

基因體晶片 (CMA)

檢測圖表說明

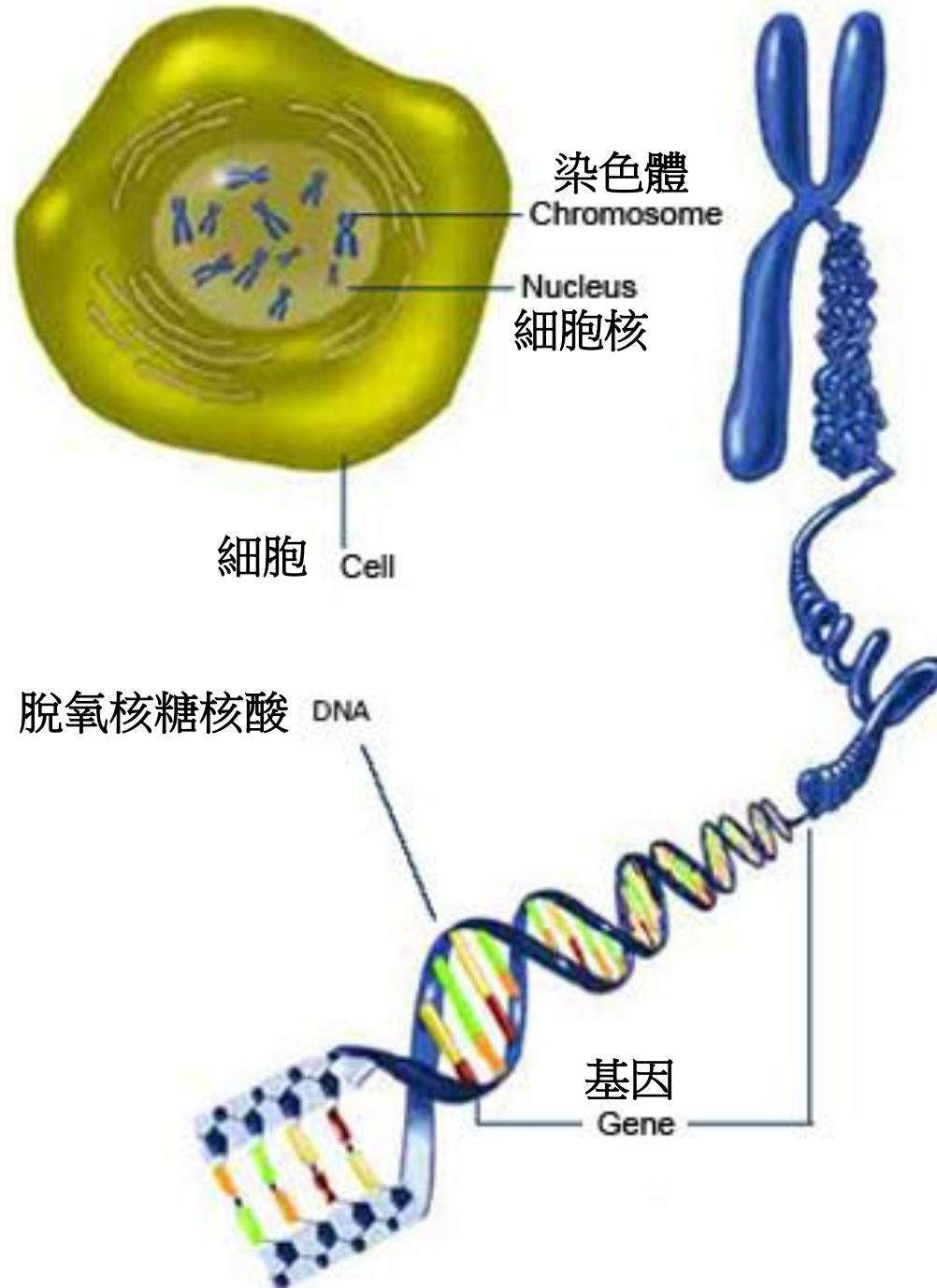
Diagrams for Chromosomal Microarray (CMA) testing



