Prenatal Screening for Down Syndrome

(Source of information provided by the Hospital Authority) (Dec 2019)
This leaflet is intended to help you understand Down syndrome, the prenatal screening tests for Down syndrome offered by ***Hospital Authority (HA)****, and to help you decide whether you want to have a screening test or not.*

What is Down syndrome?

Down syndrome is a genetic condition that typically causes some level of learning disability and certain physical characteristics. Some children with Down syndrome have additional health problems such as heart defects with varying severity. With specialist care and education, some children with Down syndrome can integrate into mainstream schools and lead semi-independent lives.

Down syndrome is caused by the presence of an extra copy of chromosome 21 in a baby's cells. It occurs by chance at conception and there is no evidence that anything done before or during pregnancy causes the syndrome. About 1 in 700 pregnancies will have the chance to carry a baby with Down syndrome and the probability increases with the pregnant woman's age. Antenatal screening for Down syndrome can help identify the condition before birth.

What is the purpose of knowing if my baby has Down syndrome before birth?

This would allow parents to be well-informed and be prepared to discuss with doctors about the options in 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 chance of having a baby with Down syndrome. The test does not harm you or your baby. It provides an estimated chance of your baby having Down syndrome, which is a more accurate estimate than that derived from your age alone.

The Hospital Authority provides 2 tier screening tests.

First tier screening would be done 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 nuchal translucency from 11 to 13 weeks and 6 days of gestation. A blood test will be performed on the same day to measure your Pregnancy Associated Plasma Protein A (PAPP-A) and human chorionic gonadotropin (hCG) levels. The 1st trimester screening test detects up to 90% of Down syndrome pregnancies. Additionally, abnormally high nuchal translucency is known to be associated with other chromosomal and congenital birth defects.

Second trimester screening -

If you are pregnant for more than 14 weeks and less than 20 weeks, you will undergo the blood test from 16 to 19 weeks and 6 days for alpha-fetoprotein (AFP), human chorionic gonadotropin (hCG), estriol (E3) and inhibin-A. This 2nd trimester screening test detects around 80% of Down syndrome pregnancies.

About 5% of women will get a positive screening result, meaning the chance of having a baby with Down syndrome is high, while about 95% will get a negative result, meaning the chance of having a baby with Down syndrome is low.

The screening test result will be available within 2 weeks after the blood test. If your test result is negative, then your report will be filed in your medical notes until your next antenatal visit. If the test result is positive, you will be informed and contacted to make an appointment for further counseling for the second tier screening or diagnostic testing.

Second tier screening is “Non-invasive Prenatal Test” which is a blood test based on the identification and counting of DNA fragments in maternal plasma originating from the placenta. A “High Risk” or “non-reportable” test result would be counselled for diagnostic testing for confirmation study.

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 any other chromosomal abnormalities. 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 from 11 to 14 weeks and amniocentesis is usually performed between 16 to 20 weeks. There would be a slight increase in miscarriage risk after either procedure compared to 0.8% of pregnancies that did not have the procedures during the same gestational period.

What will happen if the foetus is confirmed to have a genetic condition?

The doctor will explain to you and your partner about the nature of the genetic condition, 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 organizations and be better prepared for the birth of your baby. With specialist care and education as well as the support from the community services, children with Down syndrome can live semi-independently. If the pregnancy is not more than 24 weeks of duration, you may also seek advice from your doctor about the possibility of termination of pregnancy under the laws in Hong Kong.

Does a negative screening test result guarantee a baby without a genetic condition?

No. A negative screening test result indicates that the chance of your baby having Down syndrome is low, but it does not guarantee a baby without a genetic condition. A foetal morphology scan is still advised as a screen negative result does not guarantee the absence of foetal or other pregnancy related problems which occur in ≈2% of pregnancies. The availability of this foetal morphology scan is however limited in HA so you may need to approach private doctors or hospitals offering this service.

Can I ask for a diagnostic test for Down syndrome if the screening test result is negative or if I prefer not to be screened first?

In HA, a diagnostic test e.g. chorionic villus sampling or amniocentesis will only be offered to women with positive Down syndrome screening test results. Women with negative first tier screening test result or who prefer direct diagnostic test should approach private doctors or hospitals.

Points to note

- Down syndrome screening is voluntary.
- Down syndrome screening is only available in first trimester for multiple pregnancies.
- You are advised to join one screening program only.
- A positive Down syndrome screening test report does not indicate that your foetus has Down syndrome. A negative screening test report cannot guarantee that your foetus is completely free of Down syndrome or other genetic conditions.