Bahrain Medical Bulletin, Vol. 26, No. 2, June 2004

Single Ventricle with Mitral and Aortic Atresia

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We report the detailed autopsy findings in a male neonate who died due to a rare congenital cardiac anomaly, namely, a single ventricle with associated mitral and aortic atresia.

Bahrain Med Bull 2004;26(2):

Congenital cardiovascular malformations occur in about 5 to 8 of every 1,000 live births, and the percentage in stillborn infants is probably even higher[1]. Between the 5th and 8th weeks of embryonic life, the primitive heart tube undergoes a process of folding, remodeling with the formation of valves, and septation that transforms its single lumen into the four chambers of the definitive heart. Although the cause and pathogenesis of most cardiovascular defects is not well understood, genetic errors, environmental teratogens or altered haemodynamic forces may be responsible in most cases[1].

The "univentricular heart" is a generic term for a group of congenital cardiac anomalies where the entire atrioventricular junction is connected to only one chamber in the ventricular mass. This single ventricular chamber receives the entire venous return to the heart. A second ventricular chamber, if present, will lack any atrioventricular connection, and hence be rudimentary[2]. Earlier, Van Praagh et al[3] used the terms single, common and univentricular heart interchangeably. Subsequently, this condition or its variants have been described by various terms: single ventricle, common ventricle, rudimentary ventricle, cor triloculare biatriatum, single ventricle with rudimentary outflow chamber, double-inlet ventricle[4].

We describe the clinical and autopsy findings in a male neonate with a single ventricle and associated mitral and aortic atresia, keeping in view the rarity of the condition and the interesting complex combination of abnormalities.

THE CASE

A male neonate born by a full-term normal delivery in a peripheral hospital was admitted at the M.S. Ramaiah Medical Teaching Hospital, Bangalore, India, at 4 days of age, with shock and respiratory distress. The neonate measured 49cm in length, weighed 3500gm and appeared markedly cyanosed. X-ray chest showed cardiomegaly. The neonate expired soon after admission.

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AUTOPSY FINDINGS - At autopsy the neonate showed marked cyanosis. No external congenital anomaly was detected. The heart was normally situated, enlarged in size and weighed 18g. The major portion of the anterior surface of the heart was formed by the right atrium and the ventricle. The aorta was situated posteriorly and to the right of the pulmonary trunk. The ascending aorta was much narrower in diameter than the pulmonary trunk. The arch vessels appeared normal (Figure 1). On dissection of the heart, the venoatrial connections were found to be normal. The right atrium was markedly dilated. The foramen ovale was patent. The right atrioventricular valve was tricuspid with normal-appearing cusps, and was connected to a single ventricular chamber which had trabecular patterns of neither a left ventricle nor a right ventricle (indeterminate type) (Figure 2). The pulmonary trunk was arising from this single ventricular chamber and appeared normal in caliber with normal branching into the pulmonary arteries. The pulmonary valve was normal. No communication could be traced between the single ventricle and aorta. The aortic valve was atretic and imperforate with absence of cusps. The ascending aorta had a caliber of only 2mm. There was a patent ductus arteriosus in the usual position.

Figure 1. The anterior surface of the heart is formed mainly by the right atrium and the ventricle. The ascending aorta is very narrow in diameter (arrow). The arch vessels appear normal.

Figure 2. Single ventricle. Note the dilated right atrium, small left atrium and absence of the left atrioventricular valve (mitral valve).

The pulmonary veins opened into the left atrium which was very small in size. The left auricular appendage was, however, normal in size. The mitral valve was atretic with absence of cusps and there was no communication between the left atrium and the single ventricle. The blind-ending left atrium was separated from the single ventricle by an atrioventricular sulcus (Figure 2). There were no prominent descending coronary arteries on account of the absent interventricular septum.

The lungs showed features of bronchopneumonia. The other organs were normal.

A final diagnosis of a congenital cardiac anomaly, namely a single ventricle with associated mitral and aortic atresia, was made. On enquiry, the mother denied any history of drug or radiation exposure during her pregnancy. A genetic study could not be performed on the neonate due to the high costs involved.

DISCUSSION

Thiene et al[5] stressed the importance of accurate differentiation of ventricular morphology for accurate classification of the single ventricle. Three morphologic forms of single ventricle are well recognized and have been described in detail by Amplatz and Moller[4].

Left ventricular type, usually with a rudimentary chamber of right ventricular type. This is the most frequent type (75%). It was classified as Type A by Van Praagh et al[3,6], and is also called "double-inlet left ventricle". The rudimentary chamber lies anteriorly and may be located to either the right or left. In 90% cases of left ventricular type of single ventricle, transposition of the great vessels coexists, and outflow obstruction is frequent.

