

# Pulmonary valve agenesis-tetralogy of Fallot-absence of ductus arteriosus syndrome

INSERM

## Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Pulmonary valve agenesis-tetralogy of Fallot-absence of ductus arteriosus syndrome. ORPHA:101206*

Pulmonary valve agenesis-tetralogy of Fallot-absence of ductus arteriosus syndrome is a rare congenital heart malformation characterized by a tetralogy of Fallot (pulmonary stenosis, overriding aorta, ventricular septal defect and right ventricular hypertrophy), complete absence or rudimentary pulmonary valve that is both stenotic and regurgitant and an absence of the ductus arteriosus. It presents prenatally with cardiomegaly, polyhydramnios, fetal heart failure, hydrops fetalis and fetal demise or postnatally with cyanosis and respiratory failure due to bronchomalacia secondary to bronchial compression from dilated pulmonary arteries. It is frequently associated with 22q11 deletion.