



**A Rare Association; Pulmonary Artery Sling and
Mowat-Wilson Syndrome**

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**Nadir Bir Birliktelik; Pulmoner Arter Sling ve
Mowat Wilson Sendromu**

ABSTRACT

Pulmonary artery sling anomaly, where the left pulmonary artery originates from the right pulmonary artery is a rare type of vascular ring that can cause respiratory difficulties, cough, wheezing, stridor, feeding difficulties and repetitive vomiting due to tracheal and esophageal compression. We present a 14-year-old patient with dysmorphic facial appearance and mild mental retardation who underwent an echocardiography for murmur and was diagnosed with pulmonary artery sling anomaly.

Key words: Mowat Wilson syndrome, pulmonary artery sling, vascular ring.

ÖZET

Sol pulmoner arterin sağ pulmoner arterden çıktığı pulmoner sling anomalisi nadir bir vasküler halka tipi olup trakeaya ve özafagusa baskı yaparak solunum sıkıntısı, öksürük, hırıltı, stridor, beslenme problemleri ve tekrarlayan kusmalara yol açabilir. Bu yazıda dismorfik yüz yapısı, hafif mental retardasyon nedeniyle izlenen ve üfürüm nedeniyle yapılan ekokardiyografide pulmoner arter sling anomalisi saptanan 14 yaşındaki bir olgu sunulmuştur.

Anahtar Kelimeler: Mowat Wilson Sendromu, pulmoner arter sling, vasküler halka.

INTRODUCTION

Pulmonary artery sling anomaly, where the left pulmonary artery originates from the right pulmonary artery is a rare type of vascular ring that can cause respiratory difficulties, cough, wheezing, stridor, feeding difficulties and repetitive vomiting due to tracheal and esophageal compression. This pathology was first described by Glaevecke and Doeble during an autopsy and the term pulmonary sling was first used in 1958 by Contro et al. (1). This paper presents the case of a 14-year old patient with dysmorphic facial appearance and mild mental retardation who underwent an echocardiography for murmur and was diagnosed with pulmonary artery sling anomaly.

CASE REPORT

A 14-year old girl with dysmorphic facial appearance, mild mental retardation and murmur was referred to the pediatric cardiology department for evaluation of additional cardiac pathologies. The patient's history included a term 2200 g normal vaginal birth, no postnatal problems, wheezing attacks in infancy, occasional vomiting and constipation which improved by 6 years of age. Due to her dysmorphic facial appearance the patient was followed by another center, detailed genetic, neurologic and cardiologic evaluations were made but the echocardiography which was performed by an outside center did not identify any pathology. Karyotype analysis was normal and there was no deletion on 23 chromosome subtelomeric regional FISH analysis. Cranial magnetic resonance imaging (MRI) and abdominal ultrasound were within normal limits. Additionally, one year before applying to our clinic, the patient had been hospitalized for acute rheumatic fever. On physical examination the patient had low-set ears, microcephaly, narrow chin, deep eye sockets, strabismus, hypertelorism and wide-bridged nose (Figure 1). S1 and S2 were normal. Heart rate was 95/min and rhythmic. Femoral pulses were normally palpated. A more definite 2/6 systolic murmur was heard at the left upper parasternal region. Respiratory sounds were normal. There was no tachypnea or dyspnea. Second echocardiographic examination in our center determined a mild mitral and moderated aortic regurgitation with a wide main pulmonary artery. The bifurcation of pulmonary artery could not be visualized. Color-flow Doppler showed a vascular structure extending to the left from the posterior of the right pulmonary artery and contrast-enhanced 64-multislice computed

tomography (CT) confirmed this vascular structure as left pulmonary artery. CT showed the left pulmonary artery originated from the right pulmonary artery and minimally compressed the trachea (Figure 2). It was thought that the pulmonary artery sling anomaly combined with the dysmorphic symptoms may indicate Mowat Wilson syndrome. Mowat Wilson syndrome is often accompanied by Hirschsprung syndrome but a barium enema was normal. Cranial MRI was repeated and there was no agenesis of the corpus callosum. The pressure on the trachea was slight and there were no definite symptoms in the patient's respiratory system. So surgery was not planned and the patient is being followed up as an outpatient.

