Impact of 1st trimester anomaly scan on un-natural history of CHD

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Prenatal prevalence of congenital heart defects (CHD) is higher in comparison to postnatal prevalence. True incidence is however unknown due to frequent intrauterine demise in early post-conceptual stages in fetuses with severe CHD and associated co-morbidity.

Objective: Analysis of spectrum of CHD, severity of associated abnormalities and outcome of fetuses diagnosed with CHD in first and second trimester screening (1TS and 2TS respectively).

Methods: Total of 510 fetuses diagnosed with CHD (127 in 1TS, 383 in 2TS) undergone ultrasound cardiac and anomaly scan and genetic assessment including karyotype analysis.

Results: Out of 127 fetuses diagnosed with CHD within 1TS (11+0 to 13+6 weeks of gestation) chromosomal and/or extracardiac anomalies were found in 85(67%) fetuses and CHD with univentricular circulation (UV) in 54(43%). Seven (6%) fetuses died in utero (IUD), pregnancy was terminated (TOP) in 108(85%) and only 12(9%) were born (11 of those with biventricular CHD). Similarly, out of 383 fetuses in 2TS (14+0 to 28+0 weeks of gestation) chromosomal and/or extracardiac abnormalities were found in 117(31%) fetuses and CHD with UV circulation in 50(13%). Four (1%) fetuses died in utero, TOP was in 115(30%) and 264(69%) were born (258 with biventricular outcome and 227 without associated co-morbidity).

There were significantly less chromosomal and/or extracardiac abnormalities (p<0.0001), CHD with UV outcome (p<0.0001), IUD (p=0.0027) and TOP (p<0.0001) in the group of CHD diagnosed within 2TS.

The frequency of atrioventricular septal defect (p=0.0139), hypoplastic left heart (p<0.0001), pulmonary atresia (p<0.0010) and tricuspid atresia (p=0.0097) was significantly higher in 1TS, while detection of transposition of great arteries was higher (p=0.0033) in 2TS.

Conclusion: We confirmed that 1TS screening has significant impact on outcome of pregnancies with CHD as more severe forms of CHD and higher co-morbidity result in increase of TOP. In 2TS transposition of great arteries and less severe forms of CHD are more likely to be diagnosed. These fetuses have better postnatal outcome due to more frequent biventricular circulation and less associated co-morbidities. In severe CHD with associated co-morbidity diagnosed in 2TS, families are more keen to continue with pregnancy as opposed to those diagnosed with similar lesions at 1TS.