Case Report

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A complex case of univentricular heart with multiple congenital malformations diagnosed in a newborn: a case report and literature review

Huda Hossin[a,*], Mouhammed Sleiay[a], Joudi Kouran[c], Raghad Mahmoud Alsuliman[d], MHD Anas Kojok[c], Nour Mohamad Najeb Alkhateb[a], Suha Giselle Ghanem[e], Marah Mansour, MD[f,g], Tamim Alsuliman, MD[h], Saleh Takkem, MD[b]

Introduction: Univentricular heart disease is a relatively rare condition that affects infants, with a prevalence ranging from 0.05 to 0.1 per 1000 live births. It is characterized by an abnormality in the structure of the heart, specifically the presence of only one main pumping chamber (ventricle) instead of the usual two.

Presentation of case: In this particular case, a newborn male was diagnosed with double-inlet left ventricle (DILV), a specific form of univentricular heart disease. Following his birth, he exhibited symptoms of central cyanosis (a bluish tint to the skin due to poor oxygenation) and difficulties with breastfeeding. Clinical evaluation, along with a heart ultrasound, confirmed the need for palliative surgery. At the age of 6 months, the patient is scheduled to undergo the Glenn procedure, a surgical intervention that aims to redirect blood flow to the lungs and improve oxygenation.

Clinical discussion: Given the complexity of double-inlet single ventricle anomalies, there are multiple differential diagnoses that need to be considered for accurate diagnosis, including conditions such as tricuspid atresia, large ventricular septal defect and corrected transposition of the great arteries with ventricular septal defect.

Conclusion: Early intervention in the immediate postnatal period plays a crucial role in improving survival rates and reducing long-term complications. It is, therefore, essential to continue researching and refining treatment approaches.

Keywords: Blalock-Taussig shunt, case report, congenital heart disease, Glenn Shunt, single ventricle

Introduction

Double-inlet left ventricle (DILV) is a relatively uncommon condition with a prevalence ranging from 0.05 to 0.1 per 1000 live births. It constitutes ~1% of all congenital cardiac anomalies and is observed in approximately 4% of neonates diagnosed with congenital cardiac disease (CHD)[1]. Echocardiography and cardiovascular magnetic resonance (CMR) are the established imaging techniques used for assessing single ventricle function in patients with Fontan circulation[2]. Prenatal diagnosis of patients with single ventricle (SV) has an important role in obtaining better results, as it helps to improve the clinical condition prior to surgery and to reduce consequences[3]. The therapeutic options for cyanotic heart defects are staged palliation [the Blalock-Taussig shunt (BTS)], superior Cavopulmonary anastomosis, and Fontan anastomosis) and heart transplantation. Stem cell therapy may present a hopeful treatment and is expected to achieve progress in treating some congenital heart diseases[4,5]. Unfortunately, the number of infants with CHD who are at risk for heart failure is increasing; however, advances in diagnostic methods, surgical procedures, and postoperative care for newborns with heart structural abnormalities may give hope to 90% of these infants to live for many years[6,7]. This case study aims to display a real-life scenario involving treatment options for infants with DILV.

HIGHLIGHTS

- Double-inlet left ventricle (DILV) is a relatively uncommon condition with a prevalence ranging from 0.05 to 0.1 per 1000 live births.
- Prenatal diagnosis of patients with single ventricle (SV) has an important role in obtaining better results as it helps to improve the clinical condition prior to surgery and to reduce consequences.
- Making procedures in the early postnatal period may have an Important role on survival and morbidity.

[a]Faculty of Medicine, [b]Department of Cardiology Department, Alwatani Hospital, Hama University, Hama, [c]Faculty of Medicine, Damascus University, [d]Faculty of Medicine, Syrian Private University, Damascus, [e]Faculty of Medicine, Al Andalus University for Medical Sciences, [f]Faculty of Medicine, Tartous University, Tartous, [g]Division of Colon and Rectal Surgery, Department of Surgery, Mayo Clinic, Rochester, MN and [h]Hematology and Cell Therapy Department, Saint-Antoine Hospital, AP-HP Sorbonne University, Paris, France

H.H., J.K., R.M.A., MHD.A.K., M.S., N.M.N.A., S.G.G., M.M. are co-first authors.

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*Corresponding author. Address: Faculty of Medicine, Hama University, Hama, Syrian Arab Republic. E-mail: hudoosh238439hossin@gmail.com (H. Hossin).

