



Postnatal DNA chip

Postnatal DNA 檢測晶片

什麼是DNA 檢測晶片?

香港中文大學婦產科與美國貝勒醫學院遺傳醫學實驗室合作提供DNA檢測晶片服務。

DNA檢測晶片服務使用由貝勒醫學遺傳學實驗室所提供的染色體微陣列分析(CMA)。

CMA是一個新的分析染色體方法，通過利用基因晶片技術(arrayCGH)檢測多種遺傳性疾病，它覆蓋整個基因組約180K的寡核苷酸，並以高解像度分析。DNA檢測晶片可以檢測超過130種基因疾病、及因序列重複或缺失而引起之遺傳性疾病。

CMA 涵蓋範圍搜索：
<https://www.bcm.edu/geneticlabs/disorder.cfm>

檢測方法

利用兩種不同的螢光染料分別標記測試樣本及對照樣本的DNA再進行雜交，比較兩種不同螢光強弱差異並對結果進行分析，通過量成像方法和分析軟件以確定測試樣本中每個目標DNA序列的有否缺失、重複或是正常的拷貝數目。

DNA 檢測晶片的限制

DNA檢測晶片是一個新的和敏感的測試，但只能探測到的基因的缺失或重複。即使測試是非常準確，但並不是每一個基因的異常都可以檢測得到。

這個測試不能檢測染色體平衡易位、倒位、低比例鑲嵌型或點突變。

誰有需要做DNA 檢測晶片?

- 先天性缺陷，不明原因的發育遲緩 / 智力發展遲緩，自閉症譜系障礙，多發性先天異常的患者
- 復發性流產的夫婦

檢測樣本及報告

所需樣本#：血液(或唾液/皮膚活檢)

- 成人: 5 毫升
- 小童: 3 毫升
- 嬰兒: 1 毫升

檢測樣本須連同父母血液樣本一同檢測

報告：15個工作天內可取

若想知道更多資料，可向您的專科醫生查詢 或 歡迎致電2632-4219 或 請瀏覽網頁

<https://www.fetalmedicine.hk/en/oscarbooking.asp>



The Department of Obstetrics and Gynaecology
The Chinese University of Hong Kong

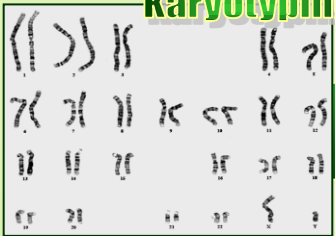
香港中文大學 婦產科學系



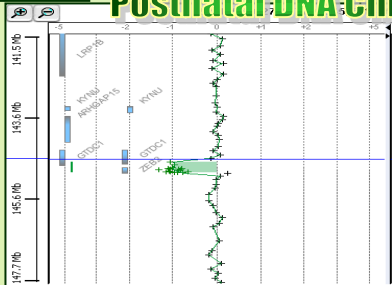
Table 1: CMA comparison to Karyotyping
 表 1 : CMA 與傳統染色體核型分析的比較

	Karyotyping 傳統染色體核型分析	Postnatal DNA chip Postnatal DNA 檢測晶片
Coverage	Aneuploidies and major structural chromosomal abnormalities more than 5Mb	Aneuploidies, structural chromosomal abnormalities and over 130 recognized genetic syndrome
覆蓋	非整倍體、染色體主要結構異常 (>5Mb)	非整倍體、染色體結構異常及超過130種已知基因疾病
Others 其他	-	Couple's blood are required 須連同父母血液樣本一同檢測

Karyotyping



Postnatal DNA Chip



What is Postnatal DNA Chip?

The Department of Obstetric and Gynaecology of the Chinese University of Hong Kong in collaboration with Baylor College of Medicine in the United States to offer Postnatal DNA Chip service. Postnatal DNA Chip service is using Chromosomal Microarray Analysis (CMA) designed by the Baylor Medical Genetics Laboratories. CMA is a new analyzing chromosomes method for a large number of genetic disorders by utilizing Array-based Comparative Genomic Hybridization (aCGH) technique. It has approximately 180K oligos covering the whole genome at high resolution. The Postnatal DNA Chip can detect over 130 recognized genetic syndromes and other rare genetic disorders reported to be associated with mental retardation and/or physical problems. Additionally it includes common micro-deletion / duplication syndromes. The CMA coverage can be search at <https://www.bcm.edu/geneticlabs/disorder.cfm>

Methodology

Genomic DNA from the test sample and a control sample are differentially labeled with fluorescent dyes and hybridized to the oligos. Results are analyzed to assist in identifying each targeted-DNA sequence as loss of copy number (deletion), gain of copy number (duplication) or normal copy number by using quantitative imaging methods and analytical software.

Limitations

The Postnatal DNA Chip is a new and sensitive test but it can only detect the gain or loss of genomic materials. There are not every genetic abnormality can be detected by CMA even the test is very accurate. This test can not detect low level mosaicism, balanced translocations, inversions, or point mutations that may be responsible for the clinical phenotype.

Who will be referred to perform this test?

- Patients who have dysmorphic features, unexplained developmental delay / intellectual disability, autism spectrum disorders, multiple congenital anomalies
- Recurrent miscarriage couples

Specimen & Reporting

Specimen requirement*:

Blood in EDTA tube (or saliva/skin biopsies)

- Adult: 5 ml
- Chlid: 3 ml
- Infant: 1 ml

*Patient samples with the parents' EDTA blood

Reporting time: Within 15 working days

For more details, please refer to your clinician or contact us at 2632-4219 or visit our website:
<https://www.fetalmedicine.hk/en/oscarbooking.asp>