Case Report

AN UNUSUAL CASE OF CONGENITAL ANOMALY OF THE HEART

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RESUMEN

La creciente utilización de diagnósticos invasivos y procedimientos de intervención en enfermedades cardiovasculares han enfatizado la importancia de entender y documentar mejor el tipo y la frecuencia de las variaciones vasculares. Los defectos del corazón se encuentran entre las anomalías congénitas más comunes. Estas pueden ser simples o complejas. En la mayoría de las anomalías congénitas del corazón se requiere intervención quirúrgica para garantizar la vida. Los médicos y cirujanos deben ser educados sobre las distintas formas de enfermedades congénitas del corazón para facilitarles su manejo. En este artículo reportamos un caso de un recién nacido de 28 días de raza india al cual se le detectó, durante la autopsia, una anomalía muy poco frecuente. Esta consistía en la presencia de una sola aurícula y un sólo ventrículo comunicados por un sólo orificio aurículo-ventricular. Casos como este son incompatibles con la vida.

***Palabras clave**: tabique interauricular, tabique interventricular, ventrículo, aurícula, canal aurículo-ventricular*

ABSTRACT

The increasing use of invasive diagnostic and interventional procedures in cardiovascular diseases makes it important that the type and frequency of vascular variations are well documented and understood. Congenital heart defects are among the most common of all birth defects. They can be thought of being common or complex lesions. In most cases of complex congenital heart defects surgical intervention is required in order to sustain life. Surgeons and Physicians need to be informed of the various forms of congenital heart diseases in order to be able to manage such conditions. We report an unusual case found on autopsy of a 28 days old male East Indian neonate, who had single atrial and single ventricular chambers of the heart, which were connected by common atrio-ventricular orifice. This congenital heart defect has to be documented. Such cases are usually incompatible with life.

***Key words**: inter-atrial septum, inter-ventricular septum, ventricle, atrium, atrio-ventricular canal.*

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INTRODUCTION

The primordial heart forms in the cardiogenic plate located in the cranial end of the embryo. Angiogenic cell clusters which lie in the horse shoe shaped configuration in the plate coalesce to form two endocardial tubes. These tubes are then forced into the thoracic region due to cephalic and lateral foldings where they fuse together forming a single endocardial tube. The endocardial tube consists of three layers; the myocardium, cardiac jelly and endocardium. The endocardium can be divided into primordial heart chambers starting caudally at the inflow end: the sinus venosus, primitive atria, primitive ventricle and bulbus cordis. The endocardial tube begins to grow and undergoes dextral looping. Septa begin to grow in the atria, ventricle and bulbus cordis to form the right and left atria, right and left ventricles and two great vessels — the pulmonary artery and the aorta. Partitioning is completed by the end of the eight week and the fetal heart is formed.

Any slight deviation from the normal pattern of development can lead to various types of congenital heat defects, which includes; atrial septal defect (ASD), ventricular septal defect (VSD), patent ductus arteriosus (PDA), pulmonary stenosis (PS), aortic stenosis (AS), coarctation of the aorta, tetralogy of Fallot, transposition of the great arteries, atrio-ventricular septal defect, persistent truncus arteriosus, tricuspid atresia, pulmonary atresia, total anomalous pulmonary venous connection, hypoplastic left heart syndrome, double outlet right ventricle, single ventricle, Ebstein's anomaly and dextrocardia. The diagnosis of congenital heart defects includes careful physical and cardiovascular examination, chest radiography, electrocardiography, echocardiography, cardiac catheterization, cineangiography and cardiac MRI.

Although many early investigators speculated on the pathogenesis of these malformations, the precise developmental processes underlying the lesions remain poorly understood. It has been established that both the environment and genetic factors play important causative roles.

CASE REPORT

We present a 28 days old male neonate of East Indian ethnicity admitted to the emergency department with a two days history of poor feeding and dyspnea. There was no history of vomiting, abdominal distention, fever, cyanosis or constipation. The patient was not on any medication. There were no known allergies and nutrition consisted of breast milk and formula.

The neonate was full term with the mother having normal spontaneous vaginal delivery. The mother denies any alcohol / tobacco use during pregnancy. There were no abnormalities noted at birth. There was no history of complicated labour or post natal problems. The mother and father had four children including the patient and denied any history of medical ailments in the family. The mother also denied any history of complications in previous pregnancies.

