior rotation of the glenoid and medial translation of the convex side of the scapula in contact with the inferolateral portion of the omovertebral bone[3,4,6]. The inward rotation of the scapula leads to downward facing of the glenoid cavity and limited shoulder abduction to 90º or less and is associated with cosmetic impairment combined with muscle atrophy of the trapezius, levator scapulae and the rhomboids[1,3,4,6,11]. Progressive periscapular muscle stiffness further increases glenohumeral joint stiffness[12]. Associated deformities may also be present, such as shorter clavicle, deformities of the cervical and thoracic spine and rib cage, an omovertebral bone between the cervical spine and the scapula (25-50%) and muscular atrophy leading to functional impairment of the shoulder joint[1,2,9].

The most widely used classification system in Sprengel deformity is the Cavendish classification system, which was first described in 1972, and it is based on severity of the deformity and the grade of scapula elevation[1,3,4,6,11]. Rigault et al further classified patients with Sprengel deformity according to the vertebral level of protrusion of the superior and medial aspect of the scapula in plain X-rays[1,3,6].

The omovertebral bone, like the cleithrum of a bony fish, consists of a rhomboid- or trapezoid- shaped, fibrocartilaginous, or osseous connection between the superomedial surface of the scapula and the lamina, spinous or transverse processes of the cervical spine, from the level of C4 to the level of C7. It was observed in 1883 by Willet and Walsham, and it is present in 16% to 65% of patients with Sprengel deformity[13-15]. The omovertebral bone either forms a joint with the scapula or presents an ossified bridge connecting the scapula with the lower cervical spine. It is not known if it first develops from the vertebra, the scapula or the periscapular muscles and it is usually unilateral[13,14]. The excision of the omovertebral bone is performed at the age of 3 to 8 years for cosmetic and functional improvement with lower risk of brachial plexus injury[13,14,16,17,18].

The relation of Sprengel deformity to other congenital anomalies is the result of specific genetic disorders. Klippel- Feil syndrome is observed in 7% to 42% of patients with elevated scapula[1,3,6,10,11,19]. Other associated anomalies and syndromes include congenital scoliosis (35-55%), spina bifida occulta, cervical spine deformity, syringomyelia, posterior congenital fusion of the thoracic spine, spinal dysraphism, absent or fused ribs (16-48%), malformation of the cardiopulmonary system (4-14%) and the genitourinary system (35%), colon abnormalities, cleft palate, facial anomalies, Poland syndrome (15.9%), Arnold-Chiari malformation, myelomeningocele, Mobius syndrome, X-linked hydrocephalus, diastematomyelia and mental disturbance syndrome[1,6,8,20].

Non operative treatment of Sprengel deformity is indicated in patients with grade 1 or 2 cosmetic deformity according to Cavendish classification and mild restriction of shoulder motion[1,10,11]. Conservative treatment includes rehabilitation programs and participation in sports[11]. Evaluation of the patients' shoulder range of motion, cosmetic appearance and psychological development should be performed every year until they are skeletally mature[1,11].

Surgical treatment is performed in children, 3 to 8 years old with higher grades of aesthetic deformity (Cavendish grades 3 and 4)[1,3,10,11,17]. In case of concomitant spinal abnormalities, surgical treatment is suggested at 2 years of age followed by Sprengel deformity surgery[21]. Surgical options include partial scapulotomy or resection of the elevated scapula and the omovertebral bone and caudal translocation of the scapula in reference to the contralateral scapula after muscle detachment and reinsertion[17]. Brachial plexus and first rib decompression is performed before scapula mobilization with resection osteotomy of the mid-clavicle[1].

The most popular and effective surgical techniques for scapula relocation in Sprengel deformity repair are the Woodward and Green procedures followed by scapular osteotomy and partial scapulectomy[1,17,21,22]. Muscles' origin (Woodward procedure) or insertion (Green procedure)

translocation is important for prevention of varus position of the glenohumeral articulation. Brachial plexus injury could be avoided by a single- level clavicular osteotomy through a small incision[1,11,18,22,23]. Scapulectomy was first described by Wilkinson and Campbell[1]. A similar surgical technique was later described by Mears[1,21]. Partial scapulectomy also seems to improve shoulder abduction[1]. In case of presence of the omovertebral bone, extra- periosteal removal is performed[17].

Several studies demonstrated the results of the Woodward and Green procedures in patients with Sprengel deformity. Oner et al showed great improvement in shoulder abduction and flexion in 17 patients (18 shoulders) at 62.9 months postoperatively[24]. Yamada et al also showed great increase in shoulder flexion and abduction and cosmetic improvement in 7 patients with a mean age of 50.9 months at 53.1 months after modified realignment of scapula rotation whereas Agarwal et al demonstrated the effect of rib anomalies on the clinical outcomes 4 years after vertical scapulectomy in 8 patients with Sprengel deformity with a mean age of 6.8 years[25,26]. Clinical outcomes were better in patients with simple or no rib anomaly[25].

