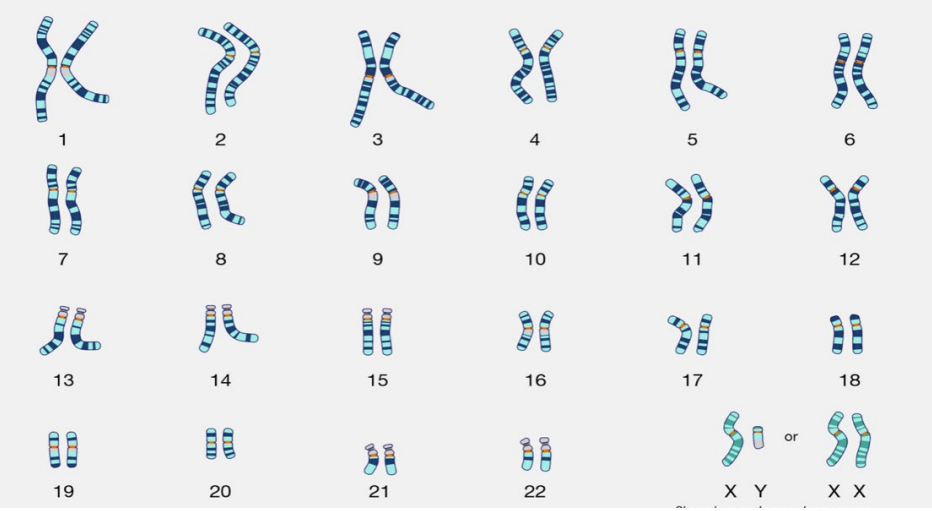
Women's Health Circle
NIPT (Non-invasive prenatal testing) Screening

Background to NIPT- the Genome



A normal genome consist of 23 pairs of chromosomes, including one pair of sex chromosomes (XX for female, XY for male).

NIPT tests screen for additional/ missing material by analysing a marker called cell free DNA in your blood. This marker is released by the early placenta and contains **fetal DNA**.

NIPT testing cannot screen for every genetic condition- but can provide accurate information about the risk of more common genetic disorders.

As NIPT is a **screening test**, if you receive a high risk result you will be counselled about further **diagnostic testing**.

There is more than one type of NIPT testing!

Different pathology providers offer slightly different variations of NIPT testing.

At the Women's Health Circle, we offer Percept and Panorama testing on site with one of our doctors.
Each test is combined with an ultrasound to confirm ongoing viability of your pregnancy prior to paying for testing.
We recommend completing this testing around 10-11 weeks.

*Please note that **Panorama** tests can only be done Monday- Thursday before 2pm for transport reasons

Percept screens for the three most common aneuploidies (chromosomal problems) and sex chromosome disorders. **Panorama** provides the option of testing for triploidy and other microdeletion syndromes.

Aneuploidies	**Detection rate – PERCEPT**	**Detection rate- PANORAMA**
Down Syndrome (T21)	99%	99%
Edward's Syndrome (T18)	98%	98%
Patau Syndrome (T13)	99%	99%
Triploidy (three copies of all chromosomes)	Not available	>99%
Fetal Sex (optional)	99%	99%
Sex chromosome disorders		
Turner's syndrome (X0)	95%	95%
Klinefelter's syndrome (XXY)	Limited data	73%
Triple X (XXX)	Limited data	73%
Jacob's syndrome (XXY)	Limited data	73%
Extended panel	**Not available**	**Optional**
Di George Syndrome- 22q11.2		95.7%
Prader Willi Syndrome		93.8%
Angleman Syndrome		95.5%
Cri du chat		99%
1p36 deletion		>99%

Which one should I choose?

- Everyone is different and has different preferences about genetic screening. A Panorama test can screen more accurately for sex chromosome disorders, microdeletions and triploidy.

Percept	Panorama
• Reassured in a low risk result for the most common aneuploidies • No family history of genetic conditions • Previous pregnancies uncomplicated • You are less likely to consider interrupting the pregnancy due to diagnosis of a rare genetic condition	• Want maximum information • Family history of genetic disorders • Would consider interrupting the pregnancy due to diagnosis of a genetic syndrome • Previous pregnancy terminated due to diagnosis of a genetic syndrome • Have another child affected by a genetic condition

What if I get a high risk result?

