Partial Pectoralis Major Muscle Aplasia: A Case Report

Pektoralis Major Kası Kısmi Aplazisi Olgu Sunumu

Murat Üzel[1], Ercan Çetinus[2], Erkal Bilgiç[1], Alp Ömeroğlu[3], Gönül Parmaksız[4], M. Ali Ergun[5]

[1]*Department of Orthopedic Surgery, Medical Faculty of University of Kahramanmaras Sutcu Imam. Kahramanmaras, Turkey*
[2]*Department of Orthopedic Surgery, Ministery of Health Haseki Research and Training Hospital. Istanbul, Turkey*
[3]*Department Of Radiology, Ethica Incirli Hastanesi, Incirli Pelinli Sk. No:23 Bakirkoy, Istanbul, Turkey*
[4]*Department of Pediatrics, Imamoglu State Hospital, Adana, Turkey*
[5]*Medical Genetic Department, Medical Faculty of Gazi University, Ankara, Turkey*

SUMMARY

The most common congenitally absent muscles are the pectoralis major and minor muscles. When associated with syndactyly and microdactyly, it is referred to as Poland's syndrome.

A patient reported here had partial congenital absence of the pectoralis muscles identified by clinical examination, ultrasonography, CT and MRI. This case was investigated with Doppler ultrasonography to find any vascular abnormality and results are presented here of his family pedigree and genetic analyses. The anomaly did not impair functions of the upper extremities. No treatment was required.

KEY WORDS: *Shoulder; Muscle aplasia; pectoralis majour muscle; pectoralis minor muscle; Ultrasound; Doppler studies; Computed tomography; Magnetic resonance Doppler studies; Computed tomography (CT); Magnetic resonance (MR)*

ÖZET

Doğuştan yokluğu en sık görülen kaslar pektoralis major ve minor kaslarıdır. Sindaktili ve mikrodaktili eşlik ettiğinde Poland sendromu olarak adlandırılır.

Bu çalışmada pektoral kasların kısmi yokluğu klinik muayene, ultrasonografi, BT ve MR görüntüleme ile ortaya konan bir hasta sunuldu. Bu olgu bir damar anomalisi olup olmadığını araştırmak için dopler ultrasonografi ile araştırıldı. Soyağacı ve genetik analiz sonuçları da sunuldu. Anomali üst ekstremite fonksiyonlarını bozmamıştı. Tedaviye gerek yoktu.

ANAHTAR KELİMELER: *Omuz; Kas yokluğu, Pektoralis major kası; Pektoralis minor kası; Ultrason; Dopler çalışmaları; Bilgisayarlı Tomografi; Manyetik rezonans*

Corresponding Author:
Murat UZEL, M.D. Associate Professor
Department of Orthopedic Surgery, Medical Faculty of University of Kahramanmaras Sutcu Imam, Yorukselim mh. Hastane cd. No:32 - 46050 Kahramanmaras
Phone: (0344) 221 23 37
Fax: (0344) 221 23 71
e-mail: drmuratuzel@hotmail.com

Introduction

About the congenital anomalies of muscle development, deficiency of the pectoral muscles, especially the sternocostal portion, is more frequently seen than any other groups.[1] When associated with syndactyly and microdactyly, it is referred to as Poland's syndrome.[1,2]

The child reported here had partial congenital absence of the pectoralis muscles identified by clinic examination and imaging modalities. His family pedigree and genetic analysis results are also presented.

Case Report

A 9-year-old boy, S.Ç., was referred to us for orthopeadic consultation from our pediatric department. The parents of patient were consanguineous. The family pedigree showed the sixth degreee consanguinity between his parents (Figure 1). Family history was negative concerning hand and thoracic muscle abnormalities.

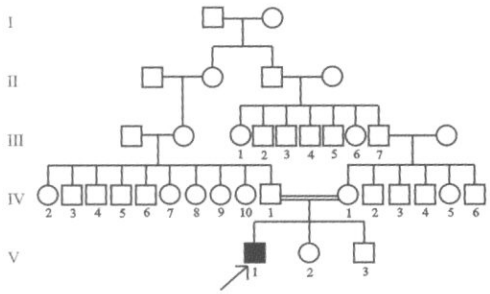

Figure 1.
Family pedigree showing the sixth degreee consanguinity between his parents.

