Carleton UNIVERSITY
Canada's Capital University

BIOL 1103
FOUNDATIONS OF BIOLOGY I

10. Hereditary genetics (Ch. 14 -15)

- Mendel and the birth of genetics
- Inheritance

Iain McKinnell – Dept. Biology

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10. Hereditary genetics (Ch. 14 -15)

- **Mendel and the birth of genetics (Ch. 14.1 – 14.2)**
- Inheritance

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Gregor Mendel

- Founder of genetics
- Augustinian monk (1822-1884)
- First to use scientific method to study inheritance

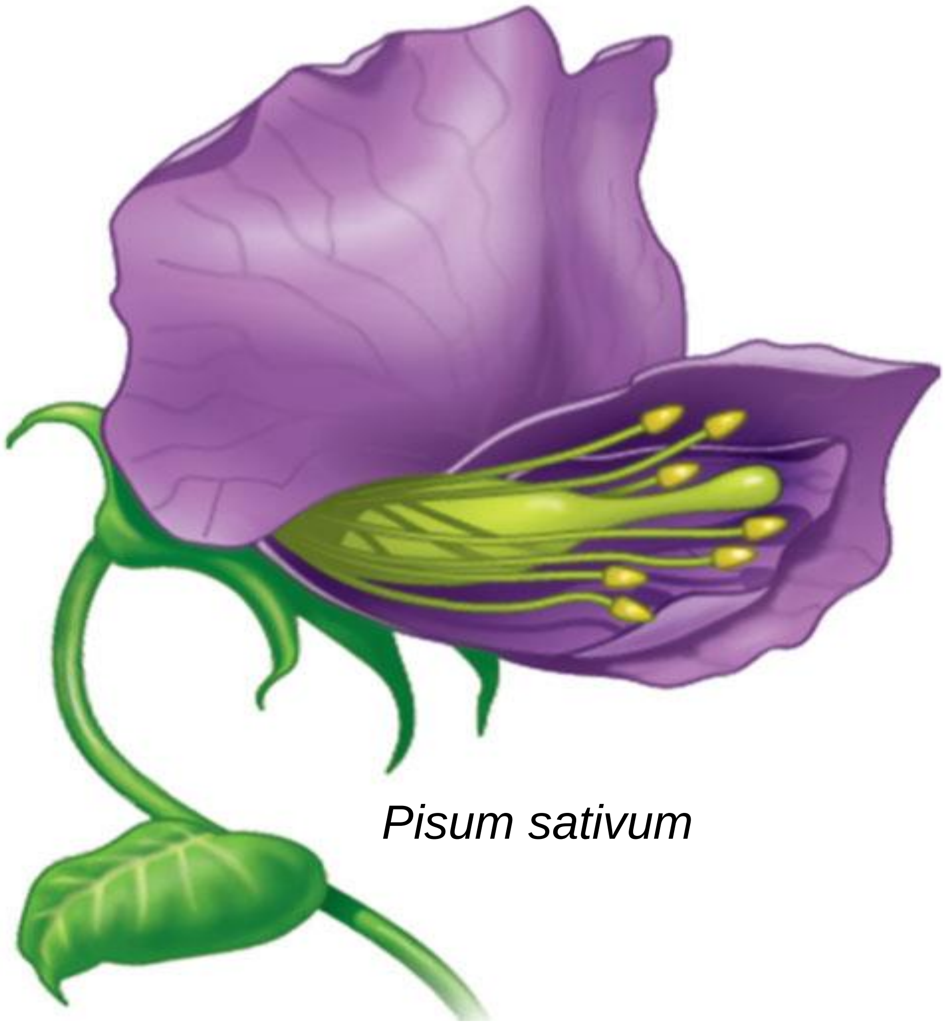
http://0.tqn.com/d/biology/1/S/l/e/3244238.jpg

Blending theory of inheritance

- Popular belief until about 1900
  - Hereditary traits blend evenly in offspring through mixing of parents' blood
- Does not explain some observations:
  - Extremes do not gradually disappear
  - Offspring sometimes have different traits than either parent

Mendel's experimental model – the garden pea

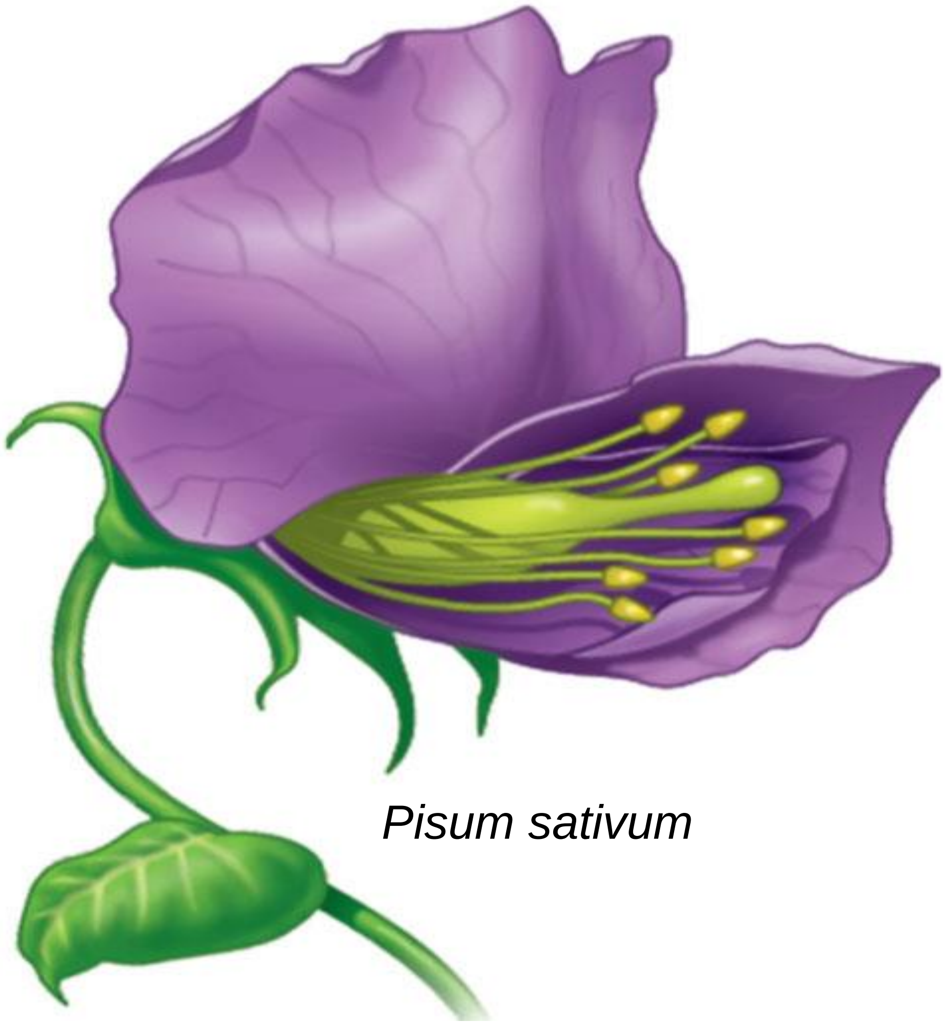
- In 1866, Mendel
  - correctly argued that parents pass on to their offspring discrete "heritable factors" and
  - stressed that the heritable factors (today called genes), retain their individuality generation after generation.
- A heritable feature that varies among individuals, such as flower color, is called a **character**.
- Each variant for a character, such as purple or white flowers, is a **trait**.



Pisum sativum

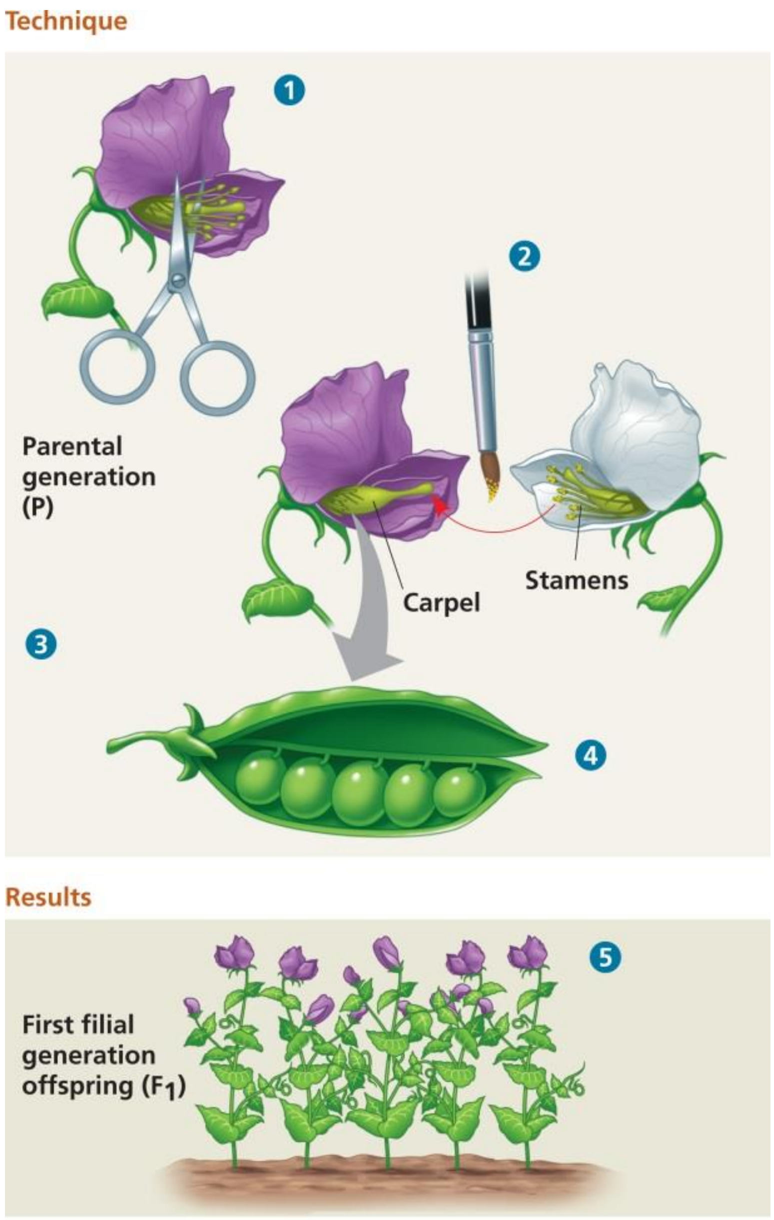
Mendel's experimental model – the garden pea

- **True-breeding varieties**
  - Self-fertilized plants (same trait each generation)



Pisum sativum

Mendel’s experimental model – the garden pea



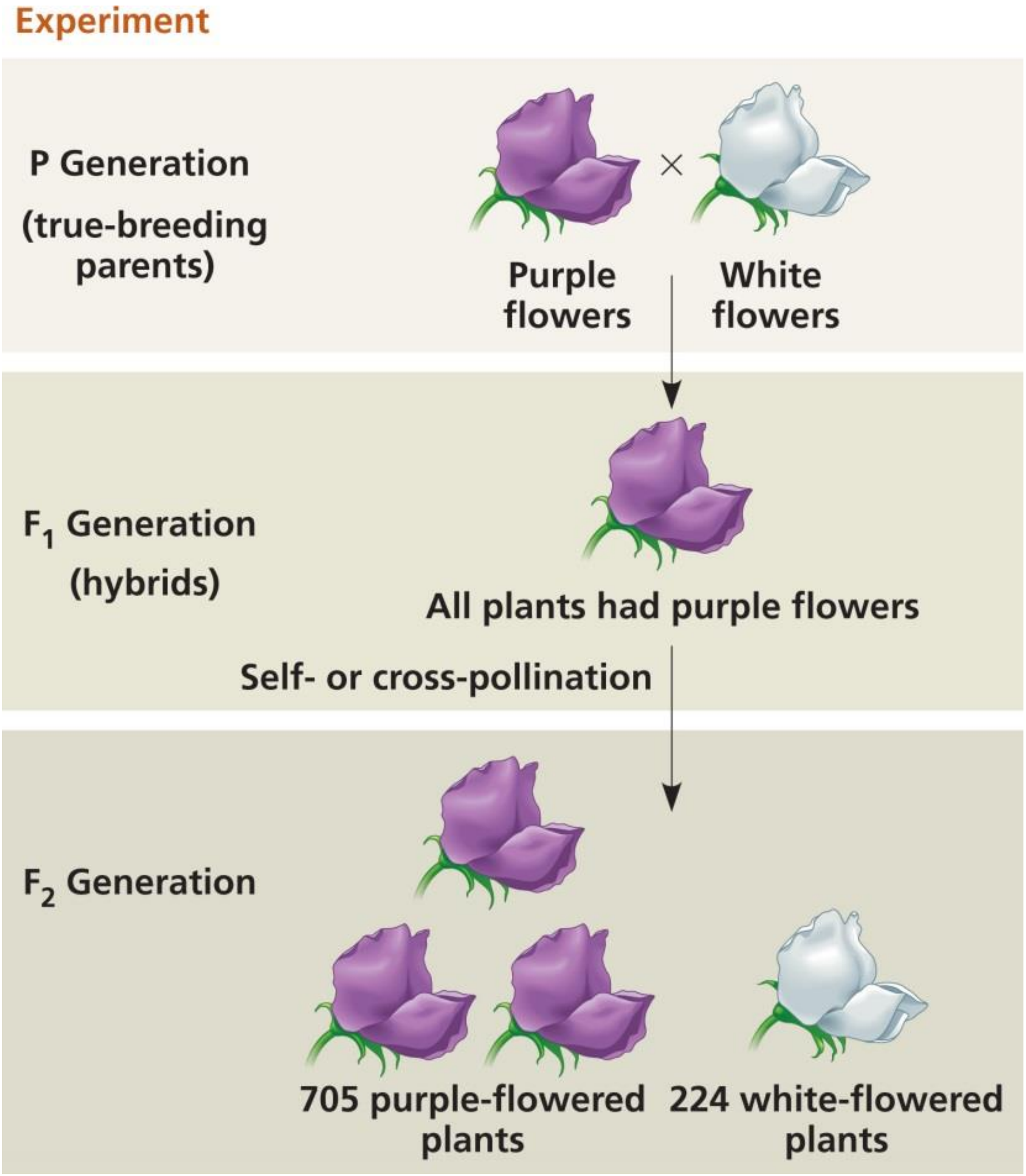
- **True-breeding varieties**
  - Self-fertilized plants (same trait each generation)
- Easy to **cross**
  - **Cross-pollination between parents**

Mendel's experimental model – the garden pea

- Offspring of two different varieties are **hybrids**.
- True-breeding parental plants are the
  P generation (parents).
- Hybrid offspring are the **F_1 generation (filial)**.
- A cross of F_1 plants produces an
  F_2 generation.

Mendel - inheritance of a single character

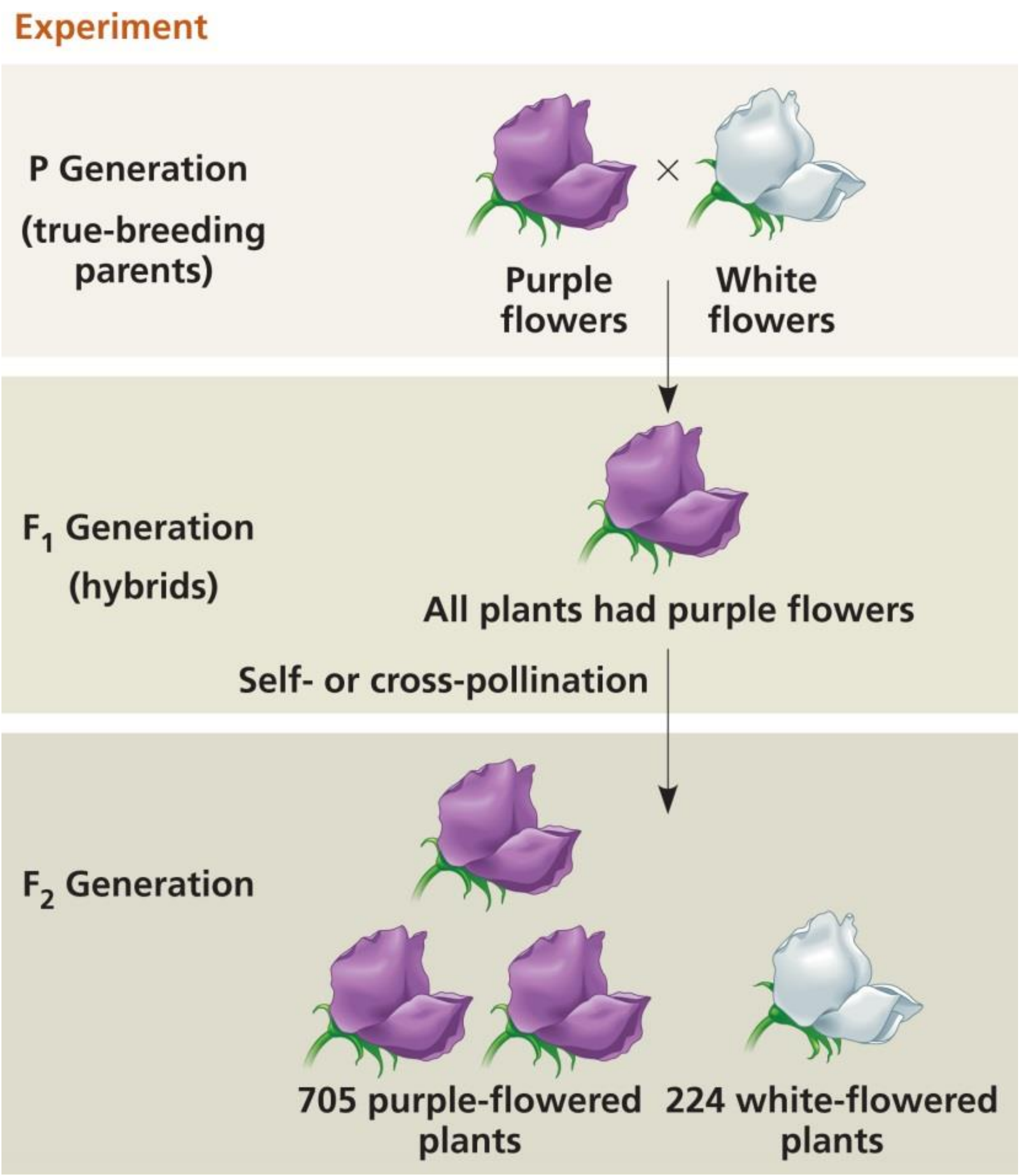
- A cross between two individuals differing in a single character is a **monohybrid cross**.
- Mendel crossed a plant with purple flowers and a plant with white flowers.
  - The F_1 generation produced all plants with purple flowers.
  - A cross of F_1 plants with each other produced an F_2 generation with ¾ purple and ¼ white flowers.



Mendel’s law of segregation describes the inheritance of a single character

- The all-purple F_1 generation did not produce light purple flowers, as predicted by the blending hypothesis.

- Mendel needed to explain why
  - white color seemed to disappear in the F_1 generation and
  - white color reappeared in one quarter of the F_2 offspring.



Mendel’s law of segregation describes the inheritance of a single character

- Mendel observed the same patterns of inheritance for six other pea plant characters.

Table 14.1 The Results of Mendel’s F_1 Crosses for Seven Characters in Pea Plants

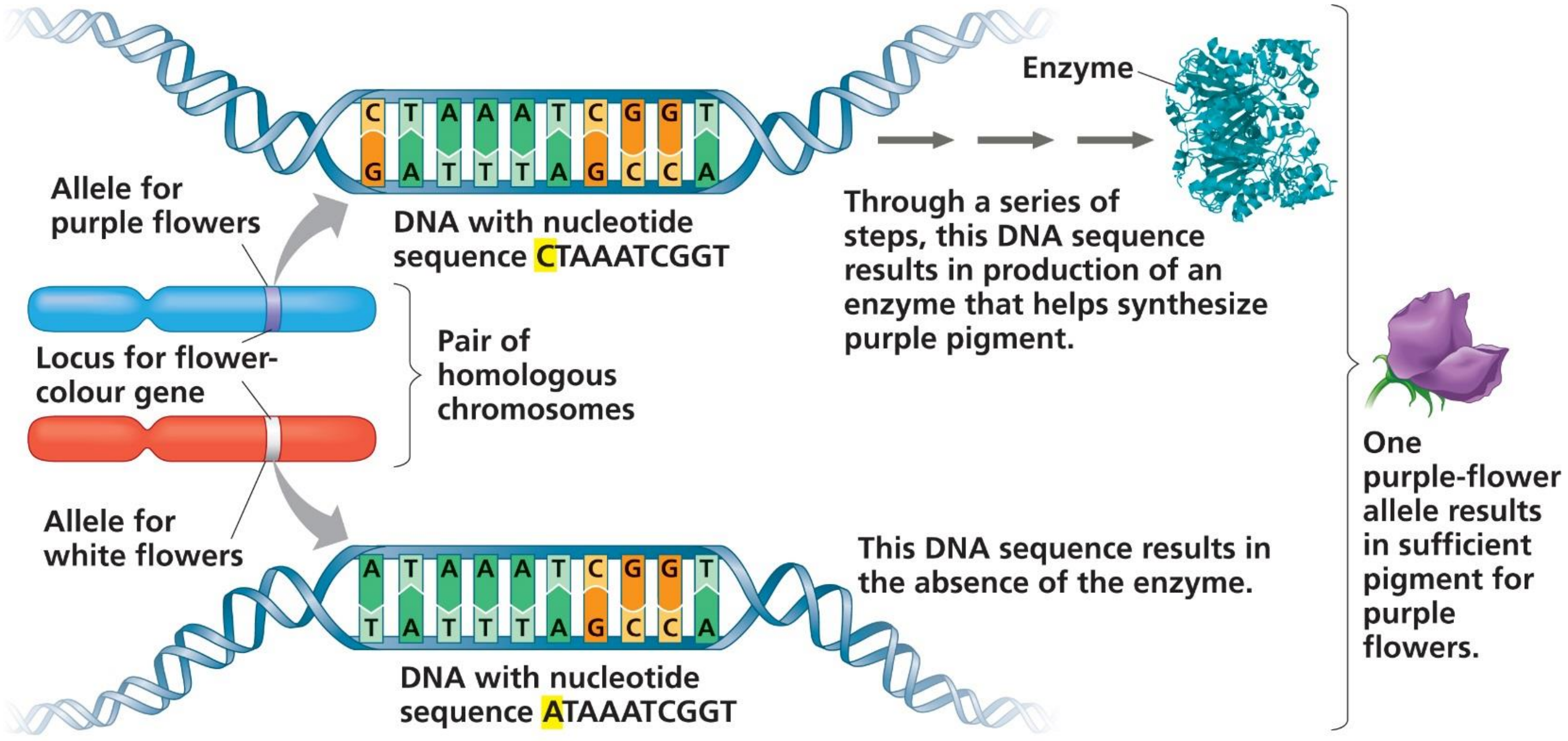
Character	Dominant Trait	×	Recessive Trait	F_2 Generation Dominant: Recessive	Ratio
Flower colour	Purple 	×	White 	705:224	3.15:1
Seed colour	Yellow 	×	Green 	6022:2001	3.01:1
Seed shape	Round 	×	Wrinkled 	5474:1850	2.96:1
Pod shape	Inflated 	×	Constricted 	882:299	2.95:1

Table 14.1 The Results of Mendel’s F_1 Crosses for Seven Characters in Pea Plants

Character	Dominant Trait	×	Recessive Trait	F_2 Generation Dominant: Recessive	Ratio
Pod colour	Green 	×	Yellow 	428:152	2.82:1
Flower position	Axial 	×	Terminal 	651:207	3.14:1
Stem length	Tall 	×	Dwarf 	787:277	2.84:1

Mendel's law of segregation describes the inheritance of a single character

- Mendel developed four hypotheses:
  1. **Alleles** are alternative versions of genes that account for variations in inherited characters.
  2. For each characteristic, an organism inherits two alleles, one from each parent. The alleles can be the same or different.
     - A **homozygous** genotype has identical alleles.
     - A **heterozygous** genotype has two different alleles.



Mendel's law of segregation describes the inheritance of a single character

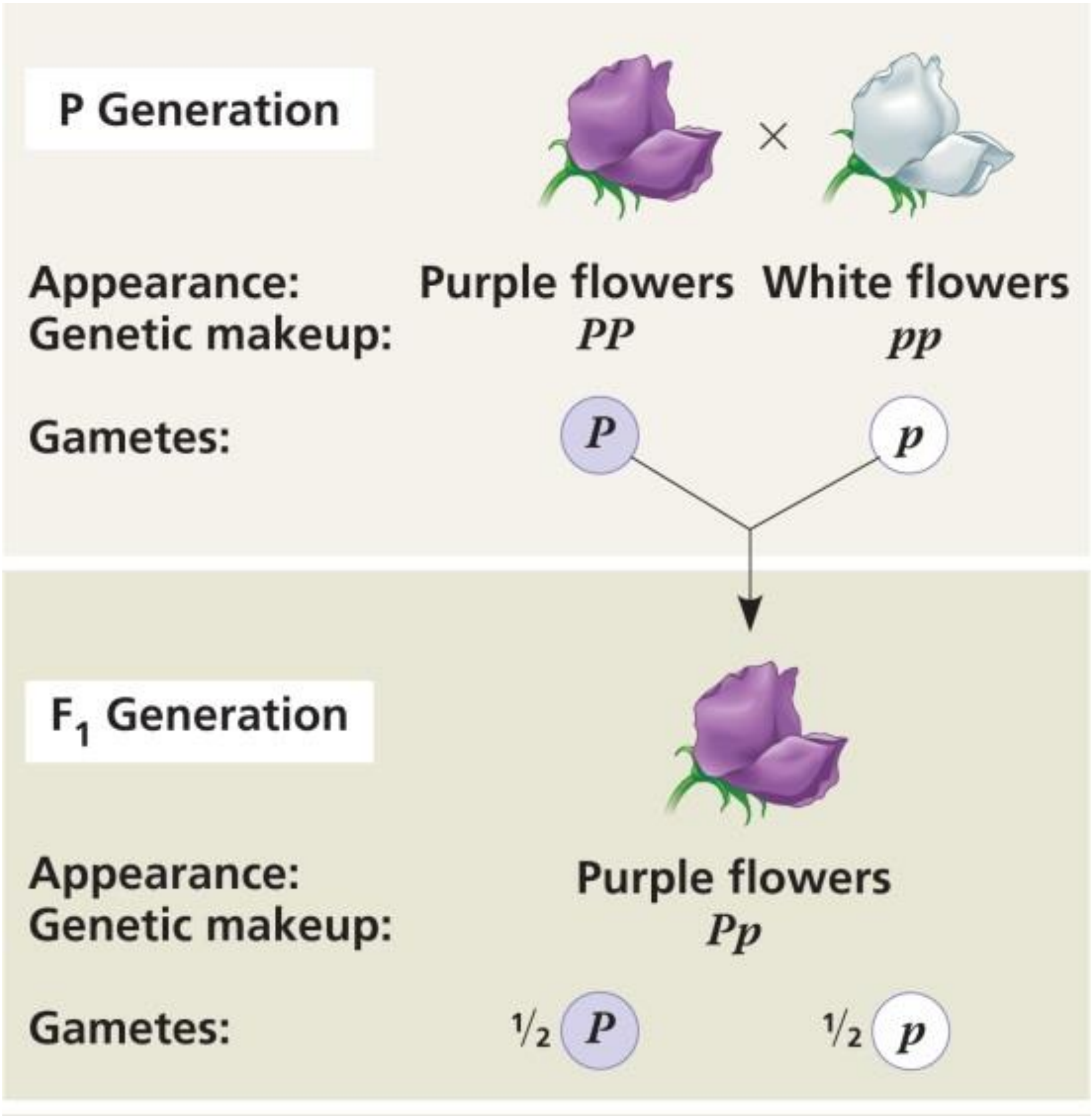
3. If the alleles of an inherited pair differ, then one determines the organism's appearance and is called the **dominant** allele. The other has no noticeable effect on the organism's appearance and is called the **recessive** allele.
    - The **phenotype** is the appearance or expression of a trait.
    - The **genotype** is the genetic makeup of a trait.
    - The same phenotype may be determined by more than one genotype.

Mendel’s law of segregation describes the inheritance of a single character

4. A sperm or egg carries only one allele for each inherited character because allele pairs separate (segregate) from each other during the production of gametes. This statement is called the **law of segregation**.

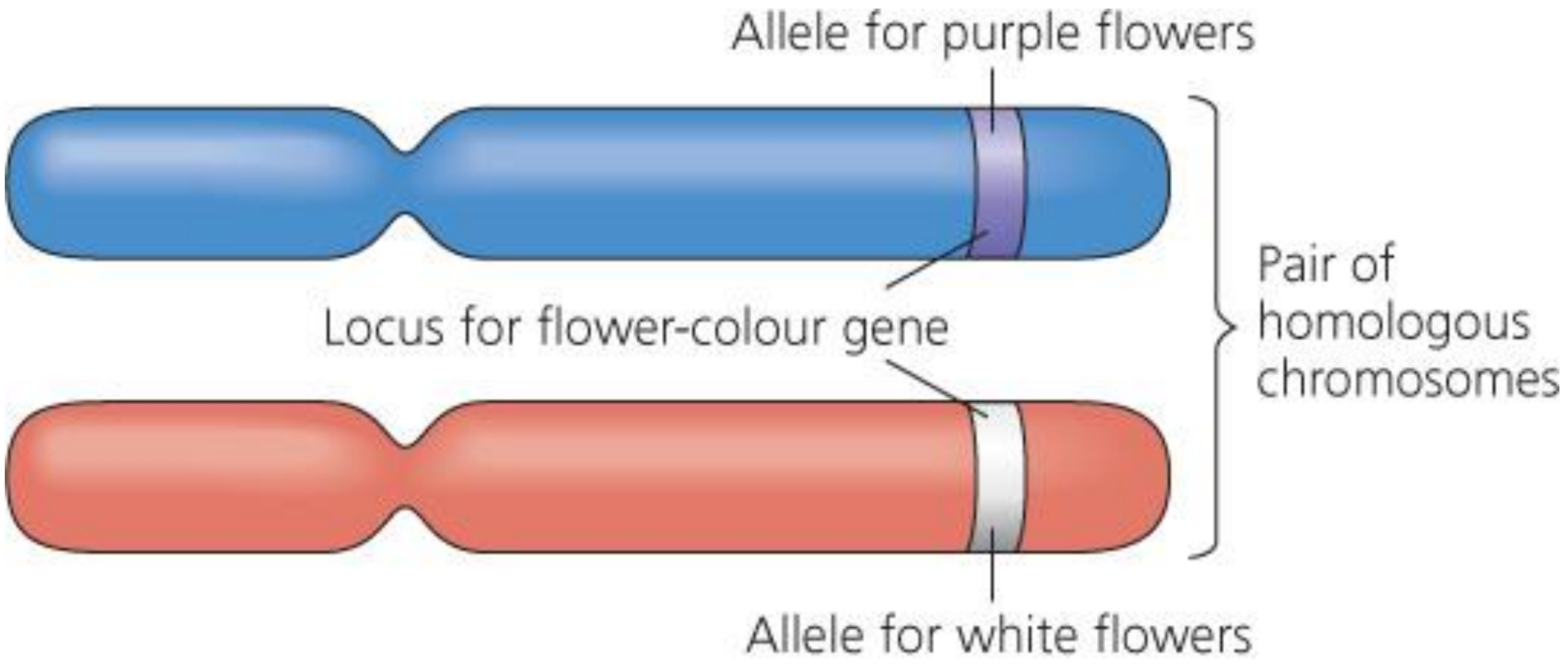
Mendel’s law of segregation describes the inheritance of a single character

- Mendel’s hypotheses explain the 3:1 ratio in the F_2 generation.
  - The F_1 hybrids all have a *Pp* genotype.