Postoperatively, the patients wear a sling for 3 weeks. At 3 to 4 weeks passive motion is allowed[6,10]. Postoperative complications include hypertrophic scarring, deformity recurrence and winging of the scapula, asymmetry, brachial plexus injury, poor functional outcomes and infection[1,10,11,17,23]. Wound complications and postoperative pain, infection rate and recovery time could be further decreased by performing the endoscopic Woodward procedure described by Soldado et al in 2017[27].

Glenoid Hypoplasia

Congenital glenoid hypoplasia is hypoplasia of the glenoid as a result of failed development of the lower part of the glenoid and the neck of the scapula[28-30]. Hypoplastic glenoid was reported for the first time by Giongo and Heupke in 1927-1928 and by Valentine in 1931 and is characterized by shallow glenoid cavity, rounded posteroinferior glenoid rim and hypertrophic posterior labrum[28,31-35]. This rare anatomic deformity can be asymptomatic, so its true prevalence is not known[29,30,32,36]. Most patients are male, and, in most cases, it is bilateral and symmetric and is an incidental finding in plain radiographs[36-38]. It may also be combined with hypoplastic, subluxated humeral head, hypertrophic glenoid labrum and widened glenohumeral joint space, hooked distal part of the clavicle and hyperplastic, elongated acromion and coracoid process[28-31,35].

Congenital glenoid hypoplasia may be associated with genetic disorders in a familial pattern as it is inherited in an autosomal dominant way and present as a clinical aspect of clinical syndromes[28,30,33,35,38,39]. The glenoid ossifies from two of the eight ossification zones of the scapula during the embryonic period. The proximal (superior) ossification center presents under the coracoid process at the age of ten years and fuses at the age of fifteen years, whereas the distal ossification zone presents between the 9th and 12th years as a horseshoe- like (U shaped) glenoid epiphysis that is thicker peripherally (glenoid rim) and thinner in its center (glenoid fossa)[33-35]. Both ossification centers are present between the age of 10 and 12 years[34,37]. Glenoid hypoplasia is the result of ossification arrest. This theory was first described by Owen in 1953[37]. According to this theory, failed development of the lower precartilage leads to replacement of hypoplastic bone by fibrocartilaginous tissue as a compensatory mechanism[28,30,34,36,38].

Patients with glenoid hypoplasia may have no symptoms or complain about painful, restricted function, increased glenoid retroversion or shoulder instability leading to degenerative changes and posterior labral tears, probably in adolescence, at second decade of life or later as glenohumeral arthritis, at fifth decade of life[28,30,34,37]. Glenoid hypoplasia has also been associated with a skin web in the axilla and hypoplastic pectoralis muscles[29,36]. Hypoplastic glenoid may appear as the only, primary lesion or be present in patients with multiple deformities such as Erb' s palsy, muscular dystrophy, spina bifida, Apert syndrome, Holt- Oram syndrome and mucopolusaccharidoses- related conditions[31,35,37,40].

Hypoplastic glenoid is usually an incidental finding in plain chest radiographs[28,29,32]. Hypoplastic glenoid can be further evaluated by CT[28,29,37]. The most reliable modality for the evaluation of the characteristics of glenoid hypoplasia is Magnetic Resonance Imaging (MRI)[28,30]. Shoulder arthroscopy reveals the absence of the inferior osseous element of the glenoid, thickening of the glenoid cartilage, labral tears and perilabral cysts[34,36,38].

The method of treatment depends on various factors, such as the age of the patient, pain, grade of degeneration and level of restriction in everyday activities[28]. Conservative treatment includes daily shoulder exercises for stabilization of the periscapular muscles and mobilization for releasing soft – tissue adhesions[28,29,34,35]. Surgical treatment is indicated in patients with persistent symptoms after six months of physical therapy and symptomatic labral tears with significant shoulder instability and painful restriction of range of motion[28,30,36].

Surgical options include labral tears repair combined with a bone graft and shoulder arthroplasty in patients with advanced degenerative disease with persistent symptoms[28,30,32,35]. Postoperative complications include non-union, resorption of graft and shoulder instability[28,34]. Postoperative rehabilitation includes passive motion at 4 weeks after surgery, active motion at 8 weeks postoperatively and recreational activities at 24 weeks after surgery[28].

