Interrupted aortic arch in a 4-year old girl

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ABSTRACT

Interrupted Aortic arch (IAA) is a lethal, rare congenital cardiovascular malformation (3:1,000,000), characterized by absence of structural connection between ascending and descending aorta. IAA is divided into 3 types based on the location of the interruption: discontinuity distal to the left subclavian artery is classified as type A, discontinuity between the left common carotid and the left subclavian artery is type B (the commonest), and discontinuity between the innominate and left common carotid artery is classified as type C (the rarest). The median age of death is 4-10 days, if untreated 75% will die within one month of birth. Longer survival is possible but 90% will die by one year old. The cause of death usually are combination of increase left to right shunt, pulmonary edema and ductal stenosis. Hereby we presented a four year old girl with IAA type A.

Keywords: IAA, aortic arch, pediatric, VSD, PHT

INTRODUCTION

Interrupted Aortic arch (IAA) is a lethal, rare congenital cardiovascular malformation (3:1,000,000). It is characterized by absence or discontinuity between two segments of the aortic arch. It means there is no direct way for rich oxygen blood leaving the heart to reach the body parts that are “downstream” from the blockage. It is thought to be a result of faulty development of the aortic arch system during the fifth to seventh week of fetal development. The commonest type of IAA is type B and the rarest type is type C.1-3

IAA usually occurs in association with ventricular septal defect (VSD) and ductus arteriosus. In newborns with interrupted aortic arch, the only way for blood to bypass the blockage is via the patent ductus arteriosus (PDA). Prior to birth, this small blood vessel permits blood to bypass the baby’s fluid filled lungs. Normally one or two days after birth, this vessel usually will close on its own. In a baby with interrupted aortic arch, if this duct close, blood can no longer reach the lower body that cause the baby to suddenly become very ill. Therefore prostaglandin is needed to keep the ductus arteriosus open until surgery is done.1-3 If the ductus arteriosus stay open, longer survival is possible but even then, 90% will die by one year old. This uncommon anomaly is highly lethal, with the median age of death being 4 to 10 days and 75% of such babies will die within one month of birth. Death occurs when the ductus closes soon after birth with congestive heart failure.4

As like other obstructive lesions in the left heart that are duct dependent, IAA tends to present with non-specific signs of heart failure simultaneous with closure of the arterial duct within the first few days of life, that cause IAA without surgery is associated with high mortality in the neonatal period.1
CASE ILLUSTRATION

ICDA, a 4 years old girl, was referred for evaluation of complex congenital heart defects (CHD) to private hospital by a pediatrician with chief complaint of exertional dyspnoe, palpitation, tachypnea and frequent respiratory infection since birth. From parents history taking the girl was known has cardiac problem and had been echocardiography at 3 months of ages but patient had not followed up since then. Parents were non consanguineous. From chest physical examination there was pansystolic murmur (PSM) grade III/VI on the left side of sternum at the 2nd intercostal space and the left scapular region in the back. The child was then had chest radiograph that showed cardiomegaly with left atrial and ventricle hypertrophy with aortic enlargement and pulmonary edema. (Figure 1)

The child then had the second transthoracic echocardiography which showed dilated left atrium and ventricle (Figure 2). Large perimembranous ventricle septal defect (PM VSD) (right/left 8.1/13.2), without aortic rim, very minimal membrane septal aneurism are shown in Figure 3; large PDA (5.5/13.3) are shown in Figure 4, pulmonary hypertension (PHT). The child then was given oral anti failure therapy consist of 12 mg furosemid, 10 mg spironolacton, 2.5 mg captopril, 50 mcg digoxin and 2.5 mg sildenafil twice daily.

