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CASE REPORT

Prenatal diagnosis of complete atrioventricular septal defect with truncus arteriosus

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Truncus arteriosus (TA) is a rare cardiac anomaly constituting less than 1% of all congenital heart defects. Its association with complete atrioventricular septal defect (AVSD) is extremely unusual and only 12 cases diagnosed postnatally or postmortem have been reported so far. We describe the first case of truncus arteriosus with AVSD to be diagnosed prenatally by fetal echocardiography. Copyright © 2007 John Wiley & Sons, Ltd.

KEY WORDS: fetal echocardiography; congenital heart disease; atrioventricular septal defect; truncus arteriosus

CASE REPORT

Screening ultrasound at 20 weeks in a 33-yr-old gravida 2 women revealed abnormal four-chamber view of the heart consistent with atrioventricular septal defect (AVSD) and mild bilateral cerebral ventriculomegaly. Fetal biometry was consistent with menstrual age and the amniotic fluid index was normal. Amniocentesis was performed and she was referred to our Institution for fetal echocardiography.

Fetal echocardiogram was performed using Phillips 5500 machine (HP, Andover, MA) with a 6–8 mHz transducer. There was normal visceral and atrial situs solitus and levocardia. Cardiac size was normal. The four- chamber view was abnormal with a moderate-sized primum atrial septal defect (ASD) and an inlet ventricular septal defect (VSD) (Figure 1). There was a common atrioventricular valve that bridged this defect. The systemic and pulmonary venous connections were normal. Both ventricles were balanced in size; the wall thickness and contractility was normal. Outflow tract views were abnormal with a single large artery overriding the VSD. The single trunk gave rise to both the aorta and the main pulmonary artery that divided into right and left pulmonary arteries—findings consistent with Truncus arteriosus (TA) Type 1 (Figure 2). There was no significant thickening of the truncal valve and the number of leaflets could not be discerned. The ascending aorta appeared normal in caliber. The arch views were difficult but there appeared to be a narrowing at the junction of transverse and descending portions of the aorta that raised suspicion for coarctation of aorta. There was a large ductus arteriosus.

Color flow and Spectral Doppler interrogation revealed the absence of atrioventricular valve insufficiency. The atrial and ventricular shunts were bi-directional. The truncal valve was competent without stenosis or insufficiency. The venous and arterial Doppler wave forms were normal. The fetal heart rate was 117 beats per min with a normal rhythm.

The family was counseled extensively. An amniocentesis revealed a male fetus with trisomy for chromosome 21.

The baby was born at 39 weeks of gestation by normal spontaneous vaginal delivery. A neonatal echocardiogram confirmed the findings of a complete AVSD and TA Type I. Atrioventricular valve attachments were noted to the crest of ventricular septum (Rastelli Type A). The right ventricle was noted to be mildly hypoplastic. In addition, a severe narrowing in the descending aorta consistent with coarctation was found. The patent ductus arteriosus was large with bi-directional shunt across it. Mild atrioventricular valve and truncal valve insufficiency was noted. Cytogenetic testing on peripheral blood confirmed Trisomy 21; no 22q11 microdeletions were detected. In view of the limited surgical options and the poor prognosis, the family opted for comfort care. Prostaglandin was discontinued and the neonate was discharged to hospice care.

DISCUSSION

TA is a congenital heart defect in which a common arterial trunk gives rise to the aorta, pulmonary arteries and the coronary arteries. It is a rare cardiac anomaly in itself constituting less than 1% of all congenital heart defects among live born infants. Similar frequency has been reported during fetal life (Allan *et al*., 1994). The association of TA with complete AVSD is exceedingly rare and only 12 cases of this association exist in the literature so far, *all* diagnosed postnatally (seven postmortem (Van Praagh and Van Praagh, 1965; Bharati *et al*., 1974; Ceballos *et al*., 1983; Butto *et al*., 1986; Kirklin and Baratt-Boyes, 1993) and five in the newborn live infants (Gumbiner *et al*., 1991; Trowitzsch *et al*., 1991; Sousa-Uva *et al*., 1994; Arai *et al*., 1995; Atik

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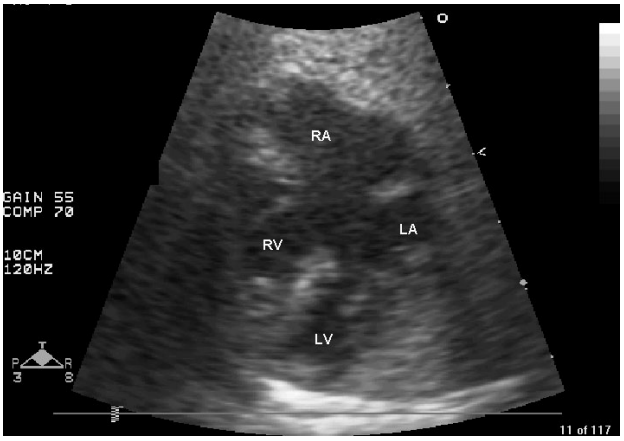

