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

Maternal age combined with	Percentage of Down's fetus identified*	Number of invasive tests need to be performed for every Down's fetus diagnosed*
Maternal Age Alone	30%	200
Second Trimester Biochemistry	60-70%	41
First Trimester NT	60-80%	41
First Trimester Biochemistry	60%	41
Combined First Trimester NT and Biochemistry	85-90%	31

*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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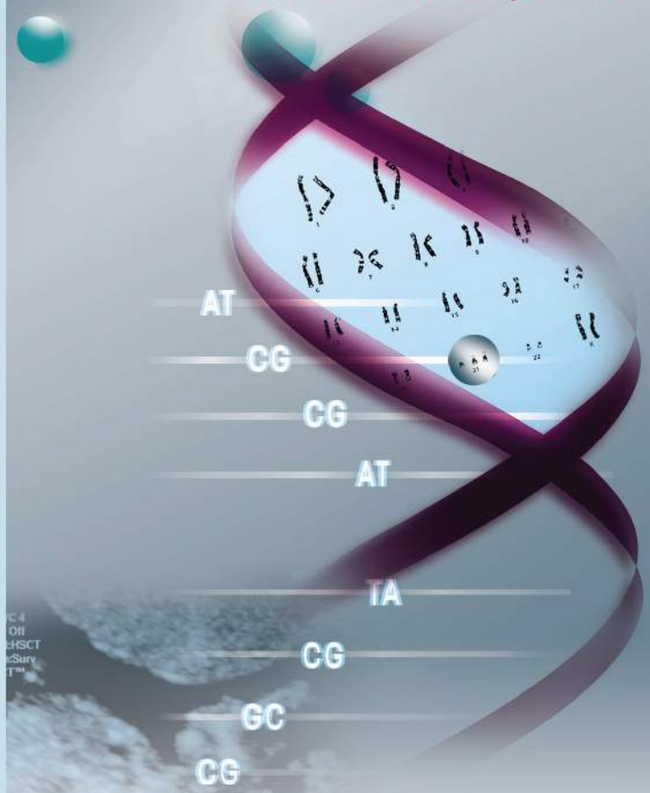
仁安醫院 UNION HOSPITAL

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弱智。身體的其他器官，例如心臟、腸道等，也可能伴有其他的缺陷。視乎弱智的程度不同，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由於他們的細胞中的染色體數目比正常人多了一條（第二十一條）。孕婦懷有唐氏綜合症胎兒的機會率會隨著年齡遞增。

孕婦年齡	胎兒患唐氏綜合症的機會率(出生時)
20	1 / 1530
25	1 / 1350
30	1 / 900
32	1 / 660
34	1 / 450
高齡孕婦	
35	1 / 360
36	1 / 280
38	1 / 170
40	1 / 100
42	1 / 55
44	1 / 30

唐氏綜合症的產前診斷

如孕婦希望在懷孕早期確定胎兒是否患有唐氏綜合症，那便必須接受羊膜穿刺術或絨毛球活檢。前者通常在十六孕週後抽取胎兒的羊水，而後者則在十一至十四孕週時抽取胎盆組織，再進行染色體分析。不論是羊膜穿刺術或絨毛球活檢檢驗，醫生都必須在超聲波儀器導引下把一支細小的針穿入子宮內，因此都有一定的危險性；大概有百分之一的孕婦在進行這些檢查後會引致流產。故此，這些檢驗並不適用於大部份的孕婦；只有當胎兒被推測為有較大的機會患有唐氏綜合症的時候，醫生才會建議孕婦進行這類檢查。

唐氏綜合症的篩查

篩查的目的在於準確地分辨出哪些胎兒有較大的機會患有唐氏綜合症，因而須要進一步接受羊膜穿刺術或絨毛球活檢。過去，醫生只能基於孕婦的年齡作為篩查唐氏綜合症的唯一方法；也就是過去所說的高齡孕婦。但眾多的醫學研究指出，孕婦年齡並非一個準確的唐氏綜合症篩查方式。在過去二十多年，研究人員發現了很多更好的篩查方式，其中最準確的要算是早孕期多項目綜合性篩查。

現時最常用的篩查方法的比較：

孕婦年齡再加上	驗測出唐氏綜合症百分比*	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*
只用孕婦年齡	30%	200
中孕期驗血檢查	60-70%	41
早孕期胎兒後頸皮下的厚度	60-80%	41
早孕期驗血檢查	60%	41
早孕期多項目綜合症篩查	85-90%	31

*任何篩查方法都會把孕婦分為低危及高危兩類。上表中第二欄的數字代表如所有分類為高危的孕婦皆接受羊膜穿刺術或絨毛球活檢時可測出患有唐氏綜合症的百分比。此數字愈高，代表該種篩查方法愈好。由於大部份被分類為高危孕婦的胎兒仍然是正常的，因此我們需要進行一定數目的羊膜穿刺術或絨毛球活檢，才會診斷出一個真的患有唐氏綜合症的胎兒，而此數字已列於上表的第三欄。此數字愈低，代表該種篩查方法愈好。

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，量度胎兒後頸皮下的厚度。此後頸皮下的厚度實為頸皮下積存的水腫，在所有的胎兒都可以看到。患有唐氏綜合症的胎兒，其頸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-hCG）。

根據孕婦的年齡、懷孕的週數、胎兒後頸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會率。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，孕婦可選擇羊膜穿刺術或絨毛球活檢檢驗確定。

陰性結果或「低危」是甚麼意思?

如果檢驗結果屬低危，表示胎兒患上唐氏綜合症的機會率相比於一般35歲的孕婦為低，但不能代表胎兒一定沒有唐氏綜合症，或其他的染色體異常。此篩查檢驗並不能預測胎兒的智力，亦不能用於診斷器官發育異常。

注意事項！

1. 早孕期多項目綜合性唐氏綜合症篩查必須在懷孕十一週起但未足十四週前進行。
2. 醫生會在進行超聲波檢驗後抽取血液樣本。
3. 量度胎兒後頸皮下的厚度所需的時間取決於多方面的因素，特別是胎兒的位置。請預留充裕的時間作檢查。
4. 醫護人員會在數天內以電話及郵遞的方式通知孕婦測試的結果。

What is Down's Syndrome?

Down's syndrome is characterized by distinctive facial features and a varying degree of mental retardation. Other structural defects such as congenital heart disease and gastrointestinal defects are common. Some individuals with Down's syndrome may be capable of performing simple work but long-term care is usually required. Down's syndrome is caused by the presence of an extra chromosome 21 in each cell. The chance of having a baby with Down's syndrome increases with maternal age.

Maternal Age (yrs)	Chance of Down's Syndrome (at birth)
20	1 in 1530
25	1 in 1350
30	1 in 900
32	1 in 660
34	1 in 450
Advanced Maternal Age	
35	1 in 360
36	1 in 280
38	1 in 170
40	1 in 100
42	1 in 55
44	1 in 30

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.