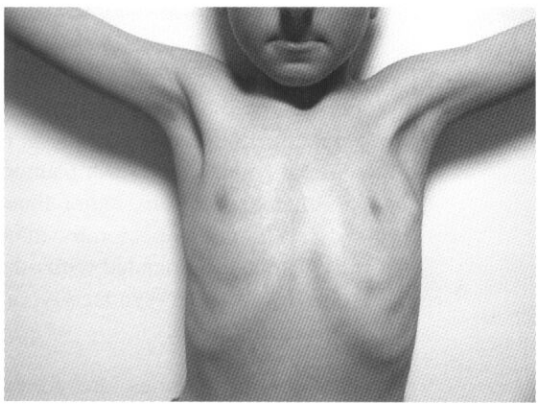

Figure 2.
The patient, a 9-year-old boy. Asymmetry between the right and left hemithorax is apparent clinically.

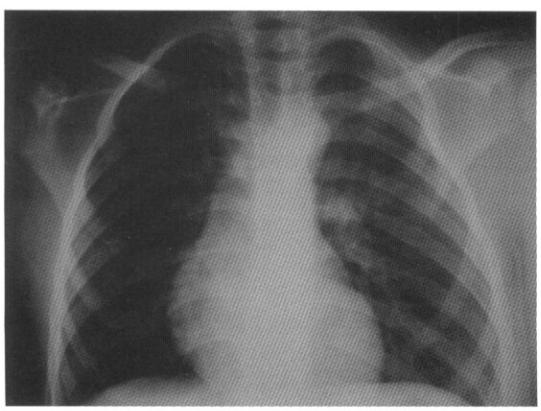

Figure 3.
AP radiograph of the thorax.

The maternal history was normal during pregnancy. Physical examination showed the absence of the sternal and abdominal portions of the left pectoralis major muscle. The clavicular portion of the muscle was intact. Range of motion was normal in the left shoulder and there was no marked difference on muscle strenght between both shoulders. Left anterior axillary fold was absent. Left nipple and areola were smaller when compared with the contralateral side. Left side of the chest was flat. On left side, scapula was elevated 15 mm higher and upper extremity was 12 mm shorter and forearm was 10 mm thinner (Figure 2).

In the radiographic examination, development of the anterior portion of the left third rib was rudimentary and the left scapula was between first and seventh ribs (Figure 3). Both hands were normal.

Ultrasonographic examination of the anterior thoracic wall was carried out with a two-dimensional, B-mode, real-time ultrasound system (Hitachi EUB-525 Hitachi Medical Corporation, Japan). A 7.5 megahertz, high - resolution, linear-array transducer was used. The anterior thoracic wall was scanned in the sections parallel and right angle to the long axis of the pectoralis major muscles. When compared with the parts of the normal pectoralis major muscle, the sternocostal and abdominal parts of the left pectoralis major muscle could not be seen. The clavicular part of it had a normal appearance. On left side, there was no pectoralis minor muscle beneath the pectoralis major and his left neurovascular bundle was seen just below the pectoralis major muscle (Figure 4). The real-time ultrasonographic examination showed clearly, the movement of the clavicular part of the pectoralis major muscle.

A computerized tomography (Toshiba CT Scanner Model TCT-600S Toshiba Corporation, Japan) of the thoracic region showed absence of the sternocostal and abdominal parts of the left pectoralis major muscle as well as

absence of the underlying pectoralis minor muscle. Left hemithorax was smaller than the right side (Figure 5).

Abdominal ultrasonography revealed that the genitourinary, gastrointestinal systems and the main intra-abdominal vascular structures were normal. The arterial doppler ultrasonography examination of both upper extremities was not significant.

