Bicuspid aortic and pulmonary valves complicated by acute aortic dissection in a highly trained athlete – Case report and review of the literature

Martin Janík1*, František Novomeský1, Lubomír Straka1, Jozef Krajčovič1, František Štuller1, Petr Hejna2

Abstract: Acute aortic dissection is typically seen in the middle aged and elderly population, the majority of the cases are related to systemic hypertension and atherogenic process. In contrast, aortic dissection-related deaths in the young are rare and are mainly associated with genetically based disorders and congenital cardiac anomalies. One of the most recognized congenital risk factors that predispose to aortic dissection is the bicuspid aortic valve. This report describes an illustrative case of acute aortic dissection in a 29-year-old athlete secondary to previously unrecognized congenital bicuspid aortic valve. Moreover, further cardiac autopsy disclosed bicuspid pulmonary valve - an exceedingly rare congenital valvular anomaly. The authors characterize and discuss, with emphasis on medicolegal approach, the mechanisms and interactions between these pathologic entities that led to the athlete’s sudden unexpected death.

Key Words: aortic dissection, bicuspid aortic valve, bicuspid pulmonary valve, athlete.
Bicuspid aortic and pulmonary valves complicated by acute aortic dissection in a highly trained athlete

Janík M. et al

Pulmonary valves are very rare and are found in approximately 0.03% of the population and may be associated with trisomy 18, tetralogy of Fallot, absent conal septum, and coarctation of the aorta [12, 13].

In this paper, we present and discuss an illustrative autopsy case of a sudden unexpected death in a young athlete due to acute aortic dissection secondary to previously undiagnosed bicuspid aortic valve. Moreover, at cardiac autopsy we discovered bicuspid pulmonary valve - an exceedingly rare congenital valvular anomaly.

CASE REPORT

A 29-year-old male presented with severe tearing chest pain with immediate onset and collapsed suddenly at work. He was unresponsive to resuscitation efforts and was pronounced dead at the scene. Information obtained prior to autopsy revealed that the decedent was a highly trained triathlete. Additionally, he had complained of progressive weakness and mild febrile illness for the several days prior to death. His family history was noncontributory.

Postmortem examination of the well-nourished athletic male showed unremarkable external findings except for marks produced by the resuscitation procedure. His height was 188 cm, and his weight was 90 kg. Macroscopically, the lungs showed congestion and mild edema with multiple, subpleural petechial hemorrhages. Sectioning of the brain demonstrated diffuse cerebral swelling. Autopsy revealed no traumatic stigmata either recent or remote. No pathological conditions or other abnormalities were identified grossly in any visceral organs except as described below. A comprehensive blood and urine toxicologic assay was negative.

Cardiac autopsy

Upon opening an apparently enlarged pericardium, approximately 970 mL of clotted and nonclotted blood was collected within the pericardial space. In situ inspection of the heart revealed full-thickness rupture (measuring 3 x 2.5 cm) of the aortic aneurysm with adjacent adventitial hemorrhage. On transverse slices, the myocardium showed mildly hypertrophied left ventricular walls, 2.4 cm in thickness (heart weight 435 g; dimensions 14 x 12 x 6.4 cm) and some dilatation of the right ventricle with no infarctions either acute or remote. Further dissection of the heart showed, directly above the aortic opening, a wide, transversely oriented,

Figure 1. (A) Bicuspid aortic valve with equal-sized leaflets and two commissures (asterisks); an apparent intimal tear directly above the aortic opening; (B) both coronary ostia arising from one sinus (asterisks); (C) Bicuspid pulmonary valve with two commissures (asterisks); (D) aneurysmatic dilatation of the ascending aorta (blue arrows).
T-shaped intimal tear (Fig. 1 A, B) with dissecting, blood-filled channel extending peripherally to the aortic arch (DeBakey I). At the level of 1.5 cm distally from the above-described tear, dissection ruptured externally into the pericardial sac. The aneurysmatic ascending aorta measured 5.6 cm in diameter (Fig. 1 D). Further examination of the aorta showed a localized mild narrowing of the lumen of the aortic arch distal to the origin of the left subclavian artery as well as patchy orange fibrolipid atherosclerotic plaques. Dissection of the valvular heart apparatus revealed both congenitally bicuspid aortic and pulmonary valves (Fig. 1 A, C). Equally-sized and shaped aortic cusps were apparently enlarged, focally thickened, with two commissures and no raphe between the undeveloped left and right coronary cusps (Fig. 1 A, B).

