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Case Report

Congenital absence of the bilateral long heads of the biceps brachii tendons in a patient with Silver-Russell syndrome ☆

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ABSTRACT

Agenesis of the long head of biceps tendon (LHBT) is a congenital anomaly not commonly reported in the literature, and bilateral absence of the LHBT is even more rare. Most cases of LHBT agenesis are found incidentally at arthroscopy or are diagnosed by magnetic resonance imaging after a history of insidious shoulder pain or anterior shoulder instability. We present the magnetic resonance imaging findings of bilateral congenital absence of the LHBT in a 37-year-old male with Silver-Russell syndrome who presented with progressive, bilateral anterior shoulder pain after failing conservative treatment strategies. This case report describes bilateral agenesis of the LHBT in association with a congenital growth disorder that has not been described previously in the literature. We provide a description of key MR imaging findings to assist in making the diagnosis, along with a discussion of potential differential diagnoses, and a review of the current literature on this topic.

Introduction

Bilateral agenesis of the long head of the biceps tendon (LHBT) is exceptionally rare with fewer than 10 cases described in the literature to date [1]. While other accompanying congenital anomalies have been reported in these patients, to our knowledge there has not been a previously reported case of bilateral or unilateral LHBT absence published in the context of a patient with Silver-Russell syndrome (SRS). The differential diagnosis of absence of the LHBT should include biceps tendon rupture, prior surgery including tenotomy and tenodesis, in addition to congenital absence. A thorough review of the patient's history and physical exam findings are key to arriving at the correct diagnosis which will help guide appropriate treatment.

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Case report

This 37-year-old male with past medical history of SRS presented with progressive, constant, left greater than right anterior shoulder pain after failing treatment with NSAIDs, intra-articular steroid injections, and physical therapy. His pain was worse with movement, lifting, and certain activities such as driving and typing. In addition to pain, the patient noted mild bilateral shoulder weakness and decreased range of motion. The patient denied any prior trauma to the shoulders, recent injuries, or prior shoulder surgery. Physical examination revealed a normal appearance of the arms with no findings to suggest a torn LHBT. The patient's medical history also included bicuspid aortic valve, hypogonadism with azoospermia, cryptorchidism with a history of right orchiectomy, atrophic left testicle, kyphoscoliosis, and congenital failure of segmentation of the T4-T6 vertebral bodies.

Bilateral shoulder MR arthrography examinations were performed on the same day which demonstrated bilateral absence of the LHBT and shallow intertubercular grooves bilaterally (Figs. 1A-D). The short head of the biceps tendon was normally developed, arising from the coracoid processes on both sides.

Discussion

SRS is a congenital imprinting syndrome most commonly (~50%) arising due to silencing of chromosome 11p15.5 region which encodes insulin-like growth factor-2, amongst other proteins [2]. About 10% of cases are due to maternal uniparental disomy of chromosome 7, and the remaining ~40% of cases are of unknown etiology. Roughly equal numbers of males and females are affected by SRS; however, there have been 9 cases of discordant monozygotic twins with a male: female ratio of 2:7, hinting at the importance of epigenetic factors in SRS [2]. The most common clinical features of SRS include short stature, triangular face, macrocephaly, intrauterine growth restriction, and body asymmetry [2]. As previously noted, this patient's congenital defects included bicuspid aortic valve, cryptorchidism, and scoliosis, which have all been documented in SRS patients. Work by Ishida et al. revealed congenital heart defects in 9 of 64 SRS patients, scoliosis in 14 of 64 SRS patients, and genital anomalies in 23 of 64 SRS patients [2]. Absence of the LHBT in these patients has not been previously documented in the literature.

