GeneSyte™

Genea

# PRE AND POST-TEST COUNSELLING FACT SHEET

## GENEA'S GENESYTE: NON-INVASIVE PRENATAL SCREENING (NIPS)

If your patient is considering GeneSyte, pre and post-test counselling is an important part of the doctor's initial consultation. Appropriate pre-test counselling will help to reduce post-test anxiety for patients.

The advantages and limitations of GeneSyte should be discussed prior to undergoing the test, along with any implications of the results.

At Genea, we want to be able to provide you with the support you and your patients may need. For this reason we offer complimentary post-test counselling with a Genetic Counsellor should the need arise.

Please contact Genea on 1300 652 657 for further information or to contact a Genetic Counsellor.

## WHAT IS GENESYTE?

GeneSyte is an advanced screening blood test that uses cell free DNA found in the mother's blood to identify the most common chromosomal conditions that can affect pregnancy.

## WHAT DOES GENESYTE SCREEN FOR?

GeneSyte screens for the most commonly occurring chromosomal conditions, offering patients peace of mind about the health of their baby. Fetal DNA in the mother's blood is analysed for the following conditions:

**Autosomal chromosomal aneuploidy:**

1. Trisomy 21-Downs syndrome
2. Trisomy 18-Edwards syndrome
3. Trisomy 13-Patau syndrome

**Sex chromosomal aneuploidy:** GeneSyte can also detect most sex chromosomal aneuplodies for the following conditions:

1. Monosomy X-Turner's syndrome
2. XXX (Triple X)
3. XXY (Klinefelter syndrome)
4. XYY (Jacob's syndrome)
5. Fetal sex (XX or XY)

There may be a possibility of incidental findings associated with this testing. These results may be discussed with your consulting doctor.

## WHEN CAN A PATIENT TAKE THE GENESYTE TEST?

The GeneSyte screening test is intended for patients at 10 weeks or greater gestation. If the patient's maternal weight is >110 kg, it is recommended to delay testing until 12 weeks.

## HOW ACCURATE ARE THE RESULTS?

GeneSyte is very accurate for the detection of Trisomy 13,18 and 21. Detection of Trisomy 21 is greater than 99% and Trisomy 13 and 18 are greater than 98%.

## WHAT ARE THE LIMITATIONS OF THE TEST?

It's very important that the patient understands that GeneSyte is an advanced screening test and not a diagnostic test. Undergoing the test and getting a negative result does not guarantee an unaffected baby.

GeneSyte screens for the most common autosomal and sex chromosomal aneuploids.

The GeneSyte test does not give information on the physical or structural abnormalities of the baby and does not look for any problems in the heart, brain or other organs. Ultrasound scan is commonly used to detect these anomalies that are not necessarily chromosomal in origin.

## WHAT WILL THE GENESYTE REPORT SAY?

The GeneSyte report will include one of the two possible results below:

**1. "No Aneuploidy Detected" (NAD):**

This "NAD" result means that none of these chromosomal conditions have been detected for T13, T18, T21. The guidelines from the Royal Australian and New Zealand College of Obstetricians and Gynaecologists (RANZCOG) recommend that no further invasive testing is required. However, it is important that your patient does not miss the fetal anomaly scan to detect any structural anomalies.

**2. "Aneuploidy Detected"**

An "Aneuploidy" detected result indicates that GeneSyte has detected one of the chromosomal conditions in the screening panel. Therefore the unborn baby is highly likely to be affected with a chromosomal abnormality. The possibility of a false result is less than 0.2%.

**RANZCOG Guidelines:** Confirmatory diagnostic testing is strongly recommended after an abnormal cfDNA result.

At Genea, a Genetic Counsellor is available to discuss the results for your patient to support and advise them as they make a decision regarding the next steps.

## WHAT IF "NO RESULT" IS ISSUED?

GeneSyte is able to issue a result in 99.9% of cases. In the rare instance that a result is not possible, you will be contacted to discuss the options moving forward for your patient.

## HOW MUCH WILL GENESYTE COST?

GeneSyte is available at a cost of $445 and includes complimentary post-test Genetic Counselling. Medicare does not currently reimburse the GeneSyte cost.

## HOW LONG DOES IT TAKE TO GET THE GENESYTE RESULTS?

Once Genea has received the blood sample in the lab, the results will take approximately **5–7 working days** and a report will be issued to the referring doctor.

## WILL GENESYTE GIVE MY PATIENTS A GUARANTEE THAT THE BABY IS COMPLETELY NORMAL?

GeneSyte does not test for other chromosomal abnormalities or genetic problems other than what is offered in the GeneSyte screening panel.

The GeneSyte test does not give information about the physical or structural abnormalities of the baby as it doesn't test for any cardiac, brain or organ anomalies.

## WHAT SUPPORT IS OFFERED TO ME AND MY PATIENTS?

As part of the GeneSyte test, complimentary post-test Genetic Counselling is available for your patients should they require this. We also have a comprehensive pathology and laboratory team that can support you in interpreting the test results or for any other questions you may have along the way.

## DOES GENESYTE TEST FOR TWIN OR MULTIPLE PREGNANCIES?

GeneSyte is able to detect the presence of a Trisomy 13, 18 or 21 in twin pregnancies, but will not distinguish which of the twins carries the chromosomal condition. An invasive procedure and diagnostic testing will be required to determine which twin is affected.

A test for the presence of the Y chromosome can also be ordered for twins.

## POST-TEST COUNSELLING FOR GENESYTE SHOULD INCLUDE THE FOLLOWING:

Careful consideration should be given to the way the test results are conveyed. The doctor should take this opportunity to explain again the implications of the results.

Appropriate follow up is needed when an abnormality is detected and referral to a Genetic Counsellor is highly recommended.

**Genea Pathology** Level 3, 321 Kent Street, Sydney NSW 2000 Australia
**p** 1300 652 687 **f** (02) 9229 6400 **w** genea.com.au **e** genea.pathology@genea.com.au

MKT02487 | May 2021