Mendelian Genetics

The laws of inheritance were derived by Gregor Mendel, a 19th century monk conducting hybridization experiments in garden peas (Pisum sativum).

Between 1856 and 1863, he cultivated and tested some 29,000 pea plants. From these experiments he deduced two generalizations which later became known as *Mendel's Laws of Heredity* or *Mendelian inheritance*.

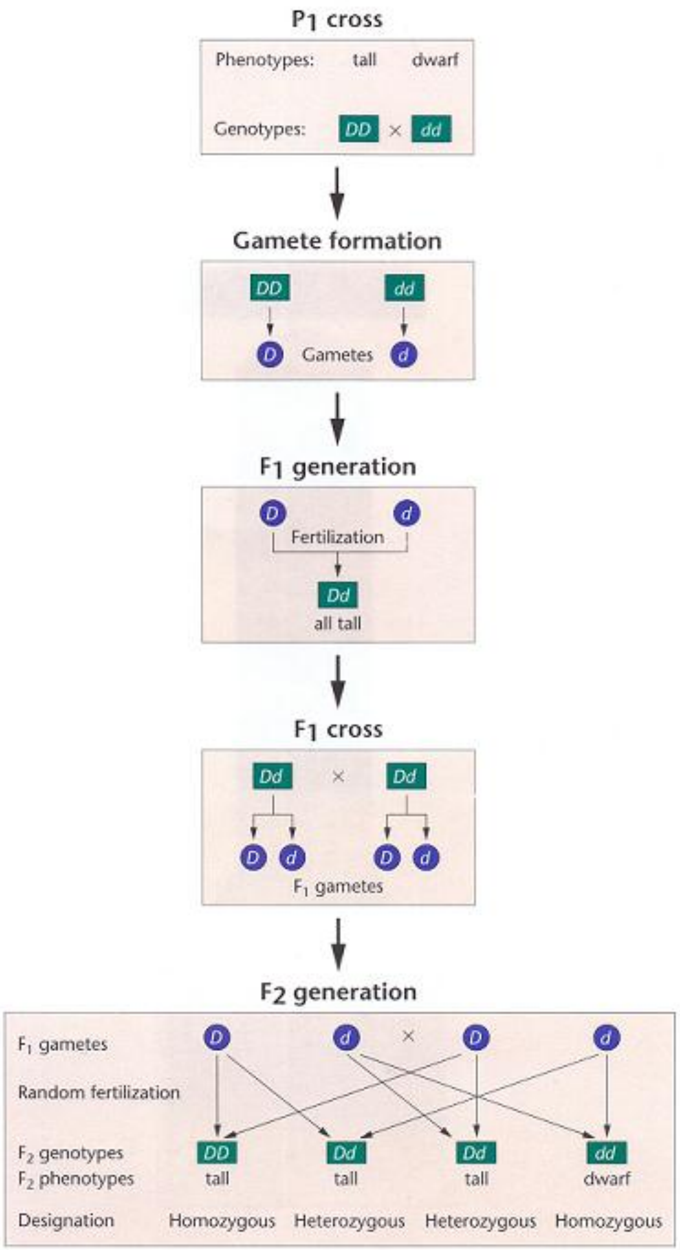
Mendel's conclusions were

largely ignored.

Fig. 3.1 Mendel's Monohybrid Results

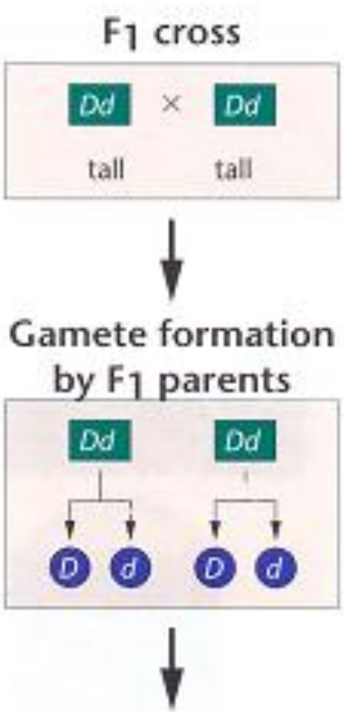
Character	Contrasting traits		F_1 results	F_2 results	F_2 ratio
Seeds	round/wrinkled		all round	5474 round 1850 wrinkled	2.96:1
	yellow/green		all yellow	6022 yellow 2001 green	3.01:1
	full/constricted		all full	882 full 299 constricted	2.95:1
Pods	green/yellow		all green	428 green 152 yellow	2.82:1
	axial/terminal		all axial	651 axial 207 terminal	3.14:1
Flowers	violet/white		all violet	705 violet 224 white	3.15:1
Stem	tall/dwarf		all tall	787 tall 277 dwarf	2.84:1

Fig. 3.2 Explanation of the Monohybrid Cross



- Gene – coding sequence of the DNA (it can be directly responsible for one trait or character).
- Allele - an alternate form of a gene. Usually there are two alleles for every gene
- Homozygous - when the two alleles are the same.
- Heterozygous - when the two alleles are different, in such cases the dominant allele is expressed.
- Dominant - a term applied to the trait (allele) that is expressed regardless of the second allele.
- Recessive - a term applied to a trait that is only expressed when the second allele is the same
- Phenotype - the physical expression of the allelic composition for the trait under study.
- Genotype - the allelic composition of an organism.

