

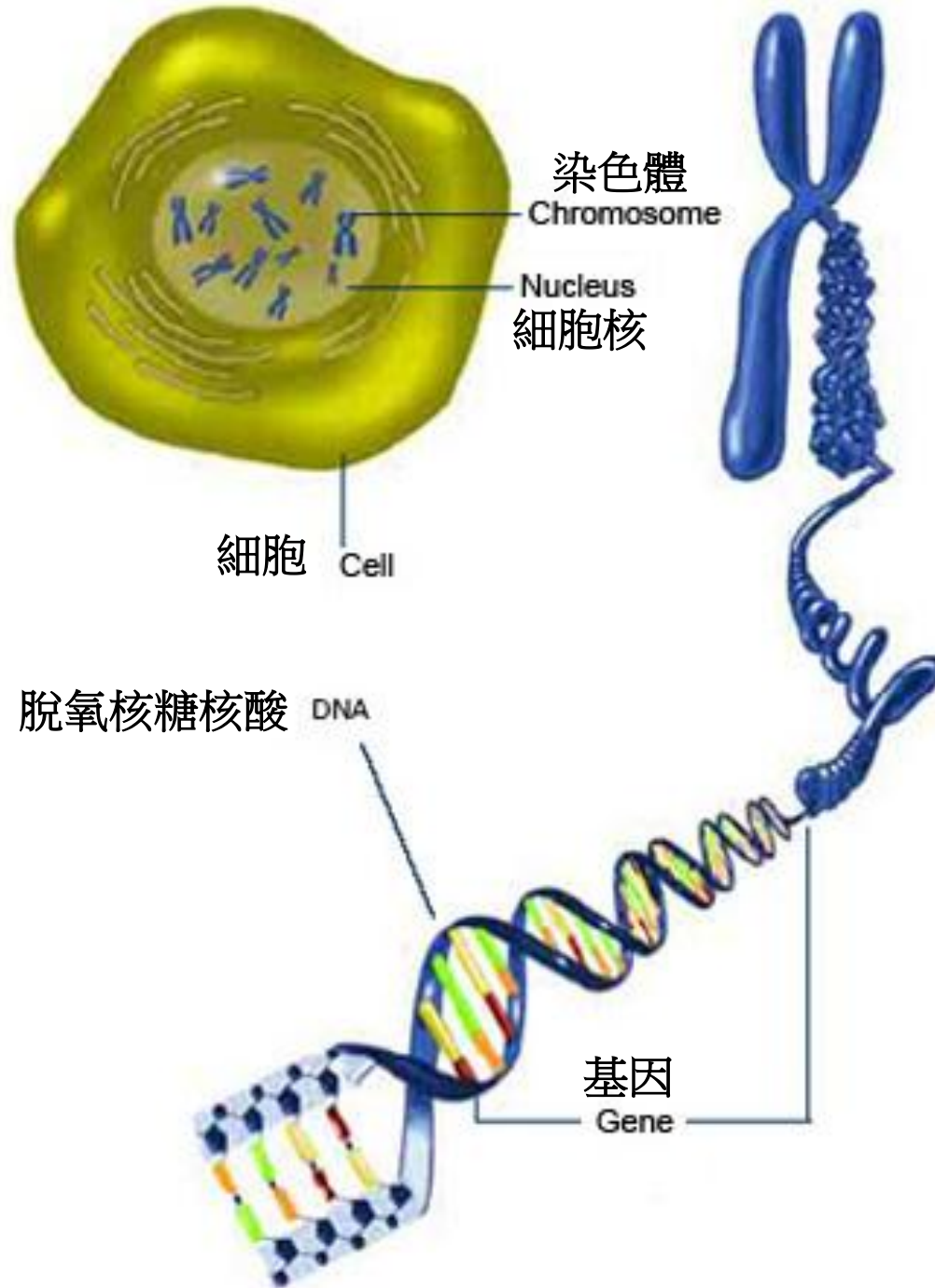
產前晶片全基因體定量分析 (aCGH)檢測圖表說明

Diagrams for Prenatal whole genome aCGH testing



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Prenatal Diagnostic and Counselling Department, Tsan Yuk Hospital



