Service Priorities and Programmes

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Early Prenatal Screening and Diagnosis of Fetal Abnormalities

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Introduction

Late detection of fetal structural abnormality limits pregnancy options and may have medicolegal implications. Since July 2010, our department has been providing universal first trimester combined screening for Down syndrome which includes an ultrasound measurement of fetal nuchal translucency and blood test for serum markers. During the ultrasound examination, we also screen for fetal abnormalities.

Objectives

The aim of our study is to explore our performance on early detection of fetal abnormalities and the pregnancy outcomes of those with fetal abnormalities diagnosed at an early gestation.

Methodology

For prenatal diagnosis of fetal structural abnormalities, our department has arranged staff training, adopted latest ultrasound technology including 3/4-dimensional ultrasound and high linear frequency linear transducer (HFLT), conducted audits to explore new means for improvement and to better understand how we have performed. When a fetal abnormality is identified during scan for nuchal translucency by midwives or doctors, the pregnant woman is referred to our maternal fetal medicine (MFM) subspecialists for rescanning either before 14 weeks or at around 18-23 weeks. Chorionic villus sampling or amniocentesis is performed if indicated. All the pregnant women are counseled by our MFM subspecialists, and trained midwives of the MFM team. Joint counseling with paediatric cardiologists or surgeons is arranged if necessary.

Result

From 2010 to 2015, we detected 68 fetuses having structural abnormalities, excluding those with only thickened nuchal translucency, during ultrasound examination at gestation 11-14 weeks. Severity of the abnormalities varied from mild, such as club feet, to severe, such as anencephaly. 19 women (27.9%) had chorionic villus sampling with 5 showing chromosomal abnormalities, while 3 (4.4%) had

amniocentesis with all showing normal results. The overall diagnostic yield was 22.7%. 44 women (64.7%) had termination of pregnancy, 22 (32.4%) had delivered the babies, 2 (2.9%) had silent miscarriage. For those with termination in our unit, abnormalities were confirmed in all abortuses. In 12 (17.6%) cases, the suspected abnormalities, like prominent renal pelvis and prominent bladder, were transient and not confirmed after birth. In conclusion, significant fetal abnormalities could be detected by trained doctors and midwives in first trimester, allowing the pregnant women to choose further genetic testing or other management options at an earlier gestation after adequate counseling.