Homologous chromosomes bear the alleles for each character

- A **locus** (plural, *loci*) is the specific location of a gene along a chromosome.
- For a pair of homologous chromosomes, alleles of a gene reside at the same locus.
  - **Homozygous** individuals have the same allele on both homologues.
  - **Heterozygous** individuals have a different allele on each homologue.

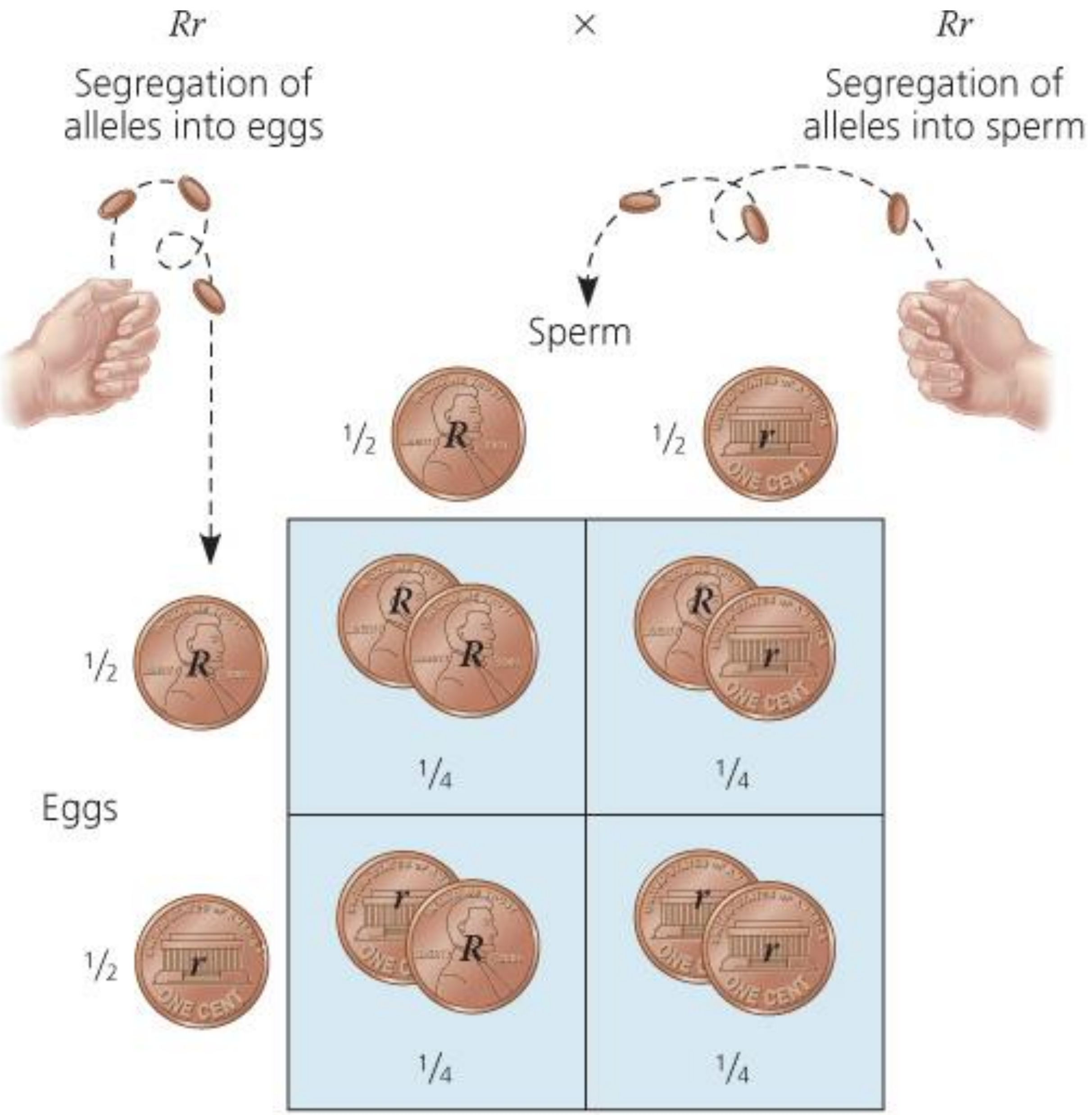


Product Rule in Probability

- Probability of two independent events occurring in succession
  - Individual probabilities multiplied

- Coin flip probabilities
  - Heads = ½
  - Tails = ½

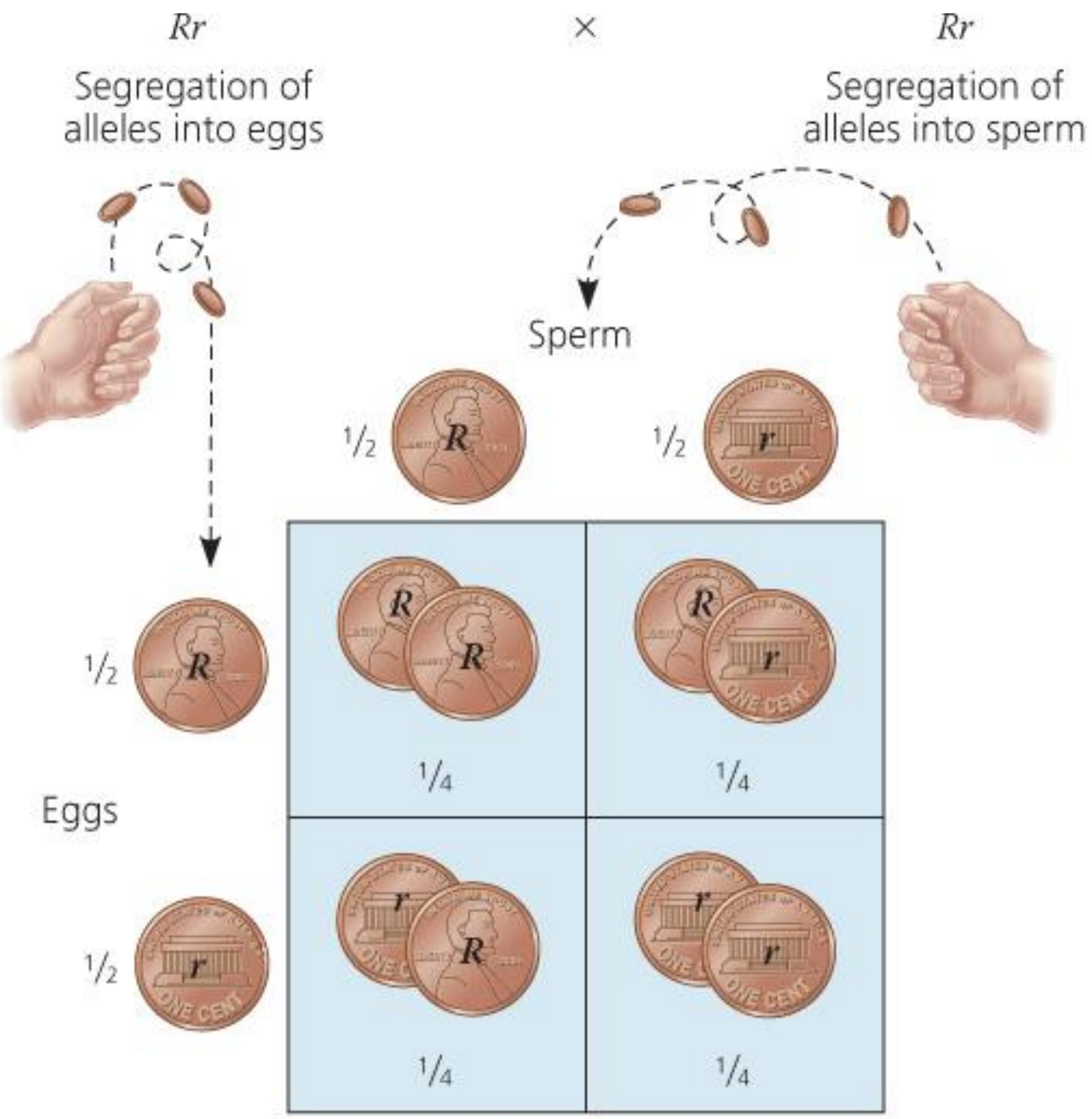
  - Two heads = ½ × ½ = ¼
  - Two tails = ½ × ½ = ¼



Sum Rule in Probability

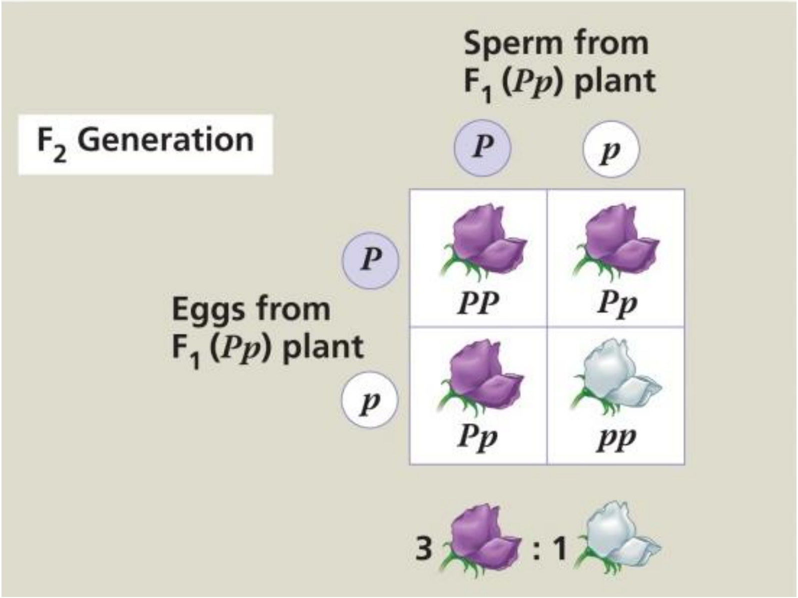
- Probability of two different events producing the same outcome
  - Individual probabilities added

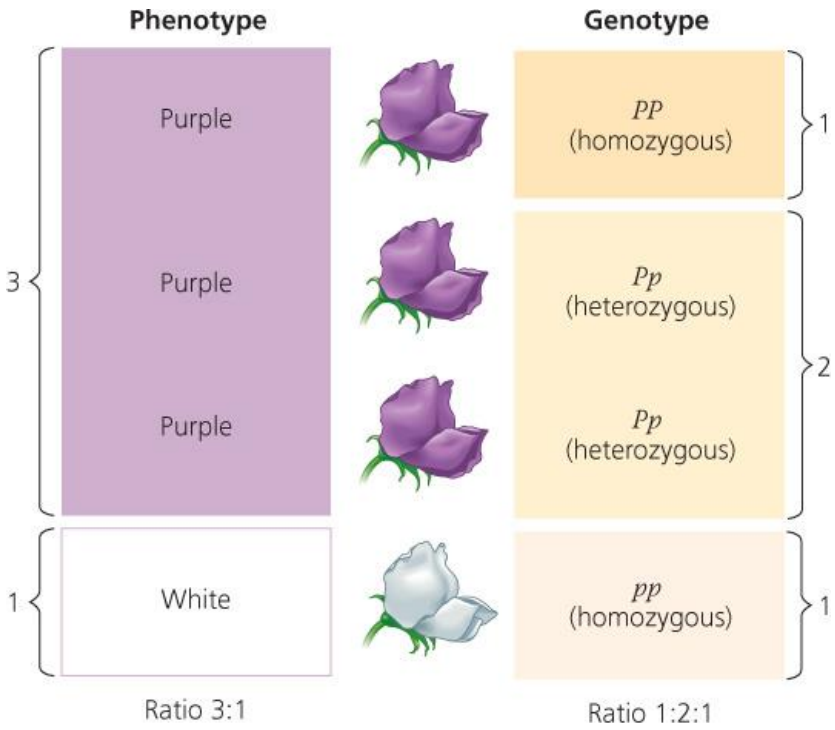
- Probability of a heads and a tails in two tosses:
  - First possibility: heads then tails
    - Heads = ½, Tails = ½ (½ × ½ = ¼)
  - Second possibility: tails then heads
    - Tails = ½, Heads = ½ (½ × ½ = ¼)
  - Total probability: ¼ + ¼ = ½



Probability in Mendel's Crosses

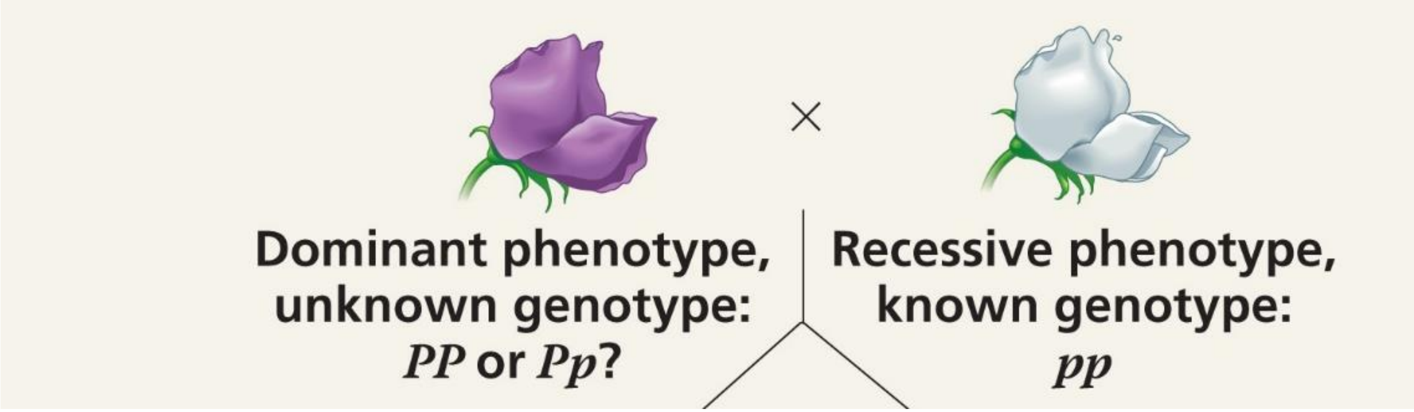
- Heterozygous cross ($Pp \times Pp$)
  - Genotype probabilities
    - *PP* zygote = ½ × ½ = ¼
    - *pp* zygote = ½ × ½ = ¼
    - *Pp* zygote = ¼ + ¼ = ½
  - Phenotype probabilities
    - Purple flowers = *PP* + *Pp* = ¼ + ½ = ¾
    - White flowers = *pp* = ¼





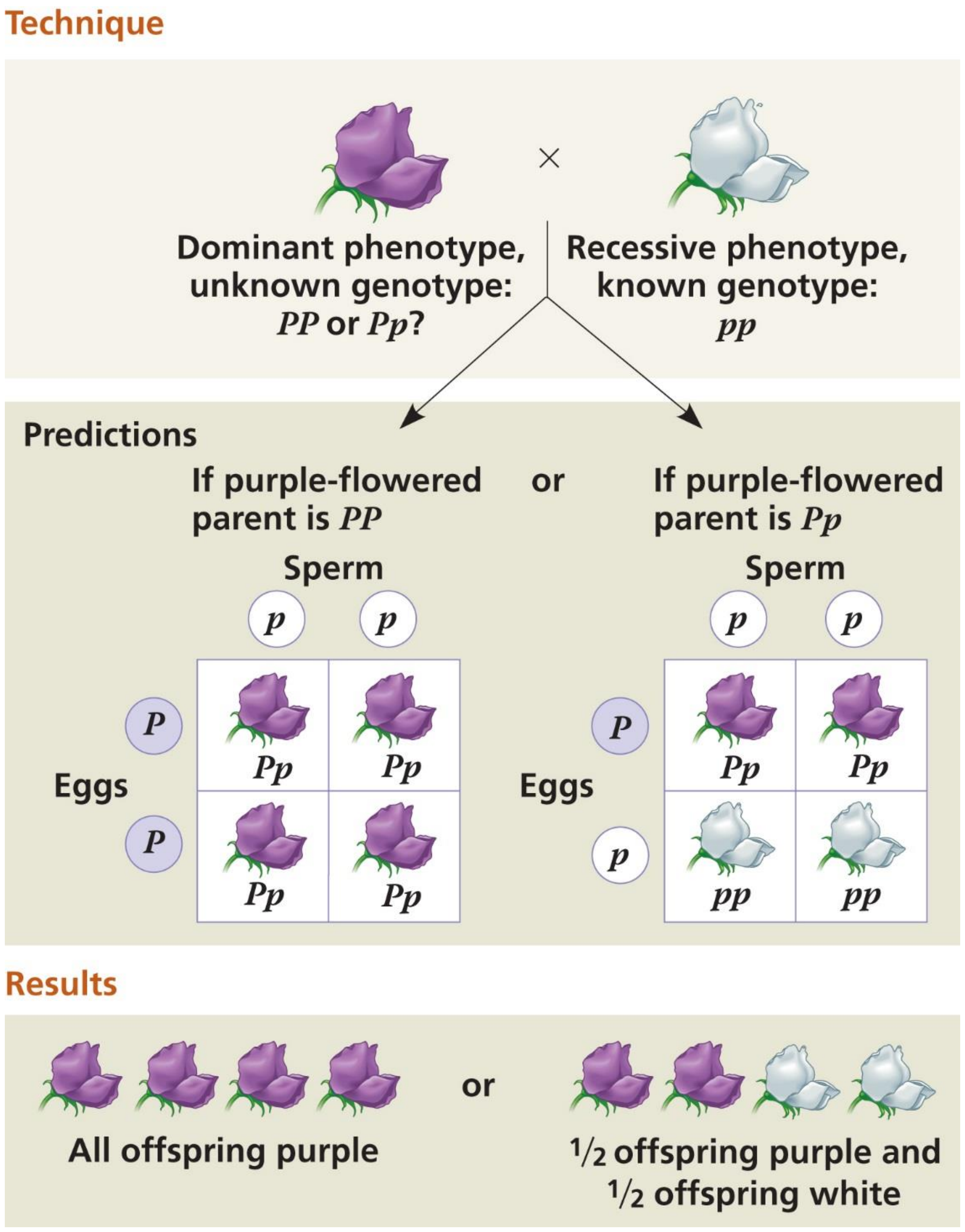
Using the testcross to determine unknown genotypes

- A testcross is the mating between an individual of unknown genotype and a homozygous recessive individual.
- A testcross can show whether the unknown genotype includes a recessive allele.



- Mendel used testcrosses to verify that he had true-breeding genotypes.

Using the testcross to determine unknown genotypes



The law of independent assortment is revealed by tracking two characters at once

- A dihybrid cross is a mating of parental varieties that differ in two characters.

- Pea shape
  - R = round
  - r = wrinkled

Pea colour

Y = yellow

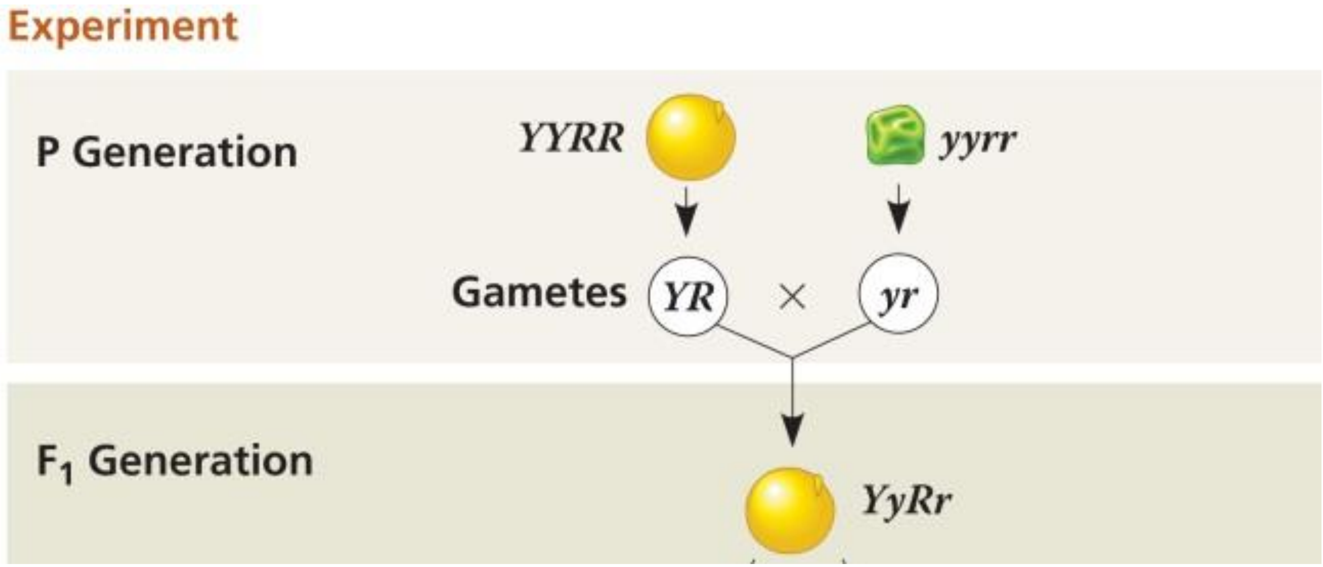
y = green

The law of independent assortment is revealed by tracking two characters at once

- Mendel performed the following dihybrid cross :

- P generation -
  - round yellow seeds (RR YY)

    x
  - wrinkled green seeds (rr yy)

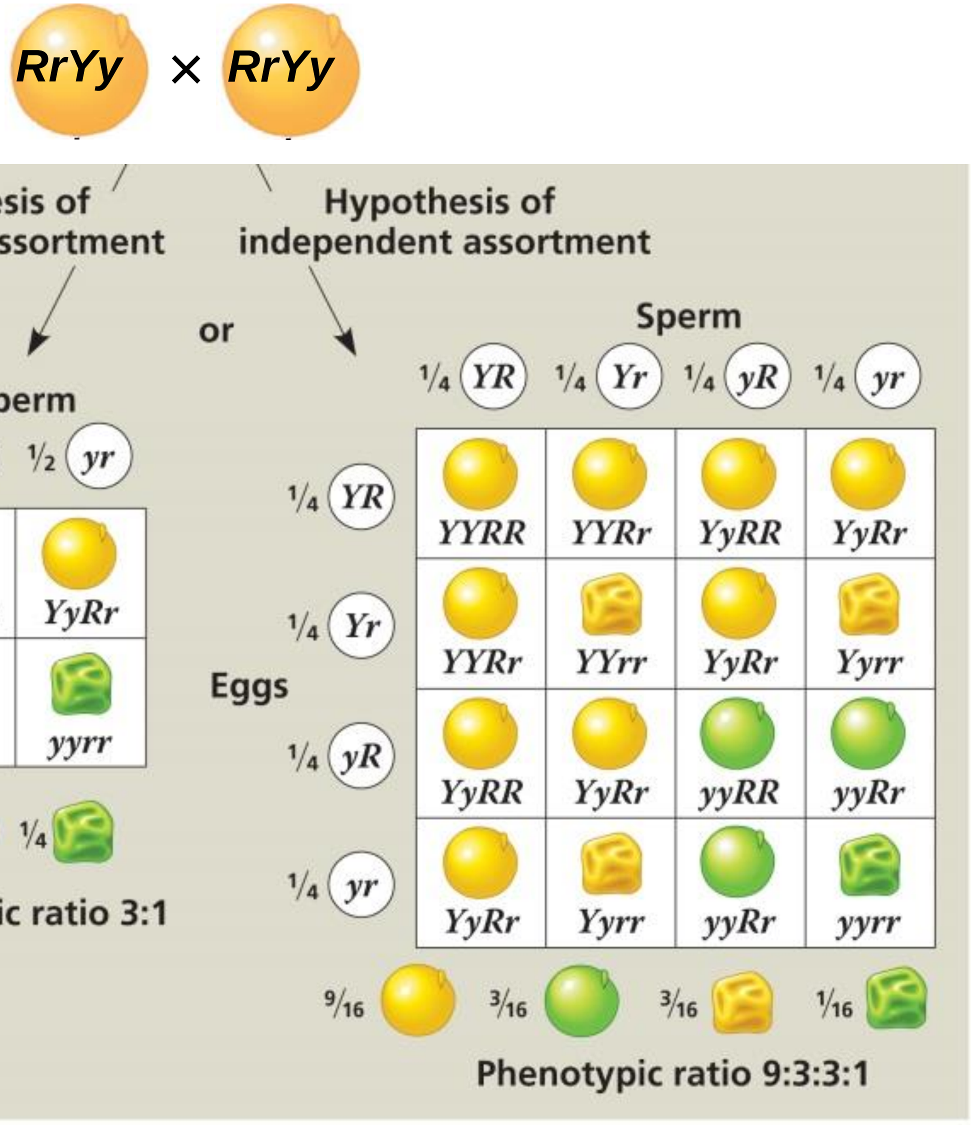


  - RR YY parent produces R Y gametes
  - rr yy parent produces r y gametes

- F1 generation: all plants with round yellow seeds
  - All offspring *Rr Yy* genotype
  - All offspring round yellow phenotype

The law of independent assortment is revealed by tracking two characters at once

- F1 generation cross:

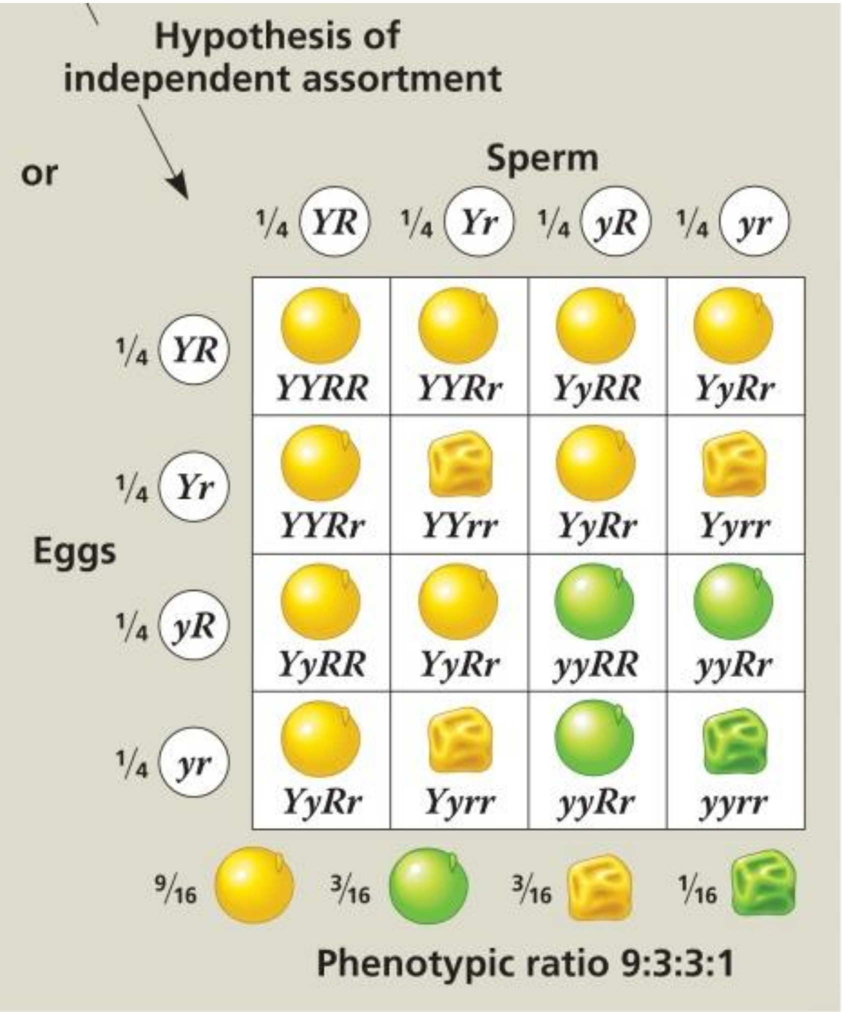


The law of independent assortment is revealed by tracking two characters at once

- Rr Yy parents produce 4 kinds of gametes
  - ¼ R Y, ¼ R y, ¼ r Y, ¼ r y

- F2 generation
  - four phenotypes
    - 9/16 = round yellow
    - 3/16 = wrinkled yellow
    - 3/16 = round green
    - 1/16 = wrinkled green

  → **9:3:3:1 ratio**

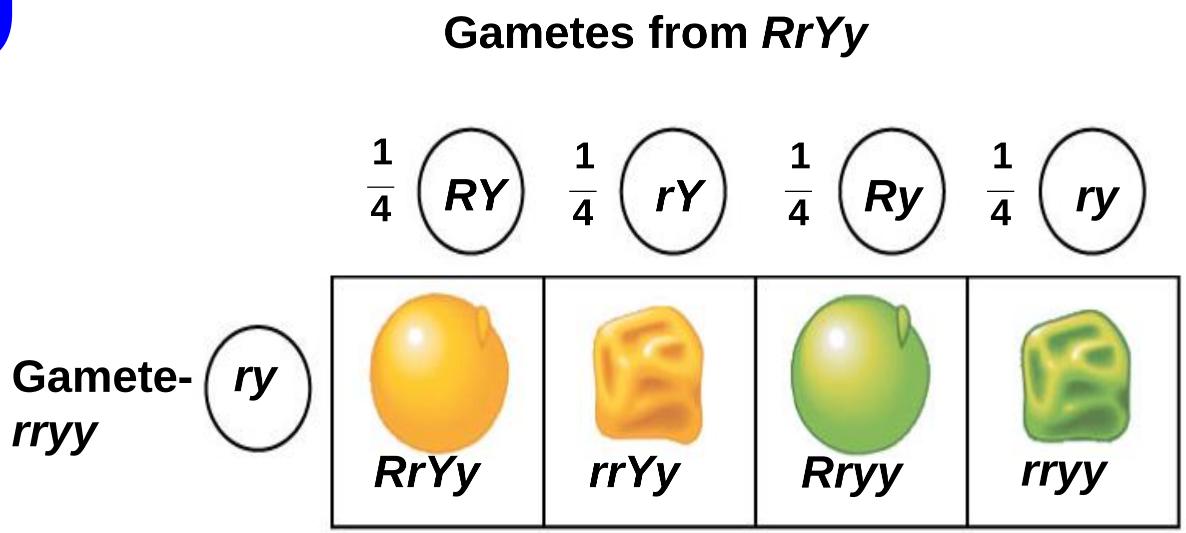


A dihybrid testcross

- Crossing…
  - Rr Yy × rr yy

- Testcross next generation
  - ¼ = round yellow
  - ¼ = round green
  - ¼ = wrinkled yellow
  - ¼ = wrinkled green

1:1:1:1 ratio



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- **Inheritance (Ch. 14.3 – 15.5)**

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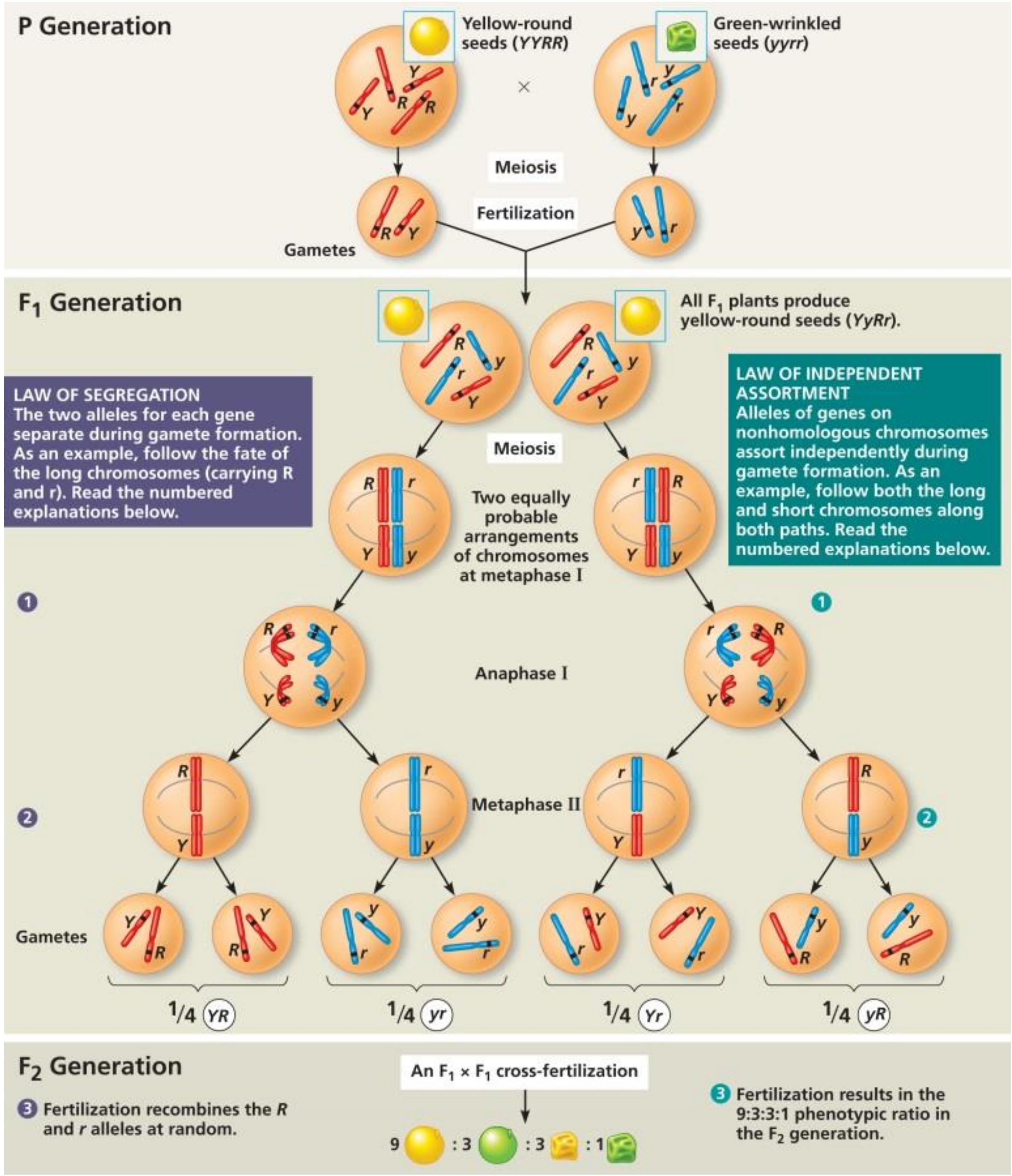
Mendel’s legacy

- Mendel’s results presented in 1866 (died in 1884)
  - Only known locally
- Work was rediscovered in early 1900s
  - Walter Sutton (1903) noted similarities between inheritance of genes and behavior of chromosomes in meiosis and fertilization
    - Chromosomes occur in pairs in diploid organisms
    - Chromosomes of each pair are separated and delivered singly to gametes
    - Independent assortment of chromosomes
    - One chromosome of each pair is derived from the male parent; one from the female parent

Chromosome behaviour accounts for Mendel's laws



Tracking genetic traits in humans

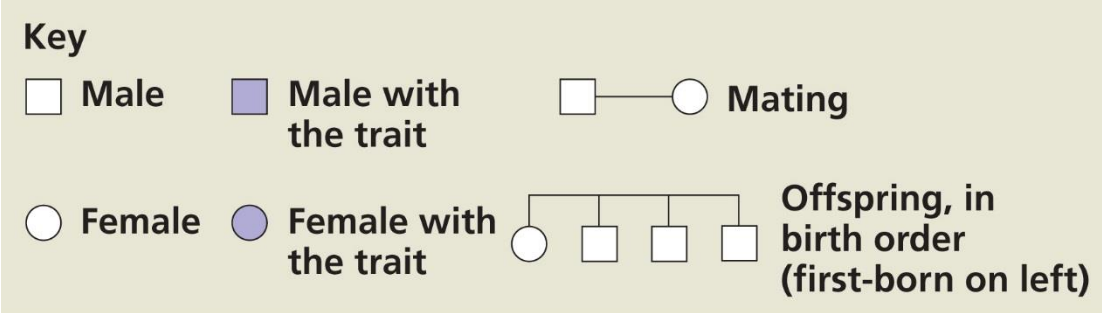
- In a simple dominant-recessive inheritance of dominant allele W and recessive allele w,
  - a recessive phenotype always results from a homozygous recessive genotype (ww) but
  - a dominant phenotype can result from either
    - the homozygous dominant genotype (WW)
    - or a heterozygous genotype (Ww).