She was scheduled for cardiac catheterization to close the PDA in private hospital. From the cardiac catheterization which lasted for about 139 minutes venography from innominate vein showed single right superior vein cava without persistent left superior vein cava (Figure 5), descending aortography (lateral projection) showed all contrast entered main pulmonary artery (MPA) via large PDA with diameter 6.6 ± 1.0 mm and interruption of descending aorta. Diameter of descending aorta was 9.0 ± 1.3 mm. Meanwhile from right ventriculography (antero posterior and lateral projection) showed large MPA, with confluent pulmonary arteries (right pulmonary artery 15.9 ± 1.9 mm; left pulmonary artery 12.4 ± 1.5 mm) (Figure 6), left ventriculography (lateral projection) revealed single large PM VSD (9.7 ± 1.1 mm), aorta drain from left ventricle, simultaneous ascending and descending aortography revealed IAA (6.4 ± 1.0 mm – 8.3 9 ± 0.9 mm) with the interruption is on distal to left subclavian artery (type A) with one collateral ascending aorta to MPA (2.8 ± 0.4 mm) (Figure 7). Ascending aortography (lateral projection) showed some contrast viewed in MPA across collateral, left aortic arch; meanwhile in antero posterior view showed suspect single ostium coronary arteries (Figure 8).
From the cardiac catheterization we concluded this girl has interrupted aortic arch (type A), large VSD, large PDA, collateral and PHT (high flow, high resistance). After then, the family was advised for further surgery in Jakarta. But the family rejected and chose palliative treatment.

DISCUSSION

Embryology of aorta is very important in the development of IAA. The development of the branchial apparatus begins during the second week of gestation and completed by the seventh week. The apparatus consist of 6 branchial arches in the wall of the foregut. The branchial arches are numbered 1 to 6 from cephalad to caudal. Each of the branchial arches connects paired dorsal and ventral aortas. Although the classic Rathke diagram shows 6 aortic arches, in reality the arches appear and disappear at different times. The 6 branchial aortic arches normally develop into the thoracic aorta and its branches. The first 2 arches involute before development of the sixth arch, and the fifth arch is atretic or never fully develops. The third arch and portions of the ventral and dorsal aortic arches contribute to the head and neck arteries. The fourth arch becomes the aortic arch, and the pulmonary arteries develop from the sixth branchial arches (Figure 9). On the right side, the dorsal contribution of the sixth arch disappears, and on the left it persists as the ductus arteriosus. The intersegmental arteries migrate and form the subclavian arteries. In the embryo, six pairs of aortic arches connect the two primitive ventral and dorsal aortas.1,2

 Interruption of the aortic arch of any type has no important impact on the fetal circulation. This is not surprising in light of the fact that less than
CASE ILLUSTRATION

10% of the fetal cardiac output is usually distributed through the isthmus of the aortic arch. The lower body is perfused prenatally through the ductus arteriosus. Following birth, the lower body of the newborn with interrupted aortic arch continues to be adequately perfused as long as the ductus remains patent and pulmonary resistance remains high.3-4

Auscultation is usually unhelpful. Often a gallop rhythm is present, and the heart sounds are usually easily audible, the second being closely split. An ejection click may indicate the presence of associated bicuspid aortic valve, but this is non-specific. If a murmur is present, it is often pansystolic or mid-systolic and of low intensity, indicating the non-restrictive nature of the ventricular septal defect.3-4 In this case, we heard pansystolic murmur (PSM) grade III/VI on the left side of the sternum at the 4th intercostal space and the left scapular region in the back that was compatible with auscultation in VSD.

Meanwhile, chest radiography demonstrated cardiomegaly with the left atrial and ventricle hypertrophy which is the effect of left to right shunt in VSD, that placed the volume overload to the left atrium and left ventricle, causing it to become hypertrophied and increasing the blood flow to lungs that ends to pulmonary edema.5 Chest radiography from IAA patient usually show large heart with cardiomegaly present in 90% of neonates and increased pulmonary vascular markings with pulmonary edema.3-5 In this case, our patient’s chest radiograph was compatible with literature, we found cardiomegaly with left atrium and ventricle hypertrophy, aortic enlargement and pulmonary edema.