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傳統染色體核型分析:利用顯微鏡檢測染色體

Conventional karyotyping: examination of chromosomes under the microscope



染色體組型圖譜

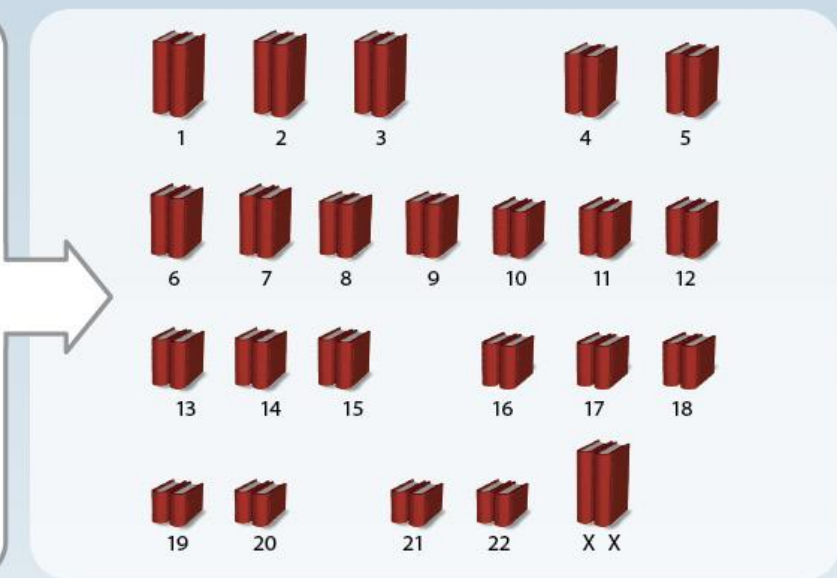
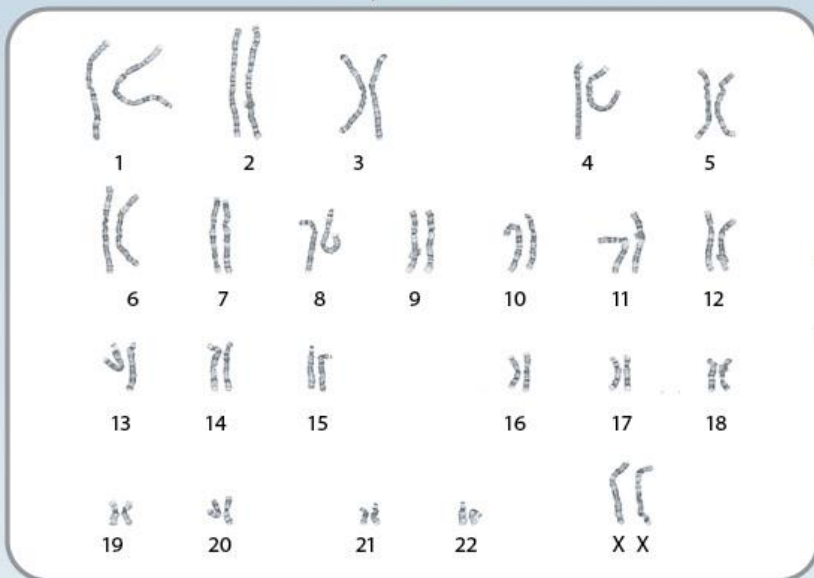
TYPICAL FEMALE AND MALE KARYOGRAMS

