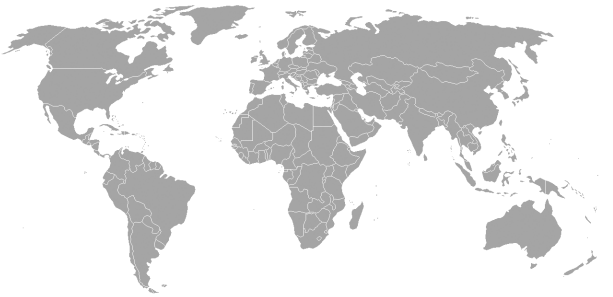
Telescopic Aortic Arch

A New Entity in Marfan Syndrome



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ABSTRACT

Telescopic aortic arch is a consequence of aortic dissection in Marfan's syndrome, which has not been reported previously. This paper presents the first case of telescopic aortic arch secondary to chronic aortic dissection, as an incidental finding of a forensic autopsy. Here we present a sudden death of a 20 years old male due to cardiac tamponade sourcing from the rupture of ascending aorta. At autopsy, there was a haemopericardium of 800 cc, which comprised blood and clot. The ascending thoracic aorta was dilated and a 1.2 cm ragged linear complete rupture was noticed on the medial side of the front wall of ascending aorta. Examination of aorta showed a second/inner aortic arch just as a tube lying inside the aortic arch. The inner aortic arch was arrised 2.5 cm above aortic valves and lasted at the beginning of the descending aorta. It was 10 cm in length with a lumen 2 cm in diameter and was associated with truncus brachiosephalicus, left common carotid, and left subclavian arteries.

Key words: *Marfan syndrome; telescopic aortic arch; chronic aortic dissection; aortic rupture; sudden death*

Teleskopik Arkus Aorta: Marfan Sendromunda yeni bir Antite

ÖZET

Teleskopik arkus aorta, Marfan sendromunda aortik diseksiyon sonucu oluşabilen ve daha önceden rapor edilmemiş bir antitedir. Burada bir adli otopside tespit edilen ve kronik aort diseksiyonuna ikincil ilk teleskopik arkus aorta olgusu bildirilmiştir. 20 yaşında bir erkekte assendan aorta rüptüründen kaynaklanan kalp tamponadı sonucu ani ölüm olgusu sunulmuştur. Otopside, serbest ve pıhtılı kandan oluşan 800 cc'lik bir hemoperikardium olduğu dikkati çekmiştir. Assendan torasik aortanın dilate olduğu ve ön duvar mediailinde 1,2 cm'lik tam kat duvar rüptürü olduğu tespit edilmiştir. Aort muayenesinde, arkus aorta içerisinde bir tüp gibi yerleşmiş ikinci bir arkus aorta olduğu dikkati çekmiştir. İçteki bu ikinci arkın aort kapaklarının 2,5 cm üzerinden başladığı ve desseden aorta başlangıcında sonlandığı; ayrıca 10 cm uzunluğunda ve 2 cm çapında olup trunkus brakiosefalikus, sol karotis kommonis ve sol subklavian arterle ilişkili olduğu saptanmıştır.

Anahtar kelimeler: *Marfan sendromu, Teleskopik arkus aorta, Kronik aort diseksiyonu, Aort rüptürü, Ani ölüm*

INTRODUCTION

Deaths due to rupture of thoracic aorta are not uncommon. The rupture of aorta is commonly caused by an aneurysm, which might be secondary to trauma, infections, valve, and arch anomalies, genetic disorders and atherosclerosis (1,2). An arch anomaly obstructing the aorta might lead dilatation or aneurysms in proximal part of ascending aorta and consequently aortic ruptures. Marfan's syndrome is a common autosomal dominant genetic disease with a prevalence of 1 in 5,000, in Europe (3). Various patterns of organ involvement are encountered, in affected individuals. Undiagnosed patients usually die from acute aortic dissection or rupture (3,4). Marfan's syndrome remains as an important cause of aortic dilatation or dissection because of connective tissue weakening. In some cases, no cardiovascular problems are encountered until an aortic aneurysm or aortic dissection occurs in ascending aorta, which is mostly fatal. A review of the literature revealed no aortic dissection resulted in telescopic shape of aortic arch, in Marfan's syndrome. In this paper, we present sudden death of a case of Marfan's syndrome characterized by a second/inner aortic arch nested inside the morphologically normal aortic arch.