On initial examination, the neonate was grunting with evidence of recession (crepitations). A pansystolic murmur was heard, being loudest in the lower left sternal border. It was also noted that there were decreased bowel sounds on the left side of the abdomen. Initial assessment was that the neonate may have had congestive cardiac failure secondary to a ventricular septal defect. A septic screen was performed to rule out sepsis. The patient was treated with furosemide and antibiotics. A chest x-ray was also ordered. The chest x-ray revealed right ventricular hypertrophy and pulmonary edema.

As time progressed, the patient became noticeably worse with decreased oxygen saturation. As a result the patient was placed on oxygen and transferred to the intensive care unit. As time went on, the patient's condition continued worsening, exhibiting signs of cyanosis, motting and increased capillary refill time. Warming blankets were implemented, but his condition was not improved. After further assessment, cardiomegaly was noted in addition to hypothermia. Patient became acidotic and was treated with sodium bicarbonate, sedated and intubated.

After a while the patient was noted to have bradycardia. Attempts were made to resuscitate the patient but there was no response to drug therapy and cardio-pulmonary resuscitation. The patient finally died and based on the symptoms his condition was clinically diagnosed as congenital heart disease with hypoplastic left heart syndrome or with possible critical pulmonary stenosis.

The patient exhibited characteristic findings for both diseases and therefore an autopsy was ordered to confirm them. Special emphasis was given to the examination of the heart, during which a routine morphometry of the heart was also conducted. During measurements the size of the heart was as follows: the maximal length from the base to the apex was 56 mm, the width – 39 mm and the thickness – 28 mm. The distance between the coronary groove and the apex was

36 mm and between the groove and the highest point of atrial wall – 20 mm, length and width of the right auricle were 17x13 mm and similar dimensions for the left one – 12x9 mm. The weight of the dry heart with its great vessels was 21.96 g. The right border of the heart was slightly acute, whereas the left border was obtuse and rounded. While both the right and left coronary groves were present and occupied by coronary arteries and accompanying veins, the inter-ventricular groove with its content was observed only on the posterior surface (Figure 1) and the anterior interventricular groove was absent. The anterior interventricular branch of the left coronary artery was very short (12 mm) ending in the upper third of the anterior ventricular wall. The 'left dominance' form of distribution of the coronary arteries was present. The posterior interventricular artery and three posterior ventricular branches were originating from the circumflex branch of the left coronary artery.

The great arteries (Figure 1) at the base of the heart were surrounded by the two auricles, and the aortic arch had its usual three branches (brachiocephalic trunk, left common carotid and left subclavian arteries). The pulmonary trunk was dividing into the right and left pulmonary arteries. The site of drainage of the caval veins into the heart was atypical in this case. It was shifted to the left side of the posterior atrial wall for about 20 mm from the cardiac crux. The opening of the inferior vena cava was very close to the superior vena cava opening and to the posterior part of the left coronary groove. The

