

基因體結構與遺傳原理

Genome Structure & Genetic Principles

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演講綱要

- 孟德爾單基因遺傳學
- 染色體遺傳學說與與細胞減數分裂之染色體行爲
- 染色體結構，基因體組成，基因結構與功能
- 多基因遺傳模式
- 遺傳疾病種類與再發率















孟德爾(Gregor Mendel) 創立了遺傳學(Genetics)

- 在1865年,孟德爾發表了兩篇關於豌豆育種的研究論文。
- 這些研究論文包含了今日遺傳學最基礎的原理。



孟德爾的實驗

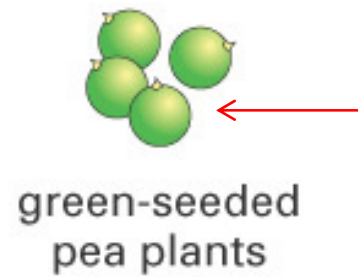
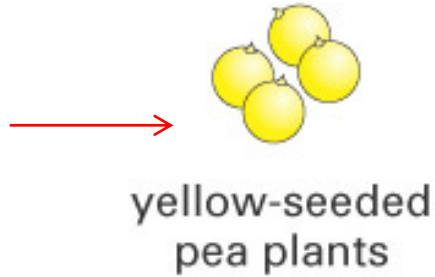
Mendel's Experiments

	Seed shape	Seed color	Flower color	Flower position	Pod shape	Pod color	Plant height
One form of trait (dominant)	 round (<i>R</i>)	 yellow (<i>Y</i>)	 purple	 axial flowers	 inflated	 green	 tall
A second form of trait (recessive)	 wrinkled (<i>r</i>)	 green (<i>y</i>)	 white	 terminal flowers	 pinched	 yellow	 short

表現型
Phenotype

基因型
Genotype

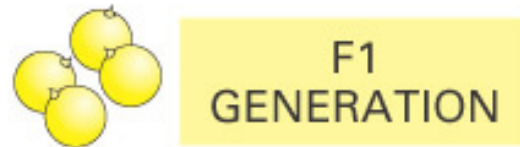
純種品系
Pure line



自體授粉
**Self-
fertilization**

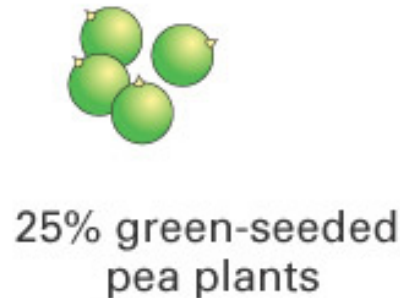
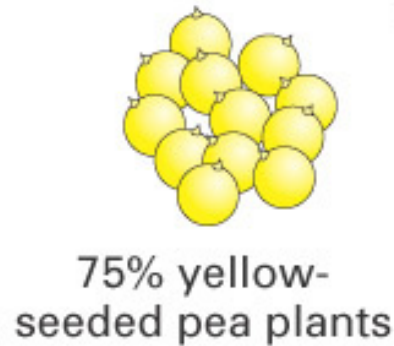
CROSS-
FERTILIZATION 交叉授粉