Fig. 3.3 Use of Punnett Square



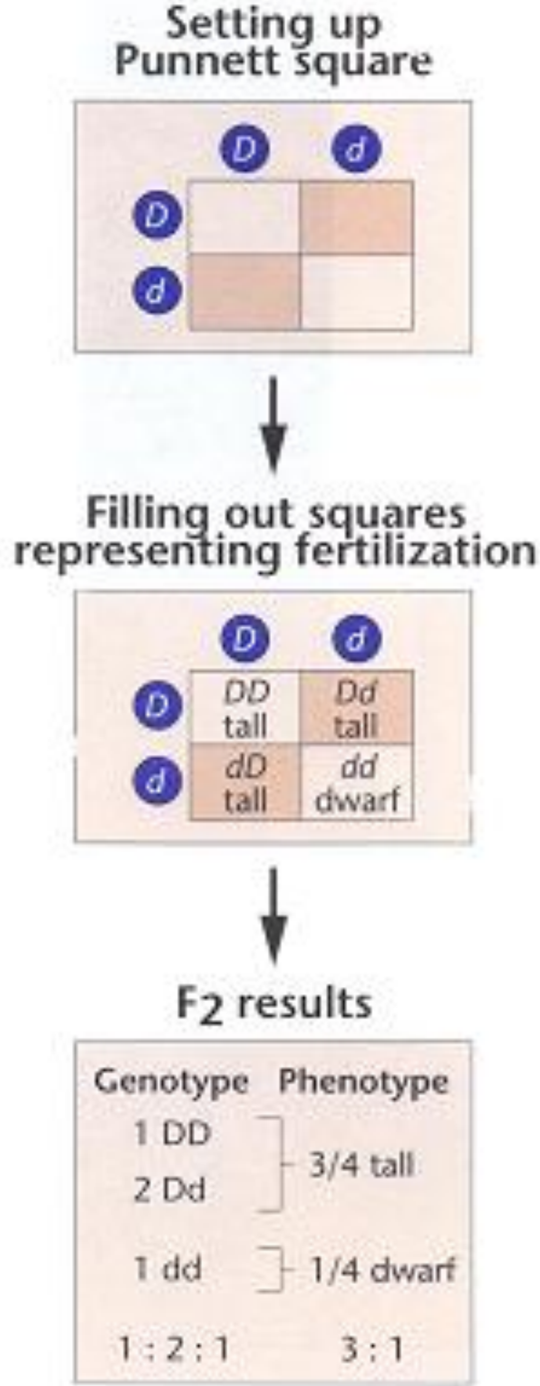


Fig. 3.4 Monohybrid Test Cross