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Case presentation

A few hours old newborn male was admitted to the Department of Pediatrics by their parents with a complaint of central cyanosis and poor breastfeeding several hours after birth. Upon clinical examination, a systolic apex murmur 2/6 was heard. The chest X-ray showed cardiomegaly, and the cardiac ultrasound revealed dextrocardia with a double-inlet single ventricle (left), and the great arteries are parallel. The aorta is anterior, and the pulmonary arteries are posterior and very small with an atretic pulmonary valve. There is a small PDA that is filling the pulmonary arteries. The patient also has a right aortic arch and a stretched patent foramen ovale (PFO). Laboratory test results showed haemoglobin at (11 g/dl), leucocytes (11 000 mcl), c-reactive protein (CRP) (11 mg/dl), blood glucose (70 mg/dl), and creatinine (0.6 mg/dl). The clinical condition of the newborn and the heart ultrasound required palliative surgery (BTS), which is an artificial connection consisting of a left subclavian artery to the left pulmonary artery. The original pulmonary artery measured 5mm, the left artery was 3.5mm, and the right artery was 4 mm. This is a surgical procedure that is intended to be temporary. It aims to enlarge the pulmonary artery and reduce cyanosis, both by increasing pulmonary flow. The patient is under preparation for the Glenn procedure at the age of 6 months to improve pulmonary flow.

Discussion

Congenital heart disease (CHD) is a heart structural abnormality that arises during foetal development[8]. Globally, the incidence rate of CHD is 8.22 per 1000 individuals, with a growth rate of 10% every 5 years[7]. The prevalence of CHD in the US is estimated at 500 000 cases, with an incidence of 1 in 100 in children and adolescents with a genetic or chromosomal basis.

Maternal viral infections during the first trimester of pregnancy, maternal drug use, and excessive alcohol consumption during pregnancy are factors that improve this disease[8]. CHD has been linked to an increased risk for these defects starting in the embryonic period, a higher incidence of congenital and acquired brain lesions, and delayed neural development. Single or dual ventricle abnormalities are possible manifestations of CHD. Compared to individuals with biventricular CHD, those with single ventricular CHD had a worse prognosis in paediatric patients[7].

Furthermore, CHD may be present even in the absence of specific symptoms such as dyspnoea, reduced exercise tolerance, exhaustion, heart murmur, rapid breathing, breathing difficulties, bluish discoloration of the skin, or low blood pressure, when circulation and oxygenation are inadequate[8–10]. With very complex congenital heart defects, the neonate only presented with cyanosis and weak breastfeeding several hours after birth. Also, a 2/6 systolic murmur was heard at the apex. The blood test results showed elevated levels of CRP. Some studies suggest a correlation between high-level CRP and CHD[11,12].

Due to the complex nature of these anomalies, it is suspected that there is more than one differential diagnosis for double-inlet single ventricle, like a large ventricular septal defect, corrected transposition of the great arteries with a ventricular septal defect, atrioventricular canal, complete transposition of the great arteries with a ventricular septal defect, double outlet right ventricle, tricuspid atresia, and single function ventricle secondary[13,14]. CHD diagnosis is made through a variety of techniques and is detected using an echocardiogram or transesophageal echocardiogram, chest X-ray (CXR), cardiac catheterization, electrocardiography, and MRI methods[8]. According to our case, the CXR and three-dimensional (3D) echocardiogram were enough to show these very complex defects (Figs. 1–4).

The complications of this disease may be extremely serious and include oesophageal varices, arrhythmias, long-term cyanosis, heart failure, renal dysfunction, and pulmonary hypertension[15–18]. The difficulty of "single ventricle" anatomy and physiology in this example, as well as the many problems after palliative treatments, are both illustrative. If a patient has a single ventricle and pulmonary atresia, the only way for deoxygenated blood to reach the lungs is through a patent ductus arteriosus or a systemic aorta that supplies pulmonary collaterals. As a newborn, this patient had an "emergency palliative" classic Blalock-Taussig shunt performed, allowing blood to flow from the aorta to the pulmonary artery for lung oxygenation[19].

