

Most pregnancies are normal but about 2% are complicated by congenital anomalies. Down syndrome is one of the anomalies that can be detected before birth. About 1 in 700 newborn babies have Down syndrome, which occurs most often by chance and is seldom inherited from parents. This leaflet is to help you understand Down syndrome, available tests offered by the Hospital Authority (HA), and to help you decide whether you want the screening tests for Down syndrome or not.

What is Down Syndrome?

Most of us have 23 pairs of chromosomes in each cell. Individuals with Down syndrome have one extra chromosome 21 in their cells. Down syndrome is a condition with mental handicap and certain physical features different from other people. Some children with Down syndrome have abnormality such as heart defects.



Is there any advantage knowing if my baby has Down syndrome before birth?

This would allow parents and doctors to discuss options to the best interest of the family.

Can I know whether my baby has Down syndrome before birth?

Yes. A logical approach is to undergo a screening test to assess your risk (or chance) of having a baby with Down syndrome. The test does not harm you or your baby. It provides an estimated risk of your baby having Down syndrome, which is a more accurate estimate than that derived from your age alone.

The HA provides two screening tests according to the duration of pregnancy:

First trimester screening

If you are pregnant for less than 14 weeks, you will undergo an ultrasound examination for measurement of fetal size as well as fetal nuchal translucency at 11 to 13 weeks and 6 days of gestation. A blood test will be performed at the same time to measure the Pregnancy Associated Plasma Protein A (PAPP A) and human chorionic gonadotrophin (hCG) levels. 1st trimester screening detects around 90% of fetuses with Down syndrome.



Second trimester screening –

If you are pregnant for more than 14 weeks and less than 20 weeks, you will undergo the blood test at 16 to 19 weeks and 6 days for alpha-fetoprotein (AFP), human chorionic gonadotrophin (hCG), estriol (uE3) and Inhibin A. The 2nd trimester screening test detects around 80% of Down syndrome pregnancies.

How do I know if my screening test result is positive or negative?

The screening test result will be available within 1 week after the blood test. About 5% of women undergoing Down syndrome screening test will get a positive result. If your test result is positive, hospital staff will call you and arrange an appointment for counseling. If your test result is negative, you will read your report at the next antenatal visit.

If my screening test result is positive, how can I confirm if my baby has Down syndrome before birth?

You can undergo a diagnostic test in the form of chorionic villus sampling or amniocentesis. A diagnostic test will tell you accurately whether the baby has Down syndrome or not. This involves introducing a needle under ultrasound guidance into the uterus to draw placental tissue or amniotic fluid for chromosome study. Chorionic villus sampling is usually performed before 13 weeks and amniocentesis is usually performed between 16 to 20 weeks. Procedure related fetal miscarriage rate is 0.1-0.2%.

If you are worried about procedure related miscarriage risk, you can consider having non-invasive prenatal testing (NIPT) which has a higher detection rate of fetal Down syndrome than first and second trimester screening tests. This test is currently provided by private doctors and hospitals.

What will happen if the fetus is confirmed to have Down syndrome?

The doctor will explain to you and your spouse about the nature of the chromosomal abnormality, its impact on the fetus and the risk in future pregnancies. You can discuss with your doctor about available support from the hospital and other organizations and be better prepared for the birth of the baby. With special care and education, children with Down syndrome can live semi - independently.

However, if you prefer not to continue with the pregnancy after thorough consideration, you may discuss with your doctor about the possibility of legal termination of pregnancy before 24 weeks of gestation.

Does a negative screening test result guarantee a normal baby?

No. A negative screening test result indicates that the chance of your baby having Down syndrome is small, but it does not guarantee a normal baby.

Can I ask for a diagnostic test if the screening test result is negative?

No. In HA, a diagnostic test e.g. chorionic villus sampling or amniocentesis will only be offered to women with positive Down screening test results. Women with negative Down screening test results requesting diagnostic test or women who prefer direct diagnostic test should approach private doctors or hospitals.

Points to note:

- Down syndrome screening test is voluntary.
- Only first trimester Down syndrome screening test is available for multiple pregnancies.
- Join one screening program only. Repeated screening may result in confusion and unnecessary anxiety.
- A positive Down syndrome screening test report does not indicate that the fetus has Down syndrome. A negative screening test report cannot guarantee that the fetus is completely free of Down syndrome or that the fetus is 100% normal.



Prenatal Down syndrome screening

Tsan Yuk Hospital