單型律
Law of
uniformity

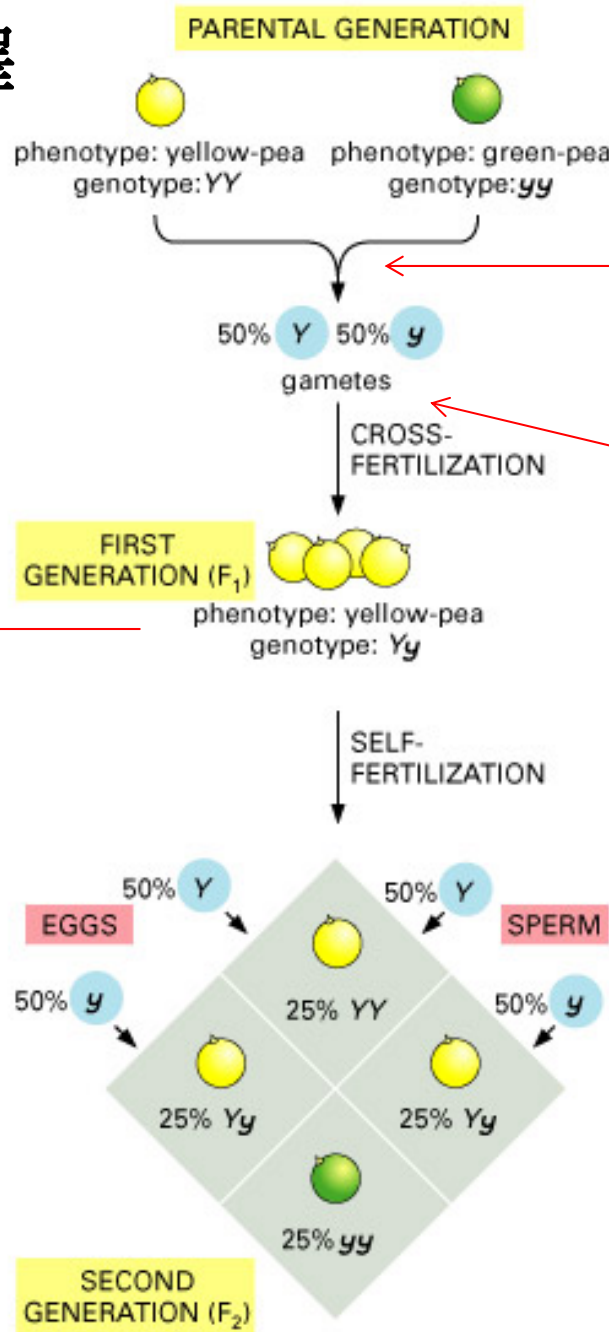


100% yellow-seeded pea plants

CROSSED TO
THEMSELVES



孟德爾如何解釋他的實驗結果



分離律
Law of segregation

配子
Gamete
如: 卵子或精子

對偶基因
allele
顯性(的)
dominant
隱性(的)
recessive

基因型與表現型的關係
(genotype-phenotype relationship)-
遺傳學的主題

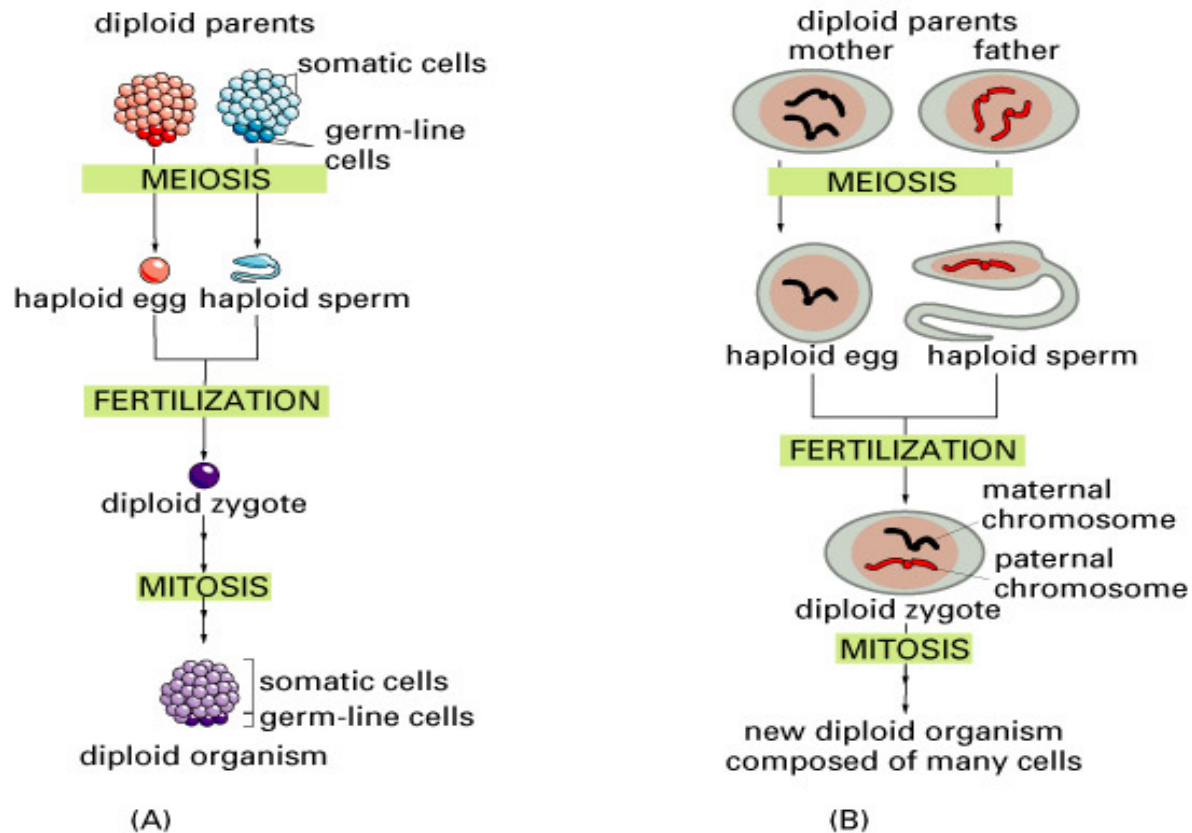
表現型
Phenotype
基因型
Genotype

手繪顯微鏡下細胞分裂(cell division) 時的染色體(chromosomes)



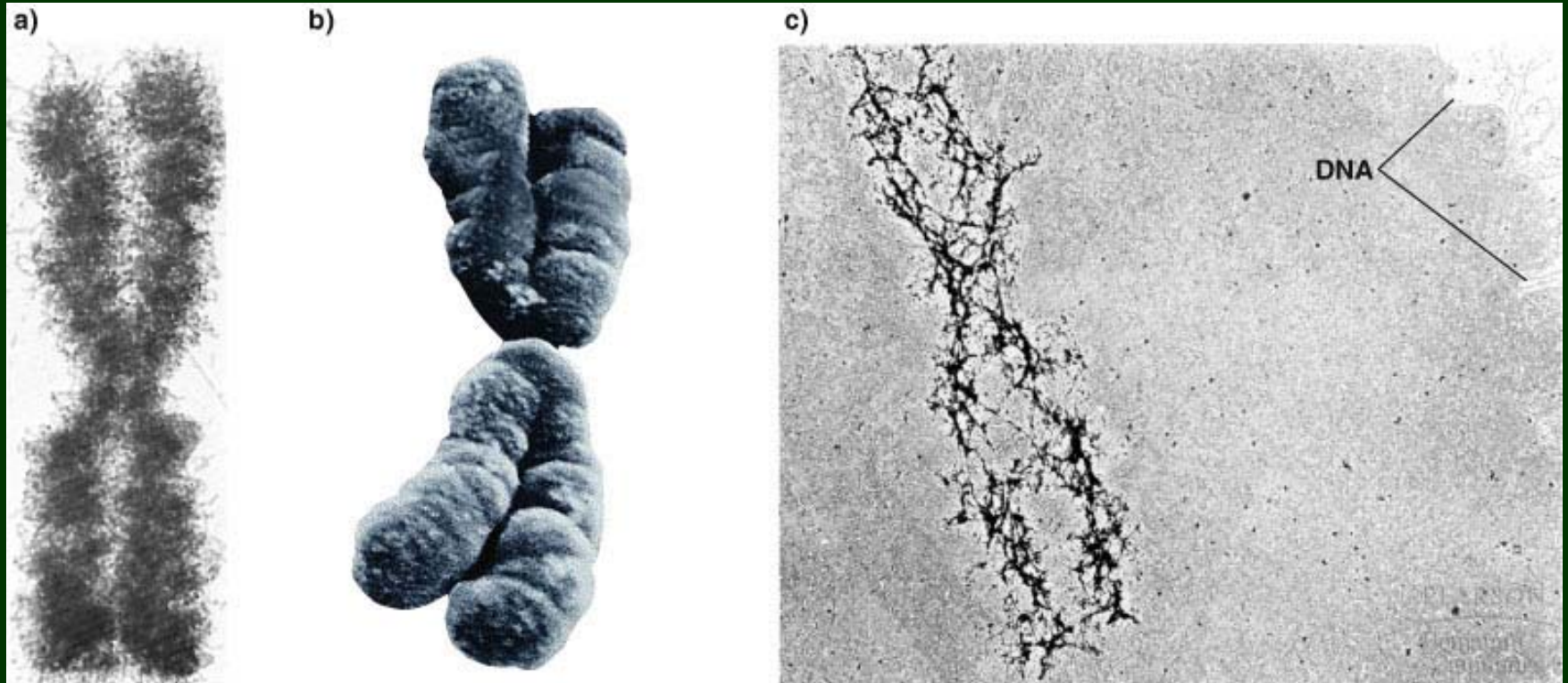
Adopted from Nature Review Genetics

生殖母細胞分裂產生配子過程中染色體的分配

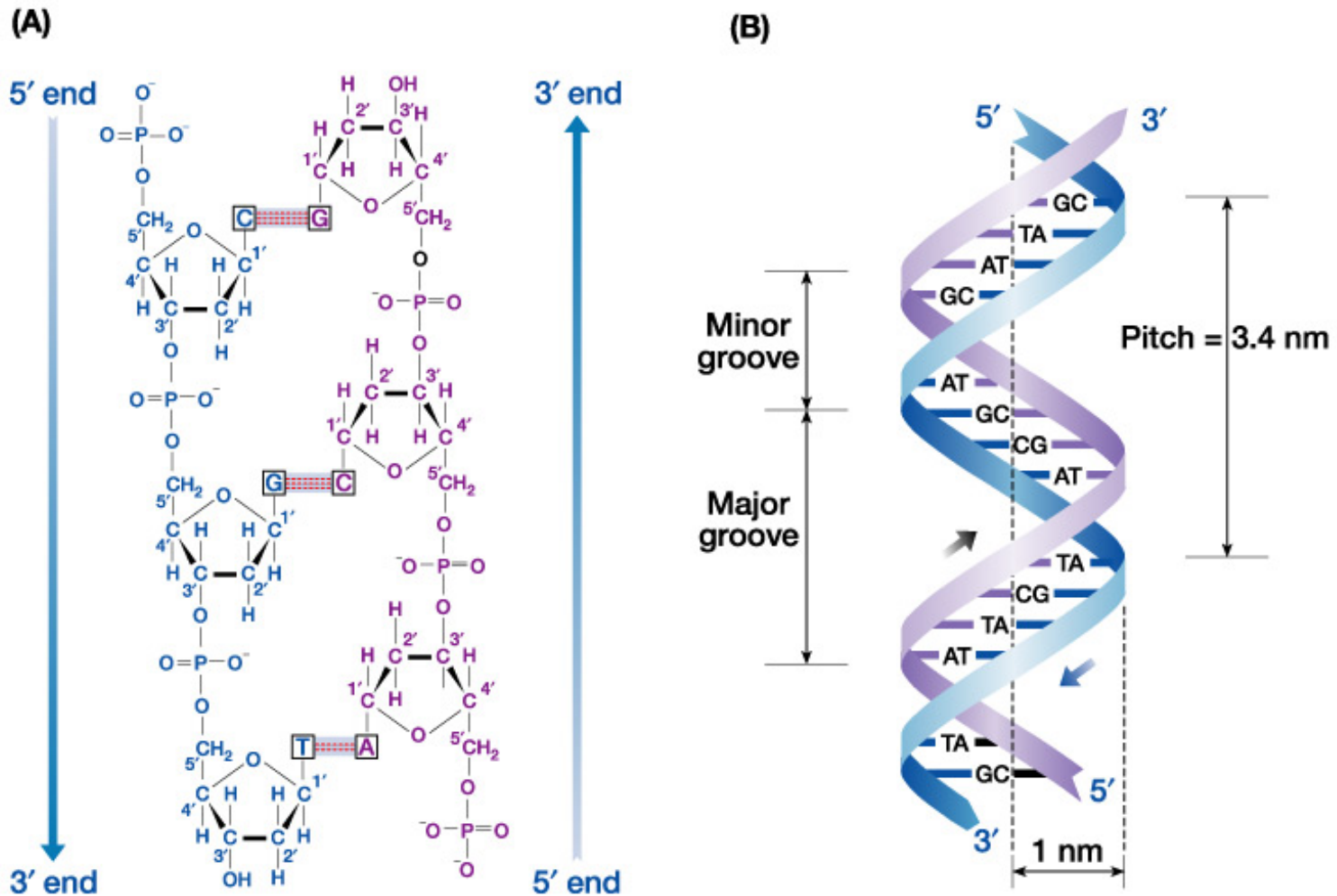




電子顯微鏡(electron microscope)下的 染色體(chromosome)