Accurate diagnostic imaging evaluation is important to correctly characterize aortic and cardiac anatomy and define the exact type of IAA. Anatomic features that must be identified include the following: location and length of the aortic vascular defect, caliber of the thoracic aorta proximal and distal to the interruption, branching pattern and origins of the great vessels, location and patency of the ductus arteriosus, appearance of the ventricular outflow tracts, and presence of any other cardiac anomalies.8

Multiple imaging techniques have been described in the evaluation of patients with suspected IAA. At present, chest radiography and echocardiography are the first line imaging modalities used in diagnosing congenital aortic arch anomalies, particularly in children. Echocardiography is considered to be the primary imaging technique for the workup of this entity. It has the advantages of being portable and not using ionizing radiation.6

Although this imaging technique usually provides excellent anatomic definition of the heart, evaluation of the aorta and great vessels can occasionally be limited. Thus, echocardiography may or may not be able to define the exact site of the aortic arch interruption and its relationship to the origins of the great vessels.6 That’s the reason why in this case, we couldn’t find aortic anomaly from echocardiography.

Interrupted aortic arch (IAA) is defined as a total anatomical and luminal discontinuity between ascending and descending aorta. In 1959 Celoria and Paton classified the lesion into three types, based on the site of aortic arch interruption. In type A, the interruption is distal to left subclavian artery. In type B, the interruption is between the left common carotid and left subclavian on the site of the aortic arch interruption and its relationship to the origins of the great vessels.6 That’s the reason why in this case, we couldn’t find aortic anomaly from echocardiography.

IAA may occur as a simple or complex anomaly. In simple IAA, only ventricular septal defect (VSD) and patent ductus arteriosus (PDA) are seen. The complex form is associated with truncus arteriosus, transposition of great artery, double outlet right ventricle, aortopulmonary window and functional single ventricle. Obstruction of the left ventricle outflow tract is also common.9-11 This case only showed a simple IAA and VSD and PDA.

The large ventricle septal defect that was found in echocardiography also present in this child, makes the blood shunted from the left side to the
right side of the heart. This shunting caused blood flow increment to the lungs, which can leads to congestive heart failure as well. Patients with VSD have two major risks. The first of it is infective endocarditis, an infection of the heart that can be fatal if not properly recognized and treated. The second is leakage of aortic valve, which may eventually require surgical repair or replacement.

If this large VSD has not been repaired, patient will usually have high blood pressure in the lungs resulted from the left to right shunt lesion. The pulmonary blood flow will increase, which is direct transmission of systemic pressure to the pulmonary artery and compensatory pulmonary vasoconstriction (pulmonary arterial hypertension) and ended to Eisenmenger syndrome, a condition in which the blood pressure in the lung becomes so high that low oxygen blood mix with rich oxygen blood causing the patient to develop cyanosis.

The ductus arteriosus that connects the left pulmonary artery to the descending aorta also have important implication in pulmonary overcirculation and the left heart volume overload that will decrease lung compliance and resulted in increase work of breathing. This two factors explain the shortness of breath, PHT and bluish appearance of this girl.

Chest radiography and echocardiography are the first line imaging modalities used in diagnosing congenital aortic arch anomalies particular in children. Exquisite anatomic display of cardiovascular structures can also be made by catheter angiography, MDCT and MRI. In this case our reasons we did echocardiography combine with cardiac catheterization to our patient are echocardiography suffers from operator and patient dependent factors and also limited access to entire thoracic aorta. Although MDCT is non invasive but its time consuming, need prolong sedation and doesn't provide sufficient information regarding the esophagus and trachea.

In this patient her cardiac catheterization report almost the same finding as in her echocardiography; only its clearly describe there is an interruption of aortic arch at the distal to left subclavian artery with collateral from ascending aorta to MPA and PHT from simultaneous ascending and descending aortography. Survival of this girl from infancy to toddlerhood relies on the development of collateral vessel, which is essential for the maintenance of distal flow and develop when there is a patent ductus arteriosus which will close gradually.

Actually IAA can be diagnosed in prenatal period by ultrasound. The advantage of prenatal diagnosis is that Prostaglandin E1 can be started immediately after birth to keep the ductus arteriosus open. This allows blood flow to the lower body until surgery is done to reestablish continuity of the aortic arch. If ductal closure occurs abruptly or is not recognized rapidly, the child will soon become profoundly acidic and anuric as perfusion of the lower body becomes entirely depend on the collateral communications between the two separate aortic systems.

Unfortunately this patient was not diagnosed prenatally, but she managed to survive because of the PDA and collateral.