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

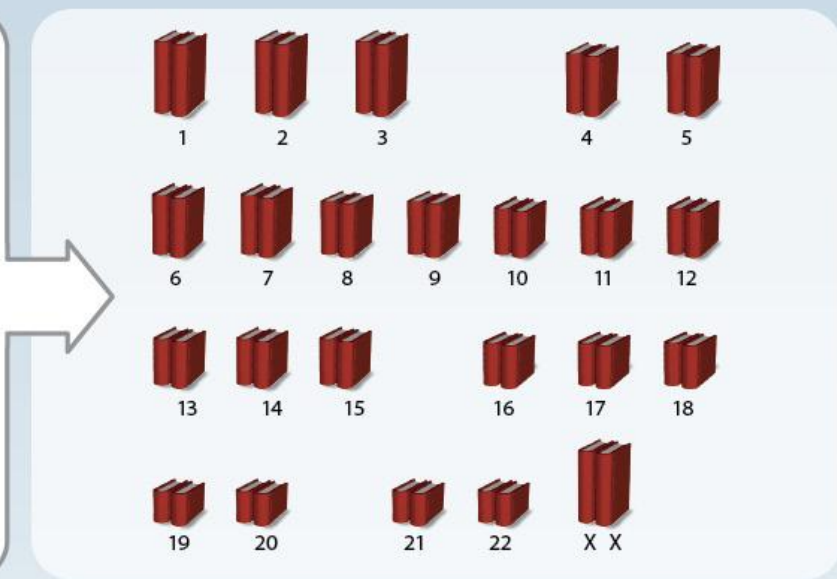
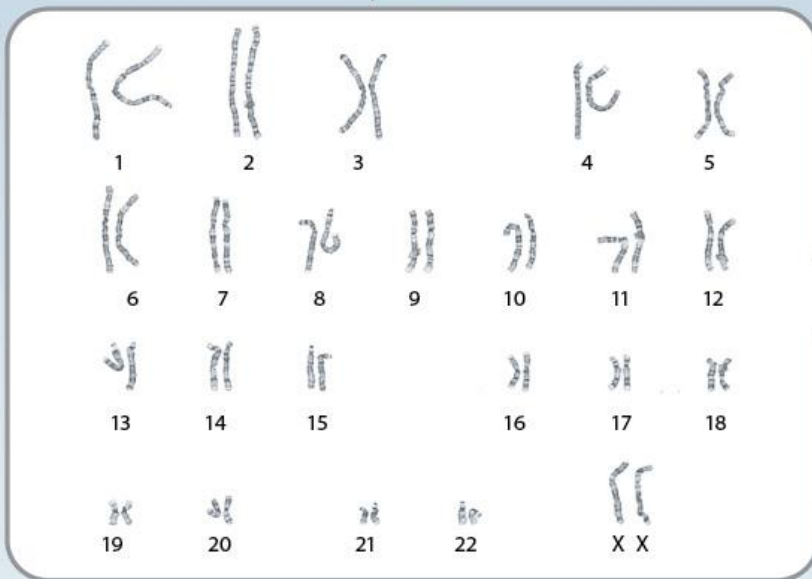
TYPICAL FEMALE AND MALE KARYOGRAMS

