Genomic Diagnostics

Generation Non-invasive Prenatal Testing

A guide for parents-to-be

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What is the *Generation* range of non-invasive prenatal tests?

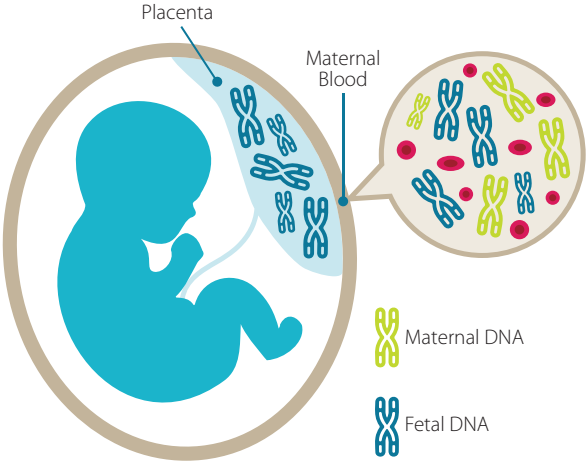
Generation is non-invasive prenatal testing (NIPT) that screens for chromosomal changes that can affect your baby's future health. NIPT uses a blood sample from the mother to determine the chance of a chromosomal condition in the developing baby. Using ***Generation*** may help avoid more invasive prenatal testing by amniocentesis and chorionic villus sampling.

Why *Generation* NIPT?

- ✓ **Simple and safe:** a one-tube blood test that poses no risk to your baby
- ✓ **Convenient:** available at a collection centre near you from as early as the 10th week of pregnancy
- ✓ **Fast results:** from 3-5 days from the sample arriving at the laboratory*
- ✓ **Reliable:** expect a result the first time, eliminating stress and time wasted in recollection
- ✓ **Accurate** (detecting Down syndrome with a >99.9% accuracy) and backed by extensive scientific data**

How does *Generation* work?

Our genetic information is found in our DNA, on string like structures called chromosomes. Healthy people usually have 46 chromosomes, made up of 23 chromosome pairs (numbered 1 through 22, and either XX or XY).



Extra or missing chromosomes can lead to chromosomal conditions that cause miscarriage or result in the birth of a child with mental or physical disabilities.

Fragments of DNA, called cell free DNA, are found in our blood. During pregnancy, some of the cell free DNA in the mother's blood comes from the baby's placenta. By sampling the mother's blood, ***Generation*** tests this cell free DNA from the placenta to determine the chance that the baby has a chromosomal condition.

What does *Generation* detect?

Genomic Diagnostics provides you with three options for NIPT: ***Generation, Generation 46,*** and ***Generation Plus***. Please discuss with your doctor which one is right for you.

All ***Generation*** options look for too few (missing) or too many (extra) copies of chromosomes. All options can also tell you the sex of the baby if that is something you want to know.

The standard ***Generation*** option screens for the most commonly seen chromosomal conditions:

- Down syndrome (trisomy 21)
- Edwards syndrome (trisomy 18)
- Patau syndrome (trisomy 13)
- Sex chromosome disorders (conditions caused by missing or extra sex chromosomes, X and Y)

Generation 46 screens for changes in all 46 chromosomes, including trisomy 21, 18 and 13 and the sex chromosome number changes screened in the standard ***Generation*** option. It not only screens for extra or missing whole chromosomes but also for extra or missing parts of the baby's chromosomes. Changes in chromosomes other than 21, 18, 13, X and Y are rare, but can provide your doctor with important information on the health of your pregnancy.

Generation Plus screens for all the conditions included in the standard ***Generation*** option, as well as a range of specific small microdeletion syndromes where very small bits of the chromosome are missing, including Di George syndrome (otherwise known as 22q11 deletion syndrome). It does not test all 46 chromosomes.

More information on the chromosomal conditions tested by ***Generation*** can be found on our website.

Is *Generation* accurate?

NIPT is a highly accurate screen for chromosomal conditions and is much better than older style screening tests at identifying whether a baby is or isn't affected by the conditions being tested. However, sometimes a pregnancy can be identified as high risk by NIPT but the baby does not have a chromosomal condition. The chance of this happening often depends on the age of the mother and the specific condition. The fact that this can happen is the reason that all high risk results should be confirmed by a diagnostic test (amniocentesis or chorionic villus sampling) to clarify if the baby does or does not have a chromosomal abnormality.

All of this can be explained by the genetic counsellor during genetic counselling that is available for all high risk NIPT results from Genomic Diagnostics.

Is the *Generation* test right for me?

Generation offers parents-to-be a new choice to obtain important information about the health of their developing baby, simply, accurately and at any time from 10 weeks throughout the pregnancy, with little or no risk to you or your baby.

The Royal Australian and New Zealand college of Obstetricians and Gynaecologists (RANZCOG) recommend that all pregnant women, regardless of risk status, be offered the opportunity for discussion and choice regarding prenatal screening for common chromosomal conditions**.

You may particularly wish to consider ***Generation*** NIPT 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" maternal 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)

It is recommended that you continue to have ultrasound scans as they can detect physical abnormalities which cannot be detected by the ***Generation*** screen.