Humerus Varus

Humerus varus is defined as the disruption of ossification of the growing ends of the humeral bone, which results in the medial migration of the humerus with relation to its

longitudinal axis. A physeal bar may be present in the inferomedial plate as the result of the growth disruption and it is combined with metaphyseal narrowing. Humerus varus is one of the two most common varus deformities in long bones. Isolated congenital varus deformity of the humerus is extremely rare, and it may be caused by genetic and metabolic disorders. Bilateral, symmetrical varus deformity is also suggestive of intra- uterine injury of the plate of the epiphysis at the upper part of the humeral bone leading to congenital humerus varus[41-43].

Patients usually present for cosmetic reasons because of the shortening of the humerus. The shortened humerus is the result of the partial arrest of the proximal epiphyseal plate. The reduced acromiohumeral distance leads to subacromial impingement and decreased forward flexion and abduction[41-43].

The radiographic characteristics of humerus varus were first defined in 1935 by Kohler et al[42]. In 1991 Herschkovitz et al also demonstrated the varus humeral head, the shortened humerus, and the lytic defects of the shaft cortex as the main characteristics of the growth plate arrest in congenital humerus varus deformity[43]. CT and MRI are also important tools in preoperative planning for detecting any additional components to varus deformity[42]. Surgical treatment of humerus varus should avoid disruption of the epiphyseal plate and soft tissue injury while minimizing the risk of postoperative nonunion or prolonged immobilization[42].

Congenital Deltoid Fibrosis

Congenital deltoid fibrosis or contracture is an infrequent congenital deformity with bilateral, symmetric presentation and delayed diagnosis because of symptomatic presentation at children aged between eight and twelve years[44-47]. Fibrosis is more common in the middle deltoid fibers. As the contracture becomes more extensive and the length of the band does not increase, patients present with painful restriction of shoulder abduction[44,46,47]. Contracture for a long time creates a fibrous cord- like band in the skin and as it becomes more extensive it may lead to anterior migration of the humerus with relation to the glenoid[44,45,47]. Clinical examination and radiographic evaluation reveal scapular winging, laterally rotated scapula and beading of the acromion[45-47]. The characteristic skin dimpling in the middle portion of the deltoid also excludes brachial plexus palsy, long thoracic nerves paresis and scapula tumors from the differential diagnosis[44,47].

Surgical treatment includes proximal deltoid release or distal in case of extensive contracture combined with excision of the fibrous band[44,45]. Operative treatment offers pain relief and improves function Postoperative rehabilitation is necessary for deltoid muscle strengthening 2 to 3 months after surgery[46]. Postoperative complications include recurrence (0-6%), intramuscular hematoma (0-5%), decreased power of the deltoid, inadequate muscle release, shoulder deformation and wound complications (0-16%)[44].

Os Acromiale

Os acromiale is caused by failed fusion of the primary acromial ossification zones and was first described by Gruber in 1863[48,49]. It is usually an incidental finding, and it is considered as a predisposing factor for rotator cuff tears and subacromial impingement[49,50]. Os acromiale is more common in Africans (frequency rate 18.2%) and African- Americans (11-20%) and in the male gender[48-52]. The frequency rate is up to 18.2 (mean rate 7-8%) in the general population[48,49,51]. Os acromiale is bilateral in 24% to 62% of the patients[50,51,53].

Several ossification centers are responsible for the formation of the acromion, which starts at the age of 12 to 14 years[52,54]. Three of four elements, the anterior acromial element which is called the “preacromion”, the middle element known as the “mesoacromion” and the posterior element, the “metaacromion” are the result of fusion of several ossification centers forming a triangle-shaped bone[48,49]. This epiphyseal bone fuses with the fourth element, the “basiacromion” at the age of 22 to 25 years[51]. The final element, basiacromion, fuses with the spine of the scapula in children aged 12 years. The bridging procedure of these elements begins from posterior to anterior[55]. Failed ossification of the preacromion, mesoacromion, metaacromion and basiacromion after the age of 25 leads to the anatomic deformity of os acromiale[48,49,51,56].

Genetic predisposition and mechanical recurrent overload or even a single traumatic event during acromion development are the main causes of os acromiale as they terminate the osseous bridging of the bony elements[51,52]. According to Lieberson Classification, there are four types of os acromiale based on the nonunion site[54].The site of os acromiale is defined based on the element anteriorly to the nonunion site which failed to fuse[49,51]. Meso-type os acromiale is the most common type (range 75-94.4%) followed by pre- type os acromiale (15%) whereas meta-type os acromiale acromion is the less common type (2%)[48,49,51].

Os acromiale can be identified as an incidental finding in plain radiographs[48,50,51,57]. Differential diagnosis includes coracoacromial ligament ossification and acromial spur fracture[53,57]. Ultrasound and CT could give further detail about the morphological characteristics of the bony margins[49,57]. However, the more specific imaging modality for diagnosis and assessment of os acromiale is MRI[48,49,51,52].

