Letter to the Editor: Total anomalous pulmonary venous connection to an unroofed coronary sinus diagnosed in a fetus with associated spinal muscular atrophy type I.

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We report a first case of total anomalous pulmonary venous connection (TAPVC) to unroofed coronary sinus diagnosed antenatally, which had rapid fatal neonatal course due to spinal muscular atrophy (SMA) type I (Werdnig-Hoffmann disease).

A 23-year old woman, gravida 1, from non-consaguineous relationship, was referred for fetal echocardiography at 23 weeks' gestation because of 'dilated abdominal aorta'. The initial scan showed dilatation of the inferior vena cava at its entrance to the right atrium. Follow up scan at 26 weeks revealed abnormal pulmonary venous connection to a confluence postero-inferior to the left atrium. From the confluence, pulmonary venous return was directed towards the region of the coronary sinus, although the roof could not be fully visualised. Pulmonary venous return seemed to enter the left atrium but in addition, colour flow mapping showed streaming of pulmonary venous flow towards the inferior vena cava, in keeping with the observation at the first fetal cardiac scan (Fig.1A). The diagnosis of TAPVC with unroofed coronary sinus was made (Fig.1B-C). There was also increased nuchal thickening, pre-nasal oedema and single umbilical artery. The family declined invasive testing.

A male infant was delivered at 37+3 weeks. Postnatal echocardiogram and CT scan confirmed the findings and also demonstrated a superior sinus venosus defect (Fig.2,3S). Genetic testing carried out due to generalised hypotonia and pathological reflexes revealed homozygous mutation in the survival motor neuron gene 1 (SMN1) confirming the presumptive diagnosis of SMA. Only one copy of the SMN2 gene was present. The infant rapidly became dependent on mechanical ventilatory support and failed extubation on several occasions. Following discussion with the family, the infant was extubated and died soon thereafter.

Prenatal diagnosis of TAPVC is rare with only eight out of 424 cases being identified prenatally from a multicentre study[1,2]. TAPVC to unroofed coronary sinus has never been reported in a fetus. Postnatally, there is sparse published data (Fig.4S).[3] The diagnosis is usually an incidental finding with concomitant congenital cardiac anomalies. Haemodynamically, it leads to right to left shunt at atrial level. Like all inter-atrial communications, it may cause symptoms of right heart failure, atrial arrhythmias and pulmonary hypertension in late adulthood.

Our patient was also diagnosed with SMA type I. This association has never been reported. SMA is a rare autosomal recessive neurodegenerative disease affecting motor neurons in the spinal cord causing postnatal degeneration and subsequent weakness of skeletal muscles, with feeding and breathing difficulties. Type I represents the most severe form of the disease beginning during the first 3 months of life and associated with early mortality. The SMN2 gene is responsible for the severity of the disease. It encodes an identical protein as the SMN1 gene but with impaired function. The number of copies of SMN2 influences the amount of fully functioning SMN protein. The worst types of SMA are associated with only one copy of SMN2 protein and there is a powerful association with congenital heart defects.[4] It is interesting to speculate that the SMN2 gene may be involved in cardiogenesis in these patients.

References.

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Conflict of interest. None

Figures Legends.

Figure 1. Prenatal echocardiogram at the 26 weeks' gestation shows: A. Significant dilatation of the inferior vena cava at its entrance to the right atrium; B-C. Abnormal pulmonary venous connection into unroofed coronary sinus. IVC = vena cava inferior, LA = left atrium, LPV = left pulmonary veins, RPV = right pulmonary veins

Fig 1A.

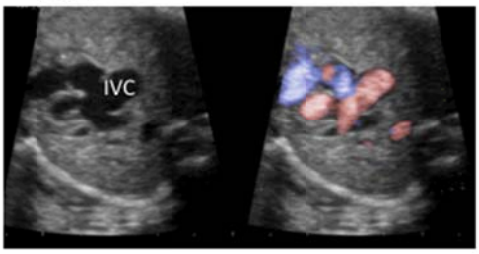


Fig 1B.

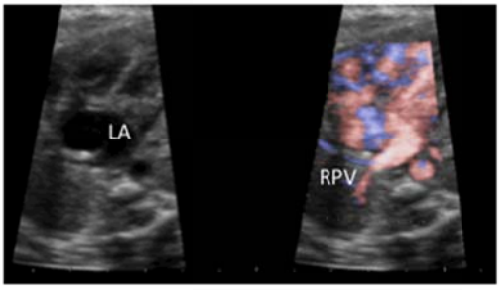


Fig 1C.

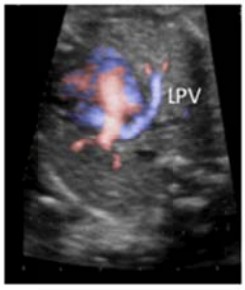


Figure 2A-B. Postnatal echocardiogram shows large atrial septal defect and abnormal connection of the pulmonary veins. AO = aorta, ASD = secundum atrial septal defect, ASD/sec = secundum atrial septal defect, ASD/svs = sinus venosus superior atrial septal defect, LV = left ventricle, SVC = superior vena cava

Fig 2A.

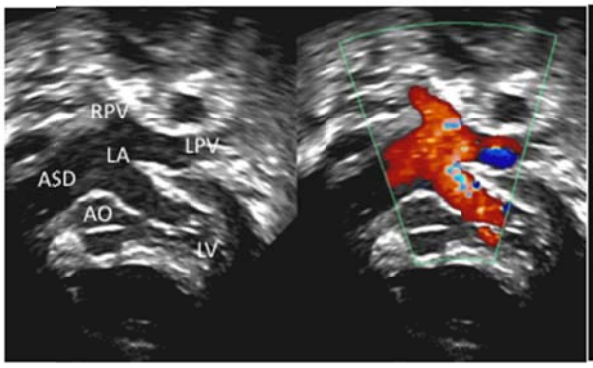
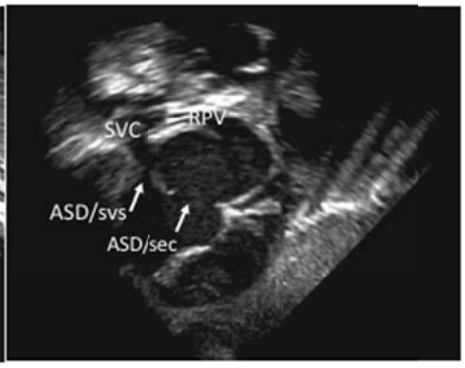


Fig 2B.



Supplementary material.

Figure 3S. Postnatal CT scan showing abnormal connection of the right and left pulmonary veins to the postero-inferior aspect of the left atrium.

Figure 4S. Schematic pictures showing A. normal cardiac anatomy; B. total anomalous venous connection to coronary sinus; C. total anomalous venous connection to unroofed coronary sinus. CS = coronary sinus, PVC = pulmonary venous confluence, RA = right atrium