MP2-24

Prenatal Diagnosis And Outcome For Fetuses With Congenital Absence of The Pulmonary Valve

Babaoglu K. (1), Uzun O.(2), Deveci M.(1), Kayabey O.(1)
Kocaeli University Medical Faculty Kocaeli Turkey (1); University Hospital of Wales Cardiff Wales UK(2)

Objective: Absent pulmonary valve syndrome (APVS) is a rare congenital anomaly with its hallmark feature of a rudimentary and dysplastic pulmonary valve. In most instances, it is associated with severe dilatation of the main and branch pulmonary arteries (PA) owing to the combination of severe pulmonary stenosis and regurgitation. The aim of this study is to review the spectrum of the prenatally detected APVS and its outcome after diagnosis.
Methods: Clinical data and echocardiographic findings of 11 cases with a fetal diagnosis of APVS between 2008 and 2013 were analyzed in this retrospective two-center study. Collected parameters included: gestational age at referral; associated cardiac, genetic and non-cardiac fetal abnormalities; maximum diameters of the aortic and pulmonary annuli in addition to the main and branch pulmonary arteries.
Results: Median gestational age at diagnosis was 21 weeks. Four subtypes of APVS were observed: (1) with tetralogy of Fallot (TOF) (n=6; 54%); (2) isolated (n = 3; 27,7%); (3) with CAVSD (n=1; 9,9%); and (4) with VSD (n=1; 9,9%). Ductus arteriosus was restricted in 5, absent in 3, and large in 3 fetuses. Two pregnancies were terminated. Two fetuses are still to be delivered. One fetus was stillborn who had trisomy 18. Of the six live births, one neonate died following birth due to severe hypoxia, 3 died after surgery, one remains well after operation, and the last patient is on medical follow up without operation. The presence of ductus arteriosus is not associated with survival. The genetic survey was abnormal in 27,7% of fetuses (trisomy 18 in one, and 22Q11microdeletion in two)
Conclusion: Prenatal diagnosis of the APVS is rather straightforward because of its typical features of a dilated main pulmonary and branch arteries, and color Doppler detection of severe stenosis and insufficiency of the functionally absent pulmonary valve. Outcome of antenatally APVS is poor and survival is mainly determined by the presence of respiratory symptoms due to bronchio-tracheal obstruction in this study.