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早孕期多項目綜合性
唐氏綜合症篩查
Combined First Trimester Screening
of Fetal Down’s Syndrome

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甚麼是唐氏綜合症？

唐氏綜合症，俗稱蒙古症。患者都有相同的外貌特徵，以及不同程度的先天性智能低下。腦部的其他器官，例如視覺、聽覺等，也可能伴有其他的缺陷。視頻長度的疾病程度不同，患者或可從事簡單的工作，但一般都需要家人的長期照顧。唐氏綜合症的成因是他們的細胞中的染色體多了一條（第二十一條）。孕婦懷有唐氏綜合症胎兒的機率會隨著年齡遞增。

<table>
<thead>
<tr>
<th>妊娠年齡</th>
<th>胎兒患唐氏綜合症的機會率 (出生時)</th>
</tr>
</thead>
<tbody>
<tr>
<td>20</td>
<td>1 / 1530</td>
</tr>
<tr>
<td>25</td>
<td>1 / 1350</td>
</tr>
<tr>
<td>30</td>
<td>1 / 900</td>
</tr>
<tr>
<td>32</td>
<td>1 / 660</td>
</tr>
<tr>
<td>34</td>
<td>1 / 450</td>
</tr>
<tr>
<td>高齡孕婦</td>
<td></td>
</tr>
<tr>
<td>35</td>
<td>1 / 360</td>
</tr>
<tr>
<td>36</td>
<td>1 / 280</td>
</tr>
<tr>
<td>38</td>
<td>1 / 170</td>
</tr>
<tr>
<td>40</td>
<td>1 / 100</td>
</tr>
<tr>
<td>42</td>
<td>1 / 55</td>
</tr>
<tr>
<td>44</td>
<td>1 / 30</td>
</tr>
</tbody>
</table>

現時最常用的篩查方法的比較：

<table>
<thead>
<tr>
<th>篩查方法</th>
<th>妊娠年齡</th>
<th>產婦年齡</th>
<th>每診斷一個唐氏綜合症的胎兒需要進行的羊膜穿刺術或絨毛球活檢的數目</th>
</tr>
</thead>
<tbody>
<tr>
<td>只用產婦年齡</td>
<td>30%</td>
<td>200</td>
<td>31</td>
</tr>
<tr>
<td>中孕期血清檢查</td>
<td>60-70%</td>
<td>41</td>
<td></td>
</tr>
<tr>
<td>早期孕期胎兒頭皮下的厚度</td>
<td>60-80%</td>
<td>41</td>
<td></td>
</tr>
<tr>
<td>早期孕期血清檢查</td>
<td>60%</td>
<td>41</td>
<td></td>
</tr>
<tr>
<td>早期孕期多項目綜合症篩查</td>
<td>85-90%</td>
<td>31</td>
<td></td>
</tr>
</tbody>
</table>

任何篩查方法都需將無助的低危及高危兩類。上表中第二欄的數字代表所有受試者為高危的產婦接受羊膜穿刺術或血液檢查即可篩出所有唐氏綜合症的百分比。此數字愈高，表示篩查方法愈好，因為大部份受試者篩出的唐氏綜合症的胎兒仍然是正常的，因此我們需要進行一項顯示的羊膜穿刺術或血液檢查，才會診斷出一個真的唐氏綜合症的胎兒，而此數字已列於上表的第三欄。此數字愈低，表示該篩查方法愈好。

甚麼是早孕期多項目綜合性唐氏綜合症篩查？

早孕期多項目綜合性唐氏綜合症篩查包括以下二項檢驗，並須在懷孕十一週起但未足十四週前進行：

1. 利用高解像超聲波儀器，量度胎兒頭皮下的厚度。此後頭皮下的厚度愈為頭皮下的面積，在所有的胎兒都可以看到。患有唐氏綜合症的胎兒，其頭皮下的厚度通常較為厚。
2. 我們會抽取孕婦血液的樣本，檢驗其中的甲型懷孕血漿蛋白 (PAPP-A) 和游離性乙型人類絨毛膜性腺 (free β-hCG)。

根據孕婦的年齡、懷孕的週數、胎兒後頭皮下的厚度及血液測試的結果，醫生就能計算出胎兒患有唐氏綜合症的機率。大部份測試的結果都會是陰性 (低危)，也就是說胎兒患有唐氏綜合症的機會非常低。大概有百分之五至六的測試結果會是陽性 (高危)，其中已包含百分之八十至九十的唐氏綜合症的胎兒。

