Non-invasive pre-natal testing

A GUIDE FOR PARENTS-TO-BE

Call 1800 822 999 for more information
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 also screens for gender identification and is available from as early as the 10th week of pregnancy, for both singleton and twin pregnancies.

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 (from 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)

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.
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.

*Generation* looks for too few (missing) or too many (extra) copies of chromosomes, which are often associated with these disabilities. The most commonly seen include an extra copy of a chromosome called a trisomy. *Generation* also screens for abnormalities of the sex chromosomes and can identify the gender of your baby at your request.

Generation screens for:

**Common Trisomies:**
- ✓ Trisomy 21 Down syndrome
- ✓ Trisomy 18 Edwards syndrome
- ✓ Trisomy 13 Patau syndrome

**Sex chromosome abnormalities*:**
- ✓ Monosomy X Turner syndrome
- ✓ XXY Klinefelter syndrome
- ✓ XXX Triple X
- ✓ XYY Jacobs syndrome

**Gender Identification**

*Sex chromosome aneuploidy testing is available for singleton pregnancies only

If you and your healthcare provider choose Generation Plus, you can also screen for the following genetic conditions:

- **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* 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 overeating 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.
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.

<table>
<thead>
<tr>
<th>chromosome</th>
<th>Observed Sensitivity</th>
<th>Observed Specificity</th>
</tr>
</thead>
<tbody>
<tr>
<td>Trisomy 21</td>
<td>99.1%</td>
<td>99.9%</td>
</tr>
<tr>
<td>Trisomy 18</td>
<td>98.3%</td>
<td>99.9%</td>
</tr>
<tr>
<td>Trisomy 13</td>
<td>98.2%</td>
<td>99.9%</td>
</tr>
<tr>
<td>Monosomy X</td>
<td>95.0%</td>
<td>99.0%</td>
</tr>
<tr>
<td>XX</td>
<td>97.6%</td>
<td>99.2%</td>
</tr>
<tr>
<td>XY</td>
<td>99.1%</td>
<td>98.9%</td>
</tr>
</tbody>
</table>

✓ **It’s tested in Australia.**

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

![0.1% Test Failure Rate](image)

✓ **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

If the sex chromosome option is ordered, 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,⁶ if ordered. It does not test for, or report all, genetic and non-genetic problems that may be present in a baby.

¹Sex chromosome aneuploidy testing is available for singleton pregnancies only

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**TM**

*With superior technology
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.

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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 generationNIPT.com.au)
3. Contact our Customer Care Team on 1800 822 999 to make a booking at one of our conveniently located collection centres and pay over the phone.
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** with proven studies\(^1,2,3,4\) simple, safe, true.
What does Generation cost?

The Generation non-invasive prenatal test does not qualify for a Medicare rebate. Payment is made at the time of booking your appointment. The total out-of-pocket costs to the patient are as follows:

- **Generation** $395
- **Generation Plus** $495

Generation is the most accessible NIPT screen in Australia

To learn more about the Generation prenatal test, please visit www.generationNIPT.com.au

References


Continue your pregnancy healthcare journey with Cell Care

Begin your pregnancy health care journey with Generation NIPT. Then stay at the forefront of medical science by storing your baby’s cord blood and tissue stem cells.

Cell Care, Australia’s largest and most experienced cord blood and tissue bank, is partnering with Generation to make NIPT accessible to more Australians.

Order Generation NIPT, then store your baby’s stem cells with Cell Care and receive up to $250 off cord blood and tissue storage.

How does it work?

- Order
  - **Generation** $395, or
  - **Generation Plus** $495
- Receive results
- Receive Cell Care Generation rebate voucher
- Order cord blood and tissue storage with Cell Care
- Redeem Generation rebate voucher of up to $250*

For more information and pricing, visit generationNIPT.com.au

*Upon enrolment with Cell Care, clients who have purchased Generation or Generation Plus will be eligible for a rebate of $200 off the price of cord blood or $250 off the price of cord blood and tissue. For full terms and conditions visit www.generationNIPT.com.au. This offer may be used in conjunction with other Cell Care offers.
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 receipt in our lab

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