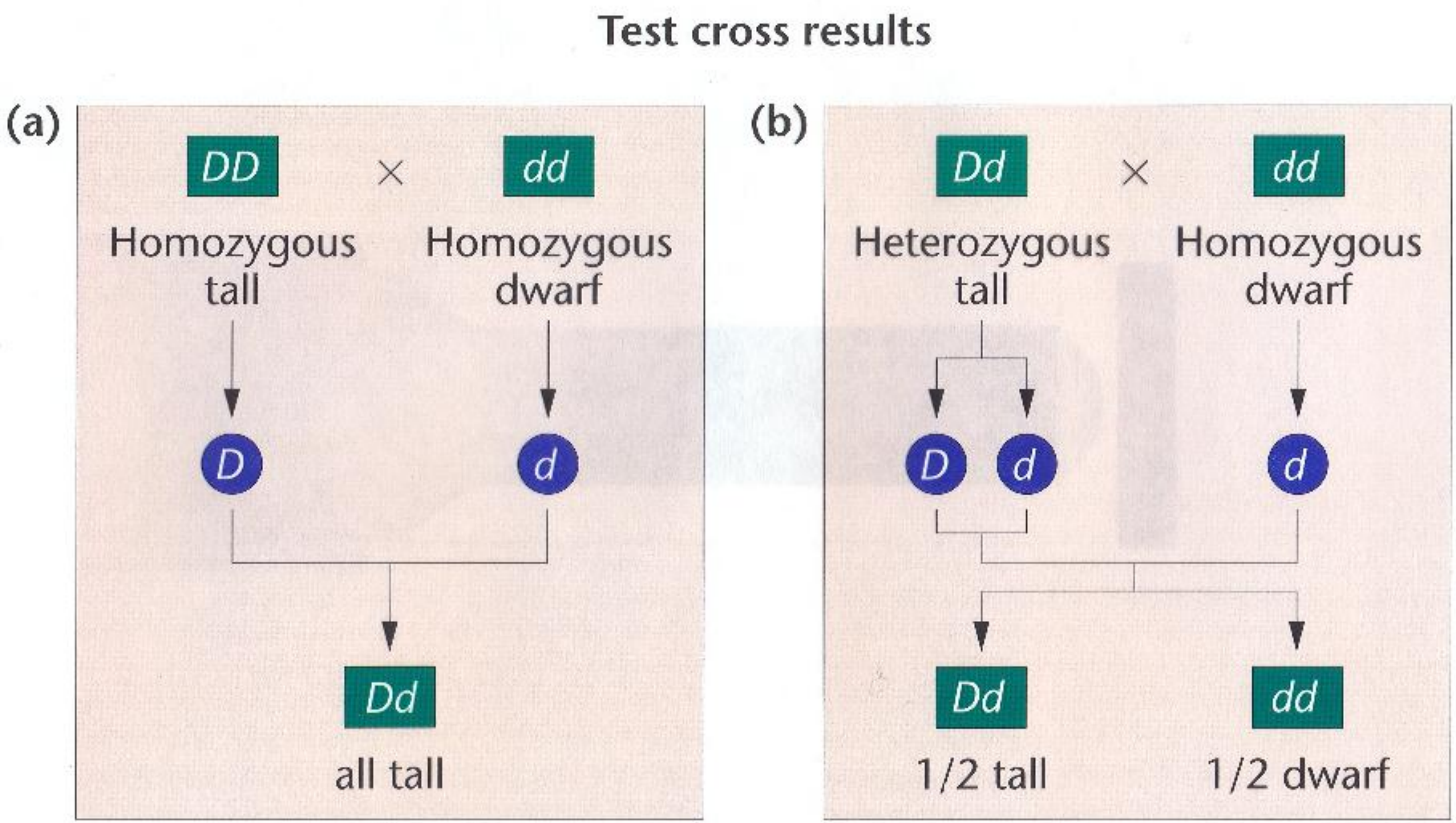


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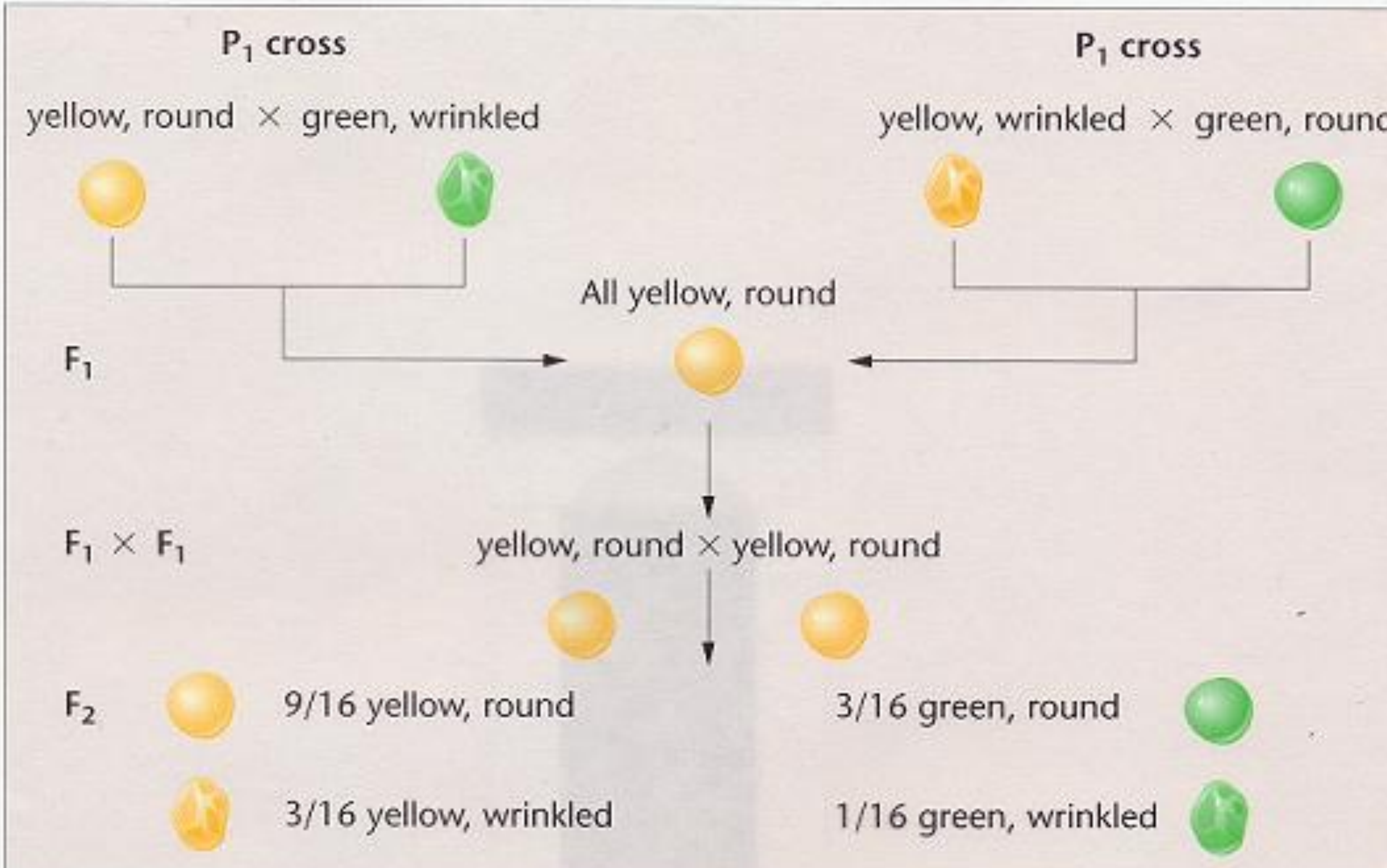
