Cardiac abnormality suspected at less than 9 weeks
Case report

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**Background**

CHD is a leading cause of infant mortality, with an estimated incidence of about 4–13 per 1000 live births\(^6\)–8. Between 1950 and 1994, 42% of infant deaths reported to the World Health Organization were attributable to cardiac defects\(^9\). Structural cardiac anomalies were also among the abnormalities most frequently missed by prenatal ultrasonography. To date the detection rate of major congenital abnormalities remains to be around 50%.

Cardiac examination at the first trimester of pregnancy requires special training. A well-trained obstetrician using high-resolution ultrasound equipment can assess the fetal heart at 11-13 weeks with a high degree of accuracy (around 95%)\(^1\).

Half of major fetal cardiac defects could be detected in the first trimester if NT and DV Doppler are used to select 2.7% of the general pregnant population for extended fetal echocardiography. NT of 3.5 mm is generally taken as the cut off for special cardiac scan.

**Case Report**

33 years old, Caucasian, BMI 21.6, in her first pregnancy and with no past medical history of significance, was sent for ultrasound scan because of early pregnancy bleeding. CRL was 20.1 mm (8+4 weeks) with positive fetal heart at 155/min.

However, with close look, considerable part of the heart was still not intra-thoracic (Figure 1-2). We suspected the possibility of ventral wall defect and ectopia cordis.

**Hence, we arranged a scan in 2 weeks time for re-assessment.** We rescanned her at 10+4 weeks (Figure 3-4), dated by our previous measurement. CRL was at the 8th centile (34 mm), the hear was completely intra-thoracic, but the fetus was severely hydroptic. There was obvious omphalocele, but regarded as physiological because of the early gestation. We decided to rescans her in a week time, and thus she would be legible for screening and give chance for clearer picture of the heart. The woman accepted the plan, and attended at 11+4 weeks (Figures 5-9). CRL was around the 10th centile, and still with severe hydrops, and NT was measured as 8 mm. The omphalocele increased in size. At the heart examination, there was obvious disproportion between the left and right ventricles with absence of flow throw the mitral valve. We put a diagnosis of HLHS. The woman was counselled for the possibility of major cardiac defect as the cause of hydrops fetalis, as well as the possibility of associated chromosomal abnormalities, namely trisomy 18, specially with the presence of large omphalocele. She opted for invasive procedure, and uncomplicated CVS was performed. The PCR confirmed trisomy 18 (Edward syndrome). The woman then opted for termination of pregnancy.

**Conclusion:**

Careful examination of the fetus at gestation as early as 9 weeks may give clue for developing fetal structural abnormalities.

First trimester fetal echocardiography is possible, but requires special training.

The association between trisomy 18 and HLHS has been evidenced by this case.