Nowadays, a bidirectional Glenn shunt, also known as a superior cavopulmonary shunt, is done at around 6 months of age. Between the ages of 18 months and 4 years, Fontan surgery is performed to separate the pulmonary from the systemic circulations[20]. The child is currently programmed for surgery with the Glenn shunt, while also removing Blalock-Taussing shunt. In 1971, Fontan and Baudet presented a cutting-edge surgical technique for treating tricuspid atresia. They were either the systemic vein-to-pulmonary artery anastomoses that Glenn and Pato first documented in 1954 or the systemic artery-to-pulmonary artery anastomoses that Blalock and Taussig first described.

Fontan and Baudet developed a 5-step procedure that involved a right atrial-to-pulmonary artery anastomosis to achieve a comprehensive cavopulmonary connection. Atrial contraction is not required for proper operation, according to experimental analysis of atrial function in the Fontan circuit. On the basis of this theory, Kawashima and colleagues carried out the first total cavopulmonary shunt, completely excluding the right atrium.

The traditional Fontan procedure required substantial intra-atrial surgery, which led to right atrial distension and pressure elevation. This frequently resulted in sinus node dysfunction and atrial tachyarrhythmias. Compared to patients with an entire cavopulmonary connection excluding the right atrium, which affects 20% of patients, patients with atrio-pulmonary type Fontan experience late atrial arrhythmias in 69% of cases. The most frequent arrhythmia is atrial flutter. Atrial tachyarrhythmias are less frequent as a result of the conversion of an atrio-pulmonary Fontan connection to an extracardiac Cavopulmonary connection[19].

Increased pressure at many locations, including the liver, splanchnic beds, and mesenteric network, results in protein-losing enteropathy (PLE) and encourages protein leakage into the intestinal lumen[21]. Protein C, protein S, and anti-thrombin III levels have all decreased. It has also been shown that platelet reactivity has increased. Cirrhosis and hepatic congestion are prevalent, but hepatic adenoma and hepatocellular cancer are less frequent[20]. With a 3% early postoperative mortality rate, the current surgical success of the Fontan procedure for single ventricles is excellent. At 10 and 20 years, the actuarial rate is 80%

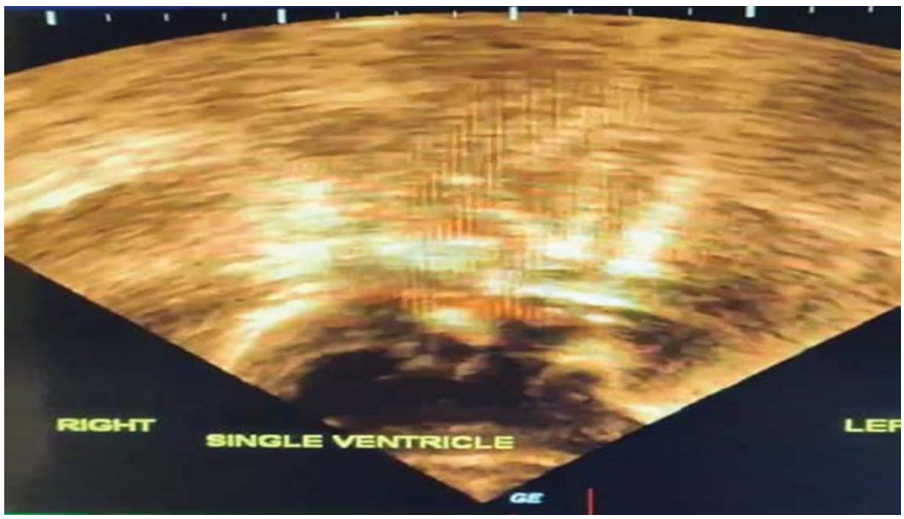

Figure 1. Subcostal view showing the single ventricle.