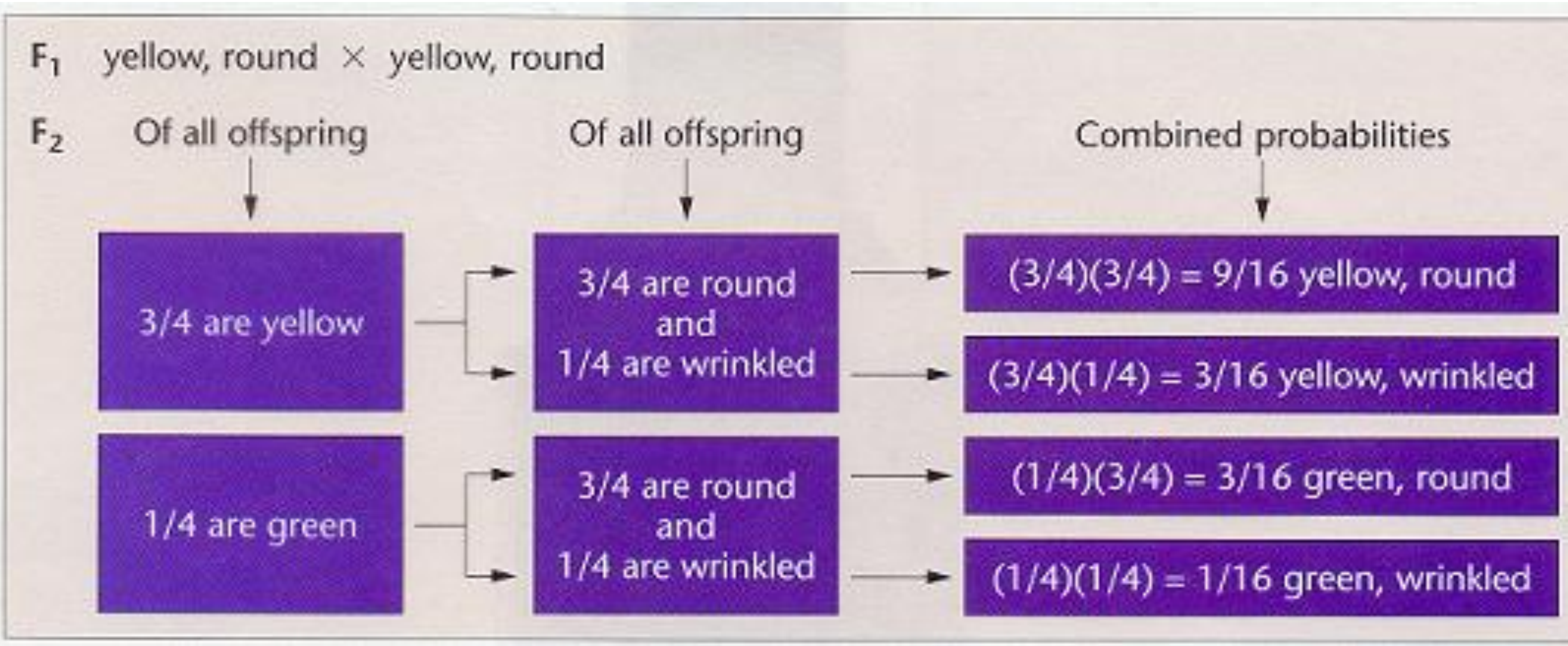
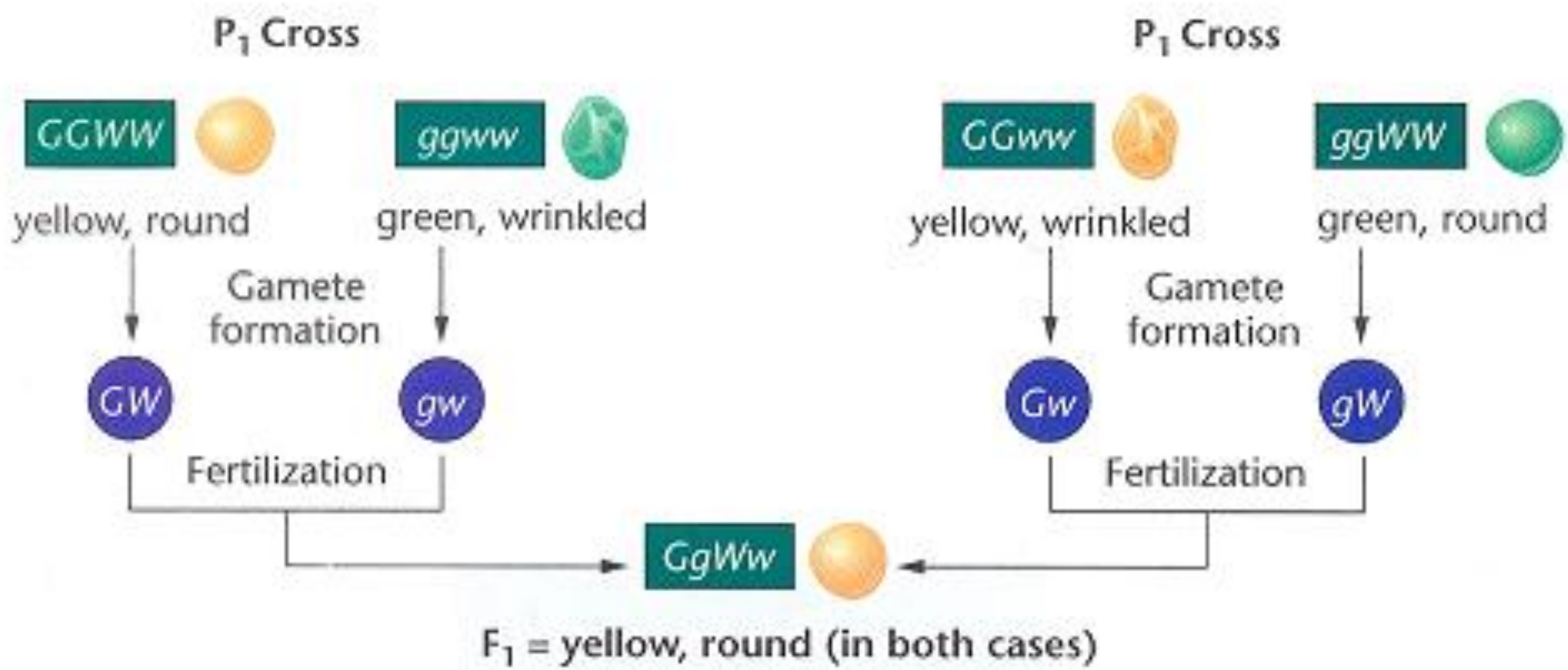
Fig. 3.5, 3.6 Mendel's Dihybrid Cross

