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Guidelines For First Trimester Ultrasound Examination: Part II

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1 INTRODUCTION

Recent advances in ultrasonographic technology enable detailed studies and evaluation of the rapidly developing embryo in vivo. It is at present the most accurate and reliable method for the evaluation of first trimester pregnancies and their complications. However, the precise role of ultrasound in the first trimester is still in evolution, mainly because of rapid development and availability of newer generations of ultrasonographic equipment, which enables better and earlier visualization of embryonic structures.

2 AIMS

Part II of this guideline examines:

- the current status of first trimester ultrasound examinations in diagnosing and screening for fetal abnormalities
- the advantages, disadvantages and the cost-effectiveness of offering a routine first trimester ultrasound examination
- safety issues

3 DIAGNOSIS AND SCREENING FOR FETAL STRUCTURAL ABNORMALITIES

With the developments of higher resolution ultrasound machines, much more detailed morphological examination of developing fetuses can be achieved in the first trimester. The prenatal diagnoses of a long list of different structural abnormalities have been reported during the first trimester of pregnancy[1]. When used as a routine screening method between 11-14 weeks of gestation among low risk populations, the detection rates for structural abnormalities have been reported to range from 22.3%[2] and 59.0%[3] to 64.7%[4]. A consistent finding of all studies was that a significant number of additional fetal abnormalities were detected in subsequent follow up studies at 18-20 weeks and this ranged from 17.6%[4] and 22%[3] to 48.2%[2]. Therefore, even if an early fetal anomaly scan were offered, the second trimester scan should not be abandoned.

Because of the small size of the fetus at this gestation, demand on the quality of the ultrasound machine and experience of sonographers are much higher than that required for an 18 – 20 weeks ultrasound examination for fetal anomalies. It was suggested that adequate sensitivity in screening for major malformations by early ultrasonography requires a learning curve of 3-4 years[5].

Furthermore, the normal morphology of the fetus changes with gestational age due to embryological development. Sonographers who perform first trimester ultrasound examinations, therefore, should familiarize themselves with such normal embryological variations. In this regard, herniation of the fetal bowel at the umbilical insertion is normal before 11 weeks of gestation. This physiological herniation should not be mistaken as an omphalocoele. Another common mistake is the diagnosis of hydrocephalus in the first trimester, when the cerebral ventricles are normally very prominent. These mistakes will obviously cause unnecessary anxiety to parents and may even lead to unwarranted termination of pregnancies.

Despite technical difficulties, early prenatal diagnosis of fetal abnormalities in the first trimester remains attractive. It is a common belief that earlier diagnosis of major abnormalities enables earlier intervention such as induced abortions, and therefore less psychological trauma to the parents. However, this supposed advantage must be viewed from other perspectives. It is known that a significant proportion of abnormal fetuses abort spontaneously in the late first trimester and early second trimester. Earlier diagnosis will therefore expose a group of women who will have aborted spontaneously to the unnecessary and painful dilemma and guilt of choosing a therapeutic abortion. Studies on spontaneous abortions in wanted pregnancies did not show any difference in the pattern and intensity of grieving following first trimester miscarriages compared with second trimester miscarriages[6].

At present, the use of first trimester ultrasound examination as a routine to screen for fetal structural abnormalities is not recommended except in research settings or in highly specialized centers. If fetal abnormalities are detected or suspected during a first trimester ultrasound examination performed for other purposes, referral to a tertiary centre for further evaluation is advisable.

4 NUCHAL TRANSLUCENCY MEASUREMENT IN SCREENING FOR FETAL CHROMOSOMAL ABNORMALITIES

The value of using nuchal translucency (NT) measurement as a screening tool for fetal Down syndrome had been the focus of heated debate over the last decade.

Initially, a fixed cut-off was proposed. Early studies examining the implementation of NT screening for fetal Down's Syndrome between 8 and 14 weeks and using cut-offs of 2.5mm to 3mm, reported a wide variation in sensitivities from 33% to 90%.[7] Since the size of NT is also known to increase with gestational age, a simple cut-off is now more commonly replaced by individualized risk assessment adjusted for gestational age and maternal age[8].

Critics were concerned about the large variation in reported detection rates, ranging from 29% to 91%[9]. Such large variation, however, was most likely a result of significant differences in methodology and training of sonographers[10]. Furthermore, the measurement of the NT needs a good quality ultrasound machine and the methodology should be standardized. A proper sagittal view is essential but not adequate, as care must be taken to distinguish between the fetal skin and amnion. Hyperextension or hyperflexion of the fetal spine will result in over- and under-estimation respectively. Placement of the calipers needs to be standardized and has been found to account for a large part of the inter- and intra-observer variation. Specific training and practice in NT measurements are needed to achieve consistent results.

