genomic diagnostics

A Healius company

Generation NIPT

Setting the benchmark in prenatal testing

What is Generation?

The Generation suite of NIPT options incorporates Generation, Generation 46 and Generation Plus.

Non-Invasive Prenatal Testing (NIPT) represents a major advance in screening and risk assessment for chromosomal abnormalities.

These are highly sensitive, accurate, non-invasive prenatal screening tests for fetal chromosomal and subchromosomal aneuploidies. The tests use whole genome sequencing (WGS) to analyse circulating cell-free fetal DNA from a maternal blood sample from as early as 10 week's gestation.

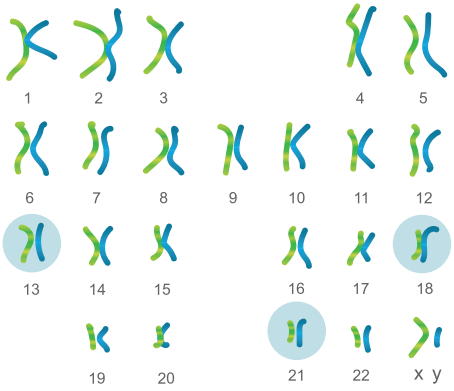
Multiple published studies of thousands of pregnant women have demonstrated the clinical utility and benefit of WGS-based NIPT, regardless of age and risk category, for both singleton and twin pregnancies.[1,2,3]

Clinical best practice guidelines from Australian and international medical societies recommend that available prenatal screening tests, including NIPT, be discussed and offered to all pregnant women.[4,5]

Our suite of Generation NIPT tests is used to identify pregnancies at increased risk of chromosomal disorders and other pregnancy complications.

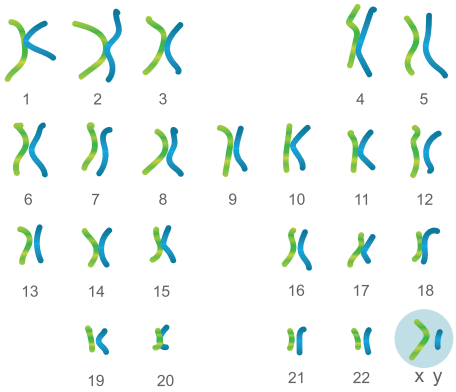
Common trisomies:

Down syndrome (T21), Edward syndrome (T18) and Patau syndrome (T13)



Sex chromosome conditions (SCA):

Including Turner syndrome (monosomy X) and Klinefelter syndrome (XXY), and XYY syndrome



Rare chromosomal and subchromosomal aneuploidies (gains or losses of whole or part of a chromosome):

Associated with significant complications such as miscarriage, IUGR, and UPD, and specific syndromes.

Generation NIPT has the lowest reported test failure rate

Our Experience* <0.5% Test Failure Rate

Test failures matter in NIPT, as they increase the risk of false negative and false positive results. There is the potential to increase false negative results if no action is taken following a test failure. A higher rate of aneuploidy in test failure samples also means that there is potentially increased invasive test utilisation for those returning a "high risk" result with other testing modalities.

Test failures also lead to increased stress for the patients, longer turnaround times and increased clinician visits, with high failure rates demonstrated for redraws from these patients.[6]

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

Available Generation testing options

Generation

Generation screens for the most commonly seen and tested chromosomal abnormalities including:

- Trisomy 21 (Down syndrome), Trisomy 18 (Edwards Syndrome) and Trisomy 13 (Patau syndrome)
- Specific sex chromosome aneuploidies (monosomy X, XXX, XXY and XYY)
- Fetal sex

The ***Generation*** test is appropriate for aneuploidy screening in unselected/low risk patients.

Testing is performed in Australia and is NATA/RCPA accredited, with results available 3-5 days from the sample arriving at the laboratory*.

Generation 46

Generation 46 expands on the Generation screen to include detection of aneuploidy for all 23 pairs of chromosomes:

Screening of the whole genome for chromosomal and subchromosomal aneuploidy >7Mb, including trisomies 21, 18, and 13 included in the ***Generation*** screen.

- Specific sex chromosome aneuploidies (monosomy X, XXX, XXY and XYY)
- Fetal sex

The ***Generation 46*** test is appropriate for aneuploidy screening in unselected/low risk patients. It is optimised to screen for gains and losses of at least 7Mb across the genome (excluding chromosomes X and Y). Screening for specific microdeletion syndromes is addressed by the ***Generation Plus*** test.

