

Prenatal diagnosis and outcome of fetuses with absent pulmonary valve syndrome

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Objectives: to describe prenatal echocardiographic evaluation and outcome of absent pulmonary valve syndrome (APVS) at a single tertiary care center.

Background: APVS is a rare congenital malformation defined by a rudimentary pulmonary valve with significant regurgitant flow. It is usually associated with tetralogy of Fallot (APV/TOF), microdeletion 22q11 and arterial duct agenesis. APVS in the setting of an intact ventricular septum (APV/IVS) and a patent ductus arteriosus is less common. Reported mortality rates for patients affected is still high.

Method: we included all patients with a prenatal diagnosis of APVs between January 1995 and December 2015.

Results: among 12 fetuses with diagnosis of APVs, 8/12 (66%) had APV/TOF and no arterial duct and 4/12 (33%) had APV/IVS. None presented tricuspid atresia. Median gestational age was 35 and 38.5 weeks, respectively at last echo and at birth. None resulted affected by 22q11 deletion. In the APV/TOF group, all had concurrent severe pulmonary stenosis and significant PAs dilatation. 2/8 (25%) elected for termination of pregnancy and 2/8 (25%) died in the neonatal period for cardiopulmonary arrest. The other 4/8 patients with APV/TOF presented respiratory distress and cyanosis immediately after birth requiring respiratory support but they were all discharged and underwent surgical repair at a median age of 6 months. In the APV/IVS group all presented dilated and hypokinetic right ventricle, significant pulmonary regurgitation but only mild stenosis and PAs of normal size. One of them presented unilateral hydronephrosis. At birth 2/4 neonates with APV/IVS were asymptomatic whereas the other 2/4 required respiratory support for severe cardiorespiratory distress and showed a wide ductus arteriosus with an unusual course and morphology ('aortopulmonary window' like). They both underwent surgical ductal closure and aortic arch reconstruction through a median sternotomy. Overall, among patients with APVS who survived the neonatal period, 7/8 are alive and well and one was lost at a median follow up of 40 months.

Conclusions: outcome after fetal diagnosis of APVS is improving for actively managed patients. In patients with APV/IVS early surgical closure of wide patent ductus arteriosus allows to improve the hemodynamic condition and to delay the homograft valve implantation.

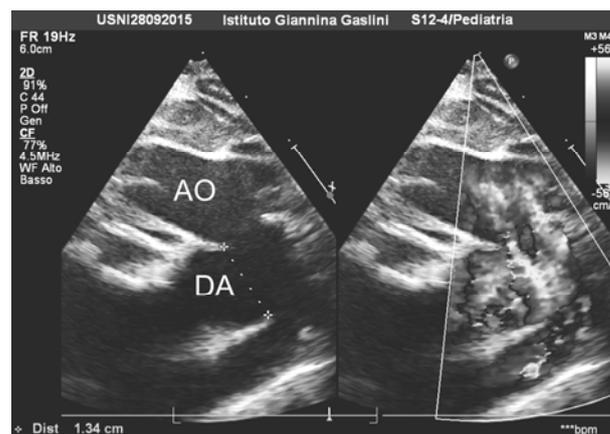


Fig. 1
APV/IVS with wide patent ductus arteriosus (aortopulmonary window-like).
AO: Aorta; DA: Ductus Arteriosus