Using standard methodology and trained sonographers, Snijders et al in a large multicenter study under the auspices of the Fetal Medicine Foundation involving 96,127 singleton pregnancies, including 326 with trisomy 21, reported a detection rate of 77% at a 5% false positive rate[11], which is comparable, if not better, than that of second trimester biochemical screening test. Therefore, the reliability of a NT screening program relies on the strict adherence to a standard protocol[10].

Amongst trained sonographers, NT measurement has been shown to be reproducible, and the inter- and intra-observer errors were 0.62mm and 0.54mm in 95% of cases respectively[12]. The size of NT is also known to increase with gestational age[13]. In clinical practice, NT measurement is usually combined with maternal age, gestational age, and previous history of chromosomal defects to calculate the individualized risk of fetal Down syndrome.

More recently, NT has also been combined with other methods to refine the estimation of patient-specific risk of Down syndrome. If combined with first trimester biochemical screening using PAPP-A and free β-hCG, it is possible to achieve a sensitivity of 90% for a 5% false positive rate[14] which is further improved to 97% for the same false positive rate if absence of fetal nasal bone is factored into the equation[15].

Other than trisomy 21, increased NT is also associated with other chromosomal abnormalities, major defects of the heart and great arteries, and a wide range of skeletal dysplasias and genetic syndromes[10].

There are currently many effective screening methods for fetal Down syndrome, and NT is one of them. However, whether screening for Down syndrome should be offered and which test to be used depends on the desire of pregnant women, availability of appropriate equipment, and the experience of the clinicians and sonographers. If one decides to use NT as a screening test, one must ensure that such test is to be performed only by trained sonographers using good quality ultrasound machines following a standardized protocol within the appropriate gestational period.

When conducted according to accepted standards of quality, first trimester NT measurement is an effective screening method for fetal chromosomal abnormalities. Such a test, however, should only be performed after proper patient counseling.

5 THE ADVANTAGES, DISADVANTAGES AND COST-EFFECTIVENESS OF OFFERING A ROUTINE FIRST TRIMESTER ULTRASOUND EXAMINATION

First trimester ultrasound examination can confirm the presence of a live intrauterine pregnancy as early as six weeks in normal pregnancies. It can also exclude early pregnancy complications. Many women find this reassuring.

Furthermore, routine ultrasound in early pregnancy appears to enable better gestational age assessment and earlier detection of multiple pregnancies[16]. Accurate gestational age estimation has been shown to decrease the rate of induction of labour for apparent post-term pregnancies and to reduce average hospital stay. However, whether this could lead to a reduction in perinatal mortality has yet to be proven.

The role of using first trimester scans for screening of fetal structural abnormality has been discussed above. Although controversial, it is unlikely that first trimester ultrasound examination could totally replace second trimester ultrasound examinations in the detection of fetal abnormalities.

In summary, if resources allow, a routine first trimester ultrasound examination can be offered.

6 SAFETY ISSUES

Ultrasound is a form of energy and therefore is capable of inducing tissue effects, of which temperature rise and cavitation are most well known[17-18]. Although bioeffects might be beneficial, significant hazardous biological effects of ultrasonic exposure have been demonstrated by in vitro and animal studies, including tissue heating, cellular alteration, teratogencitiy and changes in fetal biometry[19-20]. On the contrary, most epidemiological and population-based studies in human, which involved mostly B-mode examinations, have failed to demonstrate any significant adverse fetal effects associated with prenatal sonography[21]. Many international professional bodies, including the International Society of Ultrasound in Obstetrics and Gynecology, believe that the use of B-mode and M-mode prenatal ultrasonography, due to its limited acoustic output, appears to be safe for all stages of pregnancy[22].

On the other hand, Doppler ultrasound is associated with much higher bioeffects, especially when applied to a very small region of interest[23]. These examinations should be used in first trimester only if clinically indicated, which in normal clinical practice is rare.

7 RECOMMEDATIONS

7.1 **At present, the use of first trimester ultrasound examinations to screen for fetal abnormalities is not recommended except in research settings.**

7.2 **If resources allow, a routine first trimester ultrasound examination for estimation of gestational age and for earlier diagnosis of multiple pregnancy can be offered.**

7.3 **The use of NT for screening of chromosomal abnormalities should only be performed after proper counseling.**

7.4 **Colour Doppler and pulsed Doppler examinations should be avoided in the first trimester as far as possible.**

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This guideline was produced by the Hong Kong College of Obstetricians and Gynaecologists as an educational aid and reference for obstetricians and gynaecologists practicing in Hong Kong. The guideline does not define a standard of care, nor is it intended to dictate an exclusive course of management. It presents recognized clinical methods and techniques for consideration by practitioners for incorporation into their practice. It is acknowledged that clinical management may vary and must always be responsive to the need of individual patients, resources, and limitations unique to the institution or type of practice. Particular attention is drawn to areas of clinical uncertainty where further research may be indicated.