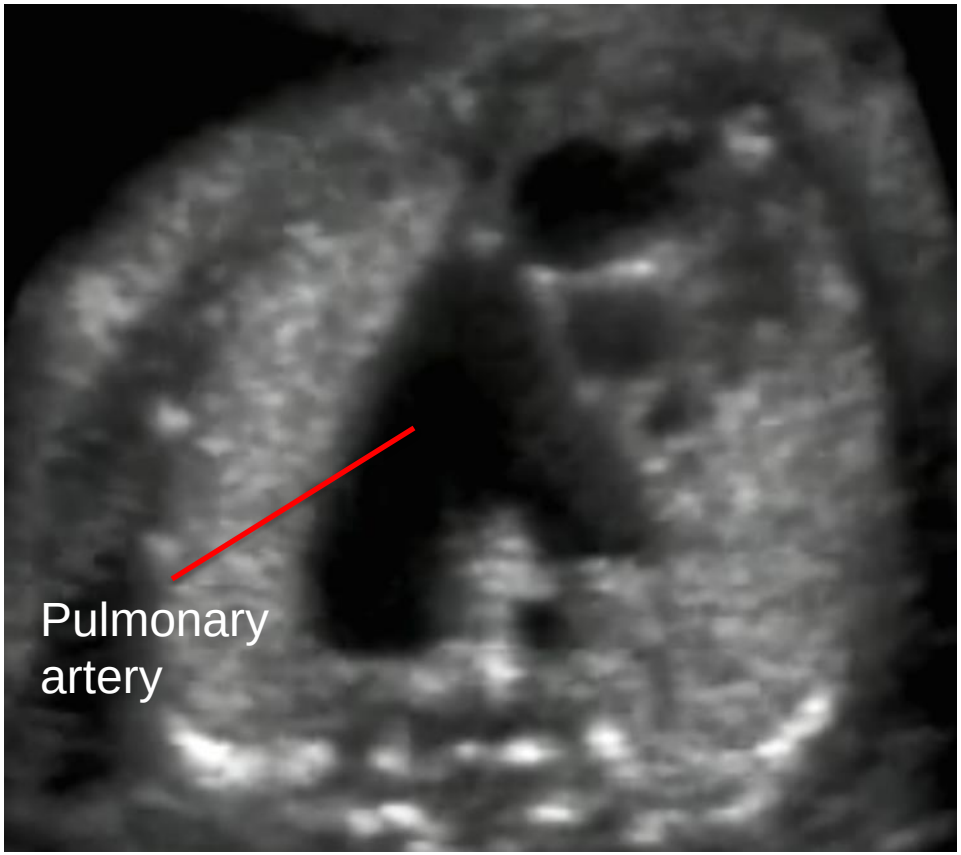
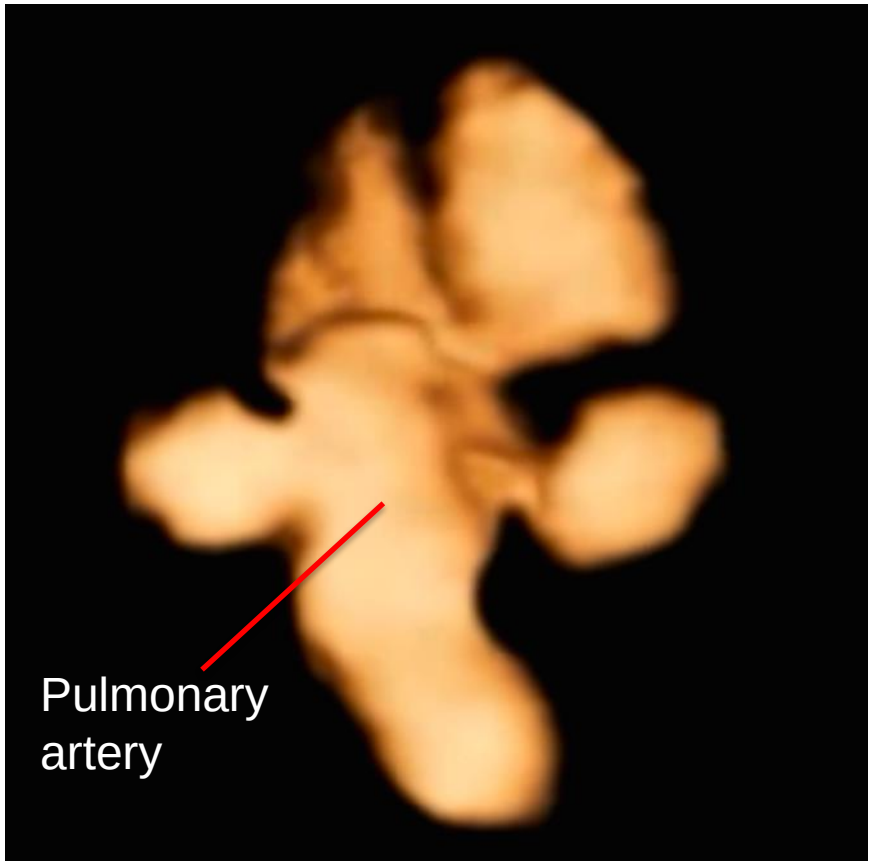
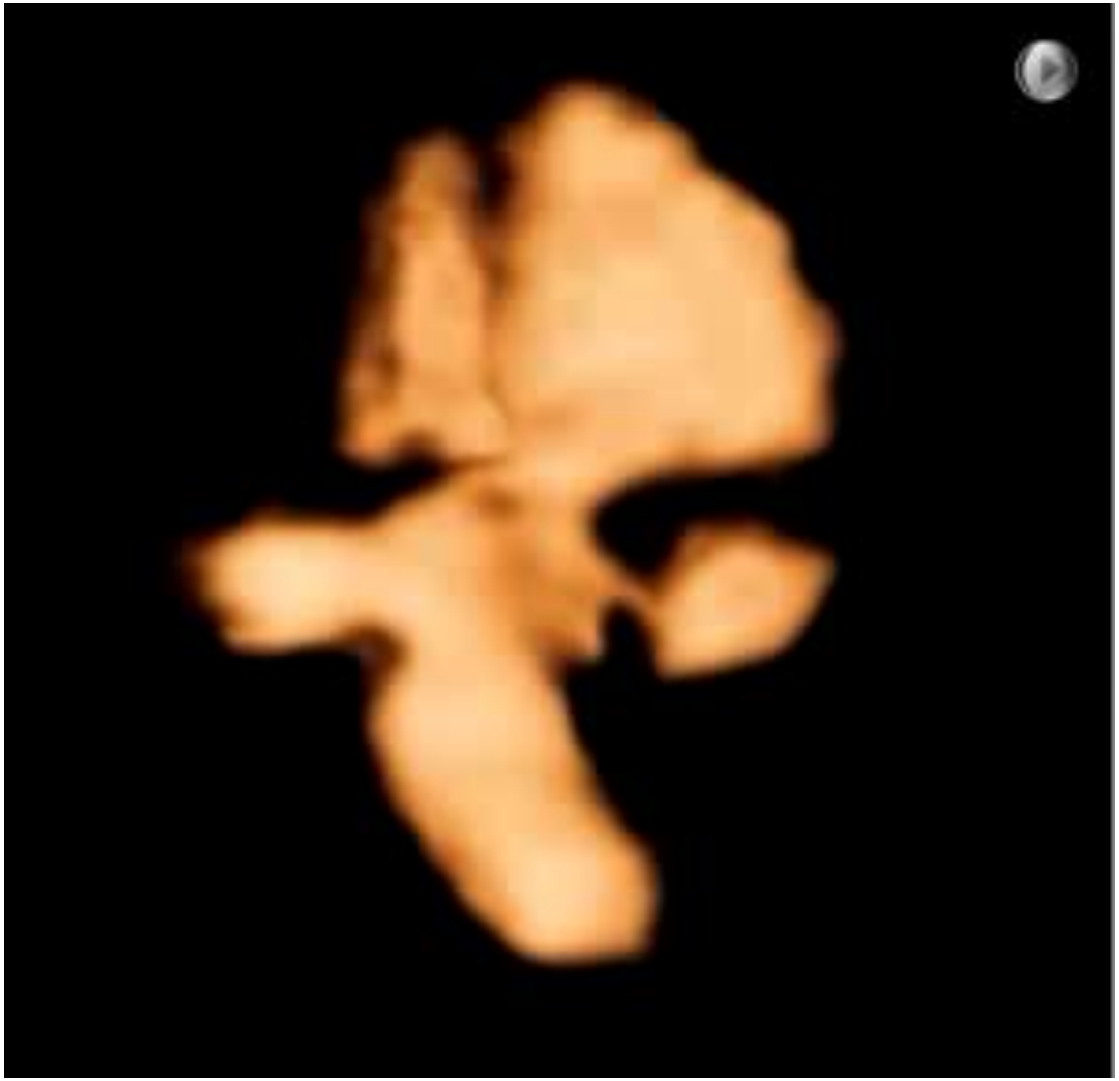
Absent Pulmonary Valve Syndrome





