產前唐氏綜合症篩查

絕大多數胎兒是正常的，大約2%的新生兒出生後發現有先天缺陷。新生兒中，大約700人之中有一名會有唐氏綜合症。大部份的唐氏綜合症是偶然發生，極少是由父母遺傳所致。唐氏綜合症是其中一種可以在產前檢測到的先天缺陷。本單張會幫助你了解唐氏綜合症及醫院管理局所提供的唐氏綜合症產前篩查方法，並幫助你決定是否參加測試。

甚麼是唐氏綜合症？

大部份人的體內細胞是有23對染色體的，唐氏綜合症是由於在第21號染色體多一條所致。唐氏綜合症的兒童有智力發育障礙和獨特的外觀，亦可能會有結構異常，如先天性心臟病。

胎兒出生前知道是否有唐氏綜合症有甚麼好處？

在產前知道胎兒是否有唐氏綜合症，可以幫助父母和醫護人員從家庭的角度進行充分的討論。

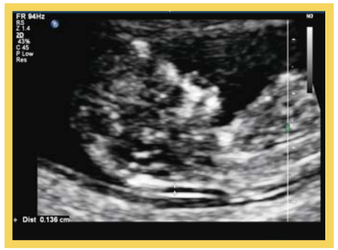
我能否在生產前知道胎兒是否有唐氏綜合症？

可以。 合理的方法是進行一個篩查測試去計算胎兒有唐氏綜合症的風險(或機會)。這項測試不會對你和你的胎兒造成傷害，它會告知你胎兒有唐氏綜合症的預測風險度。這比單獨使用年齡計算出的預測風險度更為準確。

醫院管理局現根據孕期之長短提供兩種篩查測試：

◆ 早孕期篩查

如果你懷孕小過十四週， 可在懷孕十一至十三週六日時進行超聲波檢查，量度胎兒週數及頸皮厚度，並同時抽血驗妊娠相關血漿蛋白A (PAPP-A) 和絨毛膜促性腺激素 (hCG) 水平。唐氏綜合症早孕期篩查的檢出率約80-90%。



◆ 中孕期篩查

如果你已經懷孕超過十四週， 可在十六至十九週六日時抽血驗甲胎蛋白 (AFP) 和絨毛膜促性腺激素 (hCG) 水平。唐氏綜合症中孕期篩查的檢出率約60-70%。

我如何得知唐氏綜合症篩查結果是陽性或陰性？

報告在檢驗後兩週內有結果。上述測試中，約有5%的孕婦會呈陽性報告，表示胎兒有唐氏綜合症的機會較高。若你的檢驗結果呈陽性，醫院職員會致電閣下並安排回院作產前諮詢。若你的檢驗結果為陰性，你會於下一次產前檢查時才閱讀該報告。

如果篩查測試結果呈陽性，我如何在產前證實嬰兒是否有唐氏綜合症？

你可以接受診斷性測試，如絨毛膜檢查或羊膜穿刺。診斷測試能準確驗出胎兒是否有唐氏綜合症。這需要在超聲波引導下，將一支幼針放入子宮腔內抽取胎盤絨毛組織或羊水作染色體檢測。抽取絨毛通常在十三週前進行，羊膜穿刺通常在十六至二十週內進行，抽絨毛或抽羊水測試後流產機會率約0.5 — 1%。倘若你憂慮測試後流產的風險，可以考慮非入侵性胎兒基因檢驗，其胎兒唐氏綜合症檢測率比早孕期和中孕期篩查為高。目前有部分私家醫生和醫院提供此項檢驗。

如果證實胎兒有唐氏綜合症該怎麼辦？

醫生將會與你和你的配偶解釋染色體異常的性質，它對胎兒的影響和將來妊娠的風險。你可以和醫生討論有關醫院和其他機構可以給你的支援，為孩子的出生作更好的準備。經特別照顧及教導的唐氏綜合症兒童亦可過半獨立的生活。

假若你在審慎考慮後決定終止懷孕，你也可與醫生討論是否在24週前進行合法人工流產。

篩查測試結果呈陰性，可以保證胎兒正常嗎？

不可以。篩查測試結果呈陰性，只表示胎兒有唐氏綜合症的機會較低，並不能保證胎兒是正常的。

我可否直接選擇診斷性測試而不先進行篩查測試？

不可以。醫院管理局只提供診斷性測試如絨毛膜檢查或羊膜穿刺予唐氏篩查呈陽性報告之孕婦。篩查後呈陰性報告而要求作診斷性測試的孕婦，及要求直接進行診斷性測試之孕婦，可自行到醫院管理局外之機構作安排。

請注意

- 這個篩查測試是自願性質的。
- 多胎妊娠的唐氏綜合症篩查，祇在早孕期進行。
- 你應只選擇一種篩查測試方法。如果你選擇多重篩查方法，報告結果將會令你困惑，並增加不必要的焦慮。
- 陽性篩查報告並不代表胎兒有唐氏綜合症。
- 唐氏綜合症篩查測試結果呈陰性並不保證胎兒一定沒有唐氏綜合症或一定是完全正常的。

如有任何查詢，請聯絡醫護人員

以上資料由瑪麗醫院婦產科及贊育醫院提供。

瑪麗醫院
Queen Mary Hospital

贊育醫院
Tsan Yuk Hospital

產前唐氏綜合症篩查
PRENATAL DOWN SYNDROME SCREENING

瑪麗醫院 Queen Mary Hospital	Pamphlet Topic & Department Code
Ownership	QMH/Obstetrics & Gynaecology-25(OBS.17)
Last review/revision	Feb/2016
Approval	HKWC Information Pamphlet Working Group
Distribution	As requested

PRENATAL DOWN SYNDROME SCREENING

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 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?

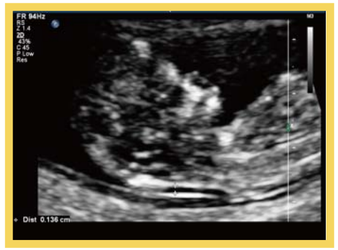
This would allow parents and doctors to discuss options to the best interest of the family.

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 80-90% of fetuses with Down syndrome.



Second trimester screening

IIf you are pregnant for more than 14 weeks, you will undergo the blood test at 16 to 19 weeks and 6 days for alpha-fetoprotein (AFP) and human chorionic gonadotrophin (hCG). The 2nd trimester screening detects around 60-70% of fetuses with Down syndrome.

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

The screening test result will be available within 2 weeks 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 0.5-1%.
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.

Please approach our nursing staff for details

Information provided by Department of Obstetrics & Gynaecology, Queen Mary Hospital and Tsan Yuk Hospital.