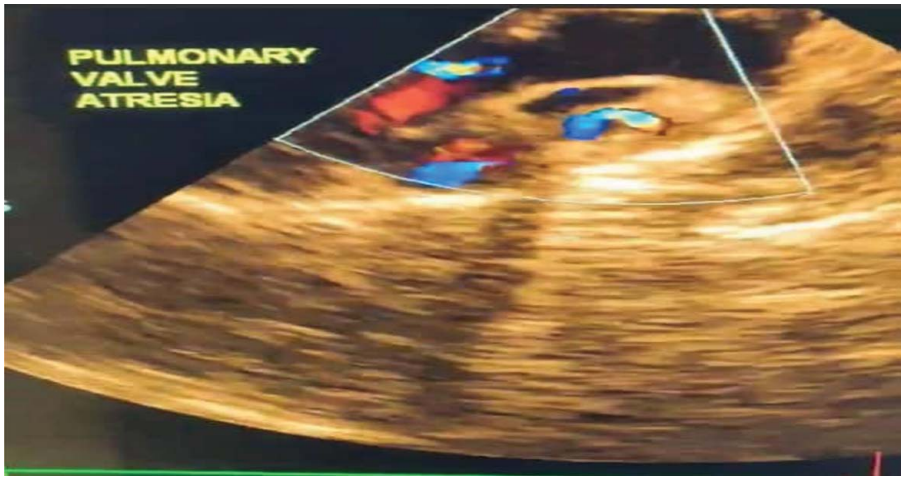

Figure 2. Suprasternal view showing the patent ductus arteriosus and small pulmonary arteries.

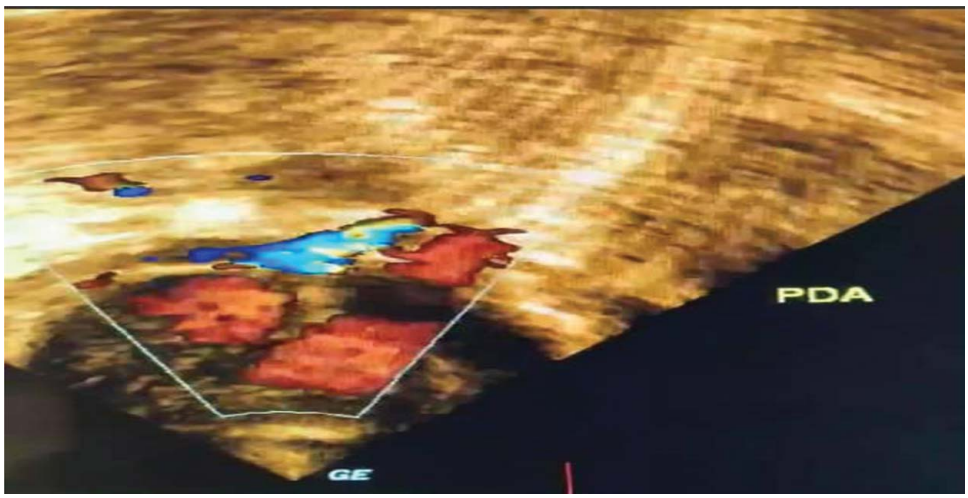

Figure 3. Subcostal view showing the patent ductus arteriosus (PDA).

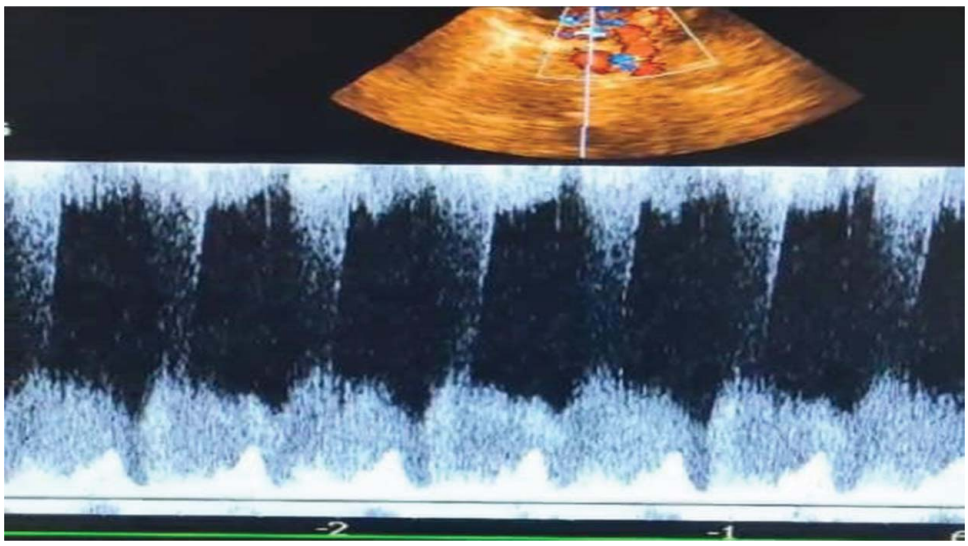

Figure 4. Patent ductus arteriosus flow.

and 69%, respectively; 49% of patients needed another procedure[19].