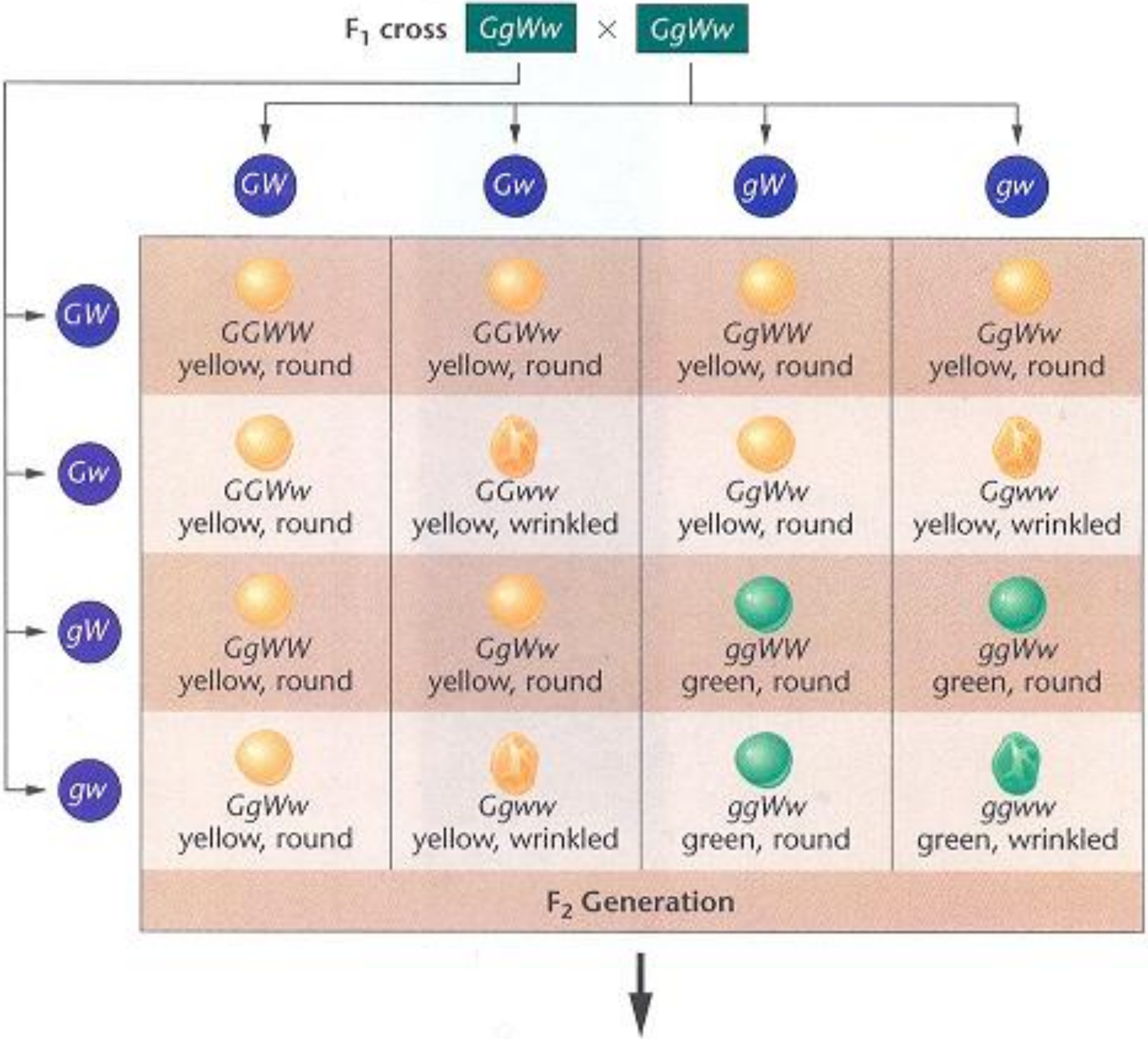




F_1 yellow, round × yellow, round
F_2
Of all offspring
Of all offspring
Combined probabilities
3/4 are yellow
1/4 are green
3/4 are round and 1/4 are wrinkled
3/4 are round and 1/4 are wrinkled
(3/4)(3/4) = 9/16 yellow, round
(3/4)(1/4) = 3/16 yellow, wrinkled
(1/4)(3/4) = 3/16 green, round
(1/4)(1/4) = 1/16 green, wrinkled

