

A Case of Sudden Death Due To Aortic Dissection in a 13-Year-Old Patient

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Abstract: A thirteen-year-old boy suddenly collapsed in junior-high school. His medical history included congenital hydronephrosis, inguinal hernia, asthma and atopic dermatitis. Furthermore, his skin was vulnerable to trauma. On arrival, he was in a state of cardiopulmonary arrest with dilated non-reactive pupils. The initial rhythm was asystole. A Marfanoid body and extreme features with exophthalmos and dolichocephaly were observed. Rapid ultrasound revealed cardiac tamponade. Urgent thoracotomy and pericardiectomy, which were performed to release the hematoma, failed to obtain a return of spontaneous circulation. Autopsy imaging revealed dissection of the ascending aorta. Because the present case had Marfanoid body features, exophthalmos due to craniosynostosis, and suffered a fatal aortic dissection at 13 years of age, Loeys-Dietz syndrome was clinically suspected. We did not receive parental consent to perform an autopsy or a genetic analysis. Health examinations at school are very important for a sporadic case because an early diagnosis, close monitoring and early surgery may prolong the life of affected individuals.

Keywords: Marfanoid; Loeys-Dietz syndrome; fatal aortic dissection

INTRODUCTION

Sudden death from aortic dissection with ascending aortic aneurysm is an uncommon but important finding in all series of sudden deaths in young patients [1]. Individuals who are at risk of sudden death from aortic dissection include those who have any of a variety of conditions in which structural weakness predisposes the ascending aorta to pathological dilation under prolonged periods of increased wall stress. These conditions include Marfan syndrome, Loeys-Dietz syndrome (LDS), bicuspid aortic valve, and the vascular form of Ehlers-Danlos syndrome [1]. LDS shares many of the clinical features of Marfan syndrome, which is an autosomal dominant connective tissue disorder caused by mutations in the fibrillin-1 gene. LDS is a newly discovered connective tissue disease caused by mutations in the transforming growth factor β receptor gene [2]. The cardinal features of LDS consist of craniofacial features, which are characterized by widely spaced eyes due to craniosynostosis, bifid uvula and/or cleft palate, and cardiovascular diseases, such as aortic root dilatation or aortic dissection, arterial tortuosity and aneurysms [2]. The natural history of LDS is characterized by aggressive arterial disease and a high incidence of pregnancy-related complications, including death and uterine rupture [3]. We herein report a case of sudden death due to aortic dissection in a 13-year-old boy with suspected LDS.

CASE PRESENTATION

A thirteen-year-old boy suddenly collapsed after yelling during rest time at recess in junior-high school. His medical history included congenital hydronephrosis, inguinal hernia, asthma and atopic dermatitis. A genetic study at birth did not reveal a causative gene for his congenital hydronephrosis. Furthermore, his skin was vulnerable to trauma; however, the cause of the condition was unknown. He had normal intelligence. When the school teacher checked him, he did not display spontaneous breathing. Basic life support and automated external defibrillator were applied, but the patient's initial rhythm was not shockable (Figure 1). When the emergency medical technicians initially checked him, pulseless electrical activity was detected. After tracheal intubation, the securing of a venous route and two infusions of adrenaline (1 mg), the patient was transferred to our hospital by a physician-staffed-helicopter. On arrival, he remained in a state of cardiopulmonary arrest with dilated non-reactive pupils. The initial rhythm was asystole. A Marfanoid body and extreme features with exophthalmos and dolichocephaly were observed. Rapid ultrasound was performed to investigate the cause of shock and hypotension revealed cardiac tamponade (Figure 2). Urgent thoracotomy and pericardiectomy, which were performed to release the hematoma, failed to obtain a return of spontaneous circulation. Autopsy imaging revealed dissection of the

ascending aorta (Figure 3). Wrist and thumb signs, hind-foot deformity, dural ectasia, reduced upper segment to lower segment and increased arm/height, mild scoliosis, and reduced elbow extension were observed, which fulfilled the revised Ghent criteria for Marfan syndrome (Figures 4, 5) [4]. In addition, his cephalic index was 66% (normal 74-83%) with

exophthalmos (Figure 6); thus, a diagnosis of dolichocephaly due to craniosynostosis was made. We did not receive parental consent to perform an autopsy or a genetic analysis.

The electrocardiogram shows sinus rhythm with ST elevation.



Fig-1: An electrocardiogram at the scene

The ultrasonography showed cardiac tamponade with a high echoic mass, suggesting hematoma.



