Why has prenatal screening been offered to me?

Prenatal screening is offered to all pregnant women because of the small chance that a pregnancy has a chromosome condition or other physical condition. Together, these fetal conditions can sometimes be referred to as fetal abnormalities or anomalies.

While most women will have healthy babies, in about 1 in 25 pregnancies (about 4%) a baby will be born with a condition that may require medical attention. These conditions vary a lot, from very mild to very severe, and can affect physical and/or intellectual development.

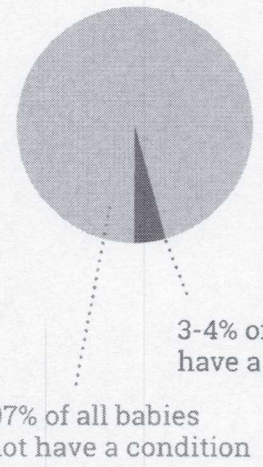


Figure 1: Most babies are born healthy.

Prenatal screening tests are used to identify those pregnancies with a higher chance of being affected by such conditions. No single test checks for all fetal conditions.

You can decide to have screening:

- because you want to know more about your pregnancy
- to prepare yourself for a baby with a condition
- so you have the option of terminating an affected pregnancy.

There are different types of tests available and they vary in how accurate they are, when they are performed and what information they give.

All prenatal screening is your choice. You can decide not to have any prenatal screening.

Chromosome conditions

For healthy development, humans need 46 chromosomes, or 23 pairs. Any extra or missing chromosomes, or any change in the structure or arrangement of the chromosomes, can affect normal development.

The chance of giving birth to a baby with a chromosome condition decreases as the pregnancy develops. This is because many pregnancies affected by chromosome conditions will miscarry naturally. It is not possible to predict which pregnancies will miscarry.

Down syndrome (trisomy 21)

Down syndrome is the most common chromosome condition found during pregnancy. On average it affects 1 in every 400 pregnancies and is caused by an extra copy of chromosome 21.

The chance of giving birth to a baby with Down syndrome increases with a woman's age, but pregnancies of younger women can also be affected (Figure 2).

People with Down syndrome have characteristic physical, medical and intellectual features. Most have some degree of intellectual disability, yet are able to participate in school, work and social life. Some can be severely intellectually disabled and require full time care. Babies with Down syndrome are also more likely to have problems with their heart and digestive system. It is not possible to predict the level of disability during pregnancy.

There is no cure for Down syndrome but early intervention, such as individualised educational programs, can support development.

For information see Down Syndrome Australia: http://www.downsyndrome.org.au

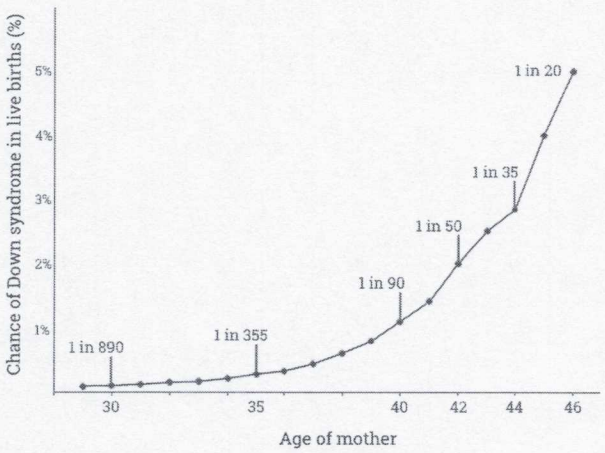


Figure 2: Chance of giving birth to a baby with Down syndrome by age of mother.

Edwards syndrome (trisomy 18) & Patau syndrome (trisomy 13)

Edwards syndrome occurs approximately 1 in every 1600 pregnancies and is caused by an extra copy of chromosome 18.

Patau syndrome occurs approximately 1 in every 3400 pregnancies and is caused by an extra copy of chromosome 13.

Pregnancies affected by these conditions show many physical problems. Infants born with Edwards or Patau syndrome have severe physical and intellectual disability and survival beyond one year of age is rare.

As with Down syndrome, the chance of having a pregnancy affected by Edwards or Patau syndrome increases with a woman's age but pregnancies of younger women can also be affected.

Sex chromosome conditions

One pair of our 46 chromosomes are called the sex chromosomes. Females usually have two X sex chromosomes (XX) and males have one X and one Y sex chromosome (XY).