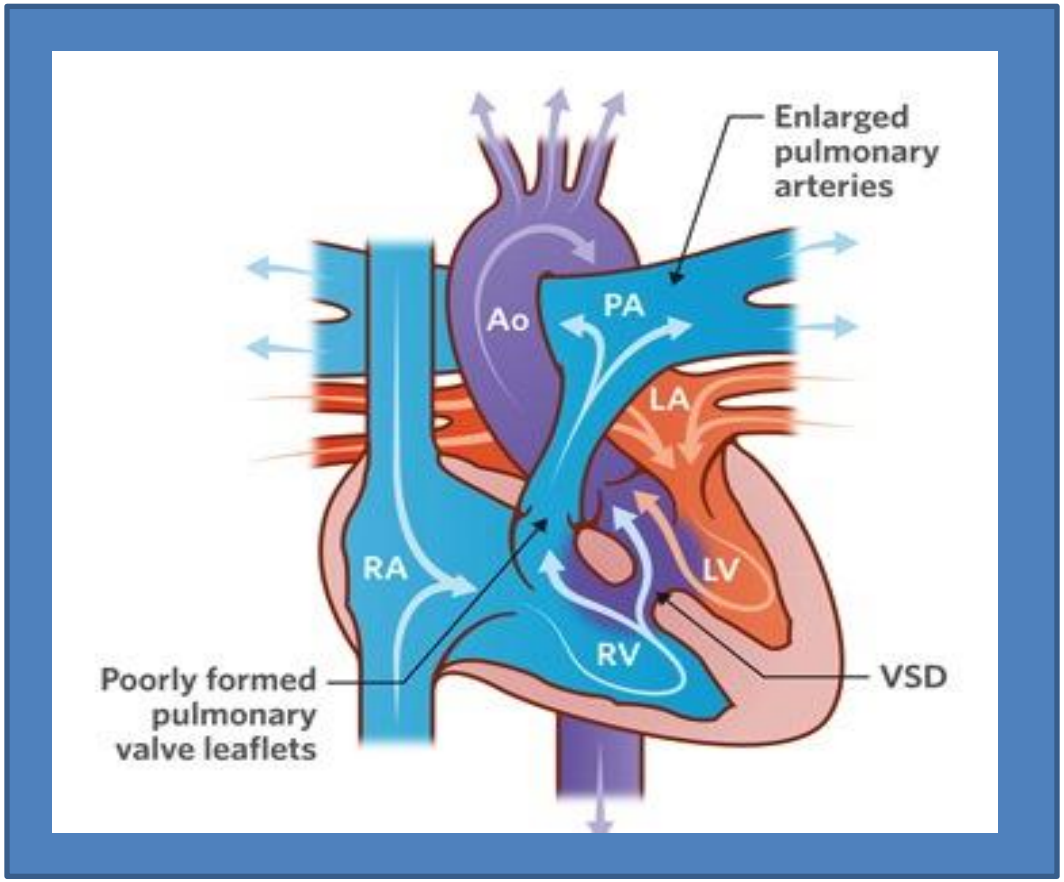
Absent Pulmonary Valve Syndrome (APVS) is a rare cardiac abnormality characterized by the existence of a dysplastic or rudimentary pulmonary valve, associated in most instances with a severe dilation of the pulmonary trunk and branches.

