



香港中文大學 婦產科學系



Department of Obstetrics and Gynaecology
The Chinese University of Hong Kong

FetalSeq

version 1.0

胎兒測序

版本 1.0



什麼是FetalSeq v1.0 胎兒測序？

FetalSeq v1.0 胎兒測序是基於二代測序平台的最新檢測胎兒基因缺失、重複的方法。此檢測與傳統胎兒DNA芯片(Fetal DNA Chip)相比，更能準確及全面地檢測出致病性變異。

Enquiries 查詢電郵及電話: obsgyn@cuhk.edu.hk

(852) 3505 1557 (general enquiries 一般查詢)

(852) 3505 4416 (appointments 預約診症)

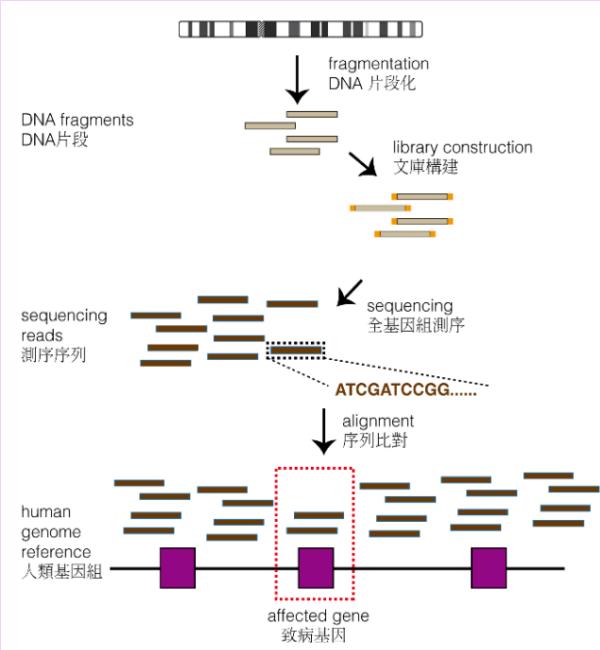
For more details, please refer to your clinician or visit our website 如需獲取更多資料，可向您的專科醫生查詢，或請瀏覽我們的網頁：

http://www.obg.cuhk.edu.hk/_services/laboratory_service/ngs/

檢測流程



圖一 (Figure 1)



誰有需要做此項檢測？

1. 曾出現流產或死胎的夫婦；
2. 所有有需要進行侵入性產前檢測的孕婦，如因為胎兒超聲結構異常、唐氏綜合症篩查或胎兒無創DNA檢測屬高風險、高齡妊娠等；
3. 患有發育和智力遲緩，自閉症譜系障礙或先天性多發性畸形的兒童。

選擇此項技術的好處

FetalSeq v1.0 胎兒測序相比 Fetal DNA Chip :

1. **更廣泛與均勻**：全基因組範圍內檢測 50kb 以上的變異，包含部份 Fetal DNA Chip 無探針覆蓋區域的疾病相關變異信息；
2. **更準確及細緻**：檢測更微細的基因組缺失/重複變異，更準確地檢測變異範圍，有助於判斷是否包括致病基因；
3. **需要更少DNA樣本量**

*Dong et al. *Genet Med*. 2016 Sep;18(9):940-8

*Dong et al. *Curr Protoc Hum Genet*. 2017 Jul 11;94:8.17.1-8.17.16.

病例分享 (微缺失/微重複個案)

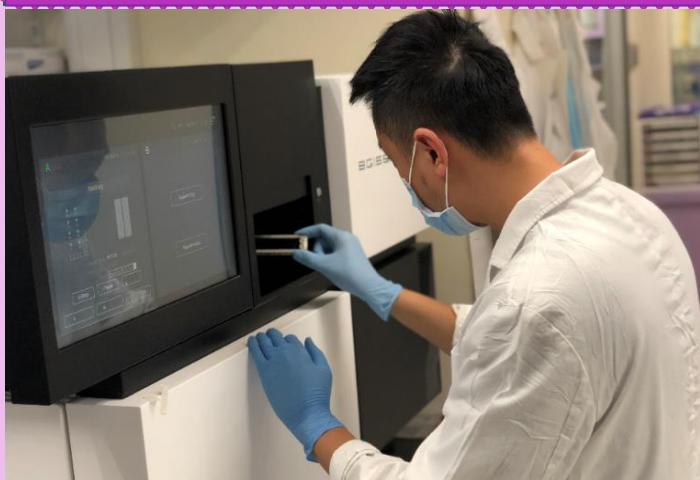
圖二 (背面) 的外圈展示了 FetalSeq v1.0 胎兒測序能夠一次性檢測全基因組範圍 (23對染色體) 的微缺失 (紅色柱指示位置) 或微重複 (藍色柱指示位置)；而內圈為6號染色體的放大展示圖，證明 FetalSeq v1.0 胎兒測序可同時檢測 germline (存在於全部細胞) 和 mosaic (嵌合型；僅存在於部分細胞，綠色箭頭指示位置) 的變異。