The aortic valve circumference measured 7.9 cm. There was evidence of mild aortic valve insufficiency. Macroscopically, smooth, mildly edematous valvular endocardium showed focal areas of yellowish fibrolipid deposits. Both coronary ostia arose from one sinus, distribution and dimensions were normal (Fig. 1 B). The examination of the right ventricular outflow tract revealed the presence of the bicuspid pulmonary valve (Fig. 1 C).

The pulmonary leaflets were equally-sized, thin, with two commissures. The pulmonary valve circumference was 5.6 cm. No other congenital heart defects, valvular vitia, or major vessels abnormalities were present.

Microscopic examination of the aortic leaflet, both periaortic and aortic perivalvular fibroadipose tissue showed reactive inflammatory process to the dissection that had invaded into the adjacent aortic wall (Fig. 2 A-C). Focal microcystic medial degeneration, fibers disorganization, and aortic intramural hematomas were also identified (Fig. 2 D). Other cardiac structures or aortic segments were not microscopically compromised.

The cause of death, based on the above-described autopsy findings, was massive hemopericardium resulting from ruptured aortic dissection sustained as a complication of bicuspid aortic valve.

DISCUSSION

The estimated incidence of aortic dissection has been found to vary from 5 to 30 per million individuals per year. From a morphological viewpoint, aortic dissection is defined as the splitting of aortic wall between the outer and middle portion of the tunica media either with or

Figure 2. (A) perivalvular tissue with acute reactive inflammatory infiltrate and focal hemorrhage (H&E, x10); (B) periaortic adventitial tissue with acute inflammatory process that invades into the adjacent aortic medial wall (H&E, x10); (C) dense inflammatory reaction to the dissection (H&E, x10); (D) aortic intramural hematoma (H&E, x10).
without intimal tear. The latter type of aortic dissection is rarer and is called bloodless dissection [14, 15]. In both types of aortic dissection, the process rarely involves the whole circumference of the aorta.

In most cases, nevertheless, dissection of the aorta originates and propagates owing to an intimal tear. In such a scenario, blood from the aortic lumen penetrates through the intimal tear into the vessel wall and causes separation of layers of the tunica media. The resulting spreading of dissection is possible owing to the pulse-dependent pressure of bloodstream, either in a backward or forward pattern. Dissection may re-enter the aortic lumen, creating a “double-barrel” aorta or may, through adventitial tear, give rise to hemopericardium, hemothorax, or even hemoretroperitoneum.

Secondary dissection-associated complications commonly include peripheral ischemia due to obstruction/compression of aortic tributaries or severe aortic regurgitation (the second most common cause of death, after aortic rupture in persons with aortic dissections) [16].

Recently, Hostiuc et al. reported two cases of aortic dissection associated with dissection and rupture of the pulmonary artery [17].

Etiologically, several aortic wall-related diseases, including atherosclerosis, systemic hypertension, hereditary connective tissue diseases, syphilis, Erdheim-Gsell cystic degeneration of media, and infective aortitis are predispositions for the development of aortic dissection or rupture [18, 19]. Additionally, cocaine and amphetamine abuse, the pregnancy/postpartum period, and iatrogenically related aortic wall injuries such as cardiopulmonary resuscitation, catheter-based diagnostic and therapeutic interventions have been recognized as a precipitating factors of aortic wall dissection or rupture [20–26]. Lastly, fatal aortic tear may occur in the setting of congenital cardiac diseases with the most recognized being bicuspid aortic valve and aortic coarctation [27].

The bicuspid valve is usually composed of two unequally sized cusps, with the presence of a central raphe that results from fusion of the commissures. The morphological configuration of the bicuspid valve depends on which commissures have fused. The most common manifestation involves a fusion of the right and left cusps, followed by fusion of the right and noncoronary cusps. The least common pattern comprises fusion of the left and noncoronary cusps. In rare instances, the leaflets are symmetrical and rapheless (as demonstrated in our case).