Fig-2: Rapid ultrasonography on arrival to investigate the cause of shock and hypotension

The imaging shows the collapse of the ascending aorta with mediastinal hematoma (arrow: aorta).

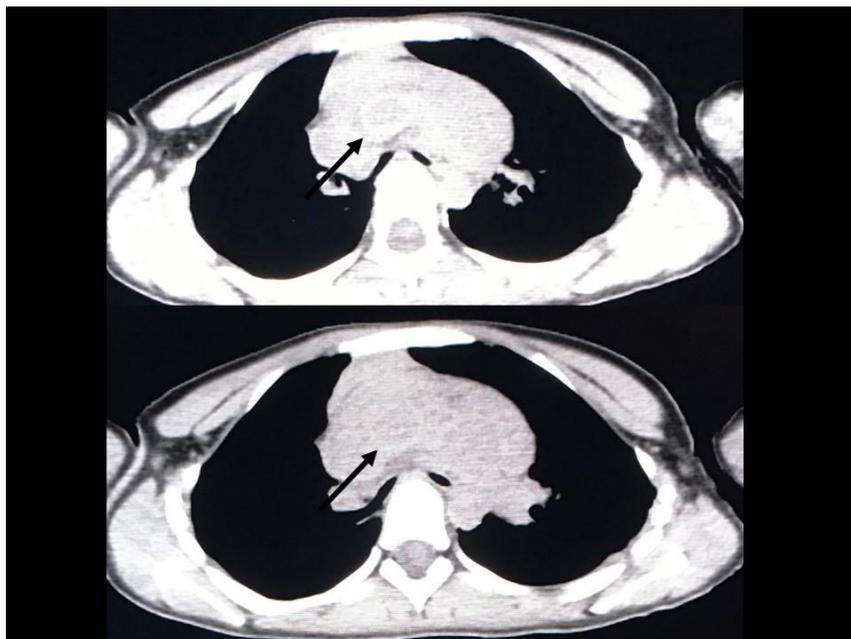


Fig-3: Autopsy imaging of the chest

Wrist and thumb signs, and hindfoot deformity were observed.



Fig-4: The features of the patient's extremities

The image shows dural ectasia (black arrow: spinal cord) and mild scoliosis. The left hip joint seems

to show acetabular protrusion; this was not confirmed by a professional orthopedician.

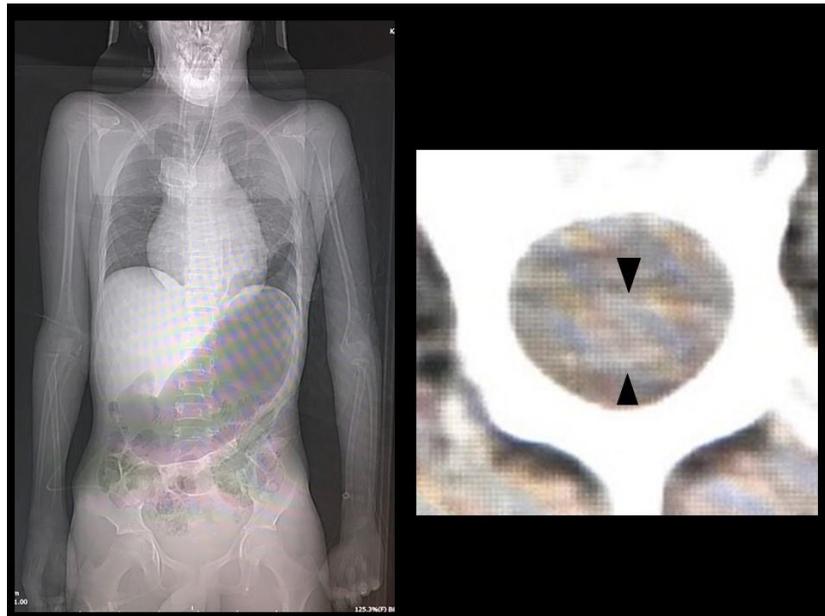


Fig-5: Radiological imaging of the trunk.

The image shows exophthalmos. The cephalic index was 66%.