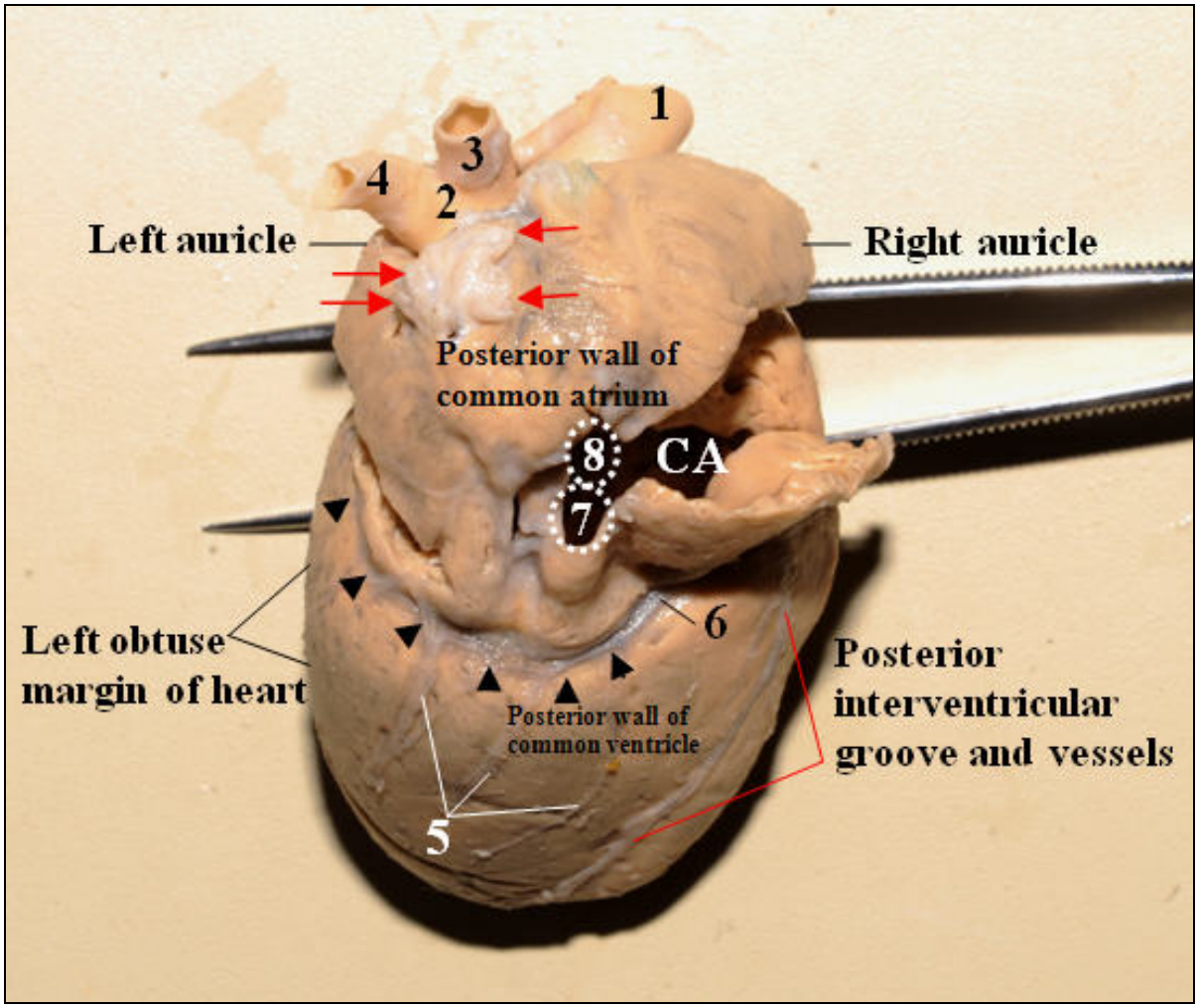

Figure 1- Heart of newborn infant showing great vessels. Four pulmonary veins (red arrows) and two caval veins (circular dash lines) open into the common atrium. (Posterior view). 1-arch of aorta, 2-pulmonary trunk, 3-right pulmonary artery, 4-left pulmonary artery, 5-posterior vessels of common ventricle, 6-coronary sinus, 7-opening of inferior vena cava, 8-opening of superior vena cava, CA-common atrum, Black arrowheads-atrioventricular (coronary) groove.

opening of the four pulmonary veins, which looked underdeveloped, were located more to the left and almost at the top of the atrial wall just behind the upper end of the pulmonary trunk.
For examination of the cardiac chambers an incision was made along the right border of the heart. It was observed (Figure 2) that the interior of the heart contained only one single atrium (common for the right and left atria) and one single ventricle (common for the right and left ventricles). Interatrial and interventricular septa of the heart were not present. The common atrial and ventricular chambers of the heart were interconnected by one single atrioventricular orifice, which had no cusps. The thickness of the common atrial wall was 2-3 mm, whereas the ventricular wall, at the right border of the heart, was 4 mm and at the left border – 5 mm tick. Inside the common ventricle there were two papillary muscles (anterior and posterior) located on the respective walls. Both aorta and pulmonary trunk were arising from the single ventricle (Figure 3), and superior and inferior cava veins and the pulmonary veins were draining into the common atrial chamber. The aortic orifice of the common ventricle had diameter 9 mm and the length of the ascending aorta was 18 mm. The ascending aorta had three