Sex chromosome conditions occur when there is a missing X chromosome, an extra copy of the X chromosome or an extra copy of the Y chromosome.

Examples include:
Turner syndrome (also called monosomy X or 45,X); Klinefelter syndrome (47,XXY); triple X (47,XXX) and Jacob syndrome (47,XYY).

These conditions affect individuals in a variety of ways. Many have a normal quality of life and often remain undiagnosed in the general population.

Women of any age can have a pregnancy affected by a sex chromosome condition.

Types of prenatal tests for chromosome conditions

Prenatal tests fall into two groups: screening tests and diagnostic tests.

Screening tests
These tests give an estimate of the chance that a pregnancy is affected by a certain condition (they are not diagnostic tests).

If the screening test shows a high chance, a diagnostic test is offered. This will confirm if a condition is present or will reassure that the pregnancy is not affected.

Screening tests require a blood sample from the mother and/or an ultrasound scan. They are available to women of all ages.

Diagnostic tests
Diagnostic tests will give a definitive (yes or no) answer. They can confirm if a condition is present or reassure that the pregnancy is not affected.

Diagnostic tests are performed on samples of the placenta or amniotic fluid from around the developing baby. These samples are collected by two procedures: chorionic villus sampling (CVS) and amniocentesis.

As these procedures have a very small risk of miscarriage (above the natural miscarriage rate), diagnostic tests are usually offered to women who have a greater chance of having an affected pregnancy (e.g. advanced maternal age, family history, or have received a high chance screening result).

This booklet and the web tool focus on screening tests. More information about diagnostic testing can be found on page 28.

Combined first trimester screening (cFTS)

Combined first trimester screening adds different measures together to provide an estimate of the chance a pregnancy is affected by Down syndrome, Edwards syndrome or Patau syndrome.

cFTS is not diagnostic, but it can predict Down syndrome in 85-90% of cases. cFTS does not identify neural tube defects.

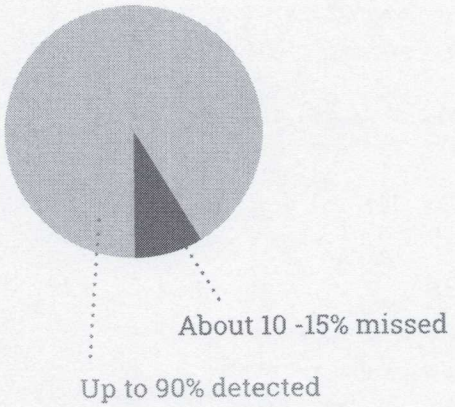


Figure 3: Detection rate of Down syndrome using combined first trimester screening.

The measures are:

1. Maternal blood, taken between 9-13 weeks gestation to measure chemicals PAPP-A and free beta-hCG.
2. A nuchal translucency ultrasound (NT scan) (11 to 13 weeks).
3. Maternal age, weight and gestation.

In some cases, an additional measurement, called the nasal bone, is included. The presence or absence of the nasal bone (seen on ultrasound) is added to the combined score.

Combined first trimester screening does not give a 'yes' or 'no' answer. Based on the result however, a decision can be made about diagnostic testing.

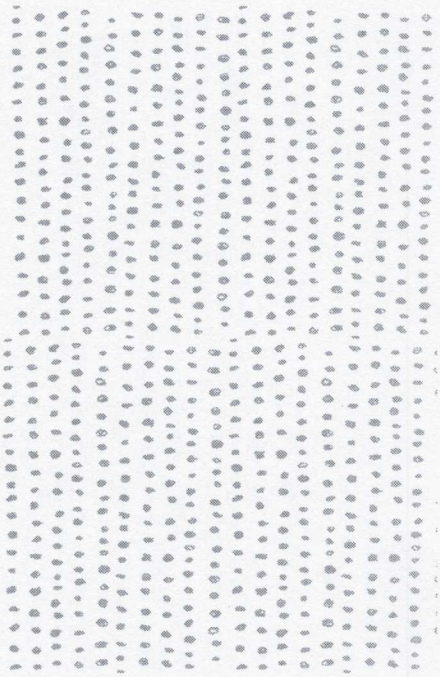
For information about how combined first trimester screening test results are reported, see page 21.

