Uhl's anomaly detected in-utero

Abstract

Uhl's anomaly is rarely diagnosed antenatally. It is a condition characterised by partial or complete absence of the right ventricular myocardium and the heart conducting system. We present a case of Uhl's anomaly diagnosed during fetal life on prenatal ultrasound, with eight years postnatal follow-up.

Keywords: antenatal, fetal, Uhl's anomaly, ultrasound.

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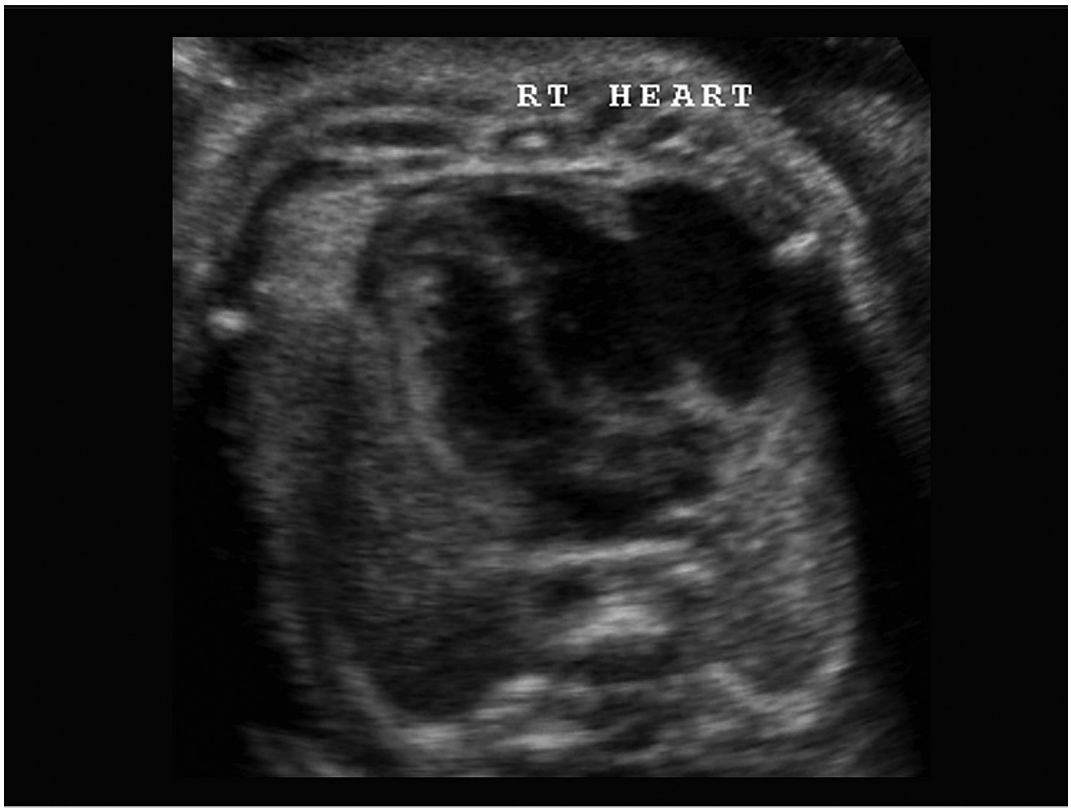

Figure 1: Fetal echocardiograph showing gross cardiomegaly and a large aneurysmal segment in the free wall of the right ventricle.

Case report

A 37-year-old woman (gravida 1 para 0) was referred to San Ultrasound for Women at Sydney Adventist Hospital for a three-dimensional ultrasound examination at 29 weeks gestation. Her antenatal course had been unremarkable and the 19 week fetal morphology scan performed elsewhere detected no obvious fetal abnormality.

The parents requested a three-dimensional ultrasound examination to see the baby's face for a bonding experience. There was no other clinical indication. The fetal biometry showed an estimated fetal weight of 1402 g, which corresponded to the 50th centile for 29 weeks gestation, and the fetus was biophysically well. The fetal heart, however, was noted to be enlarged. The right ventricle was grossly distended with a hypokinetic lateral wall. The ventricular septum was intact and the valves were functioning normally with no regurgitation. The left ventricle was completely normal in size, shape, and function; and both outflow tracts were appropriately connected. The fetal heart rate was 155bpm and it was in sinus rhythm with occasional ectopic beats. No other fetal abnormality was detected.

The patient was referred for formal fetal echocardiography by a paediatric cardiologist. The following abnormal findings were confirmed: gross cardiomegaly, frequent ectopic beats, large aneurysmal segment in the free wall of the right ventricle, and some degree of cardiac impairment (Figure 1). The lateral wall of the right ventricle appeared to be muscular but

severely hypokinetic; these findings are most consistent of Uhl's anomaly.