Widow's peak No widow's peak

- Wild-type traits, those prevailing in nature, are not necessarily specified by dominant alleles.

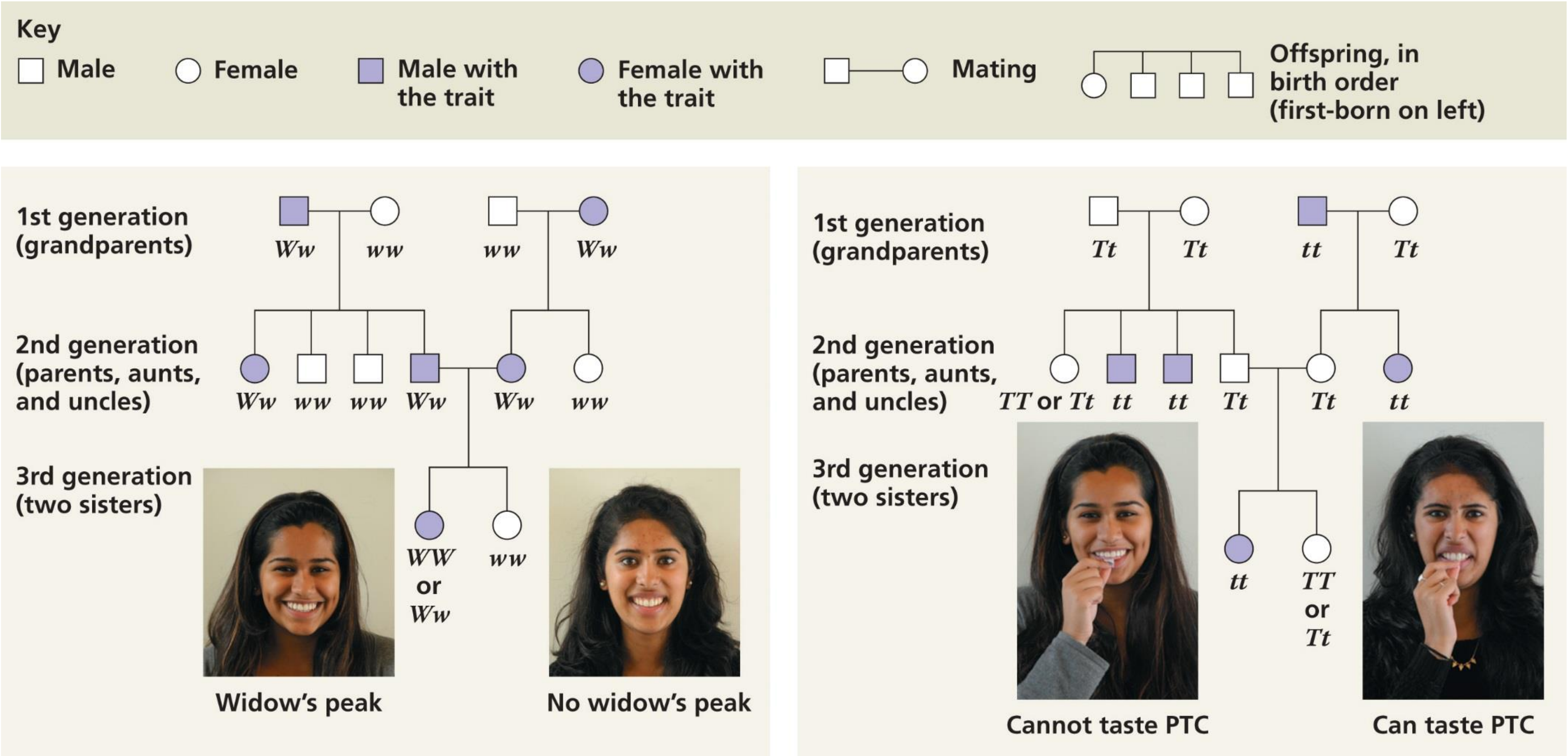
Tracking genetic traits in humans

- The inheritance of human traits follows Mendel's laws.
- A pedigree
  - Shows the inheritance of a trait in a family through multiple generations,
  - demonstrates dominant or recessive inheritance, and
  - can also be used to deduce genotypes of family members.



Widow's peak   No widow's peak

Tracking genetic traits in humans

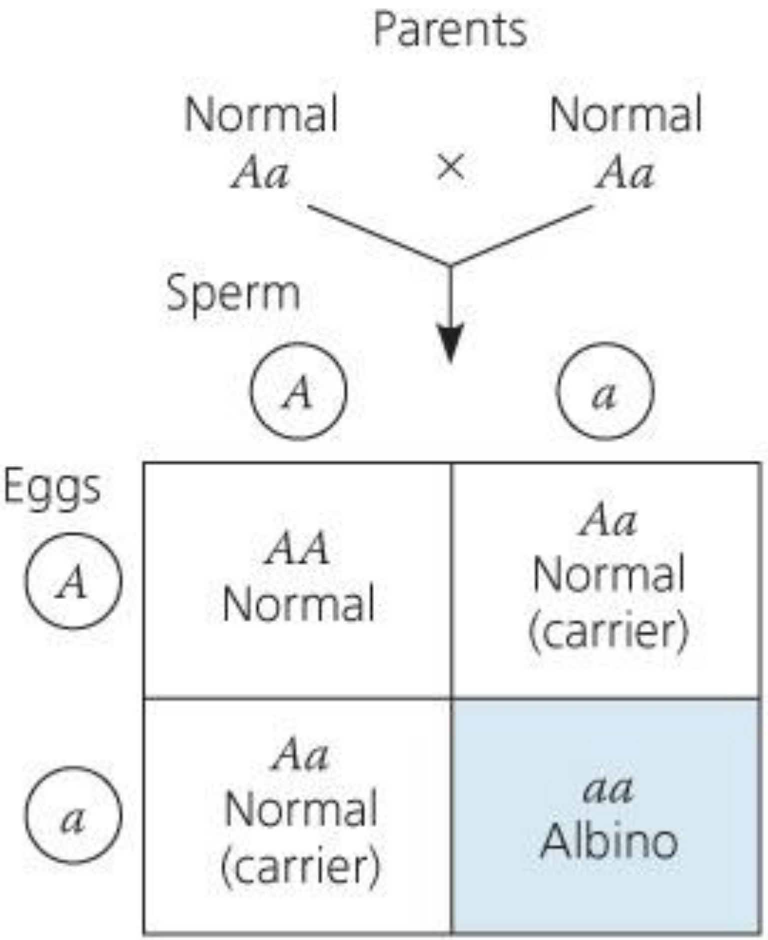


(a) Is a widow's peak a dominant or recessive trait?

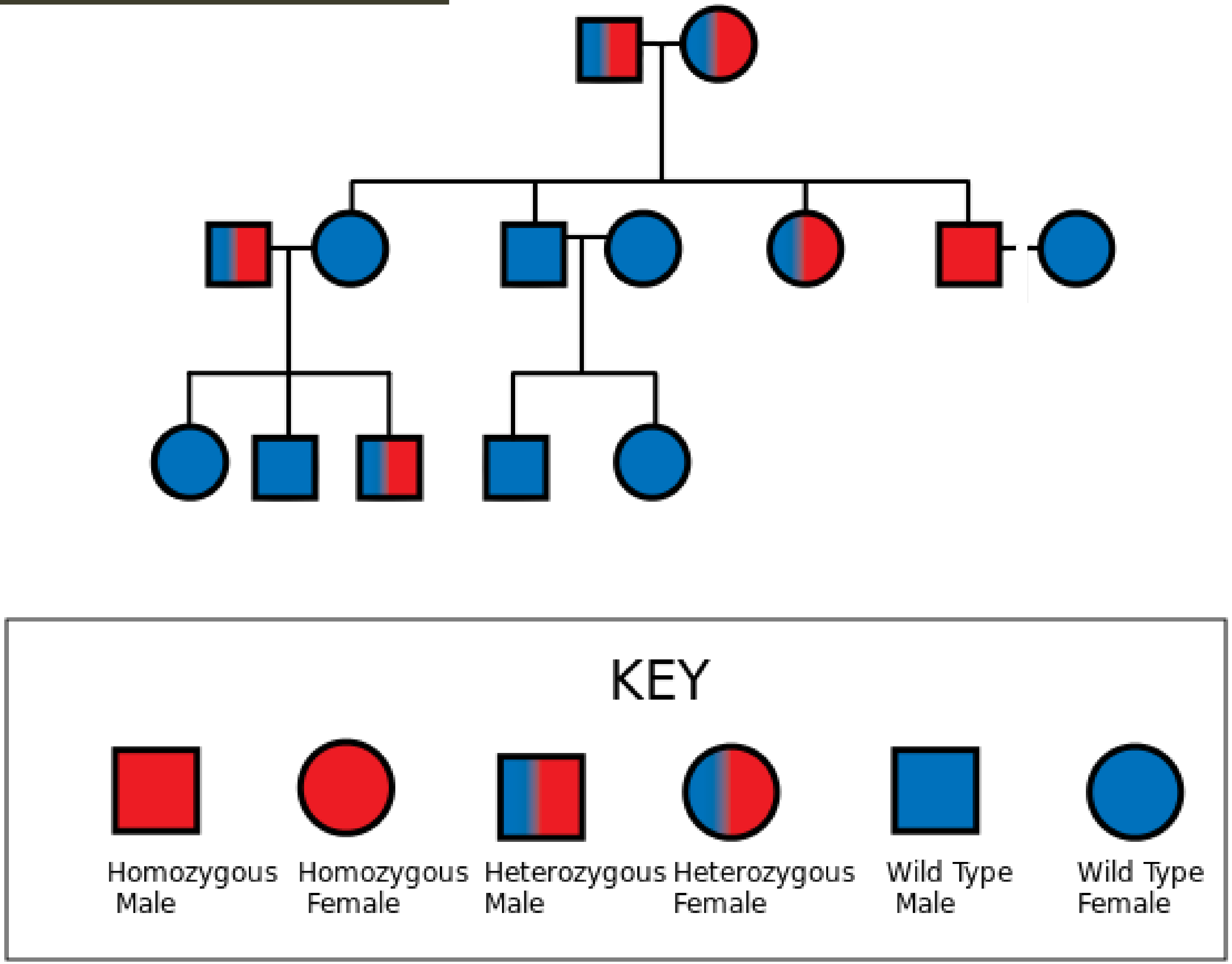
(b) Is the inability to taste a chemical called PTC a dominant or recessive trait?

Many inherited disorders in humans are controlled by a single gene

- Inherited human disorders may show recessive inheritance
  - two recessive alleles are needed to show disease,
  - heterozygous parents are carriers of the disease-causing allele, and
  - the probability of inheritance increases with inbreeding, mating between close relatives.



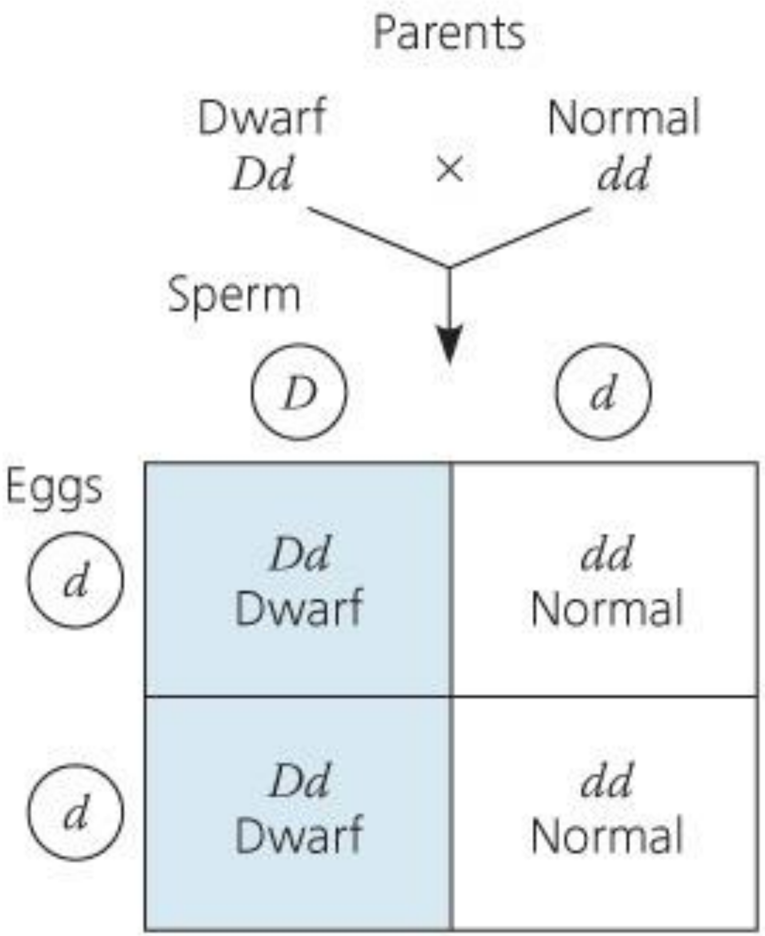
Autosomal recessive pedigree chart



http://commons.wikimedia.org/wiki/File:Autosomal_Recessive_Pedigree_Chart.svg

Many inherited disorders in humans are controlled by a single gene

- Or dominant inheritance in which
  - one dominant allele is needed to show disease and
  - dominant lethal alleles are usually eliminated from the population.



Michael Ciesielski Photography

Many inherited disorders in humans are controlled by a single gene

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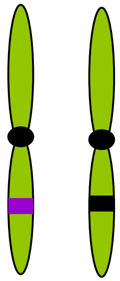

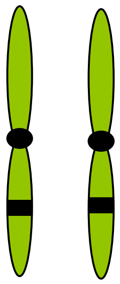

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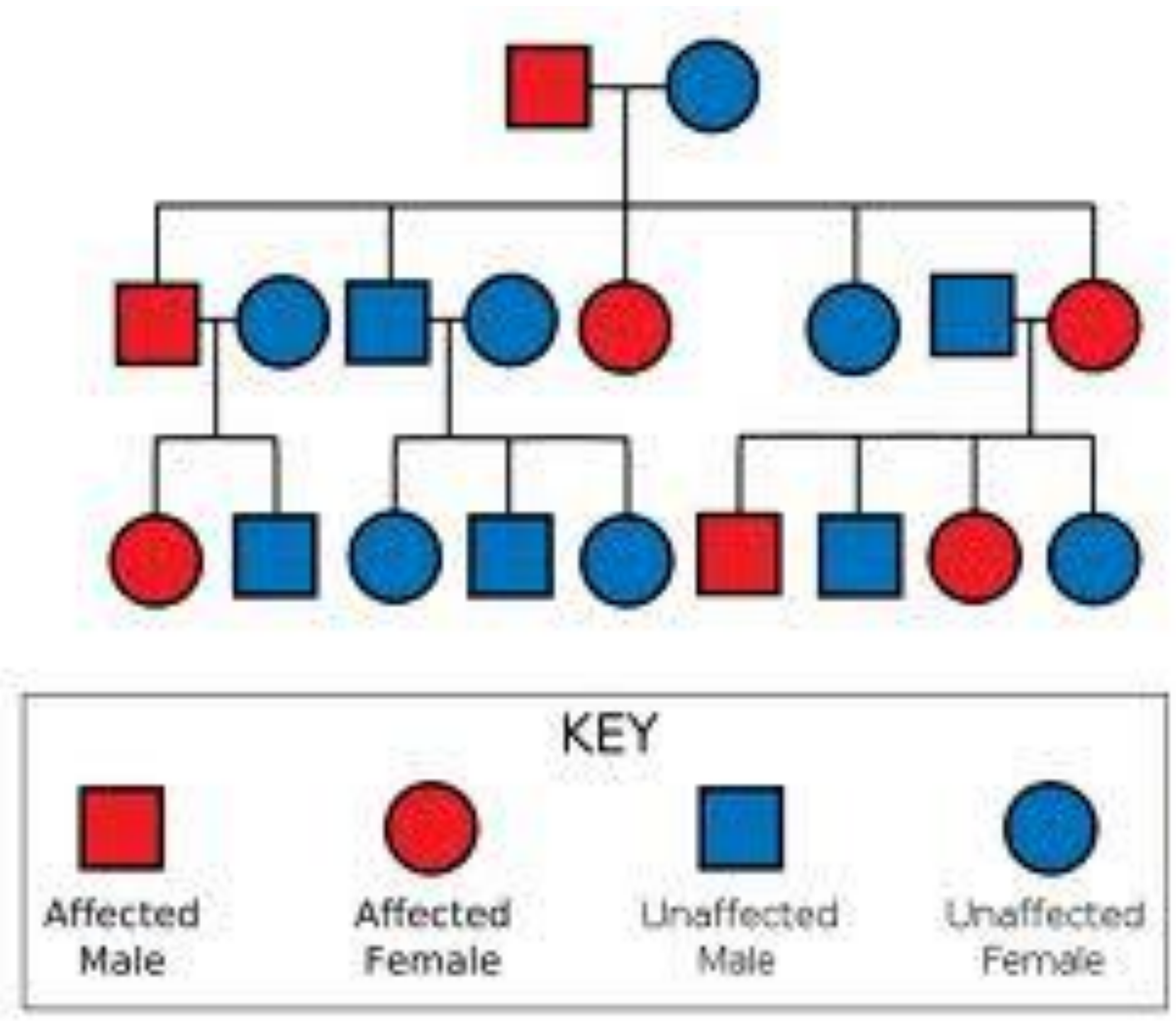

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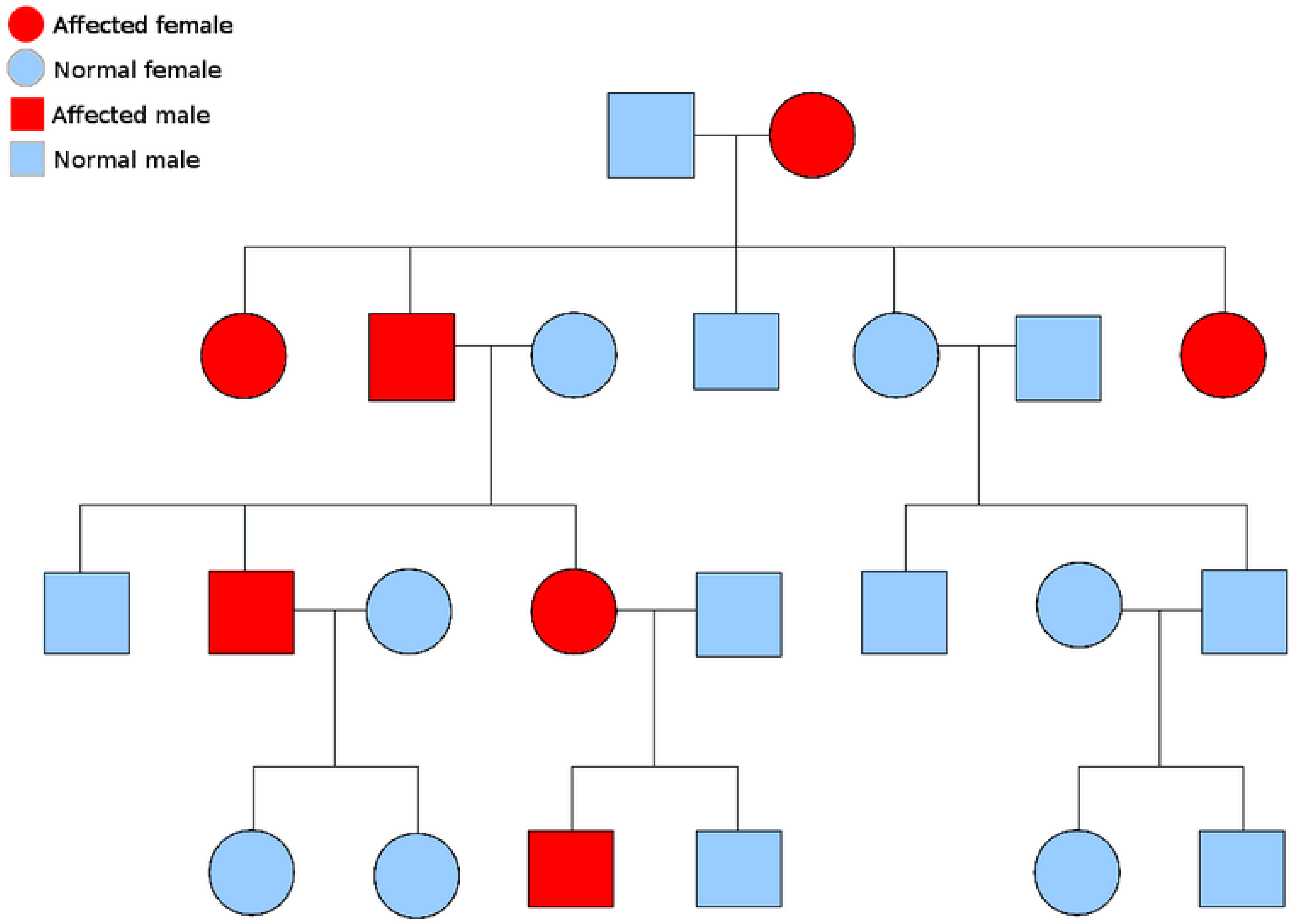
Autosomal dominant pedigree chart



http://commons.wikimedia.org/wiki/File:Autosomal_dominant.png

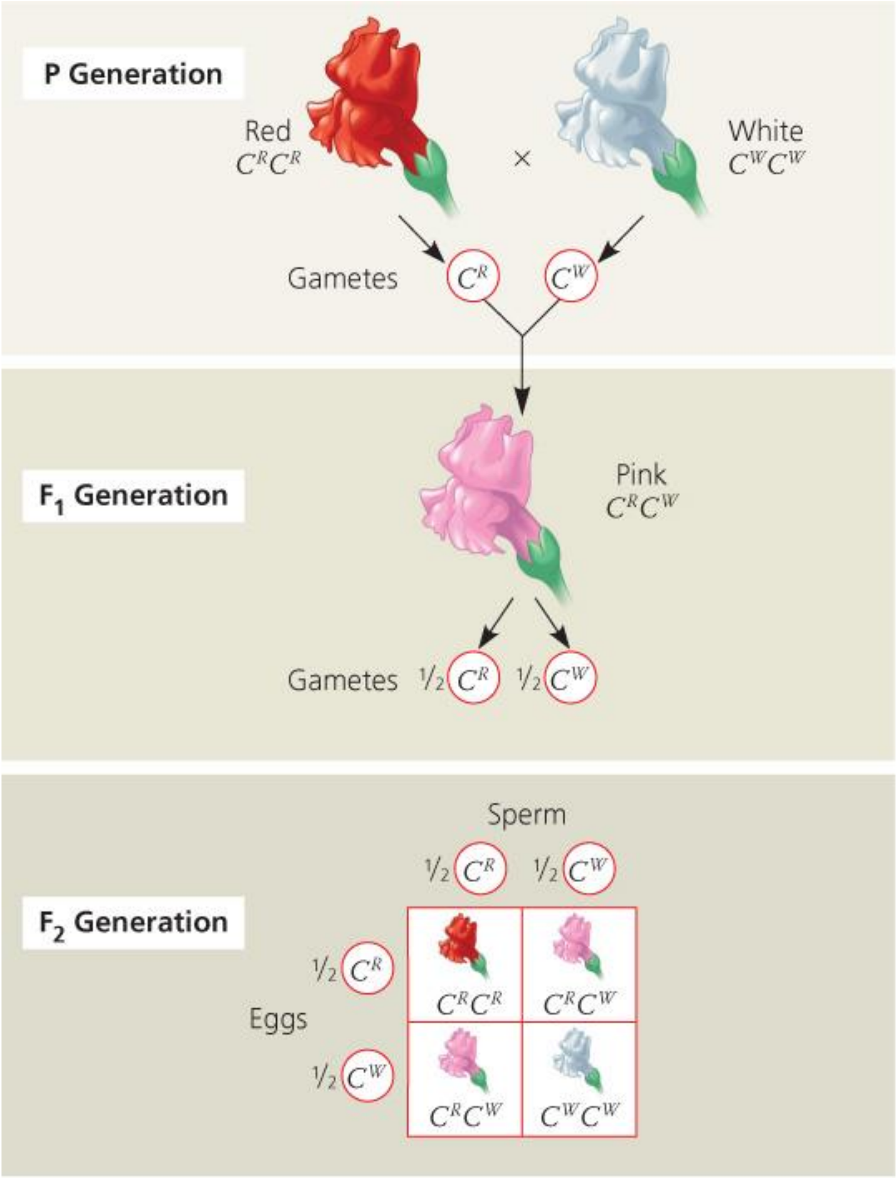
Complex inheritance patterns - Additions to Mendelian genetics

- In incomplete dominance, dominant alleles do not completely mask recessive alleles
- In codominance, the effects of different alleles are equally detectable in heterozygotes
- In multiple alleles, more than two alleles of a gene are present in a population

Incomplete dominance results in intermediate phenotypes

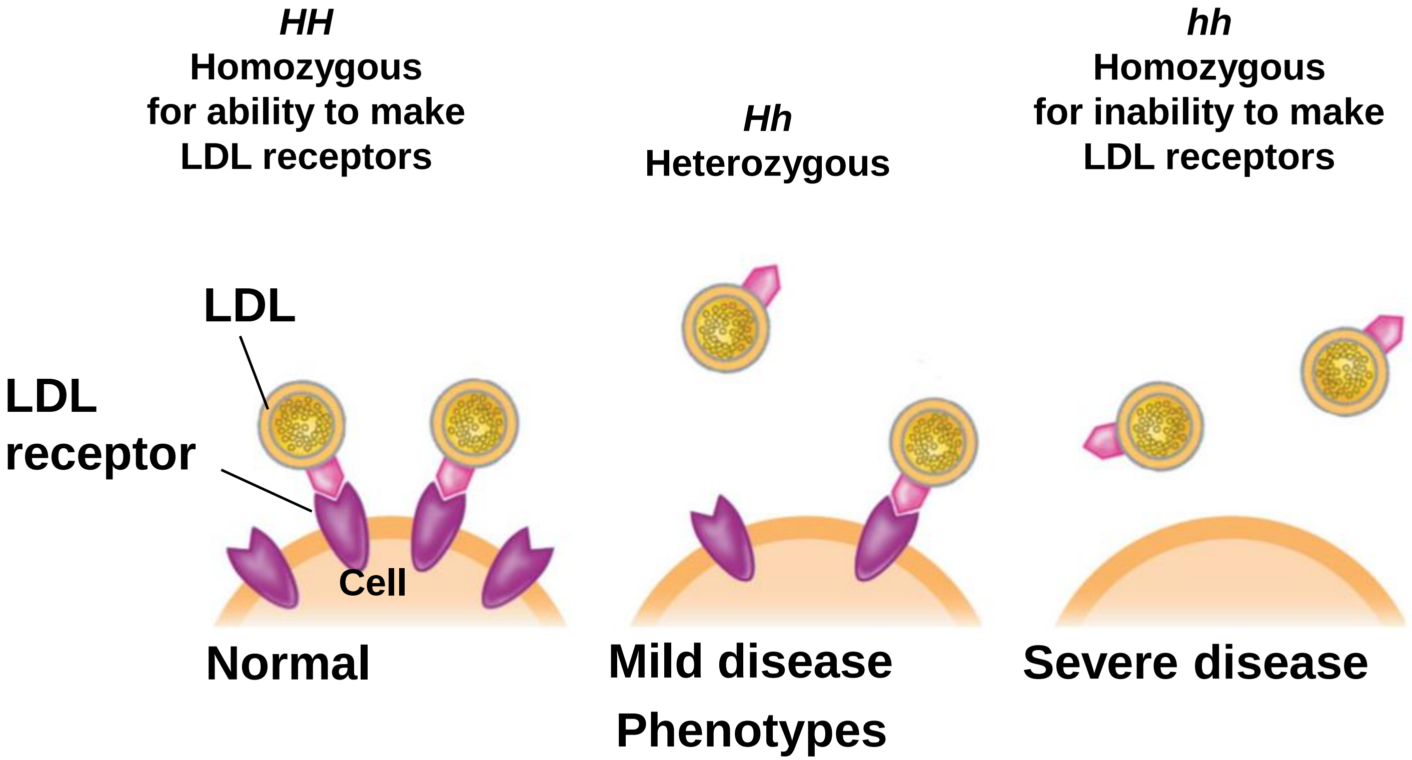
- Mendel's pea crosses always looked like one of the parental varieties, called complete dominance.
  - For some characters, the appearance of F1 hybrids falls between the phenotypes of the two parental varieties. This is called incomplete dominance, in which:
    - neither allele is dominant over the other
    - expression of both alleles occurs.