Testing is performed in Australia and is NATA/RCPA accredited, with results available 3-5 days from the sample arriving at the laboratory*.

Generation Plus

Generation Plus screens for the most commonly tested chromosomal abnormalities from the Generation test, as well as more rarely occurring genetic abnormalities including:

- 22q11 deletion (DiGeorge syndrome)
- 15q11 deletion (Angelman/Prader-Willi)
- 1p36 deletion syndrome
- 4p deletion (Wolf-Hirschhorn syndrome)
- 5p (Cri-du-chat)

The ***Generation Plus*** test should be considered when there are specific clinical indications for an increased risk of one of the test microdeletion syndromes. This includes ultrasound imaging suggesting a specific microdeletion syndrome, and previous history of a pregnancy diagnosed with, or a child affected with, one of these conditions.

This test has been optimised to specifically detect the microdeletion syndromes above, but does not include genome-wide screening.

Testing is performed in an accredited laboratory in California, with results available 9-13 days from sample arriving at the laboratory*.

All Generation Options

- Testing can be performed from 10 weeks until the end of the pregnancy
- Testing can be performed on singleton, egg donor and surrogate pregnancies
- Testing is available for twin pregnancies for ***Generation*** and ***Generation 46*** but excludes SCA screening and fetal sex determination
- Genetic counselling is available free of charge for some patients with high risk (aneuploidy detected) results. Please contact us for further information.

Generation, Generation 46 and ***Generation Plus*** do not test for any genetic conditions not listed above, such as family specific mutations (such as cystic fibrosis). Testing for these conditions may be available by invasive methods. Please contact us if you require further information about this. Non-genetic conditions (such as neural tube defects) are also not tested by NIPT.

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

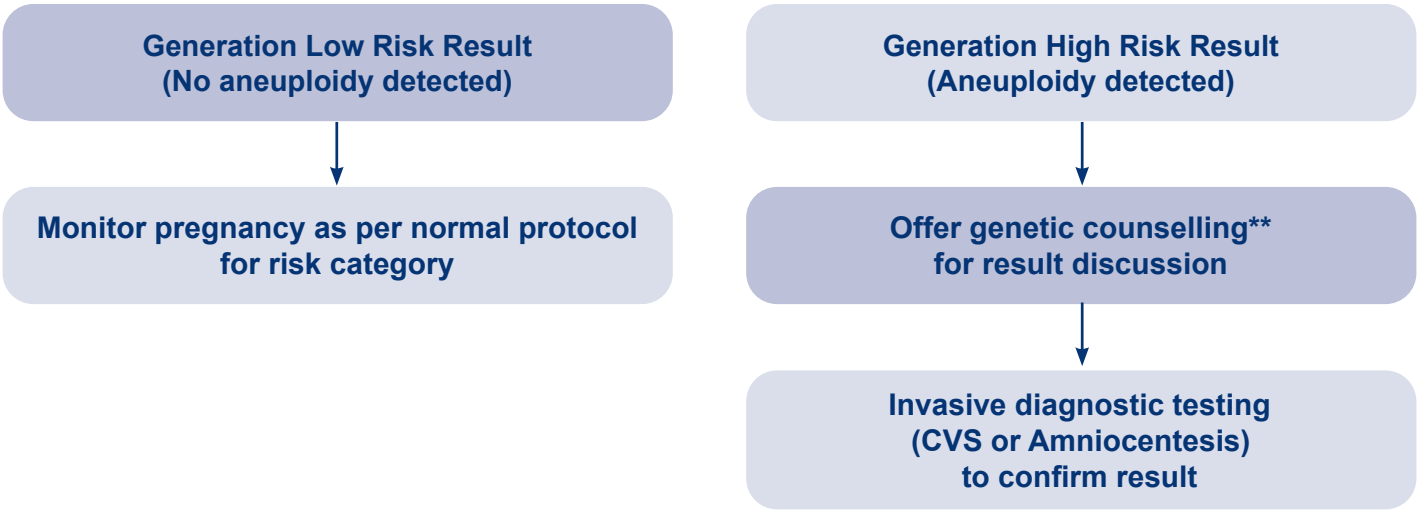
Accuracy of Generation NIPT

NIPT for fetal chromosomal aneuploidies has the highest detection rate and lowest false positive rate of available prenatal screening methods. Combined with the lowest reported failure rate, Generation NIPT ensures that more patients will avoid invasive diagnostic procedures.

