

Absent pulmonary valve syndrome diagnosed by fetal echocardiography

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Prenatal echocardiographic diagnosis of heart defects is important because it permits counseling of the parents with regard to prognosis and treatment options and prepares the medical team for the treatment postnatally. A male infant with absent pulmonary valve syndrome diagnosed prenatally at 22 weeks' gestation is reported. This congenital anomaly is characterized by absent or rudimentary pulmonary valve cusps, conoventricular septal defect, and massive dilatation of the pulmonary arteries. Soon after delivery the infant developed cyanosis and respiratory distress. The infant was placed in prone position for the relief of bronchial compression and nasal continuous positive airway pressure was (CPAP) started. Although the clinical status of the infant improved after supportive treatment, he deteriorated acutely and died at the age of five days. Fetal diagnosis remains an integral part of successful management of children with heart disease. Despite the potential benefits of prenatal diagnosis, it is hard to show significant improvement in mortality, especially in severely affected cases.

Key words: fetal echocardiography, prenatal diagnosis, absent pulmonary valve syndrome.

Congenital heart disease (CHD) is the most common form of severe congenital abnormality, with an incidence of 8 per 1,000 live births, and accounts for >20% of perinatal deaths resulting from congenital malformations¹⁻³. Prenatal diagnosis of CHD is important for proper perinatal and neonatal management. Today fetal echocardiography is indicated in high risk pregnancies, but a significant proportion of CHD occurs in fetuses with no definable risk factors. The reported sensitivity of fetal echocardiography has ranged between 70-85% in some series^{1,4}. Conotruncal anomalies, including double outlet right ventricle, tetralogy of Fallot, transposition of the great arteries, absent pulmonary valve syndrome and truncus arteriosus, can be diagnosed by prenatal echocardiography with a high degree of accuracy⁴. Although routine fetal screening by fetal echocardiography in low-risk obstetric populations is not currently the standard of care, standard four chamber view of the fetal heart is recommended as screening tool⁵.

Absent pulmonary valve syndrome (APVS) is characterized by absent or rudimentary pulmonary valve cusps, conoventricular septal defect, and massive dilatation of the pulmonary arteries⁶⁻⁷. Some infants with the severe form of this syndrome die early in the newborn period owing to severe respiratory distress due to compression of the tracheobronchial tree by massively dilated pulmonary arteries. Early diagnosis and supportive care in the newborn period are important for the survival of these severely affected infants.

Case Report

A 21-year-old primigravida pregnant woman was referred to our hospital because of cardiomegaly detected during routine prenatal obstetric ultrasonography performed at 18 weeks' gestation. No risk factors indicating high risk, including diabetes and hypertension, were present. On admission at 22 weeks' gestation, fetal echocardiogram demonstrated cardiac apex on the left side, atrioventricular

and ventriculoarterial concordance with situs solitus, and right ventricular dilatation with left and right ventricular diastolic diameters of 9.8 mm and 13.4 mm, respectively. Inlet ventricular septal defect, agenesis of the pulmonary valve cusps and massive dilatation of pulmonary arteries (13-18 mm) consistent with APVS were detected (Fig. 1). Color and continuous Doppler examination demonstrated acceleration of pulmonary blood flow velocity of 2.5 m/sec and massive pulmonary valve insufficiency (Fig. 2). Fetal heart rate was 127/minute. On prenatal ultrasonography no associated malformation was detected. On

follow-up at 28 weeks' gestation, minimal pericardial effusion and grade 1-2 tricuspid insufficiency were detected. Ventricular systolic function was at the lower limit. At 37 weeks' gestation left and right ventricular diameters were found dilated with enlarged left and right pulmonary arteries (Fig. 3). After each routine follow-up visit the pediatric cardiologist, neonatologist, and obstetrician discussed the clinical status, delivery time and delivery mode of the fetus. The prognosis of the disease and termination of pregnancy were discussed with the parents but they did not accept termination of pregnancy.



Fig. 1. Modified five chamber view of fetal heart at 22 weeks' gestation demonstrating enlarged right ventricle (rv), inlet ventricular septal defect and overriding of aorta to the interventricular septum.



Fig. 3. Aneurysmal dilatation of the pulmonary artery with markedly enlarged main pulmonary artery (pa) bifurcating into left and right pulmonary arteries and dilated right ventricle (rv). The pulmonary valve annulus is stenotic, and no discrete pulmonary valve is detected.

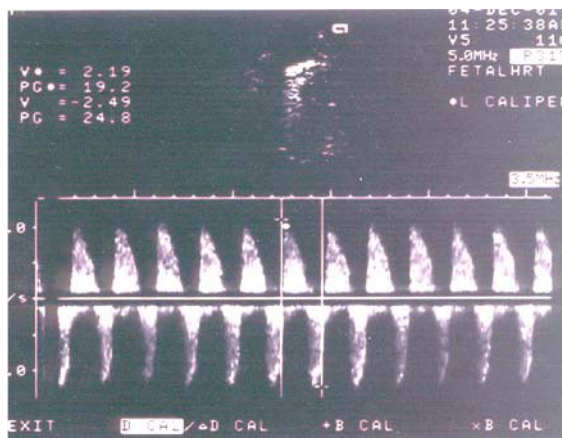


Fig. 2. The continuous wave Doppler signal in the axis of main pulmonary artery showing systolic and diastolic regurgitant jets of approximately 2.19-2.49 m/sec bidirectionally.

