



A GUIDE FOR PARENTS-TO-BE ON

Generation[®]

a new era in prenatal testing

NON-INVASIVE PRENATAL TESTING

1800 822 999



Western[®]
Diagnostic
PATHOLOGY

A revolutionary advance



What is the **Generation**[®] non-invasive prenatal test?

Non-invasive prenatal testing (NIPT) is a revolutionary advance in prenatal screening which can detect genetic material (DNA) from the placenta in a blood test from the mother. In the past, the ability to test DNA from the fetus required much more invasive methods such as amniocentesis or placental biopsies, which are not without risks to mothers and their babies. NIPT is a simple and highly accurate test which may help avoid more invasive techniques of prenatal testing.

The **Generation**[®] non-invasive prenatal test (NIPT) screens for the most common chromosomal abnormalities that can affect your baby's future health using a simple blood test. The **Generation**[®] NIPT is available from as early as the 10th week of pregnancy, for both singleton and twin pregnancies.

Your doctor may recommend the **Generation**[®] Plus test which also tests for microdeletions when there are reasons to do so.

How does the **Generation**[®] test work?

During pregnancy, some of the baby's DNA from the placenta crosses into your bloodstream. A sample of your blood is drawn from you, and the **Generation**[®] NIPT tests this DNA to identify certain chromosome conditions in your pregnancy. The **Generation**[®] 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 accurately count the number of chromosomes present and determine if there are too many or too few copies of the tested chromosomes in your baby.

in prenatal screening

Is the **Generation**[®] test right for me?

The **Generation**[®] 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 (at 10 weeks), with little or no risk to their pregnancy.

This screening test may be an option for you to consider if:

- You are 35 years or older at the time of delivery (32 years or older for a twin pregnancy)
- You have an abnormal or “positive” serum screen
- Your ultrasound shows concerns or abnormalities with fetal growth and/or development
- You have a personal or family history suggestive of a chromosome disorder (e.g. Down syndrome)

Clinical best practice guidelines from Australian and international medical societies recommend that all pregnant women, regardless of risk status, be offered the opportunity for discussion and choice regarding NIPT and other available prenatal screening and diagnostic tests^{1,2,3,4,6,7,8}.

What kind of conditions can the **Generation**[®] test detect?

Chromosomes normally come in pairs. Healthy people have 23 pairs of chromosomes, with one pair which determines sex. Men normally have an XY pair of sex chromosomes, and women normally have an XX pair of sex chromosomes. Any more or less can lead to mental or physical disabilities, with different levels of severity.

The **Generation**[®] NIPT looks for too few (missing) or too many (extra) copies of chromosomes, which are often associated with these disabilities. The most commonly seen and tested for include an extra copy of chromosome 21 (Down syndrome), or an extra copy of chromosome 18 (Edwards syndrome), or chromosome 13 (Patau syndrome) and sex chromosome aneuploidies, all of which can be accurately detected with the **Generation**[®] test.

Why the **Generation**[®] test?

Compared to similar options, the **Generation**[®] prenatal test offers accurate, near diagnostic information, rather than calculating chances or risk scores. It does not carry the risk of complications that an invasive procedure can.

✓ **It's simple.**

A single tube of blood drawn from your arm;

✓ **It's convenient.**

You can have blood collected in one of our collection centres from as early as 10 weeks;

✓ **It's accurate**^{1,2,5}.

	Observed Sensitivity	Observed Specificity
Trisomy 21	99.1%	99.9%
Trisomy 18	98.3%	99.9%
Trisomy 13	98.2%	99.9%
Monosomy X	95.0%	99.0%
XX	97.6%	99.2%
XY	99.1%	98.9%

✓ **It's tested in Australia.**

Your **Generation**[®] test is performed in Australia. (**Generation**[®] Plus tests are performed in California).

✓ **It's reliable.**

It has the lowest test failure of any NIPT (0.1%)¹;

Our Experience*

<0.1%

Test Failure Rate

✓ **It's fast.**

Your healthcare provider will receive your **Generation**[®] test results within 5-7 days from sample collection**. The **Generation**[®] Plus test including microdeletions is reported 9-14 days from collection due to sample transport times to California.

* Based on internal testing data.

** Regional collections may take slightly longer.

The **Generation**[®] test was chosen for development by Genomic Diagnostics based on a careful evaluation of its quality and proven scientific performance.