Although bicuspid aortic valve is primarily a valvular disorder, it is important to realize that this anomaly also plays a role in development of aortic wall abnormalities, with aortic dilatation and risk of aortic dissection with rupture. From a histopathological viewpoint, fibrosis, elastin fragmentation, and cystic degeneration of media have been found upon examination of the aorta in persons who died of a dissecting aneurysm [28, 29]. However, Larson and Edwards found a prevalence of aortic medial degeneration in only 18% of 161 persons with aortic dissections among 21,105 autopsies [30]. More specifically, bicuspid valve studies have reported structural abnormalities of thoracic aortic tissue, including decreased fibrillin, elastin fragmentation, matrix disruption, and apoptosis [10].

Though the mechanism of bicuspid semilunar valves remains obscure, it is thought to originate from an endocardial cushion defect which may be due to abnormal cell migration, signaling pathways and, lastly, by genetic predisposition (NOTCH1 gene) [31]. More recent experimental studies have shown that metalloprotease ADAMTS5 deficiency blocks the excavation of endocardial cushions during aortic and pulmonary valve development [32].

Aortic dissection usually occurs at a younger age and in previously asymptomatic persons with BAV. The incidence of fatal aortic dissection in individuals with BAV is approximately 5%, while the risk of dissection is highest when the aortic diameter is more than 5 cm and with concomitant systemic hypertension [33].

Besides the BAV-associated aortic stenosis/insufficiency, it is acknowledged that the aortic bicuspid valve is a well-recognized substrate for bacterial endocarditis, predominantly in children and young adults. The valvular inflammatory process is highly destructive and may be associated with bacterial aortitis as the bacterial elements invade into the aortic wall. Approximately 10–30% of individuals with bicuspid aortic valve develop endocarditis. Microbiologically, the most frequent microorganisms are Staphylococcus spp. and Streptococcus viridans. Saint-Martin et al. reported a case, where a ruptured subaortic pseudoaneurysm caused sudden death [34]. The decedent in that case report had a history of bicuspid aortic valve with recent Staphylococcal endocarditis.

While the bicuspid pulmonary valve is commonly associated with other congenital heart diseases, isolated bicuspid pulmonary valve is extremely rare, with less than 10 cases reported in the literature. The true incidence of bicuspid pulmonary valve, however, remains unknown perhaps due to fact that the clinical course is usually benign [35]. Recently, Luk et al. reported autopsy case of a 70-year-old man with bilateral congenitally bicuspid aortic and pulmonary valves [36]. Both valves showed multifocal nodular thickening and fibrosis. There was markedly greater dilatation of the pulmonary artery, compared to the aorta, as well as an absence of symptoms. A functionally normal bicuspid aortic valve is clinically asymptomatic and does not usually represent a limitation for physical activity. The stress of regular and intense exercise on an abnormal aortic valve may, however, accelerate the development of complications and sudden
death is an appreciable risk, whether the individual is symptomatic or asymptomatic [37]. Therefore, identifying and accurate staging of the bicuspid aortic valve in athletes, through strict cardiological follow-up, is of vital importance in order to prevent the potential adverse consequences of training [38, 39]. Athletes with bicuspid valve who develop aortic valve insufficiency and aortic dilatation should completely avoid vigorous sporting activities, given the increased risk of aortic rupture [37]. Furthermore, because the aortic dilatation rate may significantly differ between individual patients, only specific follow-up measurements can determine individual size-dependent aortic risk [8].

In the postmortem assessment, cardiac imaging modalities such as computed tomography, CT angiography, and MRI have proven to be beneficial, particularly in cases of suspected aortic dissection, endocarditis, or myocardial infarction [40–42]. In sudden cardiac deaths of young, previously healthy individuals, detection of primary underlying pathology may have tremendous implications for cardiologists, genetic counselors and families such as aortic dissections associated with congenital and genetically based diseases. In such cases, conventional organ-by-organ dissection, augmented with various ancillary laboratory studies such as histopathology, immunochemistry, microbiologic cultures, and molecular diagnostic evaluation are warranted and highly recommended.

In summary, we have reported an illustrative autopsy case of acute aortic dissection complicated by massive hemopericardium in the setting of previously unrecognized bicuspid aortic valve. Moreover, pathologic findings included mild left ventricular hypertrophy and aortic coarctation, additional bicuspid aortic valve-related comorbidities and precipitating factors in producing aortic dissection and/or rupture. Apart from above mentioned left-sided cardiac abnormalities, autopsy showed an uncommon congenital bicuspid pulmonary valve, although this finding did not play a role in the mechanism of death.

References


