SINGLE INLET UNIVENTRICULAR HEART WITH TRUNCUS ARTERIOSUS AND ATRIAL SEPTAL DEFECT

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Case report

SUMMARY
An early neonate with unremarkable antenatal and birth history presented to our hospital on the third postnatal day with cyanosis and features of shock. Differential diagnosis of congenital heart disease and septicaemia were considered. The baby succumbed, despite inotropic and ventilator support, 2½ hours post admission. Necropsy revealed multiple congenital cardiac anomalies comprised of single inlet univentricular heart (UVH), truncus arteriosus (TA) and atrial septal defect (ASD) without heterotaxy or associated anomaly of other organs. Truly [structurally and functionally] Univentricular hearts of indeterminate morphology are exceedingly rare and more commonly a second rudimentary or hypoplastic accessory ventricle is present. Occurrence of UVH together with rare and distinct defect like truncus arteriosus and atrial septal defect which have different embryogenesis is exceptional. UVH and TA are both total mixing lesions with total admixture of pulmonary and systemic venous return and as pulmonary vascular resistance drops in the early neonatal period the pulmonary blood flow, in the setting of unobstructed pulmonary out flow, markedly increases leading to cardiac failure finally culminating in death. Prenatal diagnosis of CHD’s is possible by fetal echocardiography with a high degree of accuracy and termination of pregnancy can be an option for CHD’s associated with a dismal outcome.

Key words: Atrial septal defect, congenital heart defect, truncus arteriosus, univentricular heart.

1. INTRODUCTION
An extremely rare case of an early neonate born with multiple congenital cardiac anomalies comprised of single inlet univentricular heart (UVH), truncus arteriosus (TA) and atrial septal defect (ASD) is presented. UVH and TA are individually uncommon and occur respectively with an incidence of 0.05–0.1 % and 0.5 – 0.9 per 10,000 live births. [1] The simultaneous occurrence of such unique and complex isolated cardiac defects challenges one’s understanding of cardiovascular pathophysiology and hemodynamics and to the best of our knowledge has been rarely reported previously. The neonate’s autopsy findings and cardiovascular pathophysiology are discussed along with a brief review of univentricular heart.

2. CASE REPORT
A baby born vaginally after uncomplicated pregnancy to a primigravida aged 25 years after 3½ years of non consanguineous marriage at 37 weeks of gestation presented to our hospital at 60 hours of life with complaints of poor feeding and lethargy. At admission the baby was cyanosed with features of shock, precordial systolic murmur and soft hepatomegaly. Chest X-ray revealed cardiomegaly. Differential diagnosis of congenital heart disease and septicaemia were considered. The baby succumbed, despite inotropic and ventilator support, 2½ hours post admission. Necropsy revealed a well developed mature, male, and appropriate for gestation age early neonate weighing 2.5 Kg with mild diffuse cyanosis and no external congenital anomaly. Internal examination revealed normal situs of lungs and abdominal organs. The heart was situated in its normal thoracic position, measured 5.6 cm transversely and weighed 24 g. Its external surface revealed absent anterior and posterior interventricular groove and a single arterial trunk replaced the great arteries [Figures 1 A, B]. Segmental sequential analysis of the heart was done. Analysis of the atrial segment revealed atrial situs solitus with no significant abnormality in the venoatrial connections. The superior and inferior caval veins opened into the sinus venarum of the “ morphological “ right atrium that possessed a broad based, blunt ending appendage and the four pulmonary veins opened via a common confluence into the left posteriosuperior part of the relatively small sized “ morphological “left atrium which possessed a tubular appendage [Figure 1 A, B]. The interatrial septum revealed a large (1 cm) ostium secundum atrial septal defect. Analysis of the atroventricular (AV) connections revealed a single AV valve (4.9 cm in circumference) connecting the right atrium to the ventricular mass [Figure 2A]. The left atrium had a completely muscular floor without even a rudiment of AV junction. The valve possessed 3 cusps, one originating from the diaphragmatic parietal ventricular wall and two originating...
ing from sternocostal aspect, which were anchored by chordae originating from multiple poorly delineated papillary muscles. Analysis of the ventricular mass revealed a solitary and indeterminate ventricle with a single coarsely trabeculated apical component, and rough inner surface with coarse ridges and thick muscle bundles (bridges) percolating throughout the ventricular cavity. Extensive examination failed to reveal any other rudimentary chamber [Figure 2B]. Examination of the ventriculo arterial junction revealed a single arterial trunk (1.1 cm in diameter), originating from the anteriorly placed outlet of the solitary ventricle, which gave rise to the pulmonary and coronary arteries and continued as the aorta giving off innominate, left carotid and left subclavian arteries [Figure 1A, 2A]. The trunk was guarded by a nonstenotic four cusp valve and the pulmonary arteries, each measuring 0.6 cm in diameter arose from separate orifices over its posterior aspect. There was no evidence of other congenital anomalies. An anatomic diagnosis of single inlet univentricular heart (UVH) with truncus arteriosus [Collett and Edward’s type II] (TA) and atrial septal defect (ASD) was made.