Images by P.Volpe



Clip 01

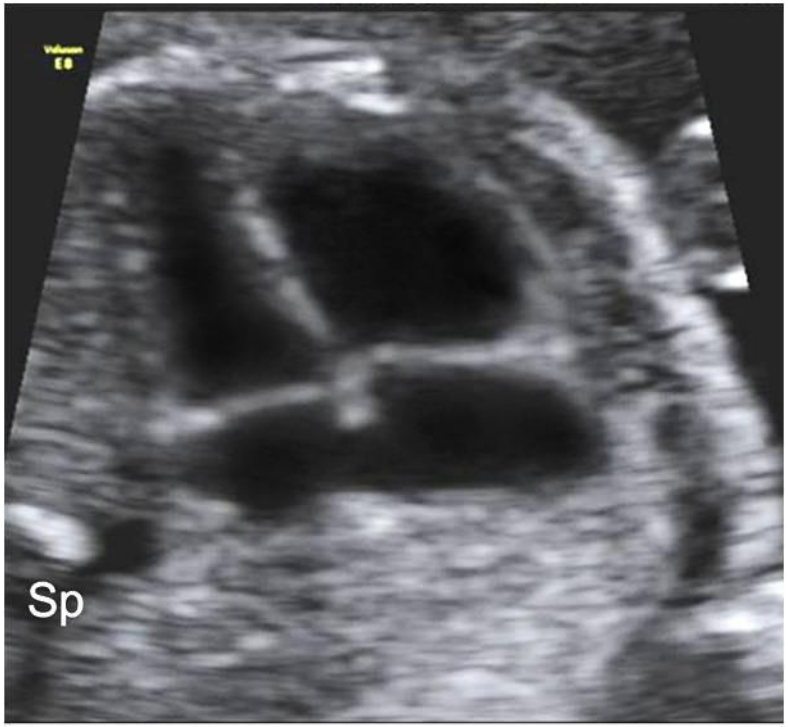
Associated anomalies

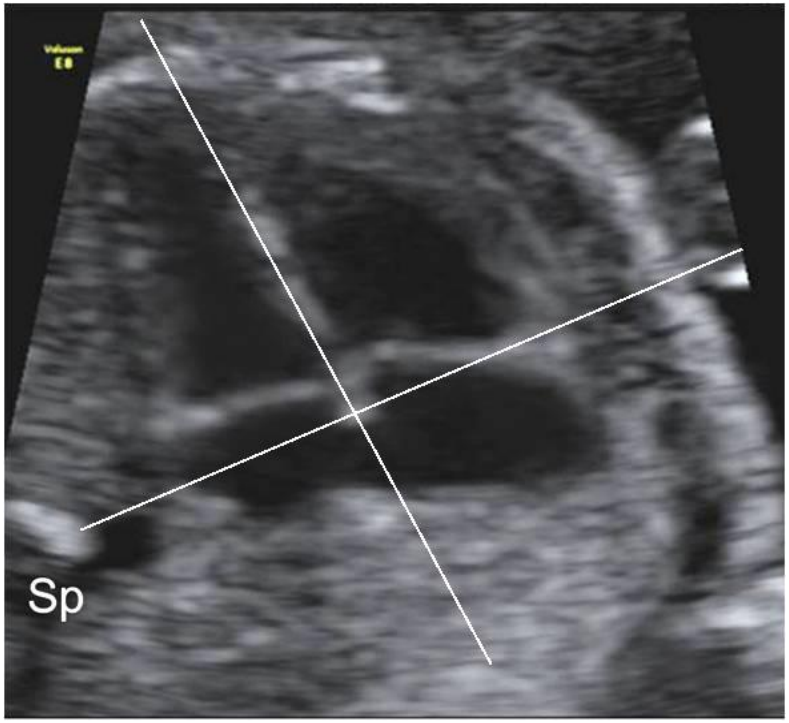


The main association of APVS is with tetralogy of Fallot and arterial duct agenesis; however in extremely rare cases this abnormality has been described in isolation or associated with tricuspid atresia.

Images by P.Volpe

Diagnosis: the four chamber view





In the four-chamber view an evident cardiomegaly (dilated right ventricle due to volume overload from the insufficient pulmonary valve) and a left axis deviation of the heart are the two key features of APVS

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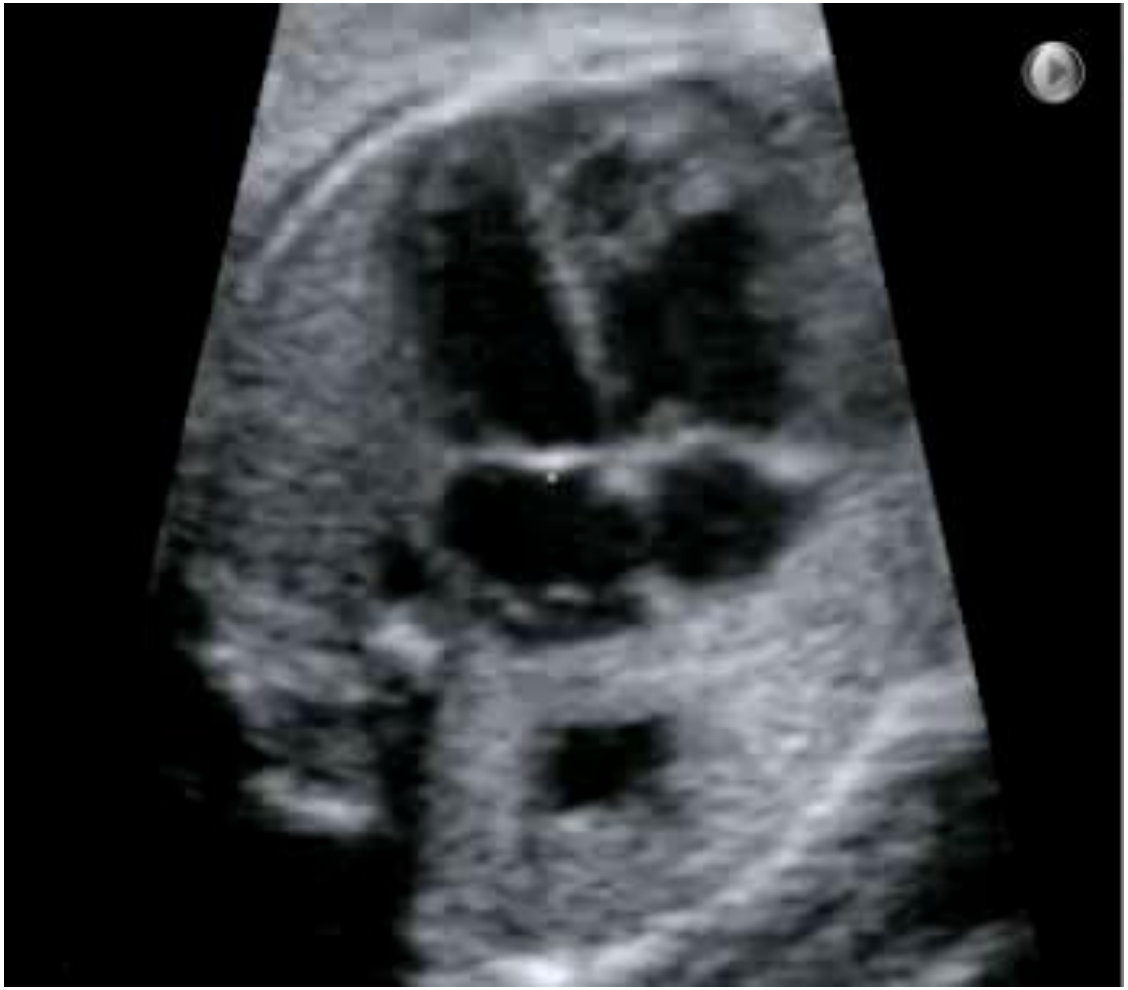
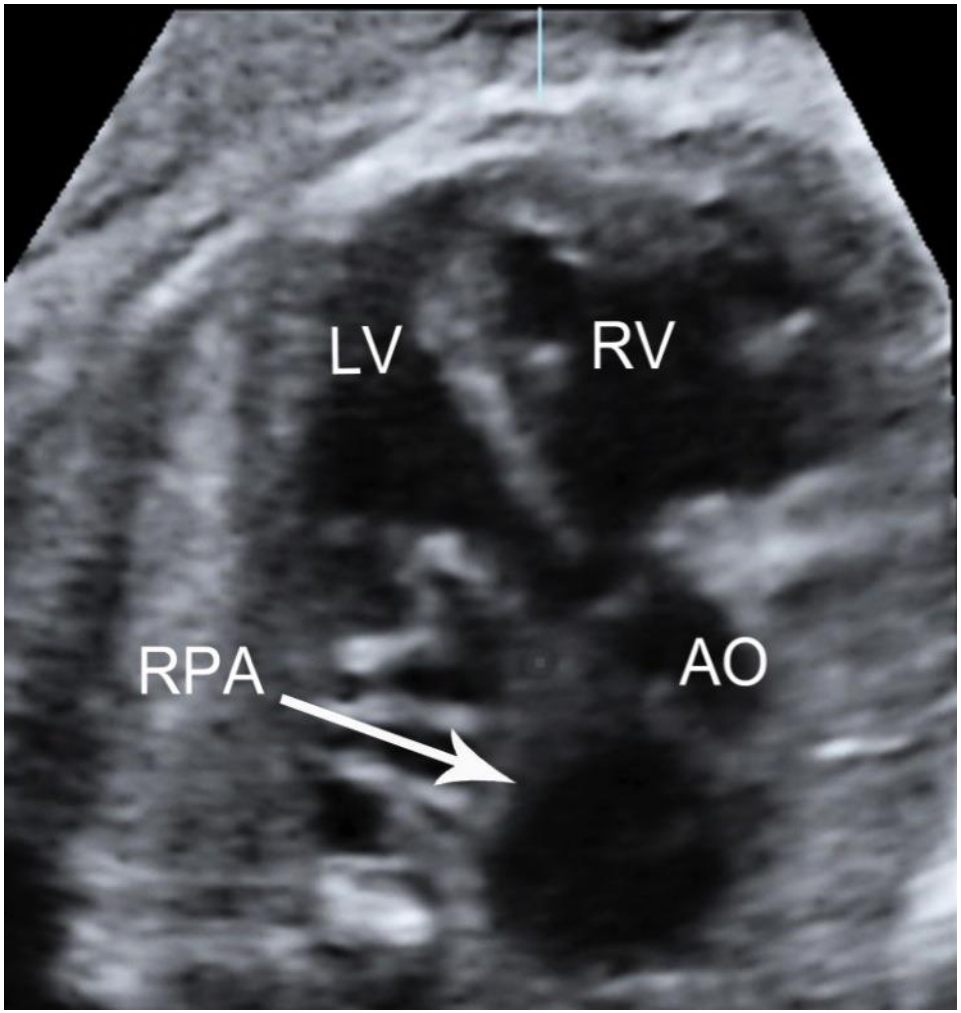
Clip 02



