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Inherited Stanford A DeBakey I Aortic Dissection: Unveiling Genetic Predispositions and Surgical Management

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Abstract

BACKGROUND: Aortic dissection refers to the disruption of the medial layer of the aorta caused by intramural hemorrhage. This leads to the separation of the layers of the aortic wall, resulting in the establishment of a true lumen and a false lumen, which may or may not be connected. The prevalence of aortic dissection ranges from 0.2% to 0.8%. Additionally, a notable percentage, surpassing 20%, of individuals exhibit a familial inclination towards thoracic aorta dissection.

CASE PRESENTATION: A male patient in his middle age, 41 years old, with risk factors of uncontrolled hypertension, obesity, and a familial propensity to aortic dissection, presented to the emergency department exhibiting symptoms of chest pain. It characterized the sensation as having a tearing quality and indicated that it extended towards the abdomen region. The results of the test revealed several notable findings. A blood pressure reading of 235/133, a wider mediastinum and cardiomegaly on the chest X-ray, a slight increase in Hs-Troponin levels, and an electrocardiogram showed sinus rhythm with left ventricular hypertrophy. He underwent a computed tomography scan, which revealed the existence of an aortic dissection that extended from the ascending to the descending aorta without any aortic valve involvement. He was assessed with a Stanford A. DeBakey I aortic dissection, and underwent successful therapy for aortic arch replacement with the elephant trunk procedure, and plans to undertake an endovascular procedure for the descending aorta in the upcoming period.

CONCLUSION: Aortic dissection is a pathological disorder that may exhibit hereditary inheritance, and the choice of surgical technique is dependent on the specific underlying disease.

Introduction

Aortic dissection is a potentially lethal condition characterized by poor prediction, necessitating immediate emergency intervention. The issue at hand continues to provide a significant clinical challenge and is associated with a documented reduced 5-year survival rate, particularly in cases of acute nature. Acute type A aortic dissection (ATAAD) is widely recognized as the most fatal form of dissection, primarily affecting the ascending aorta. Aortic dissection is characterized by the presence of an intimal tear, which facilitates the passage of blood beyond the tear and into the medial layer of the aorta. This occurrence initiates the separation of the intima layer, resulting in the formation of a dissection flap. This, in turn, causes the actual lumen to separate from a newly formed pseudo lumen. Aortic dissection has been correlated with several underlying medical diseases that augment the risk, encompassing genetic disorders, inflammatory vasculitides, as well as other factors such as pregnancy, trauma, prior heart surgery, stimulant misuse, and infection [1], [2], [3], [4].

The yearly frequency of type A aortic dissection is approximately 3 cases/100,000 individuals. A documented 5-year survival rate ranges from 55% to 85% in cases of acute type A and type B aortic dissection. Additionally, the operational mortality rate has been reported to be as low as 12%. The first death rate for untreated aortic dissection is estimated to be around 1% every hour, whereas by the 3rd day, it increases significantly to approximately 50%. Following discharge from the hospital, a significant proportion of fatalities, ranging from 31% to 66%, can be attributed to complications related to aortic dissection in instances classified as type B. It is worth noting that type B aortic dissection is often linked with a comparatively lower risk when compared to type A aortic dissection. Approximately two-thirds of instances involving aortic dissections are observed in males, whereas a significant proportion of patients, reaching 20%, report a familial inclination towards thoracic aorta dissection [3], [4].

Pain emerged as the prevailing clinical manifestation of acute aortic dissection, exhibiting substantial heterogeneity in terms of its

characteristics and anatomical presentation. Aortic dissection frequently presents with uncontrolled hypertension, which is widely regarded as a prominent modifiable risk factor. Type A necessitates surgical intervention, whereas type B, despite its intricate nature, can be efficiently addressed with pharmacological therapy [3], [4].

Case Illustration

A 41-year-old male of middle age presented at the emergency department with sudden chest pain. This individual exhibits several risk factors, including uncontrolled hypertension for over 10 years, severe obesity classified as class III with a body mass index of 50.8 kg/m^2, and a familial history of aortic dissection. Specifically, his father underwent Bentall surgery 10 years ago due to a Stanford A. aortic dissection.