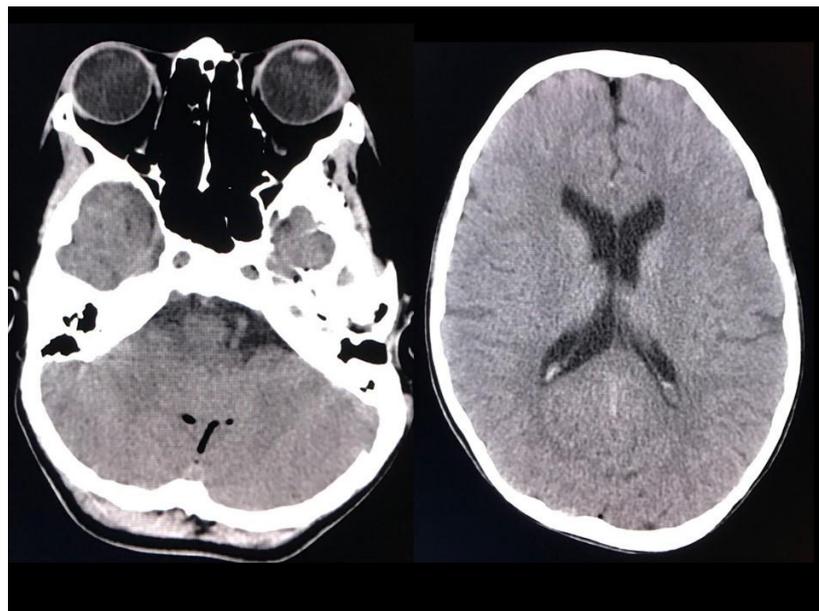


Fig-6: Radiological imaging of the head

DISCUSSION

Chiu *et al.* reported the current epidemiological profile of 2329 patients with Marfan syndrome who were registered in the Taiwan National Health Insurance database [5]. The average annual mortality rate was 0.23%, with most deaths occurring due to cardiac events (including dissection and sudden death). Aortic dissection occurred in 226 patients (10%)—62% of whom were male—at a mean age of 36 years. The probability of freedom from dissection at 20, 40, and 50 years of age was 99%, 80%, and 66%, respectively. In contrast, the natural history of LDS is characterized by fatal aggressive arterial disease, with

death occurring at a mean age of 26 years. Individuals with LDS type I (with craniofacial manifestations) underwent cardiovascular surgery earlier (13 vs. 26 years) and died at a younger age (22 vs. 31 years) in comparison to patients with LDS type II (minimal or absent craniofacial features) [3]. Although enophthalmos is a systemic feature of Marfan syndrome, the patient in the present case had exophthalmos due to craniosynostosis [4]. In addition, the patient in the present case suffered a fatal aortic dissection at 13 years of age. Based on these findings, Loays-Dietz syndrome was clinically suspected [3]. Shprintzen-Goldberg Syndrome is also characterized by

Marfanoid features and craniosynostosis [6]. However, the pathway of transforming growth factor β receptor was recently demonstrated to play a key role in the pathogenesis of Shprintzen-Goldberg syndrome, similar to Loeys-Dietz syndrome [7]. Although some reports of congenital hydronephrosis as a complication of Marfan syndrome, no such reports have been published in relation to Loeys-Dietz syndrome [8]. In the present case, we did not obtain permission to perform a genetic analysis, so we could not clinically diagnose Marfan syndrome or Loeys-Dietz syndrome.

What countermeasures exist to prevent sudden death? If a baby's parent has Marfan syndrome or Loeys-Dietz syndrome, a genetic analysis is recommended. If the baby displays genetic transformation, such as fibrillin-1 or transforming growth factor β receptor, routine echocardiography, including aortic root measurements is required [3]. In addition, magnetic resonance angiography or CT with three-dimensional reconstruction from the head to pelvis should be performed to identify arterial aneurysms and arterial tortuosity throughout the arterial tree [3]. β -blockers are the current gold standard for prevention and treatment in all patients, including children; however, titration is necessary to achieve a heart rate after submaximum exercise of <100 beats/min in patients of >5 years of age [9]. For young children with the most severe systemic manifestations of LDS, the surgical repair of the ascending aorta should be considered once the maximal dimension exceeds the 99th percentile and the aortic annulus exceeds 1.8–2.0 cm, allowing for the placement of a graft of sufficient size to accommodate growth [3]. In cases in which a baby's parent does not have Marfan syndrome or Loeys-Dietz syndrome, it is important to initially assess the clinical features based on the Ghent criteria. However, Faivre *et al.* reported that the majority of the clinical manifestations of Marfan syndrome increased with age, which emphasized the poor applicability of the international criteria to this diagnosis in childhood and the need for follow-up monitoring in cases in which Marfan and Marfan-like syndrome are clinically suspected, similar to the present case [10]. Accordingly, health examinations at school are very important because an early diagnosis, close monitoring and early surgery may prolong the life of affected individuals [11].

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