Most patients with os acromiale are asymptomatic[53,56]. However, young patients with overhead activity may present with superior or anterolateral acromial pain, pain at night, muscle weakness and limited range of motion[48,49]. Os acromiale may be slightly mobile in some cases because of almost complete fusion[51,54]. Either micromotion and inflammation at the nonunion site or dynamic outlet-based subacromial impingement of the unfused segment during arm elevation could cause painful loss of range of motion[49,51,54,56,58]. A direct trauma to the shoulder region

or bone removal of the unfused region during subacromial decompression could also cause micro- instability at the previously stable os acromiale and pain[48]. Gross motion of the anterior part of the acromion would be highly suggestive of symptomatic os acromiale[49].

Conservative management includes medicine and avoidance of repetitive trauma during overhead and throwing activities for at least 6 months[56,59]. Physical therapy should apply to the protocol of subacromial impingement and may be helpful as deltoid attachment spreading to non- ossified sites of the acromion offers stability during non- operative treatment[56,59].

Surgical treatment is considered after 6 to 12 months of failed conservative treatment[55,58-60]. Surgery is performed in children with persistent pain and unstable os acromiale and includes internal osteosynthesis or removal of the unfused element[56,58-64]. Excision of the sclerotic edges of the two elements is necessary to achieve fusion[48,49]. Acromioplasty could be avoided by the upward- tilted repair of the anterior element of os acromiale to increase the subacromial space[49,56]. In arthroscopically assisted surgery, the use of biodegradable screws with strong polyethylene sutures is associated with better outcomes avoiding complications related to hardware. Excision of os acromiale is indicated in case of a small preacromion fragment to prevent deltoid dysfunction[52,56,58,65,66]. Arthroscopically assisted reduction and internal fixation offers minimal deltoid injury and postoperative weakness and better cosmetic results[49,51,58].

Congenital Clavicle Pseudarthrosis

Congenital pseudarthrosis of the clavicle is an infrequent disorder, which is the result of disruption of the continuity of clavicular diaphysis[67]. It is a genetic condition, less common in males than females[68]. The ossification of the clavicle first begins in the 5th week of the embryonic period. The failure of fusion of the internal and external ossification centers of the clavicle leads to the above anomaly[69]. Other etiological factors are the compressive forces exerted by vascular compression as well as the accessory ribs[70]. In most cases it appears on the right clavicle, while in 10% of cases it occurs bilaterally[71]. Less commonly it occurs on the left side in case of situs inversus[69,72,73].

The first case of was reported in 1910 by Fitzwilliams in the context of clavicocranial dysostosis and twenty years later by Saint-Pierre[69,74]. Patients present with a visible, not painful protrusion in the diaphysis of the clavicle until the age of eight years[72,75,76]. Rarely, this abnormality is associated with the thoracic outlet syndrome[77]. Regarding the radiological features, the sternal end of the clavicular bone is larger and is located anterosuperiorly, while the distal end of the clavicle is located inferiorly directed upward and medially. At the site of the pseudarthrosis, the anteroposterior view of the clavicle reveals rounded scleral bone[71,78]. The differential diagnosis includes birth injuries of the clavicle, which are usually accompanied by rib fractures, neurofibromatosis and cleidocranial dysostosis[75,79]. Surgical management includes excision of the unfused bone and internal osteosynthesis using autologous bone graft (Masquelet technique). Postoperative rehabilitation includes shoulder immobilization for 3 weeks[80-83].

Cleidocranial dysplasia

Cleidocranial dysplasia is a genetic condition named by Marie and Sainton in 1898. It is the result of autosomal dominant inheritance and includes disorders of the shoulder girdle and the viscerocranium that has the shape of an inverted pear[84-86]. It occurs with the same frequency in both sexes and mainly affects the skull, the distal end of the clavicle, the ribs, the teeth and the pubic symphysis[86,87]. Patients present with a longer neck, narrowing of the space between the axillae, scoliosis, glenohumeral instability and lengthening of the proximal and distal epiphyses of the metacarpals and metatarsals[85,88]. Surgical treatment is indicated in patients with secondary neurovascular lesions due to clavicle deformities[89].

Conclusion

Congenital disorders of the shoulder girdle, though rare, present a wide range of clinical manifestations, from mild deformities to significant functional impairments. Early diagnosis is crucial for managing these conditions effectively and improving patient outcomes. This narrative review highlights the importance of understanding the underlying pathophysiology, clinical presentations, and diagnostic challenges associated with these disorders. Treatment approaches, whether conservative or surgical, should be individualized. Ongoing studies are needed to increase diagnostic accuracy and therapeutic interventions. Further genetic investigation is the key to prevention and primary management of congenital disorders of the shoulder girdle.

