NUHS
National University Health System

National University Centre for Women & Children

Screening for Chromosomal Abnormalities

Purpose of Screening Tests

Every pregnancy is associated with a small risk of having a genetic disorder, which are abnormalities involving the genetic makeup of the baby.

The most common genetic disorder is Down syndrome, where there is excessive genetic material from an extra chromosome 21. Likewise, Patau syndrome and Edwards syndrome are genetic disorders caused by excessive genetic materials from an extra copy of chromosome 13 or 18 respectively.

These genetic disorders can result in moderate to severe intellectual disability and frequently, heart defects and other physical abnormalities. They can happen to an individual who does not have any family members with the condition.

Screening tests are available to assess the risk of the foetus having the above disorders.

Types of Screening Tests

A number of tests are available. They are done at different times of a pregnancy and have different detection rates.

1. Nuchal Translucency Scan
2. Nuchal Translucency and First Trimester Serum Screening (Combined Screening)
3. Non-Invasive Prenatal Testing (NIPT)

Screening tests do not diagnose an abnormality — they only assess the risk or chance of having an abnormality.

1. Nuchal Translucency Scan

Nuchal translucency (NT) describes a fluid-filled (sonolucent) area at the back of the neck (nuchal region) of the foetus. Measuring the thickness of this area at a specific place is called NT measurement. This provides an assessment of the individual specific risk of having a baby with Down syndrome, Patau syndrome and Edwards syndrome, as well as other genetic conditions and structural differences. Individual risk depends on the age of the mother, the size of the baby and the NT measurements.

How is it done?

It is done through an abdominal ultrasound scan.

When should the test be done?

This test is done between 11 and 14 weeks of pregnancy.

How long does it take to obtain the result?

The result is known soon after the ultrasound scan is completed. A report of your individual specific risk will be produced.

What is the detection rate?

Using nuchal translucency screening alone, the detection rate for these chromosomal disorders is approximately 80%. A thickened nuchal translucency may also be associated with genetic conditions other than Down syndrome, Patau syndrome and Edwards syndrome, and may also be found in foetuses with a structural difference such as a congenital heart defect or certain genetic abnormalities.

2. Nuchal Translucency and First Trimester Serum Screening (Combined Screening)

The combined screening, previously known as OSCAR (OneStop Clinic for Assessment of Risk), combines ultrasound measurements of nuchal translucency (the fluid behind the baby's neck) with maternal blood tests (PAPP-A and free beta-hCG) to screen the baby for Down syndrome, Edwards syndrome and Patau syndrome.

How is it done?

It is done through an abdominal ultrasound scan to measure the nuchal translucency together with a blood test to measure the two proteins (free HCG and PAPP-A).

When should the test be done?

The test is done between 11 and 14 weeks of pregnancy.

How long does it take to obtain the result?

It takes about 4-7 days to obtain a result.

What is the detection rate?

The detection rate of the combined screening for Down syndrome, Patau and Edward syndromes is approximately 90%.

3. Non-Invasive Prenatal Testing

Non-Invasive Prenatal Testing (NIPT) analyses the DNA from the placenta circulating in the mother's blood which can be used to screen the baby for Down syndrome, Edwards syndrome and Patau syndrome. Performing an NT scan to assess the baby's structure is still recommended in addition to performing NIPT.

How is it done?

It is done by taking a blood sample from the mother.

When should the test be done?

The test can be done anytime after 10 weeks of pregnancy, but it should ideally be performed after the NT scan has been done and shows a normal baby's structure.

How long does it take to obtain the result?

It takes about 10-14 days to obtain a result.

What is the detection rate?

The detection rate of NIPT is 99% for Down syndrome, 98% for Edwards syndrome, and 92% for Patau syndrome.

What happens if a screening test is positive?

As these are screening tests, an abnormal result does not necessarily mean that the foetus has an abnormality. It only means that there is a significantly higher risk of having an abnormality. If the screening test is positive, the couple would be offered diagnostic tests such as chorionic villus sampling (CVS) or amniocentesis to rule out chromosomal defects (refer to our brochure on 'Diagnostic Tests for Chromosomal Abnormalities').