Nuchal Translucency ultrasound (NT scan)
The NT scan is performed between 11-13 weeks gestation. Sound waves are used to produce an image of the developing baby. This scan measures the thickness of the fluid filled space at the back of the baby's neck. An enlarged NT measurement may mean an increased chance for certain conditions, such as Down syndrome.

An NT scan alone is not able to accurately diagnose Down syndrome. A larger than normal NT measurement means that follow-up diagnostic testing is needed to confirm the presence of a condition or to reassure that the pregnancy is unaffected.

This early ultrasound can also:

- Confirm the due date.
- Identify twins.
- Identify if a miscarriage has occurred.
- Identify some physical features.



Second trimester maternal serum screening (2TMSS)

Second trimester maternal serum screening is a single blood test, between 14-20 weeks of gestation (best done between 15-17 weeks).

2TMSS provides an estimate of the chance a pregnancy is affected by Down syndrome, Edwards syndrome, or neural tube defects such as spina bifida.

2TMSS is not diagnostic, but it can predict between 75-80% of pregnancies affected with Down syndrome.

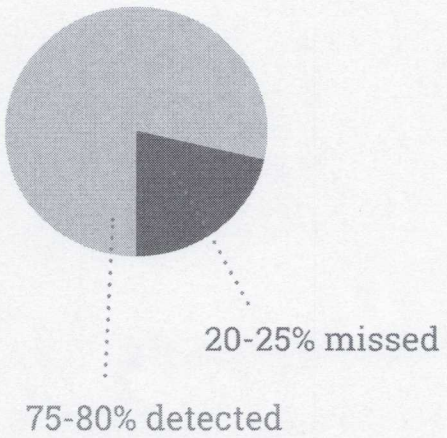


Figure 4: Detection rate of Down syndrome using second trimester maternal serum screening.

This test measures the level of four chemicals: alpha-feto protein, unconjugated estriol, free beta hCG and dimeric inhibin A. These are combined with maternal age, weight and gestation.

Women can have either combined first trimester screening or second trimester maternal serum screening depending on their gestation. It is not recommended that women have both screening tests. If you've had first trimester screening, you won't be offered second trimester screening.

Second trimester screening does not give a 'yes' or 'no' answer. Based on the result however, a decision can be made about diagnostic testing.

For information about how second trimester screening risk results are reported, see page 22.

Cell-free DNA prenatal testing (cfDNA)

(also known as non-invasive prenatal testing or NIPT)

Cell-free DNA prenatal tests give an estimate of the chance that a pregnancy is affected by Down syndrome, Edwards syndrome, Patau syndrome or sex chromosome conditions.

Some cfDNA tests can also identify the baby's sex, reveal secondary findings and can include microdeletion syndromes (see page 36).

Most cfDNA tests have a detection rate of over 99% for Down syndrome. This means that if a pregnancy is affected by Down syndrome, it will almost certainly be detected. The detection rate is not as high for the other conditions.

While cfDNA tests are more accurate than other screening tests, they are not 100% diagnostic and do not replace diagnostic testing.

Blood for cfDNA testing can be taken any time from 10 weeks gestation onwards.

It is also recommended that an NT ultrasound scan (page 13) is performed alongside the cfDNA test. This scan may identify physical conditions that the cfDNA test can't.

How is cfDNA different to the other prenatal screening tests?

Combined first trimester and second trimester maternal serum screening look at various chemicals in mum's bloodstream to predict fetal abnormalities (biochemistry). cfDNA however, looks at short pieces of DNA from the placenta (genetic), which are found in mum's blood (Figure 5).

