The 20 week scan: an opportunity to detect or miss cardiac anomalies?

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Introduction

Ultrasound screening for foetal structural abnormalities is generally recommended at 19-21 weeks of gestational age. The accuracy in detecting malformations by ultrasound, however, shows great variability among centres and operators. Congenital malformations occur in 2-4% of all births. Despite their relatively low prevalence, foetal malformations are responsible for approximately 30% of perinatal deaths in addition to considerable infant morbidity in developed countries.

We describe two complicated cases of cardiac anomalies where the diagnosis was missed during the anomaly scan between 20-22 weeks. The first case was of a baby found to have Situs inversus (SI) where the diagnosis was made only during the postnatal check up. The second case was of a baby with transposition of the great vessels (TGA). Unfortunately, this was not diagnosed during the 20 week scan and was diagnosed postnatally.

TGA is one of the most common cyanotic congenital heart defect in neonates. The hallmark feature is ventriculo arterial discordance whereby the aorta arises from the morphologically right ventricle and the Pulmonary artery arises from the morphologically left ventricle. The prevalence is 0.2/1000 live births and according to one 13 year study, the detection rate was 6.9% which improved to 25% in the last four years of the study(1). A neonate with TGA will usually present primarily with cyanosis within the first 24 hours of life.

SI alone does not have clinical features however there is an increased association with Congenital Heart Defects (CHD). One study showed 23% of babies with SI when investigating dextrocardia Of the babies with SI, 63% had structural cardiac malformations and 10% had extracardiac features(2). SI can be part of a triad with primary ciliary dyskinesia (PCD) and abnormal frontal sinuses, this is known as Kartagener’s syndrome. SI can be seen in 41% of people with PCD(3) therefore a patient with SI may present with pulmonary features and be diagnosed incidentally for SI as was the case for our first patient.

Case 1

The first patient with SI was born at term by normal vaginal delivery and did not require resuscitation at birth. He was persistently tachypnoic with perioral cyanosis and irritability. He was then admitted to Special Care Baby Unit (SCBU). His chest x-ray showed signs of congenital pneumonia and was treated with 10 days of intravenous antibiotics. After having an x-ray, it was found that he had dextrocardia and subsequent abdominal ultrasound confirmed SI. Due to the SI and pneumonia, there was a strong suspicion that the patient had primary ciliary dyskinesia as he struggled with copious chest secretions which improved with physiotherapy.

Case 2

The second patient with TGA was born at term by normal vaginal delivery and was in a good condition at birth with no resuscitation needed. There was a maternal history of hereditary angioedema and alpha thalassaemia carrier but the father was of low risk. On the postnatal ward, the patient was noted to have an apnoeic episode, followed by vomiting. During this episode she showed signs of cyanosis which lasted 30 seconds. After recovering from this episode, the patient was brought to SCBU and was found to have low pre and post-dactural saturations (70% and 85% respectively). These did not improve with oxygen. An echocardiogram was performed which showed TGA with an atrial communication and a small ventricular septal defect. An umbilical vein catheter (UCV) had been placed and Prostaglandin E had been administered, along with prophylactic antibiotics for sepsis cover.

Discussion

Early detection for cardiac anomalies, such as TGA, allows for the medical team and families to better anticipate the potential complications of TGA and it can also improve neonatal morbidity and mortality. First year mortality has been proven to be significantly lower in cases with a prenatal diagnosis of TGA compared to those without (0.0% vs 11.4%, respectively)(4). Morbidity is also significantly lower in cases with a prenatal diagnosis, those without a prenatal diagnosis had higher rates of hypoxia and renal dysfunction.

Early detection is essential in management however technical difficulties may arise in diagnosing cardiac anomalies at such an early stage. The routine antenatal scan at 20 weeks provides a four chamber view of the heart using 2-D sonography to assess structural integrity however sensitivity for detection of TGA may be lacking. The presence of an ‘T’ shaped aorta in the upper mediastium during the scan could be a novel potential marker for antenatal diagnosis of TGA. In one retrospective evaluation, the ‘T’ shaped sign (fig 1) as observed in 96.8% of cases of TGA (5). Sonographers could receive further training in identifying such a marker to allow for a prenatal diagnosis of TGA.

Due to the difficult nature of prenatally diagnosing these cardiac anomalies, healthcare professionals could apply other methods to diagnose early in the neonatal stage if they are unable to diagnose prenatally. Methods such as pulse oximetry could be utilised to minimise the risk of discharging infants with CHDs such as TGA. One retrospective cohort study demonstrated that hospitals using pulse oximetry screening diagnosed all neonates before discharge(6).

The early detection of SI carries significance due to the fact that almost half of people with PCD have SI (Kartagener’s syndrome) and although complications may not arise directly from SI, complications may be due to PCD. 70-80% of neonates with PCD present with respiratory distress and there is an increase risk of developing pneumonia(7), as we saw with our first patient. This creates a great emotional burden on the parents of babies born with such complications thus early anticipation of potential PCD, by detecting SI in the antenatal setting, may ease the shock factor parents experience upon seeing their new-born with respiratory distress

Conclusion

Whilst acknowledging the difficulty in detecting such anomalies, we must strive to improve imaging techniques in the primary setting to prepare for potential future management, ease the strain on secondary and tertiary health centres and most importantly to better prepare the families of these babies. Early detection of these anomalies would also give a choice to mothers for continuing or terminating pregnancies as some parents may not wish for their children to experience complications as a result of these anomalies. Further techniques are required to improve sensitive in detecting anomalies such as TGA and SI

References