On some occasions, the pulmonary trunk may be so dilated that it also becomes visible on the four chamber view

Images by P.Volpe

Left outflow tract view

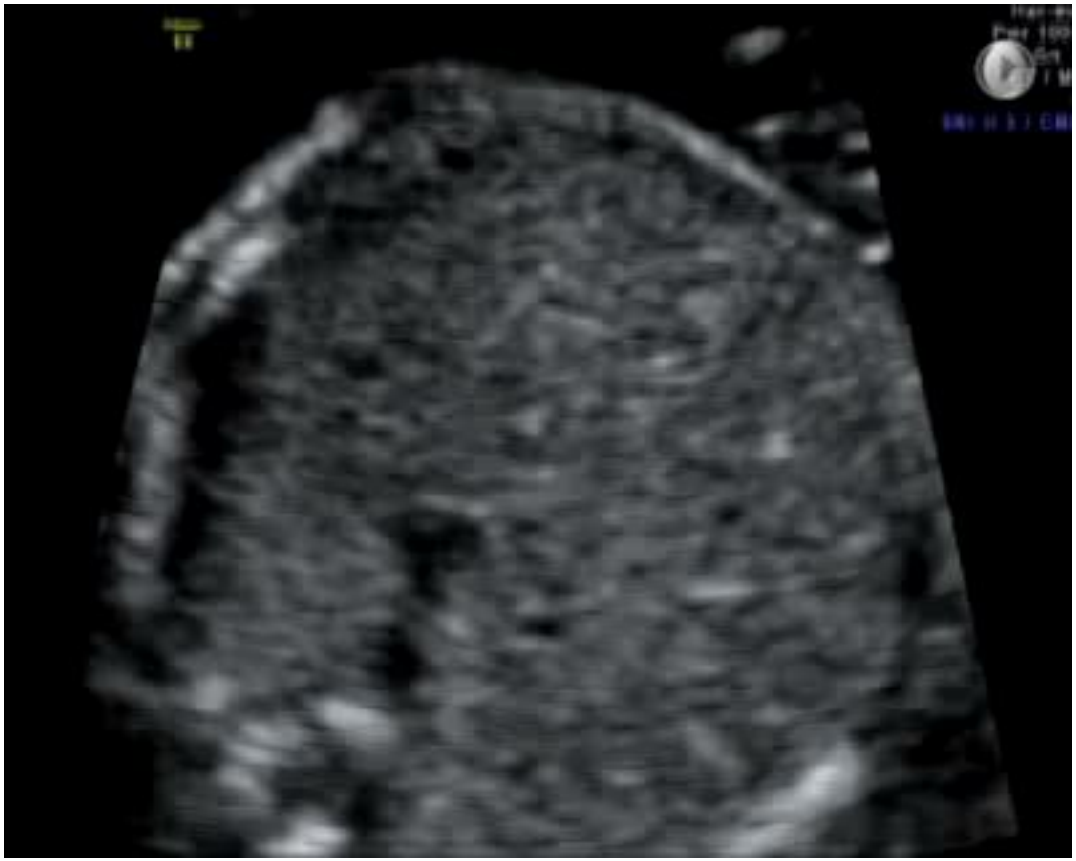


In the left outflow view, a malalignment VSD with an overriding aorta can be seen. The aortic root is not dilate unlike the classic form of TOF