DNA 雙螺旋 (DNA double helix) 結構



DNA被逐步包裝成染色絲(chromatin)及染色體(chromosome)

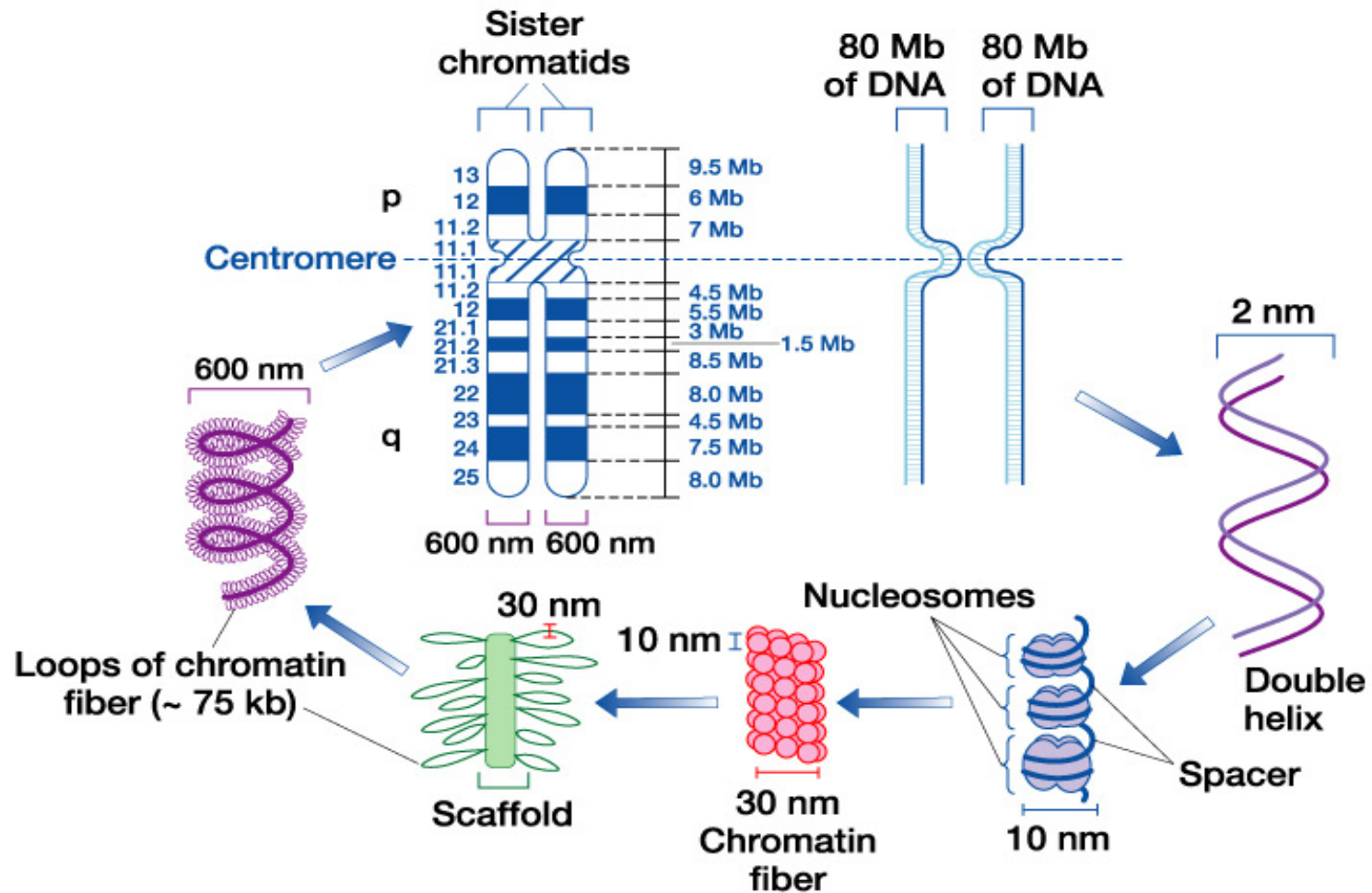
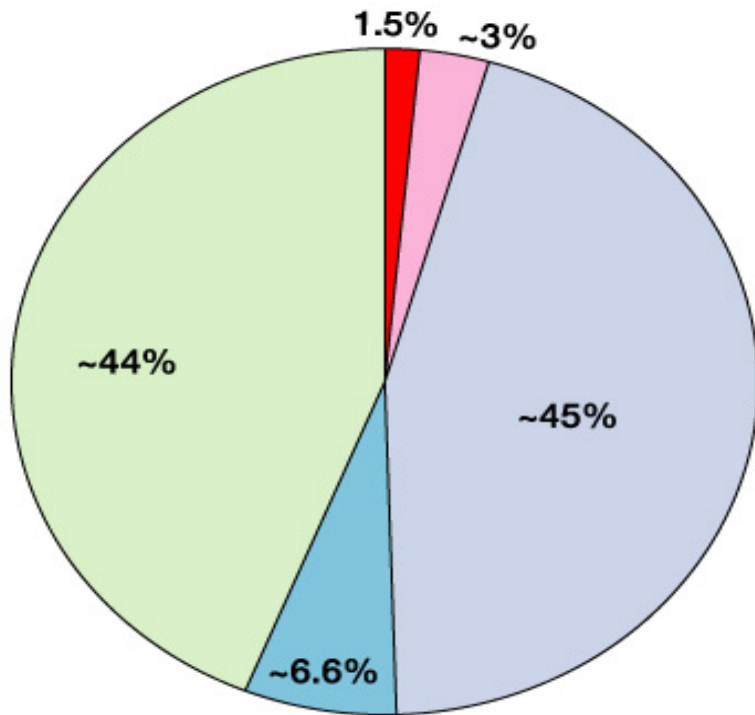


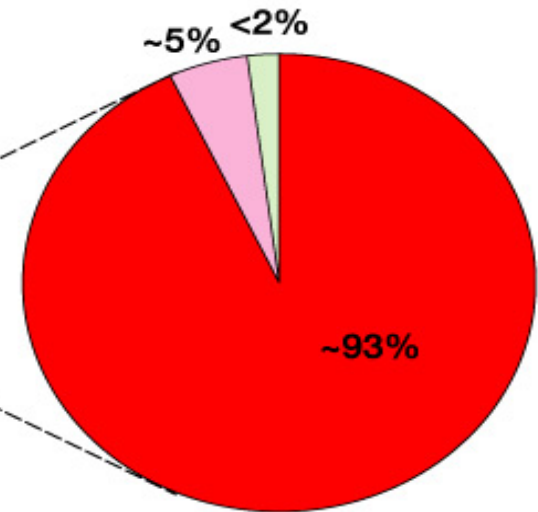
Figure 2-3 Human Molecular Genetics, 3/e. (© Garland Science 2004)

