Queensland Centre for Mothers & Babies

Making decisions about the 'nuchal scan'

While all pregnant women should be offered the combined first trimester screening (CFTS), commonly known as the 'nuchal scan', it is important to know that you can decide whether or not you want to have this test.

What is the combined first trimester screening test?

The CFTS test is performed in pregnancy to estimate the chance that unborn baby has Down syndrome (or trisomy 21), trisomy 18 and trisomy 13. Down syndrome, trisomy 18 and trisomy 13 are chromosomal abnormalities.

The CFTS test includes an ultrasound scan (done between 11 and 13 weeks+6 days gestation) and a maternal blood test. The ultrasound is used to measure the thickness of the nuchal translucency (explained below). The blood test is used to measure two proteins in the mother's blood - free ß-hCG and PAPP-A (also explained below). The ultrasound may also detect physical abnormalities that are not part of the CFTS test.

The CFTS test is a screening test. This means that it can tell you whether there is a high or low chance that your unborn baby has a chromosomal abnormality. It cannot confirm or exclude a chromosomal abnormality. Having the CFTS test does not increase your chance of having a miscarriage.

All women have a 3% chance of having a baby born with an abnormality (that is, 3 out of every 100 women) [1]. This means that 97% of women (that is, 97 out of every 100 women) have a baby without an abnormality.

What is a chromosomal abnormality?

Every cell in the human body contains 23 pairs of chromosomes. Chromosomes are the packages that contain our genetic material (or DNA). Normally we have 22 pairs of chromosomes numbered 1 – 22 plus one pair of sex chromosomes (XX for females, XY for males). See Figure 1.

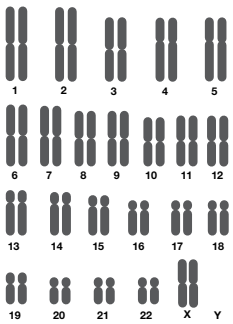

Figure 1.
A chromosomally normal female - 22 pairs of chromosomes and two X chromosomes

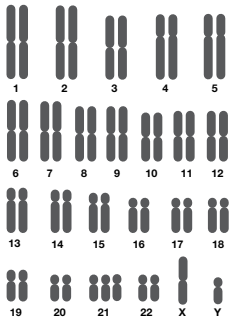

Figure 2.
A male with Down syndrome – three chromosome 21s, one X and one Y chromosome

A chromosomal abnormality is when there is a change in the number or structure of the chromosomes. For example, Down syndrome is the result of a having three chromosome 21s (see Figure 2).

What is Down syndrome?

Down syndrome is the most common chromosomal abnormality in newborns. The chance of a woman having a baby with Down syndrome increases with age, however young women can also have a baby with Down syndrome.

All individuals with Down syndrome have an intellectual disability, which can range from mild to severe [2, 3]. It is not possible to determine the level of intellectual disability from any tests in pregnancy. Approximately 50% of babies with Down syndrome are born with a heart defect and approximately 7-11% are born with a

gastrointestinal defect that may need surgery [2-5]. Some people with Down syndrome will complete school, gain work, form friendships and live semi-independently [2, 3]. Others, who are more severely affected, will need constant care. There is no cure for Down syndrome, however childhood interventions (e.g. physiotherapy and special education) can be beneficial in supporting an individual to meet his/her maximum potential [2, 3].

What is trisomy 18 and trisomy 13?

Trisomy 18 and trisomy 13 are less common than Down syndrome. Trisomy 18 is the result of having three chromosome 18s and trisomy 13 is the result of having three chromosome 13s. Women pregnant with a baby with trisomy 18 or 13 commonly miscarry early in pregnancy. Babies born with trisomy 18 or 13 usually only live for a short time after birth (due to severe abnormalities), however rarely a baby with trisomy 18 will survive longer than 12 months. The chance of a woman having a baby with trisomy 18 or 13 increases with age.

How is the chance of Down syndrome calculated?

The chance of Down syndrome is calculated using [6]:

- The woman's age
- The pregnancy gestation
- The nuchal translucency thickness, and
- The level of free ß-hCG and PAPP-A in the mother's blood.

The nuchal translucency is a pocket of fluid at the back of the unborn baby's neck that can be seen around the 12th week of pregnancy. This fluid is reabsorbed later in pregnancy, which is why the ultrasound is done between approximately 11 and 13 weeks+6 days gestation. A thicker nuchal translucency indicates a higher chance of a chromosomal, structural or other genetic abnormality, however some babies with a thick nuchal translucency in pregnancy will be born healthy. Not all babies with a chromosomal, structural or other genetic abnormality will have a thicker nuchal translucency in pregnancy.

Some care providers also examine the unborn baby's nose bone. If the nose bone does not appear bright on ultrasound, the chance of Down syndrome is increased.The nose bone measurement is new to CFTS testing, therefore only some doctors and sonographers (a person trained to perform ultrasounds) are accredited to do this. The assessment of the nose bone can only be incorporated into the calculation of Down syndrome if the doctor/sonographer is accredited to do this. Some women assume that the reporting of 'nasal bone present' indicates that this has been part of the risk calculation when in fact this is often not the case.

How is the chance of trisomy 18 and 13 calculated?

The chance of trisomy 18 and 13 is calculated using the same information as for Down syndrome, however the calculation for trisomy 13 also takes into account the baby's heart rate [7].

