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Case Report

Fatal cardiac anomaly of unguarded mitral orifice with asplenia syndrome

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ABSTRACT

We report the case of a newborn baby with an unguarded mitral orifice associated with asplenia syndrome, double-outlet right ventricle, dysplastic tricuspid valve, and pulmonary stenosis. This case was accompanied by severe tricuspid regurgitation and severe right ventricular hypertrophy. The patient had a fatal clinical course due to severe hypoxia and congestive heart failure. Unguarded mitral orifice is a rare disease in which there has been no previous report of lethal clinical course during the neonatal period. Prior reports stated that unguarded mitral orifice was a new constellation of defects and that its etiology and embryology could be classified in the same category because of similar associated malformations of double-outlet right ventricle and pulmonary stenosis or atresia. However, the present case was diagnosed on autopsy as also having asplenia syndrome. Therefore, it is possible that the genetic etiology of unguarded mitral orifice in this case was different from cases of non-heterotaxy.
<**Learning objective:** Unguarded mitral orifice is a rare disease that might be associated with asplenia syndrome and dysplastic tricuspid valve. If unguarded mitral orifice is associated with such defects, the clinical course can be fatal. Therefore, when this diagnosis is recognized, the physician should explain the possibility of neonatal death and plan the treatment of such a case to include grief therapy for the family.>

Introduction

Unguarded mitral orifice (UMO) is an extremely rare disease characterized by the complete absence of mitral valve leaflets and tensor apparatus (chordae tendineae and papillary muscles) at the mitral annulus and severe thinning of the left ventricular free wall. Only five cases have been reported in the English literature [1–4]. All cases (except for the most recent case of hypoplastic left heart syndrome) were accompanied by double-outlet right ventricle, atrioventricular discordance, and pulmonary stenosis/atresia [1–3]. All five cases survived through the neonatal period and infancy. The present report describes the case of a patient with UMO who died on the second day of life due to severe heart failure and hypoxia. This patient had UMO, dysplasia of the tricuspid valve, and hypertrophy of the right ventricle, leading to circulatory collapse and death.

Case report

The family of the patient provided permission to publish the features of this case. The identity of the patient has been protected.

The male fetus was referred to Osaka Medical College Hospital at 36 weeks' gestation because a cardiac anomaly was suspected. An ultrasound study interpreted the anomaly as double-outlet right ventricle, hypoplastic right ventricle, pulmonary atresia, and complete atrioventricular septal defect (cAVSD). However, UMO was not acknowledged and so we misinterpreted the diagnosis as cAVSD at that time. Severe common atrioventricular valve regurgitation and cardiomegaly with cardiothoracic area ratio of 56% were observed, so he was delivered at 36 weeks' gestation via emergent Cesarean section with a birth weight of 2398 g. Because

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of severe hypoxia and bradycardia, he was resuscitated by intubation and oxygen inhalation and was admitted to the neonatal intensive care unit.

On admission, blood pressure was 56/29 mmHg, and oxygen saturation was 70% (with an FiO_2 of 100%). A Levine grade II/VI systolic murmur was audible at the right upper sternal border. Blood gas analysis revealed severe hypoxia and lactic acidosis (pH 7.02; lactate 97 mg/dL). Chest radiograph showed a markedly enlarged cardiac silhouette with cardiothoracic ratio of 69% and decreased pulmonary vascular markings.

Postnatal echocardiography revealed ambiguous atrial sidedness and showed that the inferior vena cava and pulmonary vein drained into a left-sided atrium. Both great arteries arose from the right ventricle, and the pulmonary valve and main pulmonary artery were hypoplastic with severe subpulmonary stenosis. Ductus arteriosus was not detected.

A small secundum and a large primum atrial septal defect were present, but the ventricular septum appeared intact. Only a ridge, but no mitral valve leaflets or tensor apparatus were seen at the left atrioventricular junction (Fig. 1A and B). The left-sided morphologic left ventricle appeared dilated and thin-walled, with poor contractility (Fig. 1C). Doppler examination of the left atrioventricular junction revealed free regurgitation (Fig. 1D). Severe regurgitation of a dysplastic right-sided atrioventricular valve was observed (Fig. 1D). The right ventricular wall was markedly hypertrophied, and the right ventricular chamber volume was diminished (Fig. 1C).