人類基因體的組成 (Organization of human genome)



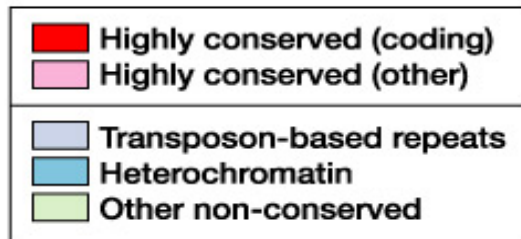
Nuclear genome
(24 linear double-stranded DNA molecules – 3200 Mb; ~30 000 genes)

細胞核
基因體

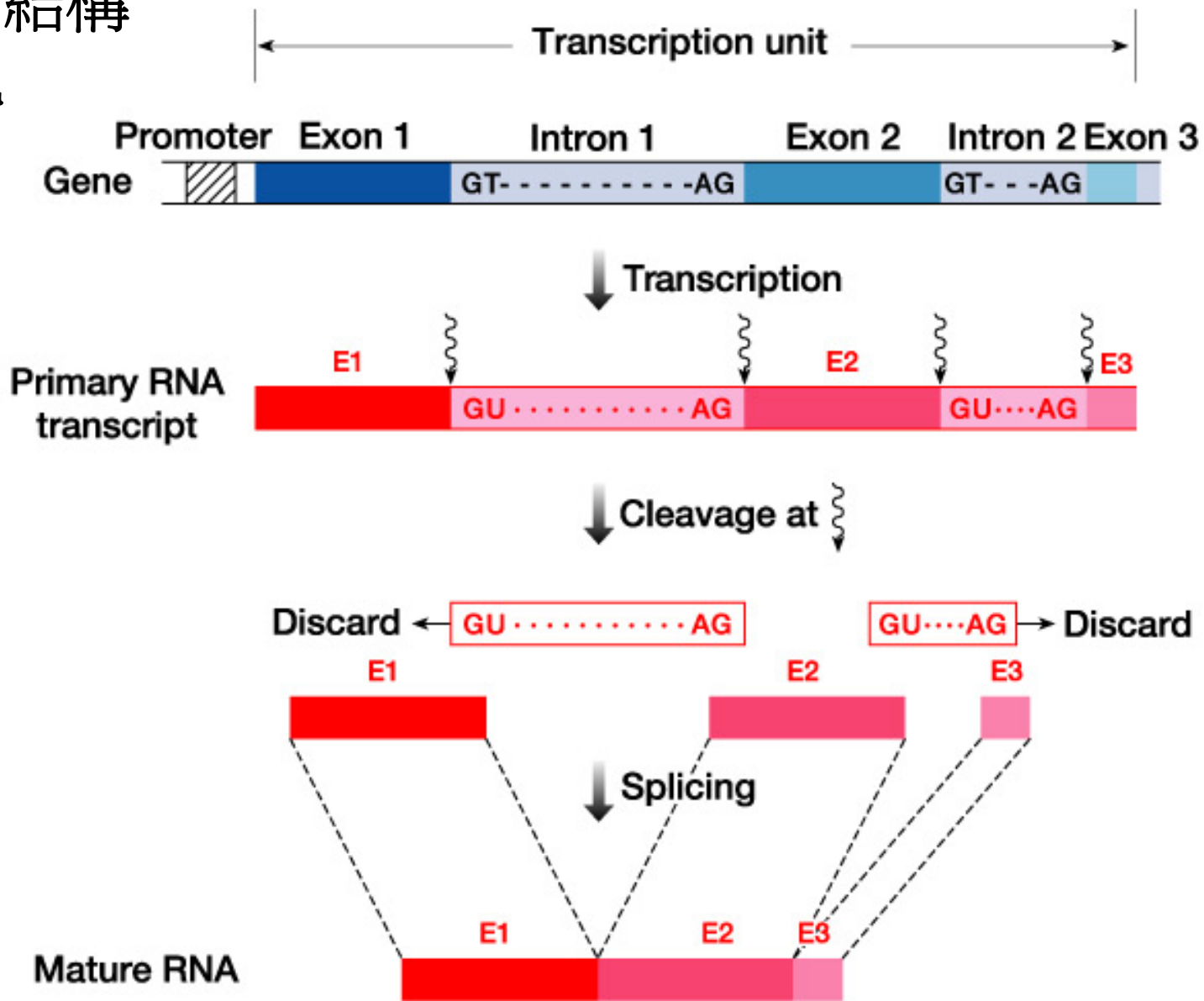


Mitochondrial genome
(1 circular double-stranded DNA 16.6 kb; 37 genes)

粒線體
基因體



基因的結構 與表現



分子生物學(molecular biology)的 中心理論(central theory)

