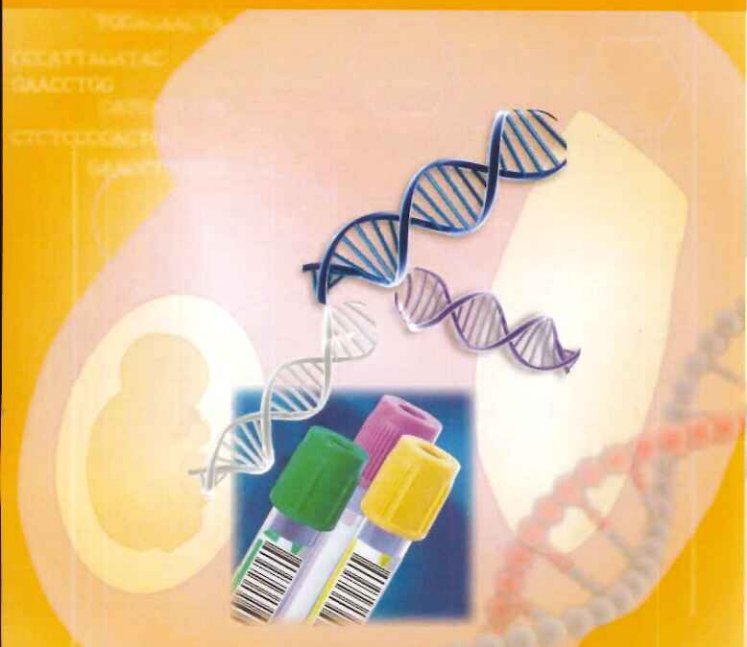




## 無創性胎兒染色體篩查 (NIPT)



本冊子內容並非包含所有有關NIPT的資料，您必須經過醫生詳細的諮詢後才可進行NIPT。本學系保留一切更改或刪除單張內容之權利而不作另行通知。

### 唐氏綜合症

唐氏綜合症是由於人體細胞內第21對染色體多出一條所致，它的發病率為1/800，是最常見的染色體異常及遺傳性智力障礙疾病。早孕期唐氏綜合症篩查（超聲波量度胎兒頸皮厚度+孕婦血液生化檢查）可以檢出約90%的唐氏綜合症，但每100個正常胎兒中將會有5個被列為高風險。現時有一種更準確的篩查方法已供臨床應用，就是「無創性胎兒染色體篩查」(NIPT)。

## 什麼是 NIPT?

### 篩查方法

每個正常人體細胞內都有23對由DNA組成的染色體。母體的血液中，除了自己的DNA外，還含有小量的胎兒DNA，NIPT就是藉這些胎兒DNA來作唐氏綜合症篩查。如果胎兒患有唐氏綜合症，化驗結果就會顯示第21對染色體DNA的數量比正常增加。

**NIPT可以檢出高達99%的唐氏綜合症，而每1000個正常胎兒中只有約1個會被誤列為高風險。**除此之外，它還可以檢出愛德華氏綜合症（第18條染色體多出一條）、巴陶氏綜合症（第13對染色體多出一條）及特納氏綜合症（只有一條性染色體[X]）。

現時醫院管理局並未有提供此項篩查服務，閣下可於香港中文大學胎兒醫學組私家症診所或其他私家診所或醫院自費進行此項篩查。

### 篩查孕週

胎兒孕週滿10週便可進行篩查

### 報告發出時間

抽血後大約7個工作天便可發出篩查報告

### 篩查費用(包括諮詢、超聲波檢查及抽血)

港幣\$5,700起

(包括染色體21「唐氏」18, 13 XY + 7種染色體微缺失症侯群)

## 其他NIPT相關資料

### 超聲波確定孕週

針對各項唐氏綜合症篩查，準確的孕週計算是十分重要的，所以建議孕婦先進行早孕期超聲波檢查確定胎兒的孕週。香港中文大學胎兒醫學組私家症診所亦可提供此項服務，如胎兒孕週滿10週，更可即時進行NIPT篩查。