CASE

The deceased was a 20 year-old male of weight 70 kg and height 190 cm. While having a very slow motor-bike ride, with his girlfriend, the deceased stops the bike and collapses on the ground. The girlfriend had no traumatic sign but the deceased was not able to speak and had shortness of breath and he died within 15 minutes, on the way to hospital. The deceased had been diagnosed to have a heart murmur during his childhood; however, he had not undergone any further diagnostic procedure or medical intervention. He had lived a healthy life, with unknown history of Marfan's syndrome. At corpse examination, as slight traumatic signs, there were superficial abrasions on the left shoulder, elbow. He also had pectus excavatum 7 cm in depth, long thin fingers, high-arched palate and irregularity of teeth. At autopsy, abdominal organs were placed minimally lower than normal level because of marked pectus excavatum. However, significant findings on internal examination were confined to the cardiovascular system. The heart was slightly enlarged weighing 390 g. The left ventricle showed concentric hypertrophy, but there was no evidence of chamber dilatation. The myocardium showed no evidence of recent or remote ischemic damage. Coronary artery anatomy and the cardiac valves were normal, apart from slight thickening of mitral and tricuspid valves, related to myxomatous degeneration. There was a haemopericardium of 800 cc, which comprised blood and clot. The ascending thoracic aorta was aneurysmatic/dilated and a 1.2 cm ragged linear rupture, with extension into pericardium, was noticed on the medial side of front wall of ascending aorta. Examination of aorta revealed an abnormality of aortic arch caused by a perfect/bloodless chronic aortic dissection restricted to aortic arch. A second minor/inner aortic arch was noticed just as a tube nested inside the aorta, forming a telescopically double barreled aortic arch (Figure 1). As aortic arch has a telescopically double barreled shape there were inner and outer lumens. The inner lumen was arising from 2.5 cm above aortic valves and lasted at the beginning of the descending aorta. It was approximately 10 cm in length with an entirely open lumen, having wide entrance and exit, 2 cm in diameter. The inner lumen was the true lumen while the outer was the false lumen of a dissecting aortic aneurysm. The diameter of ascending aorta was 5 cm, while it was 4.3 cm in the outer lumen of the aortic arch. Inner and outer lumens were associated with each other and both of them were associated with brachiosephalic artery, left common carotid artery, and left subclavian artery (Figure 2). There were no any other abnormalities in organs and vessels. The family members of deceased were stated to be free of cardiovascular anomalies. There were not any additional traumatic findings or any other pathologic changes that could be attributable to any other pathology. A complete toxicological analysis was also performed, and no trace of illicit drugs or alcohol was found in the blood and organ samples. Due to macroscopic anatomopathologic findings the case was diagnosed as Marfan's syndrome complicated by dissection of aortic arch forming a telescopic aortic arch.

DISCUSSION

Marfan's syndrome, with a number of mutations on the fibrillin gene, is a well-known cause of aortic dissection. Patients with Marfan's disease have a 50 percent risk of developing aortic dissection during their lifetime. Five percent of all aortic aneurysms and dissection cases caused by Marfan's disease (5). The atrioventricular valves are mostly involved and thickening of the atrioventricular valves is common. Aortic aneurysm and dissection remain the most life-threatening manifestations of Marfan's syndrome. The firstly affected part is almost always the aortic root and dissection can remain isolated or propagate along the length of the descending aorta, as previously classified. Acute aortic dissection is invariably characterized by radiating severe chest pain, while chronic aortic dissection usually present insidiously, mostly without chest pain. The mechanism of death usually includes rupture into the pericardial sac with subsequent pericardial tamponade (4,6,7).

Parallel with the literature regarding aortic dissection in Marfan's syndrome, the presented case, with unknown history of Marfan's syndrome, was stated to be completely healthy and asymptomatic till death. The external and internal examination findings obtained in autopsy were also truly compatible with previously reported cases of Marfan's syndrome. However, the anomalous formation of aortic arch caused by chronic dissection was unique for Marfan's syndrome. The anomalous formation presented in this report did not matched to previously described aortic arch anomalies, and it was not a branching abnormality as well. In the presented case, examination of aorta revealed an abnormality of aortic arch caused by a perfect chronic aortic dissection restricted to aortic arch. A second minor/inner aortic arch was noticed just as a tube nested inside the aorta, forming a telescopically double barreled aortic arch. As aortic arch has a telescopically double barreled shape there were inner and outer lumens. To our knowledge, this entity has not been described previously and the authors denominated it as "telescopic aortic arch" since the anomalous structure shaped by an inner aortic lumen nested inside the aortic arch of the deceased. The inner lumen was the true lumen while the outer was the false lumen of a dissecting aortic arch. Despite being structurally functional, the inner/minor aortic arch was an occlusive anomaly contributing dilatation and consequently rupture of ascending aorta. As a limitation for the investigation, since it was a forensic case, the cause of death as cardiac tamponade secondary to pathologic rupture of ascending aorta was satisfactory for the public prosecutor and no further histopathologic or molecular genetic investigations for medical purposes were performed.

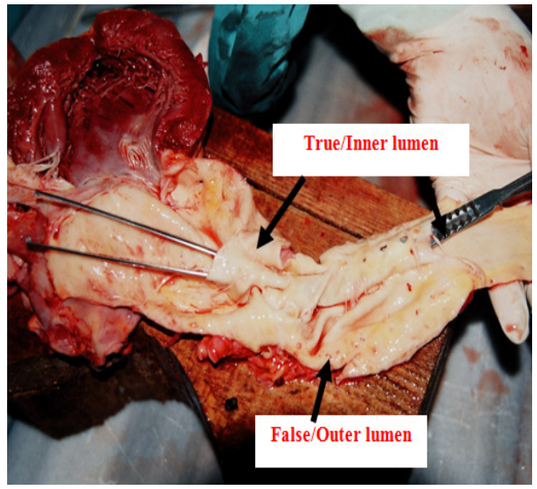

***Figure 1**. Telescopic aortic arch formation due to chronic dissection*

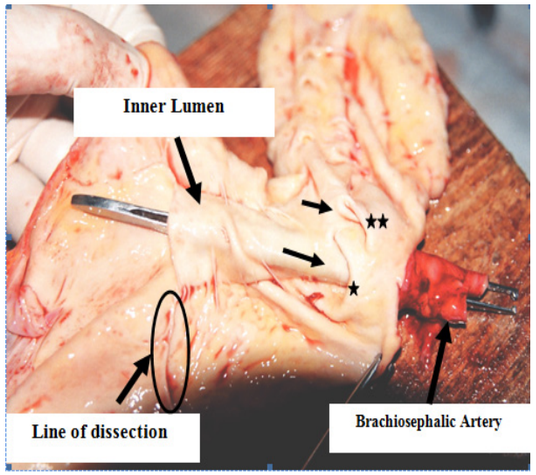

***Figure 2**. Both of inner and outer lumens are associated with the branches (*Brachiosephalicus Artery,**Left Common Carotid Artery*

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