DNA



RNA



蛋白質

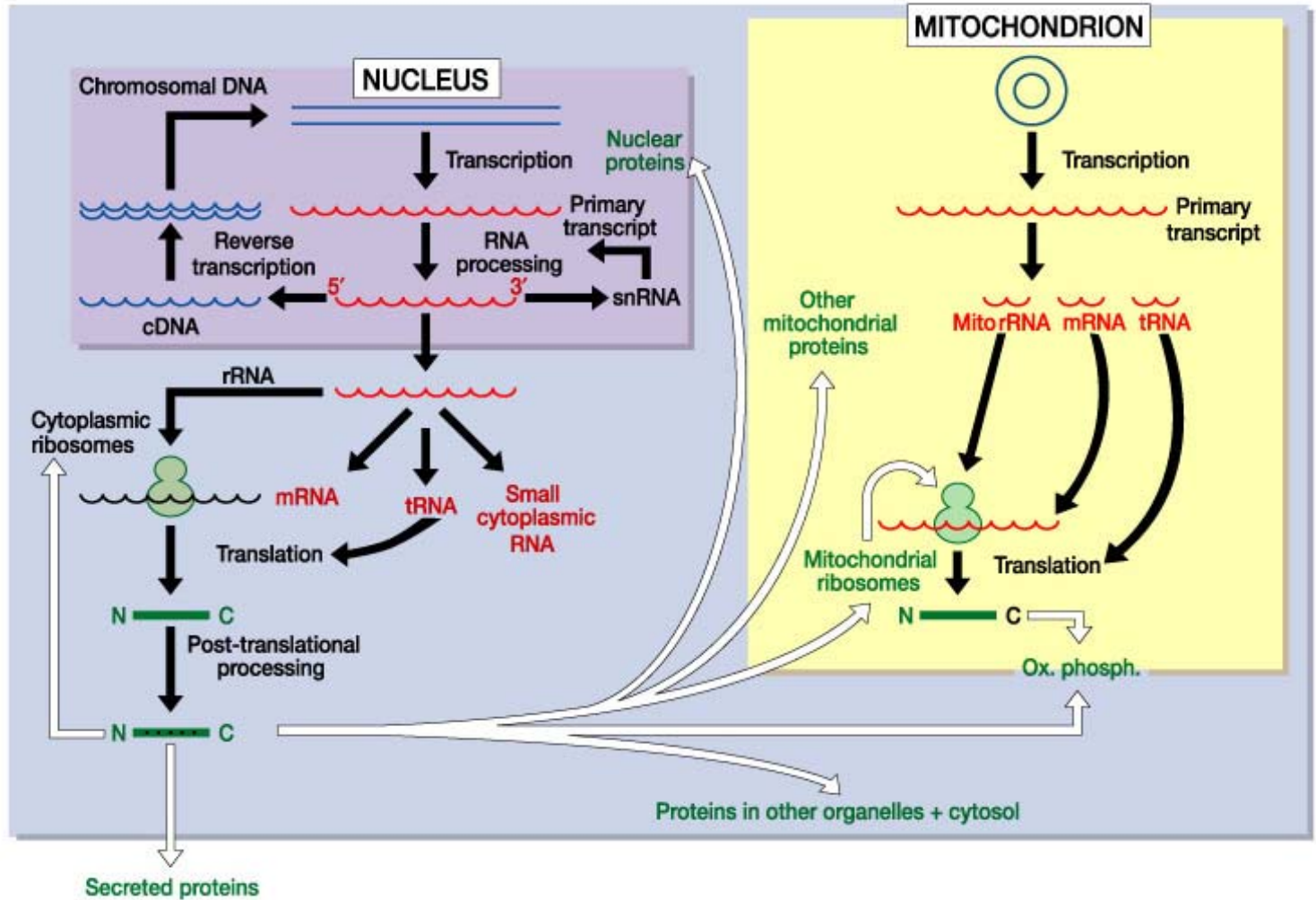
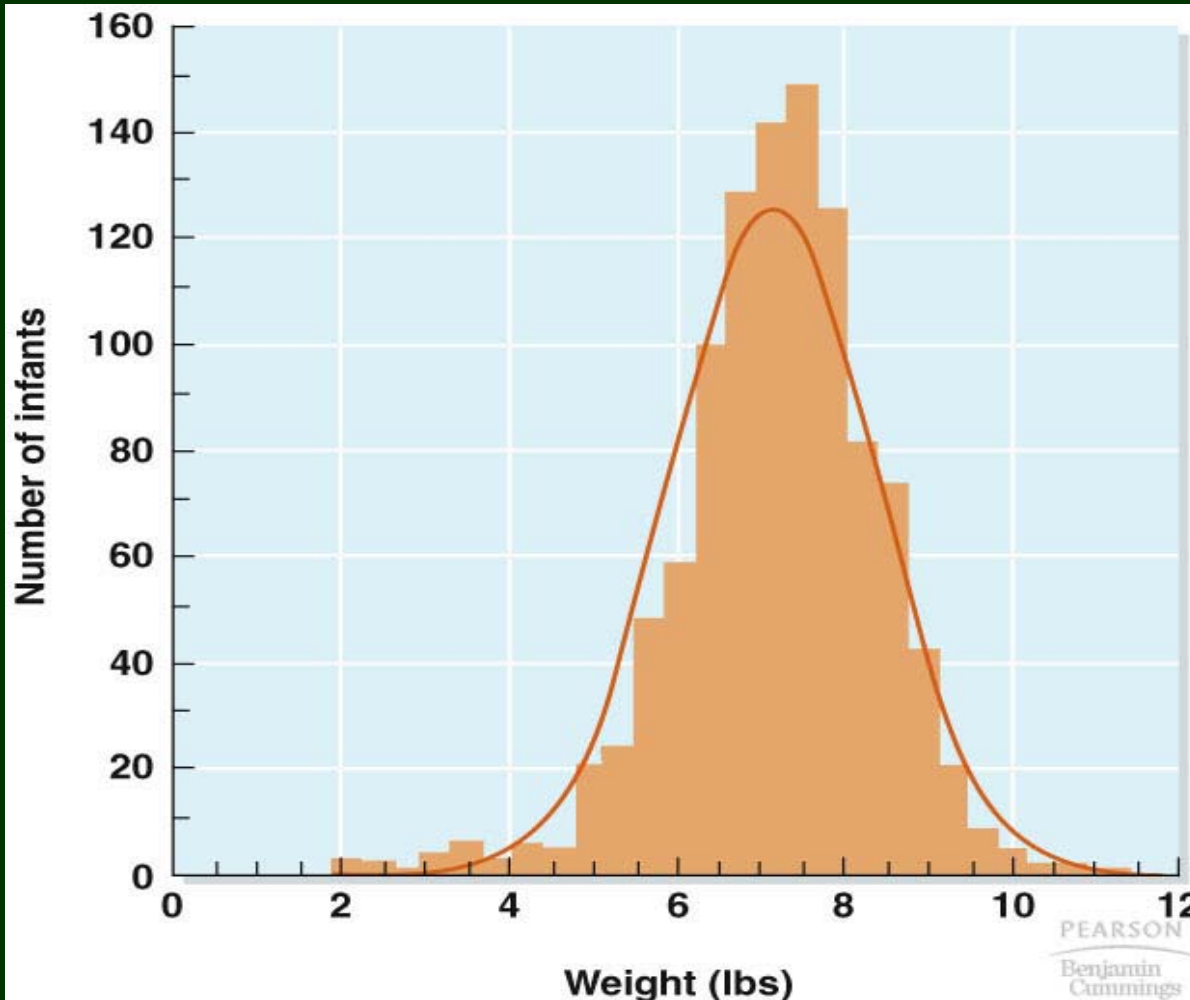


Figure 1-11 Human Molecular Genetics, 3/e. (© Garland Science 2004)

人類嬰兒出生體重分佈之直方圖(histogram)

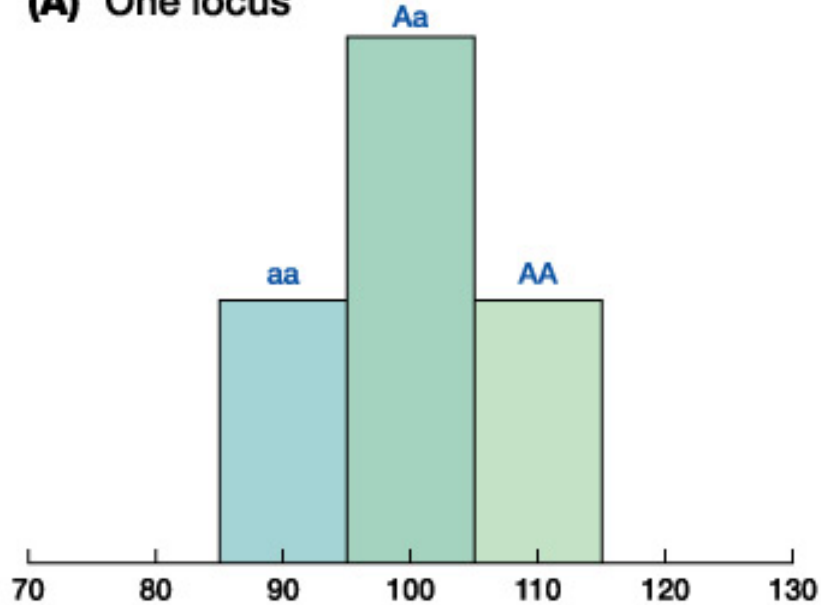


表現型是
連續性的特徵
(continuous trait)

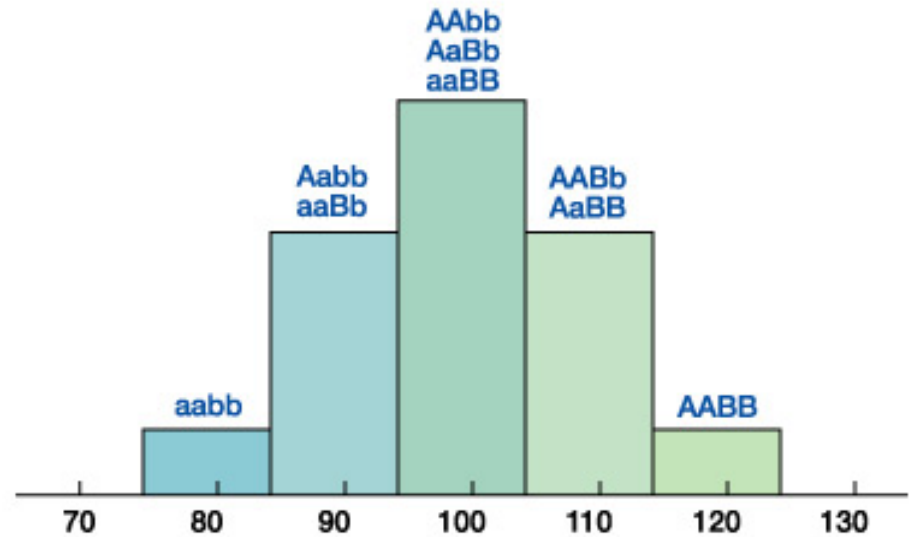
而不是孟德爾遺傳
學所研究之
二分法的特徵
(dichotomous trait)



