ILLUMI SCREEN

Next generation prenatal testing
delivered by Healthscope NZ

A guide for parents-to-be on noninvasive prenatal testing

Accurate answers about your baby's health— simply, safely, sooner

What is the Illumiscreen prenatal test?

The Illumiscreen prenatal test is a simple blood test that screens for the most common chromosomal abnormalities that can affect your baby's future health.

A sample can be drawn in your doctor's office as early as the tenth week of pregnancy. It may help you avoid more invasive procedures, such as amniocentesis or chorionic villus sampling (CVS), which can pose risks to you and your baby. The Illumiscreen test is available for both singleton and twin pregnancies.

*Test results are usually reported back to your health care provider within approximately 1 week.

How does the test work?

A sample of your blood is drawn, and the DNA from you and your baby is tested. The Illumiscreen test takes a deeper approach to the science, using an advanced technology called "massively parallel sequencing" to analyse millions of DNA fragments per sample and count the number of chromosomes present. It then uses a special SAFeR™ calculation method to determine if there are too many or too few copies of these chromosomes in your baby.

*The Illumiscreen prenatal test for chromosomes 21, 18, and 13, is available for singleton and twin pregnancies. Sex chromosome aneuploidy testing is available for singleton pregnancies only. An optional test for twin pregnancies tests for the presence of the Y chromosome.

What kind of conditions can the test detect?

Chromosomes normally come in pairs. Healthy people usually have 23 pairs of chromosomes, the last pair of which determines sex. Men normally have an XY pair of sex chromosomes. Women normally have an XX pair of sex chromosomes.

The Illumiscreen prenatal test looks for too few or too many copies of chromosomes. Missing or extra copies of chromosomes are called “aneuploidies” and are often associated with mental or physical disabilities with different levels of severity. The most commonly seen aneuploidies include **trisomy 21** (Down syndrome), **trisomy 18** (Edwards syndrome), and **trisomy 13** (Patau syndrome), all of which can be screened for with the Illumiscreen test.

If your health care provider chooses, the Illumiscreen prenatal test can also be used to identify sex chromosome abnormalities, 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.

Is the Illumiscreen test right for me?

The Illumiscreen test offers parents-to-be a new choice to obtain important information about the health of their developing baby, simply, accurately and in the first trimester, with little or no risk to their pregnancy.

This screening test is usually offered to pregnant women identified by their doctor to be at risk for fetal aneuploidy. It may be an option for you if you have a confirmed singleton or twin pregnancy of at least 10 weeks’ gestational age.

The confidence you seek—with fewer risks

What are my current testing options?

Today, there are a number of genetic testing options available for expectant women and their health care providers.

Traditional screening tests are used to predict the chance of a pregnancy having certain chromosomal conditions. Some screening tests require more than one office visit to perform multiple blood draws and ultrasound examinations at very specific times during pregnancy. Results from screening tests usually provide a "risk score" (such as 1 in 500 or 1 in 50) that describes the chance of a baby developing a certain chromosome problem. These tests do not provide definitive answers and can have a relatively high rate of "false negative" or "false positive" results. But not all screening tests are the same in terms of accuracy and convenience.

Diagnostic tests, such as amniocentesis or CVS, can accurately determine whether a pregnancy has **trisomy 21** (Down syndrome) or other chromosomal conditions. However, the invasive nature of these procedures poses a risk of complications, including miscarriage.

Why should I choose the Illumiscreen test over other tests?

The Illumiscreen test sheds much-needed light on the chromosomal health of your unborn baby—providing the reassurance of reliable answers no other screening test can. Compared to similar options, the Illumiscreen prenatal test offers accurate information, rather than calculating chances or risk scores. In addition, it does not carry the risk of complications that invasive procedures do.

It also:

Uses a simple, single blood draw from your arm—just 1 tube of blood is all that's needed.

Can be taken conveniently in any of our collection centres—as early as 10 weeks.

Provides reliable answers about the most common chromosomal abnormalities.