Images by P.Volpe

Outflow tracts

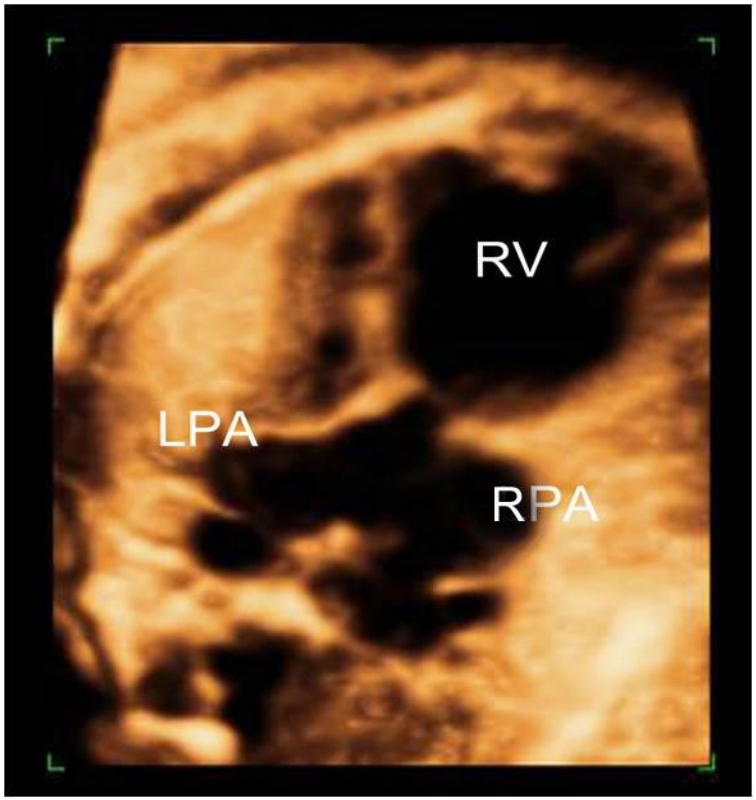
Clip 03

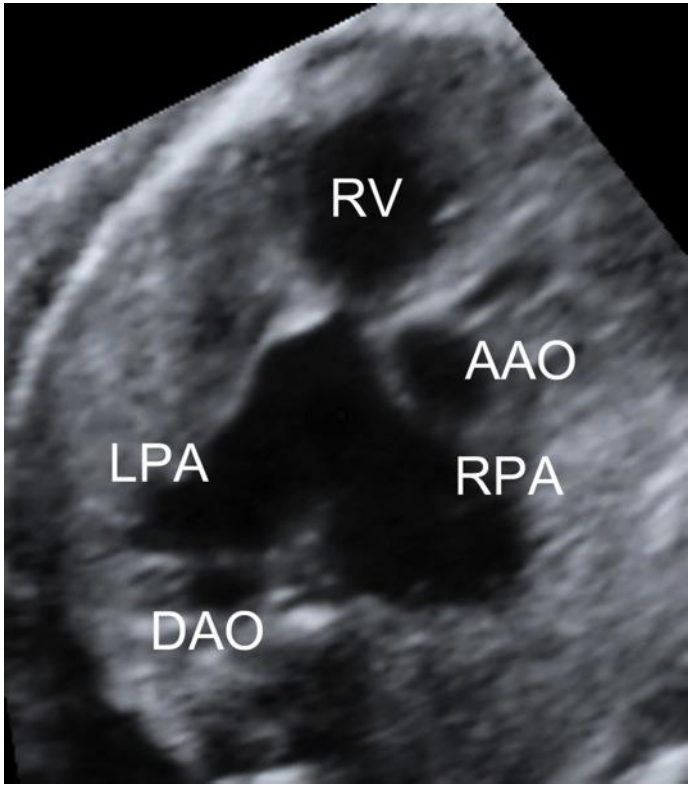


Sweeping further cephalad from the left outflow view, a severe dilation of the pulmonary trunk and branches is evident.

Images by P.Volpe

Pulmonary artery

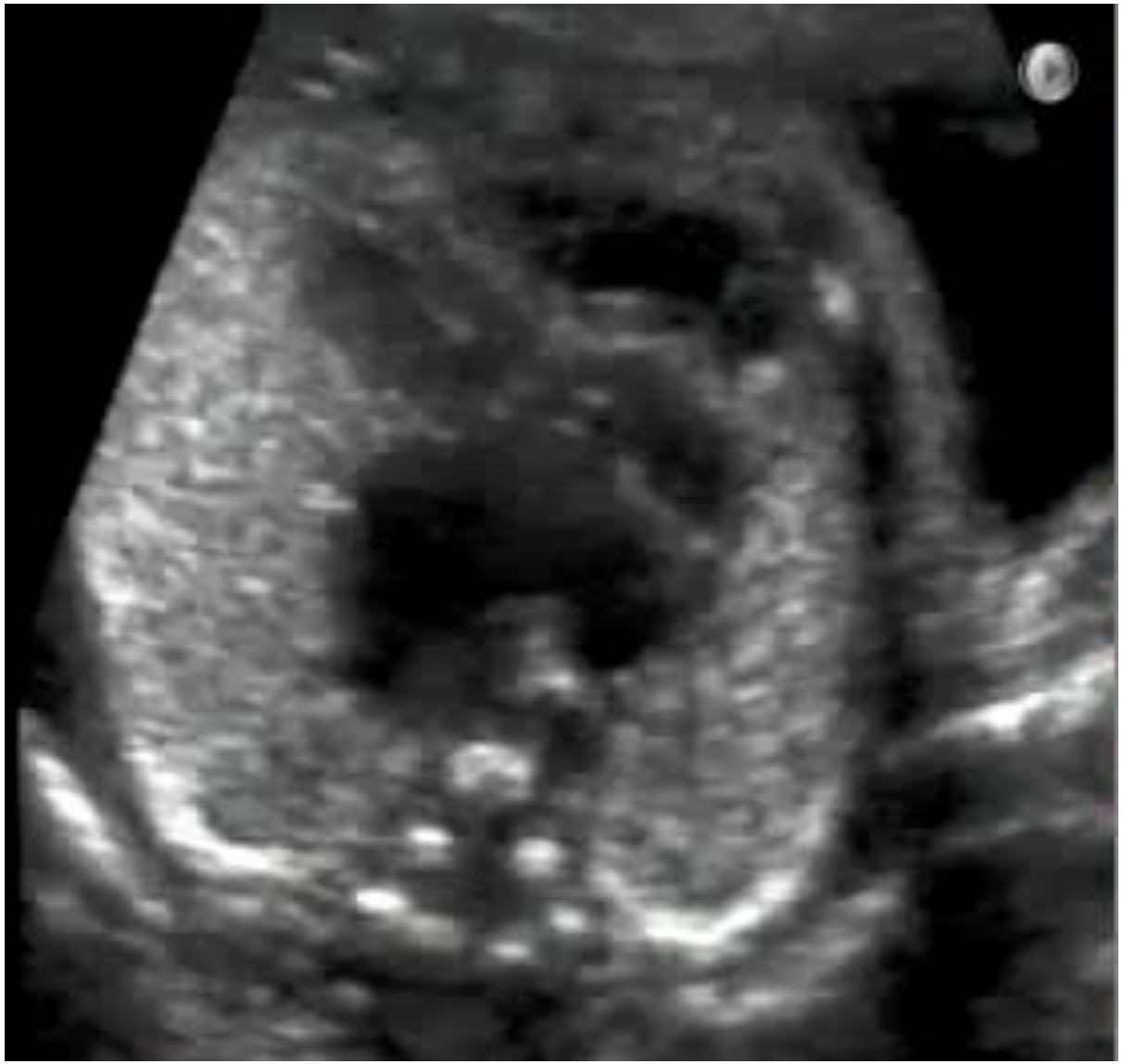




The US aspect of the dilated right and left pulmonary arteries is so typical that, after being seen once, it will never be forgotten