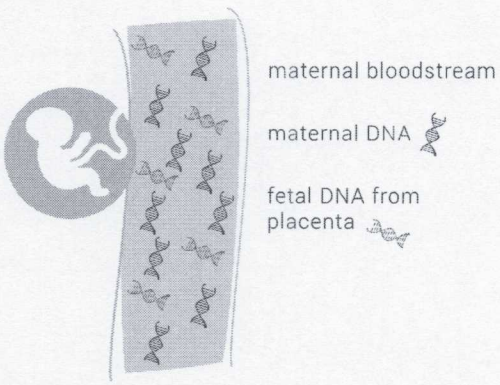
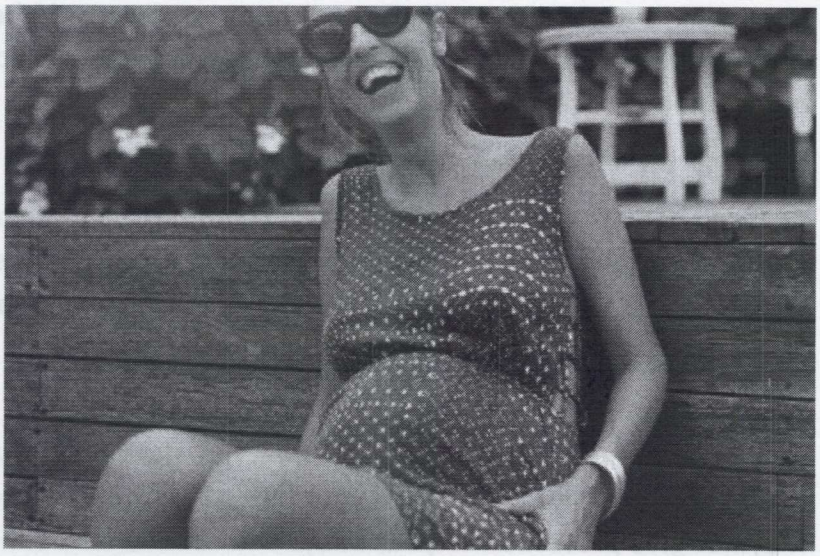


Figure 5: Cell-free DNA from the placenta found in the maternal bloodstream

Whooping cough vaccine for pregnant women



If you're pregnant, it's best to have a whooping cough (pertussis) vaccine. This vaccine also protects you against diphtheria and tetanus and boosts your immunity if you've already been vaccinated against these. It's available free of charge for pregnant women from 13 weeks of pregnancy until they give birth.

The ideal period to be vaccinated is from 16 weeks of pregnancy to 27 weeks. The later you're vaccinated, the more likely it is that only you (and not your baby) will be protected, especially if you're vaccinated within six days of giving birth.

You should have a vaccination during each pregnancy, even if you had a childhood whooping cough vaccination or one during a previous pregnancy.

Whooping cough (pertussis)

Whooping cough (pertussis) is a very contagious illness that causes bouts of severe coughing, which can last up to three months. It's dangerous for a baby under 1 year old to get whooping cough as they may get very sick and need to go to hospital. Some babies who get whooping cough will die.

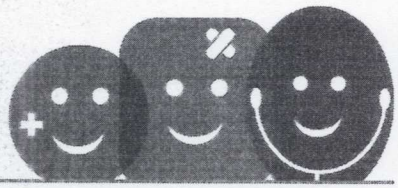
Whooping cough vaccinations

If you get whooping cough while you're pregnant or after your baby is born, there's a high chance you'll pass it on to your baby. Therefore, you should consider having the vaccine to reduce your risk of getting whooping cough. If you have the vaccine, it's also thought that you'll pass on some immunity from whooping cough to your baby.

The vaccine is available free of charge for all pregnant women from 13 weeks of pregnancy until they give birth.

It's also free of charge to parents or primary caregivers of babies admitted to a neonatal intensive care unit or specialist care baby unit for more than three days.

Your baby should still have their normal course of vaccinations starting at six weeks.



Other family members

All other people in your household and other close family members (such as grandparents) should have a whooping cough vaccination as they could be at risk of passing it on to your baby. The vaccine isn't subsidised for adults, but it's free for children as part of the normal childhood vaccination programme.

Safety of the vaccine during pregnancy

The United States Advisory Committee on Immunisation Practices (ACIP) has looked carefully at the information available on whooping cough vaccine in pregnant women and has recommended that pregnant women should be vaccinated during a whooping cough outbreak.

As with all vaccines, there's a small risk of side effects. For this reason, you'll need to stay at the practice for 20 minutes after receiving the vaccine.

If you can't or choose not to have the vaccine during pregnancy, you should consider having it within two weeks of the birth of your baby. This will still protect you from whooping cough but may not give protection to your baby. The vaccine isn't subsidised after you've given birth.

Getting the vaccination

Call your general practice team for an appointment to have the vaccination. Let them know you're coming in for the pertussis booster vaccine as they may need to order it, which can take one to two days.

For more information on whooping cough, visit www.immune.org.nz.

Written by the Canterbury Immunisation Provider Group. Adapted by HealthInfo clinical advisers. Last reviewed July 2017. Last updated July 2019.