選擇此服務需要注意的事項

1. 目前此檢測方法可檢測染色體數目異常、基因組微缺失、微重複變異；
2. 限制：不可檢測類型為染色體平衡易位、倒位、單親二倍體、低比例嵌合體及單核苷酸突變；
3. 即使報告結果正常，胎兒或患者亦有機會患有其他此技術沒有覆蓋的基因變異。



FetalSeq version 1.0



What is FetalSeq v1.0?

FetalSeq v1.0 utilizes an innovative next-generation sequencing platform for a more comprehensive, precise assessment of pathogenic copy-number variants detection, compared to Fetal DNA Chip.

How is the test carried out?

Pre-test Counseling

DNA sampling
(e.g. chorionic villus sampling or amniocentesis)

FetalSeq v1.0 testing
[refer to Figure 1 (back)]

Report (within 10 working days)

Advantages of FetalSeq v1.0

Compared with Fetal DNA Chip, FetalSeq v1.0 can detect deletion(s) and duplication(s):

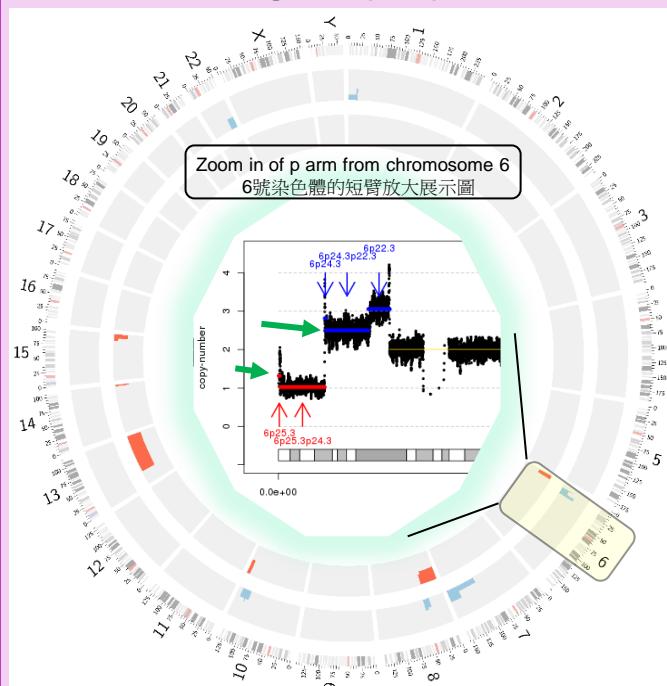
1. **More comprehensively:** on a genome-wide scale at a higher resolution (>50kb) with evenly distributed sequencing reads, including some cryptic disease-related regions not covered by Fetal DNA Chip;
2. **More precisely:** in a non-targeted manner, which reduces bias and improves detection of precise boundaries of deletions/duplications;
3. **Requiring less DNA input.**

*Dong et al. *Genet Med.* 2016 Sep;18(9):940-8

*Dong et al. *Curr Protoc Hum Genet.* 2017 Jul 11;94:8.17.1-8.17.16.

Case example (Case of microdeletions/microduplications)

Figure 2 (圖二)



Case example (Cont.)

(Case of microdeletions/microduplications)

Outer circles of **Figure 2** illustrate FetalSeq v1.0 in detecting deletions (indicated by red bars) and duplications (blue bars) in genome-wide scale (23 pairs of chromosomes). Inner circle is the zoom-in figure of chromosome 6, demonstrating the feasibility of detecting germline and mosaic deletions/duplications (green arrows).

Who will benefit from this test?

1. Couples with miscarriage or stillbirth;
2. Women who want invasive prenatal testing due to fetal anomalies in ultrasound examination, high risk from Down's Syndrome Screening/non-invasive prenatal testing or advanced maternal age;
3. Children with developmental delay, intellectual disabilities, autism spectrum disorders or multiple congenital anomalies.

Important notes for this test

1. This test detects chromosomal aneuploidies, microdeletions and microduplications;
2. Limitation: It is unable to detect balanced translocations, inversions, uniparental disomies, low-level mosaicism and single-nucleotide variants;
3. Normal test results do not exclude other genetic causes that are undetectable by FetalSeq v1.0.