♀ female

完整的百科全書集

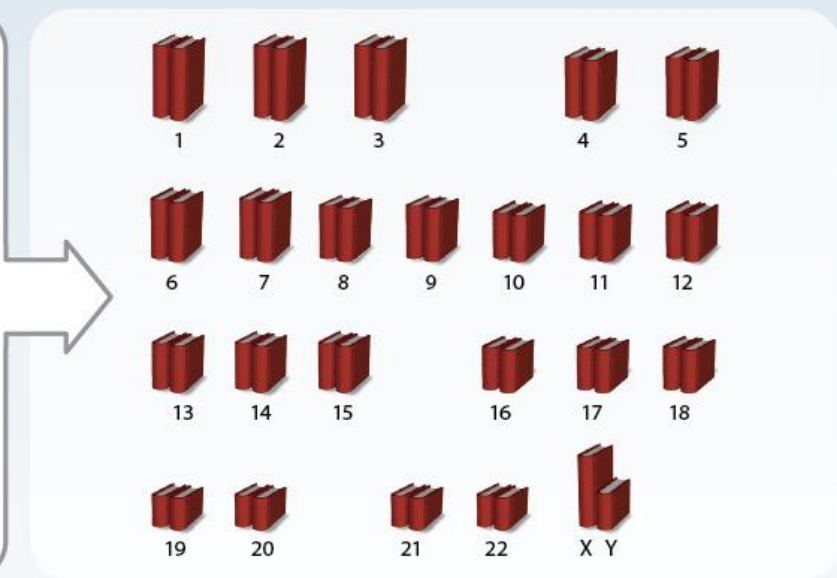
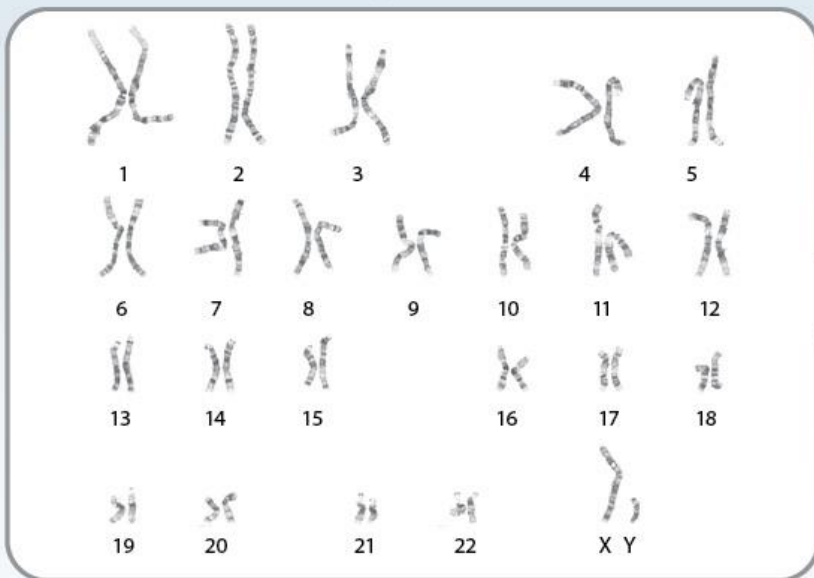
COMPLETE ENCYCLOPEDIA SET

女性
46,XX



♂ male

男性
46,XY



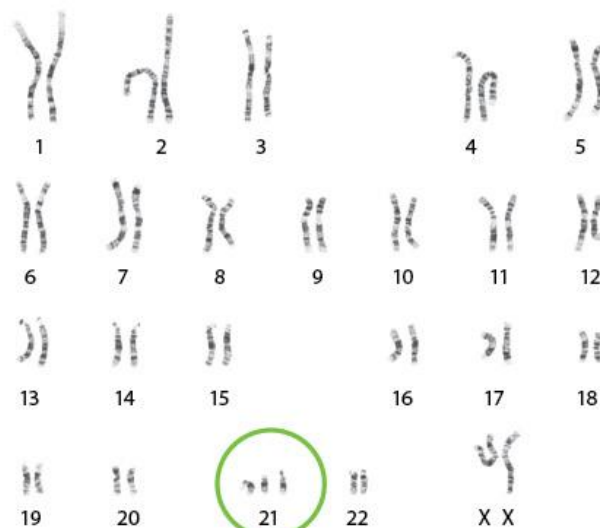
異常染色體組型圖譜

ABNORMAL KARYOGRAMS

Gain of chromosome material: **Trisomy 21** (*Down syndrome*)

染色體增多

+21



Loss of chromosome material: **5p-** (*Cri du chat syndrome*)