A male infant weighing 3,880 g was born at a gestational age of 40 weeks via spontaneous vaginal delivery. A neonatologist and a pediatric cardiologist were ready in the delivery room. Soon after delivery the infant developed cyanosis and respiratory distress. His heart rate and blood pressure on admission were 128/minute and 84/45 mm Hg, respectively. Peripheral circulation was poor, pulses were palpable and grade 4/6 to-and-fro murmur was audible at the left sternal border. The liver was palpable 3 cm below the right costal margin. No morphological anomaly was detected on clinical examination. The chest X-ray showed cardiomegaly with a cardiothoracic ratio of 65%, and marked prominence of the upper left cardiac border due to enlarged pulmonary infundibulum. Right hilus was prominent due

to dilated right pulmonary artery; peripheral pulmonary vascular markings were normal. Arterial blood gases revealed a moderate respiratory acidosis. Serum electrolytes were normal and calcium level was 8.9 mg/dl. Karyotype analysis was normal.

Postnatal echocardiography revealed extensive dilatation of right ventricle and right atrium, inlet ventricular septal defect of 11 mm diameter, and overriding aorta. Pulmonary valve cusps were absent with huge dilatation of the pulmonary arteries. The diameters of right and left pulmonary arteries were 24 mm and 22 mm, respectively. Grade 2-3 pulmonary insufficiency was detected. Left ventricular function was normal.

The infant was immediately taken to the NICU, placed in prone position for the relief of bronchial compression and nasal continuous positive airway pressure (CPAP) was started. After starting CPAP and positioning, the clinical status and arterial blood gases of the infant improved. On day 3 enteral feeding was started and tolerated. However, the patient deteriorated acutely and died at the age of five days. During hospitalization the heart rate and SPO₂ values of the infant varied between 104-158/min and 68-84%, respectively. The latest blood gases revealed normal CO₂ and low O₂ tension.

Discussion

Accurate echocardiographic diagnosis in utero is important because it permits counseling of the parents with regard to prognosis and treatment options and prepares the medical team for the treatment postnatally^{3,8,9}. A detailed cardiac scan can usually be performed in specialized centers with expertise in the diagnosis of CHD. It is important to detect high risk pregnancies and refer them to specialized centers. However, it is known that a significant proportion of CHD occurs in fetuses with no definable risk factors. Perhaps the most important indication for a fetal echocardiography is the suspicion of an abnormal heart on a screening obstetric sonogram. If cardiac screening is confined to the four chamber view, approximately 70% of the cardiac anomalies can be detected¹⁰. If the great arteries are also examined, 90% of major heart disease cases would be detectable prenatally⁹. Ideally, a complete fetal echocardiography is performed at 16 to 20 weeks' gestation because adequate identification of

cardiac structures is generally not possible before that time³. The major benefit of the fetal echocardiography is to be able to provide the family with information regarding the prognosis of the specific defect, so that further management can be planned and coordinated with the multidisciplinary team consisting of a pediatric cardiologist, neonatologist, obstetrician and cardiovascular surgeon.

Absent pulmonary valve syndrome (APVS) is characterized by dysgenesis of the pulmonary valve, severe pulmonary insufficiency, and massive enlargement of pulmonary arteries, and occurs in approximately 3% of cases of tetralogy of Fallot⁶. Tracheobronchomalacia is present due to compression from the massive branch pulmonary arteries. Patients are divided clinically into two groups: neonates with severe respiratory distress, and older children who survived the neonatal period. The patient reported here is in the former group. Neonates in this group typically present soon after birth with severe respiratory distress, cyanosis, and hyperinflation due to tracheobronchial compression^{6,11}. Mechanical ventilation with high positive end-expiratory pressure may help to stent open airways and improve gas exchange. In this patient we did not intubate the infant but rather preferred nasal CPAP, avoiding the risks of intubation and ventilator-related injury. The patient's clinical status improved after supportive treatment. However, despite vigorous, conservative medical treatment the patient deteriorated acutely and died at the age of five days.

Despite the potential benefits of prenatal diagnosis, it is hard to show significant improvement in mortality, partly because fetal echocardiography preferentially detects more severe forms of CHD which have a high mortality rate.

In conclusion, fetal diagnosis remains an integral part of the successful management of children with heart disease. It is clear that early fetal echocardiography is feasible and that complex CHD such as APVS can be detected in early pregnancy. Postnatal management of the severely affected infants with APVS is difficult. Despite prompt and optimal supportive care severely affected infants die early during the newborn period owing to severe respiratory distress, feeding difficulties and cardiovascular compromise.

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