References

1. Harvey EJ, Bernstein M, Desy NM, Saran N, Ouellet JA. Sprengel deformity: pathogenesis and management. J Am Acad Orthop Surg 2012;20(3):177-86.
2. Khan Durrani MY, Sohail AH, Khan I, Hashmi P. Sprengel's Deformity. J Ayub Med Coll Abbottabad 2018;30(1):135-7.
3. Dhir R, Chin K, Lambert S. The congenital undescended scapula syndrome: Sprengel and the cleithrum: a case series and hypothesis. J Shoulder Elbow Surg 2018;27(2):252-9.
4. Öner A, Aşansu MA, Akman YE. Sprengel Deformity: Comprehensive Evaluation of Concomitant Spinal and Extraspinal Anomalies in 90 Patients. Spine (Phila Pa 1976) 2020;45(18):E1150-E7.
5. Pröls F, Ehehalt F, Rodriguez-Niedenführ M, He L, Huang R, Christ B. The role of Emx2 during scapula formation. Dev Biol 2004;275(2):315-24.
6. Pargas C, Santana A, Czoch WL, Rogers KJ, Mackenzie WG. Sprengel Deformity in Biological Sisters. J Am Acad Orthop Surg Glob Res Rev.2020;4(4).
7. Bavinck JN, Weaver DD. Subclavian artery supply disruption sequence: hypothesis of a vascular etiology for Poland, Klippel-Feil,