A meta-analysis by Kumar et al. found that 85.7% of patients with LHBT absence had shoulder pain and 37.1% had anterior shoulder instability [1]. Due to this presentation, it is common for these patients to be evaluated for labral injury. This patient was noted to have normal glenoid labra and glenohumeral joint capsules bilaterally at MR arthrography. Further inspection revealed LHBT absence with shallow intertubercular grooves. It is important to look distally when the LHBT is absent, as the most common reason for absence of the LHBT proximally is complete rupture with distal retraction [3]. Complete rupture of the LHBT can result in a "Popeye" deformity on physical exam—a mass-like lesion in the upper arm representing distal retraction of the biceps musculotendinous unit due to loss of tension/rupture at the origin [4]. However, in this patient, due to no history of trauma, bilaterality of findings, anatomic morphologic findings, and lack of a clinical Popeye sign, an alternate diagnosis of bilateral complete LHBT tears with retraction was considered very unlikely.

Further considerations when evaluating patients with LHBT absence should include prior surgical history. Common surgical treatments include tenotomy and tenodesis. Tenodesis is growing as the preferred strategy in most instances as the literature suggests slightly better post-operative supination strength, fewer Popeye deformities, and less subjective cramping/pain [5]. Tenotomy, however, is still widely performed due to its historical use, quick intraoperative times, and similar clinical outcomes.

Due to the low prevalence of LHBT absence, we are unaware of any publications evaluating the efficacy of different management strategies. Given the similar presenting symptoms, many of these patients are treated with a similar approach to those with LHBT tendinopathy; starting with conservative management with NSAIDs, intraarticular steroid injections, and physical therapy [6].

The radiographic presence of shallow intertubercular grooves has been noted in patients with absent LHBT and may serve as a helpful marker in making the diagnosis [1]. However, shallow intertubercular grooves may also be seen as a normal anatomic variant [3]. Shallow intertubercular grooves are also associated with anterior shoulder instability [7].

Many variations of the LHBT have been observed. The most common variation is the presence of a supernumerary head, which has an estimated prevalence of 15.4% in one study of 175 cadavers [8]. Supernumerary heads of the LHBT are also associated with variations of the musculocutaneous and median nerves, which may be associated with nerve compression and neurological symptoms [3,9]. Despite the prevalence of LHBT variations, its absence, and particularly bilateral absence, has not been commonly reported in the literature.

It has long been established that mesenchyme can differentiate into connective tissues including skeletal muscle and tendon [10,11]. However, it was not until recently that cases of LHBT absence were recorded in patients with other congenital defects, a discovery that shed light on the origins of the LHBT. Absence of the LHBT has been associated with congenital anomaly syndromes, including one case of VATER (vertebral, anal atresia, tracheoesophageal fistula, and renal anomalies) complex association [12]. Other congenital anomaly associations include radial ray, upper limb deformities, spina bifida occulta, inguinal hernia, and undescended testes, some of which were also noted in this patient, as previously described [1].

Recent histological evaluations have demonstrated that the LHBT arises from the mesenchymal interzone between weeks 6 and 8 of fetal development [13]. Mesenchymal interzones are dense and compact zones of cells that form articular chondrocytes and other joint tissue between the 7th and 12th week of development [14]. Much is still unknown

