早孕期一站式
唐氏綜合症篩查

First Trimester One Stop Clinic for Assessment of Risk for Down's Syndrome

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唐氏綜合症是甚麼？

唐氏綜合症是最常見的遺傳病之一。患者的細胞比正常人多出一條第21條染色體，導致先天性弱智，外貌亦與常人有明顯分別；其他器官結構也可能出現缺陷，例如先天性心臟病、腸塞等。唐氏綜合症患者一般可活到成年。經特別訓練，患者或可担任簡單工作；但由於智能的障礙，他們大都需要長期接受照顧。

What is Down's syndrome ?

Down's syndrome is one of the commonest genetic disorders, caused by the presence of an extra chromosome 21 in each cell. Affected individuals are characterized by distinctive facial features and mental retardation. There may also be other structural defects, such as congenital heart diseases and defects in the bowel. Individuals with Down's syndrome usually can live to adulthood. Some may perform simple work with training but long-term care is usually required.

孕婦年齡與嬰兒患有唐氏綜合症的機會率之關係

胎兒患有唐氏綜合症並非只發生於年逾35歲的「高齡產婦」身上，所有孕婦都有機會懷有唐氏綜合症胎兒；不過，孕婦年齡越大，機會就越高。

How related is the maternal age to the risks of having babies affected by Down's syndrome?

Down's babies are not confined to women who are older than 35 years of age. Rather, a woman of any age does have a chance of having an affected baby. However, the older the woman, the higher the probability her baby is affected.

孕婦在預產期之年齡 Maternal age at childbirth	嬰兒患有唐氏綜合症的機會 Chance of baby with Down's syndrome
20	1 in 1500
25	1 in 1300
30	1 in 900
35	1 in 350
40	1 in 100
45	1 in 25

唐氏綜合症的產前診斷

要準確診斷胎兒是否患有唐氏綜合症，目前的做法是從胎盤「絨毛球活檢」或胎水「羊膜穿刺術」檢查胎兒細胞，數算其染色體的數目。要詳細清楚兩種手術的情況，可參閱有關小冊子。

這兩種方法一般對孕婦及胎兒都很安全，但仍屬於入侵性檢查，就算由資深醫生進行，亦存有少量風險，例如增加胎兒流產機會約0.5-1%。故若胎兒患有唐氏綜合症的機會不高，一般不建議孕婦接受這些入侵性檢查。

傳統上，只為三十五歲或以上之高齡孕婦進行上述檢查。但這項措施只能在產前發現所有唐氏綜合症病例的30%，原因是年紀較輕產婦的胎兒亦有機會患有唐氏綜合症。因此，醫學界一直研究更準確的方法來評估胎兒患有唐氏綜合症的機會。

Antenatal diagnosis of Down's syndrome

Accurate diagnosis of Down's syndrome can be made in the antenatal period by sampling either the placental tissue (chorionic villous sampling) or the amniotic fluid (amniocentesis) to count the number of chromosomes. For details of each procedure, please refer to the pamphlet on these procedures.

Both procedures are very safe in general, although they carry a small miscarriage rate of 0.5-1% even under experienced hands. As a result, they are usually offered only to women with increased risks of having babies with chromosomal anomalies.

Traditionally, only women aged 35 or above are offered the option of these procedures. However, this policy of screening only "older women" can just pick up 30% of all Down's babies since younger women may also carry Down's babies. The medical profession has tried to find ways to identify more accurately women who have high risks for giving birth to a Down's baby.

「早孕期一站式唐氏綜合症篩查」是什麼？

「早孕期一站式唐氏綜合症篩查」在懷孕十一週至十四週之前進行，以下列因素計算胎兒患有唐氏綜合症的風險：

1) 孕婦年齡
2) 孕婦血清中之妊娠相關血漿蛋白A (PAPP-A)水平
3) 孕婦血清中之游離人絨毛膜促性腺激素(free-beta hCG)水平
4) 胎兒頸皮下透明層的厚度

What is OSCAR ?