What is included in the *Generation* report?

Your NIPT report will include one of two possible results for chromosomes 21, 18, 13, and the sex chromsomes (X and Y). If you have a ***Generation 46*** or ***Generation Plus*** NIPT, it will also include information on other chromosomes and microdeletions.

Low risk result - this means that it is very unlikely that your pregnancy is affected by the chromosomal conditions screened by the test.

High risk result - this means that there is an **increased chance** that your baby has too many or too few copies of one or more chromosomes or parts of chromosomes. However, as ***Generation*** is a screening test, a diagnostic test such as amniocentesis or chorionic villus sampling (CVS) is required to confirm a high risk result.

If your result shows a high risk for having a baby with a chromosomal condition, we will arrange for a **genetic counsellor** to contact you within 48 hours on request of your doctor. They will discuss with you what the result means, the options available and answer any questions you may have. This is a free service. You should also discuss the result with your doctor.

Fetal Sex: An additional benefit to ***Generation*** is that you may choose to have the sex of your baby revealed. This is automatically reported unless you opt out on the request form.

Very rarely your ***Generation*** screen may not provide a result. There are a variety of reasons why this may happen. Your doctor will guide you through the next steps of managing your pregnancy if this occurs.

How do I organise to have a Generation NIPT?

Step 1: See your doctor to get a request

Discuss the ***Generation*** options with your doctor and complete the request form .

Step 2: Prepare for your collection

Medicare and private health insurance do NOT cover the cost of NIPT. Prepay online via generationNIPT.com.au. If you are unable to pay online contact our Customer Care Team on 1800 822 999. Visit genomicdiagnostics.com.au for current pricing on all ***Generation*** options.

Step 3: Have your sample collected

Note the receipt number on the request form and take to your closest Healius Pathology network collection centre.

Step 4: See your doctor to get your results

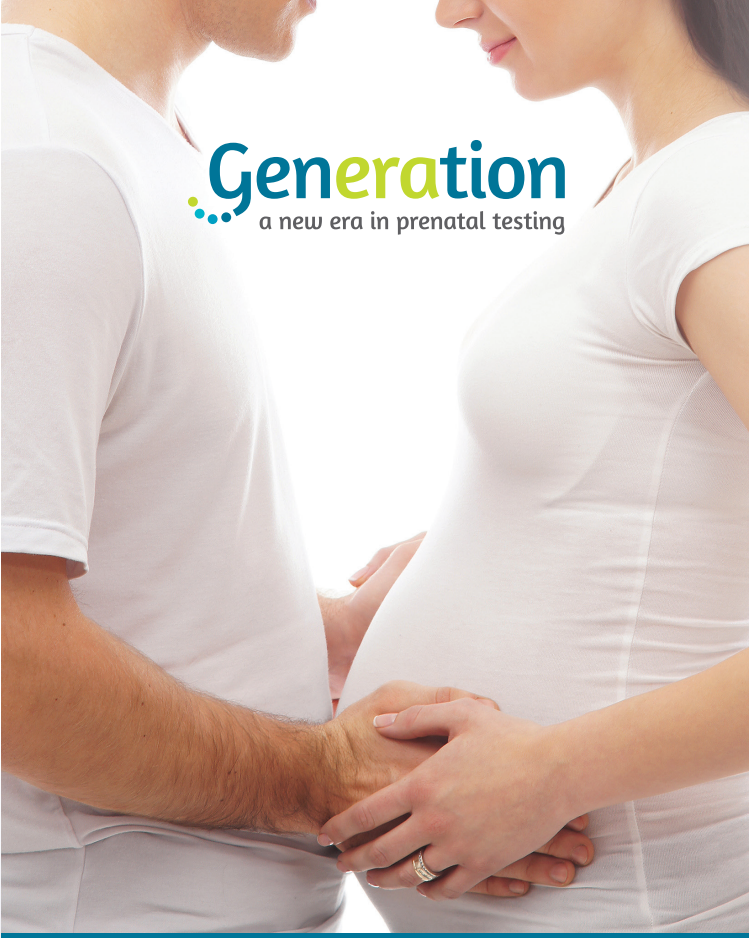
Your ***Generation*** or ***Generation 46*** results will be delivered to your doctor 3-5 days from the sample arriving at the laboratory*, or 9-13 days for ***Generation Plus***.

Does a low risk ***Generation*** result mean that my baby will be perfectly healthy?

All ***Generation*** options provide highly accurate screening for chromosomal disorders. However, ***Generation*** does not screen for all genetic and non-genetic conditions that may be present in a baby. No test can guarantee that a baby will not have any medical issues.

* Samples may take several days to reach the laboratory if coming from regional Australia, or over the weekend or a public holiday.

** Please refer to **www.genomicdiagnostics.com.au** for references and supporting scientific material.



Generation NIPT: A Quality Focus

Generation was chosen for development by Genomic Diagnostics based on a careful evaluation of quality and proven scientific performance, giving you confidence of an accurate result.

Generation NIPT also has the lowest test failure rate of any non-invasive prenatal test, providing an accurate result at fetal fraction levels of <3%.

Please refer to genomicdiagnostics.com.au for more information on ***Generation*** NIPT.

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