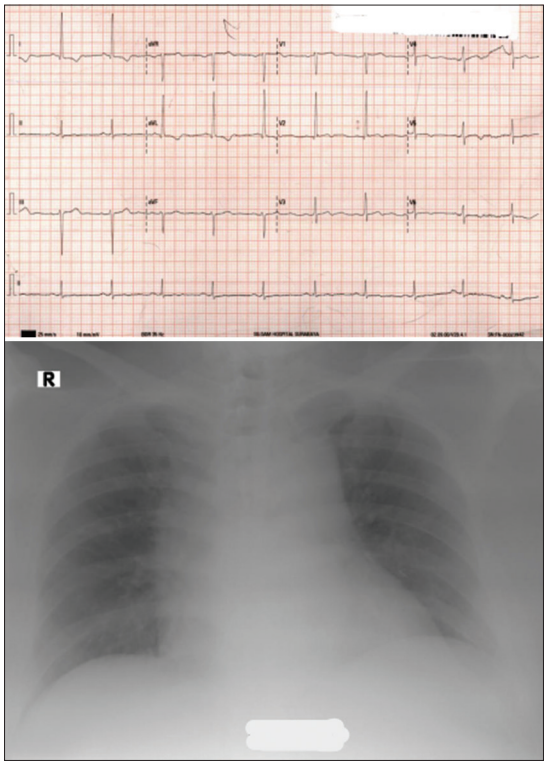


Figure 1: The initial assessment included a chest X-ray and an electrocardiogram

The pain was described as having a tearing sensation that radiated toward the abdomen area. The physical examination of the patient revealed several noteworthy observations. These included a blood pressure measurement of 235/133, the presence of a wider mediastinum and cardiomegaly as shown on the chest X-ray, a slight increase in Hs-Troponin levels, and an electrocardiogram (ECG) showing sinus rhythm accompanied by left ventricular hypertrophy (Figure 1). Based on the information gathered from the patient's medical history, physical examination, and other diagnostic tests, there exists a significant level of doubt among healthcare practitioners regarding the diagnosis of acute coronary syndrome. As a result, the medical professionals proceed to do computed tomography (CT) scans as a diagnostic process to rule out the three potential conditions in the patient stated earlier.

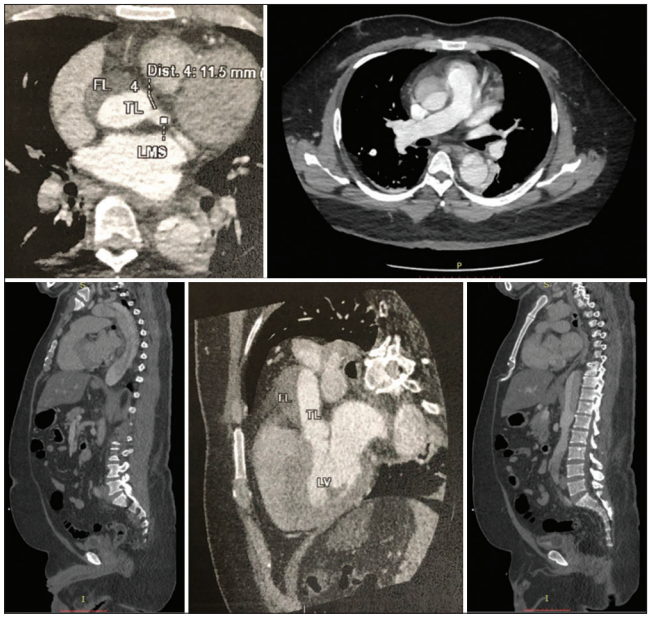


Figure 2. The CT scan revealed the presence of an aortic dissection characterized by the presence of an intimal flap. The formation of a false lumen was observed in the ascending aorta, as well as another false lumen extending from the descending aorta to the right common iliac artery

The CT scans provided evidence of a sequence of aortic dissections that spanned from the ascending to the descending aorta (Figure 2). Notably, the aortic valve appeared unaffected based on the findings from echocardiography. The patient was diagnosed with Stanford Type A DeBakey I aortic dissection. Following this, the individual underwent a surgical treatment that entailed open heart surgery, leading to a triumphant substitution of the complete aortic arch and the implementation of the elephant trunk technique (Figure 3).

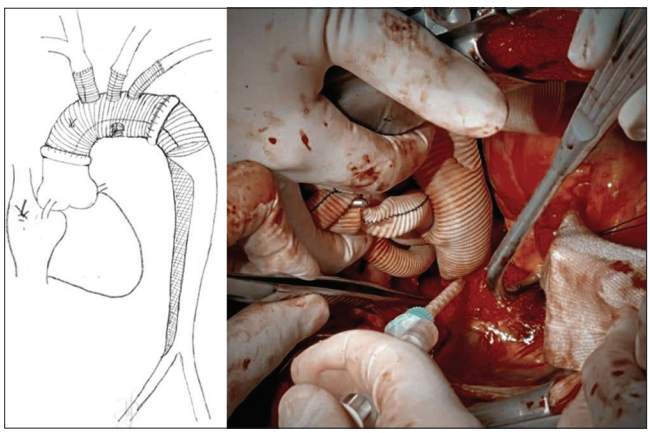


Figure 3. Total arch aortic replacement with elephant trunk procedure to manage patient with aortic dissection standford A DeBakey I

Discussion

Aortic dissection is distinguished by the existence of an intimal rip, allowing blood to flow through the tear and into the media of the aorta. Consequently, the longitudinal division of the intima occurs, resulting in the development of a dissection flap that divides the authentic lumen from a newly created false lumen. The dissection flap possesses the capacity to propagate in either an antegrade or retrograde fashion, resulting in a multitude of catastrophic outcomes that pose a significant risk to the patient's life. The aforementioned issues include acute aortic regurgitation, myocardial ischemia, cardiac tamponade, acute stroke, and malperfusion syndromes [5]. In this particular instance, the CT scan revealed the presence of intimal tears and a false lumen in the ascending and descending aorta, with no evidence of aortic regurgitation as determined by physical examination and echocardiography.