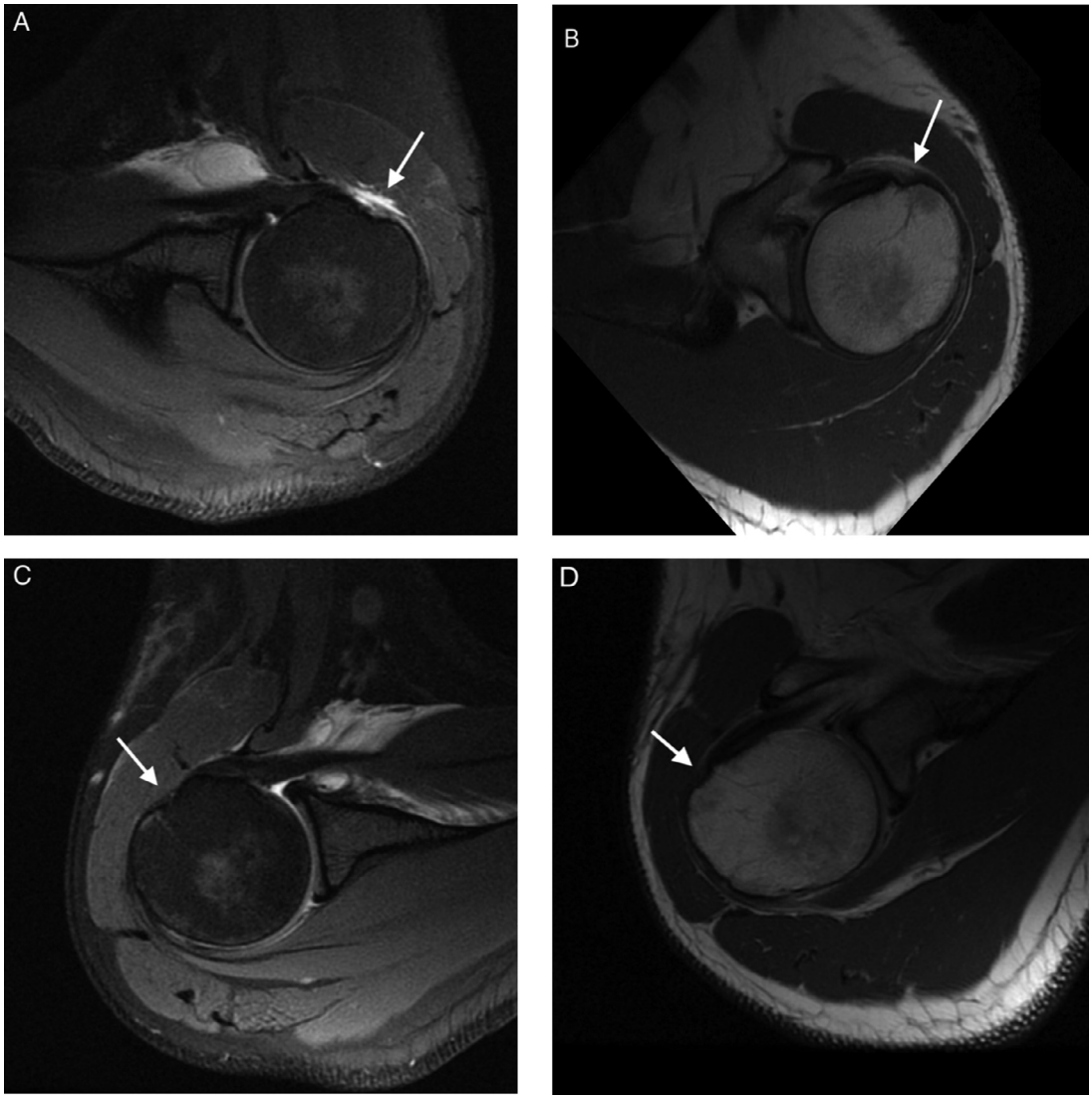

Fig. 1 – Congenital absence of the long head of the biceps tendon in a 37-year-old man with Silver-Russell syndrome presenting with progressive, bilateral anterior shoulder pain and instability after failing conservative treatment. (A) Left shoulder axial oblique PD FS MR image and (B) left shoulder axial T1-weighted MR image reveal absence of the long head of the biceps tendon with associated smooth and shallow intertubercular grooves (arrows). These findings were also present in the contralateral right shoulder as seen on (C) axial oblique PD FS MR image of the right shoulder and (D) axial T1-weighted MR imaging of the right shoulder.

about the factors that determine the location of the interzones and how joints develop into unique three-dimensional structures [14]. Future assessment of the LHBT anatomy of patients with SRS and genotyping studies with investigation into affected chromosomes may elucidate answers to these important questions.

Ethical approval

All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

Patient consent

Written informed consent was obtained from the subject described in this report.

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