What can the CFTS test tell me?

The CFTS test result will be presented as a 1 in X number. This number is the chance that your baby has a chromosomal abnormality, and will either fall into the high or low risk category:

- High risk means that the chance is greater than 1 in 300 (i.e., between 1 in 2 and 1 in 300).
- Low risk means that the chance is less than 1 in 300.

How accurate is the CFTS test for Down syndrome?

The CFTS test identifies 87% of babies with Down syndrome [6]. This means that approximately 13% of babies with Down syndrome will be 'missed' by the CFTS test.

If the nose bone is also examined, the CFTS test identifies 97% of babies with Down syndrome and 'misses' approximately 3% of babies with Down syndrome [6].

Regardless of whether the nose bone is examined or not, 5% of pregnancies where the unborn baby does not have Down syndrome are placed in the high risk category [6]. This is called a false positive result.

What is the cost of the CFTS test?

The out-of-pocket cost varies significantly between centres.

What are my options if I am high risk?

If you get a high risk result, you can choose whether or not you want further testing. The following two options provide more accurate information regarding Down syndrome or other chromosomal abnormalities:

- Non-invasive prenatal testing (NIPT)
- CVS (chorionic villus sampling) or amniocentesis (sometimes shortened to 'amnio')

What is non-invasive prenatal testing?

Like the CFTS, NIPT is a screening test. This means that it can tell you whether there is a high or low chance that your unborn baby has a chromosomal abnormality [8]. It cannot confirm or exclude a chromosomal abnormality.

NIPT can be performed after 10 weeks of pregnancy and involves the woman having a blood test. NIPT looks at the baby's genetic material in the mother's blood. Having NIPT does not increase your chance of having a miscarriage.

How accurate is NIPT for Down syndrome?

NIPT identifies more than 99% of babies with Down syndrome [8]. This means that less than 1% of babies with Down syndrome will be 'missed' by NIPT [8].

Less than 1% of pregnancies where the unborn baby does not have Down syndrome are placed in the high risk category (false positive) [8].

What is the cost of NIPT?

The cost of NIPT is continually decreasing. In 2013 in Queensland, the out-of-pocket cost was $750 or more. There is currently no Medicare or health insurance rebate. You can ask your care provider about where to access NIPT.

What is amniocentesis and CVS?

Amniocentesis and CVS are diagnostic tests. This means that they can confirm or exclude a chromosomal abnormality. These tests are offered to women who receive a high risk result from CFTS test or NIPT. Amniocentesis and CVS are currently the only ways to confirm or exclude a chromosomal abnormality.

Both of these tests involve having a needle inserted through the woman's abdomen and into the uterus. In CVS (chorionic villus sampling), small pieces of the placenta are removed and the cells are tested. Some centres will do a CVS transcervically (by inserting a thin tube through the vagina and cervix to reach the placenta). In amniocentesis, a small amount of amniotic fluid (the fluid around the baby) is removed and the cells are tested.

CVS is usually performed between 10 – 13 weeks of pregnancy [9]. Amniocentesis is usually performed after 15 weeks completed weeks of pregnancy [9]. Some centres will only perform a CVS after 11 weeks and an amniocentesis after 16 weeks. Both tests increase the chance that a woman will have a miscarriage:

- 1% of women (or 1 in 100 women) will miscarry due to a CVS

- 0.5% of women (or 1 in 200 women) will miscarry due to an amniocentesis

Some women choose to have an amniocentesis or CVS without having the CFTS test or NIPT first.

What are my options if my baby has an abnormality?

If your unborn baby has a chromosomal abnormality, you can choose to continue with the pregnancy and keep the baby or give the baby up for adoption, or you can choose to terminate the pregnancy.

How can I decide whether I want the CFTS test?

Before you make your decision, you might like to consider whether you want to know the information provided by the CFTS test. You might also like to consider what you will do with the information.

There are many reasons why women decide to have or not have the CFTS test. Some women have the test because they want reassurance about their pregnancy. Others have the test so that they can make decisions about amniocentesis or CVS and about continuing or terminating the pregnancy. Other women do not have the test because they would rather not know this information in pregnancy.

It can help to think about:

- How you would feel if you received a high risk result
- Whether you think you might consider having an amniocentesis or CVS
- How you might feel if your baby had a chromosomal abnormality
- How you feel about continuing or terminating a pregnancy for a chromosomal abnormality
- Whether you want to be able to prepare for a baby with a chromosomal abnormality
- Whether you want the option of terminating the pregnancy for a chromosomal abnormality

Other tips:

- Take your time to make a decision
- Think about what you feel are the pros and cons of the CFTS test
- Consider your personal beliefs and values
- Seek support from people around you – partner, family and friends
- Ask your care provider questions
- If you choose to have the CFTS test, it can be helpful to talk with your care provider about how you are going to receive your result before you have the test.

What if I choose not to have the CFTS test?

If you choose not to have the CFTS test, you can choose whether or not you want to have a first trimester ultrasound. This can be performed without being given information about the chance of your unborn baby having a chromosomal abnormality. It is important to know that the first trimester ultrasound may detect other physical abnormalities that can be seen by the sonographer/doctor (e.g. limb or brain abnormalities, etc). If you choose not to have the CFTS test, your pregnancy care will continue as usual.

References are available from: http://www.qcmb.org.au/media/pdf/nuchalref.pdf