DISCUSSION

The frequency of pulmonary artery sling (PAS) anomaly, within the group of vascular ring anomalies, is unknown though some studies have reported an incidence of 5% (2). In this anomaly the left pulmonary artery typically arises anomalously from the posterior aspect of the right pulmonary artery, passes over the right mainstem bronchus, and courses leftward between the trachea and esophagus to the left pulmonary hilum. Respiratory system symptoms are the most common accompanying symptoms and are revealed in 90% of patients within the first year of life. In 25% of patients dysphagia may accompany respiratory symptoms (3); 30% of PAS patients have major cardiac pathologies such as patent ductus arteriosus, secundum atrial septal defects, tetralogy of Fallot and ventricular septal defects (4). Our patient had a repeated history of wheezing episodes during infancy. There was no history of dysphagia or recurrent vomiting. There was no additional congenital cardiac pathology. Aortic and mitral valve regurgitation were attributed to acute rheumatic fever of one year ago.

Until recently there has been insufficient data on the genetic

origin of PAS anomaly. Recently PAS anomaly has been reported to accompany Mowat Wilson syndrome and Klippel-Feil syndrome (5-7). Mowat Wilson syndrome is a genetic disease resulting from heterozygote mutation and deletions in the ZEB2 gene and was described by Mowat et al in 1998. This syndrome can be accompanied by moderate to severe mental retardation, typical facial appearance, epilepsy, microcephaly, short height, Hirschsprung disease, agenesis of the corpus callosum and varying degrees of cardiac anomalies. PAS anomaly is reported in 2.9% of Mowat Wilson syndrome patients (8). While our patient could be evaluated as a Mowat Wilson syndrome with typical facial appearance, mild mental retardation, short height, microcephaly and PAS anomaly, deletion and mutation on the ZEB2 gene could not be studied due to socioeconomic reasons. Hirschsprung disease seen in 53% of Mowat Wilson syndrome patients and agenesis of corpus callosum seen in 47% were not present in our patient.

Chest x-ray, barium esophagus x-ray, CT, MRI, angiography, bronchoscopy and echocardiography are methods used to identify PAS anomaly. Chest x-ray can identify indentation of the trachea, and excessive aeration of the right lung due to right bronchus compression. Barium esophagus x-ray can identify anterior indentation on lateral x-ray which is pathognomonic for PAS (2). Echocardiography is useful to diagnose additional cardiac anomalies. Bronchoscopy can be used for additional pathologies such as tracheomalacia and tracheal stenosis. Contrast-enhanced 64-multislice computed tomography is the most useful method for the diagnosis and treatment planning. It not only shows the left pulmonary artery's origin and pathway but also shows additional tracheal pathologies (9). Our patient's diagnosis was made after the second echocardiography that showed the left pulmonary artery originated from the right pulmonary artery and CT confirmed this diagnosis. Other than slight compression on the trachea no additional airway pathology was identified on CT.



Figure 1: Dysmorphic appearance of patient compatible with Mowat Wilson Syndrome.