Determining the prevalence of aortic dissection presents a challenge due to the mortality rate ranging from 18% to 49% in patients prior to its diagnosis. Acute aortic syndrome has an incidence ranging from 2.6 to 7.7 cases/100,000 person-years, with the middle-aged comprising 15 cases/100,000. The early mortality rate associated with acute aortic dissection is exceptionally high, reaching 1%/h in the initial hours following acute type A dissection. Type A aortic dissection is most frequently observed in patients aged 50–60 years. Approximately two-thirds of cases involve aortic dissections occurring in males. However, a significant proportion of patients, surpassing 20%, assert a familial predisposition to thoracic aorta dissection [1], [3]. Based on available epidemiological data, this particular case is more prevalent among men than women, and a history of aortic dissection occurred in the patient's father, but at a younger age than usual.

Aortic dissection is frequently observed in conjunction with several underlying medical conditions, including genetic diseases. Connective tissue abnormalities, including Marfan syndrome, Loeys-Dietz syndrome (LDS), and type IV Ehlers-Danlos syndrome (EDS), are widely recognized as predisposing factors for the development of aortic aneurysms and dissections. The etiology of Marfan syndrome involves an autosomal dominant inheritance pattern resulting from a mutation in the FBN1 gene. This gene is responsible for encoding fibrillin-1, a crucial protein involved in maintaining the structural integrity of the extracellular matrix and regulating the activity of transforming growth factor beta (TGFβ). The classification of LDS encompasses six distinct subtypes, which are differentiated according to the specific gene that is afflicted. Type IV EDS, also known as the vascular variation of EDS, is distinguished by the presence of vascular fragility, which ultimately results in rupture and hemorrhage. The observed phenomenon can be attributed to a genetic mutation occurring in the COL3A1 gene, which is responsible for encoding type III collagen, a vital structural protein found in the extracellular matrix [3], [6], [7].

Non-syndromic familial thoracic aortic aneurysm and dissection (FTAAD) refers to a collection of hereditary genetic alterations that lead to a heightened susceptibility to the development of aortic aneurysms and dissections. Approximately 30% of patients diagnosed with FTAAD possess one of the 37 genes that have been linked to the pathogenesis of aneurysm formation or dissection. Like certain connective tissue illnesses, the mutant genes implicated in FTAAD are linked to the TGFβ signaling pathway or the mechanism of smooth muscle contraction [3], [8].

A dissection is distinguished by an abrupt and severe chest pain that is occasionally described as "ripping" or "tearing." The discomfort typically manifests in the retrosternal or substernal region and may extend distally or proximally as the dissection progresses. Malperfusion resulting from compromised flow in end-organ arteries is observed in approximately one-third of the patients, concomitant with the presence of symptoms. The findings of the IRAD study revealed that 15% of individuals diagnosed with acute Type A AD reported experiencing migrating pain, whereas approximately 20% of patients diagnosed with acute Type B AD exhibited the identical symptom [2], [9]. Based on established theoretical frameworks, the patient's clinical manifestation that prompted their visit to the emergency department was characterized by the sudden onset of severe chest discomfort that extended to the abdominal region.

A plain chest X-ray is not sufficiently sensitive or specific for diagnosing Aortic Aneurysm Syndrome. However, certain radiographic findings such as mediastinal widening, disruption of the aortic knob contour, calcium sign (intimal calcification separation from the aortic wall >5 mm), double density appearance within the aorta, tracheal deviation to the right, and deviation of the nasogastric tube to the right may raise suspicion of aortic dissection or suggest an alternative diagnosis for the patient's symptoms. Interestingly, it has been shown that more than 20% of patients diagnosed with AAD do not exhibit any abnormalities in the mediastinum or aortic contour. This phenomenon is more prone to manifest in people who have a dissection involving a nondilated aorta. The observation of a pleural effusion, which lacked specificity, was notably observed in individuals with prolonged durations until diagnosis [5], [10].