3. DISCUSSION

Congenital heart disease (CHD) occurs in 0.5 to 0.8 % of live births and its diagnosis is established by 1 week of age in 40 – 50 % of patients. [2] The most frequent "missed CHD’s" that lead to death or readmissions during the 1st 2 weeks of life are Coarctation of aorta and Hypoplastic left heart syndrome. [3] UVH, comprising 1 % of all CHD, denotes a wide variety of rare and complex congenital cardiac malformations whereby both atria predominantly egress into a functional single ventricle. [1,4] This definition encompasses i) Double inlet AV connections (double inlet left or right ventricle) ii) Absence of one AV connection[Mitral or Tricuspid atresia] iii) Common AV valve and only one well developed ventricle(Unbalanced AV canal defect) and iv) Only one well developed ventricle and heterotaxy syndrome. The clinical picture and long term outlook depend on associated intracardiac anomalies (pulmonary and aortic outflow obstruction and AV valve insufficiency) and morphology and function of the ventricle.[4] UVH’s are hard to diagnose with certainty and ventricular morphology is an important feature that has to be considered for diagnosis. The working definition of a ventricle is any chamber within the ventricular mass that has an apical component, which is coarsely trabeculated in the “morphological” right ventricle and fine in the “morphological” left ventricle.[5] Thus based on the morphology of apical component UVH possesses any of the three types of ventricular morphology – 60-66 % have a main chamber of left ventricular type (with fine apical trabeculation, relatively smooth walls and lack of septal chordal attachments of the AV valve) and a rudimentary right ventricle, which is anteriorly placed and either right or left sided ; 10-24 % have a main chamber of right ventricular type (with coarse apical component and chordal attachments of the AV valve to the septal surface) and a rudimentary left ventricle, which is posteriorly placed and either left or right sided ; < 10% are truly (structurally and functionally) solitary ventricles of indeterminate morphology with a single chamber exhibiting coarse apical component and thick muscle bundles percolating the cavity, as in the present case. [4, 5, 6, 7] The latter

![Figure 1: (A) Sternocostal surface of heart with absent anterior interventricular groove and a single arterial trunk emanating from the ventricle, (B) Diaphragmatic surface of heart with absent posterior interventricular groove. Aorta; BCT Brachio-cephalic Trunk, IVC Inferior Vena Cava; LAA left atrial appendage; LCC Left Common Carotid; LPA Left Pulmonary Artery; LSC Left Subclavian Artery; OPV Opening of Pulmonary veins; RAA Right atrial appendage; RCC Right Common Carotid; RPA Right Pulmonary Artery; RSA Right Subclavian Artery; SV Single Ventricle; SVC Superior Vena Cava; TA Truncus Arteriosus.](image)

![Figure 2: (A) Cut surface showing atrial segment that connects with the single ventricle via a single atrioventricular valve with 3 cusps, (B) Cut surface showing hypertrophied bridges, ridges and papillary muscles. ASD Atrial septal defect; AV Atrioventricular valve; B Bridges; LA Left atrium; RA Right atrium; P Papillary muscles; R Ridges.](image)
has to be distinguished from hearts with huge ventricular septal defect in which a remnant of the apical muscular septum separates the apical trabecular components of the morphologically right and left ventricles. [5] Hearts with solitary and indeterminate ventricles usually have double inlets and either a double or single outlet. Single inlet with absence of one AV valve, as in the present case is unusual. UVH and TA are both total mixing lesions with total admixture of pulmonary and systemic venous return and a degree of hypoxemia is present in all cases. Despite the single ventricle ejecting blood at systemic pressure into TA, the pulmonary blood flow is normal at birth because of the relatively high pulmonary vascular resistance (PVR). However as PVR drops in the early neonatal period the pulmonary blood flow, in the setting of unobstructed pulmonary outflow, markedly increases leading to tachypnea, dyspnea, failure to thrive and cardiac failure ensues. Further, in the present case, the increased systemic load produced by effect of feeding and hypoxemia could have exacerbated the cardiac failure precipitating the vicious cycle of cardiogenic shock which inevitably ended in death. Theoretically, TA with UVH is amenable to surgical palliation. After initial successful separation of the pulmonary arteries from the TA and insertion of a controlled systemic-pulmonary artery shunt, a staged reconstructive surgical approach comprised of Fontan completion operation may be required. [8] UVH may be associated with subvalvular or valvular outflow tract obstruction to great artery, hypoplastic ascending aorta, coarctation, anomalies of the atroventricular valves and heterotaxy syndromes. [1] The present case is a unique case in which structurally and functionally univentricular heart was associated with TA and ASD, without heterotaxy or associated anomaly of other organs. The embryology of the above mentioned unusual developmental defects are different and the concurrence of such anomalies within the same patient is the result of defective development of multiple distinct segments of the embryonic heart, which is exceptional and explains the extreme rarity of the condition. Prenatal diagnosis of CHD’s is possible by fetal echocardiography with a high degree of accuracy. [9] It has a great impact on management of fetal and neonatal cardiac diseases and termination of pregnancy can be an option for CHD’s associated with a dismal outcome. The above case emphasises the importance of making fetal cardiac examination a routine part of perinatal ultrasonographic examination.

Conflict of interest: none declared.

REFERENCES