Magnetic resonance (General Electric GMR max 0.5 Tesla, G Medical Systems Milwaukee, USA) also carried out on our patient, confirmed the CT findings. T1 Weigthed thorax MR scans were taken in sagittal, coronal and transverse planes. Soft tissue defect in the left side of the thoracic cage was clear. The patient had no pulmonary herniation. Ipsilateral latissimus dorsi and serratus anterior muscles and the main vascular structures were normal (Figure 6).

There was no abnormality in the pulmonary function tests. Hematologic parameters were within normal ranges. Cytogenetic analysis was reported as "46,XY".

Our patient had insignificant complaints about his deformity. No treatment was necessary.

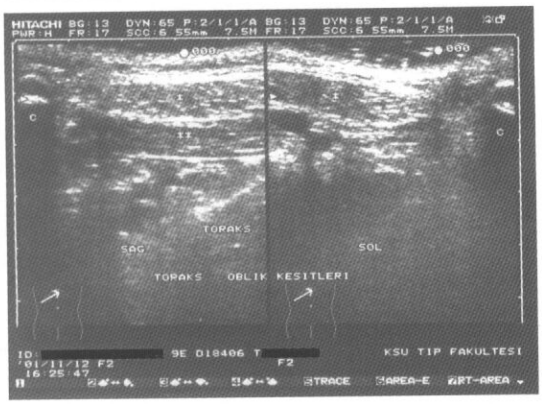

Figure 4.
Oblique section ultrasound images of the clavicular part of the pectoralis major

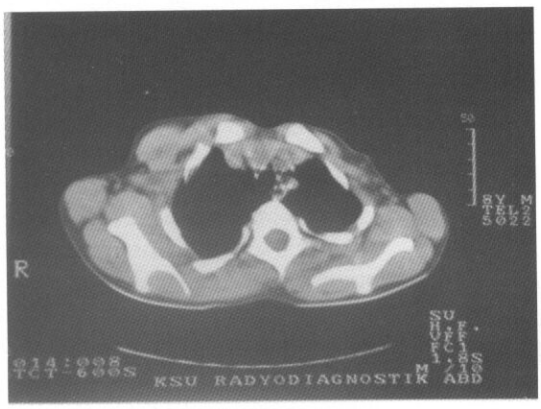

Figure 5.
CT scan showing the absent left pectoral muscles, hypoplastic left hemithorax.

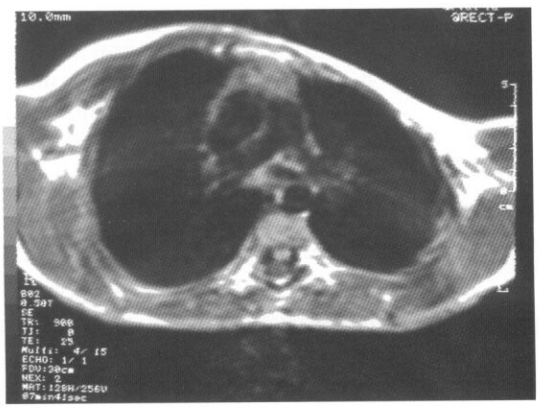

Figure 6.
Axial T1-weighted MR image shows absence of the left pectoralis major and minor muscles.

Discussion

Almost any muscle can occasionally be lacking. The abnormalities are usually unilateral and confined to a single muscle or a related group of muscles.[1] The incidence of the pectoralis major muscle defect ranges from 1 in 11 000 to 1 in 22 000 cases.[2] In Poland's syndrome, absence of the sternal head of pectoralis major is associated with webbed fingers and sometimes with absence of one phalanx in all fingers and absence of forearm flexors and general underdevelopment of the limb.[1,3] The incidence of Poland's syndrome ranges from 1 in 25 000 to 1 in 49 000 cases.[2,3] Genitourinary, musculoskeletal, gastrointestinal, cardiovascular and hematopoietic anomalies, excluding those in the thorax and ipsilateral upper extremity, sometimes coexist in this syndrome.[2,4,5] In the literature on the embryologic origin and development of the shoulder, there is no consensus on the relationship among the isolated pectoralis major muscle defect and the pectoralis major muscle aplasia associated with other anomalies without syndactyly and Poland's syndrome.