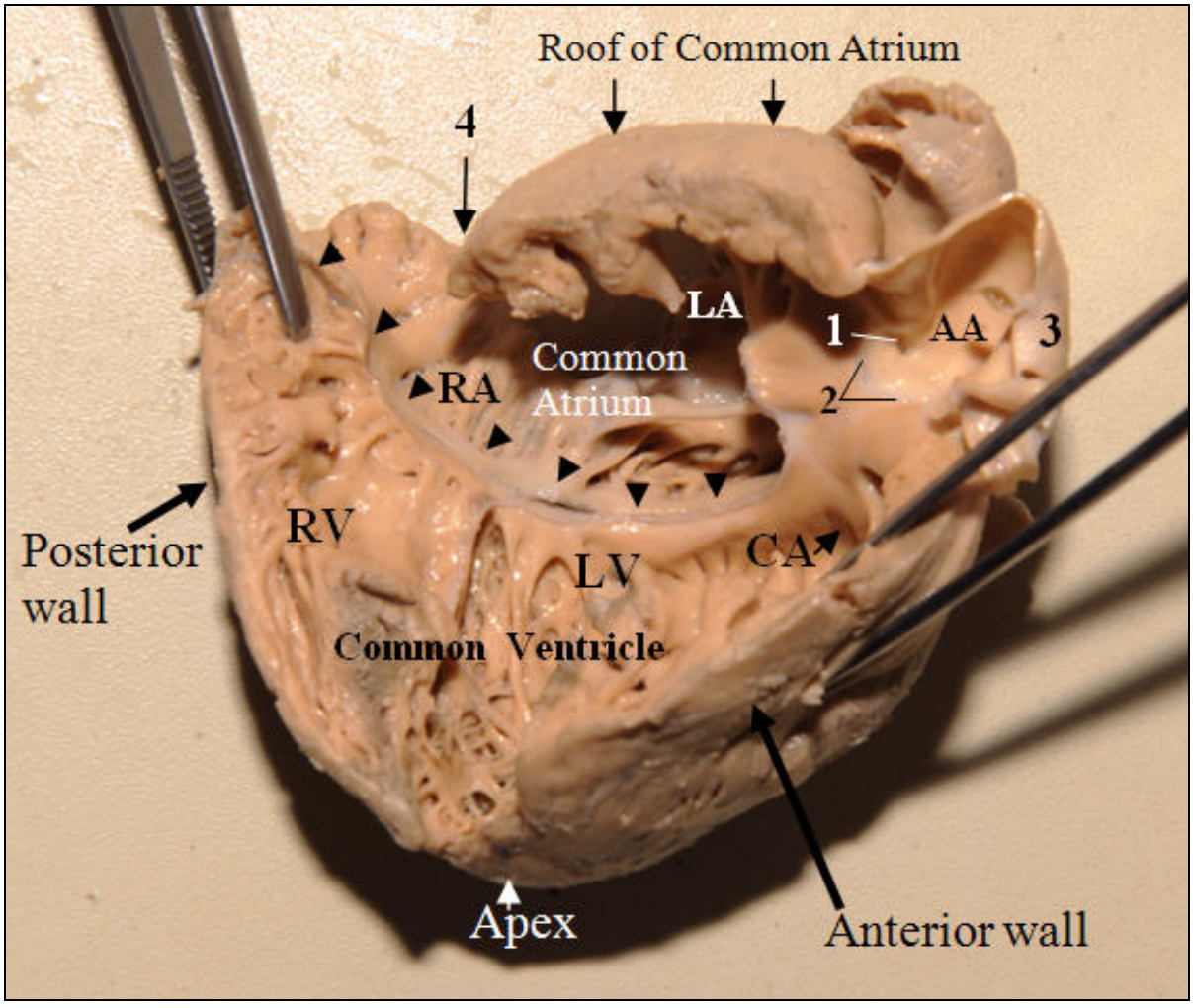

Figure 2- Heart of newborn infant showing common atrial and ventricular chambers, which communicate via common atrioventricular orifice. (Right cardiac border view). Incision was made along right border of the heart and cut sides of the wall of the common ventricle were pulled apart by two forceps. Notice absence of interatrial and interventricular septa. 1-orifice of left coronary artery, 2-base of left aortic sinus (no semilunar cusps are present), 3-arch of aorta, 4-area with openings of superior and inferior vena cava, AA-ascending aorta, CA-conus arteriosus, LA-left part and RA-right part of common atrium, LV-left part and RV-right part of common ventricle, Black arrowheads-line of common atrioventricular ring or orifice (no cusps are present).

aortic sinuses. There were not any semilunar cusps at the bases of these sinuses. The right and the left sinuses had orifices of their respective coronary arteries. The orifice of the right coronary artery was less than 1 mm in diameter, whereas the orifice of the left coronary artery (Figure 1) was almost two times larger (1.6 mm). Incision along the pulmonary trunk showed its orifice in the common ventricle measuring 6 mm. There were no semilunar cusps present at the bases of the pulmonary sinuses. The length of the pulmonary trunk from its orifice to the bifurcation was 9 mm. The diameter of each pulmonary artery was 3 mm.

