Novel diagnostic procedure

A clue to the diagnosis of TAPVD

Andrew J Jones,¹ Krzysztof Zieba,¹ Luke Starling,² Joanna Longman¹¹Neonatal Unit, Northwick Park Hospital, London, Middlesex, UK;²Paediatric Cardiology Department, Royal Brompton Hospital, London, UK**Correspondence to** Dr Andrew J Jones, andrewjones7@nhs.net**Summary**

Total anomalous pulmonary venous drainage (TAPVD) is a rare form of congenital heart disease where all four pulmonary veins drain to the systemic venous circulation. A term infant was found to have low oxygen saturations on the neonatal check and he was admitted to the neonatal intensive care unit. An increasing oxygen requirement necessitated invasive ventilation. A blood gas taken from the umbilical venous catheter (UVC) showed a pO_2 of 28.1 kPa – a finding that at the time was considered to be erroneous. An x-ray showed the UVC tip was located in the liver. The following day the baby was transferred to a cardiology centre where a diagnosis of unobstructed infracardiac TAPVD was made on echocardiography. In retrospect the unusually oxygenated venous gas was consistent with pulmonary venous return draining directly to the hepatic venous system. This could have provided a vital clue to diagnosis in a situation where an echocardiogram was not available.

BACKGROUND

Total anomalous pulmonary venous drainage (TAPVD) (or total anomalous pulmonary venous connection) is a rare form of congenital heart disease in which all four pulmonary veins drain to the systemic veins, right atrium or coronary sinus. Pulmonary and systemic venous blood mix in the right atrium causing volume loading of the right atrium and right ventricle. An atrial septal defect or patent foramen ovale (PFO) is always present and is crucial for left ventricular output.

TAPVD accounts for 1.5% of all congenital heart disease, with an overall prevalence of 6.8/100 000.¹ The anomalous venous drainage can be supracardiac, with drainage to the right superior vena cava or innominate vein; cardiac, with drainage to the coronary sinus or right atrium; or infracardiac, with drainage to the inferior vena cava (IVC) or hepatic venous system.

Clinical presentation depends on whether the pulmonary venous return is obstructed or unobstructed. If unobstructed the presentation can be insidious with failure to thrive and recurrent chest infections in infancy. Obstructed TAPVD results in an acutely unwell neonate who presents with tachypnoea, tachycardia and severe cyanosis. The natural history of obstructed TAPVD is death within weeks to months. Obstruction will eventually occur in virtually all children with infracardiac drainage, but is less common when drainage is supracardiac.

In our case venous drainage was infracardiac, initially without obstruction. This fits with the clinical picture of a well neonate with mild cyanosis picked up on routine pulse oximetry screening.

This case highlights a somewhat esoteric clue to the diagnosis of a very rare heart condition: TAPVD. However, it also demonstrates that no piece of clinical information should be disregarded, even if it cannot obviously be reconciled with the clinical picture. In a district general hospital, where the expertise to perform and interpret sophisticated diagnostic investigations may not always be immediately available, it is important to make the most of all clinical

information available to try and make a diagnosis and expedite appropriate treatment.

CASE PRESENTATION

A male infant was born by emergency caesarean section at term following an unremarkable pregnancy. He was the first child of non-consanguineous Sri Lankan parents. The decision to deliver the baby by caesarean section was made due to atypical variable decelerations in the fetal heart rate seen on cardiotocography. The baby required no resuscitation at birth. Birth weight was 3250 g and APGAR scores were 8 at 1 min, 9 at 5 min and 10 at 10 min. Intravenous benzylpenicillin and gentamicin were commenced according to local protocol in view of maternal fever during labour.

At 20 h of age a full newborn check was carried out on the postnatal ward. As is routine at our hospital, pulse oximetry was performed showing saturations of 98% preductal and 93% postductal; however the baby appeared well and had no signs of respiratory distress. Heart sounds were normal with no murmurs and femoral pulses were palpable.

On review at 24 h of age the baby remained well but oxygen saturations had fallen to 93% preductal and 88% postductal. The decision was made to admit the baby to neonatal intensive care unit for further investigation and management.