Uhl's anomaly has been linked to the cardiomyopathy known as Arrhythmogenic Right Ventricular Dysplasia (ARVD). Although Uhl's anomaly and ARVD are distinct morphological entities, it is possible that they might share a common pathogenesis.[1] Uhl's anomaly is characterised by congenital partial or complete absence of the myocardium of the parietal wall of the right ventricle.[1] Furthermore, the myocardium is composed only by surfaces of endocardium and epicardium with no interposition of adipose tissue between the layers.[2] In ARVD, however, fibrous and adipose tissue replace the myocardium of the right ventricular wall.[2] It has been considered that ARVD may be due to an autosomal recessive condition or to one of autosomal dominance with incomplete penetrance.[1] For this reason both parents had echocardiography; both studies were normal. Amniocentesis was performed for reassurance and the karyotype was normal male.

Fetal echocardiograms were conducted every two weeks, along with weekly fetal ultrasounds for growth and wellbeing. At 35 weeks gestation, the fetus was in frank breech presentation. The fetal heart was in bigeminy. A male infant of 3180g was delivered by elective Caesarean section at term at a tertiary referral hospital. The baby was born with Apgar scores of 9^{1} and 10^{5}. The baby needed no resuscitation. After birth neonatal arrhythmias were controlled with medication and the infant had no evidence of cardiac failure at discharge. The infant is now healthy nine year old boy, he has had no surgery and needs no regular medications.

Discussion

Uhl's anomaly was first described by Uhl in 1952, and is characterised by congenital partial or complete absence of the myocardium of the right ventricular wall.[3,4] The occurrence of Uhl's anomaly is very rare, and most cases are diagnosed after birth due to symptoms of heart failure or arrhythmia. The first antenatal ultrasound diagnosis of Uhl's anomaly was reported in 1988.[5] To our knowledge, only three prenatal diagnosis of Uhl's anomaly have been documented since then, because of its rarity.[6,7] Although other pathologies may also result in the absence of myocardium of either ventricle wall, Uhl's anomaly is unique as the destruction to the ventricular wall myocardium is not due to an inflammatory processes such as myocarditis or obstructive lesions in the coronary arteries leading to ischemia.[2] The pathogenesis of Uhl's anomaly first was assumed to be a failure of the right cardiogenic fold development occurring in the early growth of the human embryo.[8] However, subsequent embryonic studies failed to demonstrate this hypothesis.[8] The right ventricle does not have its own origin in a single cardiogenic fold, and the destruction or loss of the myocardium of the right ventricle seems to occur after the heart has been fully developed.[8] A possible explanation of Uhl's anomaly is that the right ventricular myocardium undergoes selective apoptosis in utero.[9,10] Apoptosis is a type of cell death that is considered a normal phenomenon in postnatal heart maturation.[9] The excessive right ventricular myocardial mass is reduced via apoptosis due to the reduction of the pressure against the right ventricle after birth.[11] In Uhl's anomaly, this process appears to occur before birth instead of after birth, and it continues until little or no right ventricular myocardium remains.

The fundamental haemodynamic fault in Uhl's anomaly is inadequate or absent right ventricle contraction.[12] The specific echocardiographic features of Uhl's anomaly reveal an enlarged and diffusely hypokinetic thin-walled right ventricular cavity with no apical trabeculation.[7] The tricuspid valve shows regurgitation with delayed valve closure and delayed diastolic pulmonary valve opening.[13] The pulmonary artery shows retrograde blood flow to the right ventricle during systole and the right atrium is dilated secondary to tricuspid valve regurgitation.[7] Additional defects described in association with Uhl's anomaly include dysplasia of the tricuspid valve, pulmonary atresia and persistent ductus arteriosus.[13]

The clinical signs and symptoms of Uhl's anomaly are quite variable; however, most Uhl's anomaly patients die in infancy or in utero, because the destruction of some critical parts of the heart. If the crista supraventricularis is destroyed, along with the free wall of the right ventricle, right ventricular failure is inevitable and ultimately fatal.[3,9] Such patients contribute significantly to the perinatal mortality of Uhl's anomaly.

The prognosis of Uhl's anomaly is poor but varies in relation to the extent of the destruction of the right ventricular myocardium and the heart conducting system. Successful cases have been reported of infants surviving surgery for critical ventricular arrhythmias due to Uhl's anomaly.[14–16] Starr, *et al*.[17] demonstrated that part or complete absence of right ventricular myocardium is itself insufficient to cause serious outcomes such as death. Such patients often survive with relatively little hemodynamic change in their infancy, and can live to an advanced age.[18]

Correct prenatal diagnosis, therefore, can be crucial in identifying patients who will require surgical intervention and specialist postnatal care. The infant we have documented in this report is currently a healthy nine year old boy; he has had no surgery and needs no regular medication for his cardiac condition. As with other congenital heart lesions, accurate prenatal diagnosis of Uhl's anomaly is important to enable optimisation of perinatal care and increase the chances of infant survival.

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