Objective

Aortopulmonary window (APW) is a rare congenital cardiac malformation, which is characterized by a communication between the ascending aorta and the main pulmonary artery before the bifurcation just above the semilunar valves. It accounts for 0.2% to 0.6% of all congenital cardiac defects. Prenatal diagnosis of APW enables early closure of the defect with a low operative mortality and improves the postnatal outcome. We report a case of APW which was diagnosed prenatally. The main purposes of this case report are to discuss the ultrasonographic diagnosis of this rare defect and to demonstrate the importance of the prenatal diagnosis.

Methods

A 23 year-old woman, gravida 2, para 1 was referred to our prenatal diagnosis and treatment unit at 24 weeks of gestation due to the detection of short femur length. Her family history was unremarkable, however first-degree consanguinity was noted between the couple. Ultrasonographic examination revealed micromelia, normal abdomen/thorax ratio and polyhydramnios. Fetal echocardiography demonstrated a normally positioned and sized heart with concordant atrioventricular and ventriculoarterial connection. On the short axis view, a communication between the ascending aorta and the main pulmonary artery was clearly visualized. The defect was just above the semilunar valves and the blood flow from right to left was detected by color Doppler. Cordocentesis was performed with the diagnosis of aortopulmonary window. Fetal karyotype was found to be normal, fluorescence in situ hybridization was negative for chromosome 22q11 microdeletion. Caesarean section was performed due to the development of maternal HELLP syndrome at 27 weeks of gestation and a 770 gram male infant was delivered with apgar scores of 2 and 6 at 1 and 5 minutes. The diagnosis of type I aortopulmonary window was confirmed by postnatal echocardiography with an additional patent foramen ovale. Surgical correction could not be performed due to prematurity and the baby died at the age of 11 day of life due to neonatal pulmonary hypertension and pulmonary hemorrhage. Fibroblast growth factor receptor (FGFR) gene analysis has not been completed.

Results

APW is a rare congenital heart defect caused by defective development of conotruncal ridges. Unlike the other conotruncal defects, association with 22q11 deletion has never been reported. APW may occur as an isolated malformation or in 25% to 35% of cases it may be associated with other structural cardiac anomalies which influence the postnatal management and outcome markedly. The most common associated cardiac anomalies are arch abnormalities, particularly interrupted aortic arch and coarctation of the aorta. The main fetal echocardiographic findings are the visualization of the normally separated semilunar valves and a defect between the ascending aorta and the main pulmonary artery. Also dilatation of the outflow tracts and the presence of pulmonary regurgitation should arise the suspicion of APW. Aortopulmonary septum should be visualized on the short axis view and the diagnosis of APW should be made by demonstrating the both semilunar valves and the defect above the valves between the ascending aorta and the main pulmonary artery. The differential diagnosis mainly includes the truncus arteriosus, which can be ruled out by the visualization of two separate semilunar valves. When pulmonary vascular resistance decrease in the postnatal period, pulmonary blood flow increase due to left-to-right shunt and this shunt causes congestive heart failure and pulmonary hypertension in untreated infants. Early closure of defect should be performed in order to prevent the development of irreversible pulmonary vascular obstructive disease. Without surgical correction, the mortality rate reaches to 40 to 50 % in the first year of life. The prognosis of APW mainly depends on the associated anomalies and the presence of pulmonary vascular diseases.

Conclusion

Prenatal diagnosis of APW allow avoidance of irreversible pulmonary vascular disease and premature death which are the inevitable outcome of the untreated APWs. Many of these risks are eliminated by making the diagnosis prenatally, planning the delivery in a tertiary center and performing the early surgical correction.