陽性結果或「高危」是甚麼意思？

如果檢驗結果屬高危，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，並可選擇羊膜穿刺術或血液檢查確定。
Prenatal Diagnosis of Fetal Down’s Syndrome

The only way to ascertain whether a fetus has Down’s syndrome or not is by doing an invasive test, either an amniocentesis or a chorionic villus sampling (CVS). Both tests involve the passage of a needle into the womb under ultrasound guidance. A CVS is performed between 11 and 14 weeks of gestation to obtain a sample of the placenta, while amniocentesis is usually performed after 16 weeks to obtain a sample of fluid around the baby. However, these invasive tests cause miscarriage in about 1% of procedures. Therefore, these invasive tests should only be performed if the risk of fetal Down’s syndrome is high.

Prenatal Screening of Fetal Down’s Syndrome

The purpose of a screening test is to identify those fetuses which are at high-risk of abnormalities. For a long time, maternal age was the only screening method for fetal Down’s syndrome. This means that pregnant women beyond a certain age (usually 35 or above) would be offered an invasive test. However, it is now clear that this screening test is not precise. Over the last two decades, many screening tests have been developed. The most sensitive method is the combination of first trimester nuchal translucency (NT) with biochemical tests.

The following table is a comparison of commonly used screening tests for fetal Down’s syndrome:

<table>
<thead>
<tr>
<th>Maternal age combined with</th>
<th>Percentage of Down’s fetus identified*</th>
<th>Number of invasive tests need to be performed for every Down’s fetus diagnosed*</th>
</tr>
</thead>
<tbody>
<tr>
<td>Maternal Age Alone</td>
<td>30%</td>
<td>200</td>
</tr>
<tr>
<td>Second Trimester Biochemistry</td>
<td>60-70%</td>
<td>41</td>
</tr>
<tr>
<td>First Trimester NT</td>
<td>60-80%</td>
<td>41</td>
</tr>
<tr>
<td>First Trimester Biochemistry</td>
<td>60%</td>
<td>41</td>
</tr>
<tr>
<td>Combined First Trimester NT and Biochemistry</td>
<td>85-90%</td>
<td>31</td>
</tr>
</tbody>
</table>

* A screening test will categorize a pregnant woman as either at low-risk or high-risk for Down’s syndrome. Figures in the second column refer to the percentage of babies with Down’s syndrome that will be identified if all high-risk women accept an invasive diagnostic test. The higher this figure is, the better the screening test. Since the majority of babies among the high-risk women are still normal, many invasive tests still need to be performed before one Down’s syndrome baby is diagnosed (figures in the last column). The lower this figure is, the better the screening test.
What is Combined First Trimester Screening of Down’s Syndrome?

The following two tests are performed between 11 weeks and 13 weeks and 6 days of gestation:

1. An ultrasound examination to measure the fetal NT, which represents the fluid accumulation behind the fetal neck. All fetuses have some fluid behind the neck, although those with Down’s syndrome tend to have more.

2. A maternal blood sample will be taken to determine the levels of two chemicals produced by the placenta, called free β - hCG (human chorionic gonadotropin) and PAPP-A (pregnancy associated plasma protein-A).

Based on maternal age, gestational age, NT thickness and results of biochemistry, pregnant women will be given a risk factor of how likely their baby is affected by Down’s syndrome, and that risk factor is specific to their pregnancy and baby. The test result will be negative in the majority of women, meaning that the risk of Down’s syndrome is low. However, the test will be positive in about 5-6% of women, which will include 85-90% of the Down’s syndrome.

What if the test result is positive?

An increased risk does not mean that the baby is abnormal. It only means that further invasive tests, such as CVS, need to be considered to confirm whether the baby is normal or has Down’s syndrome.

What does a negative test result mean?

A negative test only indicates that the risk that the baby has Down’s syndrome is low. However, it does not completely eliminate the possibility that the fetus may have Down’s syndrome or other chromosomal abnormalities. This test is NOT a test for intelligence or structural abnormalities.

If you are interested, please note that:

1. The test can only be performed from 11 weeks to less than 14 weeks pregnant.
2. The NT scan and blood test will be performed during the same visit.
3. Measurement of NT could be time-consuming, depending very much on the fetal position.
4. Results are usually available in a few days and participants will be notified by phone, and a written report.

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The Professorial Clinic is jointly run by a professor in Obstetrics and Gynaecology of The Chinese University of Hong Kong and the Head of Department of Obstetrics and Gynaecology of Union Hospital. The clinic aims to provide service for difficult obstetric and gynaecological problems.

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