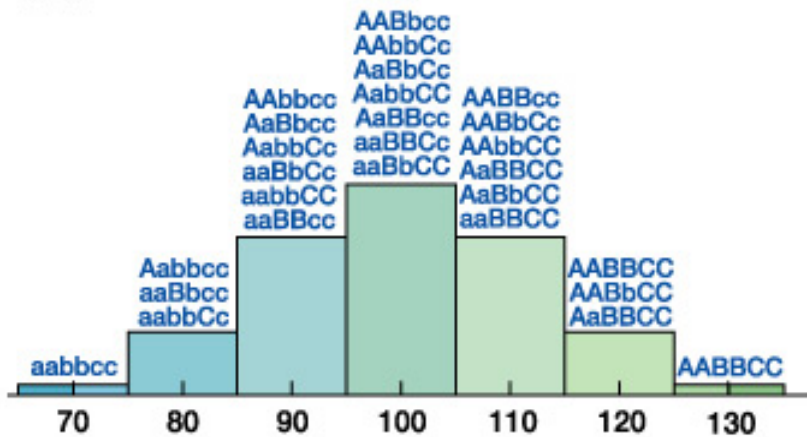
(A) One locus



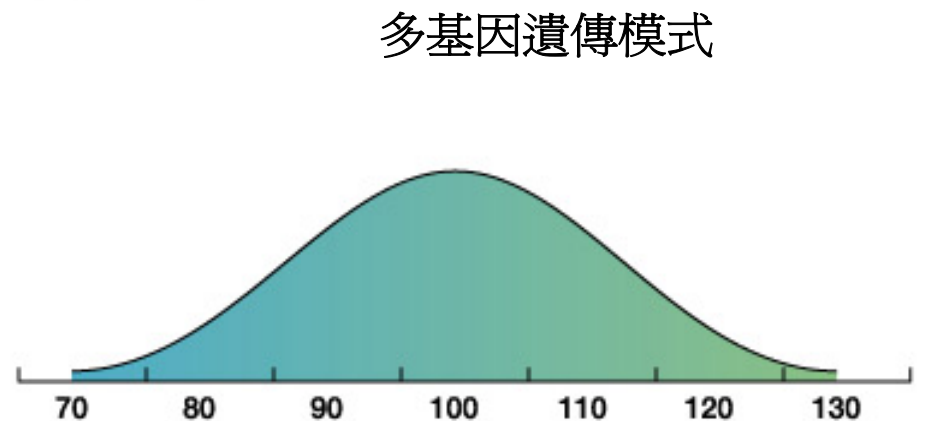
(B) Two loci



(C) Three loci



(D) Many loci

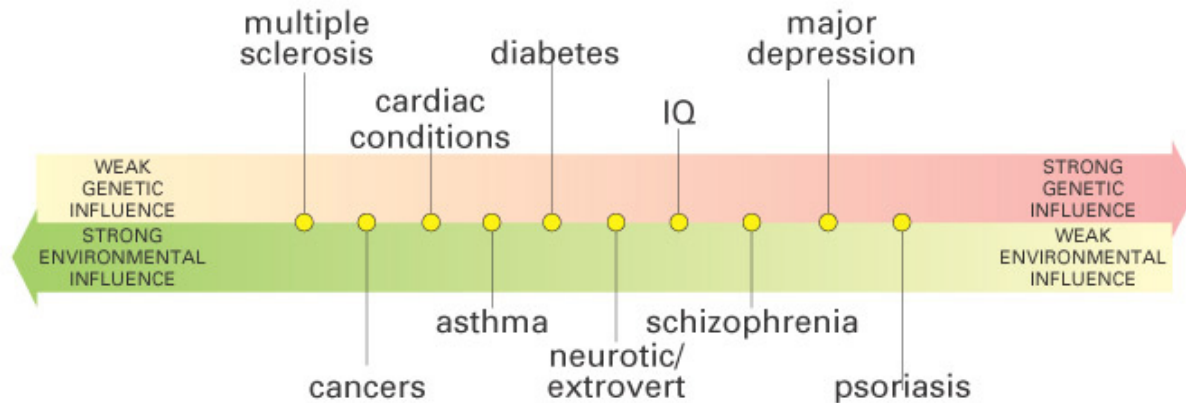


多基因遺傳模式

人類疾病與特徵受到遺傳與環境因素共同影響

多基因
Polygenic

單基因
Monogenic



遺傳疾病與再發風險(Recurrence Risk)

- 染色體異常(Chromosomal disorders)
- 粒線體基因突變(Mitochondrial mutations)
- 單基因突變(Monogenic disorders)
- 多基因遺傳模式疾病(Multifactorial or polygenic or complex diseases)
- 體細胞遺傳(Somatic cell genetics)

Online Mendelian Inheritance in Men (<http://www.ncbi.nlm.nih.gov/omim>)

OMIM Home

www.ncbi.nlm.nih.gov/omim

建議的網站 網頁快訊圖庫 從 IE 匯入

此網頁為 英文 您要翻譯網頁內容嗎? 翻譯 不需要 永遠不要翻譯英文

NCBI

OMIM Online Mendelian Inheritance in Man

Johns Hopkins University

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Search OMIM for [] Go Clear

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- Enter one or more search terms.
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NCBI is implementing changes to help you find current content in OMIM based on resources at NCBI, and then directing you to omim.org. Please be aware that you will leave NCBI to view OMIM records. Access to full records from NCBI (e.g. web, ftp, eutils) will no longer be supported.

OMIM® - Online Mendelian Inheritance in Man®

Welcome to OMIM®, Online Mendelian Inheritance in Man®. OMIM is a comprehensive, authoritative, and timely compendium of human genes and genetic phenotypes. The full-text, referenced overviews in OMIM contain information on all known mendelian disorders and over 12,000 genes. OMIM focuses on the relationship between phenotype and genotype. It is updated daily, and the entries contain copious links to other genetics resources.

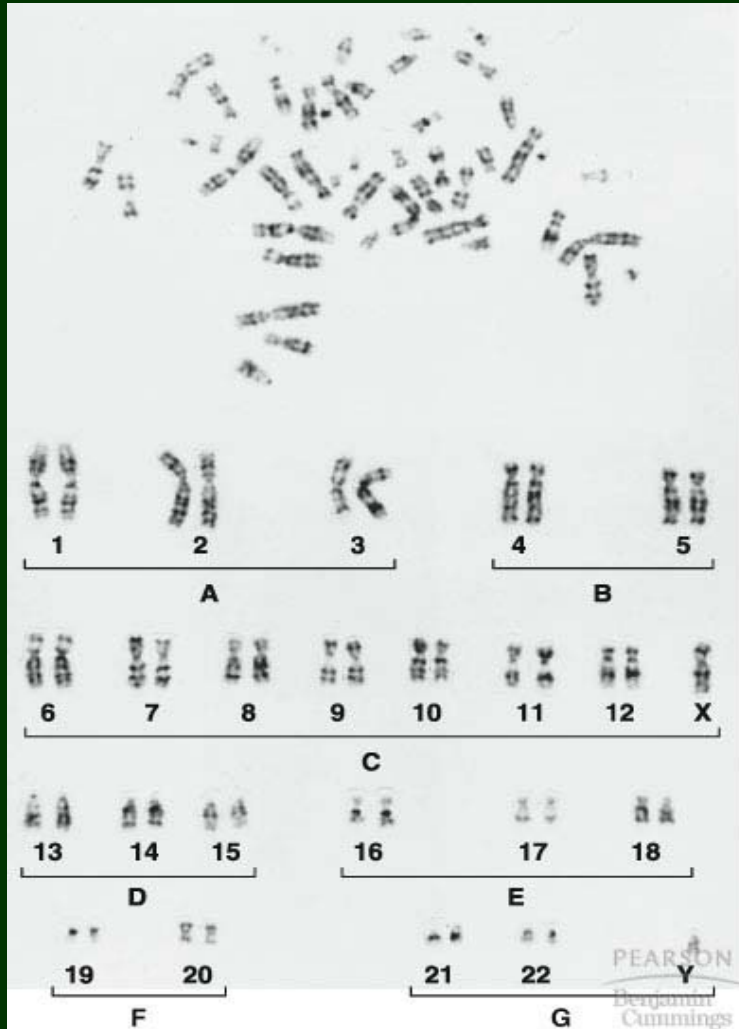
This database was initiated in the early 1960s by Dr. Victor A. McKusick as a catalog of mendelian traits and disorders, entitled Mendelian Inheritance in Man (MIM). Twelve book editions of MIM were published between 1966 and 1998. The online version, OMIM, was created in 1985 by a collaboration between the National Library of Medicine and the William H. Welch Medical Library at Johns Hopkins. It was made generally available on the internet starting in 1987. In 1995, OMIM was developed for the World Wide Web by NCBI, the National Center for Biotechnology Information.

