Points to note

- Should you have any private dating ultrasound arranged, please show us for reference. If you already have a Down syndrome screening test performed at a private clinic, it is generally recommended not to repeat the test.
- If you would like to undergo other screening or diagnostic tests, you should approach organisations outside the Hospital Authority.
- Join one screening program only. If you join more than one screening program, the results may be confusing and may cause unnecessary anxiety.
- For twin pregnancy, Down syndrome screening is available only in the first trimester.

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. This leaflet is to help you understand Down syndrome, available tests offered by Hospital Authority (HA), and to help you decide whether you want the screening tests for Down syndrome or not.

What is Down syndrome?

About 1 in 700 newborn babies have 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 occurs most often by chance and is seldom inherited from parents. Drugs, X-rays or other environmental exposure does not incur Down syndrome.
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. With special care and education, some of them can integrate into normal schools and lead semi-independent lives.

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.

How can I tell whether my baby has Down syndrome before birth?

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 projects an estimated risk of your baby having Down syndrome, which is a more accurate estimate than that derived from your age alone.

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

First trimester screening -
You will undergo an ultrasound examination at 11 to 13 weeks and 6 days of gestation. A blood test will be performed during the same period of pregnancy to measure your Pregnancy Associated Plasma Protein A (PAPP-A) and human chorionic gonadotrophin (hCG) levels. The test detects around 80-90% of Down syndrome pregnancies. This test is not suitable for you if you are pregnant for more than 14 weeks.

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 gonadotropin (hCG), estriol (uE3) and Inhibin A. The 2nd trimester screening test detects around 80% of Down syndrome pregnancies.
For the above tests, about 5% of women will get a positive result, while about 95% will get a negative result.

<table>
<thead>
<tr>
<th>Down syndrome screening test</th>
<th>1st trimester screening 11-13 weeks 6 days</th>
<th>2nd trimester screening 16-19 weeks 6 days</th>
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</thead>
<tbody>
<tr>
<td>Nuchal measurement by ultrasound examination</td>
<td>✓</td>
<td></td>
</tr>
<tr>
<td>Maternal serum PAPP-A &amp; hCG</td>
<td>✓</td>
<td></td>
</tr>
<tr>
<td>Maternal serum AFP, hCG, uE3 &amp; Inhibin A</td>
<td></td>
<td>✓</td>
</tr>
<tr>
<td>Detection rate</td>
<td>80-90%</td>
<td>80%</td>
</tr>
<tr>
<td>False positive rate</td>
<td>5%</td>
<td>5%</td>
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</table>

The screening result will be ready within 2 weeks after the blood test. If the test result is negative, you will not hear from us. The report will be filed with your antenatal test results in your antenatal record.

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. It does not guarantee a normal baby.

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

In HA, a diagnostic test will only be offered to woman with positive screening test result. Women who would like to have chorionic villus sampling or amniocentesis should approach private doctors or hospitals.
If my screening test result is positive, how can I confirm if my baby has Down syndrome before birth?

You need to undergo a **diagnostic test** in the form of amniocentesis or chorionic villus sampling. 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 amniotic fluid or placental tissue. The materials contain cells from the pregnancy for chromosomal study. Amniocentesis is usually performed between 16 to 20 weeks, and chorionic villus sampling is usually performed before 13 weeks. They are associated with a 0.5-1.0% risk of miscarriage.

Can I ask for a diagnostic test without having the screening test first?

No. Women who would like to have chorionic villus sampling or amniocentesis should approach private doctors or hospitals instead.

What will happen if the foetus is confirmed to be abnormal?

The doctor will explain to you and your spouse about the nature of the abnormality, its effect on the foetus and the risk in future pregnancies. You can discuss with the doctor about available support from the hospitals and other organisations and be better prepared for the birth of your baby. If the pregnancy is not more than 24 weeks of duration, you may also seek advice from your doctor about the possibility of lawful termination of pregnancy under the laws in Hong Kong.

Must I have the screening test?

The screening test is absolutely voluntary.