NIPT is a highly accurate screening test. This means that if there is a true chromosomal aneuploidy in the fetus, it is highly likely that it will be detected by NIPT. However, a more clinically useful statistic is the positive predictive value (PPV) which is the chance that a high-risk correctly identifies true aneuploidy in the fetus. PPV is different for every woman, and is determined by factors including maternal age, type of aneuploidy, and personal and family medical history. For example, the chance of Down syndrome following a high risk test result is 94% for a 40-year-old woman and 63% for a 30-year-old woman. This variability in PPV, due to biological and technical false positive results, means that diagnostic testing by amniocentesis or chorionic villus sampling is strongly recommended prior to making definitive decisions regarding further pregnancy management.

Genomic Diagnostics provides a free genetic counselling service to explain the individual risks to every woman who has a high-risk result.

Appropriate follow up after NIPT



**Current RANZCOG and international guidelines recommend genetic counselling for women with high risk results.

We offer a genetic counselling service for patients with high risk (aneuploidy detected) results. Please contact us for further information.

References:

1. Taneja et al. Prenat Diagn 2016 36(3);237-24 (PMID: 2671519)
2. Van der Meij et al. Am J Hum Genet 2019;105(6):1091-1101 (PMID: 31708118)
3. Gil et al. Ultrasound Obstet Gynaecol 2019;53(6):734-742. (PMID: 31165549)
4. RANZCOG guidelines for Prenatal screening and diagnostic testing for fetal chromosomal and genetic conditions, available at https://ranzcog.edu.au/RANZCOG_SITE/media/RANZCOG- MEDIA/Women%27s%20Health/Statement%20and%20guidelines/Clinical-Obstetrics/Prenatal-screening_1.pdf?ext=.pdf.
5. ACOG bulletin 226, Screening for Fetal Chromosomal Abnormalities, available at https://www.acog.org/clinical/clinical-guidance/practice-bulletin/articles/2020/10/screening-for-fetal-chromosomal-abnormalities.
6. Pergament et al. Obstet Gynecol 2014; 124(201):210–218. (PMID: 250043544)
7. Illumina Veriseq NIPT Solution V2 Kit Insert available at https://support.illumina.com/content/dam/illumina-support/documents/documentation/chemistry_documentation/veriseq-nipt-v2/veriseq-nipt-solution-v2-package-insert-1000000078751-02.pdf

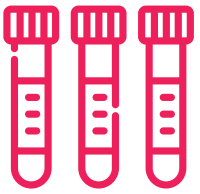
Arranging Generation testing

Step 1: Patient consultation

- Discuss Generation NIPT options with your patient
- Provide your patient with the request form and ensure Patient Consent section signed
- Request form available through patient management software, online at genomicdiagnostics.com.au or in hard copy pads

Step 2: Prepare for collection

- Patient is required to pay for their Generation test prior to having their blood taken
- Payment occurs online at generationnipt.com.au or over the phone on 1800 822 999
- At the time of payment, the patient can select their most convenient collection centre
- Patient notes their receipt number on the request form



Step 3: Sample collection

- Patient attends collection centre with their signed request form
- Blood collected
- Generation testing is performed

Step 4: Result discussion

- Results are delivered to you by preferred method
- Genetic Counselling is provided for patients with a high risk result if required
- A specific referral form is provided with all high risk results, to be completed should the patient wish to undertake counselling.

This test is NOT covered by Medicare or private health funds.

Prepayment is required before test is collected.

Why choose Genomic Diagnostics?

We're one of Australia's longest running specialist DNA testing laboratories.

We bring you depth of knowledge and experience, state of the art facilities and dedicated scientists and pathologists who care about what they do. We've been performing NIPT for over 8 years and have provided testing for many thousands of pregnancies in that time.

Quality and Accreditation

Our laboratory has held continuous accreditation to all relevant medical testing standards for all the tests we do. We pride ourselves on providing you with quality results and expert testing advice that you can rely on.

We're Convenient

We're part of the Healius Pathology network of pathology laboratories across Australia, which means wherever your patients are, there's a collection centre nearby.

Dedicated, friendly and knowledgeable Customer Care team

Our customer care team specialize in taking enquiries. With extensive experience in the field, they're on hand to provide you with the right advice, or direct your enquiry, based on your requirements.

For more information, contact us at **info@genomicdiagnostics.com.au**

1800 822 999 **genomicdiagnostics.com.au**

Healius companies

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Healius Pathology Pty Ltd (ABN 84 007 190 043) t/a Genomic Diagnostics PUB/MR/1338_V7_Sep24