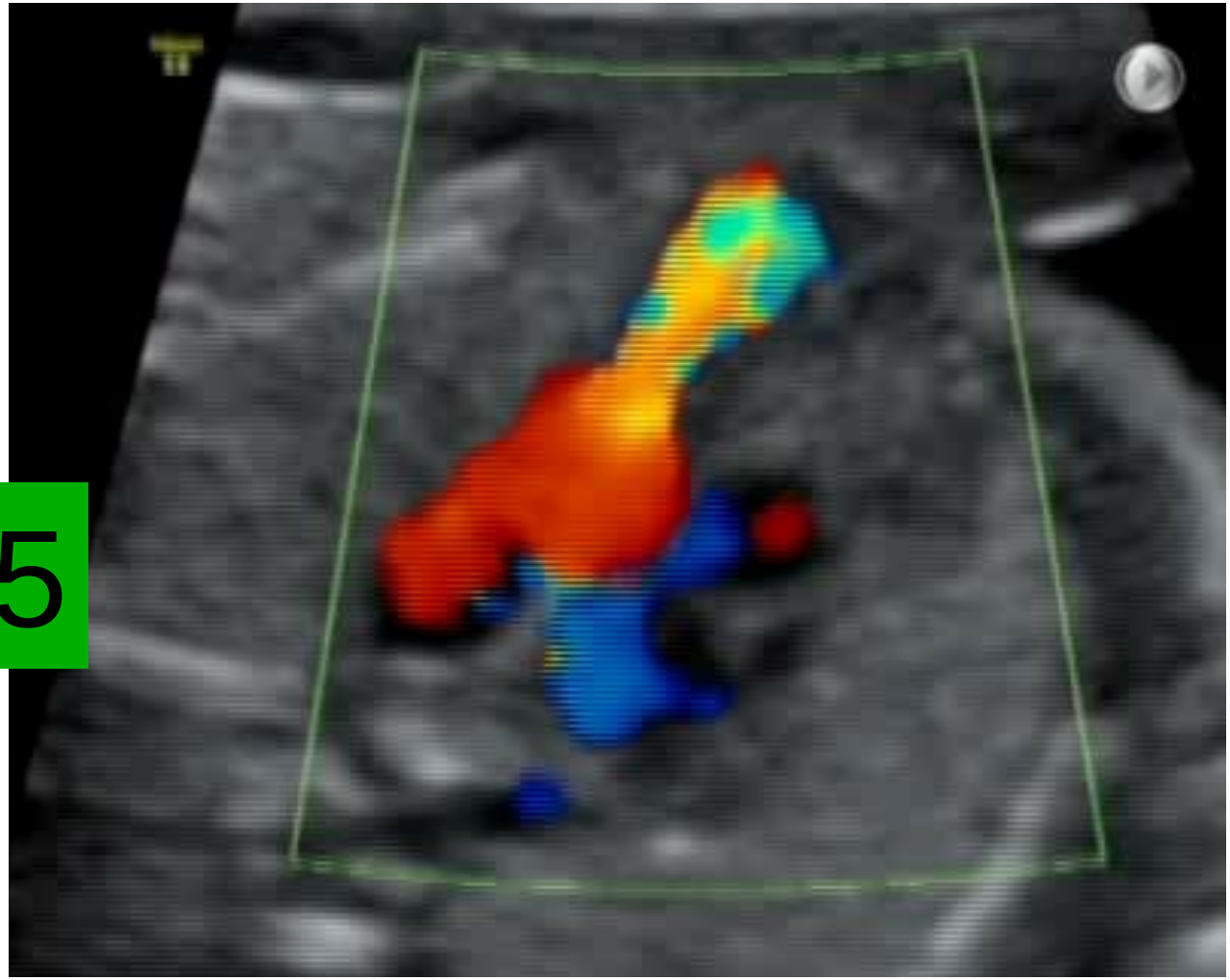
Images by P.Volpe

Clip 04



Images by R. Chaoui

Clip 05

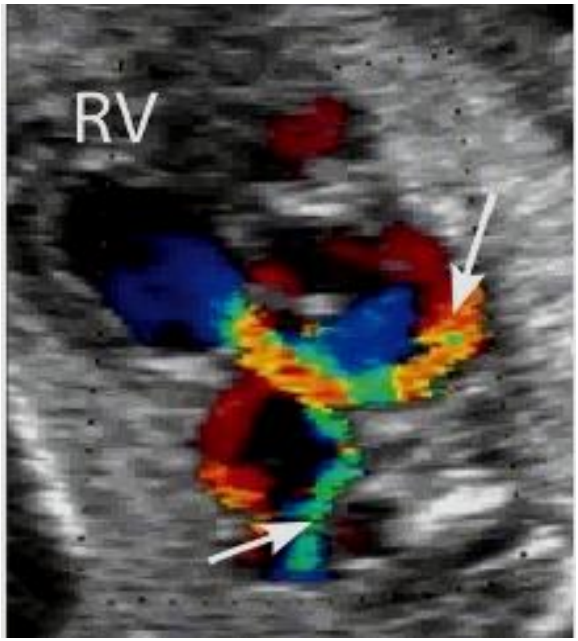


Color Doppler allows detection of the severe stenosis and insufficiency of the functionally absent pulmonary valve