Most critically, it is still essential that there be little resistance throughout the pulmonary capillary bed. Because excessive Peripheral vascular resistance (PVR) limits cardiac output in patients without a pre-pulmonary pump, and non-pulsatile flow can lead to long-term, poorly adaptive remodelling, excessive PVR is a clear contraindication to a Fontan. Fontan failure risk is further increased by ventricular dysfunction and atrioventricular valve regurgitation. Prior to the Fontan treatment, early atrioventricular valve repair may be advised[22].

DeLevaletal. proposed a significant variation in 1987 that included a prosthetic X patch to channel the inferior vena cava to the transected superior vena cava, a composite intra-atrial tunnel with the right atrial posterior wall, and an end-to-side anastomosis of the superior vena cava to the undivided right pulmonary artery. The superior vena cava and the pulmonary artery are anastomosed, much like in the intracardial lateral tunnel. Moreover, Fontan routes may be "fenestrated" by creating an Atrial septal defect (ASD) in the baffle or patch to act as an escape valve and permit right-to-left shunting, which may be advantageous immediately following the procedure. These fenestrations can eventually be closed via a transcatheter procedure if hemodynamics are favourable. The objectives of the first surgery are to produce unimpeded systemic blood flow, balanced pulmonary and systemic circulations with regulated pulmonary blood flow, and unobstructed pulmonary and systemic venous return (including unrestricted atrial level mixing of venous returns). Relief of any pulmonary venous and systemic outflow tract blockages must also be carried out as part of the first surgical procedure. The precise procedure to achieve the goals will, therefore, depend on the underlying anatomy and may involve the repair of aortic coarctation and aortic arch hypoplasia, the creation of a consistent and controlled source of pulmonary blood flow through a systemic-pulmonary shunt, the limitation of pulmonary blood flow via main pulmonary banding, or a combination of these (including the Norwood procedure)[22].

A variant of the Norwood phase that ends in a Fontan-type circulation is frequently recommended in patients with univentricular hearts and systemic outflow restriction, the most severe manifestation of which is hypoplastic left heart syndrome. Patients with hypoplastic left heart syndrome had no effective surgical alternatives less than three decades ago and often died as newborns. The Norwood stage 1 surgery, carried out within the first two weeks of life, where the primary pulmonary artery is split, the proximal end is anastomosed to the ascending aorta, the aortic arch is repaired and augmented, and the right ventricle is used to sustain pulmonary blood flow by a modified Blalock-Taussig or Gore-Tex shunt.

Variants that are "hybrid" have been described. Before the age of six months, a bidirectional Glenn shunt or hemi-Fontan is done, together with the closure of the Blalock-Taussig shunt. The stage III technique, which connects the inferior vena cava to the pulmonary artery, completes the whole cavopulmonary Fontan between the ages of 18 months and 3 years. Cardiac transplantation is an option if this staged strategy is impractical[20].

Conclusion

In conclusion, it is crucial to acknowledge that even with advancements in surgical techniques and postoperative care, individuals with univentricular heart disease continue to face substantial morbidity and mortality rates. The complex nature of this condition highlights the importance of making informed decisions in the early postnatal period. These decisions not only influence short-term survival rates but also have long-term implications for the individual's overall health and well-being. It becomes especially critical to be proactive in the absence of conclusive studies that can provide concrete guidance. This uncertainty underscores the need for ongoing research and innovation in the field of univentricular heart disease to continually refine treatment approaches and enhance outcomes for these individuals. Only through a comprehensive understanding of this complex condition can

we hope to reduce its burden and improve the quality of life for those affected. Clinical practitioners must consider the presence of complex cardiac anomalies, particularly if the patient presents with central cyanosis hours after birth.

Ethics approval and consent to participate

Not applicable.

Consent for publication

Written informed consent was obtained from the parents of the child for publication of this case report and any accompanying images and videos. A copy of the written consent is available for review by the editor of this journal.

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Author contribution

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Methods

The work has been reported in line with the SCARE criteria[2,3].

Declaration of generative AI and AI-assisted technologies in the writing process

During the preparation of this work the authors used ChatGPT in order to paraphrase some sentences. After using this tool, the authors reviewed and edited the content as needed and take full responsibility for the content of the publication.

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