Prenatal Diagnosis of Pulmonary Atresia with Intact Ventricular Septum

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Introduction:

Hypoplasia of the right ventricle is uncommon as an isolated entity. It may result from tricuspid atresia, but is most commonly secondary to pulmonary atresia with intact ventricular septum. Pulmonary atresia with intact ventricular septum is rare, accounting for 1-3% of cases of congenital heart disease. It occurs in 0.1 to 0.4 in 10,000 live births. Pulmonary atresia with intact ventricular septum has been reported to occur slightly more frequently in males.

Case Report:

A 24-year-old woman (gravida 2, para 1) was referred for an obstetrical ultrasound and fetal echocardiogram. Based on an outside ultrasound, a fetal cardiac anomaly was suspected. At the time of referral, the fetus was 28 weeks gestation by ultrasound; the date of the patient’s last menstrual period was unknown.

Fetal echocardiogram revealed a four-chamber heart correctly oriented in the left chest. The right ventricle appeared small and severely hypertrophied (Figure 1). A small pericardial effusion was also noted. A small pulmonary artery was visualized (Figure 2) with an atretic pulmonic valve. Pulsed Doppler distal to the pulmonic valve confirmed absence of flow. The tricuspid valve was present, but small. A small amount of flow through the tricuspid valve was confirmed by both pulsed and color Doppler. No tricuspid insufficiency was noted. The left atrium and ventricle were enlarged. In addition, the aortic root was significantly enlarged (Figure 3). Pulsed Doppler flow through both the mitral and aortic valves was within normal limits. Flow through the foramen ovale was correctly moving from the right atrium to the left atrium. Retrograde flow through the ductus arteriosus was also appreciated (Figure 4).

Figure 1:
Apical four chamber view of the fetal heart showing a small, hypertrophied right ventricle. A small pericardial effusion is also noted.
Figure 2: Long axis view of the pulmonary artery and it's bifurcation. The diameter of the pulmonary artery was smaller than normal for gestational age.

Figure 3: Long axis view of the dialated aortic root.
The patient denied exposure to illness during this pregnancy or any family history of a congenital cardiac anomaly. She was offered an amniocentesis, but declined. A follow up fetal echocardiogram was performed 4 weeks later which showed the heart to be essentially unchanged from the previous exam.

After delivery at term, prostaglandin was administered to maintain ductal patency, and the in-utero findings were confirmed. At surgery, a pulmonary valvulotomy in conjunction with a systemic-to-pulmonary artery anastomosis was attempted, but the infant did not survive.

**Discussion:**

Embryologically, pulmonary atresia is thought to occur following cardiac septation. It is thought to be a sporadic occurrence, but an autosomal recessive association has been suggested. Pulmonary atresia with intact ventricular septum has also been reported to occur secondary to an inflammatory process, such as Rubella.

Patients with pulmonary atresia and intact ventricular septum usually have a pulmonary valve with three fused cusps. The main pulmonary trunk is of normal or slightly increased caliber in over 50% of
cases. This differs from pulmonary atresia associated with a ventricular septal defect, in which the main pulmonary artery diameter is almost always decreased.

Right ventricular hypoplasia is usually present and is thought to be the result of an inadequate amount of blood entering the ventricle, impeding its growth. The fact that the pulmonary artery and ductus arteriosus are usually of normal caliber would imply that there was normal blood flow through the pulmonary artery at one time. However, another hypothesis would be that the reversal of blood flow through the ductus maintains patency.

Because a normal caliber main pulmonary artery does not exclude the diagnosis of pulmonary atresia, demonstrating reversal of blood flow through the ductus is essential. Retrograde blood flow through the ductus is virtually always present in pulmonary atresia with intact ventricular septum. The tricuspid valve is by definition, patent in all cases of pulmonary atresia with intact ventricular septum, but rarely normal. The tricuspid valve orifice is usually proportional to the size of the right ventricular cavity. The valve leaflets are often dysplastic, which results in varying degrees of insufficiency or stenosis.

A dilated right atrium is another finding commonly associated with this disorder. The degree of dilation is dependent on the amount of tricuspid regurgitation present or the inability of blood to enter the right ventricle.

An enlarged left heart, as was seen in this patient, results from an increased amount of blood being shunted to the left atrium because it cannot enter the right ventricle due to tricuspid stenosis. The foraminal flap may become very redundant, sometimes bulging into the left atrium to such a degree that mitral valve obstruction occurs. The outcome for infants born with pulmonary atresia and intact ventricular septum is poor. They are dependent on a patent ductus arteriosus for survival in the neonatal period. Administration of prostaglandin E1 at birth is essential to maintain patency until surgery can be performed. Surgical treatment will vary, depending on the type of associated cardiovascular anomalies. In those patients with a normal sized right ventricle, pulmonary valvulotomy would be the procedure of choice. If the right ventricle is hypertrophied but with an obvious infundibulum, a systemic-to-pulmonary arterial anastomosis with a pulmonary valvulotomy is performed. A balloon atrial septostomy also may be necessary in some cases. The long-term prognosis may depend, in part, on the size of the right ventricle because mortality rates are higher among those neonates with a severely hypoplastic right ventricle and a small tricuspid valve orifice.

References:
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