♀ female

完整的百科全書集

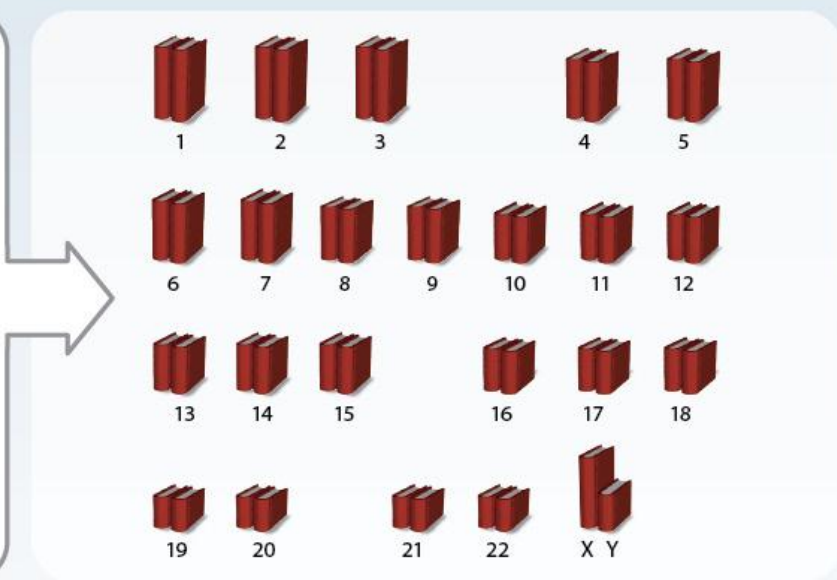
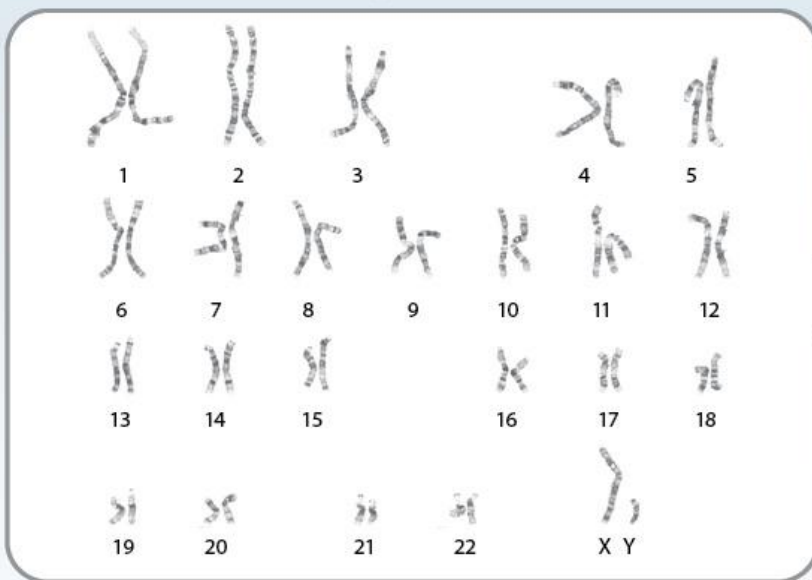
COMPLETE ENCYCLOPEDIA SET

