Abbott Pathology

The comfort of knowing

A GUIDE FOR PARENTS-TO-BE ON

Generation®

a new era in prenatal testing

NON-INVASIVE PRENATAL TESTING

1800 822 999

A revolutionary advance in prenatal screening

simple, safe, true.

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.

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.

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)

Accurate answers about your baby's health

simple, safe, true.

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.

If you and your healthcare provider request the ***Generation*® *Plus*** test, the following more rarely occurring genetic syndromes are also tested for:

- **Trisomy 9**, which is caused by an extra copy of chromosome 9. Almost all pregnancies with trisomy 9 end in first trimester miscarriage. Pregnancies with partial trisomy 9 may survive until term, but typically have significant birth defects and intellectual disabilities.
- **Trisomy 16**, which is caused by an extra copy of chromosome 16. Trisomy 16 is one of the most common causes of miscarriage. Pregnancies with partial trisomy 16 may survive until term, but are at increased risk for pregnancy complications and often have significant birth defects and intellectual disabilities.
- **Common microdeletions**, which are caused by the loss of a small piece of a chromosome.

  Some of the common microdeletions which can be detected by the ***Generation*® *Plus*** test include:

  - ***DiGeorge syndrome*** (22q11.2 deletion syndrome), which is commonly associated with heart defects, cleft palate, immune system disorders and intellectual disabilities.
  - ***Angelman syndrome***, which is commonly associated with significant developmental delay and learning disabilities, seizures and hyperactivity.
  - ***Prader-Willi syndrome***, which is commonly associated with mild to moderate intellectual disabilities, poor muscle tone and feeding difficulties in infancy that progresses to behaviour issues and compulsive over-eating in childhood.
  - ***Wolf-Hirschhorn syndrome***, which is associated with intellectual disability, characteristic facial features, seizures and delayed growth and development.
  - ***Cri-du-chat syndrome***, which is associated with intellectual disability, developmental delays, characteristic facial features and a high-pitched, cat-like cry in newborns.

The honesty you seek with superior technology

simple, safe, true.

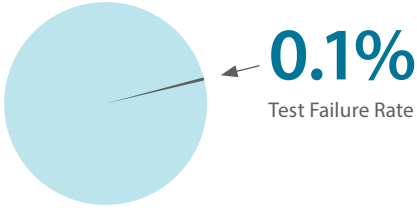
Why should I choose the ***Generation***® test over other tests?

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.**
- ✓ **It's reliable.**
  It has the lowest test failure of any NIPT (0.1%)[1];



- ✓ **It's fast.**
  Your healthcare provider will receive results within 3-7 business days from receipt in our laboratory.

How will my test be reported?

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

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
3. ***Aneuploidy Suspected*** — a borderline result, which occurs very rarely. This suggests there might be too many (or too few) copies of a chromosome present. Your provider may advise a diagnostic test for confirmation

Sex chromosome results will be reported as either *No Aneuploidy Detected* or *Aneuploidy Detected*.

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

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

The ***Generation***® test only addresses aneuploidies of chromosomes 21, 18, 13, and sex chromosomes.† It does 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

Generation®
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Results you can trust with proven studies[1,2,3,4]

simple, safe, true.

How do I know I can trust the *Generation*® test?

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[1]. 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 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 genomicdiagnostics.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 documents 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

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Results you can trust

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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. The total out pocket cost to the patient for the Generation test for chromosomes 13, 18 21 and sex chromosomes is $395 (or $450 if Generation Plus is requested) which is payable prior to sample collection by calling our Customer Care Team on 1800 822 999.

To learn more about the ***Generation***® and ***Generation***® ***Plus*** prenatal tests, please visit **www.genomicdiagnostics.com.au**

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.

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The honesty you seek

simple, safe, true

- *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 3-7 days from*

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

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