The performance of the **Generation**[®] prenatal test has been evaluated and published in numerous major studies, including clinical experience in over 34,000 patients from over 60 leading US medical research and teaching institutions¹. Those findings have subsequently been replicated in other studies^{2,3,4}, including the New England Journal of Medicine, one of the most prestigious international medical journals. These studies have found that the test performed substantially better than conventional tests under regular clinical conditions, with ~1 in 4,000 false negative results, ~1 in 500 false positive results, and the lowest test failure rate of any non-invasive prenatal test.

Your tests are performed in Australia in an accredited Australian laboratory, with this test undergoing full regulatory evaluation in the coming months.

How will my test be reported?

Your test report will include one of two possible results for chromosomes 21, 18, 13, X and Y.

1 **No Aneuploidy Detected** — means the expected number of chromosomes was found

2 **Aneuploidy Detected** — means too many or too few copies of one of the chromosomes have been identified. A diagnostic test for confirmation is recommended and should be discussed by your doctor

Do normal **Generation**[®] test results mean that my baby will be perfectly healthy?

The **Generation**[®] and **Generation**[®] Plus prenatal tests are highly accurate advanced screening tests that are non-invasive. No test, however, can guarantee a baby will not have any medical issues.

They do not test for, or report all, genetic and non-genetic problems that may be present in a baby.

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

How do I organise to have the test?

1. Make an appointment to see your medical practitioner and discuss the **Generation**[®] NIPT
2. Complete the request form with your doctor (go to www.generationNIPT.com.au)
3. Contact our Customer Care Team on 1800 822 999 to prepay and identify the most conveniently located **Generation**[®] collection centre
4. Bring the request form to your appointment. Your blood sample will be taken and sent to the lab for testing
5. Your **Generation**[®] NIPT is performed
6. Your results are delivered to your medical practitioner



Does Medicare or private health insurance cover the cost of the **Generation**[®] prenatal test?

The **Generation**[®] prenatal test does not qualify for a Medicare rebate. Payment for the **Generation** test for chromosomes 13, 18, 21 and sex chromosomes is payable prior to sample collection by calling our Customer Care Team on 1800 822 999.

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. Bianchi DW, Platt LD, Goldberg JD, et al. Genome-wide fetal aneuploidy detection by maternal plasma DNA sequencing. *Obstet Gynecol.* 2012;119:890-901.
3. Futch T, Spinosa J, Bhatt S, de Feo E, Rava RP, Sehnert AJ. Initial clinical laboratory experience in non-invasive prenatal testing for fetal aneuploidy from maternal plasma DNA samples. *Prenat Diagn.* 2013;33:569-574.
4. Bianchi DW, Parker RL, Wentworth J et al. DNA Sequencing versus Standard Prenatal Aneuploidy Screening. *N Engl J Med* 2014; 370:799-808.
5. Verinata Health, Inc. (2012) Analytical Validation of the veriFi Prenatal Test: Enhanced Test Performance For Detecting Trisomies 21, 18 and 13 and the Option for Classification of Sex Chromosome Status. Redwood City, CA.
- 6) RANZCOG Statement on Prenatal screening and diagnosis of chromosomal and genetic abnormalities in the fetus in pregnancy C-Obs 59. Endorsed by RANZCOG: March 2015
- 7) ACOG Committee on Practice Bulletins. (2007) ACOG Practice Bulletin No. 77: screening for fetal chromosomal abnormalities. *Obstet Gynecol.* 109(1):217-227.
- 8) Society for Maternal-Fetal Medicine (SMFM) Publications Committee. #36: Prenatal aneuploidy screening using cell-free DNA. *Am J Obstet Gynecol.* 2015; S0002-9378(15)00324-5.

As this brochure contains only general educational information, professional advice from your medical practitioner should be sought before applying the information in this brochure to particular circumstances. You should not rely on any information contained in this brochure without first obtaining professional advice. Prices are correct at time of printing and are subject to change without notice.





Generation[®]

a new era in prenatal testing

Generation[®] NIPT

- A simple, one tube blood test
- Safe from procedural risks of invasive procedures (amniocentesis/CVS)
- Lowest test failure rate among non-invasive prenatal tests
- Fast results—usually available to your doctor within 5-7 days from collection (**Generaton[®] Plus** results take 9-14 days from collection).

To learn more about the **Generation[®]** prenatal test please call 1800 822 999 or visit www.generationNIPT.com.au