OMIM is authored and edited at the McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University School of Medicine, under the direction of Dr. Ada Hamosh.

NLM's Profiles in Science -- The McKusick Papers [More...](#)

上午 09:17 2012/11/4

染色體核型 (karyotyping)



Thymidine
Clocemid
Staining

← **Karyotype**

Karyogram

染色體不分離

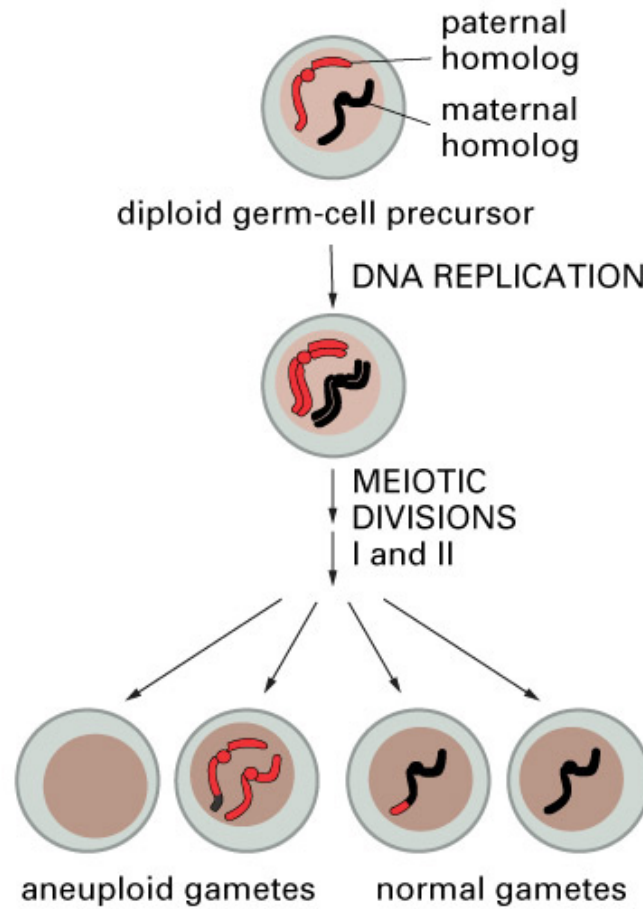


Figure 20-12 Essential Cell Biology, 2/e. (© 2004 Garland Science)

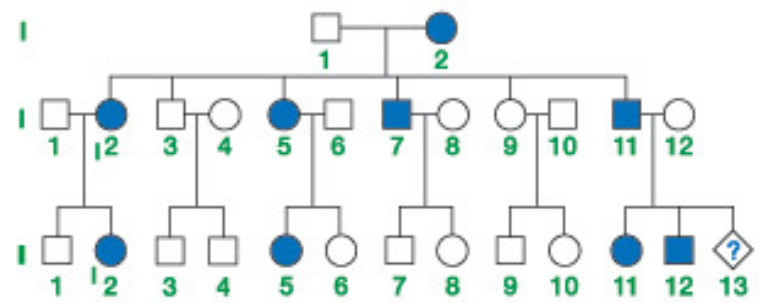
五種孟德爾單基因遺傳族譜型態 (Mendelian Pedigree Patterns)

- 體染色體顯性(autosomal dominant)
- 體染色體隱性(autosomal recessive)
- X體染色體性聯隱性(X-linked recessive)
- X體染色體性聯顯性(X-linked dominant)
- X體染色體性聯(Y-linked)



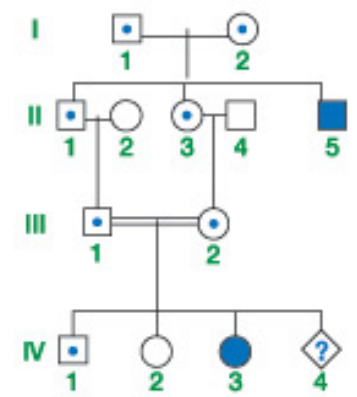
AD

(A)



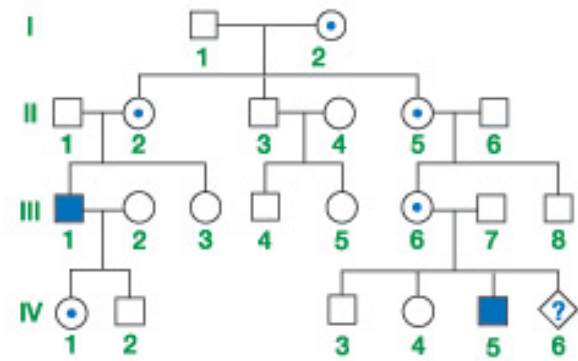
AR

(B)



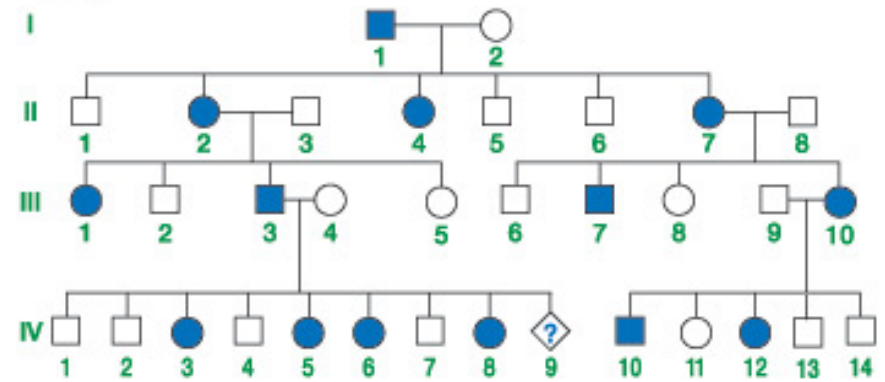
XR

(C)



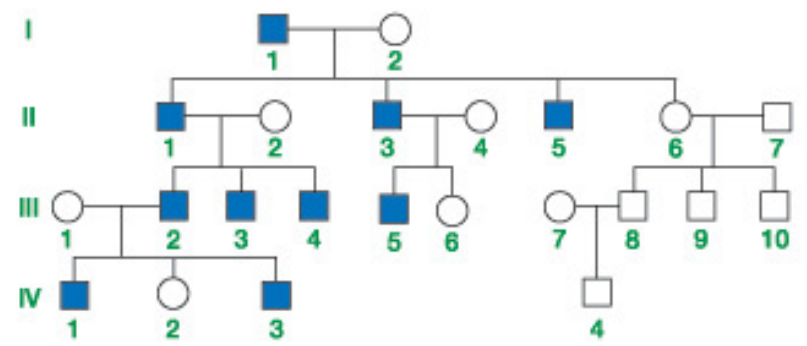
XD

(D)



