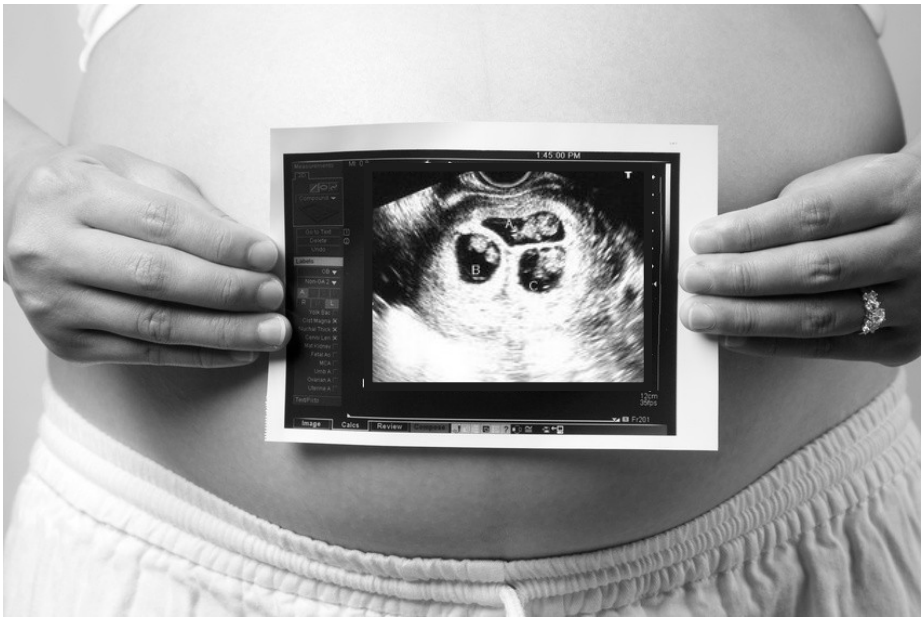
Screening for Down syndrome (Trisomy 21) in multiples



Nowadays most women will book early with a Lead Maternity Carer (LMC) once they are pregnant, particularly if a multiple pregnancy is identified on the first scan. Initial blood tests will be arranged, checking for a range of infections and immunity, which are important to the developing baby. More recently all pregnant women are also being offered an HIV test.

All of this can be overwhelming, but in addition to these tests there is also the question of screening for congenital abnormalities. This includes genetic conditions such as Down syndrome and structural abnormalities such as spina bifida.

What is Down syndrome?

Down syndrome is one of the most common causes of intellectual handicap in the developed world. It is due to a genetic abnormality which can be diagnosed on testing cells from the baby. People with Down syndrome can have associated physical problems and the average life expectancy is 60 years. The majority of families would like the opportunity to know whether a baby has this condition, as it may alter the choices which are made in the pregnancy.

Multiple pregnancy does not increase the risk of a particular baby having Down syndrome.

In multiple pregnancy where the babies are non-identical (the majority), each baby will have their own risk. In the case of twins, the risk for the woman of having one baby with Down syndrome will be doubled, compared to a mother the same age with a singleton, as each of her twins has the same risk - just as buying two lottery tickets doubles your chances of winning.

However, having a multiple pregnancy where the babies are identical, the risk for the woman of having a baby with Down syndrome is about the same as a singleton as the babies will have the same genetic make-up. If one baby has a genetic abnormality, though, they will almost certainly both have it.

Screening for Down syndrome

Screening for Down syndrome has been available in New Zealand, in one form or another, since 1968. The main test used over the last 10 years has been an ultrasound scan performed between 11 weeks, and 13 weeks and 6 days gestation. This scan measures a fold of tissue at the back of the baby's neck: the Nuchal Translucency. The Nuchal Translucency scan can identify some major structural abnormalities, though the scan at 18-20 weeks is more accurate. More recently, levels of chemicals (known as 'analytes') in the mother's blood have also been used to screen for Down syndrome. These analytes can also screen for placental problems, some types of spina bifida and rare genetic disorders.

The ultrasound scan and blood tests also screen for two other genetic conditions: trisomy 13 and trisomy 18. These conditions are ultimately lethal to the baby.

Screening tests are safe for mother and baby and can give the woman an estimate of the risk of having a baby with Down syndrome. If the risk is high, the woman can choose to have a diagnostic test. The diagnostic tests involve taking cells from the placenta (Chrorionic Villus Sampling, CVS) or the amniotic fluid (amniocentesis). Both carry a risk of miscarriage of approximately 1%.

Nation-wide Down syndrome antenatal screening programme

On the 8th February 2010, the National Screening Unit launched a New Zealand-wide Down syndrome antenatal screening programme which is free to all New Zealanders. It is different to the previous programme, as it includes the Nuchal Translucency scan and a blood test for maternal analytes at the same part of the pregnancy.

The Nuchal Translucency scan is done at 11 to 13+6 weeks gestation and the blood test at 9 to 13+6 weeks gestation. The scan result is faxed by the person doing the scan to the appropriate laboratory (in Auckland or Christchurch) and once the woman has had her blood test, the two results are combined to give an overall risk for Down syndrome. This result is then given to the woman's LMC, who informs the woman of the result.

In the case of multiple pregnancy, the tests can still be performed. The woman will need to discuss the results with her LMC.

If the result is high risk the option of diagnostic tests will still be available to women with multiple pregnancy. It is important that the woman sees an Obstetrician and Gynaecologist with experience in this area, ideally prior to proceeding with a diagnostic test. The diagnostic tests CVS or Amniocentesis

can distinguish between individual babies. However, if one baby has Down syndrome and one or more do not, the decision-making can become quite difficult.

Summary

In summary, screening for Down syndrome is available in New Zealand for women with multiple pregnancy. It is important to understand that screening does not provide certainty, but a *risk* for a condition. In the case of a high risk result, it is important that an Obstetrician and Gynaecologist with experience in this area oversees the counselling and planning.

Information leaflets

Following are two useful leaflets from the National Screening Unit regarding the new programme:

http://www.healthed.govt.nz/uploads/docs/HE2256.pdf

http://www.healthed.govt.nz/uploads/docs/HE2258.pdf

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Preventing premature labour (Website review)

By Terri-Lynn Mitchell

As you are probably aware, one of the major risks of a multiple gestation is that of premature labour. If you want to know more about how to maintain your pregnancy for as long as possible, then aside from advice from your doctor, LMC or obstetrician, the KeepEmCookin website, at www.keepemcookin.com, is a great place to find out more.

This website contains useful advice, suggested books, online resources and the latest research on preventing premature labour (a LOT of research!).

To boost morale, there are a range of forums, with topics including coping with bedrest—when I did a search using the keyword 'twins', I found a lot of personal stories of women expecting twins, and on bedrest to prevent premature labour.