FOCUS: WOMEN'S HEALTH APRJUN 2013

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First Trimester Screening & Chromosomal Disorders

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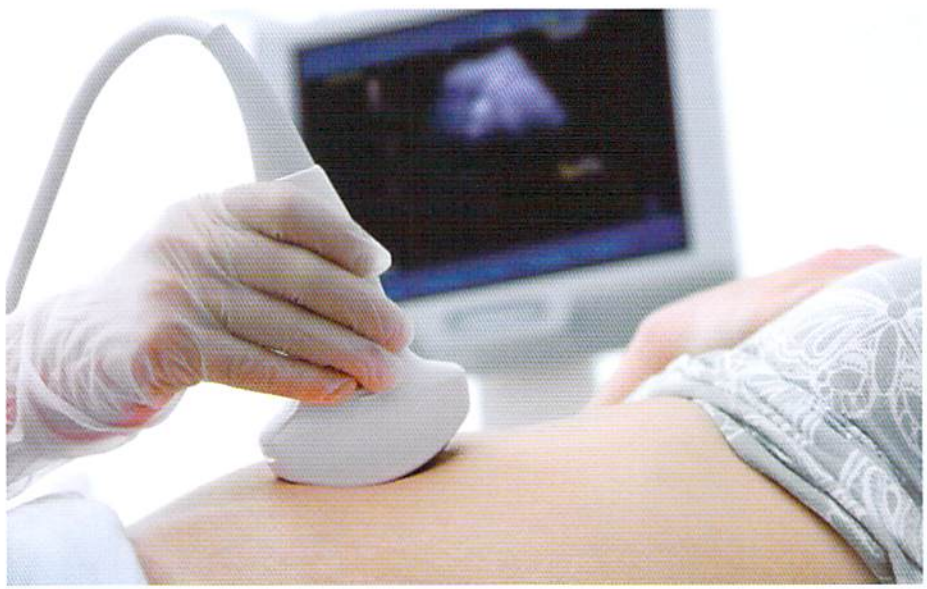
Pregnant patients above 35 are at higher risk of chromosomal disorders like Down syndrome (Trisomy 21), Patau syndrome (Trisomy 13) and Edward syndrome (Trisomy 18). In the past, age alone was used as a screening tool for prenatal diagnostic procedures like amniocentesis. The cut-off age then for indication for prenatal diagnostic procedures was previously 40 years of age and this gradually dropped to 38, 37 and then 35 over the years. However, using age alone as the sole cut-off for prenatal diagnostic testing is suboptimal as the detection rate is much lower for a given testing rate.

Now with **First Trimester Screening - FTS** (includes age, ultrasound measurements and serum tests) as a standard for the last decade, age is no longer the only factor in the calculation of the risk for the decision for the need for prenatal diagnostic procedures. This combination of screening has improved the accuracy of detection to around 95% compared to only 30% using age solely as a criterion, for a diagnostic testing rate of 2.5% to 5% for the population.

Incidence of fetuses affected with chromosomal disorders in Singapore

In a study of all Singapore births, (Tan KH et al. Singapore Med J 2005; 46(10): 545-552), the incidence of fetuses affected with chromosomal disorders was 4.35 per thousand births of which Down syndrome was 1.88 per thousand births. In terms of risks of chromosomal disorders, the 45-49 years age group (38.0/1,000 live births) showed a much higher (18.3-fold) risk than the 25-29 years age group (2.1/1,000 live births). Education of the population is necessary to ensure that mothers and the community are aware of the optimal age of motherhood in terms of such risks.



Nuchal Translucency (NT) of the fetus

One of the most important measurements is that of Nuchal Translucency (NT) of the Fetus. There is a normogram that has been created by the Fetal Medicine Foundation, UK which computes the risk of chromosomal disorders according to the measurement of NT in mm. In general, the thicker the measurement, the higher the risk of chromosomal disorders. A thickened NT also sheds light on the possibility of an increased risk for fetal death, perinatal loss, chromosome disorders like Turner syndrome and genetic diseases, including some cases of alpha thalassaemia. An increased NT is also associated with many structural malformations such as cardiac defects in the fetus.

