Foetal echocardiography: tool to predict the future of patients with congenital heart defects?

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Online publish-ahead-of-print 8 May 2008

This editorial refers to ‘Foetal echocardiographic assessment of tetralogy of Fallot and post-natal outcome † by F. Kaguelidou et al., on page 1432

Antepartum obstetrical ultrasonic evaluation is nowadays commonly used in order to detect congenital anomalies.1,2 As congenital heart disease is the most frequently encountered congenital anomaly, cardiac examination is of paramount importance to identify defects on time.

The advantage of foetal echocardiography is that ultrasound energy can be applied safely in an evolving fetus and that cardiac structures can be studied early in pregnancy (from 10 to 12 weeks by the vaginal approach, and from 16 to 18 weeks using the transabdominal approach).

Failures to diagnose congenital heart defects correctly are caused by multiple variables including ultrasound technology, sonographer experience, and mother- or foetal-related factors such as gestational age, foetal intrauterine position, or polyhydramnios. As congenital heart malformations are often associated with other cardiovascular and/or extracardiac malformations, extensive ultrasonic evaluation of the fetus is mandatory in case of a congenital heart defect.

In their interesting study, Kaguelidou and co-workers present the impact of prenatal diagnosis on postnatal outcome in a large series of patients prenatally diagnosed with either tetralogy of Fallot or pulmonary atresia with ventricular septal defects.3 In common with other studies, they report a high incidence of extracardiac malformations (46%), abnormal karyotyping (11%) and 22Q11 deletions (18%) in the embryos diagnosed to have either Fallot’s tetralogy or pulmonary atresia with ventricular septal defects.4,5 No relationship between chromosomal anomalies and postnatal outcome was established; the majority of pregnancies with chromosomal anomalies were terminated prematurely and the main reason for death of the liveborn children was the presence of extracardiac anomalies.

Impact of foetal echocardiography

Foetal echocardiography may play an important role in prenatal counselling. Kaguelidou and co-workers determined the characteristics and outcome of embryos diagnosed with tetralogy of Fallot or pulmonary atresia with ventricular septal defects. They studied 218 embryos, which is the largest number reported so far. Furthermore, they also evaluated the accuracy of identifying congenital heart defects by foetal echocardiography. In addition, foetal echocardiography was used to study anomalies of the pulmonary arterial tree. Essential in this study is that the results of the echocardiographic examination were the most important information used for prenatal counselling. It is demonstrated that in children with tetralogy of Fallot or pulmonary atresia with a ventricular septal defect, prenatally determined anatomy of the pulmonary arterial tree can be used to determine (normal size of pulmonary artery branches and presence of a main pulmonary artery) the possibility of surgical repair in the first year of life. The reliability of foetal echocardiography in diagnosing pulmonary artery anomalies was confirmed by comparison with postnatal echocardiography, angiography, surgical findings, or autopsy findings. Hence, this study emphasizes the important role foetal echocardiography may play in prenatal counselling as the results of the foetal echocardiographic examination were used to predict postnatal outcome.

Technological progress and foetal cardiac intervention

An accurate foetal echocardiographic examination is mandatory for reliable prenatal counselling. The ongoing advances in ultrasound technology may further increase sensitivity and specificity of foetal echocardiography.6 The current advancements in ultrasound technology enable the generation of dynamic three- and four-dimensional views of the beating foetal heart.7,8 Furthermore,
Foetal echocardiography facilitates the evaluation of the developing heart and identification of heart defects at a much earlier stage, in utero, thereby not only improving our insight into the mechanisms underlying congenital heart defects but also allowing a timely development of a patient-tailored treatment plan. Another intriguing consequence of early identification of congenital heart defects by foetal echocardiography is the opportunity for in utero interventions. In utero balloon dilation of aortic stenosis and hypoplastic left heart syndrome repair has already been performed successfully. It is likely that foetal cardiac interventions will improve the long-term outcome as cardiac anatomy and physiology are corrected at an early stage.

The elegant study presented by Kaguelidou and co-workers clearly demonstrates the impact of foetal echocardiography on management and outcome of patients with congenital heart defects. The authors are precise in describing foetal echocardiography, including present limitations such as insufficient image quality. Future studies are essential to define further the role of foetal echocardiography in prenatal counselling. Improvement in detection of complex congenital heart defects and guiding of in utero interventions will be a challenge for foetal echocardiography in the next decades.

Conflict of interest: none declared.

References