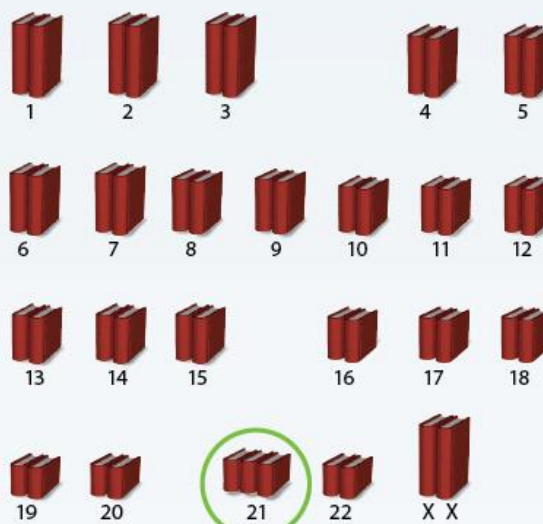
染色體減少

5p-

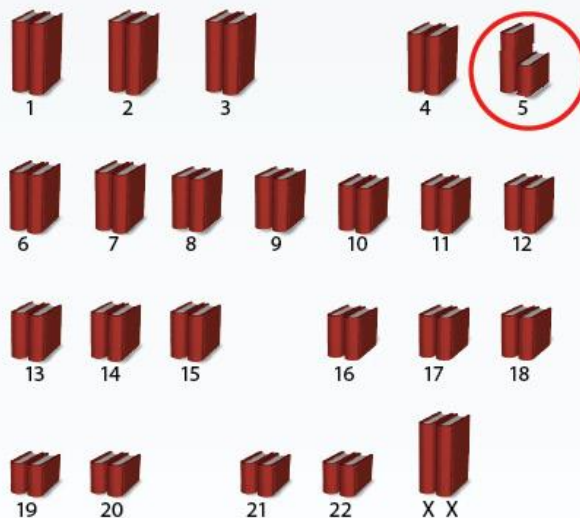


異常百科全書集

ABNORMAL ENCYCLOPEDIA SETS



第21卷額外多一本書 An extra book in Volume 21

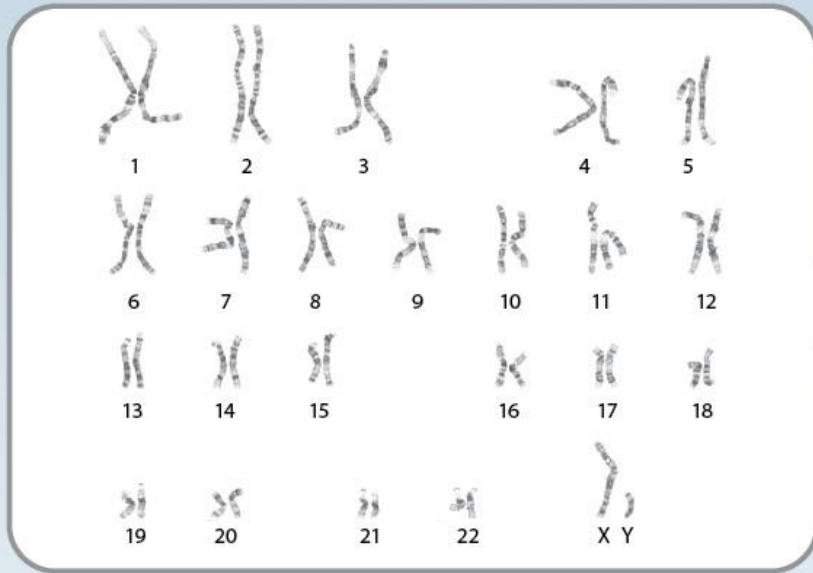


一本書在第5卷比其他短 One book shorter than the other in Volume 5

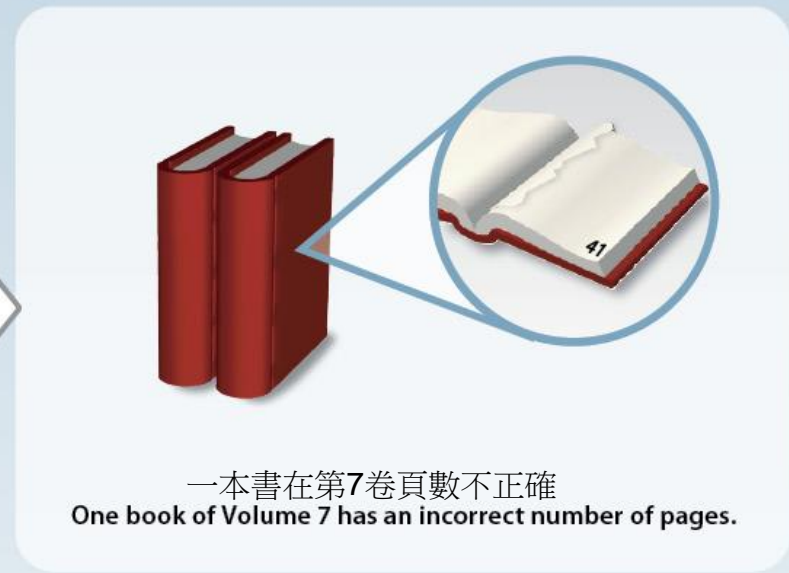


即使傳統分析顯示染色體屬正常，CMA檢測仍有可能發現傳統分析未能檢測到的基因體數量變化

NORMAL KARYOGRAM



ABNORMAL ENCYCLOPEDIA VOLUME (INSIDE)



OR



Pages within the chromosome or encyclopedia volume are removed or **deleted**.