Fig 3.7 Punnett Square with Dihybrid Cross

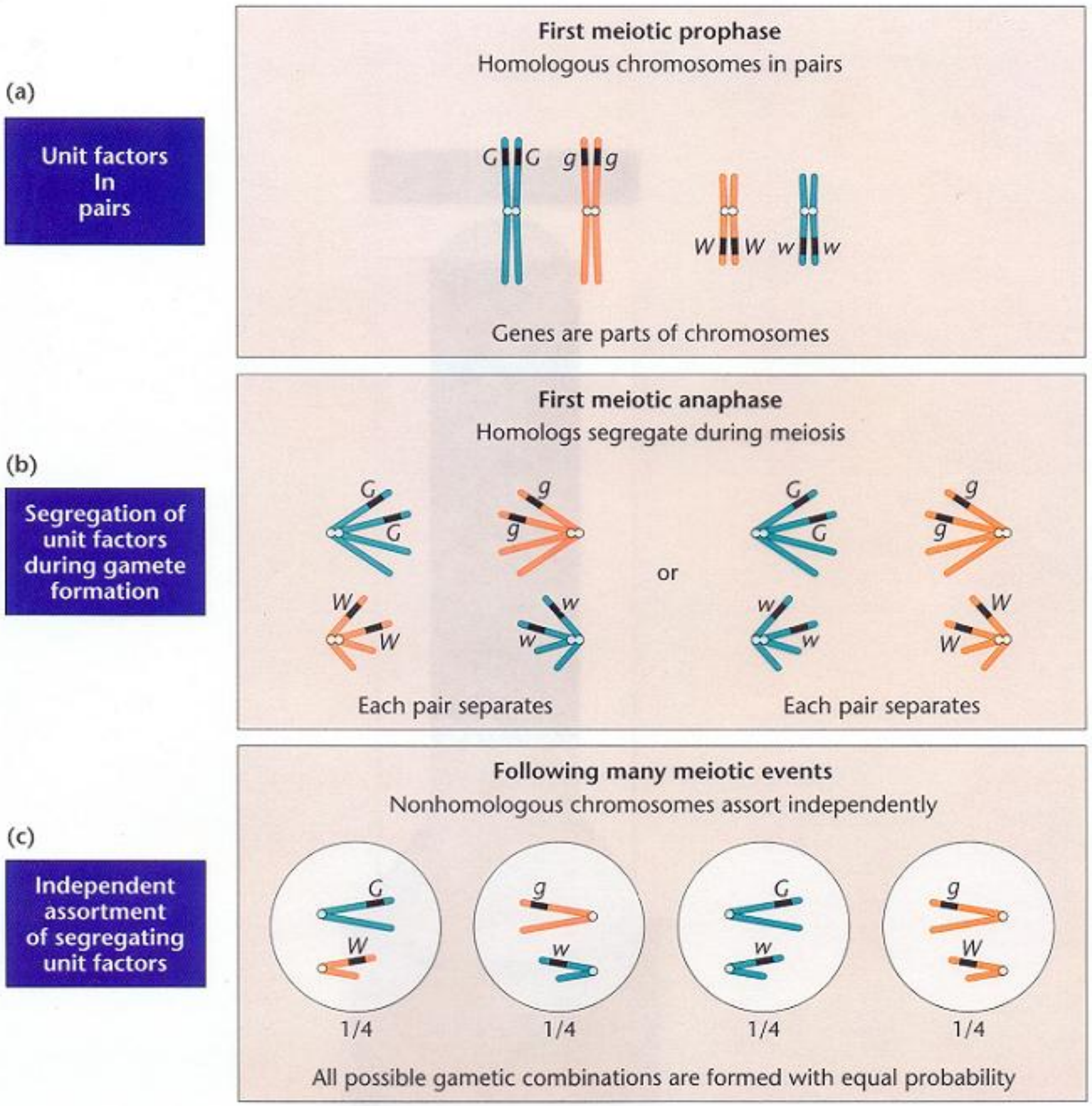




F1 cross GgWw × GgWw
GW
Gw
gW
gw
GW
GGWW yellow, round
GGWw yellow, round
GgWW yellow, round
GgWw yellow, round
Gw
GGWw yellow, round
GGww yellow, wrinkled
GgWw yellow, round
Ggww yellow, wrinkled
gW
GgWW yellow, round
GgWw yellow, round
ggWW green, round
ggWw green, round
gw
GgWw yellow, round
Ggww yellow, wrinkled
ggWw green, round
ggww green, wrinkled
F2 Generation

F_2 Genotypic ratio	F_2 Phenotypic ratio
1/16 *GGWW* 2/16 *GGWw* 2/16 *GgWW* 4/16 *GgWw*	9/16 yellow, round
1/16 *GGww* 2/16 *Ggww*	3/16 yellow, wrinkled
1/16 *ggWW* 2/16 *ggWw*	3/16 green, round
1/16 *gg ww*	1/16 green, wrinkled

Fig. 3.11 Correlation of Mendel's Postulates



MENDEL'S 3 LAWS OF INHERITANCE

- <u>Law of Dominance (Uniformity):</u> if two plants that differ in just one trait are crossed, then the resulting hybrids will be uniform in the chosen trait. Depending on the traits is the uniform feature either one of the parents' traits (a dominant-recessive pair of characteristics) or it is intermediate.

