

# Genea's Non-Invasive Prenatal Screening

GeneSyte Patient Information Booklet



 GeneSyte™





The early days of your pregnancy can be exciting and overwhelming. Imagining the new life growing inside you is exciting but wading through pages of information and advice on what tests, screens and ultrasounds you need to have can be daunting and confusing. We understand that you simply want the fastest and most accurate answers without risking your pregnancy.

## Introducing Genea's non-invasive prenatal screening - GeneSyte

Genea is a pioneer in prenatal screening with 30 years of experience in the field and a strong reputation for quality testing. Our non-invasive prenatal screening GeneSyte, is the latest addition to our suite of services for expectant mums and will provide you with prompt, accurate reassurance about the health of your unborn child.

GeneSyte is Australia's first non-invasive prenatal screening service to test samples onshore. Until now, all other non-invasive prenatal screening offered in Australia have collected samples here before sending them overseas for testing.

### Peace of mind

**Testing as  
early as  
Week 10**



**Safer with  
a simple  
blood test**



**More  
comprehensive  
results**



## How does GeneSyte work?

GeneSyte uses state of the art technology to identify chromosome conditions in your unborn child by screening your blood sample in the first trimester of your pregnancy. It's a blood test that's safe for you and your baby - a highly accurate test that removes the risk of miscarriage associated with invasive procedures such as chorionic villus sampling (CVS) or amniocentesis.

When you are pregnant, some of your baby's DNA crosses the placenta into your bloodstream. We can find it in a sample of your blood and check for the conditions which are caused by extra or missing chromosomes.



Chromosomes are long, spiral strands of DNA that carry genetic information. Humans have 46 chromosomes - 23 pairs. The first 22 pairs are numbered from 1 through to 22; the last pair determine your baby's sex - girls have two X chromosomes and boys have one X and one Y chromosome. Health and developmental problems occur when there are too many (trisomy) or too few (monosomy) chromosomes.

Genea's GeneSyte screens for the widest range of chromosome conditions offering you peace of mind about the health of your baby. With greater than 99% detection of Trisomy 21 commonly known as Down syndrome it is one of the most accurate non-invasive prenatal screening tests available. Your consulting doctor will be able to explain this further.

# GeneSyte can identify whether your baby has a high risk of:

## **Trisomy 21** Down syndrome

Carrying an extra copy of chromosome 21 causes Down syndrome, the most common chromosome disorder affecting approximately one in every 660 pregnancies in Australia. People born with Down syndrome have some characteristic physical features, some health and development challenges and some level of intellectual disability.

## **Trisomy 18** Edwards syndrome

Carrying an extra copy of chromosome 18 causes Edwards syndrome, the second most common chromosome disorder affecting approximately one in every 1,100 pregnancies in Australia. Edwards syndrome is associated with a high rate of miscarriage and babies born with the condition have a significantly shortened life expectancy, suffer heart abnormalities, kidney malformations and developmental delays.

## **Trisomy 13** Patau syndrome

Carrying an extra copy of chromosome 13 causes Patau syndrome. Approximately one in 3,000 pregnancies in Australia are affected by Patau syndrome, a condition that is also associated with a high rate of miscarriage. Babies born with Patau syndrome rarely survive beyond the first year of life and suffer heart and brain problems, eye defects and difficulties with feeding and breathing.

## **Monosomy X** Turner syndrome

The absence of an entire sex chromosome in girls causes Turner syndrome. Approximately one in every 2,500 female babies are affected by Turner syndrome, a condition which can cause sterility, characteristic physical abnormalities and vision and hearing difficulties.

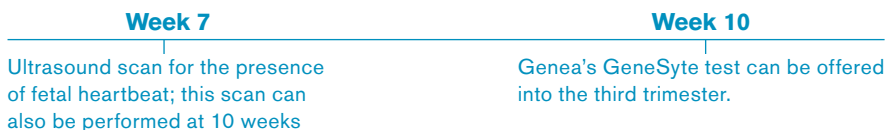
Genea's GeneSyte test also includes XX and XY testing of your baby.

## When can I be tested?

Genea's GeneSyte follows best practice guidelines for prenatal screening and, in conjunction with accredited ultrasound practices, recommends the following timeline of testing to give you the most definitive results on the health of your baby.

Advised by your doctor and based on clinical necessity, the test can be offered from as early as 10 weeks of pregnancy and into the third trimester.

The gestational dating of your pregnancy can be determined by ultrasound scan at 7 to 10 weeks or by an accurate last menstrual period date. This will be determined by your treating doctor.



## How do I get tested?

- 1.** Discuss GeneSyte with your doctor. A Genea Pathology Request Form must be completed by your doctor.
- 2.** Attend a collection site for your blood test. Blood collection is available through your health care provider or an alternative site.

You can also attend a Genea collection centre at your convenience. If you are located in NSW, ACT or Victoria please call 1300 652 687 or visit [bookings.mygenea.com.au](https://bookings.mygenea.com.au) to book an appointment near you. WA patients please call (08) 93894200.

- 4.** Make payment. A payment slip is included with the referral form for your convenience. Note: Medicare does not currently offer a rebate on this testing.
- 5.** Your results will be delivered to your consulting doctor in 5 to 7 working days\*. Your doctor or a genetic counsellor will contact you to discuss and explain your results.

\* 5 to 7 working days from the date the sample is received at Genea.

## What will my results say?

Genea's GeneSyte will provide you with clear, concise results on the health of your unborn child. Your results report will identify whether your baby is carrying any of the chromosomal conditions listed.

If the test result indicates that your baby has one of the chromosomal conditions, Genea recommends you consult with your Doctor. They may advise you to consider further diagnostic testing (CVS or amniocentesis) to confirm or disprove the result as recommended by the Royal Australian New Zealand College of Obstetricians and Gynaecologists (RANZCOG) and the Fetal Medicine Foundation of London.

Genea offers in-house Genetic Counselling for patients who need to discuss their results and the decisions they need to make. Our counsellors are highly trained and experienced in speaking with people about genetic conditions and testing.

A negative result means that none of these chromosomal conditions have been detected by this test. The guidelines, as mentioned above, recommend that no further invasive testing is required, however, it is important to talk to your Doctor about follow up scans to check for any other fetal anomalies and structural defects. These scans can be important for further assessment of the health of your pregnancy.

## Why Genea for your testing?

With 30 years of experience in fertility and helping couples achieve their dream of creating families, Genea is perfectly qualified to give you fast and accurate answers and ultimately peace of mind about the health of your pregnancy.

Having your test done through Genea ensures you receive the most comprehensive clinical coverage of the most common chromosomal conditions.

Unlike other general pathology providers, Genea only focuses on fertility, meaning you benefit from our solid expertise and interpretation of results.

GeneSyte™  
Experience the difference.

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