Case Reports

Double-Outlet Single Ventricle and an Abdominal Vascular Mass: In Utero Diagnosis with Pathological Confirmation

Walter J. Duncan,1 David George,2 Wendelin Ezzat,2 Kirk Wallace,3 and Bob Van den Beuken2

Departments of 1Pediatrics, 2Pathology, and 3Radiology, College of Medicine, Saskatoon, Saskatoon, Canada

SUMMARY. A fetal echocardiographic scan was performed when routine prenatal ultrasound screening failed to identify four cardiac chambers. The scan showed a single ventricle with an associated circoid varicosity. Because of these anomalies, amniocentesis was suggested and trisomy 18 confirmed. The presence of major cardiac structural anomaly should prompt careful and specific review of all fetal anatomy to screen for syndrome identification and consideration of amniocentesis.

KEY WORDS: Fetal echocardiography — Syndrome identification

The in utero diagnosis of congenital heart disease has become common. In many large centers, the unborn with known cardiac pathology is provided with expectant therapy to ensure that cardiac decompensation and collapse do not occur unexpectedly after delivery. This approach also provides the parents with adequate time to prepare themselves, their families, and finances for the known incipient difficulties.

In many instances, cardiologists are presented with a known genetic diagnosis (e.g., Trisomy 21) and then asked to screen the fetal heart for abnormalities. Indeed, if one reviews the published information on the frequency of diagnostic categories based on fetal echocardiography [1, 3], atrioventricular canal defects rank among the most commonly diagnosed condition. The linkage between Trisomy 21 and atrioventricular canal is well known.

We recently had an opportunity to study a fetus with a suspected cardiac anomaly; we confirmed the defect and recommended amniocentesis.

Address offprint requests to: Dr. W.J. Duncan, Division of Cardiology, Children’s Hospital of Eastern Ontario, 401 Smyth Road, Ottawa, Ontario K1H 8L1, Canada.

Case Report

A 26-year-old North American Indian woman underwent routine prenatal ultrasound screening at 28 weeks of pregnancy. The technician performing the study was unable to obtain a four-chamber view that showed normal atrioventricular relations. The transverse arch could not be seen beyond the initial two branches and was thought to be interrupted (Fig. 1A). A vascular mass having an appearance of a leash of circular vessels was seen extending on both sides of the abdominal wall at the region of the umbilicus (Fig. 1B). Color mapping and pulsed Doppler recording confirmed turbulent continuous flow in the vessels. Visceral and renal ultrasonography was normal.

The cardiac features coupled with intrauterine growth retardation prompted a referral to the clinical genetics service. Amniocentesis confirmed Trisomy 18 and labor was induced at 31 weeks by dates. The child (a 1070-g female) survived 1 h—no resuscitation was performed. Autopsy confirmed multiple anomalies consistent with Trisomy 18.

The anatomical features (Fig. 2) identified by antenatal echocardiography were largely confirmed. There were also additional features including a tiny ventricular septal defect communicating to a vestigial left ventricle, mitral atresia, and severe hypoplasia of aorta and transverse aortic arch without interruption.
Fig. 1. (A) In utero view of heart showing single ventricular chamber, with the small aorta lying anterior to the pulmonary artery. Note the apparent discontinuity of aorta beyond the first two divisions. Ao, aorta; PA, pulmonary artery; V, ventricle; At, atrium. (B) The vascular mass is seen extending on both sides of the abdominal wall. Echo-free spaces represented aneurysmally dilated vessels.

Fig. 2. View of heart before (right) and after (left) sectioning. Note the close comparison of these frames with Fig. 1A. Abbreviations as in Fig. 1; VSD, ventricular septal defect; TV, tricuspid valve.

Fig. 3. Gross appearance of vascular mass extruded on both sides of the abdominal wall.

Discussion

As ultrasound technicians and physicians in many specialties become more adept at accurate in utero diagnosis of congenital heart disease, the prospects of survival for the compromised fetus and newborn are enhanced [4]. Most commonly, cardiac pathology is isolated and not associated with syndromic abnormalities. The presence of a cardiac defect should prompt a careful and expert review of all fetal structures to allow the best possible opportunity for syndrome identification [2, 5]. As in our case, in utero growth retardation is often associated with syndromes—it is not a common feature of isolated cardiac pathology except in the face of severe cardiac failure.

References

2. Crawford DC, Chapman MG, Allan LD (1985) Echocardio-