什麼是胎兒DNA芯片 V.2?

胎兒DNA芯片是香港中文大學婦產科學系設計，於2009年率先在香港推出的產前基因檢測服務，專門用作診斷**基因序列缺失/重複**，比傳統染色體核型分析檢測範圍**更廣泛**、**更細緻**及**更快捷**，可檢測傳統核型分析不能診斷的遺傳性疾病。現推出Version 2芯片，更可診斷**單親同二體**引起的疾病。

有需要進行胎兒DNA檢測的孕婦包括：

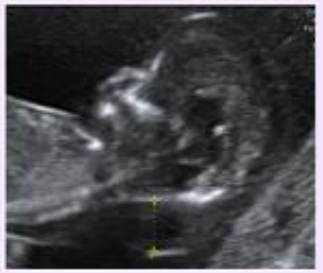
1. 超聲波檢查顯示胎兒有異常徵象
2. 唐氏綜合症篩查屬高危
3. 曾誕下先天畸形的嬰兒
4. 夫婦任何一方攜帶異常染色體
5. 不明原因死胎
6. 產前篩查屬高風險，需要做侵入性產前檢測的孕婦

檢測流程：

選擇胎兒芯片的好處：

1. **更廣泛**：除了主要的染色體異常疾病之外，芯片更可檢測出超過一百種已知的基因序列缺失/重複及由單親同二體引起的疾病。有關這些遺傳性疾病的資料，可瀏覽網頁：
https://www.obg.cuhk.edu.hk/services/laboratory-services/chromosomal-microarray-analysis/
2. **更細緻**：解像度比傳統的染色體核型分析高出五十倍，能夠診斷出顯微鏡下觀察不到的染色體異常。
3. **更快捷**：七天內便可以取得報告，傳統的染色體核型分析報告需時約二至三星期。

病例：



胎兒頸皮增厚，唐氏綜合症篩查屬高危，但核型分析正常，胎兒芯片檢測發現DNA序列缺失病Mowat-Wilson綜合症。

參考文獻：Choy KW, To KF, Chan AW, Lau TK, Leung TY. Second-trimester detection of Mowat-Wilson syndrome using comparative genomic hybridization microarray testing. Obstet Gynecol 2010;115(2 Pt 2):462-5.

選擇芯片需要注意的事項：

1. 胎兒DNA芯片不能檢測**所有類型**的基因組變異，包括平衡易位、染色體倒位、低比例嵌合體及點突變。

2. 即使報告結果正常，嬰兒亦有極少機會存在其他胎兒DNA芯片沒有涵蓋的基因變異。

香港中文大學 婦產科學系
The Department of Obstetrics and Gynaecology
The Chinese University of Hong Kong

胎兒 DNA 芯片 版本 2.0

Fetal DNA Chip version 2.0

查詢電話
(852) 5569 6412 (辦公時間內)
(852) 5600 1970 (/)

若想知道更多資料，可向您的專科醫生查詢詳情，或瀏覽我們的網頁。

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What is Fetal DNA Chip V2?

The Fetal DNA Chip testing was first launched in Hong Kong in 2009 by the Department of Obstetrics and Gynaecology of the Chinese University of Hong Kong. It is specifically designed to diagnose common micro-deletion / duplication syndromes which would not be detected by conventional karyotype analysis. The version 2 chip now also enables us to diagnose uniparental isodisomy.

Who will benefit from this test?

1. Fetal abnormalities revealed by ultrasound examination
2. Positive result of Down's Syndrome Screening
3. Previous baby with congenital abnormalities
4. Couples being chromosomal abnormalities carriers
5. Unexplained stillbirth
6. Pregnant women undergo invasive testing due to high-risk screening result

Flow:

High risk pregnancies

↓

Genetic Counseling

↓

Fetal DNA sampling (via chorionic villus sampling or amniocentesis)

↓

Fetal DNA Chip testing

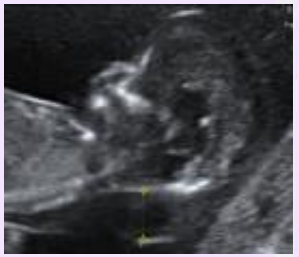
↓

Report (within 7 days)

Benefits from this test:

1. **More Comprehensive**: In addition to major chromosomal abnormalities, the Fetal DNA Chip can diagnose more than 100 recognized micro-deletion / duplication syndromes and uniparental isodisomy. For more details on these genetic disorders, please visit: https://www.obg.cuhk.edu.hk/services/laboratory-services/chromosomal-microarray-analysis/
2. **Higher Resolution**: 50-fold higher and detects much smaller abnormalities than karyotype analysis.
3. **Rapid Result**: Reporting time within 7 days. The reporting time for karyotype analysis usually takes 2 to 3 weeks.

Case example:



Fetal nuchal translucency is thickened and Down's syndrome screening test shows positive result. Karyotype analysis is normal. Fetal DNA Chip revealed a *ZEB2* deletion in chromosome 2, which is the cause of Mowat-Wilson Syndrome

Reference: Choy KW, To KF, Chan AW, Lau TK, Leung TY. Second-trimester detection of Mowat-Wilson syndrome using comparative genomic hybridization microarray testing. Obstet Gynecol 2010;115(2 Pt 2):462-5.

Important notes for Fetal DNA Chip:

1. Does not test for ALL genetic conditions: Unable to detect balanced translocations, inversions, low level mosaicism and point mutations.
2. Even the test result is normal, it cannot exclude genetic causes not detected by Fetal DNA Chip.

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(852) 5569 6412 (office hours)
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For more details, please refer to your specialist or visit our website.

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