(E)

Y

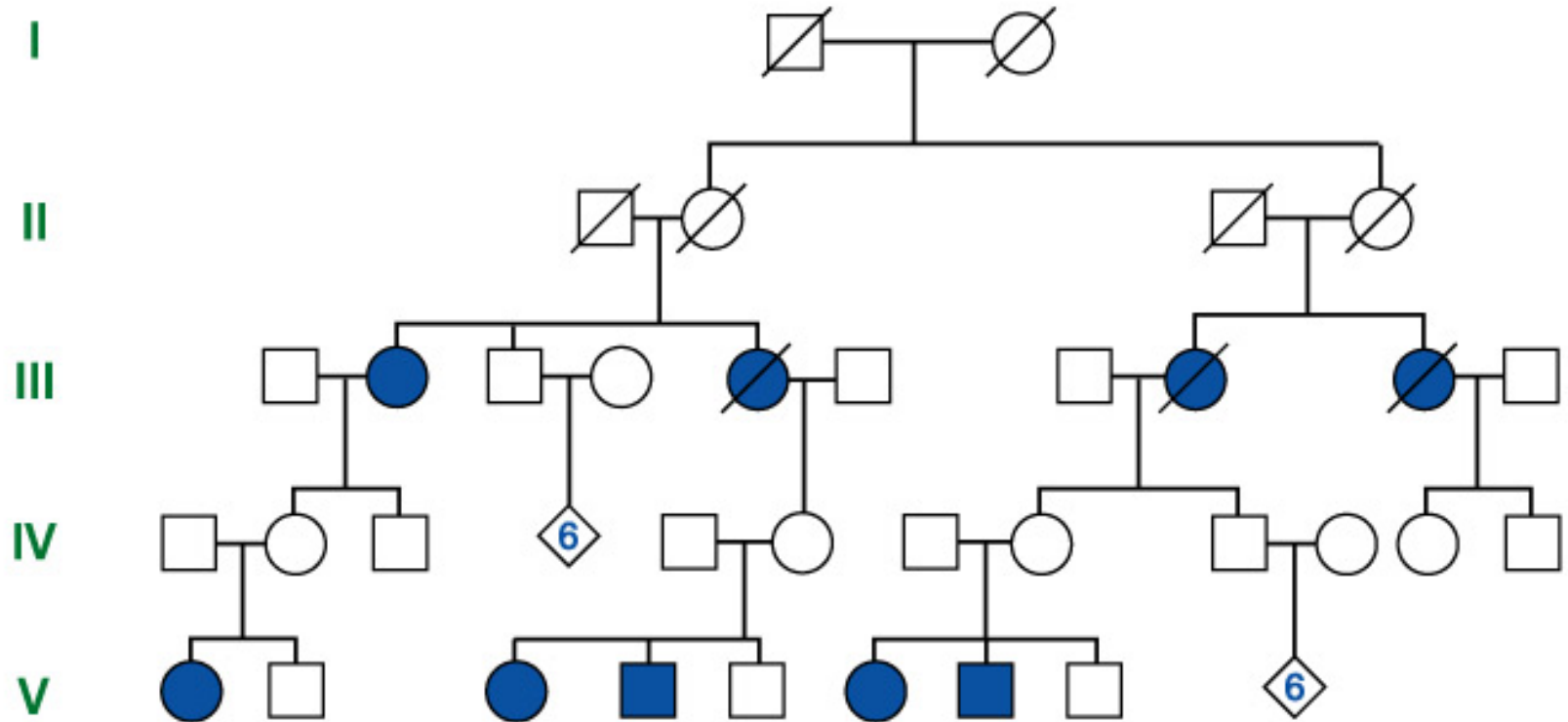


家族譜



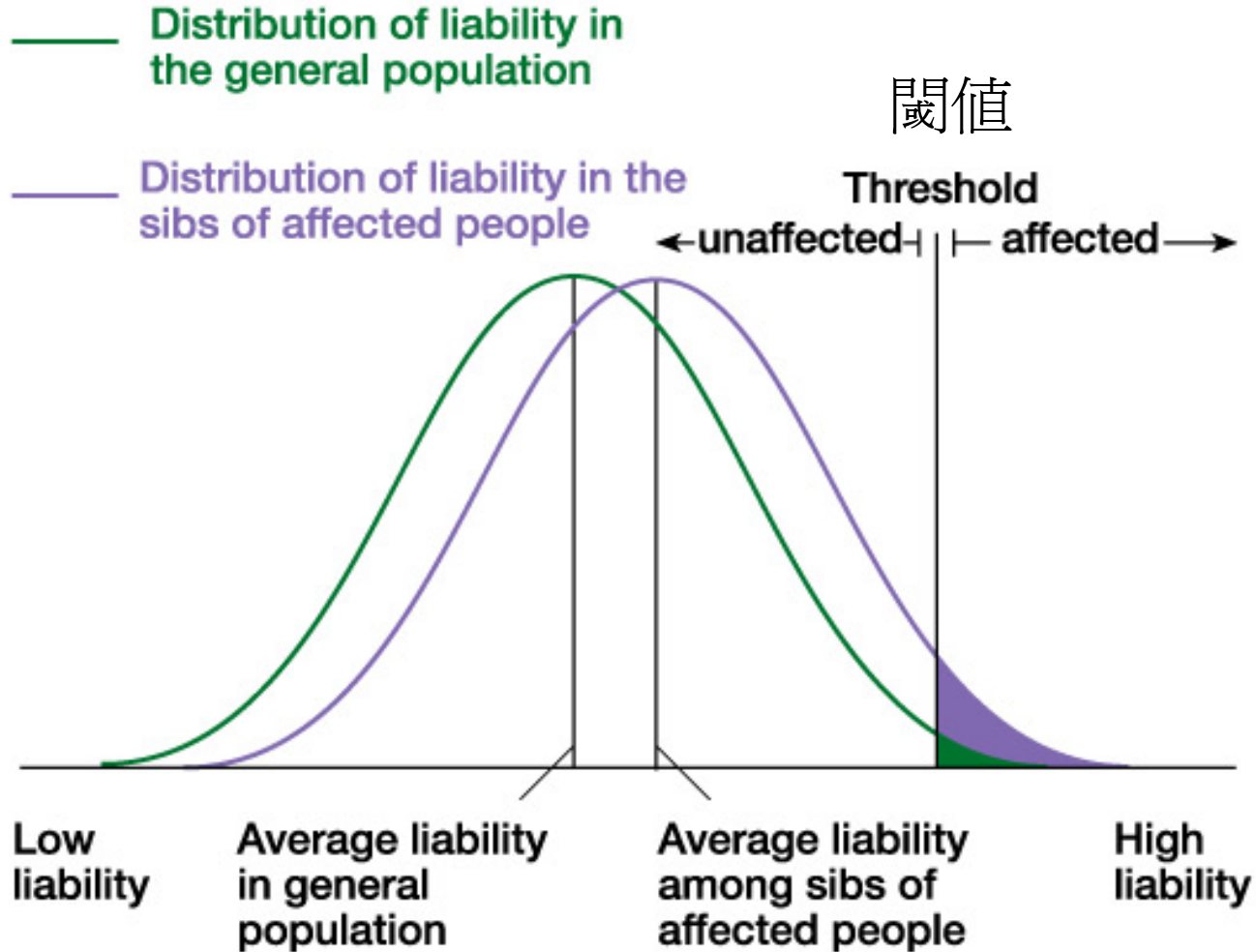
粒線體基因遺傳

(Mitochondrial Inheritance)





多基因遺傳模式



結論

- 孟德爾遺傳學是現代基因醫學的中心理論。
- DNA是遺傳的物質物質基礎。
- DNA數量與組成的變異是遺傳疾病主要來源。
- 幾乎大多數疾病都是基因與環境的交互作用所致。

attention!

*Ayers Rock, Central
Australia,
Balance of Life!*