Right ventricular type, usually with a rudimentary chamber of left ventricular type (20%). This corresponds to Type B described by Van Praagh et al[3,6], and is also known as "double-inlet right ventricle". The rudimentary left ventricle is located posteriorly. Straddling of the atrioventricular valves is common. A double outlet from the right ventricular type of single ventricle is usually present, with each great vessel arising from an infundibulum.

Indeterminate type (5%) where there are morphologic features of neither a left ventricle nor a right ventricle, and a rudimentary chamber is absent. The aorta is usually anteriorly situated in these cases.

Our case showed the rare indeterminate type of single ventricle with the aorta, however, being situated posteriorly. Anderson and Becker[7] feel that this rarest variant of the single ventricle is often the most difficult to diagnose with certainty. They noted prominent trabeculations crossing the apex of the single ventricle in these cases, a feature which was not striking in our case.

Amplatz and Moller[4] describe that in the univentricular heart, either both atrioventricular valves may connect to the single ventricle, one of the valves may be straddling, atretic, stenotic or regurgitant, or there may be a common atrioventricular valve. In our case the left atrioventricular valve was atretic.

In each type of single ventricle, the great vessel connections are variable and outflow obstruction to either great vessel is frequent. Pulmonary stenosis or atresia, aortic coarctation and interruption of the aortic arch have been observed by Amplatz and Moller[4]. They also report transposition of the great vessels in 85% of patients with a single ventricle. Our case showed a single outlet pulmonary artery, the aortic valve and ascending aorta being atretic.

Coronary arteries are a guide to the possible position of the rudimentary chamber. Becker and Anderson[8] state that anterior delimiting coronary arteries suggest the presence of a right ventricular rudimentary chamber and posterior arteries a left one. When there is no septum and no rudimentary chamber, there are no prominent descending coronary arteries, as was observed in our case.

Our case appears to show a close similarity to hypoplastic left heart syndrome (HLHS), in that the left atrium was underdeveloped and the mitral and aortic valves were atretic along with a narrow ascending aorta. However, all cases of HLHS show a left ventricle even if it may be minute[9]. In our case, a comprehensive dissection of the heart including serial sectioning of the myocardium failed to reveal even a trace of the left ventricular cavity. This finding fits well with the statement made by Amplatz and Moller[4] that about 20% of patients with single ventricle do not have even a rudimentary chamber. It would, hence, be appropriate to classify our case under the category of single ventricle with overlapping features of HLHS.

Rahimtoola et al[10] describe the haemodynamics of the single ventricle. When there is a single ventricle with no associated anomalies, the entire systemic and pulmonary venous returns flow into this ventricle from where the blood is ejected into the aorta and pulmonary artery depending on the relative resistances to the outflow. An inverse relationship exists between the volume of pulmonary blood flow and the degree of cyanosis.

In our case, owing to the severe complexity of the cardiac anomaly, the mitral and aortic atresia dominated over the other abnormalities, thus altering the haemodynamics. Because of the obstruction in the left side of the heart in the region of the mitral valve, a left-to-right shunt existed at the atrial level through the patent foramen ovale. The entire systemic and pulmonary venous returns entered the right atrium leading to its marked dilatation. From the right atrium the blood entered the single ventricle and then was ejected, essentially, into the pulmonary artery, the aortic valve being atretic. The patent ductus arteriosus was responsible for delivering the blood into the aorta and the systemic circulation. Usually, the ductus arteriosus begins to close functionally by 48 hours after birth. As a consequence of closure of the ductus arteriosus, with the sole source of systemic circulation getting obliterated, hypoxia had ensued, resulting in the death of the neonate. The admixture of the systemic and pulmonary venous blood explains the cyanosis of the neonate in our case.

A few patients with a single ventricle may survive into adulthood even without a surgical intervention[11,12]. All operations for single ventricle are palliative and are usually performed at around 2 years of age. Previously, septation of the ventricle[13] was being done, wherein the single ventricle was divided by a patch into two equal ventricular chambers. However, this procedure was found to have an operative mortality as high as 50%. In the past decade, the Glenn shunt and Fontan operation are being increasingly used[14,15,16]. These procedures involve the creation of a cavopulmonary anastomosis obligating the passage of the systemic venous blood through the healthy mature lungs directly, thus reducing the volume work of the single ventricle. Heart transplantation has also been recommended for patients with a single ventricle, as it may increase the long-term survival rate[17]. In our complex case, had the neonate survived, heart transplantation would have probably been the only treatment option available.

CONCLUSION

Single ventricle with associated mitral and aortic atresia is a rare and interesting complex congenital cardiac anomaly. It is an important cause of morbidity and mortality in early life. In a child presenting with congestive cardiac failure, an early diagnosis of this condition by angiography and echocardiography can facilitate timely surgical intervention. Unfortunately, in the male neonate that we have described, the severe cardiovascular compromise due to the multiple cardiovascular anomalies was incompatible with life, resulting in its death after a short survival.

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