Pages within the chromosome or encyclopedia volume are present in multiple copies or **gained**.

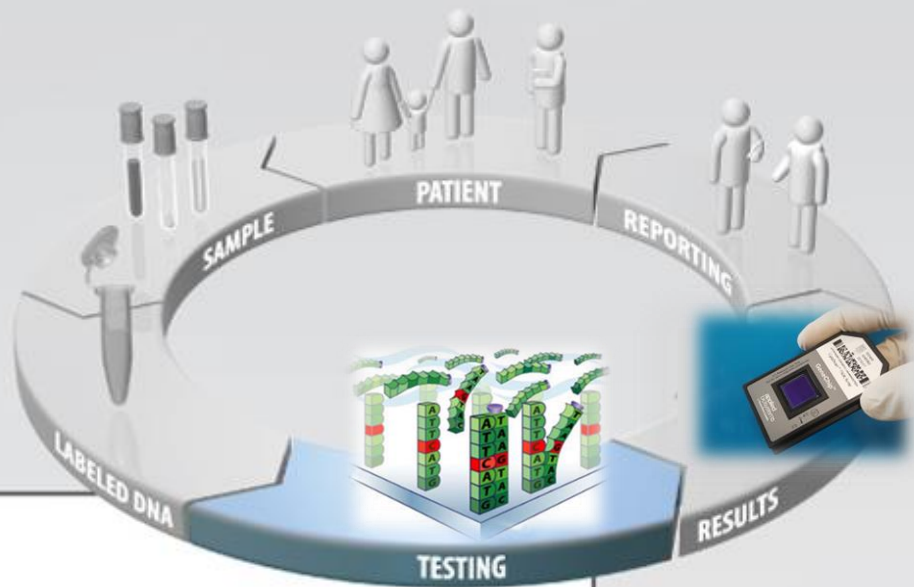
頁數被刪除 → **基因體減少**

頁數被重複 → **基因體增加**



How does CMA testing work?

如何進行基因體晶片(CMA)檢測?

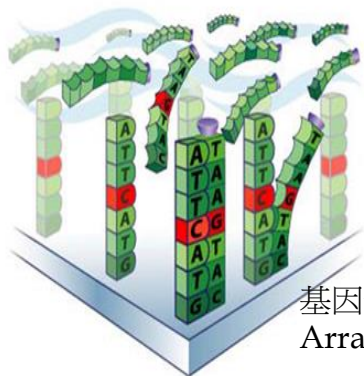


Fluorescently labeled DNA hybridize to chromosomal microarray

螢光標籤DNA在基因體晶片上作配對

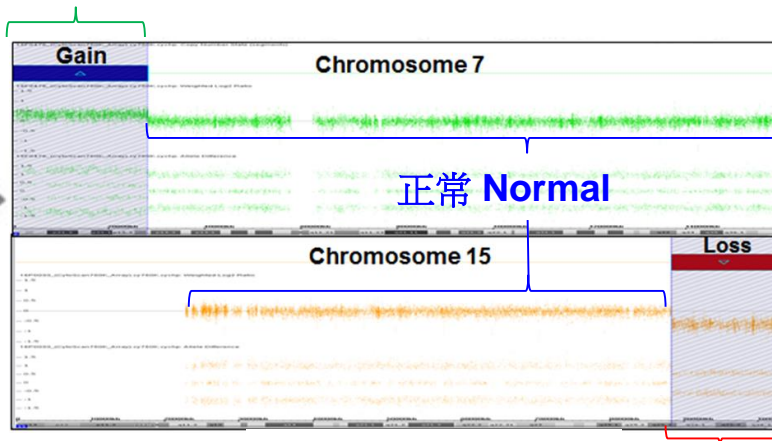
患者DNA
Patient DNA

基因體增加 DNA dosage gain



在晶片上
作配對

Hybridization



基因體減少 DNA dosage loss

