

# Generation<sup>®</sup> Non-invasive Prenatal Testing

A guide for parents-to-be



**QML Pathology**

*Specialists in Private Pathology since the 1920s*

# A revolutionary advance



## What is the **Generation**<sup>®</sup> 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**<sup>®</sup> 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**<sup>®</sup> NIPT is available from as early as the 10th week of pregnancy, for both singleton and twin pregnancies.

Your doctor may recommend the **Generation**<sup>®</sup> **PLUS** test which also tests for microdeletions when there are reasons to do so.

## How does the **Generation**<sup>®</sup> 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**<sup>®</sup> NIPT tests this DNA to identify certain chromosome conditions in your pregnancy. The **Generation**<sup>®</sup> 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.

## Is the **Generation**<sup>®</sup> test right for me?

The **Generation**<sup>®</sup> 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.

# in prenatal screening

**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.<sup>1,2,3,4,6,7,8</sup>

## What kind of conditions can the **Generation**<sup>®</sup> 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**<sup>®</sup> 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**<sup>®</sup> test.

**Generation**<sup>®</sup> **PLUS** is a separate test that can incorporate additional microdeletions.

# Why the **Generation**<sup>®</sup> test?

Compared to similar options, the **Generation**<sup>®</sup> 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**<sup>1,2,5</sup>

	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 reliable**

It has the lowest test failure of any NIPT (0.1%)<sup>1</sup>;

**Our Experience\***

**<0.1%**

**Test Failure Rate**

## ✓ **It's fast**

Your **Generation**<sup>®</sup> NIPT test is performed in Australia in an accredited laboratory, and your healthcare provider will receive results within 5-7 days from sample collection\*\*.

The **Generation**<sup>®</sup> **PLUS** including the microdeletions test, are sent to California and your healthcare provider will receive results from 9-14 days from collection due to sample transport times.

\* Based on internal testing data for Standard Generation NIPT tested and reported on shore in Australia.

\*\* 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 institutions<sup>1</sup>. Those findings have subsequently been replicated in other studies<sup>2, 3, 4</sup>, 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.

## How will my test be reported?

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

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

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**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.

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## 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, nor report, all genetic and non-genetic problems that may be present in a baby.

<sup>†</sup> Sex chromosome aneuploidy testing is available for singleton pregnancies only.

# How do I organise to have the test?

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1. Make an appointment to see your medical practitioner and discuss the **Generation®** test
  2. Complete the request form with your doctor (available at [www.generationNIPT.com.au](http://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 for **Generation®** NIPT can be taken Monday to Friday and **Generation® PLUS** can only be collected Mondays and Tuesdays
  5. Your blood sample is sent to the laboratory and your **Generation®** test is performed
  6. Your results are delivered to your medical practitioner



# Does Medicare or private health insurance cover the cost of the **Generation**<sup>®</sup> prenatal test?

Neither **Generation**<sup>®</sup> NIPT or **Generation**<sup>®</sup> PLUS are covered by Medicare and out-of-pocket fees apply.

Full payment by credit card is required prior to blood collection.

Call our Customer Care Team on 1800 822 999 (Mon-Fri, 9am-5pm AEST) to make payment and locate your nearest QML Pathology **Generation**<sup>®</sup> collection centre.

## 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.







## Generation® NIPT

- ✓ A simple, one tube blood test
- ✓ Safe from procedural risks of invasive procedures (amniocentesis/CVS)
- ✓ It has the lowest test failure of any NIPT (0.1%)<sup>1</sup>
- ✓ It is fast- **Generation®** NIPT test is **performed in Australia** and your healthcare provider will receive results within 5-7 days from sample collection\*\*.

The **Generation® PLUS** including the microdeletions test, are sent to California and your healthcare provider will receive results from 9-14 days from collection due to sample transport times.

*To learn more about the **Generation®** prenatal test please call 1800 822 999 or visit [generationNIPT.com.au](http://generationNIPT.com.au)*

As this brochure contains only general 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.



Genomic Diagnostics