Incomplete dominance results in intermediate phenotypes



Incomplete Dominance in Human Traits

- Familial hypercholesterolemia
- Normal blood cholesterol level is 120-180 mg/dl
  - Homozygous FH patients (1 in million) have total blood cholesterol 600-1200 mg/dl
  - Heterozygous FH patients (1 in 500) have total blood cholesterol of 280-500 mg/dl.



Many genes have more than two alleles in the population

- Although an individual can at most carry two different alleles for a particular gene, more than two alleles often exist in the wider population.
- Human ABO blood group phenotypes involve three alleles for a single gene.
- The four human blood groups, A, B, AB, and O, result from combinations of these three alleles.
- The A and B alleles are both expressed in heterozygous individuals, a condition known as **codominance.**

Codominance

- Neither allele is dominant over the other
  - expression of both alleles is observed as a distinct phenotype in the heterozygous individual.
  - AB blood type is an example of codominance.

(a) The three alleles for the ABO blood groups and their carbohydrates. Each allele codes for an enzyme that may add a specific carbohydrate (designated by the superscript on the allele and shown as a triangle or circle) to red blood cells.

Allele	I^A	I^B	i
Carbohydrate	A △	B ●	none

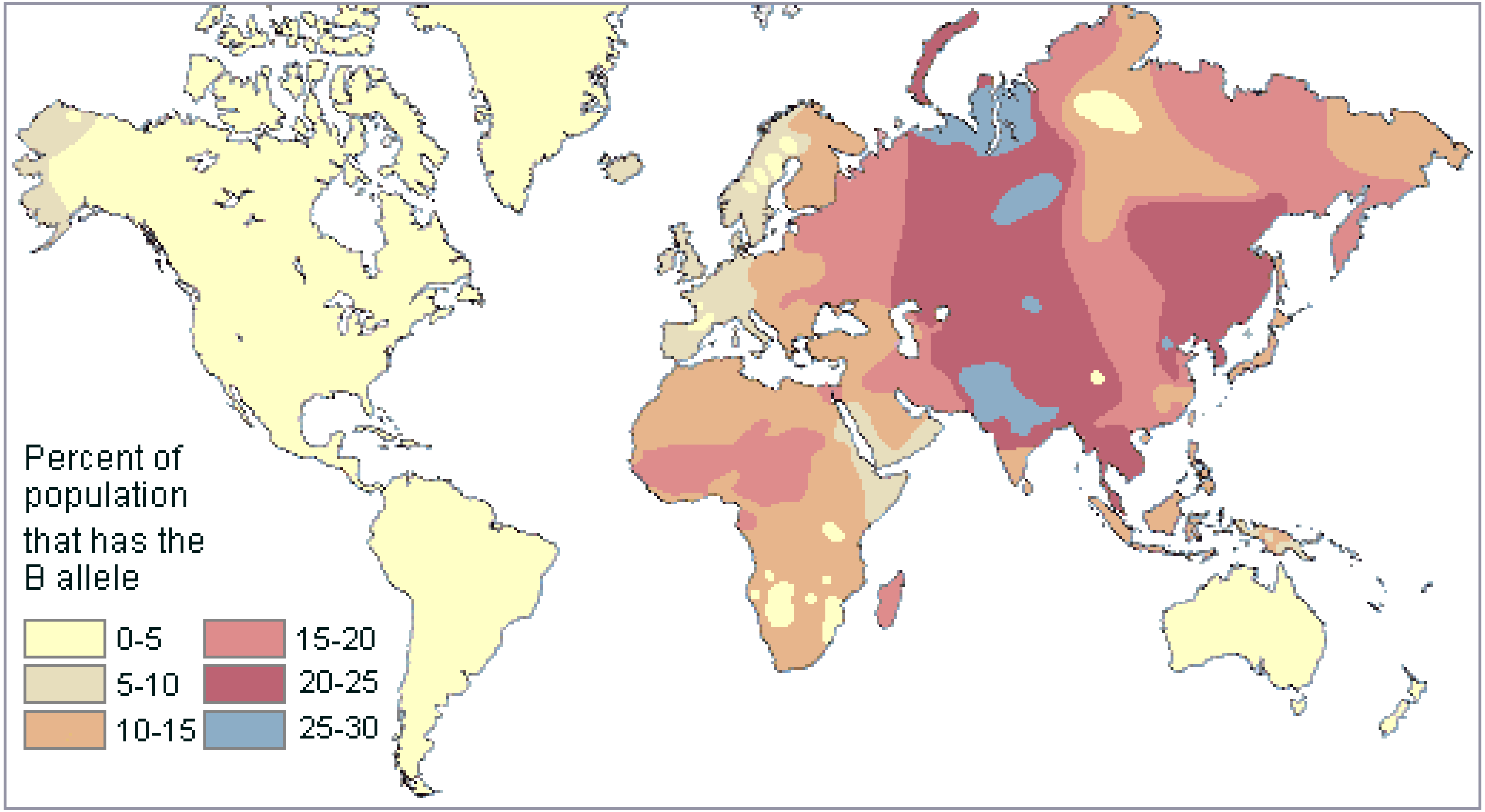
(b) Blood group genotypes and phenotypes. There are six possible genotypes, resulting in four different phenotypes.

Genotype	I^AI^A or I^Ai	I^BI^B or I^Bi	I^AI^B	ii
Red blood cell appearance				
Phenotype (blood group)	A	B	AB	O

Codominance

Blood Group (Phenotype)	Antibodies Present in Blood	Reaction When Blood from Groups Below Is Mixed with Antibodies from Groups at Left			
		O	A	B	AB
A	Anti-B				
B	Anti-A				
AB	*None*				
O	Anti-A Anti-B				

Distribution of blood types



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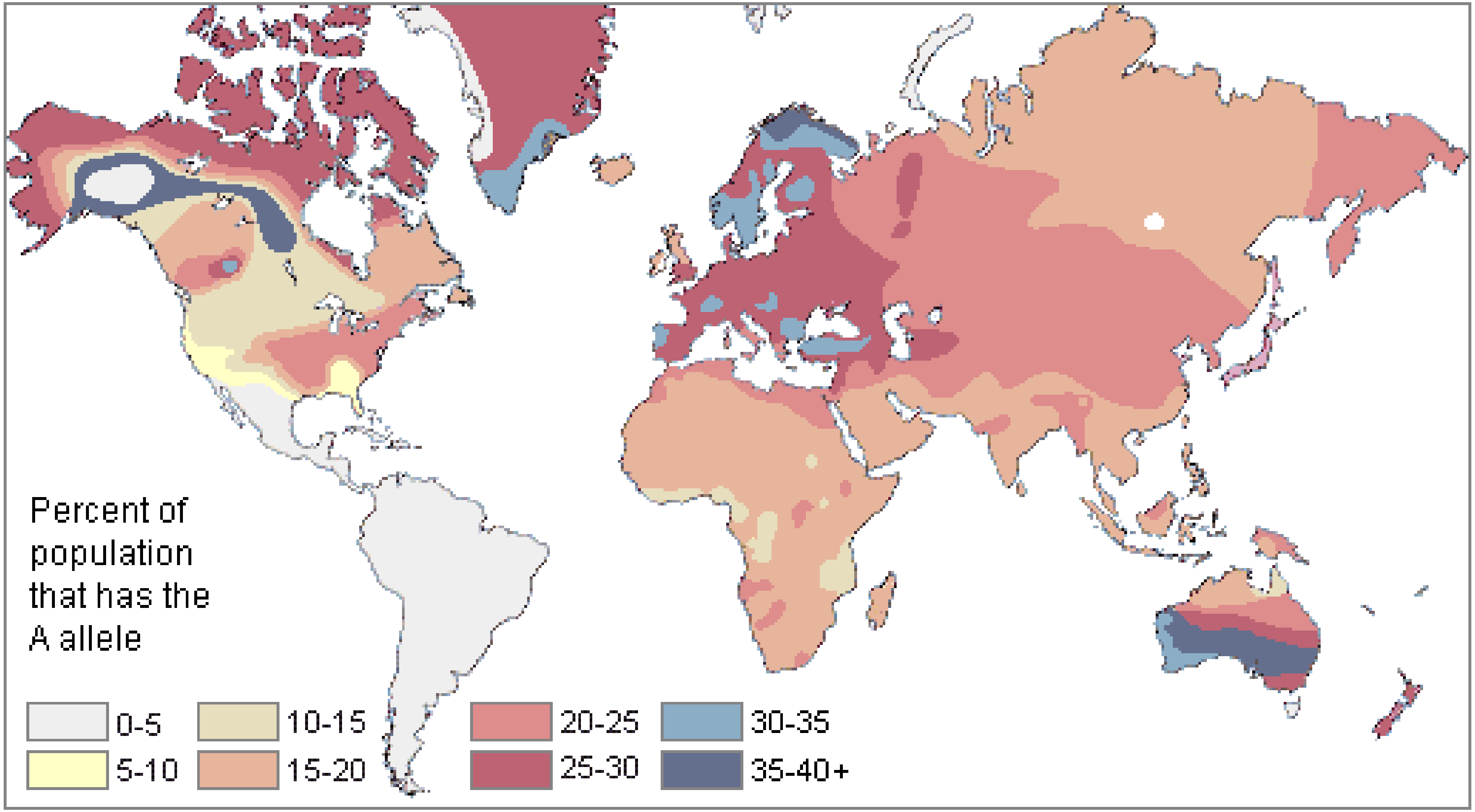
Distribution of blood types



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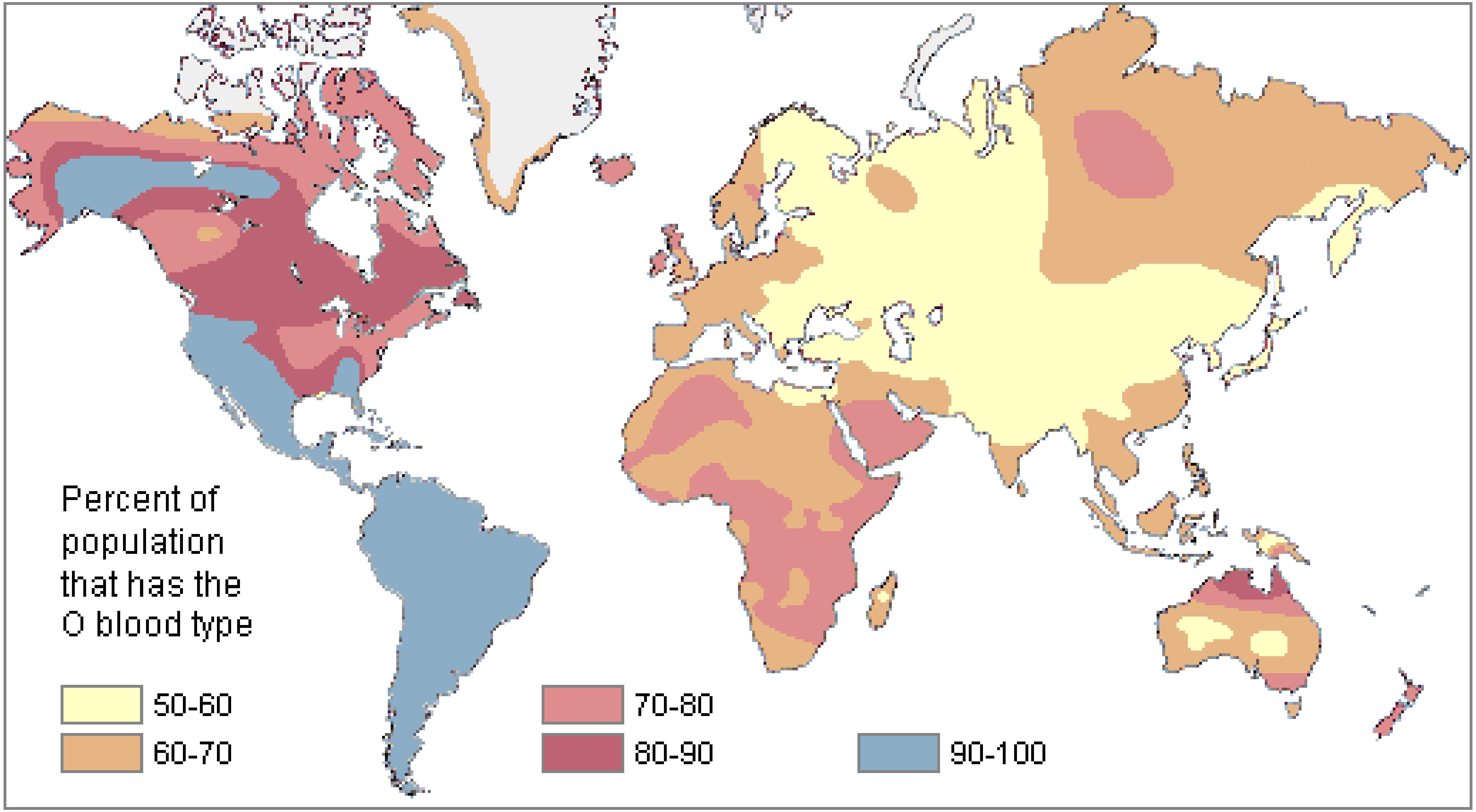
Distribution of blood types



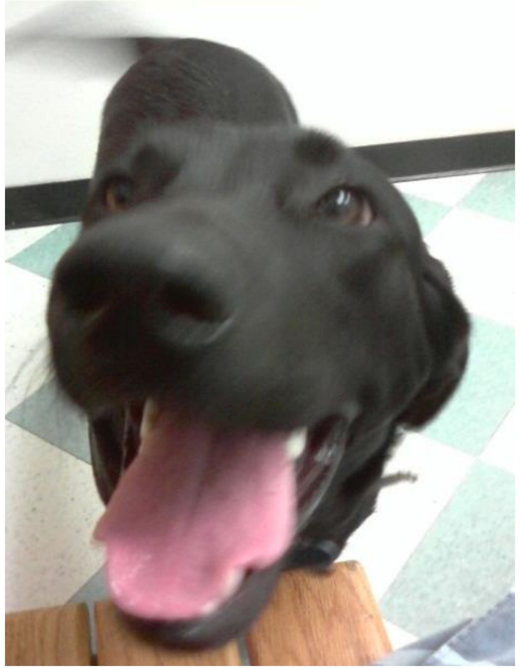
https://www2.palomar.edu/anthro/vary/vary_3.htm

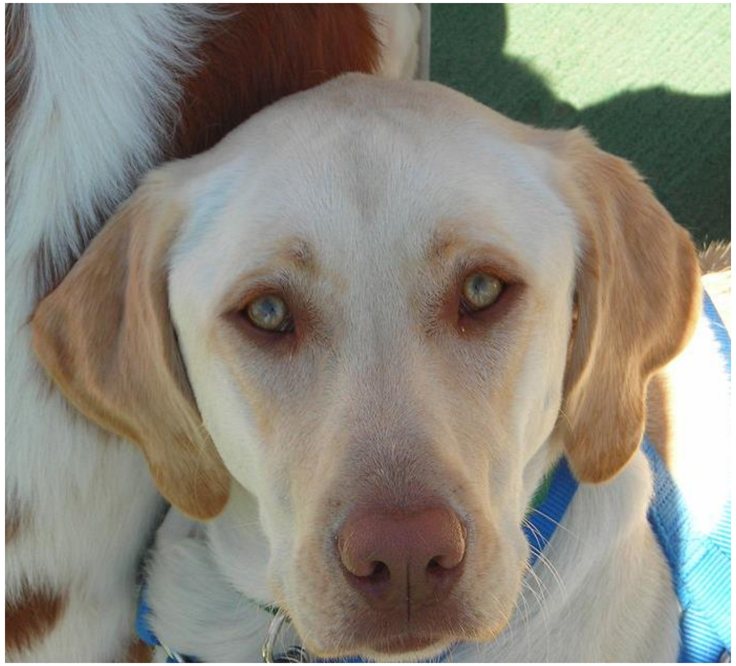
Complex inheritance patterns - Additions to Mendelian genetics

- In epistasis, genes interact, with the activity of one gene influencing the activity of another gene
- In polygenic inheritance, a character is controlled by the common effects of several genes
- In pleiotropy, two or more characters are affected by a single gene

Epistasis

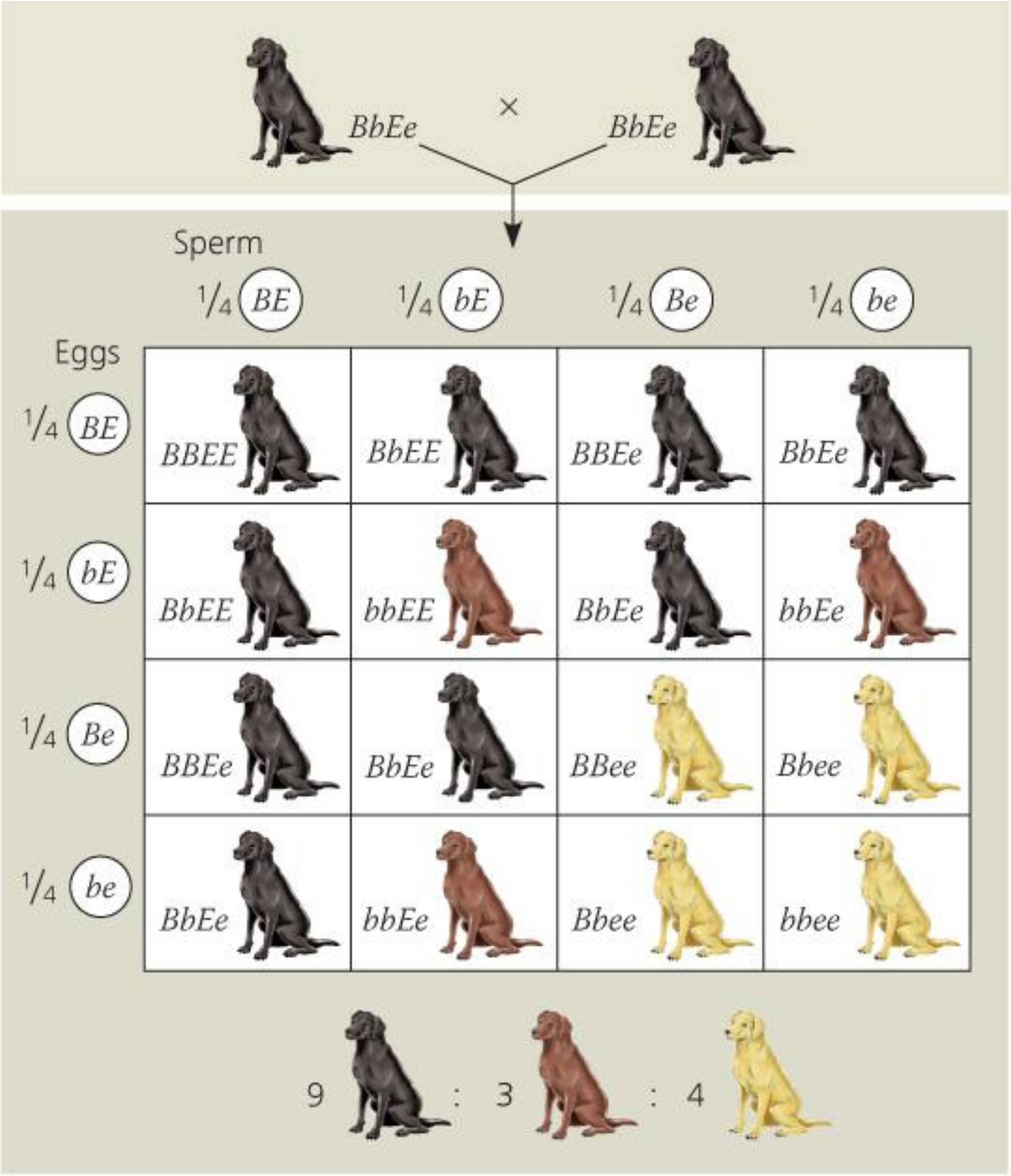
- Genes interact
  - Allele of one locus inhibits or masks effects of allele at a different locus
  - Some expected phenotypes do not appear among offspring



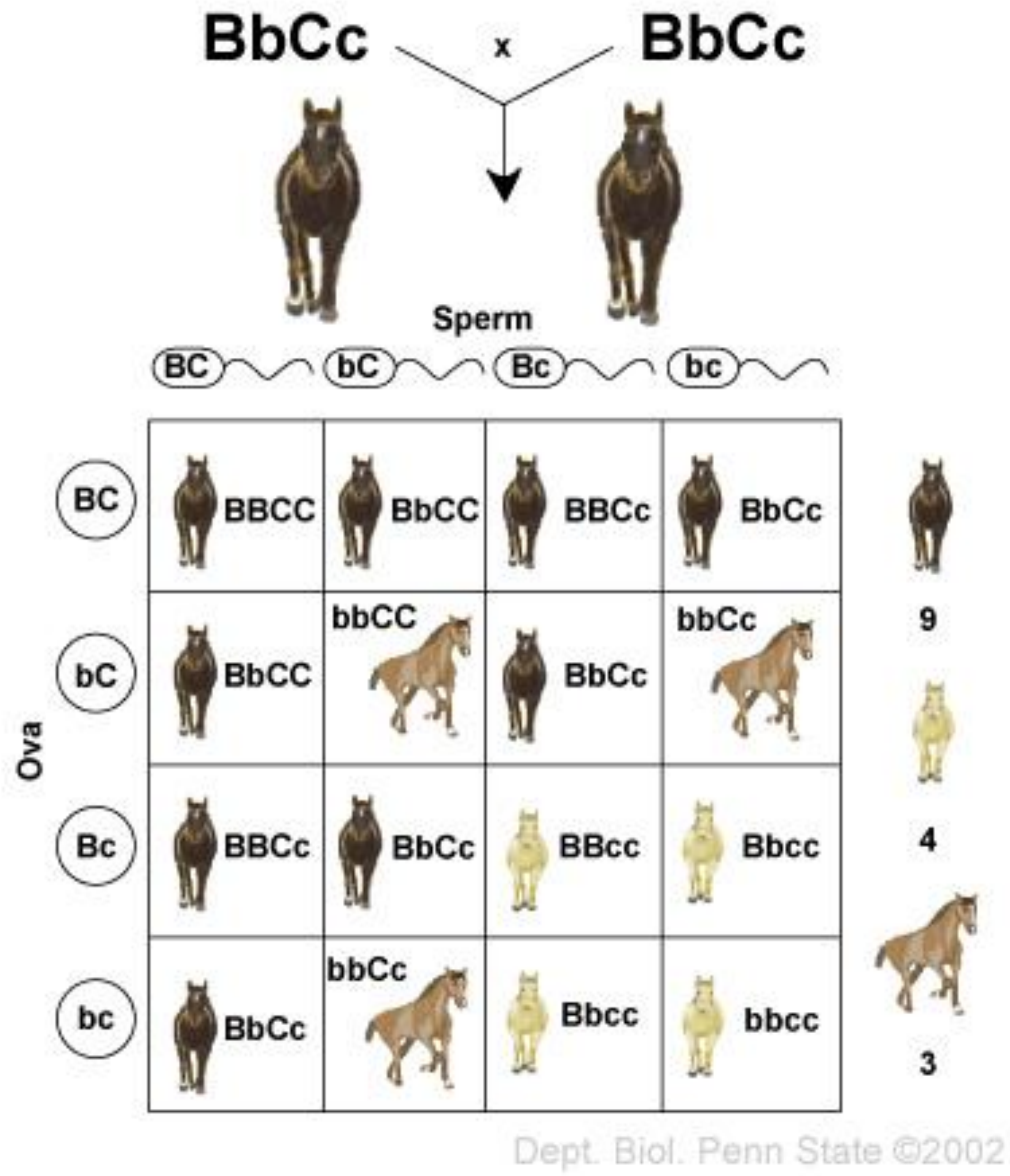


Labrador Retrievers

- Melanin pigment gene
  - *B* allele: black fur colour (*dominant*)
  - *b* allele: brown fur colour (*recessive*)
- Pigment deposition gene
  - *E* allele: pigment deposition normal (*dominant*)
  - *e* allele: pigment deposition blocked (*recessive*)
- Phenotypes
  - Black fur: *BB EE, BB Ee, Bb EE, Bb Ee*
  - Brown fur: *bb EE, bb Ee*
  - Yellow fur: *BB ee, Bb ee, bb ee*



Epistasis



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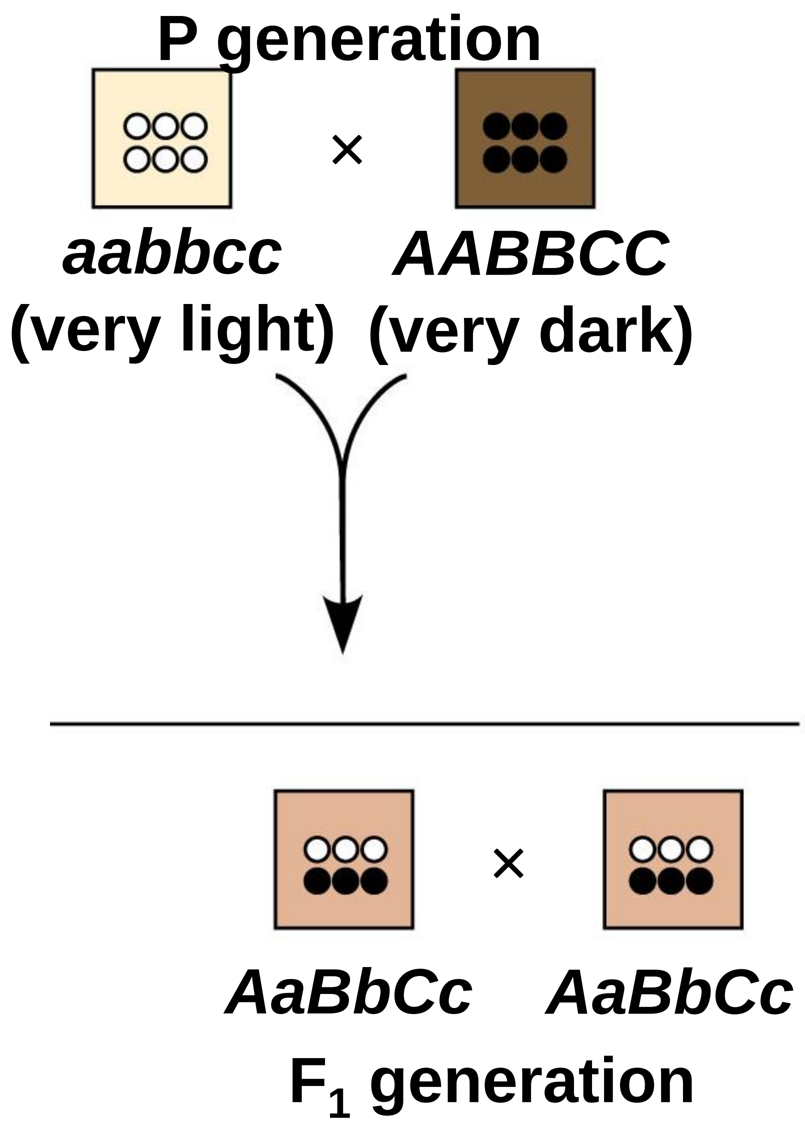
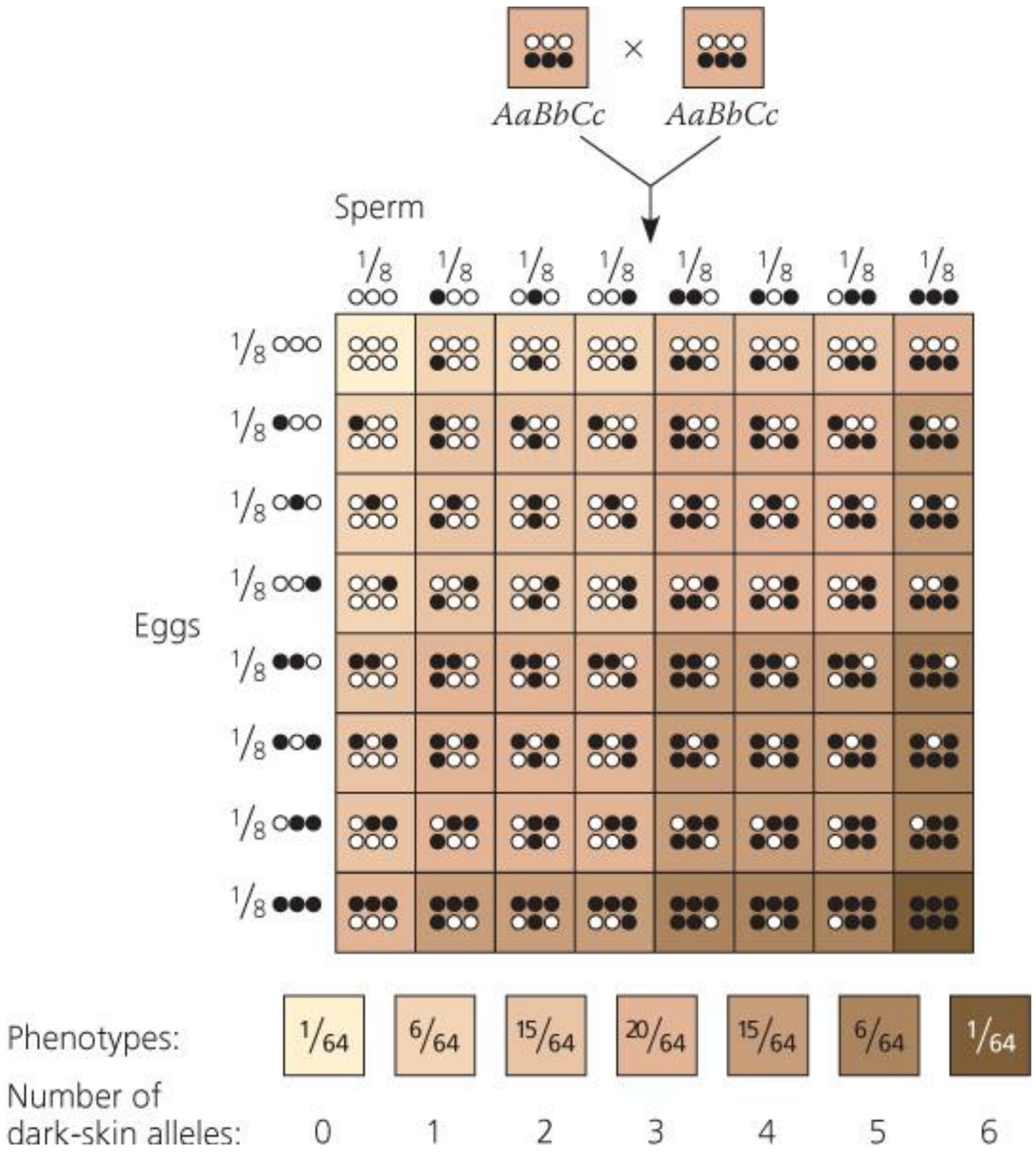
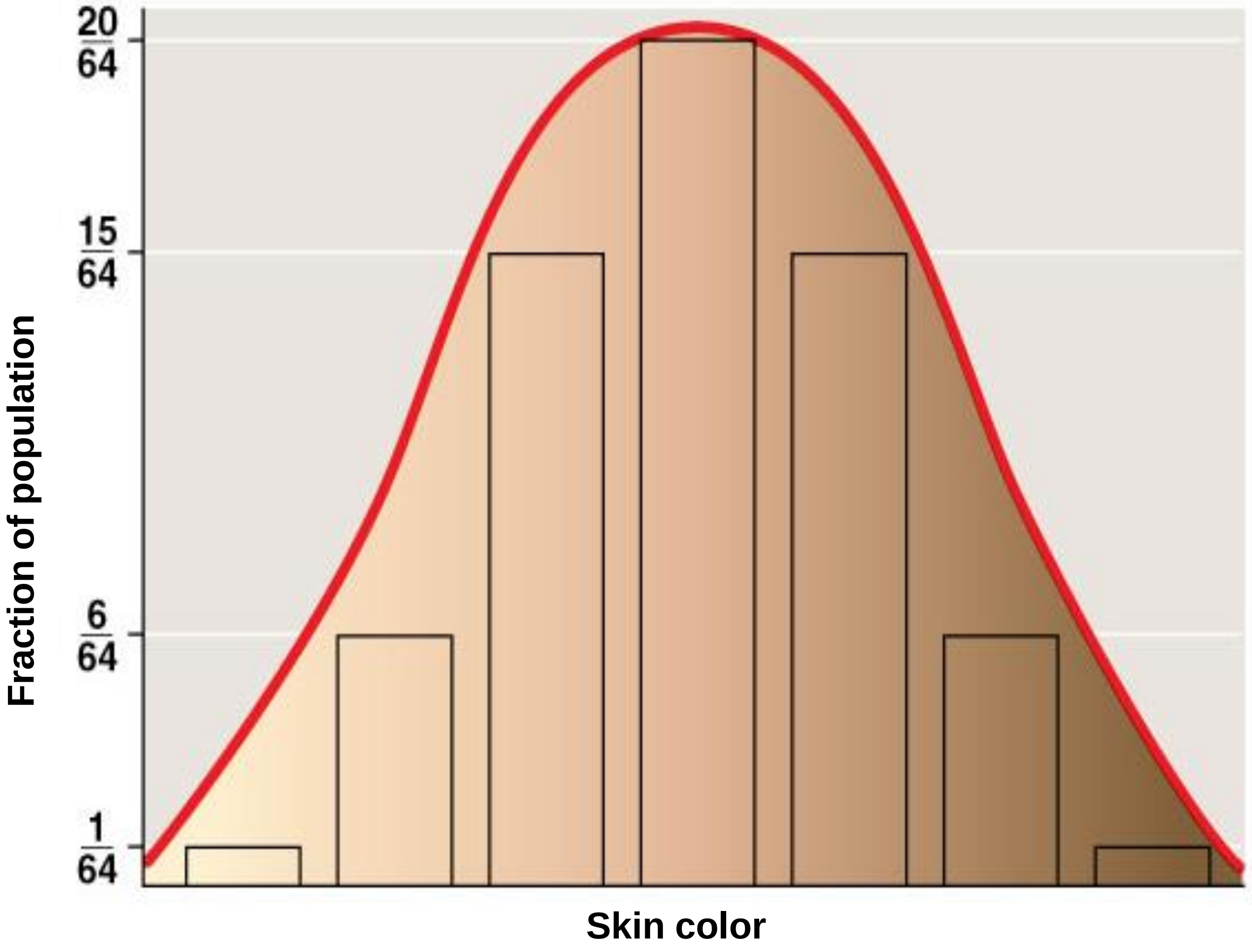
A single character may be influenced by many genes



