Congenital Dissecting Thoracic Aortic Aneurysm: A Rare Pediatric Case Report Experience

Siti Aishah Ahmad Maulana1*, Sharifah Huda Engku Alwi2, Mohd Shafie Hashim2 and Rabiatul Adawiyah Abdul Rohim1

1Faculty of Medicine, Universiti Sultan Zainal Abidin, 20400 Kuala Terengganu, Terengganu, Malaysia
2Neonatologist, Hospital Sultanah Nur Zahirah, 20400 Kuala Terengganu, Terengganu, Malaysia

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*Corresponding author: aishah1906@gmail.com
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Abstract

A congenital thoracic aortic aneurysm is very uncommon in pediatric age groups. But in some syndromes, it may be a common association. They can be asymptomatic, until patient presented with some respiratory problem and by imaging, accidentally detecting the disease. Here, we present a case of congenital thoracic aortic aneurysm complicated with dissection in a syndromic girl of a nine years old girl with a phenotypic appearance of Noonan syndrome (NS); together with cleft lip and palate, severe club foot, cardiac lesion of large Atrial Septal Defect (ASD) and intellectual impairment. She had history of a large ASD which was operated 7 years ago in one of local cardiac center. Initially she presented with rapid breathing and upper respiratory tract symptoms, CXR revealed huge upper mediastinum with cardiomegaly. Echocardiogram by consultant pediatrician showed possible of ascending thoracic aortic aneurysm. Urgent CT angiogram (CTA) aorta was performed, demonstrated very huge aortic root-ascending thoracic aortic aneurysm with small aortic dissection. No evidence of aortic leak.

Keywords: Atrial Septal Defect, CT angiogram, Noonan syndrome.

Introduction

A nine years old girl with a phenotypic appearance of Noonan syndrome (NS) which was confirm genetically in one of the local medical centre; together with cleft lip and palate, low set ears, philtrum jutting anteriorly and severe club foot, cardiac lesion of large Atrial Septal Defect (ASD) and intellectual impairment. She underwent a repair of large ASD during infancy in National Heart Institute, however she defaulted follow up few years after that. Her current presentation of moderate respiratory distress to a district hospital with short history of rapid breathing preceded with mild flu-like illness and audible wheeze thus was treated as acute exacerbation of Bronchial Asthma (AEBA) and referred to us for Non-Invasive Ventilatory (NIV) support. Retrospectively, she had recurrent visit to General Practitioners and was treated as having recurrent upper respiratory tract infection (URTI) . Hence the family were self-purchased of Metered Dose Inhaler bronchodilator and self “treated” her as such.

On examination, she was lethargic, hypotensive, tachypnoeic and tachycardic. There was hyperdynamic pericardium and displaced apex beat. Otherwise, no cardiac murmur was heard. Lungs were fairly clear Other systems are essentially unremarkable except her syndromic features as mentioned earlier. Her chest radiograph showed a presence of cardiomegaly with the right heart border occupying the right hemithorax.
**Figure 1**: CXR AP- cardiomegaly with dilated right heart border as well as the right mediastinum.

Echocardiography revealed a moderate Mitral Regurgitation (MR), with a large lesion compressed on the left ventricle (LV), possibly of dilated ascending aorta (measuring 10.3 cm x 8.3cm). No any thrombus or aortic regurgitation.

**Figure 2**: Short axis view of echo shows very dilated aortic root (as shown with arrow) with blood stasis inside.

Aortic root and ascending aorta huge aneurysm with small dissection at distal ascending aorta was diagnosed by CTA aorta.

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Figure 3: Coronal and sagittal CTA show dilated aortic root and ascending aorta with small dissection (as shown with arrow) at distal part of ascending aorta. Normal aortic arch and descending aorta.

Figure 4: Axial CTA shows the dissecting part of huge ascending aorta (arrow). Severely compressed trachea (arrow head) from huge aneurysm.

Figure 6: Volume rendering image of huge ascending aortic aneurysm.

Blood parameters were normal and not suggestive for acute infections. She was subsequently intubated for worsening respiratory distress but unfortunately we had difficulty to ventilate her afterwards as this condition was most likely due to severe compression of trachea by huge aneurysm. She subsequently succumbed to death and the cause of death was due to severe airway compromised from huge aneurysm of aortic root and ascending aorta.


Discussion

**Thoracic aortic aneurysm**

The incidence of aortic aneurysm has been estimated to be 5.9 cases per 100000 person over 30 years period and the incidence was equally in both sexes[1]. A retrospective study of 128 young patients and children (<21 years of age with TAA, 48 patients were found to have syndrome associated with TAA, or complex congenital heart disease or at least moderate aortic stenosis, as they suggest whenever TAA is found in a child or young adult there is a significant association with genetic syndrome or cardiovascular abnormalities [2,3].

Thoracic aortic aneurysm is mostly seen in the ascending aorta, but it can also be seen in the descending aorta and/or the rest of aortic branches [4,5]. Although it is rare, aortic aneurysm can be important cause of mortality in children and adolescents. Aortic aneurysm may be related to hereditary diseases (Marfan syndrome, Loeys-Dietz syndrome, Ehler-Danlos syndrome, Arterial Tortuosity Syndrome, Cutis laxa syndrome, Alagille syndrome, and Noonan Syndrome (NS), or non-genetic diseases (bicuspid aortic valve, coarctation of aorta, Tetralogy of Fallot, and aortitis syndromes). As like our patient with phenotype of NS.