The pectoral muscles are formed by a mass that is a derivation of the V-VII cervical myotome.[5] Between the sixth and seventh weeks of gestation, the pectoralis premuscle mass separates into major and minor primordium. According to Lewis, this primordial mass fails to attach to the ribs and sternum, and thus the muscles and underlying ribs do not develop. This failure of attachment may be caused by many teratogenic environmental factors or vascular insufficiency.[1,6,7] The best developed of the hypotheses on vascular etiology is the subclavian artery supply

disruption sequence, which seeks to explain Poland syndrome and absence of the pectoralis major. The location and extent of tissue abnormality is determined by the extent, location, and timing of the interruption of normal blood supply, caused by a defect of arterial formation or an injury to existing arteries.[8]

The absence of pectoralis major muscle produces little disability and requires no treatment. Marks and associates recommended a latissimus dorsi muscle flap to cover the chest wall defect. Breast reconstruction by mammoplasty is a cosmetic benefit to many female patients.[2]

Ultrasonography, CT and MRI can accurately define the muscle abnormalities. Ultrasound is the first imaging modality available for the evaluation of the muscle pathology. Its spatial resolution and definition of muscle structure is usually superior to that provided by MRI. Real-time examination elucidates some types of muscle lesions, which are occult on static examinations. The degree of functional deficit can be fully assessed using real-time sonography.[9] CT and MRI can accurately define the muscle abnormalities and therefore the muscles available for transfer in Poland syndrome. Imaging may also detect associated anomalies and can be helpful in the preoperative planning and the evaluation of the postoperative situation.[10,11]

We suggest that ultrasonography can be used prior to MRI and CT to accurately evaluate a patient with muscle aplasia. Because of many anomalies associated with pectoralis muscle aplasia, the approach to the patient should be multidisciplinary.

References

1. Sharrard WJW. Diseases and Abnormalities of Muscles and Connective Tissue: Congenital Absence of Muscles. *Paediatric Orthopaedics and Fractures*. 3rd edition Oxford Blackwell, 1993: 839-40.
2. Wood VE, Marchinski L. Congenital Anomalies of the shoulder. In: Rockwood and Matsen, eds. *The shoulder*. 2nd ed. Philadelphia: Saunders 1998: 136-8.
3. Rasjad C, Sutiaksa IG. A case report of Poland's syndrome from Indonesia. *Aust N Z J Surg* 1991; 61: 320-2.
4. Perez Aznar JM, Urbano J, Garcia Laborda E, et al. Breast and pectoralis muscle hypoplasia: A mild degree of Poland's syndrome. *Acta Radiol* 1996; 37: 759-62.
5. Wilson MR, Louis DS, Stevenson TR. Poland's syndrome: variable expression and associated anomalies. *J Hand Surg (Am)* 1988; 13: 880-2.
6. Denti M, Facchini R, Peretti G. Partial aplasia of the pectoralis major muscle with an anomalous distal insertion: two case reports. *J Pediatr Orthop* 1985; 5: 489-91.
7. Yamasaki M. Anatomical study on 2 cases of the congenital partial defect of pectoralis major and minor muscles. *Anat Anz* 1989; 168: 423-32.
8. Dietz FR, Morcuende, JA. Embryology and Development of the Musculoskeletal System. In: Morrissy, RT, Weinstein, SL, eds. *Lovell and Winter's Pediatric Orthopaedics*, 6th edition Philadelphia: Lippincott Williams and Wilkins 2006: 1-33.
9. Van Holsbeeck M, Introcaso, JH. Sonography of the muscle. Musculoskeletal Ultrasound, Mosby-Year Book, St. Louis, 1991; 13.
10. Wright AR, Milner RH, Bainbridge LC, et al. MR and CT in the assessment of Poland syndrome. *J Comput Assist Tomogr* 1992; 16: 442-7.
11. Demos TC, Johnson C, Love L, et al. Computed tomography of partial unilateral agenesis of the pectoralis muscles. *J Comput Assist Tomogr* 1985; 9: 558-9.