and Möbius anomalies. Am J Med Genet 1986;23(4):903-18.
8. Radio FC, Digilio MC, Capolino R, Dentici ML, Unolt M, Alesi V, et al. Sprengel anomaly in deletion 22q11.2 (DiGeorge/Velo-Cardio-Facial) syndrome. Am J Med Genet A 2016;170(3):661-4.
9. Georgiev GP, Groudeva V. Klippel-Feil Syndrome with Sprengel Deformity. J Radiol Case Rep 2019;13(5):24-9.
10. Nakamura N, Inaba Y, Machida J, Saito T. Use of glenoid inclination angle for the assessment of unilateral congenital high scapula. J Pediatr Orthop B 2016;25(1):54-61.
11. Stelzer JW, Flores MA, Mohammad W, Esplin N, Mayl JJ, Wasyliw C. Klippel-Feil Syndrome with Sprengel Deformity and Extensive Upper Extremity Deformity: A Case Report and Literature Review. Case Rep Orthop 2018;2018:5796730.
12. Soldado F, Di-Felice-Ardente P, Barrera-Ochoa S, Diaz-Gallardo P, Bergua-Domingo JM, Knörr J. Passive range of glenohumeral motion in children with a Sprengel's deformity. JSES Int.2020;4(3):495-8.
13. Al-Salman HT, Al-Hussien AA, Al-Ahmed IH. Cervical myelopathy secondary to omovertebral bone in the pediatric patient with Sprengel deformity. Neurosciences (Riyadh) 2021;26(1):89-92.
14. Füllbier L, Tanner P, Henkes H, Hopf NJ. Omovertebral bone associated with Sprengel deformity and Klippel-Feil syndrome leading to cervical myelopathy. J Neurosurg Spine 2010;13(2):224-8.
15. Kafadar C, Saglam M, Sonmez G, Mutlu H. Omovertebral bone associated with Sprengel deformity presented with chronic cervical pain. Spine J 2016;16(2):e43.
16. Parish JM, Holland CM, Healy AT. Sprengel Deformity with Omovertebral Bone Encroaching the Spinal Canal Causing Progressive Cervical Myelopathy: A Technical Case Report. World Neurosurg 2021;146:163-5.
17. Jiang Y, Guo Y, Zhu Z, Wang Y. Surgical management of Sprengel's deformity by a modification of Green's procedure : A single center experience. Orthopade 2020;49(3):255-9.
18. Alsiddiky AM, Rafiq Z, Bakarman KA, Alhuzaimi FS, Asif M. A Novel Modification of Woodward Procedure for Correction of Sprengel Deformity by Application of Anchoring Sutures. Indian J Orthop 2021;55(Suppl 1):189-98.
19. Nouri A, Patel K, Evans H, Saleh M, Kotter MRN, Heary RF, et al. Demographics, presentation and symptoms of patients with Klippel-Feil syndrome: analysis of a global patient-reported registry. Eur Spine J 2019;28(10):2257-65.
20. Yiyit N, Işıtmangil T, Öksüz S. Clinical analysis of 113 patients with Poland syndrome. Ann Thorac Surg 2015;99(3):999-1004.
21. Öner A, Köksal A, Çimen O, Kargin D, Albayrak A, Akman YE. Modified Woodward Technique for Sprengel Deformity and a Modification of the Cavendish Classification. J Pediatr Orthop 2020;40(8):401-7.
22. Naik P, Chauhan H. Functional improvement in patients with Sprengel's deformity following Modified Green's procedure and simplified clavicle osteotomy-a study of forty cases. Int Orthop 2020;44(12):2653-63.
23. Elzohairy MM, Salama AM. Sprengel's deformity of the shoulder joint treated by Woodward operation. Eur J Orthop Surg Traumatol 2019;29(1):37-45.
24. Patwardhan S, Sodhai V, Gugale S, Sancheti P, Shyam A. Surgical correction of Sprengel's deformity by modified Woodward procedure: outcomes in twenty eight patients. Int Orthop 2020;44(6):1143-51.
25. Agarwal A, Arkesh M, Jandial G. Sprengel's deformity correction by vertical scapular osteotomy in a paediatric age group: influence of rib cage abnormalities. Int Orthop 2018;42(9):2191-7.
26. Yamada K, Suenaga N, Iwasaki N, Oizumi N, Minami A, Funakoshi T. Correction in malrotation of the scapula and muscle transfer for the management of severe Sprengel deformity: static and dynamic evaluation using 3-dimensional computed tomography. J Pediatr Orthop 2013;33(2):205-11.