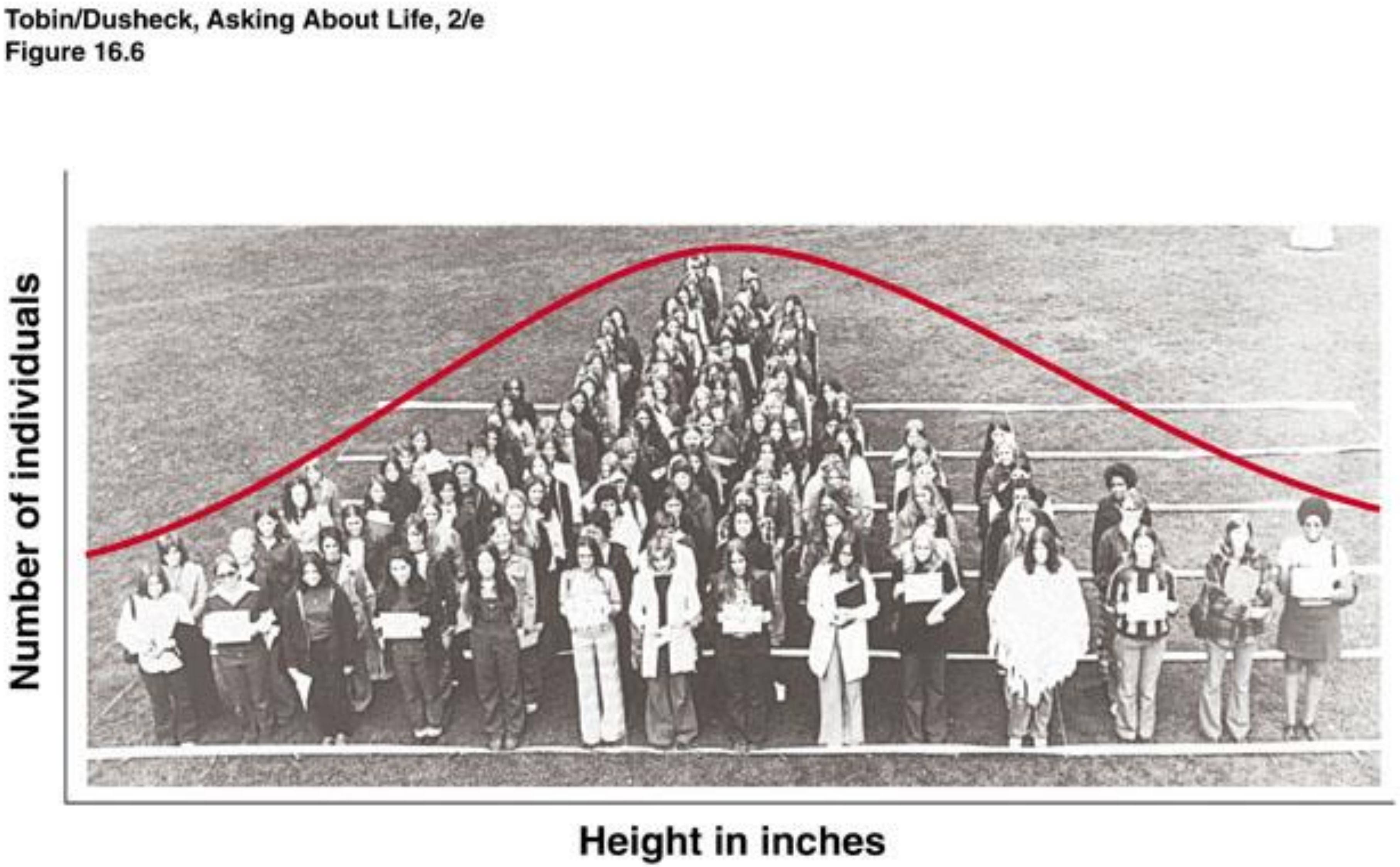
- Many characteristics result from polygenic inheritance, in which a single phenotypic character results from the additive effects of two or more genes.
  - Human skin color is an example of **polygenic inheritance.**

A single character may be influenced by many genes





Polygenic inheritance



http://blog.canacad.ac.jp/bio/BiologyIBHL1/3093.html

A single gene may affect many phenotypic characters

An individual homozygous for the sickle-cell allele

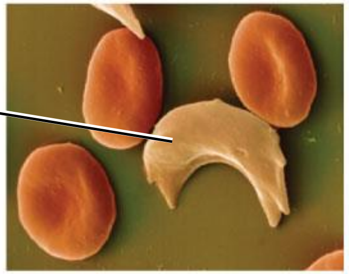
↓

Produces sickle-cell (abnormal) hemoglobin

↓

The abnormal hemoglobin crystallizes, causing red blood cells to become sickle-shaped

Sickled cell



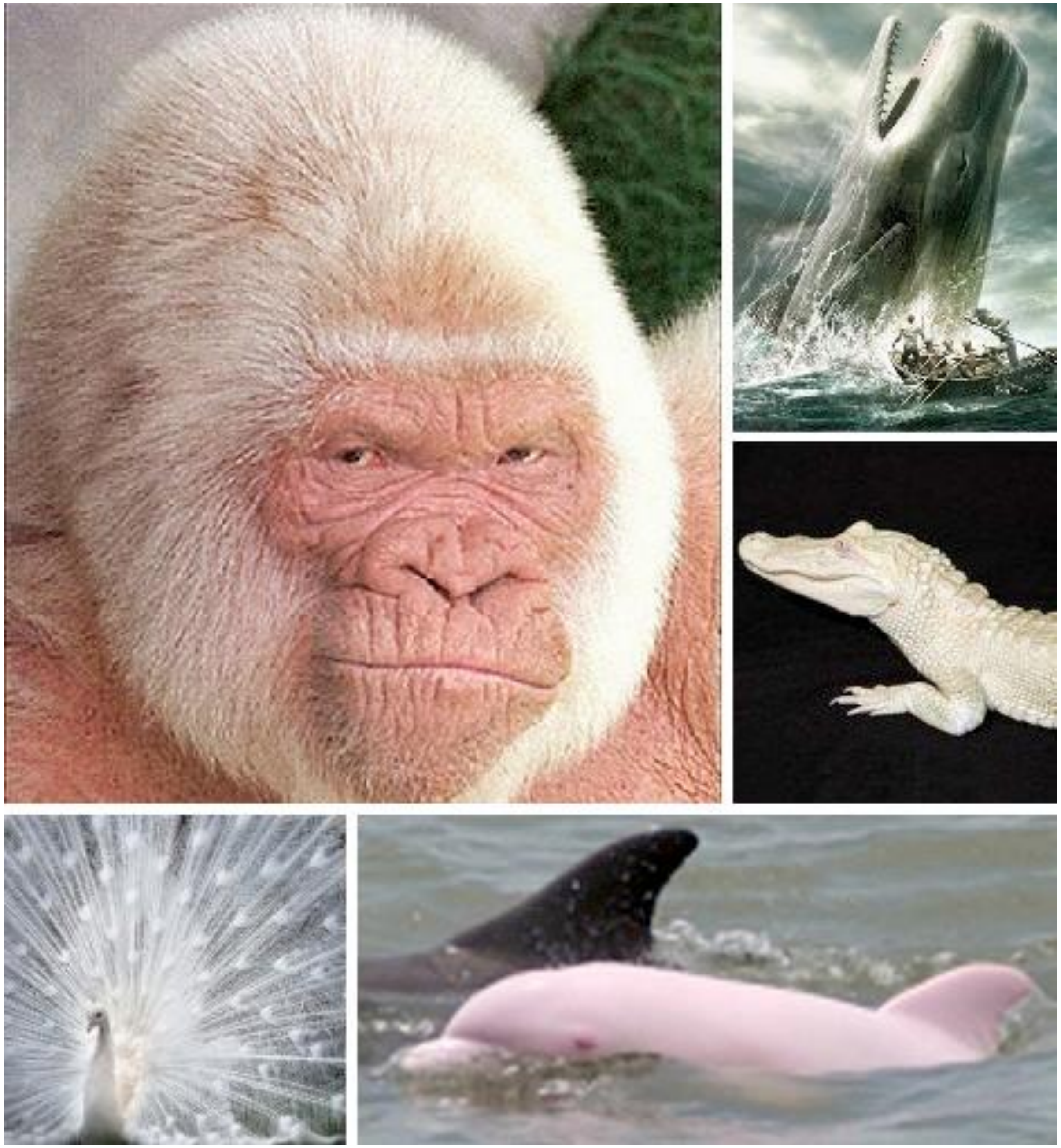
The multiple effects of sickled cells

Damage to organs
- Kidney failure
- Heart failure
- Spleen damage
- Brain damage (impaired mental function, paralysis)

Other effects
- Pain and fever
- Joint problems
- Physical weakness
- Anemia
- Pneumonia and other infections

- **Pleiotropy** occurs when one gene influences many characteristics.
  - Sickle-cell disease is a human example of pleiotropy.

Pleiotropy



http://www.bio.miami.edu/dana/250/250SS13_6.html

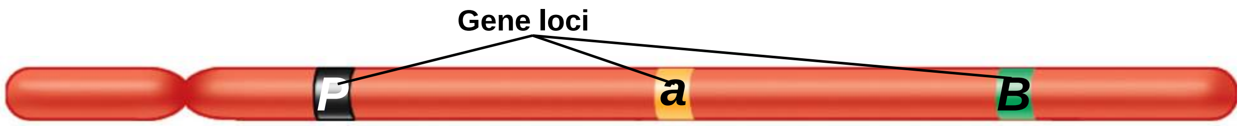
The environment affects many characters

- Many characters result from a combination of heredity and the environment. For example,
  - skin color is affected by exposure to sunlight,
  - susceptibility to diseases, such as cancer, has hereditary and environmental components, and
  - identical twins show some differences.

- Only genetic influences are inherited.

Chromosome behavior accounts for Mendel's laws

- The chromosome theory of inheritance states that
  - genes occupy specific loci (positions) on chromosomes

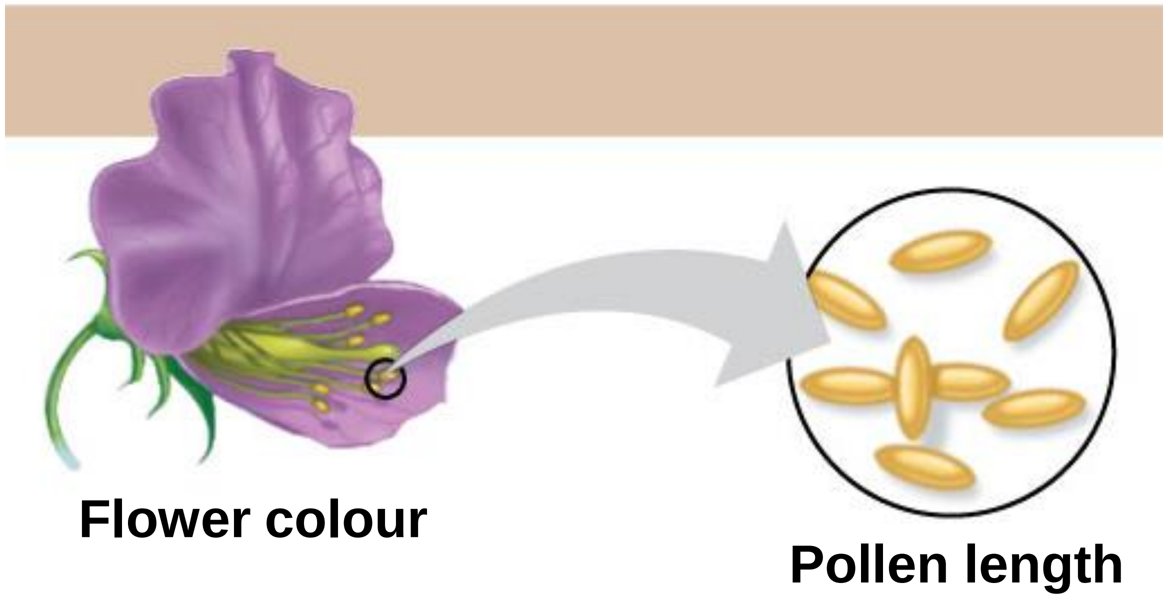


  - chromosomes undergo segregation and independent assortment during meiosis.

- Mendel's laws correlate with chromosome separation in meiosis.
  - The law of segregation depends on separation of homologous chromosomes in anaphase I.

Genes on the same chromosome tend to be inherited together

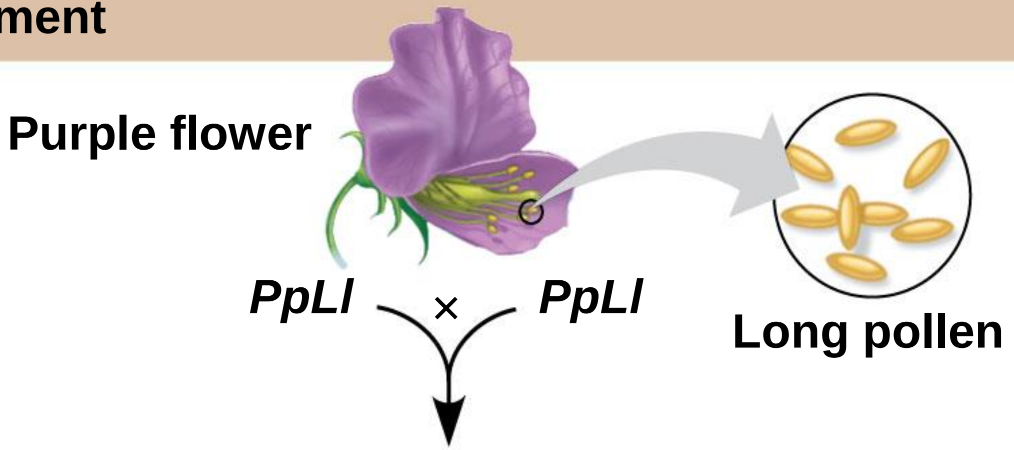
- Bateson and Punnett studied plants that did not show a 9:3:3:1 ratio in the F2 generation.
  - Purple colour dominant (*P*)
  - Red colour recessive(*p*)
  - Long pollen dominant (*L*)
  - Round pollen recessive (*l*)



Genes on the same chromosome tend to be inherited together

- Performed dihybrid cross, then ‘selfed’ the F1 heterozygotes

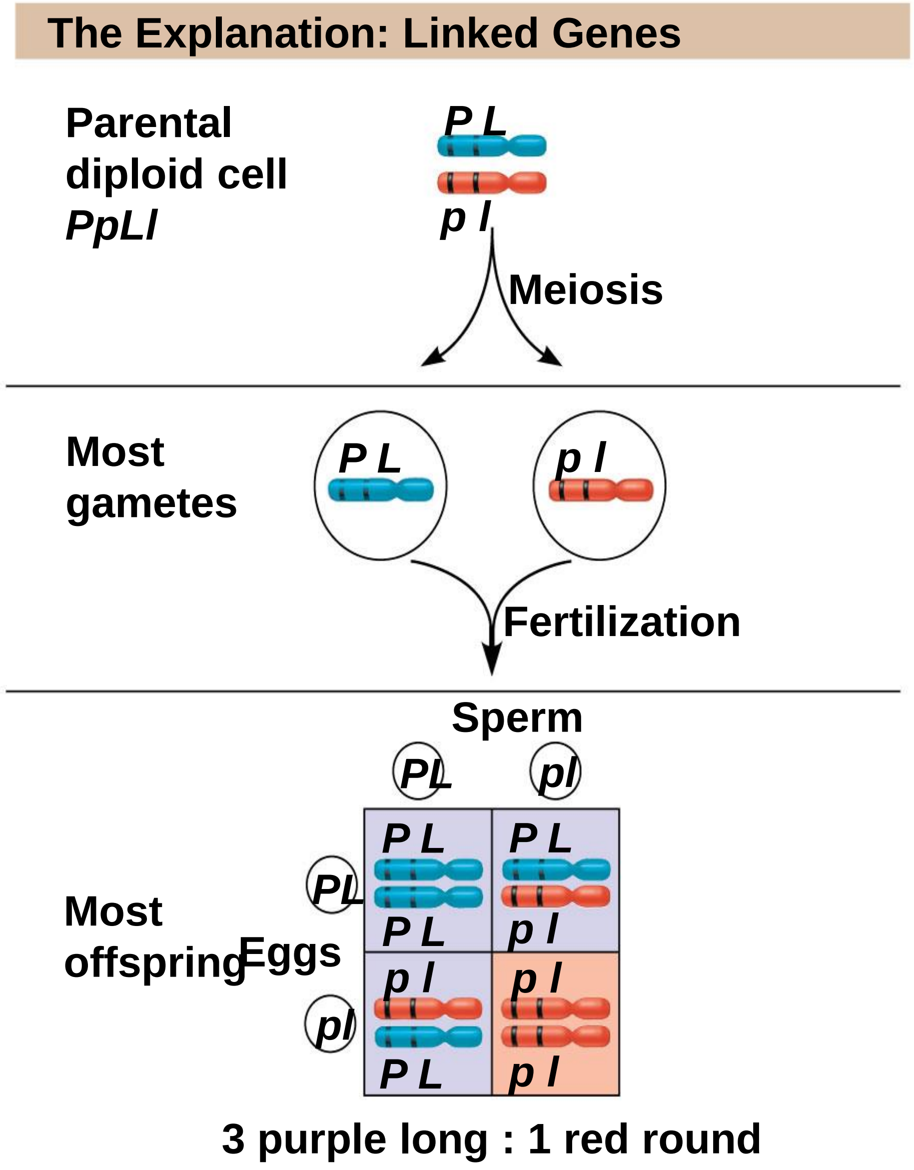
The Experiment



Phenotypes	Observed offspring	Prediction (9:3:3:1)
Purple long	284	215
Purple round	21	71
Red long	21	71
Red round	55	24

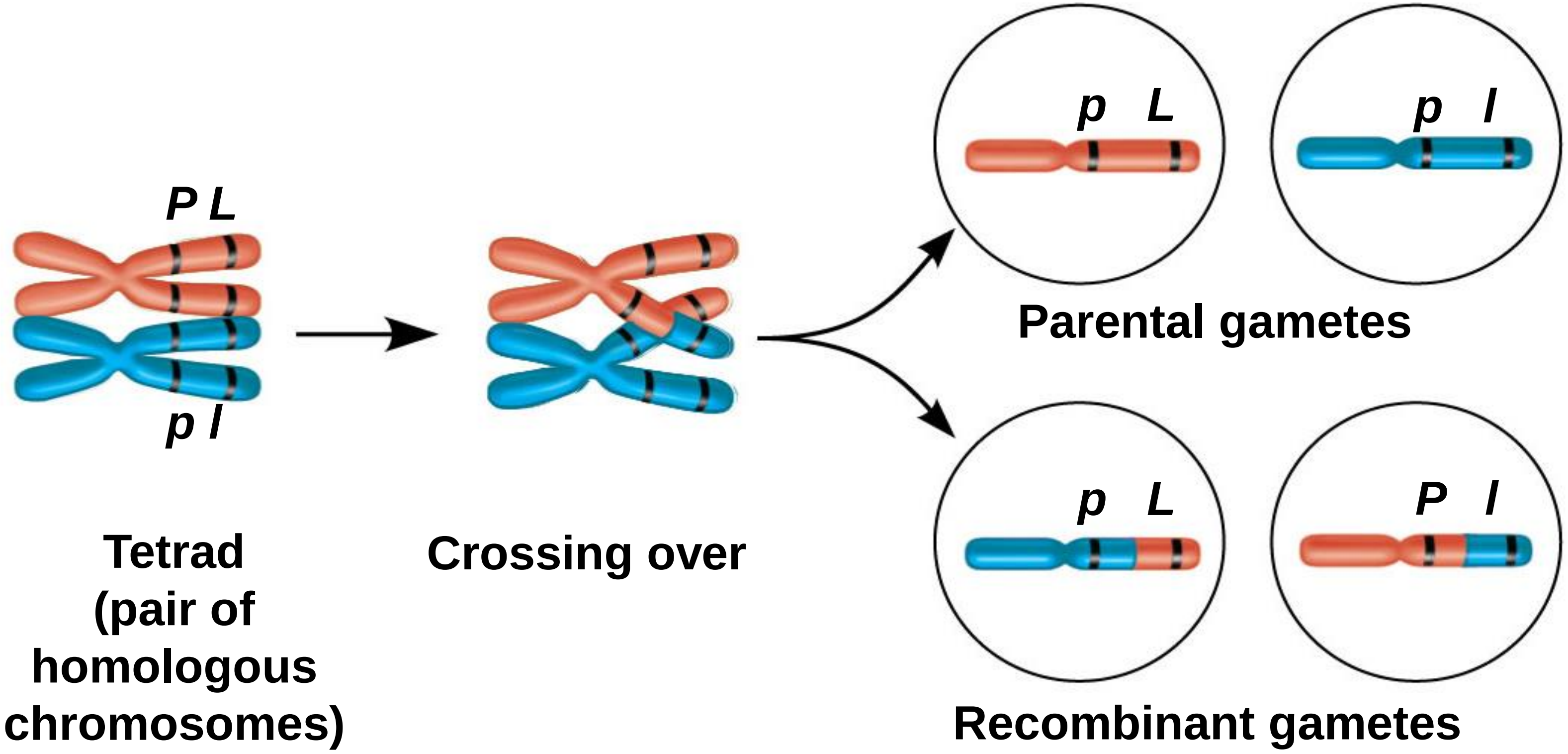
Genes on the same chromosome tend to be inherited together

- Not accounted for: purple round and red long



Crossing over produces new combinations of alleles

- Crossing over between homologous chromosomes produces new combinations of alleles in gametes.
  - Linked alleles can be separated by crossing over, forming recombinant gametes.
  - The percentage of recombinants is the recombination frequency.

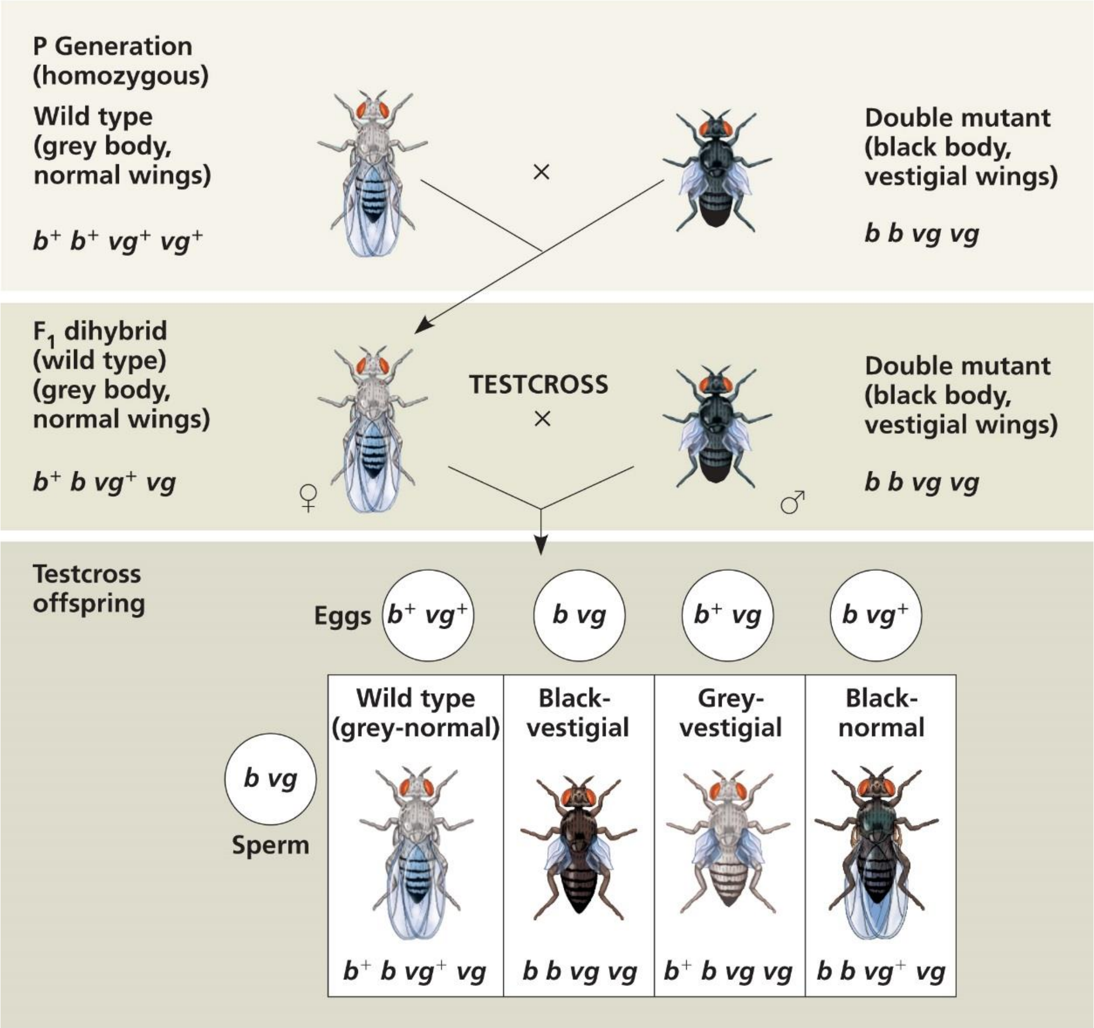


Drosophila melanogaster

- Fruit fly
  - Model organism for animal genetics
  - Compared to Mendel’s peas
  - Used to test linkage and recombination