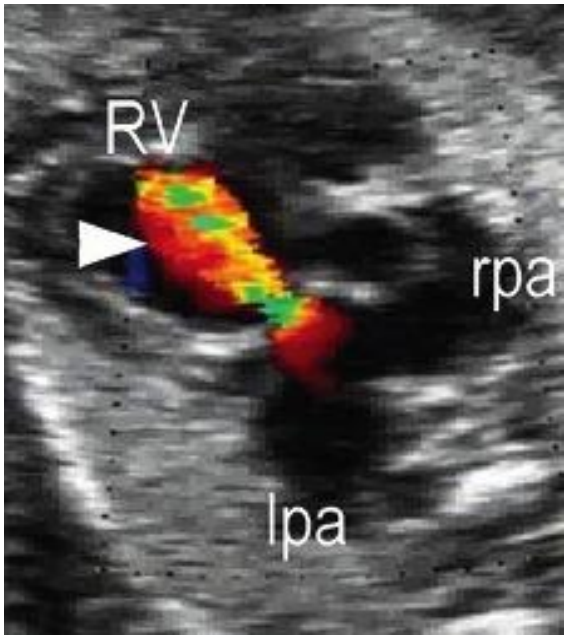
Images by P.Volpe

Doppler of pulmonary artery

Systole



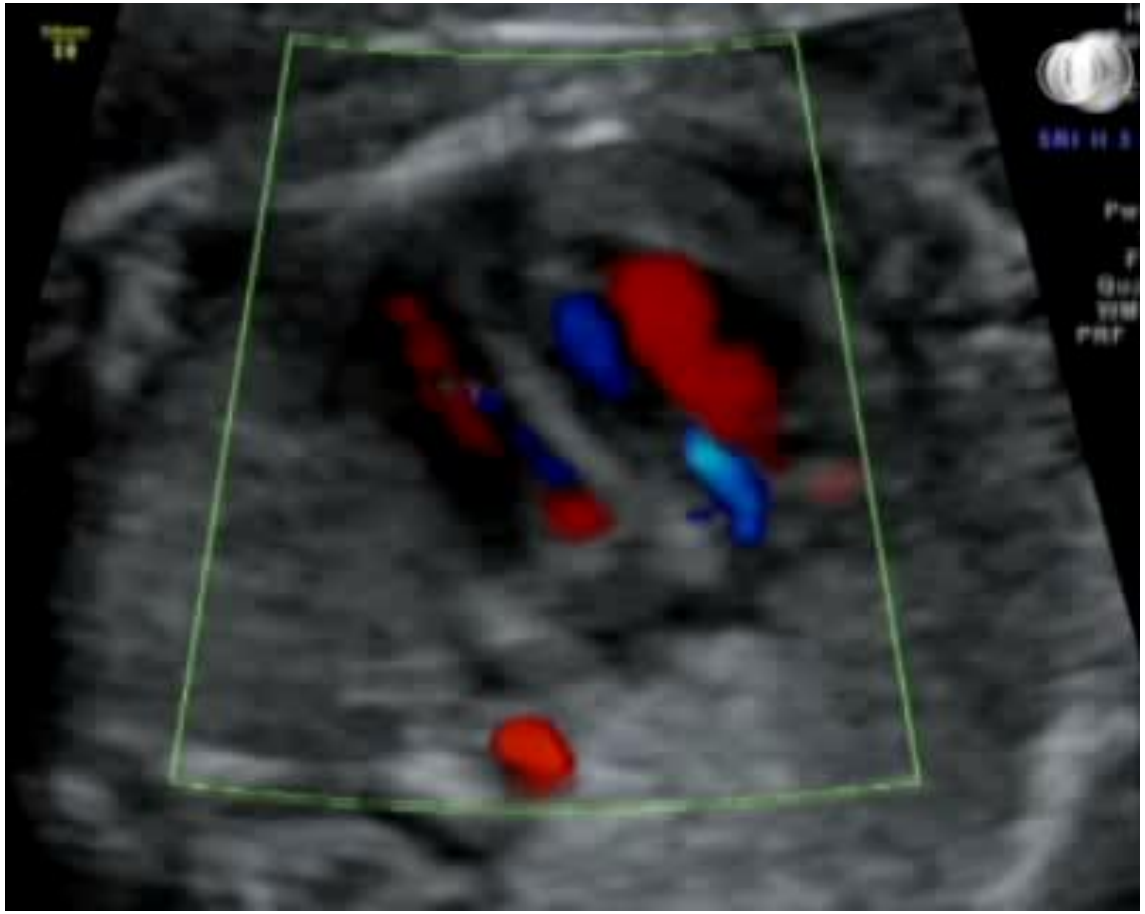
Diastole



During systole, the high-velocity and turbulent blood flow is seen entering the main pulmonary trunk and recircling along the walls of the pulmonary branches; during diastole, a severe pulmonary insufficiency is evident. This is responsible for the volume overload leading to both cardiomegaly and dilation of the pulmonary arteries.

Images by

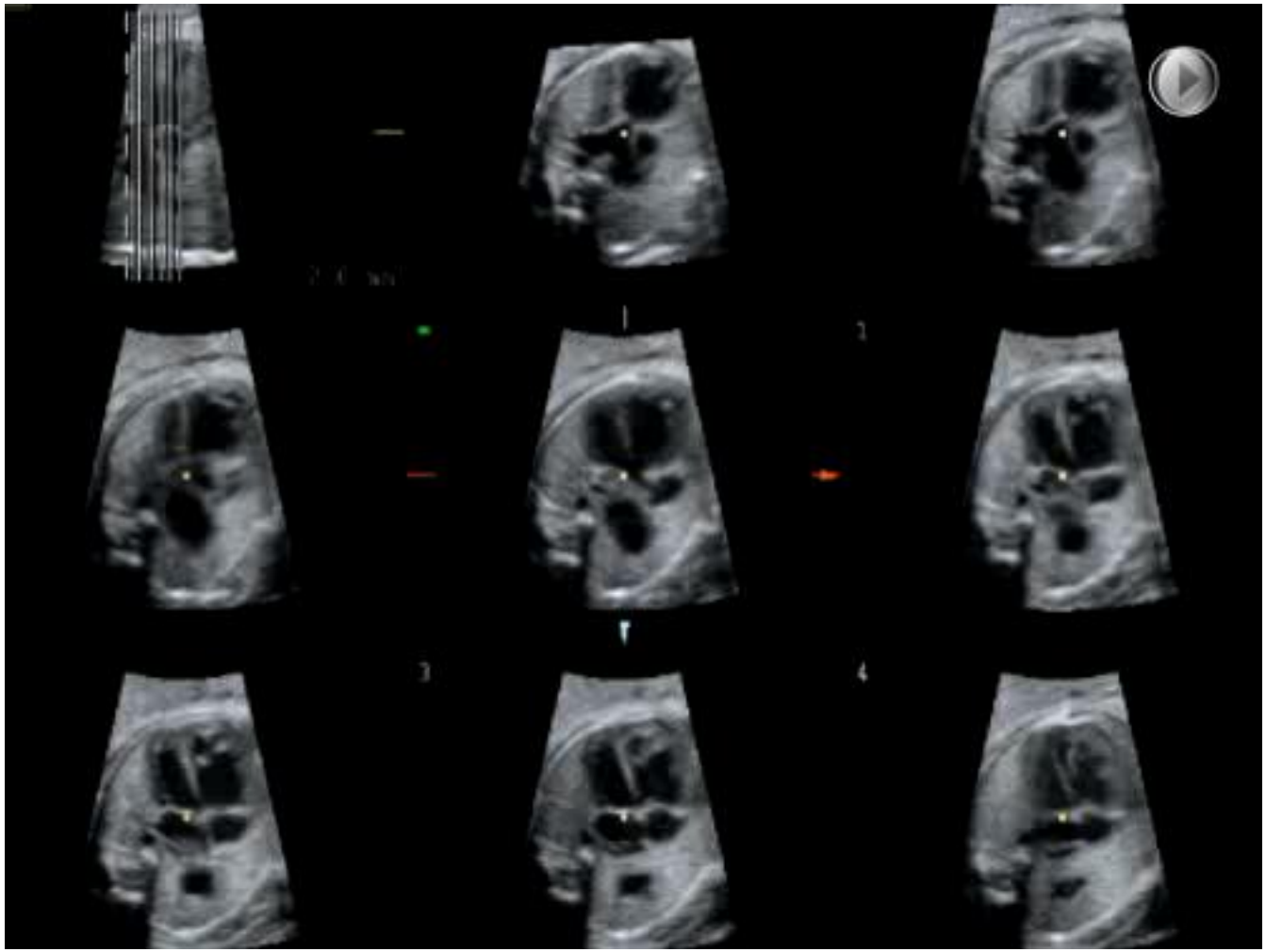
Clip 06



Associated cardiac findings include a right-sided aortic arch.
In this case the risk of association with 22q11 microdeletion is increased