27. Soldado F, Barrera-Ochoa S, Domenech-Fernandez P, Bergua-Domingo JM, Diaz-Gallardo P, Knorr J. Endoscopic Woodward procedure for Sprengel deformity: case report. J Pediatr Orthop B 2017;26(3):266-9.
28. Baca MJ, King RW, Bancroft LW. Glenoid hypoplasia. Radiol Case Rep 2016;11(4):386-90.
29. Trout TE, Resnick D. Glenoid hypoplasia and its relationship to instability. Skeletal Radiol 1996;25(1):37-40.
30. Wagner SC, Formby P, Keblish DJ. Bilateral Glenoid Hypoplasia in a College Football Athlete: a Case Report and Review of the Literature. Curr Sports Med Rep 2015;14(6):431-4.
31. Borenstein ZC, Mink J, Oppenheim W, Rimoin DL, Lachman RS. Case report 655: Congenital glenoid dysplasia (congenital hypoplasia of the glenoid neck and fossa of the scapula, with accompanied deformity of humeral head, coracoid process, and acromion). Skeletal Radiol 1991;20(2):134-6.
32. Lynch CJ, Taylor JA, Buchberger DJ. Glenoid hypoplasia: a report of 2 patients. J Manipulative Physiol Ther 2008;31(5):381-8.
33. Manns RA, Davies AM. Glenoid hypoplasia: assessment by computed tomographic arthrography. Clin Radiol 1991;43(5):316-20.
34. Nace J, Abboud JA. Hypoplastic glenoid with hyperplastic labrum: a case report. J Shoulder Elbow Surg 2009;18(3):e17-20.
35. Suryawanshi A, Mittal A, Dongre S, Kashyap N. Unilateral glenoid hypoplasia: a case report and review of the literature. Case Rep Orthop. 2011;2011:412721.
36. Weishaupt D, Zanetti M, Exner GU. Familial occurrence of glenoid dysplasia: report of two cases in two consecutive generations. Arch Orthop Trauma Surg 2000;120(5-6):349-51.
37. Currarino G, Sheffield E, Twickler D. Congenital glenoid dysplasia. Pediatr Radiol 1998;28(1):30-7.
38. Munshi M, Davidson JM. Unilateral glenoid hypoplasia: unusual findings on MR arthrography. AJR Am J Roentgenol 2000;175(3):646-8.
39. Kozlowski K, Scougall J. Congenital bilateral glenoid hypoplasia: a report of four cases. Br J Radiol 1987;60(715):705-6.
40. Owen R. Bilateral glenoid hypoplasia; report of five cases. J Bone Joint Surg Br 1953;35-B(2):262-7.
41. Davies AG. Bilateral humerus varus; with report of a case. Br J Radiol 1956;29(341):295-6.
42. Gill TJ, Waters P. Valgus osteotomy of the humeral neck: a technique for the treatment of humerus varus. J Shoulder Elbow Surg 1997;6(3):306-10.
43. Molto JE. Humerus varus deformity in Roman period burials from Kellis 2, Dakhleh, Egypt. Am J Phys Anthropol 2000;113(1):103-9.
44. Bhagat S, Bansal M, Sharma H, Jandhyala R, Amin P, Pandit J. A rare case of progressive bilateral congenital abduction contracture with shoulder dislocations treated with proximal deltoid release. Arch Orthop Trauma Surg 2008;128(3):293-6.
45. Chari PR, Rao YV, Rao BK. Congenital abduction contracture with dislocation of the shoulder in children: report of two cases. Aust N Z J Surg 1979;49(1):105-6.
46. Jhunjhunwala HR. Abduction contracture of the deltoid muscle in children. Int Orthop 1995;19(5):289-90.
47. Lian LY, Zhang LJ, Zhao Q. Deltoid contracture mimicking shoulder dislocation in a 7-year-old boy. Orthopade 2010;39(9):874-8.
48. Hurst SA, Gregory TM, Reilly P. Os acromiale: a review of its incidence, pathophysiology, and clinical management. EFORT Open Rev 2019;4(8):525-32.
49. You T, Frostick S, Zhang WT, Yin Q. Os Acromiale: Reviews and Current Perspectives. Orthop Surg 2019;11(5):738-44.
50. Hasan SA, Shiu B, Jauregui JJ. Symptomatic, Unstable Os Acromiale.