### 預約方法

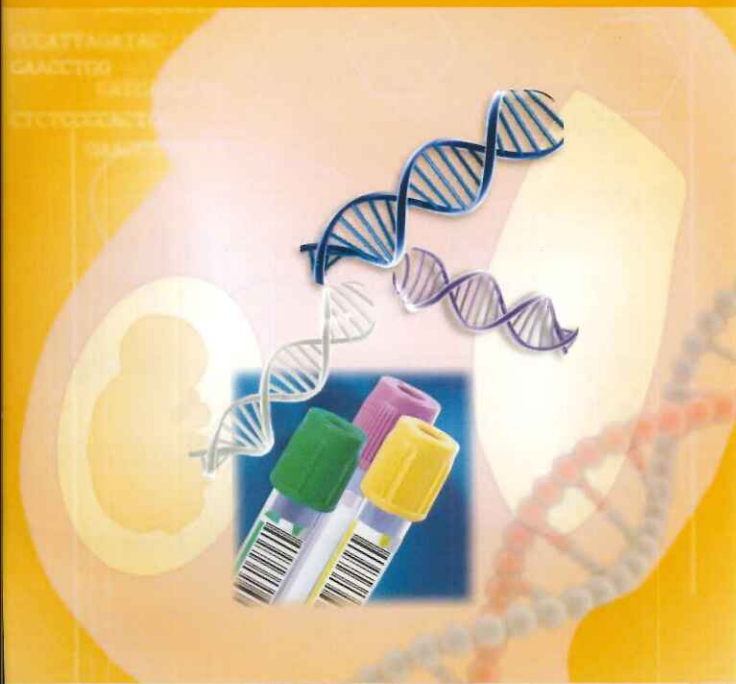
- **親臨** 威爾斯親王醫院李嘉誠專科門診南翼二樓三十號房香港中文大學胎兒醫學組辦公室
- **致電** 威爾斯親王醫院香港中文大學胎兒醫學組直線 56001970 或電話留言 35054219 (請留下姓名、電話及有興趣參加的服務)
- 填妥以下表格，**傳真**至香港中文大學胎兒醫學組 26325065
- 瀏覽以下有關此服務的網頁及在**網上預約**  
<https://www.fetalmedicine.hk/common/oscarbooking.asp>

### 預約表格 Registration Form (in block letters)

姓名 Name	
最後經期第一天 Last Menstrual Period	
預產期 Estimated Date of Confinement	
聯絡電話 Telephone Number	
電郵 Email	
轉介醫生 Referring Doctor	
請選擇 Please choose: <input type="checkbox"/> 單胎 Singleton / <input type="checkbox"/> 雙胎 Twins	



## Non-invasive Prenatal Test (NIPT) for chromosomal abnormality



### Down syndrome screening

Down syndrome is caused by an extra copy of Chromosome 21 in the human cells. It is the most common chromosomal abnormality and the most common genetic cause of mental retardation. The local incidence is around 1 in 800. The first-trimester combined Down syndrome screening test (fetal nuchal translucency NT measurement plus maternal biochemical blood test) detects 90 out of 100 Down syndrome, but around 5 out of 100 normal babies would be reported as high risk. **A more accurate screening test called NIPT (non-invasive prenatal test) is now available.**

### What is NIPT?

#### The test

People normally have 23 pairs of chromosomes in each cell. Each chromosome is made up of a coil of DNA. Besides her own DNA, mother's blood also contain small amount of fetal DNA. Therefore, it allows accurate screening for fetal Down syndrome using mother's blood. If the baby has Down syndrome, the test will show increased amount of DNA from chromosome 21.

**NIPT detects around 99 out of 100 Down syndrome, and only around 1 out of 1000 normal babies would be reported as high risk.**

It also detects Edward syndrome (an extra copy of Chromosome 18), Patau syndrome (an extra copy of Chromosome 13) and Turner syndrome (only 1 copy of Sex Chromosome [X]).

NIPT is not currently available in Hospital Authority, but is **available in The Chinese University of Hong Kong Fetal Medicine Private Clinic as well as other private clinics and hospitals in Hong Kong.**

#### Gestational age for NIPT

From 10 gestational weeks onwards

#### Reporting time

Around 7 working days after blood taking

#### Charge (consultation, ultrasound and blood test)

HKD\$5,700 up

- (Chromosome 21 (Down syndrome) 13, 18, XY + 7 microdeletion syndromes)

### Other information about NIPT

#### Dating Scan

Pregnant women are recommended to have a dating scan to determine the exact gestational age which is important for all forms of Down syndrome screening. **This service is also available in The Chinese University of Hong Kong Fetal Medicine Private Clinic. NIPT can be done at the same time if the gestational age is beyond 10 weeks.**

#### How to make an appointment

##### - In person :

The Chinese University of Hong Kong Fetal Medicine Private Clinic, Room 30, 2/F, Li Ka Shing Specialist Out-patient Clinic South Wing, Prince of Wales Hospital

##### - By phone :

Call The Chinese University of Hong Kong Fetal Medicine Private Clinic direct line at 5600-1970 or leave a message at 3505-4219 (Please leave your name, telephone number)

##### - By fax :

Complete the form on the other side of the pamphlet and fax it to The Chinese University of Hong Kong Fetal Medicine Private Clinic at 2632-5065

##### - Online :

Visit the following web pages and book online <https://www.fetalmedicine.hk/common/oscarbooking.asp>

The content of this pamphlet does not include all the information about NIPT. Doctor's counselling is essential before you undergo NIPT. Our department reserves the right to amend any information in this pamphlet without prior notice.