女性
46,XX



♂ male

男性
46,XY



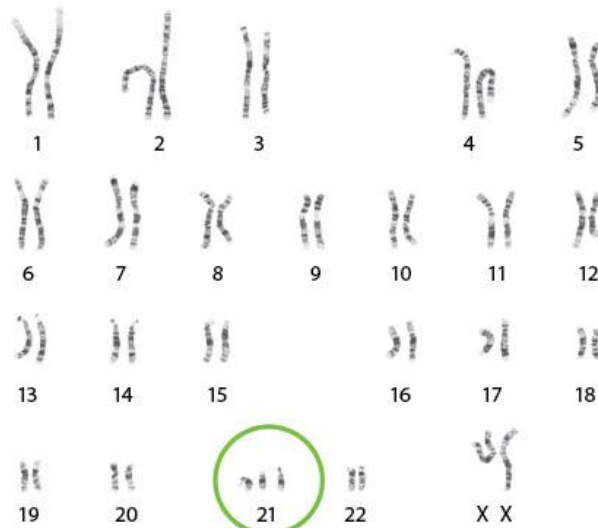
異常染色體組型圖譜

ABNORMAL KARYOGRAMS

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

染色體增多

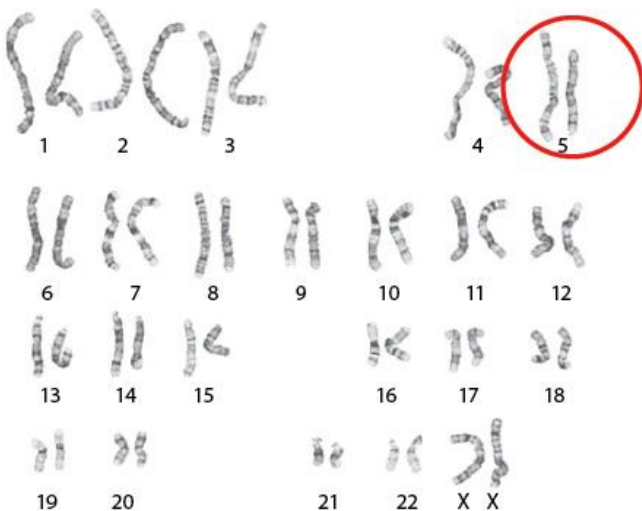
+21



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

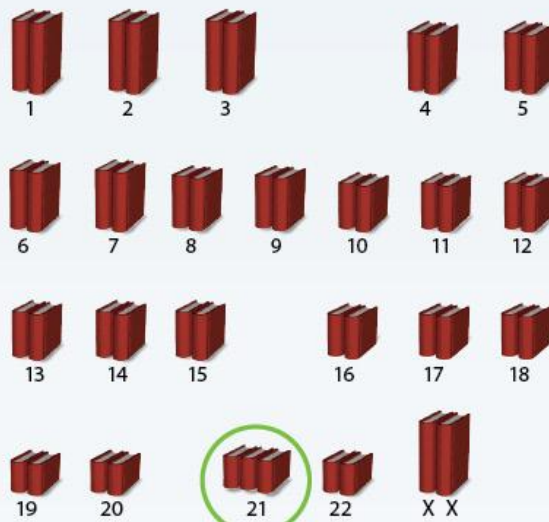
染色體減少

5p-

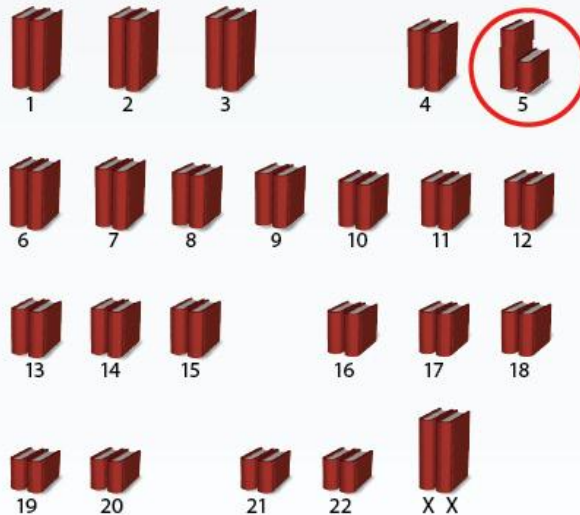


異常百科全書集

ABNORMAL ENCYCLOPEDIA SETS



第21卷額外多一本書 An extra book in Volume 21

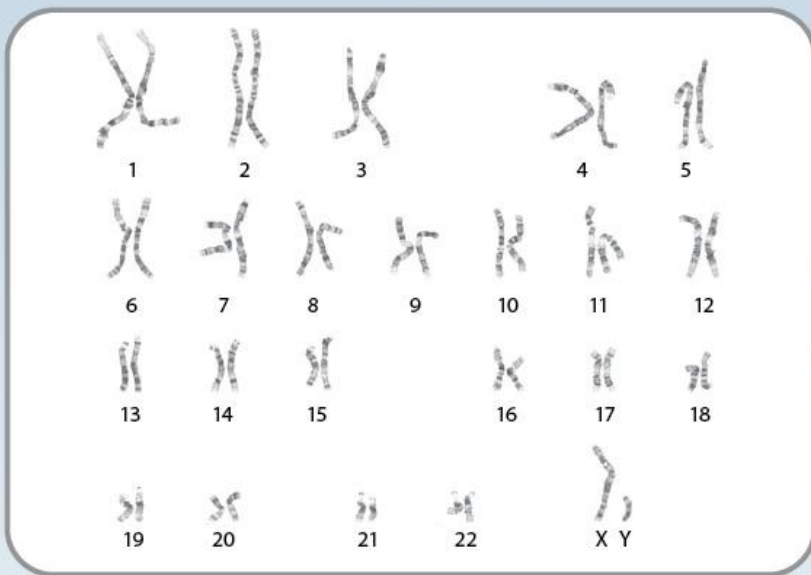


一本書在第5卷比其他短 One book shorter than the other in Volume 5

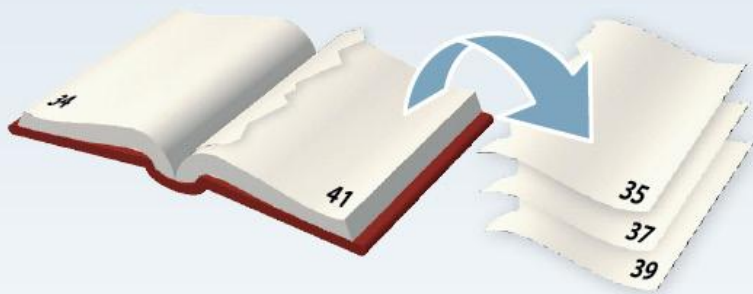