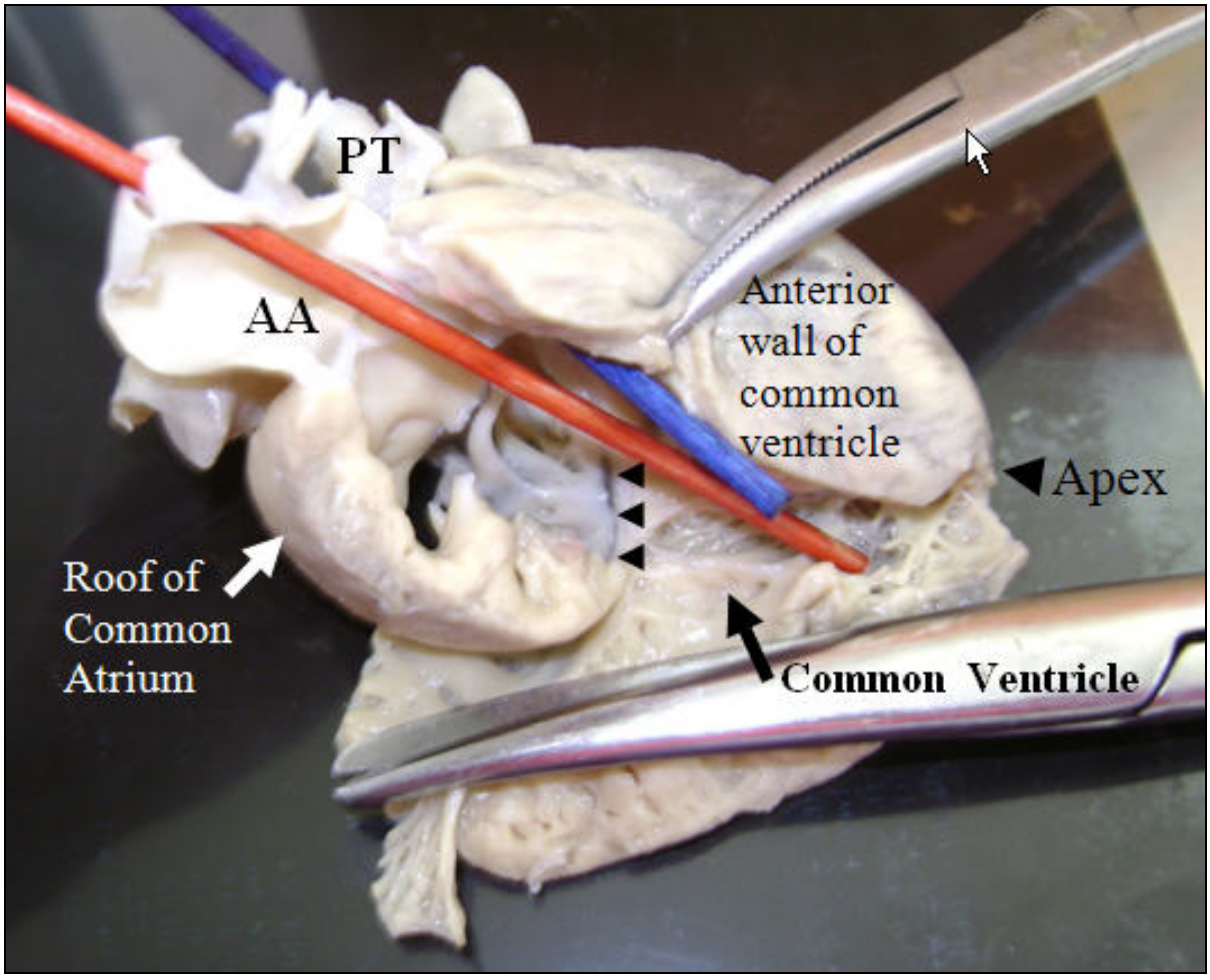

Figure 3- Heart of newborn infant showing ascending aorta (red probe) and pulmonary trunk (blue probe) arising from common ventricular chamber. (Right oblique view). Incision was made along right border of the heart and cut sides of the wall of the common ventricle were pulled apart. AA-ascending aorta, PT-pulmonary trunk, black arrowhead-line of common atrioventricular orifice.