Despite 100% oxygen inhalation, nitric oxide inhalation, and prostaglandin E1 infusion, the hypoxia did not improve. Dopamine infusion was initiated because of hypotension. On the first day of life, prostaglandin E1 infusion was discontinued because of its influence on the systemic blood pressure and because of no observed reopening of the arterial duct. After that, the patient's clinical state was relatively stable, with mild desaturation (75% at room air). On the second day of life, the patient collapsed with sudden hypoxia due to muscular subpulmonary stenosis and hypotension. Volume infusion and inotropic support temporarily improved oxygenation and blood pressure. However, metabolic acidosis continued and the baby died despite maximal intensive care support. Post-mortem autopsy was performed.

Autopsy

The thoracic and abdominal organs indicated right isomerism without a spleen. The heart was huge and was located in the midline. From the frontal view, the atrial appendages on both sides

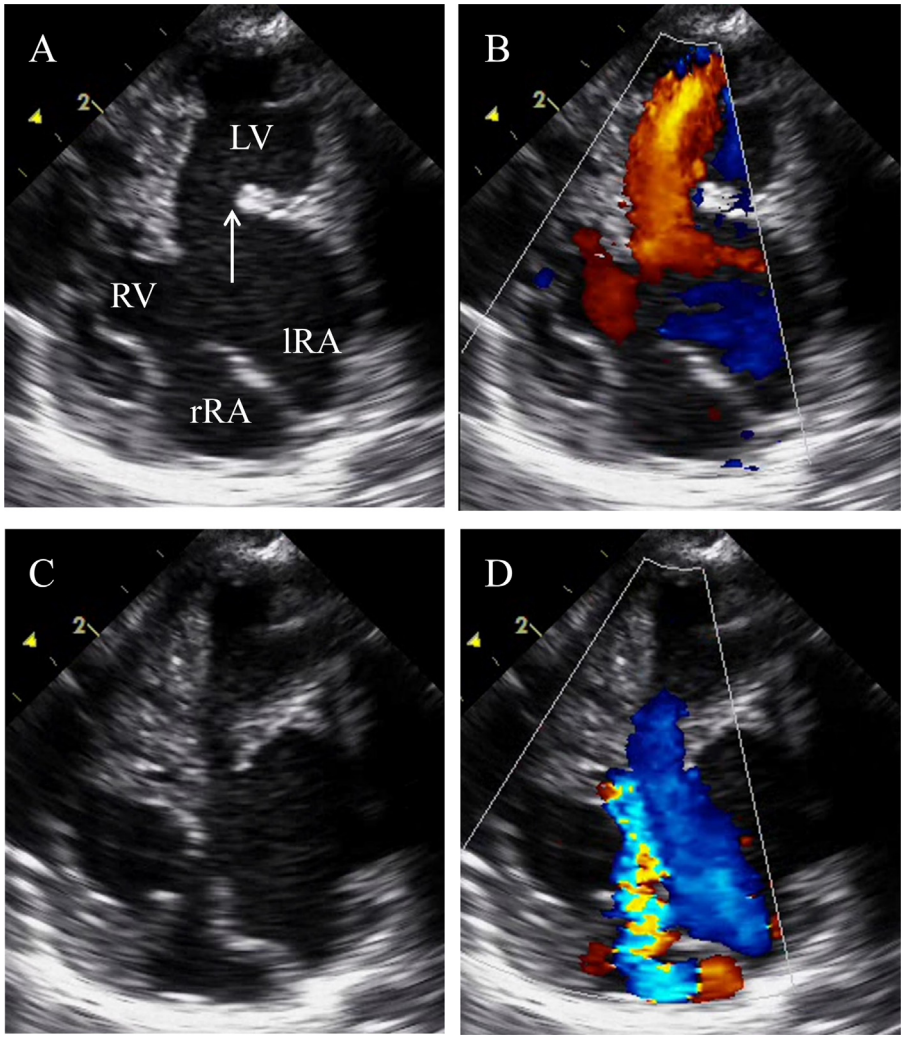

Fig. 1. Echocardiographic findings. Representative four-chamber views. (A) Transthoracic echocardiography demonstrating the unguarded mitral valve orifice (white arrow), thinned left ventricle walls, and a large primum atrial septal defect. (B) Diastolic frame demonstrating color-flow Doppler through the mitral valve and tricuspid valve. (C) and (D) Systolic frame reveals free regurgitation of the unguarded mitral orifice and severe regurgitation of the tricuspid valve. lRA, left-sided morphologic right atrium; rRA, right-sided morphologic right atrium; RV, right ventricle; LV, left ventricle.

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