IAA represents a ductal dependent congenital heart disease, which without surgery is associated with high mortality in the neonatal period. The goal of surgery is to reestablish the continuity of aorta. But the surgical approach (which consist of single or staged surgery) is still controversial, with reconstruction through an end to end anastomosis, creation of a subclavian flap, carotid turn down, and placement of an interposition graft.

Single stage repair of interrupted aortic arch was first described by Barrat-Boyes in 1972, arch continuity was established using a synthetic conduit. Single stage repair incorporating direct arch anastomosis was first described by Trusler in 1975. The goal of surgery is to reconnect the aortic arch to create the a continuous “tube” and close the ventricular septal defect. Surgery is typically performed urgently but after the patient is stabilized. The open heart surgery will be done to connect the two separate portions of aorta, close the ventricle septal defect and tie off (ligate) the patent ductus arteriosus.

Complications after interrupted aortic arch repair may include residual obstruction or stenosis (narrowing) at the aortic repair site. The aortic valve or the area below the valve are often small and may not grow, which can result in stenosis in months or years following surgery.

Both single and staged repair of IAA have improved and early mortality in some centers approaches 8-10%. However, the longterm fate of early survivors remains uncertain. There is risk for the development of restenosis at the site of aortic anastomosis and left ventricle outflow tract obstruction, those often require reintervention and represent a risk for late death.

Many literatures have recommended single stage rather than staged repair. The advantages of single stage repair over staged approach include fewer reoperation/reintervention, avoidance of pulmonary artery banding (PAB) which could accelerate subaortic stenosis and the decrease need for future arch reconstruction.

Meanwhile stages approach has been recommended in premature neonates weighting less than 1500 gram, in the presence of severe infection, intracranial bleeding, multiple organ failure and very unfavourably morphology, like the presence
of multiple VSD, transposition of great artery or single ventricle. A study in China that investigated the outcome of surgical treatment of IAA with VSD and PDA in patients over one year of age, suits with our patient who was four years old. This study said that single stage surgery of patient with IAA, VSD and PDA over one year of age can have good surgical results and functional outcomes. But must be followed with careful assessment and treatment of pulmonary artery pressure pre-operatively and post operatively.

While in another study by O’Byrne et all whose all of their subjects also underwent single stage surgery, showed that rates of reintervention were still highest in the first year of life (38% on the left ventricular outflow tract, 33% for the aortic arch, and 24% for both) and decrease in subsequent years with decrement of maximal oxygen consumption, maximal work, forced vital capacity, health status and quality of life. Subjects with IAA demonstrate a significant burden of surgery and transcatheter intervention and large magnitude deficits in exercise performance, health status and quality of life.

That means after surgery the girl must be monitored regularly and restricted from vigorous or competitive sports, only can participate in recreational sports. By both studies we can conclude that despite of surgery, the mortality rate of this girl gonna improve, her quality of life and health status gonna be compromised following the correction of her IAA and the burden of medical care to the family.

**SUMMARY**

Interrupted aortic arch is a rare congenital malformation with prevalence 3 per one million live births and typically occurs with intracardiac malformation, such as a ventricular septal defect, patent ductus arteriosus, bicuspid aortic valve, left ventricular outflow tract obstruction, or aortopulmonary window. This uncommon anomaly is highly lethal with the median age of death being 4 to 10 days, and 75% of such babies will die within one month of birth. Death occurs when the ductus closes soon after birth. Treatment and surgical intervention are vitally important to reestablish the continuity of the aortic arch.

In this case a 4 years old girl came with chief complain exertional dyspnoe, palpitation, tachypnea, grey appearance on the lower body and frequent respiratory infection since birth. Her echocardiogram showed large PM VSD without aortic rim, very minimal MSA and large PDA. Then she was scheduled for cardiac catherization to close the PDA but unexpectedly the ascending and descending aortography revealed interruption of aortic arch at distal portion to left subclavian artery (type A) with one collateral from ascending aorta to MPA, large VSD, large PDA and PHT. Then the family was advised for heart surgery in Jakarta but they refused it, finally the patient passed away 1 year later due to pulmonary hypertension.

**REFERENCES**