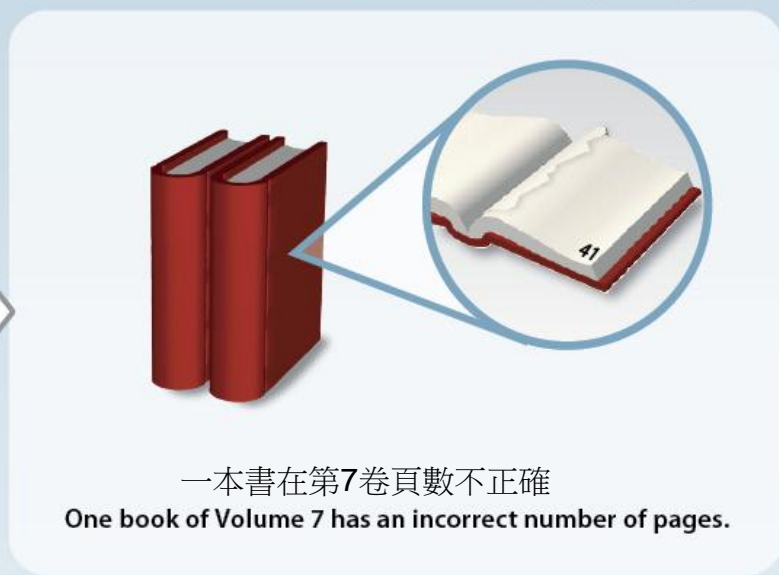


即使傳統分析顯示染色體屬正常，aCGH檢測仍有可能發現傳統分析未能檢測到的基因體數量變化

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

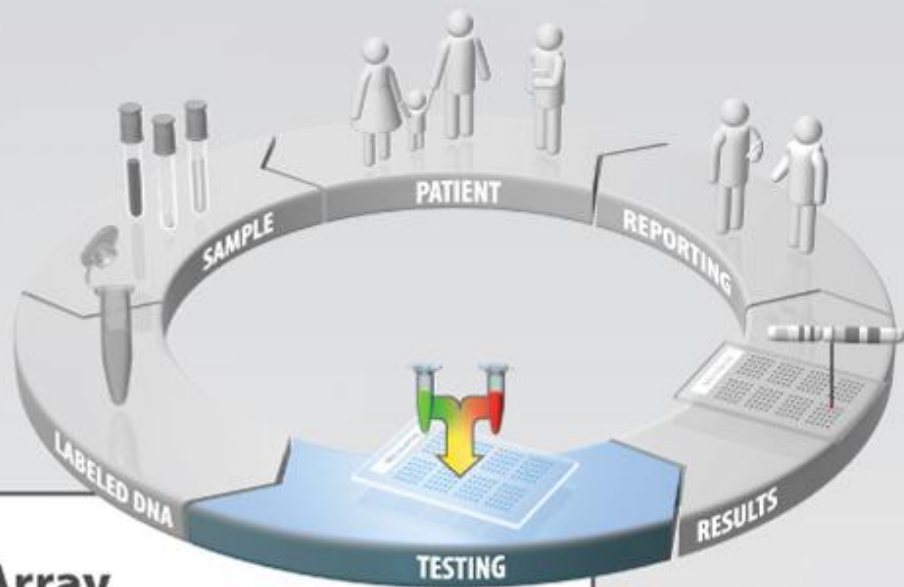
頁數被重複 → 基因體增加



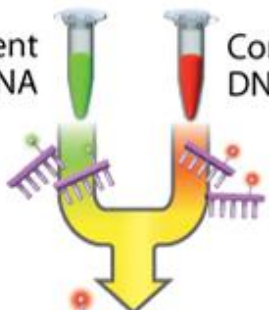
aCGH 檢測方法

胎兒基因體

正常的基因體



Patient DNA Control DNA



排列在晶片
上作配對

Hybridization

How Does Array CGH Testing Work?

Labeled patient and control DNA compete to attach or hybridize to the microarray slide.

基因體平衡

基因體減少

基因體增加

Equal hybridization

DNA dosage loss (e.g. deletion)

DNA dosage gain (e.g. duplication)