He was placed in an incubator and given supplemental oxygen via nasal cannulae at a rate of 2.5 l/min. However, the oxygen requirement gradually increased over the next 6 h with the baby needing high flow oxygen and then intubation and ventilation in order to maintain oxygen saturations above 95%.

A catheter was inserted into the umbilical vein (it was not possible to catheterise the umbilical artery).

He was ventilated using conventional mechanical ventilation in 45% oxygen. It was noted that preductal oxygen saturations were 95%, and postductal 99%. The next day the baby was transferred to paediatric intensive care at a tertiary centre for further investigation.

INVESTIGATIONS

ECG was normal. Chest x-ray was unremarkable with no evidence of pulmonary venous congestion.

C reactive protein was <10 mg/l on two occasions, 24 h apart.

The blood gas drawn from the UVC showed: pH 7.48, pCO₂ 3.17 kPa, pO₂ 28.1 kPa, lactate 1.9 mmol/l, HCO₃ 17.6 mmol/l.

A contemporaneous capillary blood gas showed: pH 7.47, pCO₂ 4.58 kPa, pO₂ 4.84 kPa, HCO₃ 24.8 mmol/l.

At 46 h of age the baby was transferred to the local cardiac centre for an echocardiogram, which showed TAPVD of the infracardiac type with the confluence draining into the hepatic vein with no sign of obstruction. Also present were a patent foramen ovale with right to left shunt and a small patent ductus arteriosus with bidirectional flow.

DIFFERENTIAL DIAGNOSIS

The differential diagnosis of a term neonate presenting with decreased oxygen saturations is wide, and includes pneumonia, transient tachypnoea of the newborn, persistent pulmonary hypertension of the newborn and congenital heart disease. In this case the presence of cyanosis in the absence of respiratory distress, the absence of lung pathology on chest x-ray and the normal chest x-ray and the normal inflammatory markers put heart disease at the top of the list of differentials. Without an echocardiogram narrowing down the diagnosis any further is difficult.

However, the presence of an unexpectedly oxygenated blood gas drawn from the umbilical venous catheter (UVC) in this context is diagnostic of infracardiac TAPVD. Correct interpretation of this result could have led to an expedited transfer to a cardiac centre. This would have been particularly important if pulmonary venous return had become obstructed and the baby had deteriorated.

In theory, the presence of a UVC in the IVC or the hepatic veins could itself obstruct venous drainage. If infracardiac TAPVD is diagnosed, the UVC should be removed.

TREATMENT

After transfer and diagnosis he was initially observed in the tertiary centre. However, the PFO became narrower leading to the development of cardiac failure. At 2 weeks of age he underwent a full surgical repair where the confluence of the pulmonary veins was anastomosed to the left atrium.

OUTCOME AND FOLLOW-UP

Postoperatively the infant was treated for sepsis and suffered a significant pulmonary hypertensive crisis requiring inhaled nitric oxide and paralysis. He was discharged from hospital 3 weeks postoperatively, feeding from the breast

with bottle top ups, and on a weaning dose of Sildenafil. He is currently doing well at home and will have regular follow up.

DISCUSSION

Two similar case reports describe diagnosis of TAPVD aided by umbilical vein catheterisation. In both cases the infant presented with cyanosis but was otherwise well. In the first case,² from the pre-echocardiogram era, the diagnosis was made at 13 h of age based solely on the high pO₂ observed in blood gases from the UVC. In the second report³ diagnosis was initially missed on echocardiogram and the high pO₂ observed in venous gases was at first disregarded as anomalous. A series of six cases of infracardiac TAPVD⁴ describes bedside umbilical venous catheterisation as being diagnostic in three cases. In our case umbilical venous catheterisation was only noted retrospectively to be of diagnostic importance; but if identified at the time then a diagnosis could have been made immediately.

Learning points

- ▶ No clinical information should be entirely disregarded, even if it cannot immediately be reconciled with the clinical picture.
- ▶ Where there is a high index of suspicion, blood gases from a UVC can be used to diagnose infracardiac TAPVD.
- ▶ Infants with unobstructed TAPVD can have no or only subtle clinical signs – in this case the baby was only identified as having a problem through routine pulse oximetry.
- ▶ Once the diagnosis of infracardiac TAPVD is made the UVC should be removed so as not to obstruct venous return.

Competing interests None.

Patient consent Obtained.

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