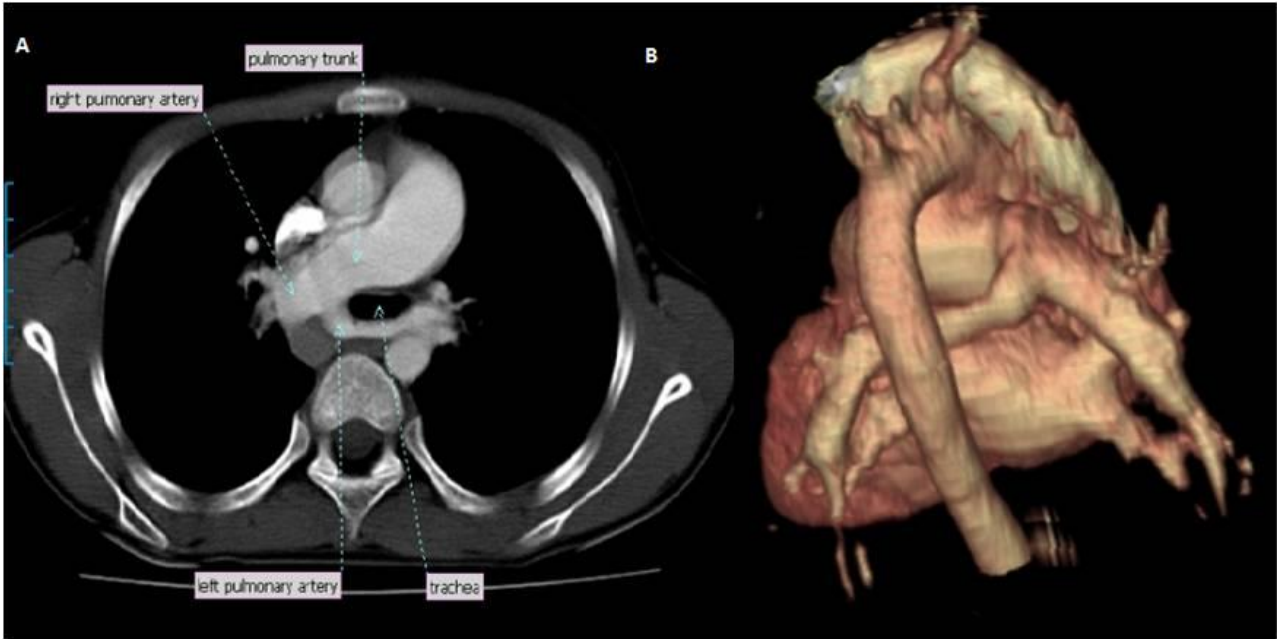


Figure 2: Contrast-enhanced 64-multislice computed tomography showing pulmonary sling.

Treatment of PAS anomaly is determined by the severity of the accompanying symptoms. In the newborn and infant period serious respiratory difficulties, frequent repeated lower respiratory infections, apnea, dysphagia, feeding difficulties or aspiration pneumonia are early surgical indications, with high mortality for patients without surgery (10). The prognosis is very good for asymptomatic cases with no surgical indications (11). For children operated for PAS 10% have airway obstruction or respiratory problems due to residual tracheal compression or tracheomalacia which may continue for months after the operation (12). While our patient had repeated complaints of respiratory problems in infancy, at the time of diagnosis she was asymptomatic and initial surgical intervention was not considered.

In conclusion children with repeated respiratory and gastrointestinal complaints must be investigated for PAS anomaly. Detailed echocardiography and if necessary tomography should be carried out, especially for children with accompanying dysmorphic facial appearance or with suspected underlying genetic syndromes.

KAYNAKLAR

- Contro S, Miller RA, White M, Potts WJ. Bronchial obstruction due to pulmonary artery abnormalities: I: Vascular sling. *Circulation* 1985; 17: 418-423.
- Dodge-Khatami A, Tulevski II, Hitchcock JF, et al. Vascular rings and pulmonary arterial sling: from respiratory collapse to surgical cure, with emphasis on judicious imaging in the hi-tech era. *Cardiol Young* 2002; 12: 96-104.
- Chun K, Colombani PM, Dudgeon DL, Haller JA Jr. Diagnosis and management of congenital vascular rings: a 22-year experience. *Ann Thorac Surg* 1992; 53: 597-603.
- Gikonyo BM, Jue KL, Edwards JE. Pulmonary vascular sling: report of seven cases and review of the literature. *Pediatr Cardiol* 1989; 10: 81-89.
- Adam MP, Schelley S, Gallagher R, Brady AN, Barr K, Blumberg B et al. Clinical features and management issues in Mowat-Wilson syndrome. *Am J Med Genet A* 2006; 140: 2730-2741.
- Streng S, Heinritz W, Zweier C, Rauch A, Rolle U, Merckenschlager A et al. Pulmonary artery sling and congenital tracheal stenosis in another patient with Mowat-Wilson syndrome. *Am J Med Genet A* 2007; 143A: 1528-30.
- Kawano Y, Tamura A, Abe Y, Kadota J. Klippel-Feil syndrome accompanied by pulmonary artery sling. *Intern Med* 2008; 47: 327.
- Garavelli L, Mainardi PC. Mowat-Wilson syndrome. *Orphanet J Rare Dis* 2007; 24: 2:42.
- Russell HM, Backer CL. Pediatric thoracic problems: patent ductus arteriosus, vascular rings, congenital tracheal stenosis, and pectus deformities. *Surg Clin North Am* 2010; 90: 1091-113.
- Yilmaz M, Ozkan M, Dogan R, Demircin M, Ersoy U, Boke E et al. Vascular anomalies causing tracheoesophageal compression: a 20-year experience in diagnosis and management. *Heart Surg Forum* 2003; 6: 149-52.
- Wang J, DING G, Zhang M, Liu M, Niu H. Clinical and imaging features of pulmonary artery sling in infants without significant hemodynamic changes. *Chin Med J* 2011; 124: 3412-3414
- Backer CL, Ilbawi MN, Idriss FS, DeLeon SY. Vascular anomalies causing tracheoesophageal compression. Review of experience in children. *J Thorac Cardiovasc Surg* 1989; 97: 725-31.