Can screen for sex chromosome conditions—(for singleton pregnancies) if ordered by your health care provider. If you're carrying twins, however, the Illumiscreen test cannot test for sex chromosome conditions. There is an option that can determine if the Y chromosome is present.

Has the lowest test failure in its class (0.1%)[1]—which means there is no need for additional blood draws and the inconvenience of another office visit.

Delivers results fast—results are usually reported 5-7 working days after sample receipt.[2]

Performance you can trust—we've got you covered

What do my Illumiscreen test results mean?

Your results will tell your doctor whether **trisomies 21**, **18**, **13** or sex chromosome abnormalities or certain microdeletion conditions (available by special request which will incur an additional cost) are highly likely to be present in your pregnancy. In the case of a positive result, your health care provider will discuss your results as well as further testing options to consider. It is recommended that no irreversible clinical decisions should be made based on these screening results alone. If a definitive diagnosis is desired, chorionic villus sampling or amniocentesis should be considered.

Your test report will include one of three possible results for chromosomes 21, 18, and 13:

No Aneuploidy Detected—This means the normal number of chromosomes was found. The result indicates low risk for chromosomal abnormalities.

Aneuploidy Detected—This means an extra copy of the tested chromosome has been identified. This result can indicate high risk for chromosome abnormalities. Your 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 provider may advise a diagnostic test for confirmation.

If the sex chromosome option is ordered, results will be reported as either *No Aneuploidy Detected* or *Aneuploidy Detected*.

How do I know I can trust the Illumiscreen test?

The performance of the Illumiscreen prenatal test was evaluated in a major validation study that involved more than 60 leading US medical research and teaching institutions. The study findings were reviewed and published in the leading journal read by obstetricians and gynaecologists.[3] A second study, published subsequently, presented the test's performance under regular clinical conditions and found similar results.[4]

Do normal Illumiscreen test results mean that my baby will be perfectly healthy?

The Illumiscreen prenatal test is a highly accurate noninvasive advanced screening test. No test, however, can guarantee a baby will not have any medical issues. The Illumiscreen test addresses aneuploidies of chromosomes 21, 18, 13, and sex chromosomes only,† when requested. It does not test for or report all genetic and non-genetic problems that may be present in a baby. If the test result is *No Aneuploidy Detected*, indicating a negative result, it does not completely rule out all potential problems with chromosomes 21, 18, and 13, or all sex chromosome aneuploidies in your baby.

Detailed discussion with your referrer before and after testing is recommended. Results of *Aneuploidy Detected* or *Aneuploidy Suspected* are considered positive, and women who receive such results should be offered invasive prenatal procedures for confirmation. A negative test does not ensure an unaffected pregnancy.

CVS and amniocentesis provide definitive diagnostic information, but their invasive nature is a potential concern.

†Sex chromosome aneuploidy testing is available for singleton pregnancies only.

The reassurance of reliable answers—simply, safely, sooner

ILLUMI SCREEN

A simple, in-office blood test

Safe from procedural risks of invasive procedures (amniocentesis/CVS)

Lowest test failure rate among noninvasive prenatal tests

Fast results—usually sent to the laboratory in 5–7 business days after sample receipt

To learn more about the Illumiscreen prenatal test, visit illumiscreen.co.nz

References

1. Bhatt S, Parsa S, Snyder H, et al. Clinical Laboratory Experience with Noninvasive Prenatal Testing: Update on Clinically Relevant Metrics. ISPD 2014 poster.
2. Data on file.
3. Bianchi DW, Platt LD, Goldberg JD, et al. Genome-wide fetal aneuploidy detection by maternal plasma DNA sequencing. *Obstet Gynecol*. 2012;119:890-901.
4. Futch T, Spinosa J, Bhatt S, et al. Initial clinical laboratory experience in noninvasive prenatal testing for fetal aneuploidy from maternal plasma DNA samples. *Prenat Diagn*. 2013;33:569-574.