DISCUSSION

Atrioventricular septal defect (AVSD) is a lesion where the atrial and ventricular separation is not complete and the separation of the mitral and tricuspid orifices does not occur. Instead there is a common atrio-ventricular junction. These lesions can be found as a spectrum of anomalies, ranging from a complete form to a partial form, or incomplete form. All forms involve an intrinsic

abnormality of the atrio-ventricular valves. AVSDs can be classified as being balanced or unbalanced. In the balanced form the atrio-ventricular junction is connected to the right and left ventricle so that blood flows equally into each ventricle. In the unbalanced form the atrio-ventricular orifice is predominantly connected to one of the ventricles, usually leading to hypoplasia of the ventricle receiving the smaller portion of the orifice (Allan, 1999).

Barry et al (1979) observed a double inlet atrio-ventricular connection to a chamber of right ventricular pattern. Variations in the mode of connection were observed. The most common variations entailed two atrio-ventricular valves or common valve communicating entirely with the right ventricular chamber. The morphological characteristics of the main ventricular chamber were noted. A rudimentary chamber was also noted in each case with left ventricular trabecular pattern. This condition can be classed as being an unbalanced AVSD.

Pulmonary stenosis (PS) is a dynamic or fixed anatomic obstruction to flow from the right ventricle to the pulmonary arterial vasculature. PS can cause via isolated valvular, subvalvular, or peripheral obstruction, or it may be found in association with more complicated congenital heart disorders (Hameed et al., 2007).

Premature closure or absence of the foramen ovale represents another theoretical cause of hypoplastic left heart syndrome because it eliminates fetal blood flow from the inferior vena cava to the left atrium. Fetal pulmonary blood flow is not sufficient for normal development of the left atrium, left ventricle, and ascending aorta (Lev et al., 1963).

The hypoplastic left heart syndrome is generally postulated to follow multifactorial mode of inheritance. The primary abnormality occurs during aortic and mitral valve development. During cardiac development, adequate flow of blood through a structure is largely responsible for the growth of that structure. With little or no blood flow because of aortic and mitral valve atresia, growth of the left ventricle does not occur. Similarly, growth of the ascending aorta does not occur because of lack of left ventricular output. The ascending aorta is perfused in retrograde manner from the ductus arteriosus functioning only as a common coronary artery (Nora, 1968).

Earlier studies found that congenital heart diseases (CHD) are the most common of all birth defects and are the leading cause of mortality in the 1st year of life. A correlation was observed between the prevalence of cases of Down syndrome, Turner syndrome, Edwards syndrome, Ellis-van Creveld syndrome and mutations in genes TBX5, GATA4 with respect to the prevalence of Hypoplastic left heart syndrome (HLHS) and Atrio-ventricular septal defects (AVSD) (Ramegowda and Ramchandra, 2005).

Hypoplastic left heart syndrome (HLHS) is a noted hypoplasia of the left heart structures, with enlargement and hypertrophy of the right heart. HLHS also demonstrates a spectrum of severity. In the most severe form, aortic and mitral valve are atretic, with a diminutive ascending aorta and markedly hypoplastic left ventricle. The left atrium is usually smaller than normal, although it may be normal in size or enlarged; it receives all pulmonary veins with rare complications of pulmonary venous stenosis (Rao et al., 1994).

The cause of AVSD is when the endocardial cushion fibroblasts fail to migrate normally to form the septum of the AVC. As a result, a deficiency of the primum atrial septum, the ventricular septum, the septal leaflet of the tricuspid valve, and the anterior leaflet of the mitral valve occurs. The position of the AVVs becomes lower than normal. The anterior leaflet of the AVV extends across the ventricular septum and is shared between the left and right ventricles. If the leaflet opens preferentially toward one ventricle, blood flow is limited to the other ventricle, causing hypoplasia of that ventricle and creating unbalance between the 2 ventricles (VanPraagh and Litovsky, 1999).

The heart was found on autopsy to have only one single anatomically distinguishable atrium and one single ventricle with common artrioventricular orifice. No interatrial and interventricular septa were present. No cusps were present at atrioventricular and semilunar valves. The common atrial and common ventricular heart chambers displayed hypertrophy. The common ventricular chamber showed right ventricular characteristics and was in direct communication with the aorta and pulmonary trunk. This condition was not found in any of the literature reviewed. This malformation may have been caused by factors responsible for similar conditions such as AVSD and HLHS. The clinical importance of this congenital defect lies in the fact that it is not compatible with life and infants with similar defects such as HLHS and AVSD have a high mortality rate. Treatment by surgical intervention and cardiac transplantation are the only options in similar conditions such as HLHS and AVSD. In cases like this if such defects are detected during intrauterine life the best option may be to interrupt the pregnancy by abortion to avoid complications after birth.

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