Linked Genes

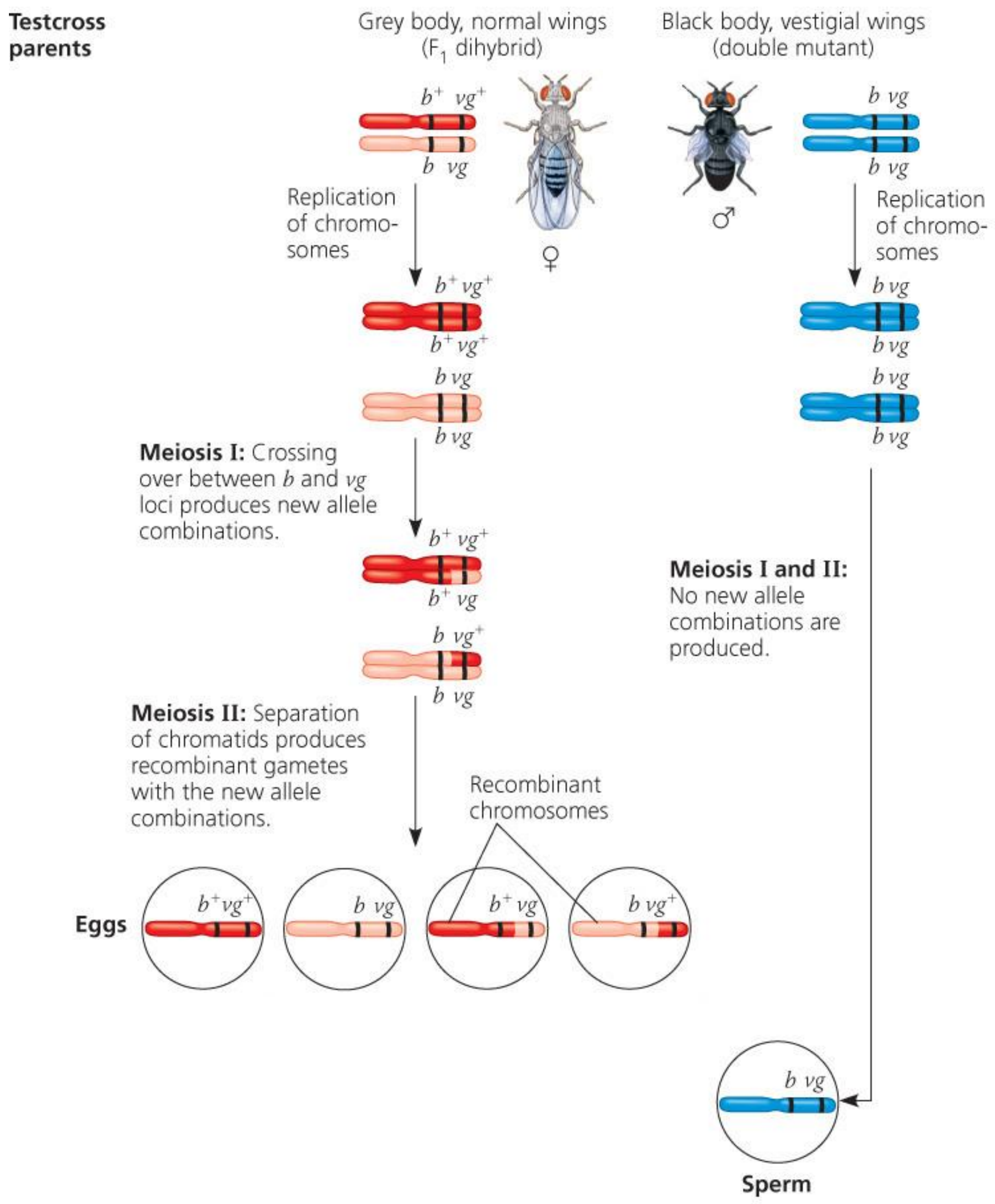
Experiment



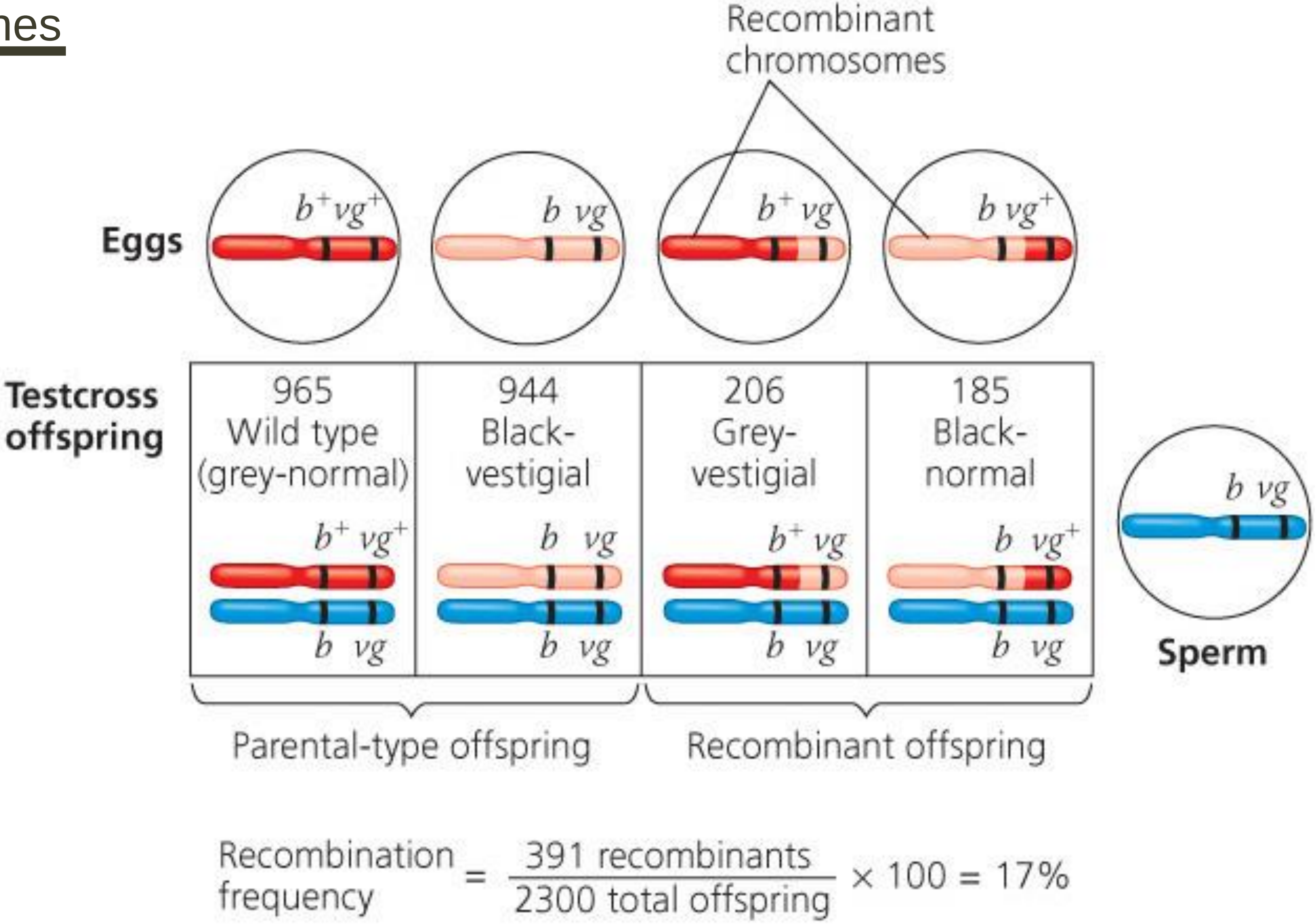
PREDICTED RATIOS

If genes are located on different chromosomes:	1	:	1	:	1	:	1
If genes are located on the same chromosome *and* parental alleles are always inherited together:	1	:	1	:	0	:	0
Results	965	:	944	:	206	:	185

Linked Genes and recombination



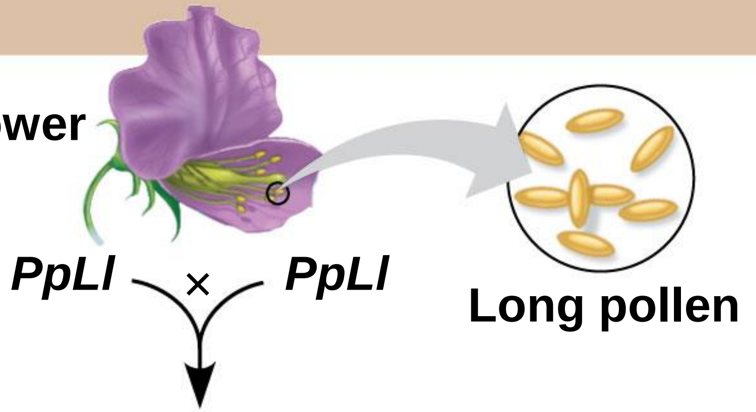
Linked Genes



Linked Genes and recombination

- What is the recombination frequency between the genes for flower color and pollen shape?

The Experiment



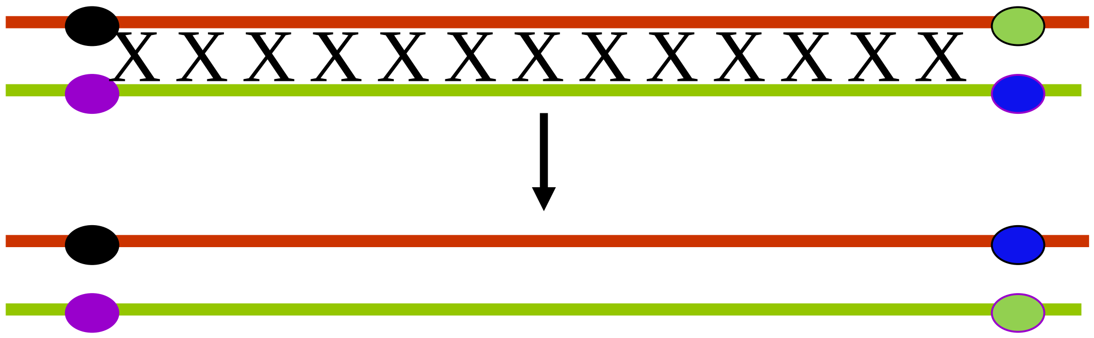
Phenotypes	Observed offspring	Prediction (9:3:3:1)
Purple long	284	215
Purple round	21	71
Red long	21	71
Red round	55	24

Recombination frequency = 42 (recombinant) / 381 (parental) *100 = 11%

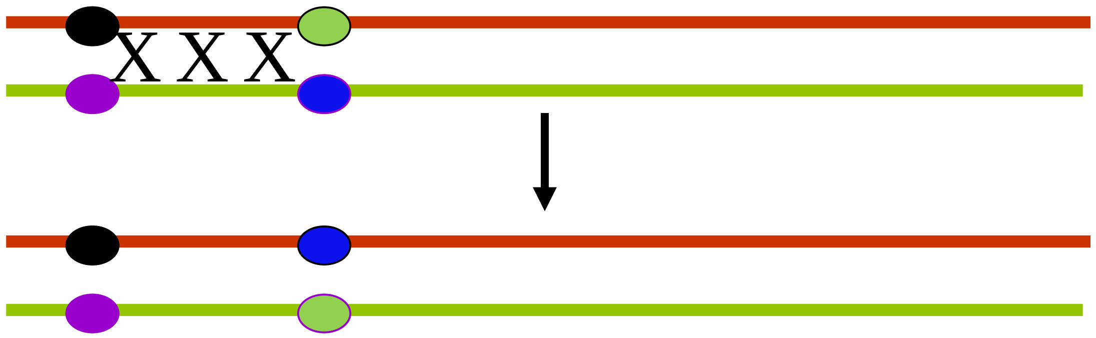
Recombination Frequency

- When examining recombinant frequency, Morgan and his students found that the greater the distance between two genes on a chromosome, the more points there are between them where crossing over can occur.

Greater distance: more potential sites of recombination……higher recombination frequency

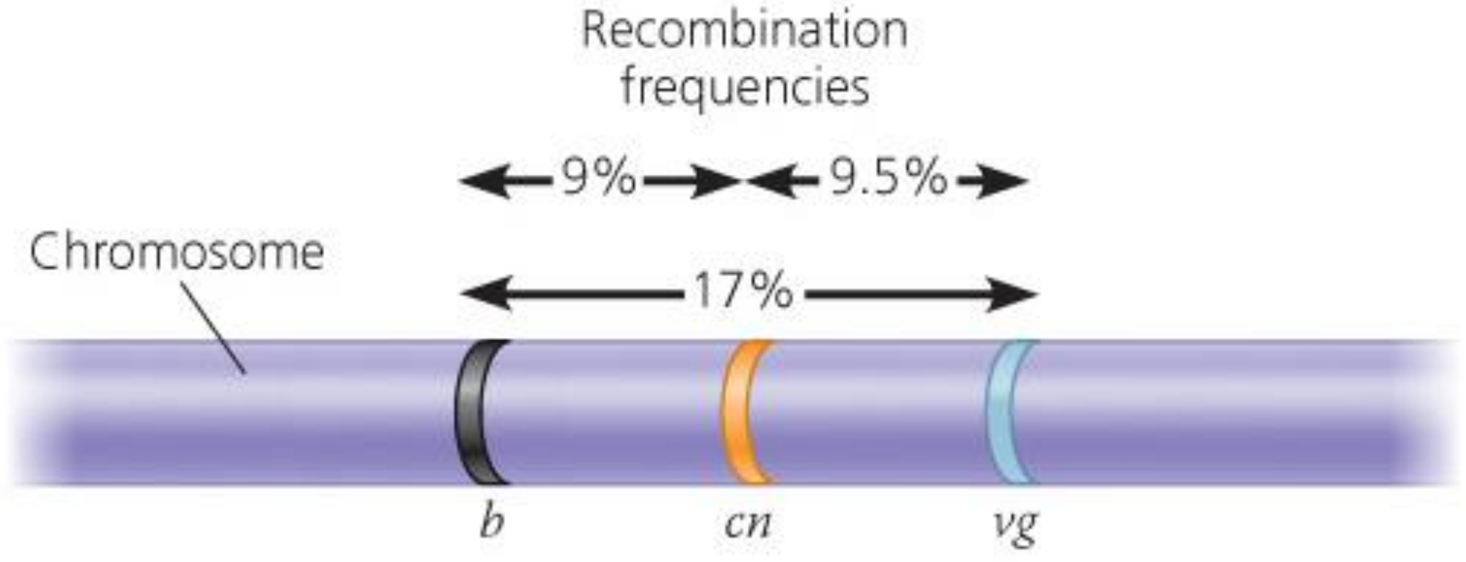


Lesser distance: fewer potential sites of recombination………lower recombination frequency



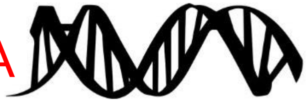
Geneticists use crossover data to map genes

- Recombination frequencies used to determine relative locations on a chromosome
  - Linkage map for genes *b*, *cn*, and *vg*:

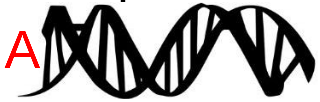


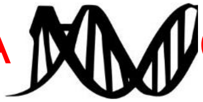
- 1 map unit = 1% recombination = 1 centimorgan

Linkage – genes on the same chromosome

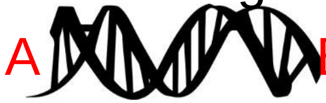
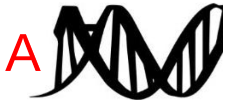
- Example: Genes A, B and C
- RF A → B = 30%
  - So A  B is 30 map units

- RF B → C = 10%
  - So B  C is 10 map units

  - Two possible arrangements...
  - A  B  C or...

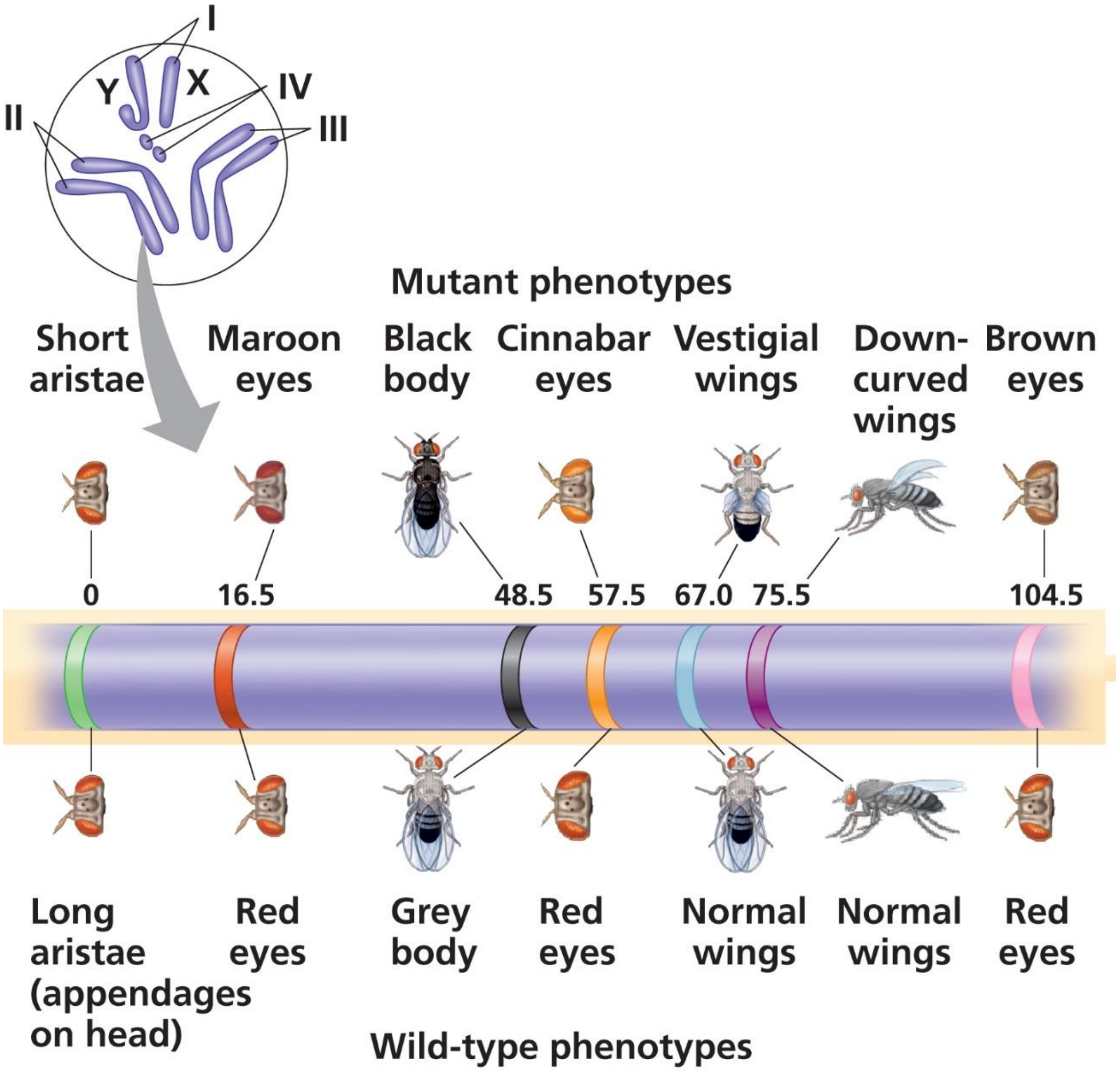
  - A  C  B

Linkage – genes on the same chromosome

- Example: Genes A, B and C
- RF B → C = 10%
  - So B  C is 10 map units
  - Two possible arrangements...
  - Option 1: A  B  C or...
  - Option 2: A  C  B
- So we test the RF between A → C
  - If RF = 40%/40 map units then option 1.
  - If RF = 20%/20 map units then option 2.

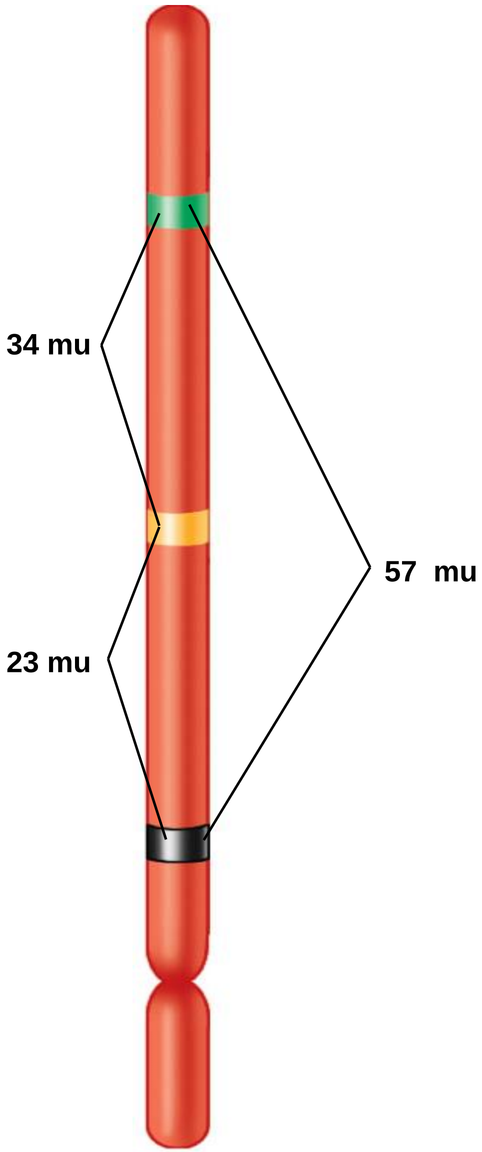
Geneticists use crossover data to map genes

- Recombination frequencies used to determine relative locations on a chromosome



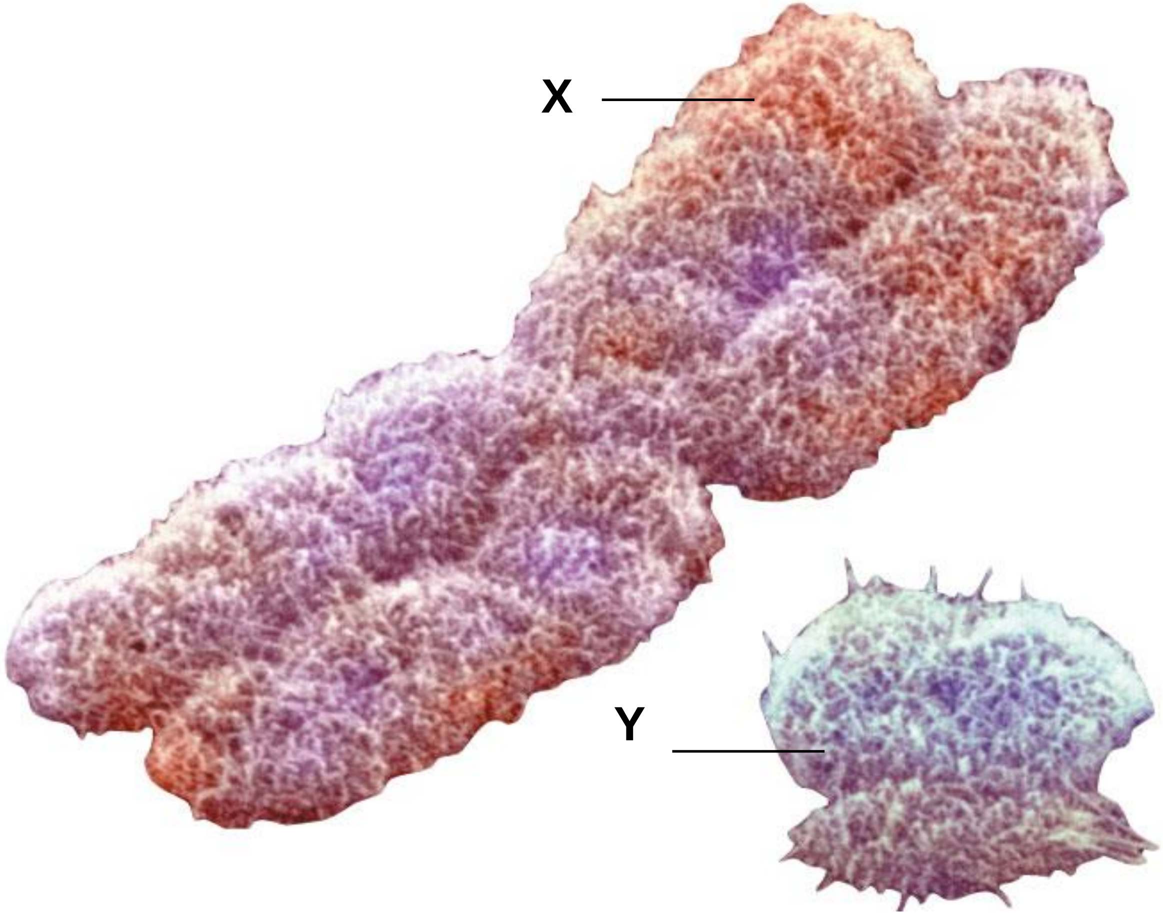
Recombination occurs often

- Widely separated linked genes often recombine
  - Seem to assort independently
  - Detected by testing linkage to genes between them



Sex-Linked Genes

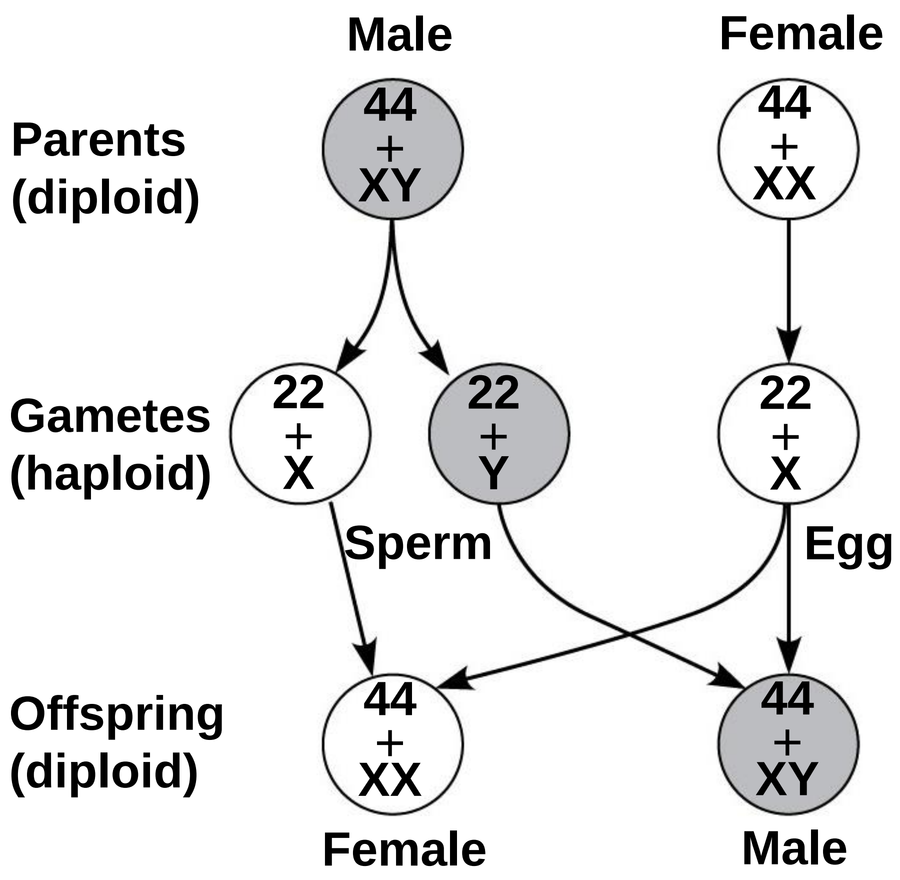
- *Many animals have a pair of sex chromosomes,*
  - *designated X and Y,*
  - *determine an individual's sex.*



Sex-Linked Genes

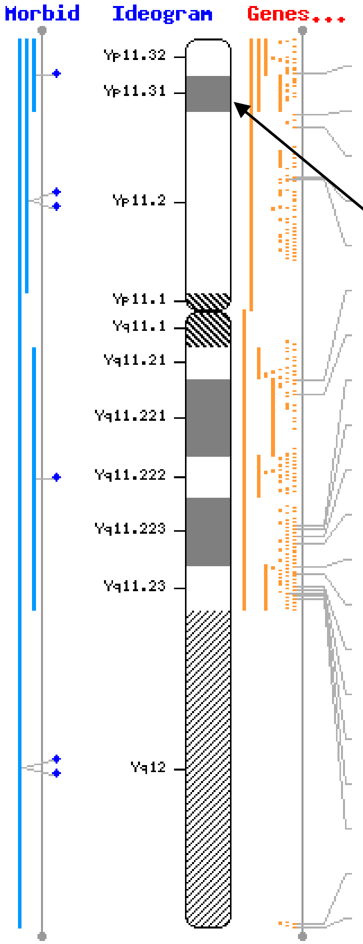
- *In mammals,*
  - *males have XY sex chromosomes,*
  - *females have XX sex chromosomes,*

- *Females are XX and males are XY in both humans and fruit flies*



Human sex determination depends on the SRY gene

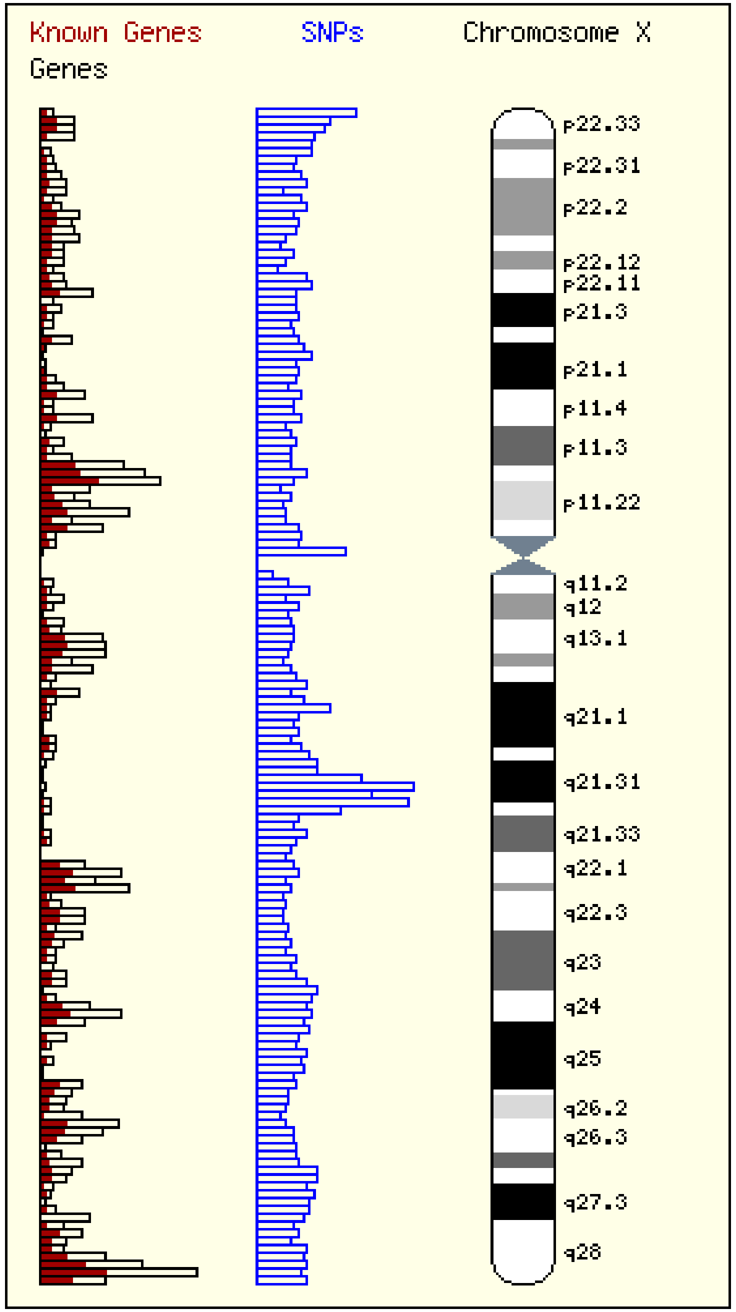
- **Y chromosome length:**
  - 57,772,954 bps
- **Known protein-coding genes:**
  - 73



SRY (TDF) : testes determining factor

The human X chromosome

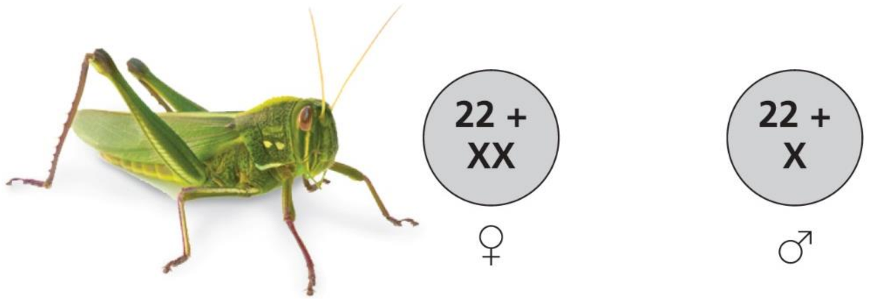
- **X chromosome length:**
  - 154,913,754 bps
- **Known protein-coding genes:**
  - 1788



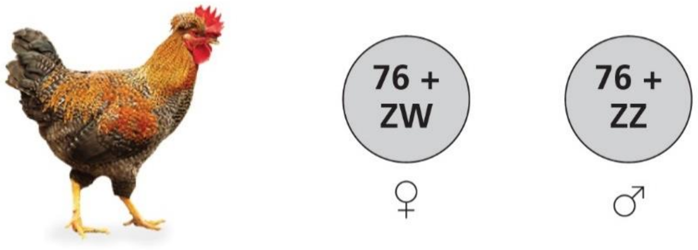
http://www.ensembl.org/Homo_sapiens/mapview?chr=X

Chromosomes determine sex in many species

- Grasshoppers, roaches, and some other insects have an X-O system
  - O stands for the absence of a sex chromosome,
  - females are XX, and
  - males are XO.