Images by P. Volpe

Clip 07

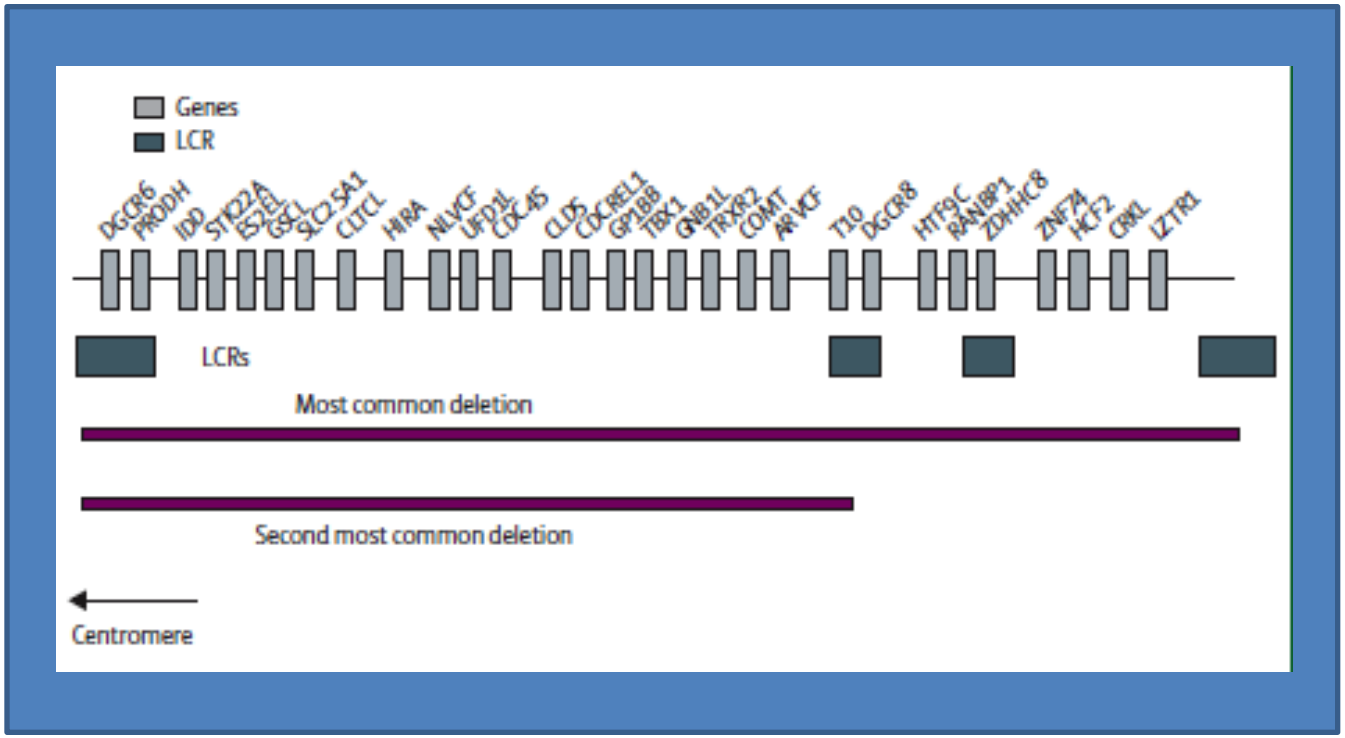


Three-dimensional tomographic imaging enables the demonstration of a *cardiomegaly* in the four chamber view, an *outlet VSD* with overriding aorta in the left outflow view, and a markedly *dilated pulmonary arteries* in the right outflow view

Images by P. Volpe

Absent Pulmonary Valve Syndrome and 22q11 microdeletion

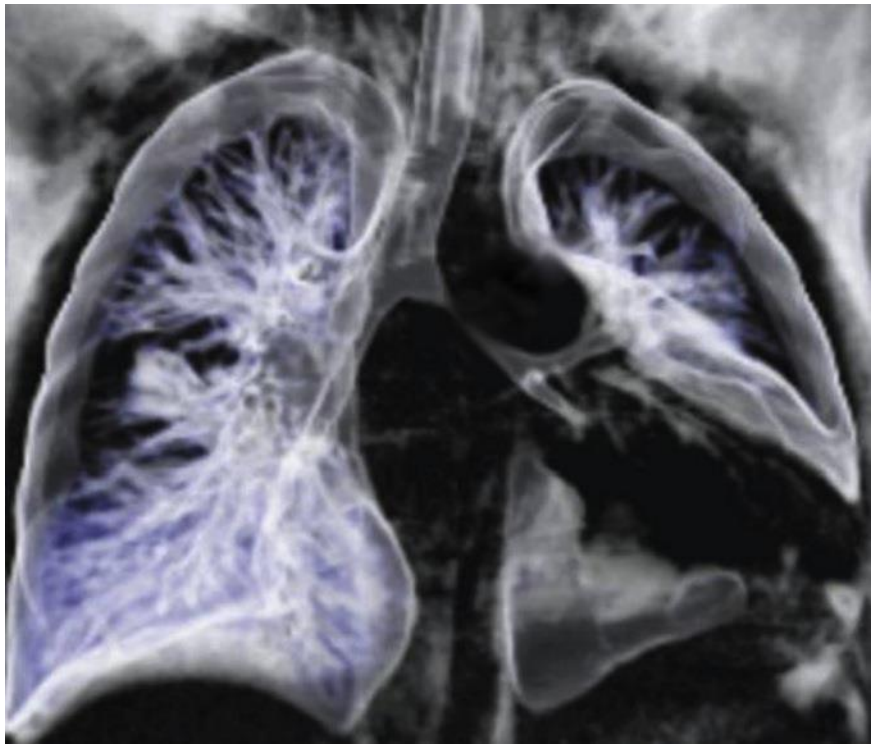
Genes in the commonly deleted region of chromosome 22



A high association with chromosomal anomalies, especially the 22q11 microdeletion (20-25% of cases) has been reported

Images by P. Volpe

The outcome of absent pulmonary valve syndrome



Prenatally diagnosed APVS is associated with a poor outcome (15-20% survival rate). However, in the more recent series the outcome of APVS seems to be significant better (up to 50% survival rate) compared with those of previous series. Main risks to children with APVS include chromosomal abnormalities (primarily 22q11 microdeletion) and respiratory symtoms due to bronchial compression by the dilated PAs

Images by P. Volpe