MENDEL’S 3 LAWS OF INHERITANCE

- Law of Segregation: It states that the individuals of the F2 generation are not uniform, but that the traits segregate. (The original traits did not “meld together”, they reappear.) Depending on a dominant-recessive crossing or an intermediate crossing are the resulting ratios 3:1 or 1:2:1. According to this principle hereditary traits are determined by discrete factors (now called genes) that occur in pairs, one of each pair being inherited from each parent.

MENDEL’S 3 LAWS OF INHERITANCE

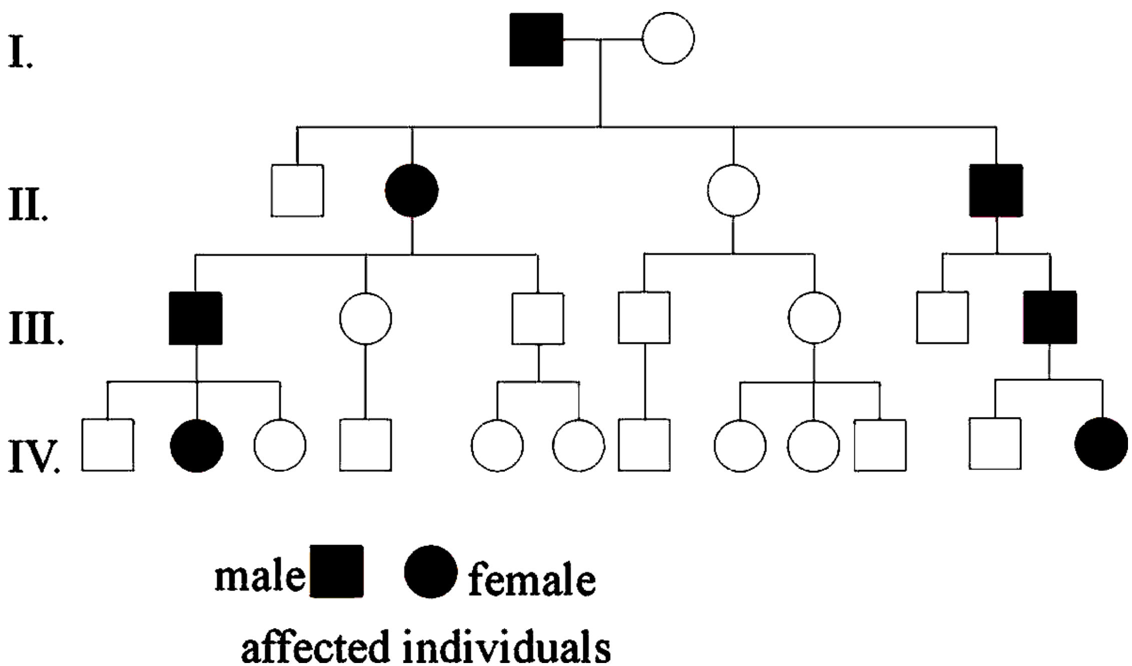
- Law of Independent Assortment (reciprocity): It says that every trait is inherited independently of the others and it thus covers the case that new combinations of genes can arise, which were not existing before. We know today that this principle is just valid in the case of genes that are not coupled, i.e. that are not located at the same chromosome.

A **Mendelian trait** is one that is controlled by a single locus and shows a simple Mendelian inheritance pattern. In such cases, a mutation in a single gene can cause a disease that is inherited according to Mendel's laws. Examples include sickle-cell anemia, Tay-Sachs disease, cystic fibrosis and xeroderma pigmentosum.

Monogenic diseases

- ***autosomal dominant diseases***
  - are caused by *gain-of-function* mutations
  - *vertical* type of inheritance
    - e.g. achondroplasia
    - Huntington's disease
    - osteogenesis imperfecta

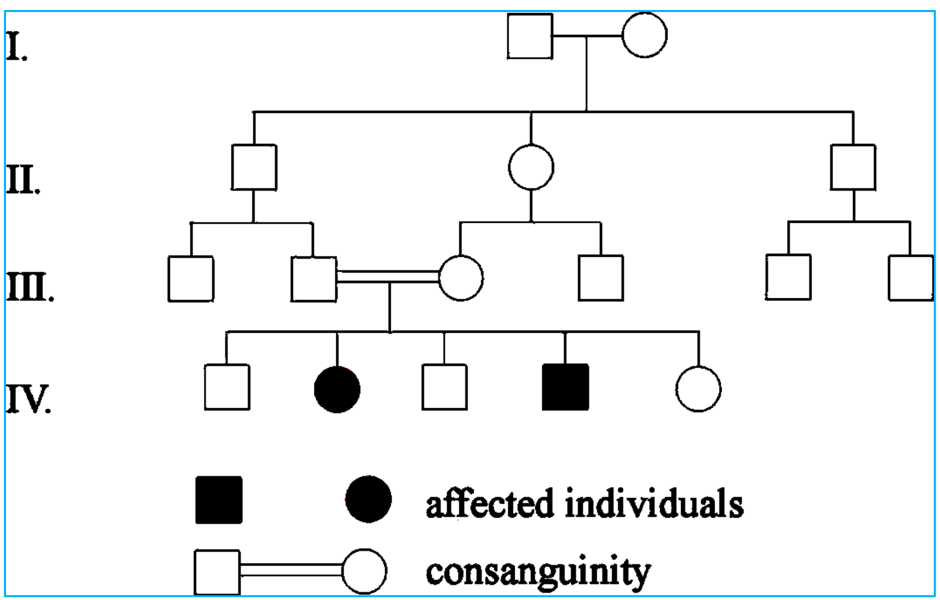
♂ Aa / ♀ aa		sperm cells	
		A	a
oocytes	a	Aa affected	aa healthy
	a	Aa affected	aa healthy