Figure 1—The apical four-chamber view of the heart in diastole showing a complete atrioventricular septal defect seen as a large defect at the crux of the heart

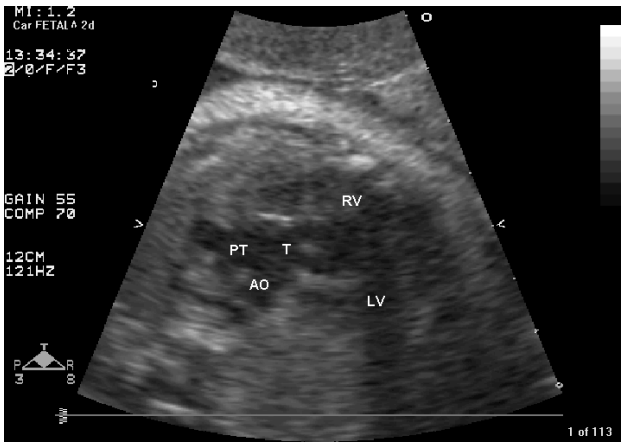

Figure 2—Outflow view showing a single semilunar valve and a single large trunk (T) giving rise to the aorta (AO) and the pulmonary trunk (PT) that divides into right and left pulmonary arteries

et al., 1999)). Prenatal diagnosis of AVSD with TA has not been reported to date and this is the first report of this association.

In a large multicenter series of 23 cases of TA diagnosed prenatally, (Volpe *et al*., 2003) eight had associated cardiac anomalies none of which included an AVSD. Similarly, no AVSD was reported among the 17 cases of TA diagnosed prenatally at Guy's Hospital, London (Duke *et al*., 2001). Other smaller series and individual reports of prenatal diagnosis of TA do not report this association as well.

Fetal diagnosis of AVSD with TA may be difficult, but careful screening of the four-chamber view and outflow tracts should help identify both components of this complex congenital heart disease. The diagnosis of TA was missed on routine obstetric screening ultrasound in our case perhaps due to failure or inability to visualize the outflow tracts. It is also likely that this 'second' defect was overlooked since an initial diagnosis of AVSD was already made. We cannot overemphasize the importance of incorporating visualization of cardiac outflow tracts in all cases for better diagnostic accuracy. It is also very important to 'look' for associated cardiac defects whenever a diagnosis of congenital heart disease is made.

Fetal diagnosis of AVSD is quite straightforward and includes an abnormal four-chamber view with a defect at the crux of the heart that is bridged by a common atrioventricular valve. As a result, the normal differential insertion of mitral and tricuspid valves is lost. The initial clue to a TA is finding a single semilunar valve and great artery that appear large and override the VSD. The differential diagnosis at this point should include TA and pulmonary atresia with VSD. The second semilunar valve and great artery are usually demonstrable in cases of other conotruncal anomalies such as tetralogy of Fallot, double outlet right ventricle and D-transposition of great arteries. Establishing the origin of pulmonary arteries from the common trunk confirms the diagnosis of TA. The pulmonary arteries are usually good sized in TA due to unrestricted pulmonary blood flow while in pulmonary atresia/VSD they are usually small and supplied retrogradely from arterial duct. Presence of abnormal semilunar valve with stenosis or insufficiency favors a diagnosis of TA rather than pulmonary atresia in which the aortic valve is usually normal. The pulmonary artery anatomy should be delineated in an attempt to classify TA into the four types as defined by Collett and Edwards (Colett and Edwards, 1949) but this may be a difficult task on a prenatal ultrasound.

While the outcome for AVSD (Crawford and Stroud, 2001; Frid *et al*., 2004) and TA (Brown *et al*., 2001; Thompson *et al*., 2001) as separate lesions is excellent, the prognosis for this combination of defects is dismal as can be deduced from the fact that there is only one other survivor with this heart defect in addition to our case. The diagnosis was made postmortem in seven cases mentioned above (Van Praagh and Van Praagh, 1965; Bharati *et al*., 1974; Ceballos *et al*., 1983; Butto *et al*., 1986; Kirklin and Baratt-Boyes, 1993). In other cases, the infants died during the newborn period before surgery was attempted—two neonates both with associated anomalies consistent with heterotaxy syndrome died in 12 h (ai *et al*., 1995) and 6 days (Gumbiner *et al*., 1991). There is one report of an infant (unrepaired) surviving until 6 months of age (Atik *et al*., 1999). More recently, surgical attempts to correct these defects have failed in one case (Trowitzsch *et al*., 1991) and there is a single report of successful surgical correction in an infant who was alive at the time of the report 3 months out from the operation (Sousa-Uva *et al*., 1994). Our patient at the time of this report was 2 months old and still alive despite the severe coarctation most likely due to continued patency of his ductus arteriosus.

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