- If you get a high result, we will contact you to arrange further counselling and discuss options for diagnostic testing (chorionic villus sampling or amniocentesis)

What happens if I receive an invalid result?

- We will discuss options of a repeat NIPT test or diagnostic testing.

How long will it take to receive results?

- Turn-around time for Percept is 5-7 days
- Turn-around time for Panorama is 10-14 days

Aneuploidies

Syndrome	Incidence	Life expectancy	Mental effects	Physical effects
Down Syndrome T21	1/2000	Reduced	Intellectual impairment	Syndromic features Heart defects Hearing and visual impairment Gastro-intestinal problems Immune deficiency Anaemia
Edward Syndrome T18	1/3600-1/10000	Significantly reduced- may not survive infancy	Severe intellectual impairment	Syndromic features Heart defects Significant motor impairment Seizures Lung/ breathing problems Gastro-intestinal problems Kidney problems
Patau Syndrome T13	1/3000	Significantly reduced- may not survive infancy	Severe intellectual impairment	Syndromic features Heart problems Significant motor impairments Cleft lip and palate- feeding problems Sex organ malformation
Triploidy	1/2000	Hours/days	-	-

https://www.cdc.gov/ncbddd/birthdefects/downsyndrome/data.html
https://www.ncbi.nlm.nih.gov/books/NBK570597/
https://www.rarechromo.org/media/information/Other%20Topics/Triploidy%20FTNP.pdf

Sex chromosome Aneuploidies

Syndrome	Incidence	Life expectancy	Mental effects	Physical effects
Turner's syndrome (XO)	1/2000	Normal		Syndromic features Heart failure Hearing problems High blood pressure Kidney problems Higher rate of thyroid problems and diabetes Issues with sexual development/ infertility
Klinefelter's syndrome (XXY)	1-2.5/1000	Normal	Mild intellectual impairment	Taller, less muscular body Larger breasts Weaker bones Lower energy level Small penis and testicles Delayed/ incomplete puberty
Triple X (XXX)	1/1000	Normal	Mild intellectual impairment	Taller than average Low muscle tone Widely spaced eyes
Jacob's syndrome (XYY)	1/1000	Normal	Mild intellectual impairment	Tall stature Macrocephaly (large head) Widely spaced eyes Seizures Infertility

https://www.betterhealth.vic.gov.au/health/conditionsandtreatments/turners-syndrome#diagnosis-of-turner's-syndrome
https://kidshealth.org/en/parents/klinefelter-syndrome.html
https://www.uptodate.com/contents/clinical-manifestations-and-diagnosis-of-turner-syndrome
https://kidshealth.org/en/parents/triple-x-syndrome.html
https://www.ncbi.nlm.nih.gov/books/NBK557699/

Microdeletions

Syndrome	Incidence	Life expectancy	Mental effects	Physical effects
Di George Syndrome	1/4000	Reduced	Mild-moderate intellectual impairment	Syndromic features Heart defects Electrolyte disturbances Kidney problems Cleft lip and palate, feeding issues Orthopaedic problems Growth problems Immune problems
Prader Willi Syndrome	1/10000	Reduced	Mild-moderate intellectual impairment Behavioural problems	Syndromic features Delayed motor development Obesity- insatiable appetite Visual impairment Reproductive problems- delayed puberty/ impaired development of genitals Skeletal problems
Cri-du-chat syndrome	1/25000-1/50000	Reduced	Moderate to severe intellectual impairment Behavioural problems	Syndromic features Heart defects Difficulties with suckling and swallowing Gastro-intestinal issues- reflux/ constipation Delayed motor development Immune deficiency Hearing impairment Skeletal problems
Angelman syndrome	1/12000	Normal	Intellectual impairment	Syndromic features Epilepsy/ seizures Delayed motor development Movement disorder (hand flapping/ jerking) Feeding problems
1p36 deletion	1/5000	Normal	Intellectual impairment	Syndromic features Epilepsy/ seizures Delayed motor development Movement disorder (hand flapping/ jerking movements) Feeding problems