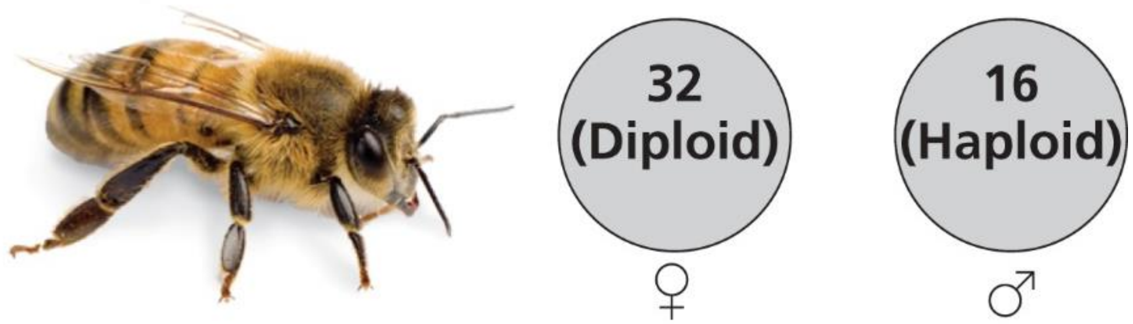


- In certain fishes, butterflies, and birds,
  - the sex chromosomes are Z and W,
  - males are ZZ, and
  - females are ZW.



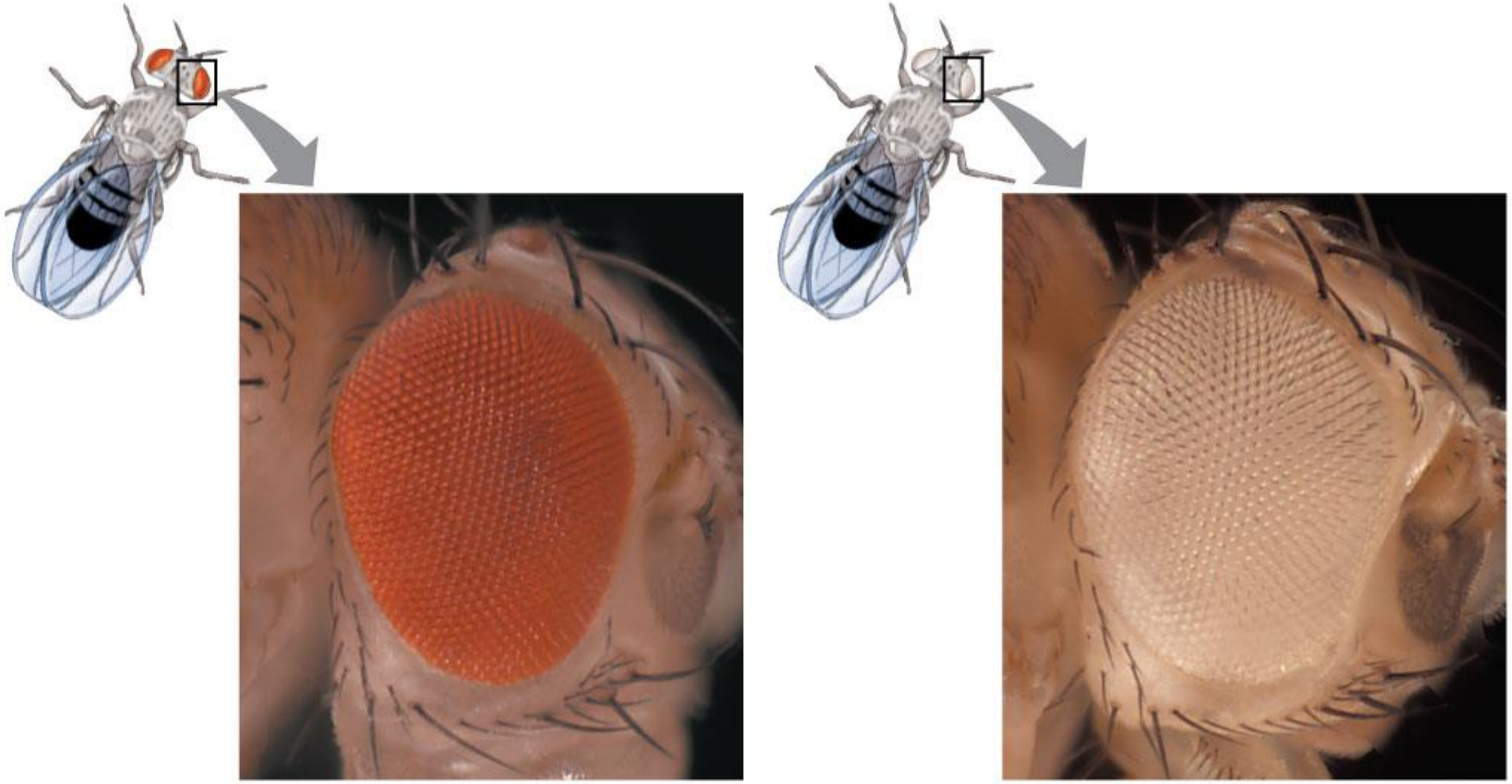
Chromosomes determine sex in many species

- Some organisms lack sex chromosomes altogether.
  - In bees, sex is determined by chromosome number.
  - Females are diploid.
  - Males are haploid.

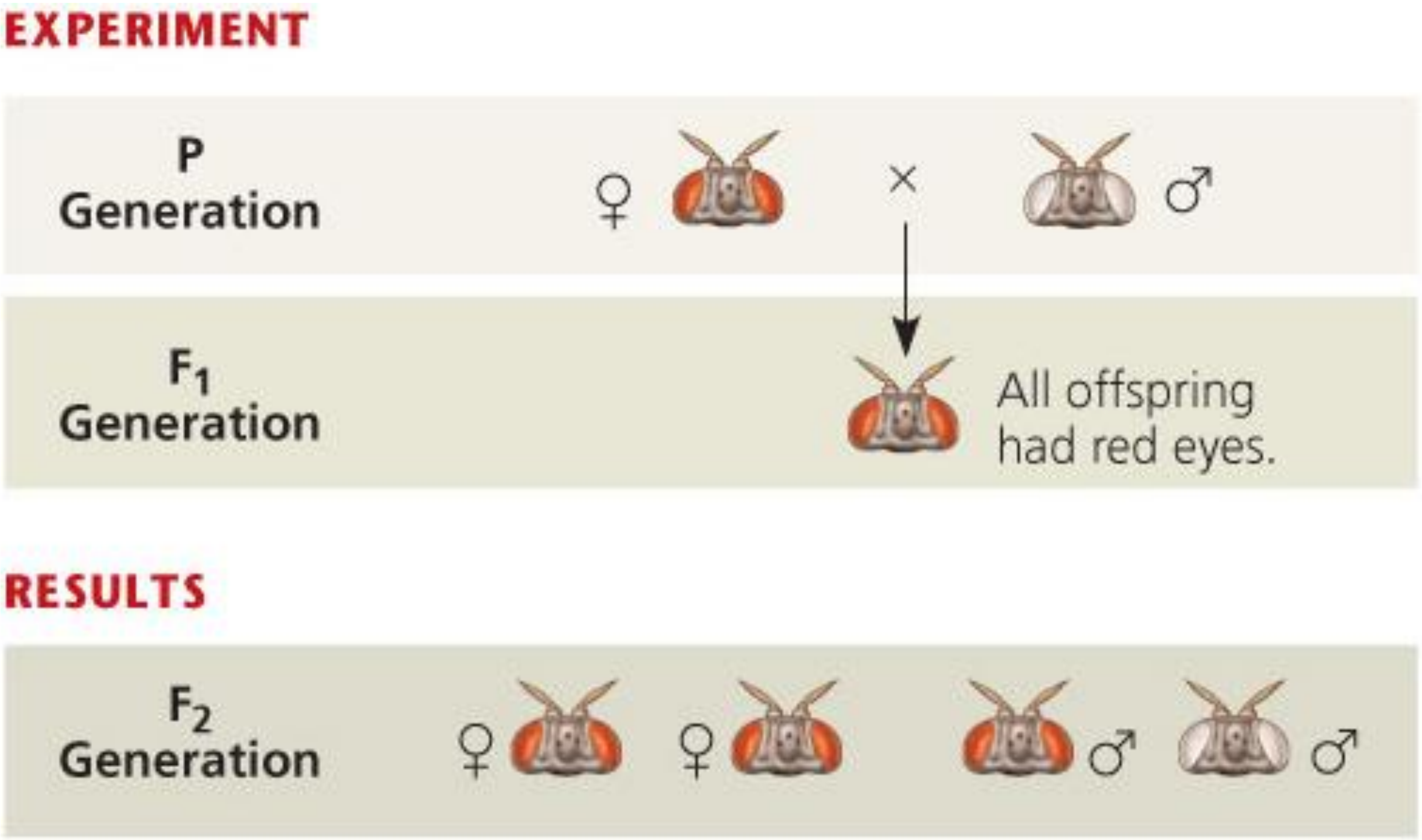


Sex-linked genes exhibit a unique pattern of inheritance

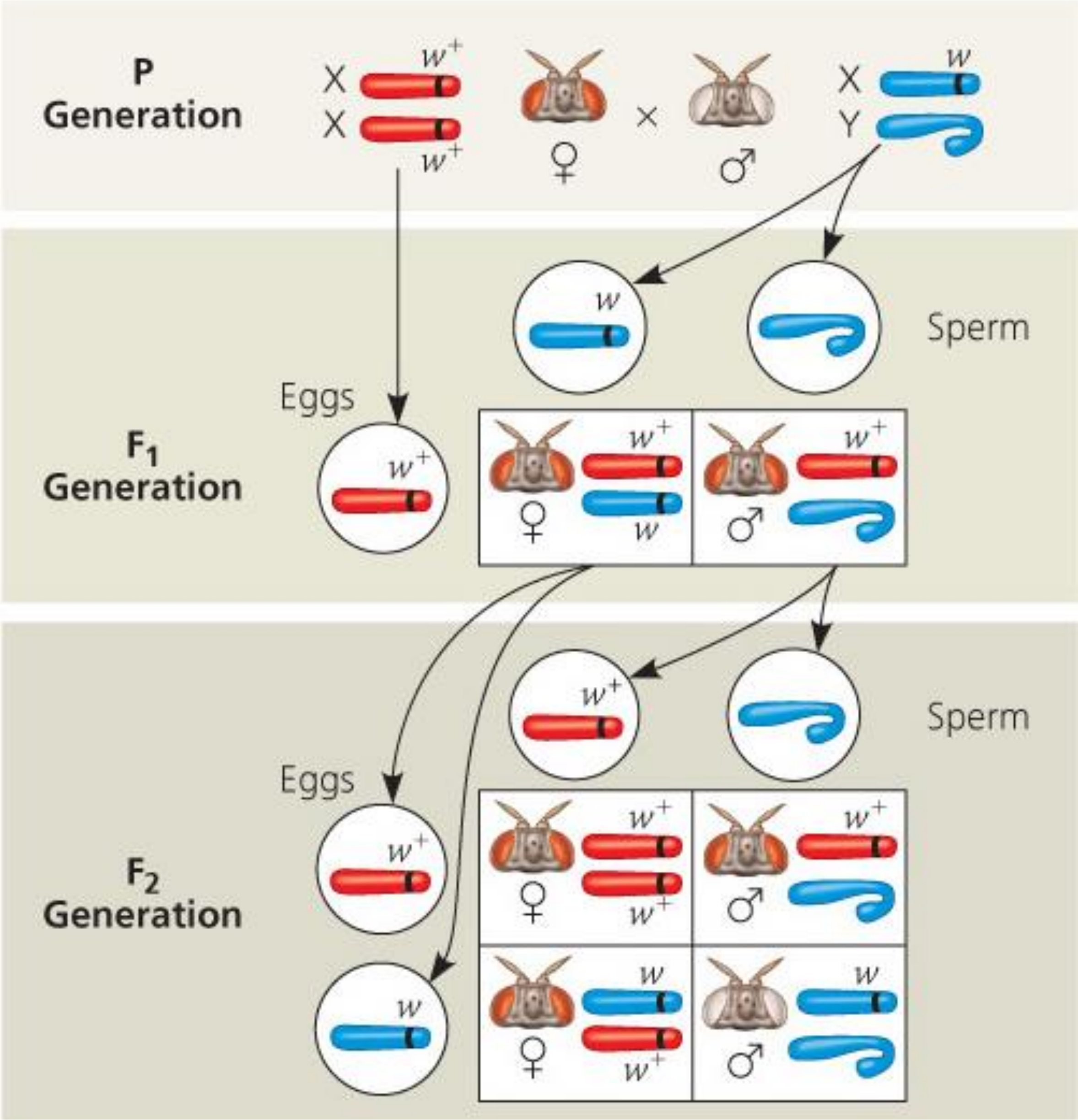
- Sex-linked genes are located on either of the sex chromosomes.
  - The X chromosome carries many genes unrelated to sex, for example eye colour.
  - The inheritance of white eye color in the fruit fly illustrates an X-linked recessive trait.



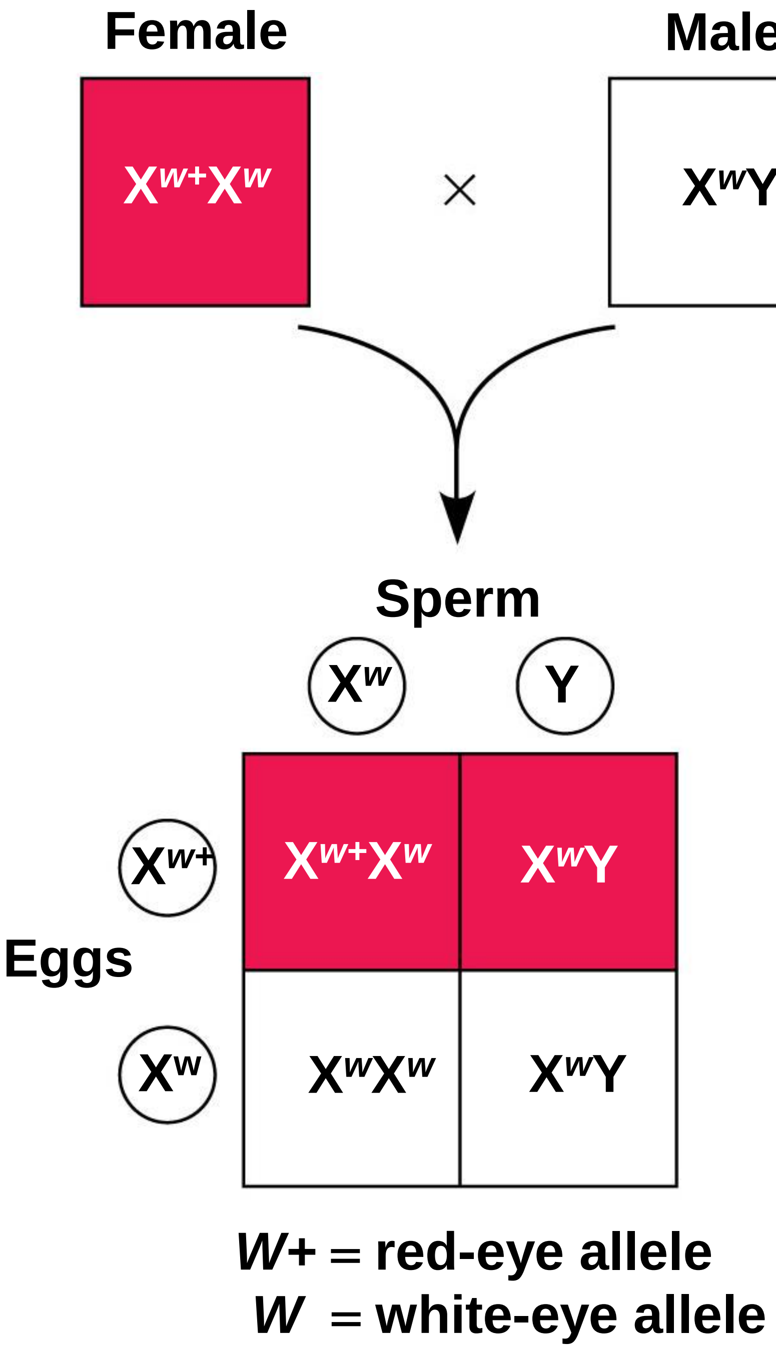
Evidence for Sex-Linked Genes



Evidence for Sex-Linked Genes



Evidence for Sex-Linked Genes



Human sex-linked disorders affect mostly males

- Most sex-linked human disorders are
  - due to recessive alleles and
  - seen mostly in males.

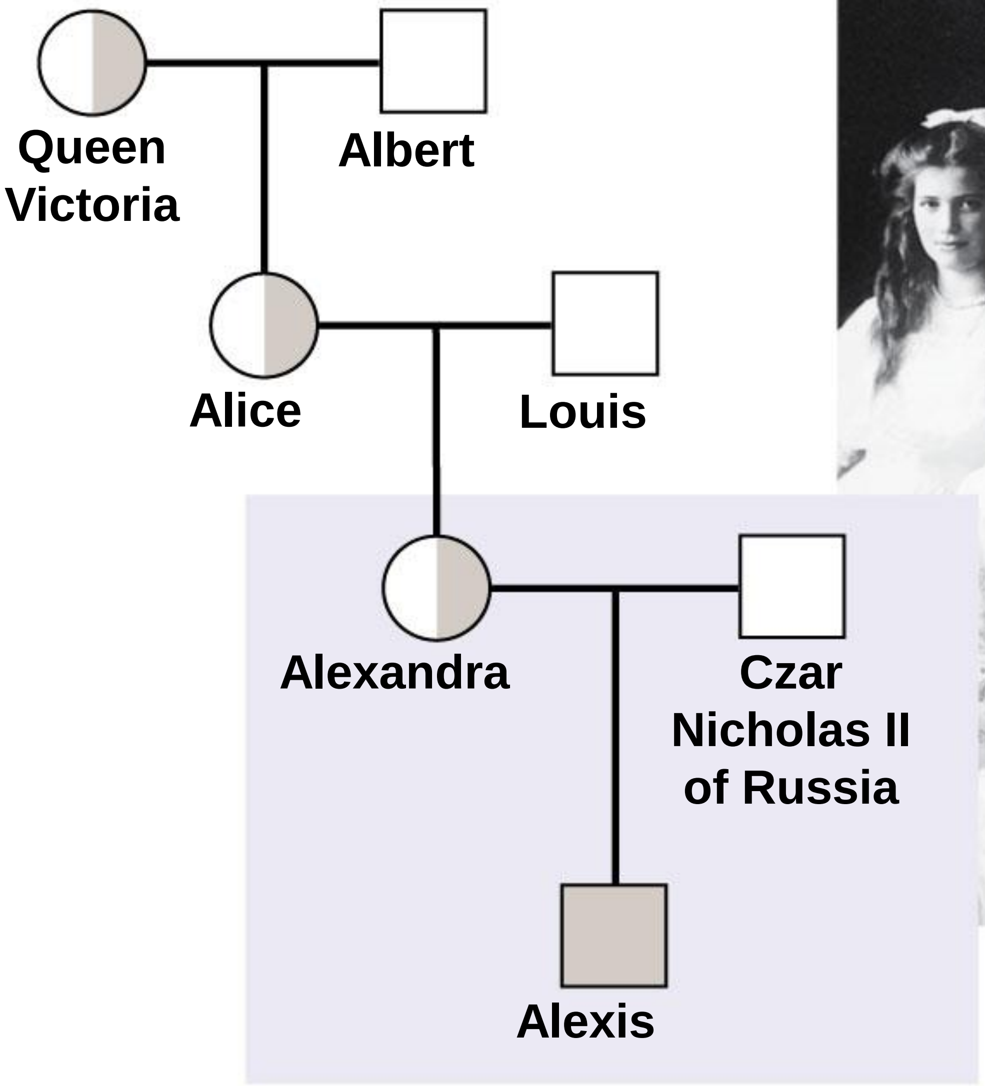
- A male receiving a single X-linked recessive allele from his mother will have the disorder.

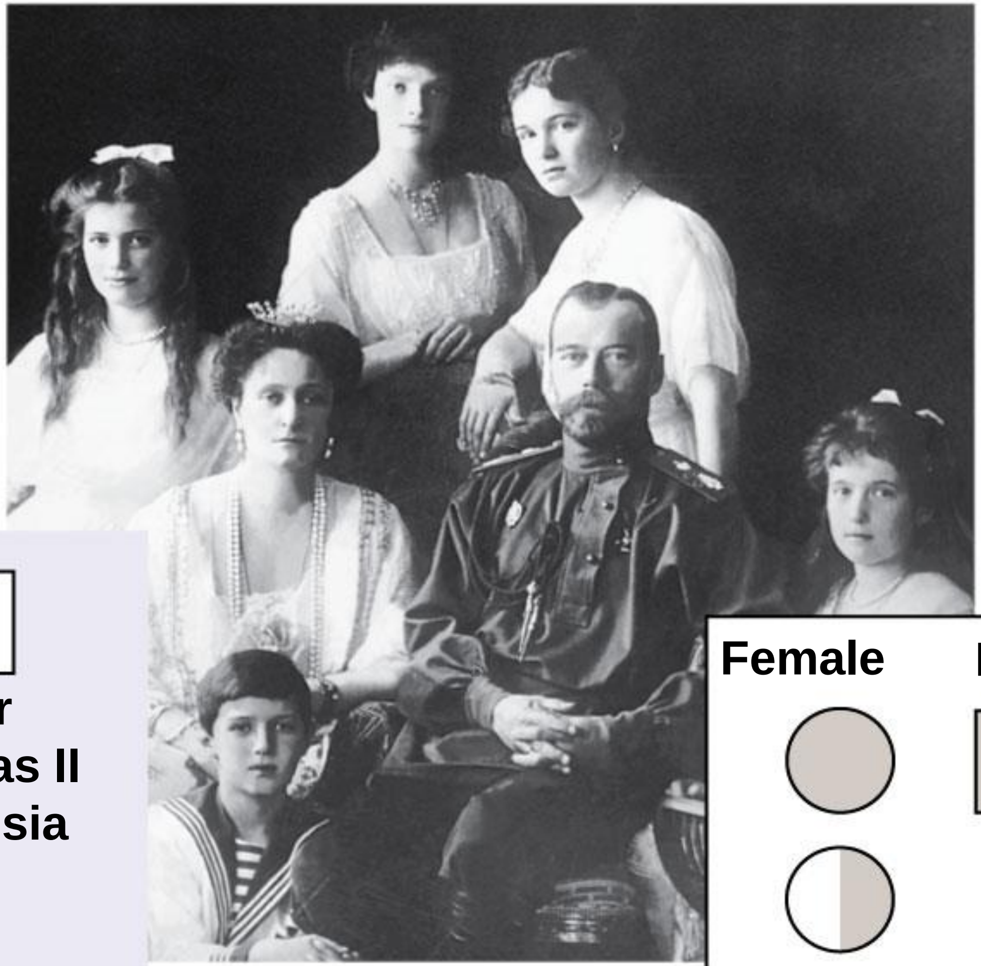
- A female must receive the allele from both parents to be affected.

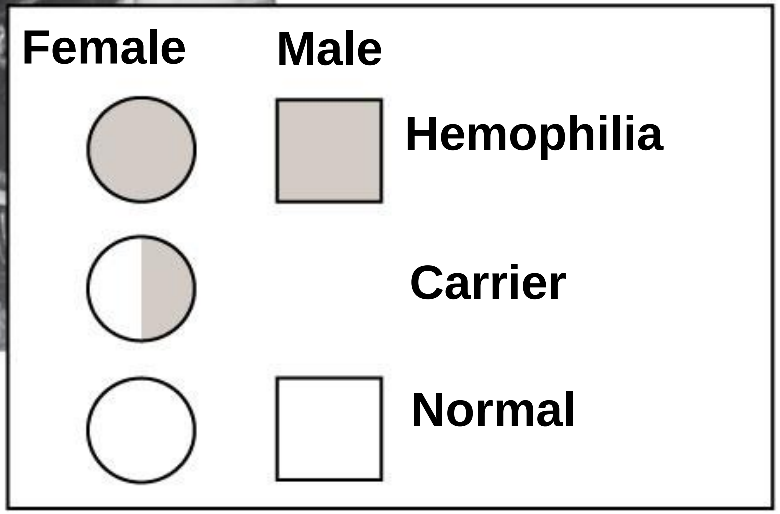
Human sex-linked disorders affect mostly males

- Recessive and sex-linked human disorders include
  - hemophilia, characterized by excessive bleeding because hemophiliacs lack one or more of the proteins required for blood clotting
  - red-green color blindness, a malfunction of light-sensitive cells in the eyes
  - Duchenne muscular dystrophy, a condition characterized by a progressive weakening of the muscles and loss of coordination.

Inheritance of Hemophilia

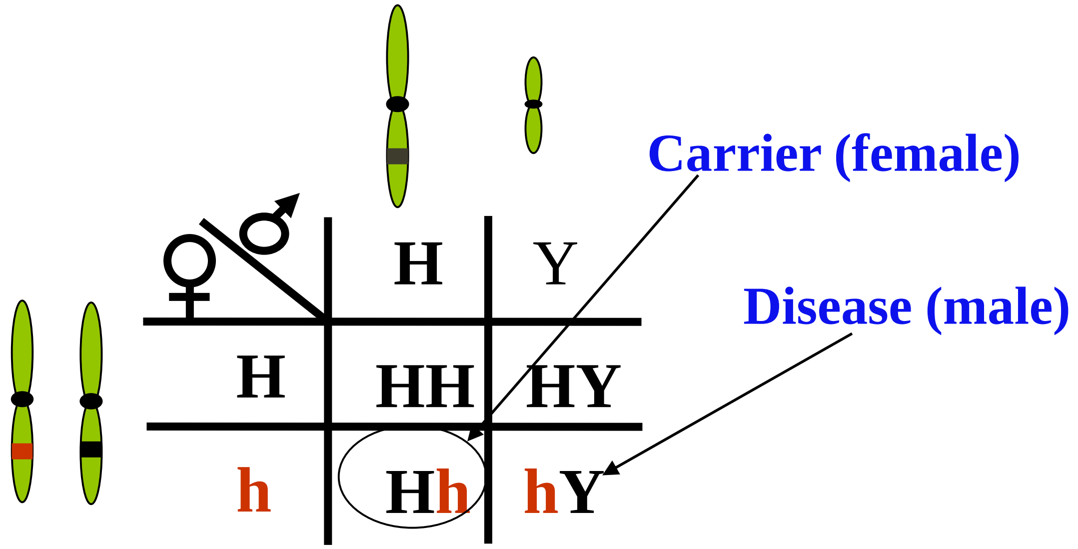


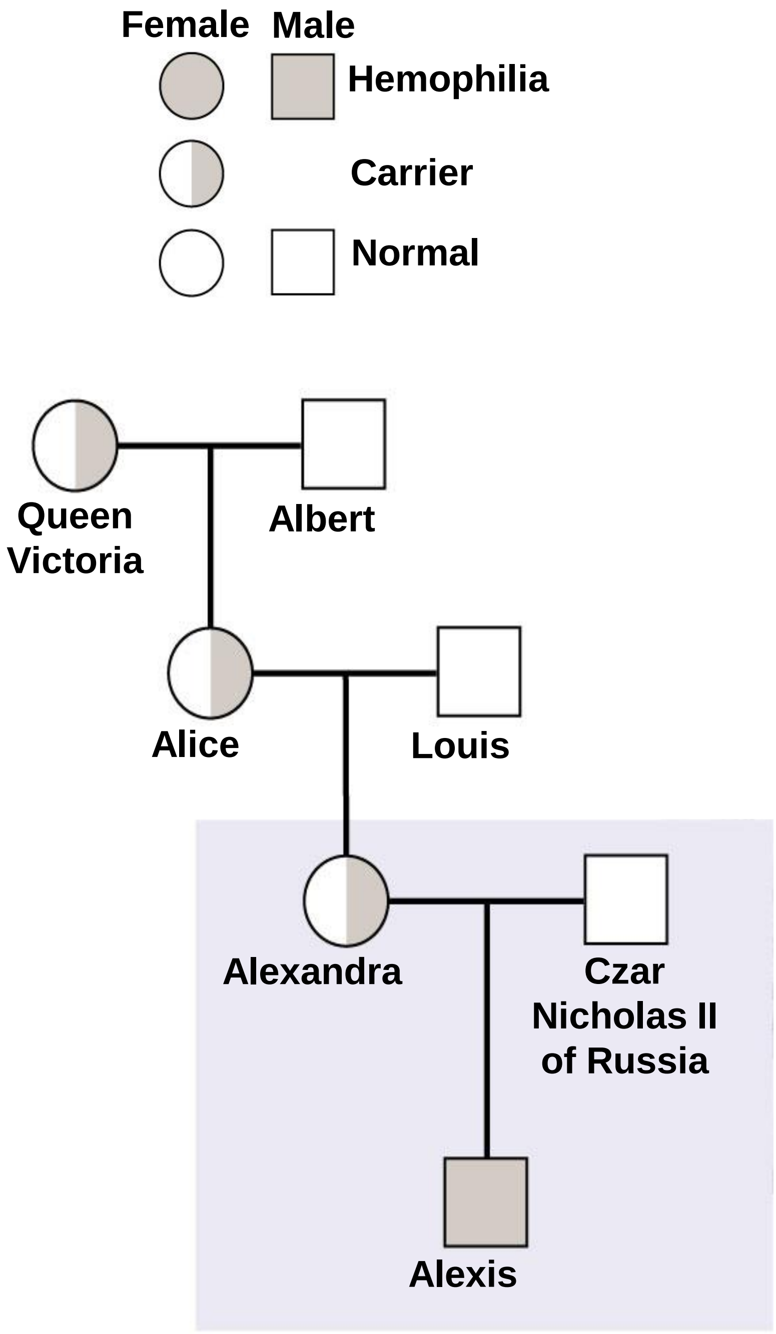




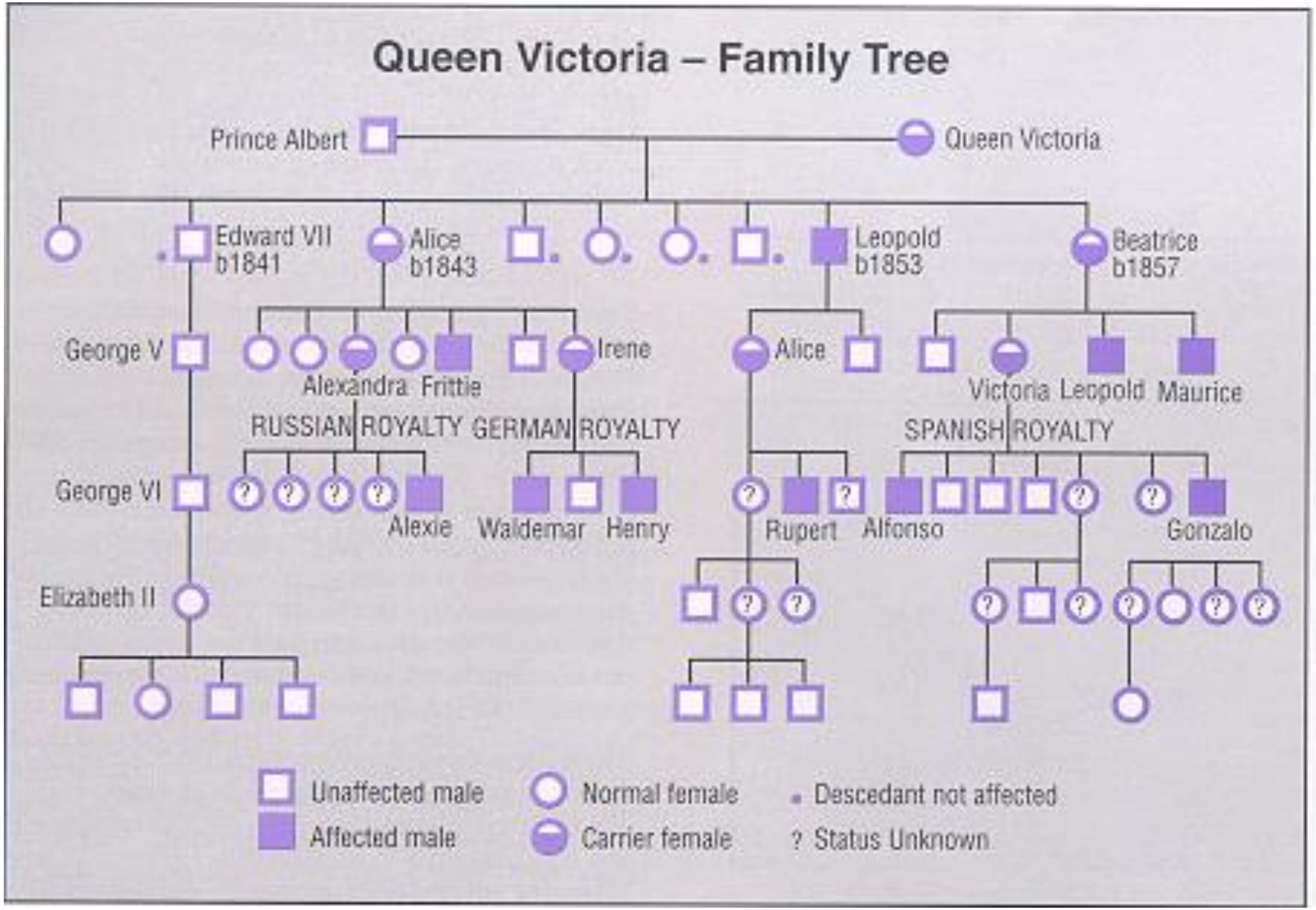
Inheritance of Hemophilia

- Recessive allele carried on X chromosome
  - Males show symptoms
  - Female heterozygous carriers - no symptoms
  - Homozygous - show symptoms