ECG is a widely employed diagnostic test that is routinely administered to people who present with complaints of chest pain. According to the statistics, it has been observed that 30% of individuals demonstrate abnormal results when an ECG shows irregularities. These irregularities can be related to hypertensive changes, impaired coronary ostia, or underlying coronary artery disease. In the context of the IRAD

series, it was noted that 42% of individuals diagnosed with ATAAD demonstrated nonspecific ST and T-wave disturbances on their electrocardiograms, indicating the presence of ischemic changes. Anticipating diagnostic delays might be expected when an ECG exhibits abnormalities, since medical professionals often prioritize the study of coronary disease as an initial diagnosis owing to its higher prevalence [10].

The utilization of imaging techniques is of utmost importance in the identification and treatment of this disorder, as it is common for dissections to be overlooked, presentations to deviate from the norm, and the potential for adverse health outcomes to increase over time. Within the framework of Investigational Research and Development (IRAD), the predominant diagnostic modality utilized was CT, accounting for approximately 69% of cases. Echocardiography was employed in 25% of the cases, while magnetic resonance imaging was utilized in 4% of the cases. CT has emerged as the preferred imaging technique for the repeated assessment of patients in ambulatory settings, hospital settings, and emergency departments when diagnosing aortic dissection. The diagnosis of acute aortic dissection is achieved with a high degree of accuracy using contrast CT, which demonstrates a sensitivity and specificity that falls within the range of 95–98%. CT is the primary imaging technique employed by physicians for evaluation purposes, especially in rural regions, owing to its extensive availability. The significance of this matter lies in the fact that an accurate diagnosis plays a crucial role in determining the appropriate course of therapy and predicting the future outcome. [5], [10], [11]

The patient underwent a surgical intervention that encompassed total aortic arch replacement and the implementation of an elephant trunk technique. The decision to forgo aortic valve replacement was made based on the absence of aortic regurgitation resulting from aortic dissection. Surgical is the preferred therapeutic approach for managing type A aortic dissection. The mortality rate linked to untreated ATAAD is reported to be 1% every hour. Prior research has established the superiority of surgical treatments over conservative care, particularly in instances where patients present with unfavorable circumstances and/or major comorbidities. The research investigated a group of 936 individuals who had been diagnosed with ATAAD and were included in the International Registry of Acute Aortic Dissection (IRAD) database. The investigation mostly concentrated on individuals within the age range of 80 years or below. The results of the study indicated a statistically significant decrease in the mortality rate among patients who underwent surgical intervention in comparison to those who got pharmacological treatment during their hospital stay. The study revealed a reduction in the rate of mortality during hospitalization among those in their eighties who underwent surgical procedures, in comparison to those who got conservative medical treatment (37.9% vs. 55.2%) [5], [9].

The primary strategy for addressing dissections confined to the descending aorta should involve drug treatment as the initial approach. It is recommended to provide timely interventional therapy for cases involving complications related to dissection, such as persistent pain or hypertension, rapid aortic enlargement, hemorrhage or contained rupture, and ischemia of distal organs [12].

The endovascular technique can be conducted in two ways: concomitantly with the initial surgery or as a separate procedure, either preceding or following the initial open operation. A multi-center randomized controlled trial was conducted, involving a total of 154 enrolled participants. This study suggests that the optimal timing for doing TEVAR surgery is between 30 and 90 days after the commencement of elephant trunk treatment. The purpose of this timing is to enhance the process of collateral revascularization of the intercostal arteries and reduce the adverse effects of the rigid and thick intimal flap on the remodeling of the aorta. The suggestion for promptly implementing interventional therapy, including an endoprosthesis, should be restricted to cases where problems resulting from dissection are evident [13]. Based on a prior investigation, the TEVAR technique was not conducted for the descending aorta during the acute onset. However, it is intended to be performed in subsequent instances, as no associated complications were observed.

Conclusion

Acute aortic dissection is an infrequent and potentially fatal disorder affecting the aorta that is accompanied by substantial morbidity and death. Around 20% of individuals who are diagnosed with Stanford A aortic dissection exhibit symptoms of an inherited familial condition. At present, noninvasive cardiac tomographic imaging holds a prominent position in the fields of primary diagnosis and treatment planning. The primary approach for managing ATAAD typically involves open surgical intervention. However, the selection of surgical and perioperative techniques may vary depending on the individual's clinical manifestation and the characteristics of the aortic condition. The postoperative care of descending aortic dissection may be pursued in the absence of any problems associated with the endovascular technique.

Authors contributions

Idea/concept: FIR/SAS. Design: FIR. Control/supervision: SAS/YAN. Data collection and processing: FIR/SAS. Extraction/Analysis/interpretation: SAS/YAN.

Literature review: FIR/SAS/YAN. Writing the article: FIR/SAS/YAN. Critical review: SAS/YAN. All authors have critically reviewed and approved the final draft and are responsible for the content and similarity index of the manuscript.

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