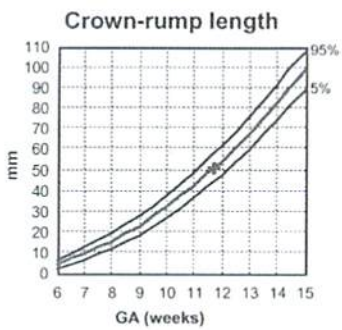
Patient Profile

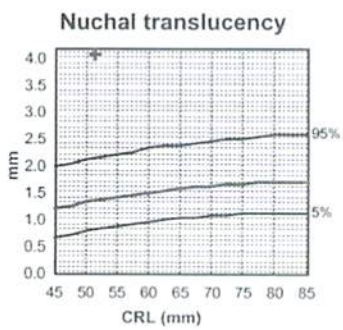
Mdm A, a 35-year-old lady had two previous first trimester miscarriages. She has no surgical or family history of note and was a non-smoker. She was booked at six weeks amenorrhea. She had ultrasound dating at 6.6 weeks and gestation corresponded to dates. She was offered first trimester screening for Down syndrome and she was keen.

Results

FTS was performed at 12 weeks gestation:
Crown-Rump Length CRL = 52 mm (equals to 11 weeks plus gestation)
Fetal heart activity present. Nuchal translucency = 4.1 mm (high)

Background risk of Down syndrome (Trisomy 21) for maternal age of 35 years = 1: 210
Adjusted risk of Down syndrome (Trisomy 21) for this patient = 1: 5
Background risk of Trisomy 13 + 18 for maternal age of 35 years = 1: 362
Adjusted risk of Trisomy 13 + 18 for this patient = 1: 9



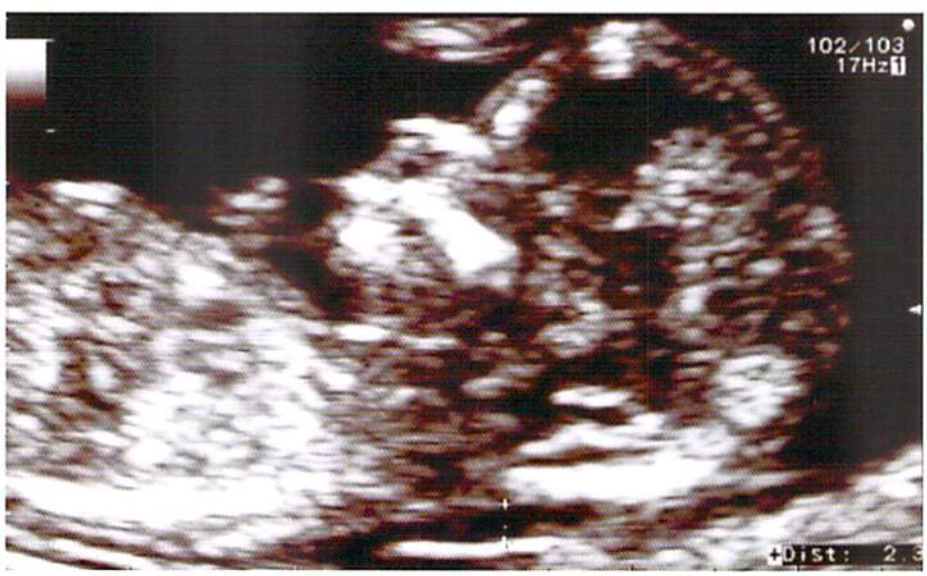


Counselling

Mdm A was counselled regarding the high risk of chromosomal abnormalities (1:5 for Down syndrome). With increased NT, there was also increased risk of structural abnormalities and increased risk of genetic syndromes.

The patient was offered fetal karyotyping namely, **chorionic villus sampling (CVS)** or **amniocentesis**. She was keen for amniocentesis and she underwent amniocentesis at 16 weeks gestation. The scan just before amniocentesis showed Cervical Skin 5.3 mm & Femur Length 16.6 mm both sides (3rd centile). The result of karyotyping revealed 47, XX, +21 (Down syndrome).

Mdm A was counselled on the results of Down syndrome and the options (continuation or termination). She opted for mid-trimester termination of pregnancy (MTPT) at 18 weeks gestation.



▲ An example of NT measurement - 2.3 mm

Referral for FTS

It is important for GPs to refer pregnant patients early for FTS as it can only be effectively performed from 11 weeks gestation to 13 weeks + 6 days (corresponding to Crown-Rump Length or CRL of 45 mm to 84 mm).

It is thus essential for the pregnant patient to seek care earlier and for women to come early to see the doctor if they suspect they are pregnant. This would ensure that their gestation can be accurately dated and properly counselled for FTS to be performed from 11 weeks to 13 weeks + 6 days.

Conclusion

FTS improves the accuracy of screening for Down syndrome and other chromosomal disorders. It is important that doctors refer patients early for the screening test and patients avail themselves early in pregnancy.

GP CONTACT
GPs can call for appointments through the Specialist Outpatient Clinic Appointment Centre at 6294 4050.