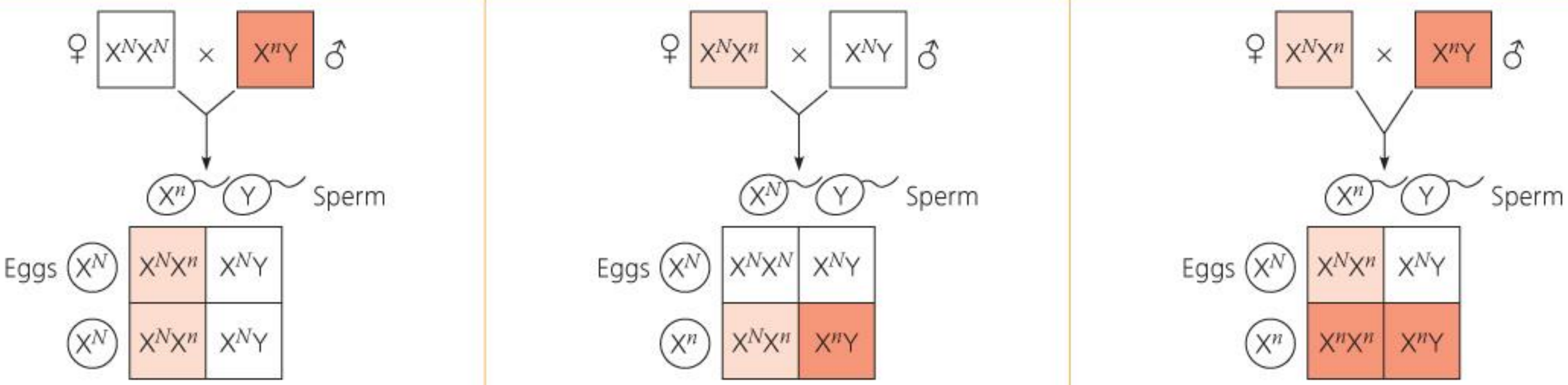


Inheritance of Hemophilia



http://www.hemophilia.org/NHFWeb/MainPgs/MainNHF.aspx?menuid=178&contentid=6

Sex-linked traits – X linked recessive

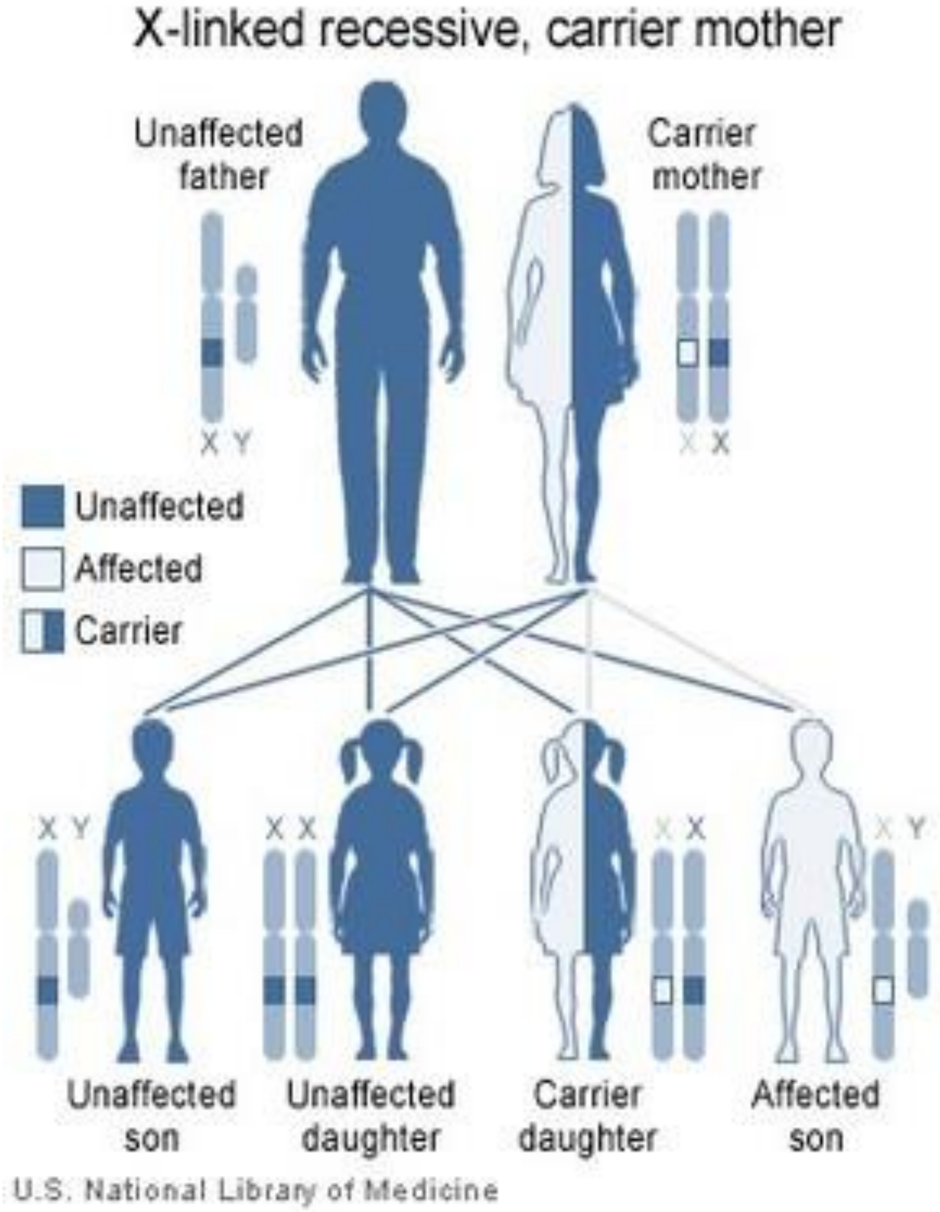


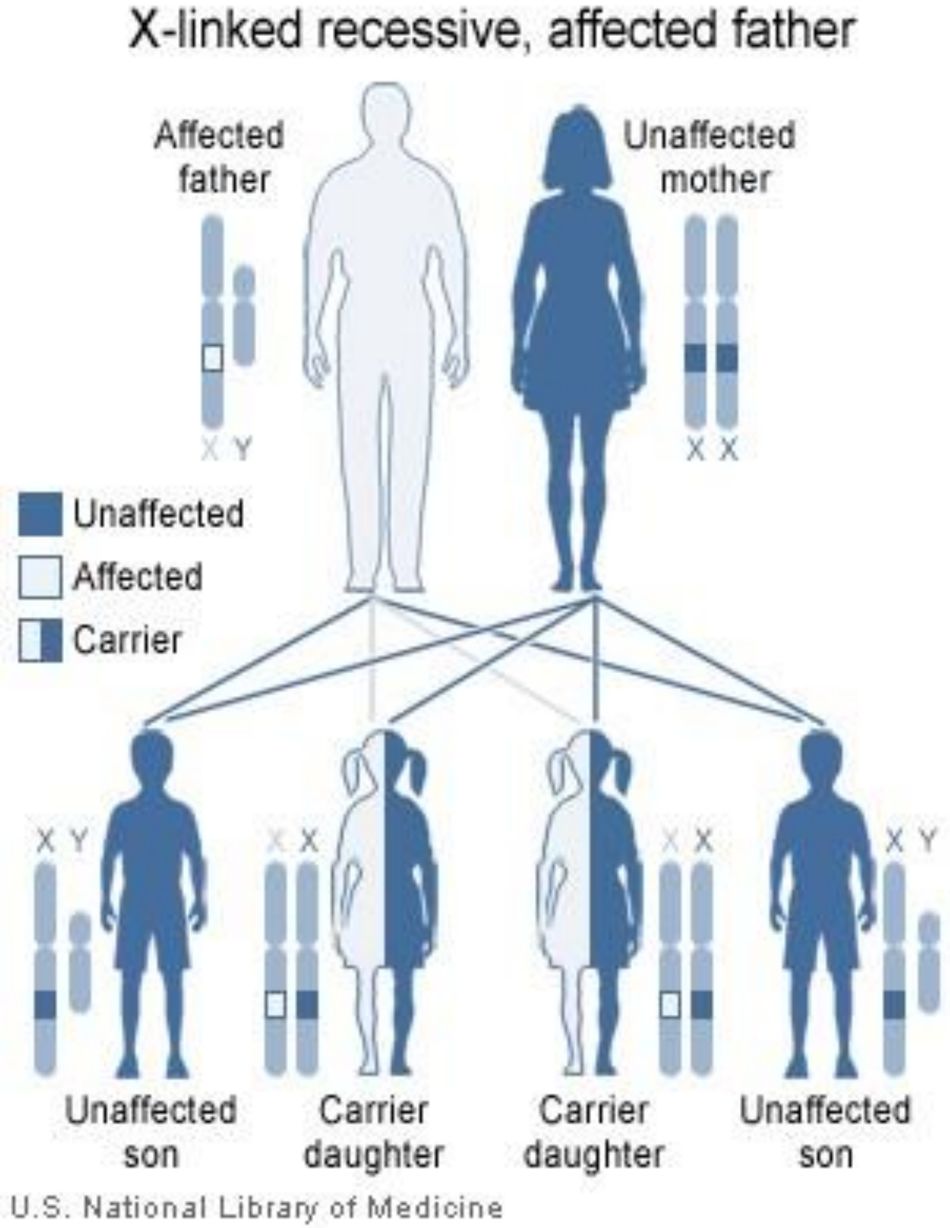
(a) A colour-blind father will transmit the mutant allele to all daughters but to no sons. When the mother is a dominant homozygote, the daughters will have the normal phenotype but will be carriers of the mutation.

(b) If a carrier mates with a male who has normal colour vision, there is a 50% chance that each daughter will be a carrier like her mother and a 50% chance that each son will have the disorder.

(c) If a carrier mates with a colour-blind male, there is a 50% chance that each child born to them will have the disorder, regardless of sex. Daughters who have normal colour vision will be carriers, whereas males who have normal colour vision will be free of the recessive allele.

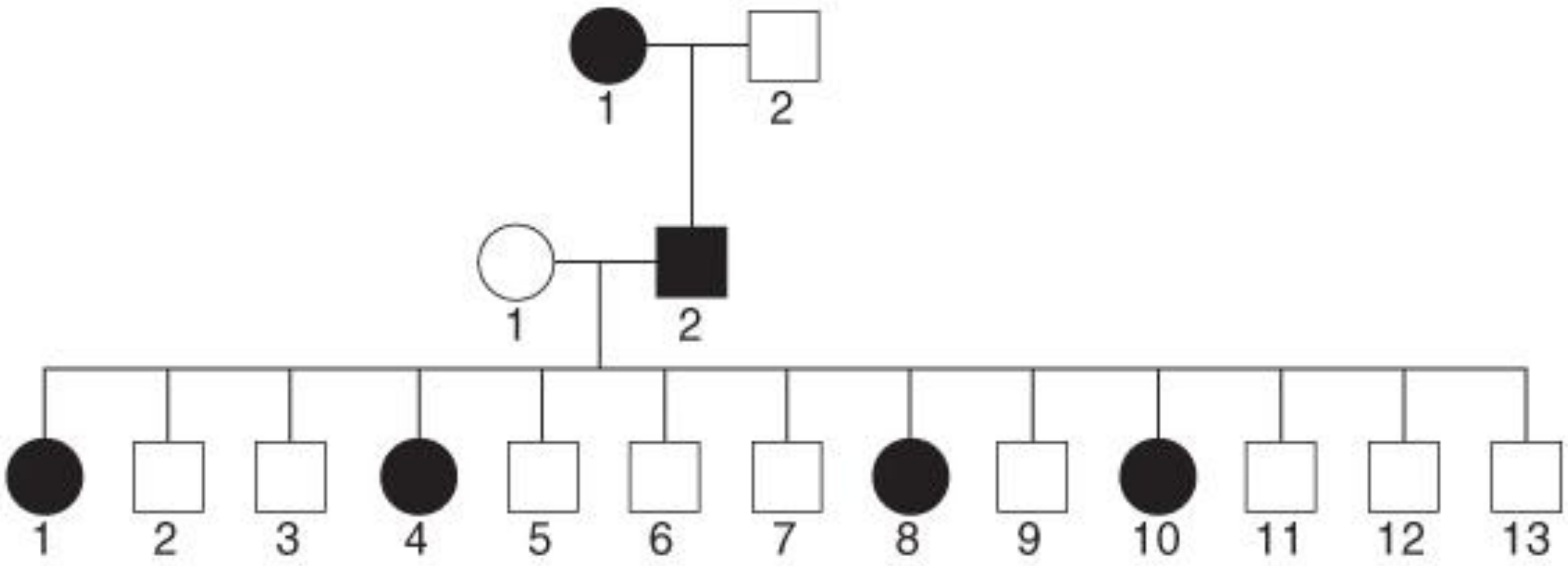
Sex-linked traits – X linked recessive





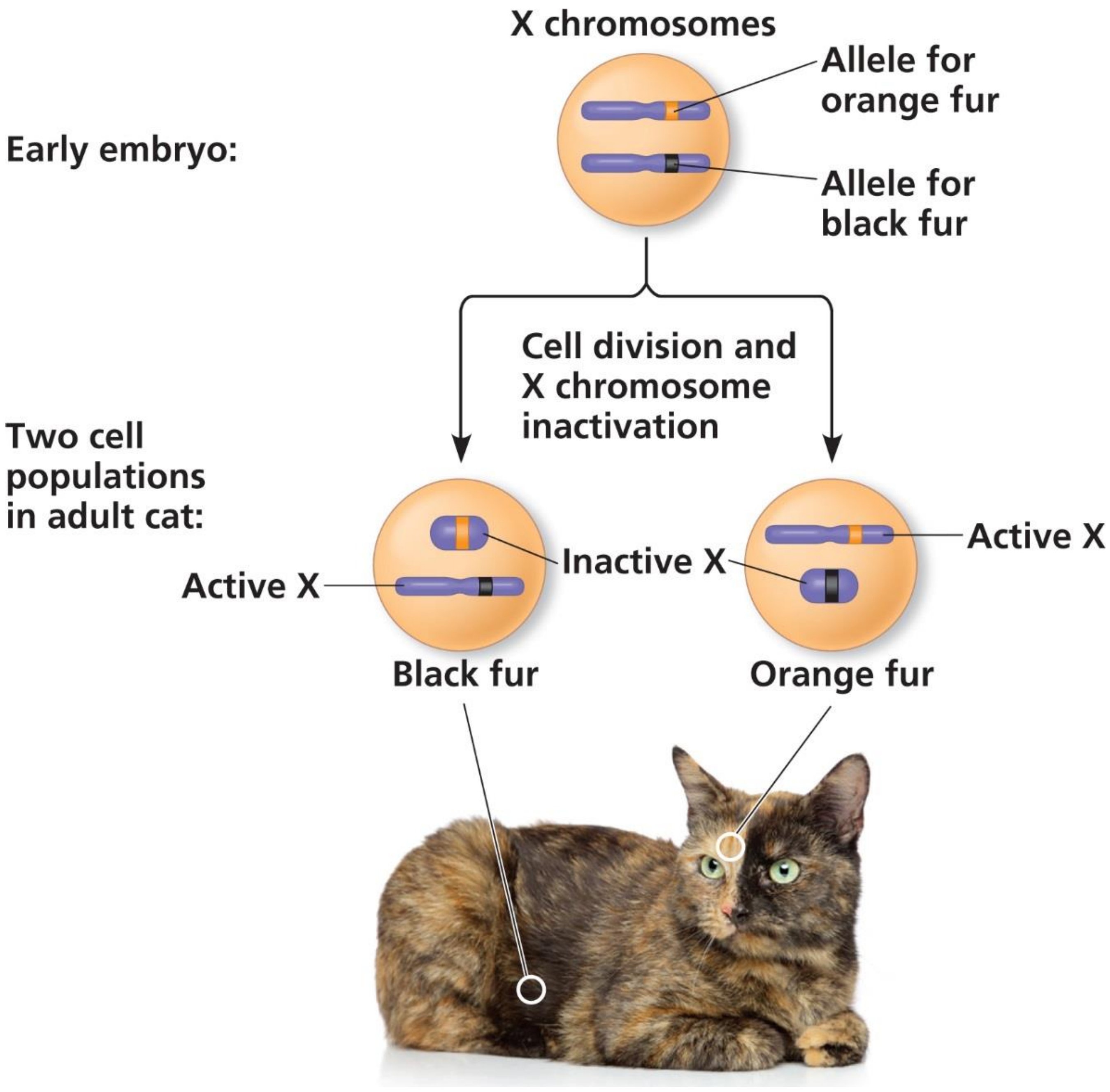
X-linked Dominant Traits

- Congenital generalized hypertrichosis



X chromosome inactivation

- In female mammals, one of the two X chromosomes is highly compacted and transcriptionally inactive (called a Barr body).
  - Either the maternal or paternal chromosome is randomly inactivated.
  - *XIST*, a long nc-RNA is a major effector of X inactivation.



Non-traditional patterns of Inheritance

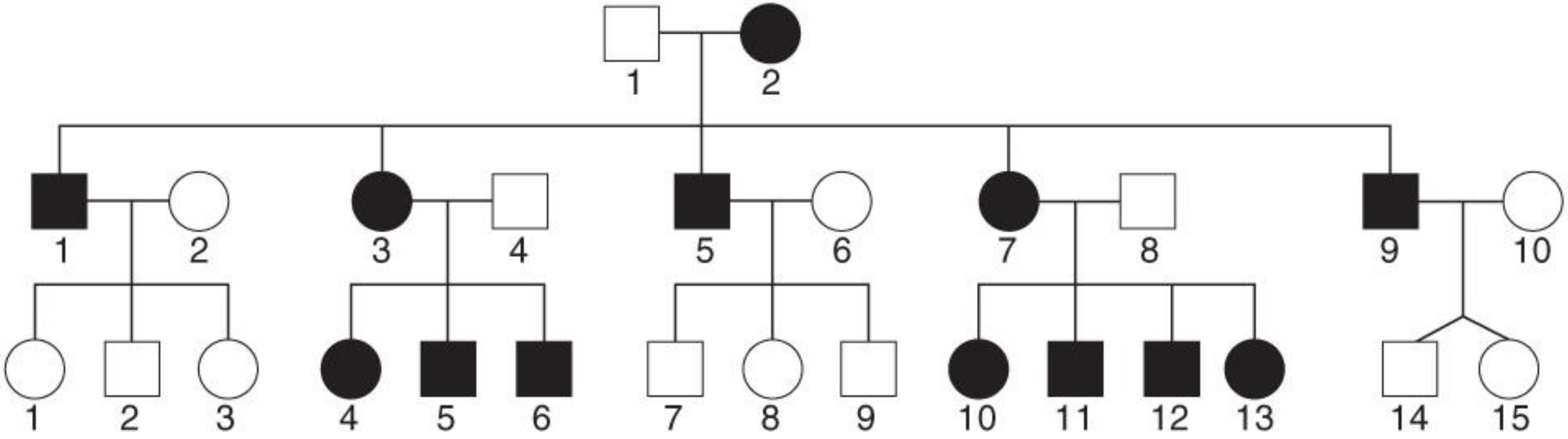
- Cytoplasmic inheritance follows the pattern of inheritance of mitochondria or chloroplasts

- Cytoplasmic inheritance follows the maternal line
  - Zygote’s cytoplasm originates from egg cell

- Mutant alleles in organelle DNA
  - Mendelian inheritance not followed (no segregation by meiosis)
  - Uniparental inheritance from female

Non-traditional patterns of Inheritance

- Mitochondrial genes are transmitted from mother to all of her offspring



https://www.genomicseducation.hee.nhs.uk/modes-of-inheritance/mitochondrial-conditions

- Ooplasmic transfer technique can enable woman to avoid transmitting a mitochondrial disorder.

Non-traditional patterns of Inheritance

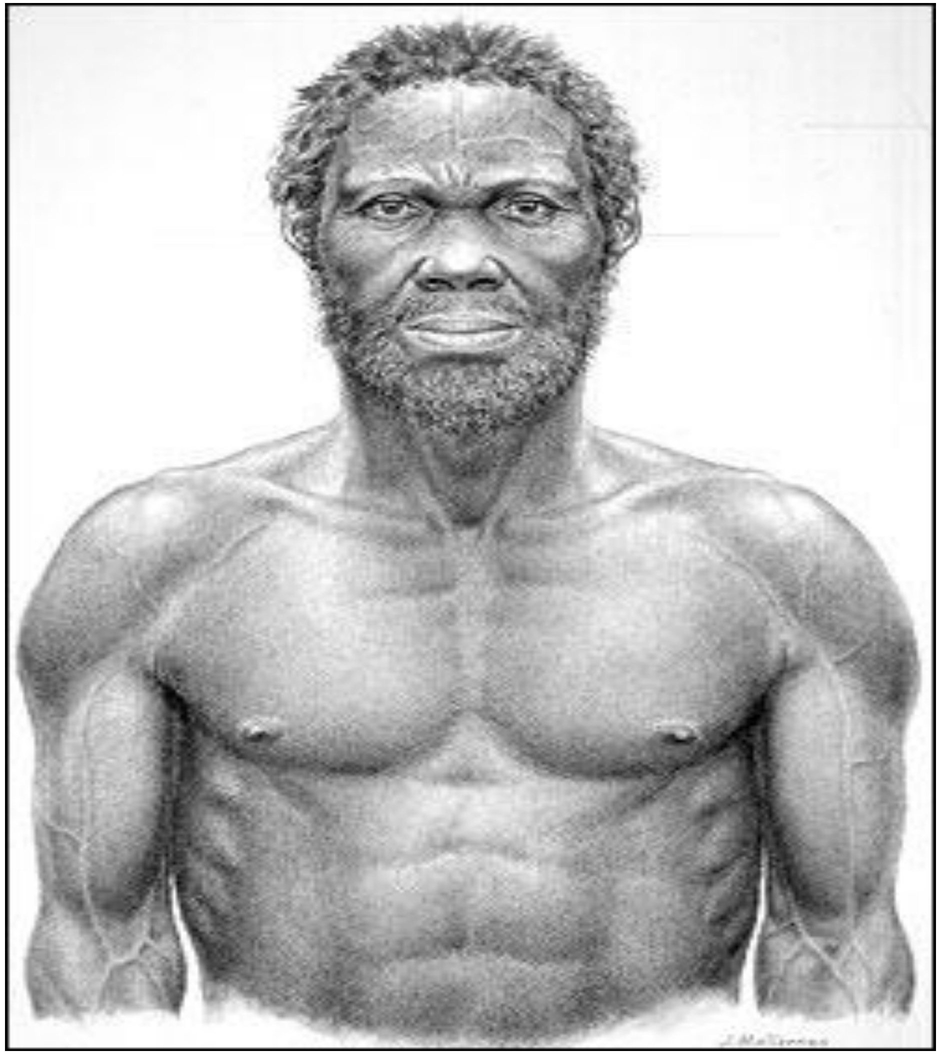
- Ooplasmic transfer technique can enable woman to avoid transmitting a mitochondrial disorder

http://news.bbc.co.uk/2/hi/health/8220553.stm

http://www.bbc.co.uk/news/health-18393682

Mitochondrial "Eve"

- mtDNA mutates at a faster rate than nuclear (no DNA repair). Approximately 2-3% per 10^6 years.
- mtDNA sequences of indigenous peoples worldwide were compared to determine the common ancestral mtDNA sequence
- Hypothesized ancestral woman lived approximately 170,000-200,000 years ago in Africa
- This is remarkably close to the date of the Homo sapiens idaltu fossils



http://www.npr.org/programs/atc/features/2003/jun/humanfossil/idaltu.html

Mitochondrial DNA ancestry tracing

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Leicester

Richard III dig: DNA confirms bones are king's

4 February 2013 | Leicester | 1579

Richard Buckley, lead archaeologist, Leicester University: "Beyond reasonable doubt, the individual exhumed is Richard III"

A skeleton found beneath a Leicester car park has been confirmed as that of English king Richard III.

http://www.bbc.com/news/uk-england-leicestershire-21063882

"Richard III earliest surviving portrait" by Unknownartist;https://commons.wikimedia.org/

The Y chromosome provides clues about human male evolution

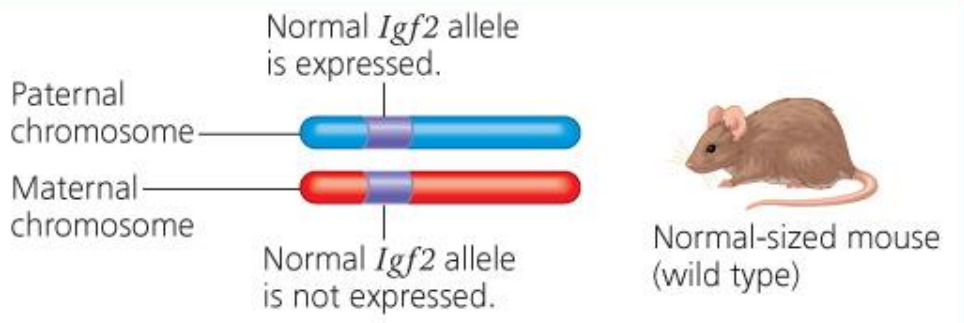
- The Y chromosome provides clues about human male evolution because
  - Y chromosomes are passed intact from father to son and
  - mutations in Y chromosomes can reveal data about recent shared ancestry.

https://commons.wikimedia.org/wiki/File:YuanEmperorAlbumGenghisPortrait.jpg

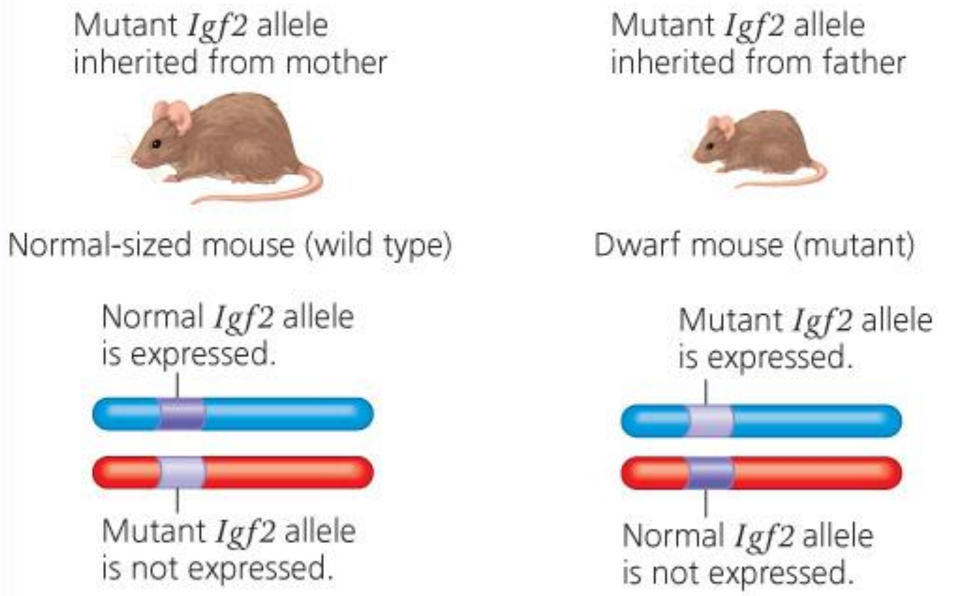
Genomic Imprinting

- Expression of an allele is determined by the parent that contributed it
  - Only one allele (from either father or mother) is expressed
  - Other allele is turned off (silenced)

  - Often, result of methylation of region adjacent to gene responsible for trait



(a) Homozygote. A mouse homozygous for the wild-type *Igf2* allele is normal-sized. Only the paternal allele of this gene is expressed.



(b) Heterozygotes. Matings between wild-type mice and those homozygous for the recessive mutant *Igf2* allele produce heterozygous offspring. The dwarf (mutant) phenotype is seen only when the father contributed the mutant allele because the maternal allele is not expressed.

Prader-Willi (PWS) and Angelman Syndrome (AS)

- Deletion of a region of Chromosome 15 (contains the PWS and AS genes).

- PWS – Normal but imprinted maternal PWS gene, paternal deletion of the region (so no functioning PWS gene).

- AS - Normal but imprinted paternal AS, maternal deletion of the region (so no functioning AS gene).

http://en.wikipedia.org/wiki/File:Pws.jpg

Phenocopy

- A trait that appears inherited but is caused by the environment, e.g. exposure to teratogens.
- May have symptoms that resemble an inherited trait or occur within families
  - Thalidomide causes limb defects similar to inherited phocomelia

Malformations due to maternal ingestion of thalidomide (Schardein 1982 and Moore 1993).

NIH –US. Public Domain

http://en.wikipedia.org/wiki/File:Phocomelia.jpg

Summary study points

- What is meant by character, trait, allele ?
  - Genotype, phenotype
  - Homozygous, heterozygous, dominant, recessive
- What is a monohybrid cross?
  - Expected phenotype ratios
- How does Mendel’s law of segregation explain the observed phenotypes?
  - Product and sum rule in probability
- What is a testcross?
  - What results would you predict if the plant were heterozygous versus homozygous?
- What is a dihybrid cross?
  - What results would you expect and why?
- What results would you expect from a dihybrid testcross?
- How does the chromosomal theory of inheritance explain Mendel’s results?

Summary study points

- How can one track genetic traits in humans
  - Dominance vs. recessive
  - Pedigree analysis
- What are the modifications to Mendel's work
  - Incomplete dominance
  - Codominance
  - Epistasis
  - Pleiotropy
  - Environment
- What is meant by gene linkage?
- How are new allelic combinations created?
- How is the percentage of recombinant offspring calculated?
- How can recombination frequency be used to determine relative gene position on a chromosome?
- How is sex determined in human embryos?
  - XX vs. XY, SRY gene
- What does it mean if a gene is sex-linked?
  - What inheritance patterns would you observe?
- What are the inheritance patterns for human disease X-linked recessive versus dominant?
- What is meant by non-traditional patterns of inheritance?