**Noonan Syndrome (NS)**

NS is a known genetic disorder caused by mutations altering proteins relevant to RAS/mitogen-activated protein kinase (MAPK) signal transduction. It is a genetically heterogeneous, pleomorphic autosomal dominant disorder [6] characterized physically by hypertelorism, a downward eye slant, and low set posterior rotated ears, short stature, short neck with webbing, cardiac anomalies, epicanthic folds, deafness, motor delay, and bleeding diathesis. Cardiac defects are recognised complication, however the incidence is unknown, mostly they are with pulmonary valve stenosis and hypertrophic cardiomyopathy. In a series of 118 patients 27% had pulmonary stenosis or dysplasia and left ventricular hypertrophy in 25% with mostly localised septal hypertrophy with 10% having secundum atrial septal defects. There were also reported cardiac abnormalities of ventricular septal defects, patent ductus arteriosus, tricuspid atresia, mitral stenosis and subaortic stenosis rare reports of aortic aneurysm in NS [6]. A retrospective study of echocardiogram in 37 patients with NS and without confounding medical conditions (age ranged from 0.6 to 32 years), they found that 27% (10 patients, p<0.01) had aortic annulus aneurysm while 21.6% (8 patients, p<0.05) had aortic root aneurysm, these were based on Z-scores>2, however the mean Z-scores for sinotubular junction and ascending aorta diameter were 0.05 and 0.19, respectively indicating of no significant different with general population [7]. Few studies individuals with NS, shows dilated aortic annulus and root were often presenting in childhood and very rare in adulthood and the dilation of the aortic annulus and root can be progressive [7,8,9]. Some case reports of patients with NS who presented with aneurysms of coronary arteries [10], intracranial arteries [11], the main pulmonary artery[12] and the descending aorta [13]. These findings reveal that the vascular involvement in NS can be very broad. More information is needed to determine if screening for aneurysm such as with magnetic resonance angiography (MRA) is appropriate for individuals with NS in general, those with particular mutations, or those with aortic root aneurysm.

**Aortic dissection**

Fatal disease do occurs in young populations predominantly with congenital heart disease, connective tissue disease or severe trauma, they are life threatening disease requiring early diagnosis and treatment [14]. Aortic dissections occur in less than 3.5% with mortality is 0.035% in young patients under 19 years old, as it most common in adolescent rather than childhood. However aortic dissection is extremely rare in NS. Only a case report in 1984 documented in an adult patient with aortic dissection of ascending aorta who presented with severe anterior chest pain, CXR showed widened mediastinum and the dissection was then confirmed by CT Angiography (CTA) [15].

Untreated aortic dissection has a higher mortality rate of 1-2% hourly for the first 48 h from symptom onset [15]. Furthermore, evaluation of predisposing factors are important particularly in the congenital causes, they can also be non-cardiac causes of aortic dissection as connective tissue disorders. These disorders usually have clear physical stigmata associated with their own condition and autosomal dominant inheritance [16].
Presenting symptom of aortic dissection is mostly severe anterior or posterior chest pain (85-95%) with first site of the pain varies and migrates in 60-80% to the exact site extend of the dissection[18]. The chest X-ray (CXR) of aortic dissection is not always specific for aortic shadow. Patients can have normal CXR in aortic dissection, but they may have symptoms suggestive of the disease [17]. However, mediastinal widening, pleural effusion, abnormal aortic contour, and cardiomegaly are predictors for the disease [15]. Some of diagnostic radiological modality offers to support the diagnosis, CTA is the first imaging of choice, other diagnostic tools are aortography, MRI, or echocardiography are helpful. Trans-oesophageal echocardiography is known to be highly sensitive and specific for detection of intimal flap aortic dissection as well as it is convenient to be performed at the bedside in the emergency room [16]. Aortic dissection is categorized as two types, as type A with involvement of the ascending aorta, while type B with restricted involvement within the aorta distal to the left subclavian artery. Like our patient, she suffered type A aortic dissection. Surgery is necessary for type A or type B dissection impending rupture, rapid progression of symptoms, or risk of malperfusion of vital organs [19].

Early operation can give better outcomes to these group depending to the severity of the disease and stability of patient condition. Unfortunately, our patient did not proceed with surgical intervention in view of her unstable condition besides huge aneurysm.

**Vascular tracheobronchial compression**

A retrospective study in 810 patients, 81 patients(10%) were found to have vascular tracheobronchial compression as the leading cause was from vascular ring or pulmonary artery sling (55.5%) rather than to the rest causes of the vascular tracheobronchial compression like abnormal enlarged or malposition cardiovascular structure (34.6%) and post operative congenital heart disease (CHD) (9.9%) in a paediatric heart centre. Many of the patient may require respiratory support due to respiratory distress, risk of morbidity is also increased during perioperative period [20]. They also emphasized, the presence of vascular tracheobronchial compression should be suspected in children with CHD if there is persistent respiratory symptoms or difficulty in weaning off respiratory support despite successful correction of underlying cardiac defects [20,21].

**Conclusion**

The earlier future identification of children or adolescent of NS with TAA complicated by airway compression may allow early medical or surgical treatment to be initiated at a younger age with possible long-term benefit and reduce number of mortality.

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**References**