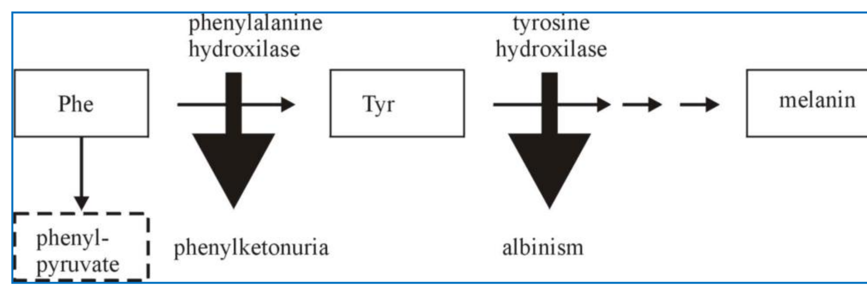


Autosomal recessive diseases

- are caused by *loss-of-function* mutations
- *horizontal* type of inheritance (Fig. 56.4., Fig. 56.5.)
- role of *consanguinity*
- e.g. :
  - cystic fibrosis
  - lysosomal storage diseases
  - phenylketonuria (Fig. 56.6.)
  - = phenylalanine hydroxylase deficiency
  - albinism
  - = tyrosine hydroxylase deficiency
  - sickle cell anaemia

♂ Aa / ♀ Aa		sperm cells	
		A	a
oocytes	A	AA healthy	Aa healthy
	a	Aa healthy	aa affected



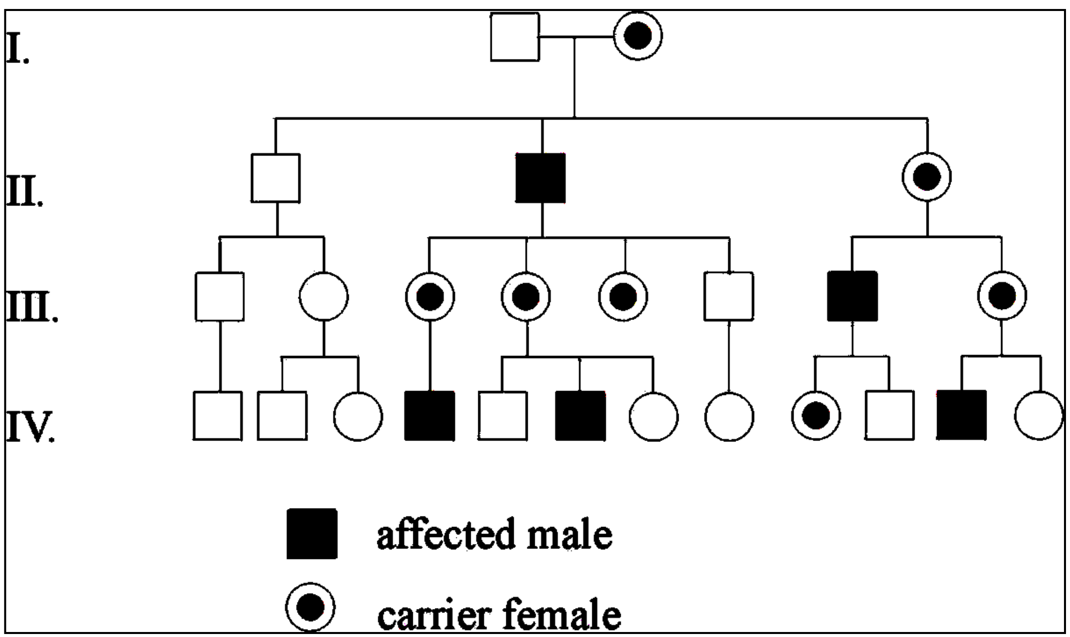


X-linked recessive diseases

- much more common in males (*hemizygotes*)
- no male-to-male inheritance
- e.g. :
  - haemophilia A
  - Duchenne's muscular dystrophy
  - colour blindness

♂ XY / ♀ X*X		sperm cells	
		X	Y
oocytes	X*	X*X carrier	X*Y affected
	X	XX healthy	XY healthy

♂ X*Y / ♀ XX		sperm cells	
		X*	Y
oocytes	X	X*X carrier	XY healthy
	X	X*X carrier	XY healthy



Polygenic diseases

- polygenic traits: *Gaussian distribution*
- *multifactorial* abnormalities:
  - inheritance
  - environmental factors
- *twin studies*
- e.g. :
  - diabetes mellitus
  - hypertension
  - schizophrenia