illumiscreen

A guide for parents-to-be

Non-invasive prenatal screening

For more information, talk to your healthcare provider, or visit our website

illumiscreen.co.nz

Learning the sex of your baby

From as early as 10 weeks, you can reliably and safely learn the sex of your baby using Illumiscreen. It is important to know that all other chromosomes will also be screened, and their results provided in the report given to your healthcare provider.

The male sex typically has an XY pair of sex chromosomes, and the female sex typically has an XX pair of sex chromosomes. Your results will provide the sex chromosome information for single pregnancies.

If you are having twins, the report will tell you if there is a Y chromosome present. The presence of a Y chromosome suggests at least one twin is male, whereas the absence of a Y chromosome suggests neither twin is male.

Illumiscreen can also identify conditions related to missing or extra sex chromosomes, such as Turner syndrome (only one X chromosome in a female) or Klinefelter syndrome (an extra X chromosome in a male). Other possible conditions are Triple X and Jacobs syndrome.

You can choose whether you want your healthcare provider to share the sex of your baby or not. There is no additional charge for including this in your report.

illumiscreen

What is non-invasive prenatal screening?

Non-invasive prenatal screening (NIPS) is a simple blood test that looks at all 23 pairs of your baby's chromosomes from 10 weeks of pregnancy onwards. The screen can safely and reliably tell you if there is an increased chance your baby will have a chromosome related condition.

The non-invasive prenatal screening service we provide is called *Illumiscreen*. Illumiscreen is available for both single and twin pregnancies and can also identify the sex of your baby*.

Results are usually reported back to your healthcare provider within 5-7 working days of the sample arriving at the testing laboratory in Auckland. We recommend discussing this screen with your healthcare provider.

* Sex chromosome information is included for single pregnancies. For twin pregnancies, only the presence of the Y chromosome is reported. See more information under 'Learning the sex of your baby'.

Illumiscreen is available from as early as 10 weeks into your pregnancy

How does the screening work?

As your baby grows, DNA (genetic material) is released from the placenta into your bloodstream. This DNA is free-floating and called cell-free DNA. When we take a sample of your blood, we can find and learn about this cell-free DNA.

Illumiscreen uses advanced technology to analyse this cell-free DNA and count the number of chromosomes present. The screen is then able to determine if your baby has 46 chromosomes, or too many or too few copies of these chromosomes.

Generally, humans have 23 pairs of chromosomes (46 chromosomes in total). Growth and development can be affected when there are more than 46, less than 46, or when some chromosomes have extra or missing pieces. This is called a *chromosomal aneuploidy*, which can lead to a chromosome related condition.

Down syndrome is likely the most well-known example of a *chromosomal aneuploidy*, which is caused by an extra copy of chromosome 21. This is known as trisomy 21 - 'tri' indicating three copies.

The three most common chromosome conditions seen in pregnancy are Down syndrome, Edwards syndrome and Patau syndrome.

The difference between a screen and a diagnostic test

It is important to note that Illumiscreen is a highly reliable non-invasive prenatal screen, and not a diagnostic test. This means it does not provide you with a definite yes or no answer. It tells you if there is an *increased chance* your pregnancy has a chromosome related condition. To confirm an increased chance result from this screen, a diagnostic test is needed. This requires either an amniocentesis procedure or chorionic villus sampling (CVS), however, the invasive nature of these procedures may pose a risk to your pregnancy. Your healthcare provider will be able to talk to you about these.

What do my Illumiscreen results mean?

Your healthcare provider will receive and discuss your results, as well as further screening or testing options to consider, if appropriate.

Your results will tell your healthcare provider whether there is an increased chance your baby will have a condition because of too many or too few chromosomes, or due to chromosomes having extra or missing pieces.

It is recommended that clinical decisions are not made based on these screening results alone. If a definitive diagnosis is desired, chorionic villus sampling (CVS) or amniocentesis should be considered.

The report provided to your healthcare provider will include one of three possible results for chromosomes:

- **No Aneuploidy Detected** — This means the expected number of chromosomes were found. The result indicates low chance for chromosome related conditions.
- **Aneuploidy Detected** — This means an extra or missing copy of a chromosome has been identified. This result can indicate an increased chance for chromosome related conditions. Your healthcare provider may advise a diagnostic test for confirmation.
- **Aneuploidy Suspected** — This is a borderline result, which occurs infrequently, and suggests there might be an extra copy of a chromosome present. Your healthcare provider may advise a diagnostic test for confirmation.

Sex chromosomes will be reported as either *No Aneuploidy Detected* or *Aneuploidy Detected*. For single pregnancies it will also show the sex of the baby Male (XY) or Female (XX).

Learn what an aneuploidy is under 'How does the screening work'.

A safe and reliable screen about the genetic health of your baby

Does a 'negative' result mean that my baby will be perfectly healthy?

If the screening result is *No Aneuploidy Detected*, it indicates a *low-chance* result, it does not completely rule out all potential issues with your baby's chromosomes.

The Illumiscreen prenatal screen is a highly reliable non-invasive advanced screening tool. No test, however, can guarantee a baby will not have any other medical or physical conditions.

Detailed discussion with your healthcare provider who referred your test, before and after testing is recommended.

Is Illumiscreen right for me?

This screen is often offered to pregnant women who have been identified by their healthcare provider to have a higher chance of having a baby with a chromosome related condition, however any parent-to-be can request this screen with the support of their healthcare provider from 10 weeks onwards.

Understanding your baby's chance of having a chromosome related condition early may aid you, your family and your healthcare provider in preparing for the arrival of your baby and making delivery management decisions.

Why should I choose Illumiscreen over other tests?

Unlike other tests which calculate chances or risk scores, the Illumiscreen prenatal screen offers highly reliable information by analysing DNA that has been released from the placenta into the mother's bloodstream.

In addition, it does not carry the risk of complications that invasive procedures do.

Other benefits of choosing the Illumiscreen prenatal screen include:

- Uses a simple, single blood sample from your arm – just 1 tube of blood is all that is needed.
- Get your blood taken from your choice of 150 collection centres conveniently located all around Aotearoa New Zealand.
- Learn about the genetic health and the baby's sex from as early as 10 weeks into your pregnancy.
- Your blood sample and the genetic information of your baby stays in Aotearoa New Zealand.
- Screens the whole genome, not just chromosomes 13, 18 and 21, which other providers are limited to.
- Receive results in as little as 5-7 working days from your blood sample arriving at our Auckland laboratory.

Available from as early as 10 weeks

To learn more about the Illumiscreen prenatal screen, and for updated pricing visit illumiscreen.co.nz

POWERED BY illumina®

A service delivered by

APHG
ASIA PACIFIC HEALTHCARE GROUP

08112022