J Am Acad Orthop Surg 2018;26(22):789-97.
51. Fischer CS, Floß M, Ittermann T, Bülow R, Völzke H, Ahrend MD, et al. Os acromiale: prevalence and associated patient-related factors-a population-based study of three thousand and fifty participants. Int Orthop 2022;46(7):1583-90.
52. Rovesta C, Marongiu MC, Corradini A, Torricelli P, Ligabue G. Os acromiale: frequency and a review of 726 shoulder MRI. Musculoskelet Surg 2017;101(3):201-5.
53. Militano V, Hughes M, Vinjamuri S, Seshadri N. Incidental detection of Os acromiale mimicking a fracture on 18F-Fluoride PET-CT. Nucl Med Rev Cent East Eur 2019;22(1):43-4.
54. Nourissat G, Calò M, Montalvan B, Parier J. Os Acromiale in Professional Tennis Players. Orthop J Sports Med 2018; 6(5):2325967118773723.
55. Arenas-Miquelez A, Hertel R. The unstable os acromiale: a cause of pain in the young athlete. JSES Int 2020;4(3):559-63.
56. Viner GC, He JK, Brabston EW, Momaya A, Ponce BA. Os acromiale: systematic review of surgical outcomes. J Shoulder Elbow Surg 2020;29(2):402-10.
57. Phanichwong P, Apivatgaroon A, Boonsaeng WS. Prevalence of Os Acromiale in Thai Patients With Shoulder Problems: A Magnetic Resonance Imaging Study. Orthop J Sports Med 2022; 10(2):23259671221078806.
58. Horton S, Smuda MP, Jauregui JJ, Nadarajah V, Gilotra MN, Henn RF, 3rd, et al. Management of symptomatic os acromiale: a survey of the American shoulder and elbow surgeons. Int Orthop 2019; 43(11):2569-78.
59. Spiegl UJ, Millett PJ, Josten C, Hepp P. Optimal management of symptomatic os acromiale: current perspectives. Orthop Res Rev 2018;10:1-7.
60. Sun Q, Cai M, Wu X. Os acromiale may be a contraindication of the clavicle hook plate: case reports and literature review. BMC Musculoskelet Disord 2021;22(1):969.
61. Atinga M, Gregor R, Selvaraj KM, Hong TF. Os acromiale open reduction and internal fixation: a review of iliac crest autogenous bone grafting and local bone grafting. J Shoulder Elbow Surg 2018;27(6):1030-6.
62. Beliën H, Biesmans H, Steenwerckx A, Bijnens E, Dierickx C. Prebending of osteosynthesis plate using 3D printed models to treat symptomatic os acromiale and acromial fracture. J Exp Orthop 2017;4(1):34.
63. Purnell JA, Bourget-Murray J, Kwapisz A, Bois AJ, LeBlanc J. Clinical results and complications following surgical management of symptomatic os acromiale: a systematic review. J Orthop Surg Res 2019;14(1):26.
64. Stetson WB, McIntyre JA, Mazza GR. Arthroscopic Excision of a Symptomatic Meso-acromiale. Arthrosc Tech 2017;6(1):e189-e94.
65. Cooper CS, Field ED, Field LD. Arthroscopic "Wallow" Procedure for Resection of Symptomatic Os Acromiale Pseudoarthrosis. Arthrosc Tech 2021;10(5):e1223-e6.
66. Lebus GF, Fritz EM, Hussain ZB, Pogorzelski J, Millett PJ. Operative Treatment of Symptomatic Meso-Type Os Acromiale. Arthrosc Tech 2017;6(4):e1093-e9.
67. Gomez-Brouchet A, Sales de Gauzy J, Accadbled F, Abid A, Delisle MB, Cahuzac JP. Congenital pseudarthrosis of the clavicle: a histopathological study in five patients. J Pediatr Orthop B 2004; 13(6):399-401.
68. Studer K, Baker MP, Krieg AH. Operative treatment of congenital pseudarthrosis of the clavicle: a single-centre experience. J Pediatr Orthop B 2017;26(3):245-9.
69. Currarino G, Herring JA. Congenital pseudarthrosis of the clavicle. Pediatr Radiol 2009;39(12):1343-9.
70. Viciano J, Urbani V, D'Anastasio R. Congenital Anatomical Variant of the Clavicle. Anat Rec (Hoboken) 2017;300(8):1401-8.
71. Quinlan WR, Brady PG, Regan BF. Congenital pseudarthrosis of the clavicle. Acta Orthop Scand 1980;51(3):489-92.
72. Behringer BR, Wilson FC. Congenital pseudarthrosis of the clavicle. Am J Dis Child 1972;123(5):511-7.
73. de Figueiredo MJ, Dos Reis Braga S, Akkari M, Prado JC, Santili C. CONGENITAL PSEUDARTHROSIS OF THE CLAVICLE. Rev Bras Ortop 2012;47(1):21-6.
74. Depaoli A, Zarantonello P, Gallone G, Di Gennaro GL, Ferrari D, Marchesini Reggiani L, et al. Congenital Pseudarthrosis of the Clavicle in Children: A Systematic Review. Children (Basel) 2022;9(2).
75. Chalfant JS, Sanchez TR. Congenital Clavicular Pseudoarthosis-How to Differentiate It From the More Common Clavicular Fractures. Pediatr Emerg Care 2019;35(2):e37-e9.
76. Cataldo F. A 7-month-old child with a clavicular swelling since birth. Diagnosis: congenital pseudarthrosis of the clavicle. Eur J Pediatr 1999;158(12):1001-2.
77. Watson HI, Hopper GP, Kovacs P. Congenital pseudarthrosis of the clavicle causing thoracic outlet syndrome. BMJ Case Rep 2013;2013.
78. van der Water L, Macken AA, Eygendaal D, van Bergen CJA. Pediatric Clavicle Fractures and Congenital Pseudarthrosis Unraveled. Children (Basel) 2022;9(1).
79. Brévaut-Malaty V, Guillaume JM. Neonatal diagnosis of congenital pseudarthrosis of the clavicle. Pediatr Radiol 2009;39(12):1376.
80. Abdellaoui H, Atarraf K, Chater L, Afifi MA. Congenital pseudarthrosis of the clavicle treated by Masquelet technique. BMJ Case Rep 2017;2017.
81. Jeon IH, Wilson I, Wallace WA. Reconstruction of congenital pseudarthrosis of the clavicle in an adult. J Orthop Sci 2006; 11(2):229-31.
82. Li J, Tang SP, Mei HB, Shao JF, Shi BJ, Wang HQ, et al. Comparison of two methods in the treatment of congenital pseudarthrosis of clavicle: multicenter experience. J Orthop Surg Res 2021;16(1):301.
83. Martínez-Aznar C, Parada-Avendaño I, Gómez-Palacio VE, Abando-Ruiz S, Gil-Albarova J. Surgical treatment of congenital pseudoarthrosis of the clavicle: Our 22-year, single-center experience. Jt Dis Relat Surg 2021;32(1):224-9.
84. Cano-Pérez E, Gómez-Alegría C, Herrera FP, Gómez-Camargo D, Malambo-García D. Demographic, clinical, and radiological characteristics of cleidocranial dysplasia: A systematic review of cases reported in south America. Ann Med Surg (Lond) 2022;77:103611.
85. Farrow E, Nicot R, Wiss A, Laborde A, Ferri J. Cleidocranial Dysplasia: A Review of Clinical, Radiological, Genetic Implications and a Guidelines Proposal. J Craniofac Surg 2018;29(2):382-9.
86. Kutilek S, Machytka R, Munzar P. Cleidocranial dysplasia. Sudan J Paediatr 2019;19(2):165-8.
87. Machol K, Mendoza-Londono R, Lee B. Cleidocranial Dysplasia Spectrum Disorder 1993.
88. Dinçsoy Bir F, Dinçkan N, Güven Y, Baş F, Altunoğlu U, Kuvvetli SS, et al. Cleidocranial dysplasia: Clinical, endocrinologic and molecular findings in 15 patients from 11 families. Eur J Med Genet 2017; 60(3):163-8.
89. Jirapinyo C, Deraje V, Huang G, Gue S, Anderson PJ, Moore MH. Cleidocranial Dysplasia: Management of the Multiple Craniofacial and Skeletal Anomalies. J Craniofac Surg 2020;31(4):908-11.