One Stop Clinic for Assessment of Risk for Down's syndrome (OSCAR) is a screening method performed at 11-14 weeks of gestation. The following parameters are obtained to calculate the risks of having a Down's baby for each individual woman:

1) the woman's age
2) her serum level of pregnancy-associated plasma protein-A (PAPP-A)
3) her serum level of free beta human chorionic gonadotrophin (free-beta hCG)
4) thickness of the baby's neck-fold (nuchal translucency thickness)

進行篩查的過程是怎樣？

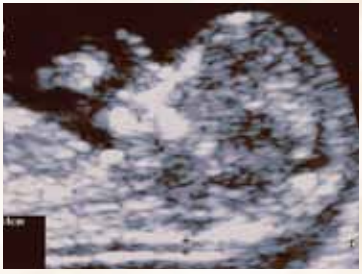
這篩查過程如下：

1) 孕婦接受超音波掃描檢查，確定胎兒的大小是否適合，並且準確量度胎兒後頸皮下透明層的厚度「頸皮厚度」(見圖一)；
2) 孕婦須作抽血檢查，以化驗PAPP-A及free-beta hCG 的水平；
3) 綜合上述數據，由電腦計算胎兒患有唐氏綜合症的機會率。

What procedures are involved in this test ?

The test involves:

1) An ultrasound examination to measure the size of the fetus and the nuchal translucency thickness (Figure 1);
2) Blood taking from the pregnant woman for assessment of the serum hormonal levels;
3) Calculation of the risk via a computer software based on the parameters



圖一
figure 1
以超聲波掃描量度胎兒頸皮厚度
Fetal nuchal translucency thickness measurement by ultrasound

如檢驗結果為高危，這代表什麼呢？

如檢驗結果為高危 (陽性)，**並不表示胎兒一定患有唐氏綜合症**，但須要考慮作進一步檢查胎兒染色體，以確定胎兒是否出現問題。作診斷的方法，包括絨毛球活檢（在孕期十一至十四週進行）或羊膜穿刺術（在孕期十六至二十週進行)。

What does it mean if the test result shows "increased risk" ?

An "increased risk" or "positive" result **does not mean that the baby is necessarily affected by Down's syndrome**. It merely indicates that further confirmatory tests need to be considered. The options include chorionic villous sampling (to be performed between 11-14 weeks of gestation) or amniocentesis (to be performed at 16-20 weeks of gestation). These tests help to confirm or refute the diagnosis of Down's syndrome.

如檢驗結果為低危，又代表什麼呢？

如檢驗結果為低危 (陰性)，表示胎兒患上唐氏綜合症的機會低。若孕婦接受此低風險，便無須冒險接受絨毛球活檢或羊膜穿刺術。然而，這檢驗是一種篩查方法而並非斷症，「低風險」並不能完全排除胎兒患有唐氏綜合症或其他染色體異常的機會。

What does it mean if the test result shows 'low risk' ?

A "low risk" or "negative" result means the chance of a Down's baby is low. If the pregnant woman accepts such low risk, there is no need for invasive prenatal diagnosis procedure, such as chorionic villous sampling amniocentesis. However, it is also important to point out that OSCAR remains a screening test. A result showing "low risk" cannot completely exclude the possibility of having a baby with Down's syndrome or other chromosomal anomalies.

「早孕期一站式唐氏綜合症篩查」的優點

1) 能發現九成患有唐氏綜合症的胎兒，為眾多篩查方法中至為有效；
2) 超聲波檢查對胎兒沒有危險；
3) 適合任何年齡孕婦；
4) 由於檢驗時週期較早，如屬高危，有足夠時間考慮是否作進一步測試；
5) 如真是不幸患上唐氏綜合症，亦有足夠時間考慮會否選擇人工流產；
6) 若選擇人工流產，於早孕期進行手術的危險性也相對較低。

Advantages of OSCAR

1) High detection rate (90%) among all existing screening programmes for Down's syndrome;
2) Ultrasound examination is safe to the baby;
3) Suitable for women of any age;
4) It can be performed in early gestation and hence plenty of time to make decision should the results be positive;
5) If the baby is confirmed to be affected, there is enough time to consider all management options;
6) If a termination of pregnancy is chosen for an abnormal baby, it carries less risks to the women when performed in early gestation.

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