The diagnostic tests are extremely accurate in detecting chromosomal abnormalities. The possibility of a false result is less than 1 in 40,000 cases.

Does it mean that my baby is normal if the test is negative?

A negative test result does not necessarily mean that the baby is normal in every aspect. Although the tests can detect some of the more common chromosomal abnormalities, it does not guarantee a normal baby. Since these are screening tests, they only assess whether the baby is at a high risk of having chromosomal disorders. There are false negatives in all these tests as the detection rates are not 100%. They do not predict other forms of foetal abnormalities such as structural abnormalities.

Scan this QR code for a video resource on Down syndrome screening in pregnancy.

About the National University Centre for Women and Children

National University Centre for Women and Children (NUWoC) is a national university specialist centre that aims to empower women, children and their families to lead healthier lives. We provide comprehensive medical and surgical services ranging from pre-conception to child and maternal health.

NUWoC comprises the Department of Obstetrics & Gynaecology (O&G) and Khoo Teck Puat – National University Children's Medical Institute (KTP-NUCMI) of National University Hospital. It focuses on the right-siting of appropriate services in the community and builds complementary services in National University Health System's (NUHS) centres of excellence – Ng Teng Fong General Hospital and Alexandra Hospital.

Through a generous gift from the Estate of Khoo Teck Puat, KTP-NUCMI established an integrated outpatient facility with medical, diagnostic and rehabilitation services for children. We are also the only public specialist centre in Singapore that offers paediatric kidney and liver transplant programmes.

For more information about us, visit www.nuh.com.sg/NUWoC

Emergency (24-hr)

Location	NUH Main Building, Zone F, Level 1
Contact	+65 6772 5000

Women's Clinic – Emerald/Ruby

Location	NUH Kent Ridge Wing, Zone D, Level 3, D03-06
Operating Hours	8.30am – 6pm (Mon to Thu), 8.30am – 5.30pm (Fri), 8.30am – 12.30pm (Sat)
Email	appointment@nuhs.edu.sg

Women's Clinic - Sapphire

Location	NUH Kent Ridge Wing, Zone D, Level 3, D03-03
Operating Hours	8.30am – 6pm (Mon to Thu), 8.30am – 5.30pm (Fri), 8.30am – 12.30pm (Sat)
Email	appointment@nuhs.edu.sg

Women's Clinic – Jade [Former Clinic G]

Location	NUH Kent Ridge Wing, Zone C, Level 3, C03-02
Operating Hours	8.30am – 6pm (Mon to Thu), 8.30am – 5.30pm (Fri)
Email	appointment@nuhs.edu.sg

Fetal Care Centre

Location	NUH Kent Ridge Wing, Zone D, Level 3, D03-04
Operating Hours	8am – 5.30pm (Mon to Thu), 8am – 5pm (Fri)
Email	appointment@nuhs.edu.sg

Clinic for Human Reproduction

Location	NUH Kent Ridge Wing, Zone D, Level 4, D04-02
Operating Hours	8am – 5pm (Mon to Fri), 8.30am – 12.30pm (Sat)

Women's Clinic @ JMC

Location	Jurong Medical Centre, Level 2
Operating Hours	8.50am – 11.30pm (Tue & Thu), 2pm – 5pm (Mon & Fri)

Jurong Clinic for Women

Location	130 Jurong Gateway, #01-231
Operating Hours	9am – 12pm, 2pm – 5pm (Mon to Sat), 6pm – 9pm (Mon to Thu)
General Enquiry	+65 6665 4277
Appointment Line	+65 6908 2222
Email	appointment@nuhs.edu.sg

GS @ NTFGH

Location	Ng Teng Fong General Hospital, Tower A – Specialist Outpatient Clinics, Level 7
Operating Hours	8.30am – 5.30pm (Mon to Fri), 8.30am – 12.30pm (Sat)
Appointment Line	+65 6908 2222
Email	appointment@nuhs.edu.sg

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Information is correct at time of printing (Aug 2025) and subject to revision without prior notice.