Genes and Inheritance

Variation

Causes of Variation

Variation

- No two people are exactly the same
- The differences between people is called VARIATION.
- This variation comes from two sources:
  - <u>**Genetic cause**</u>
    - Inherited (passed on) from the parents
  - <u>**Environmental cause**</u>
    - Influences from the environment

Variation

- Draw a table as below:

Genetic cause	Environmental cause	Mixture of causes

Now put these variations into your table:

- Height
- Size of ears
- Ability to swim
- Shape of nose
- Language spoken
- Natural hair colour
- Length of hair
- Colour of skin
- Strength
- Eye colour
- Amount of tooth decay
- Weight

Answers

Genetic cause	Environmental cause	Mixture of causes
•Shape of nose •Natural hair colour •Eye colour	•Ability to swim •Language spoken •Length of hair •Amount of tooth decay	•Size of ears •Height •Colour of skin •Weight •Strength

Variation

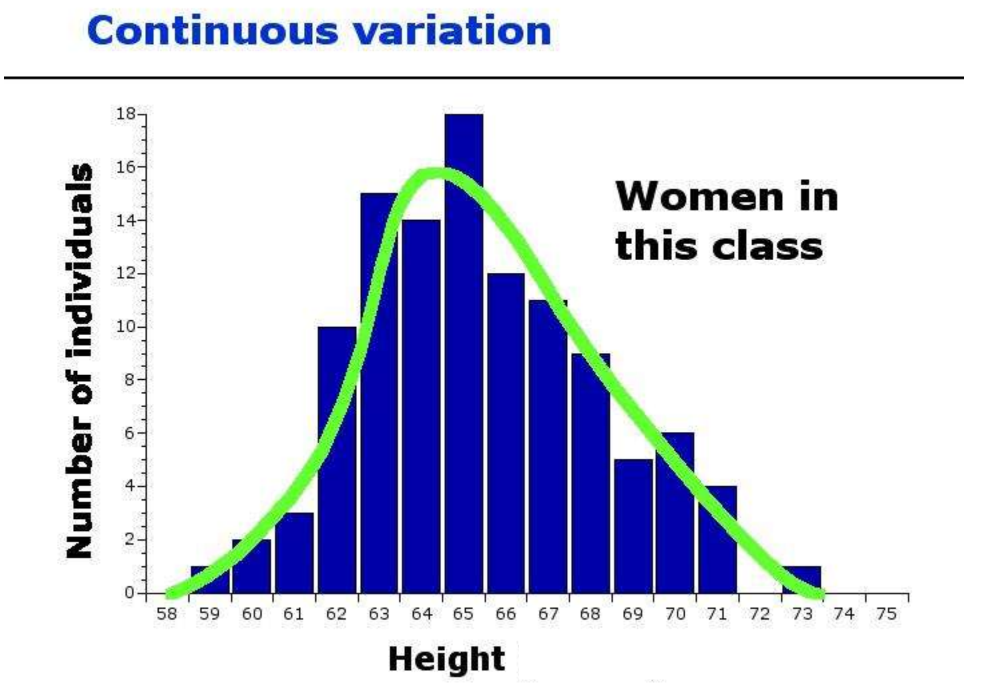
Types of Variation

Continuous and Discontinuous Variation

- Variation can also be split into continuous and discontinuous variation.
- This refers to how we organise the values of variation
  - Values = how many different types there are.

Continuous variation

- This kind of variation has a RANGE of values available.
- For example, you can be any height:

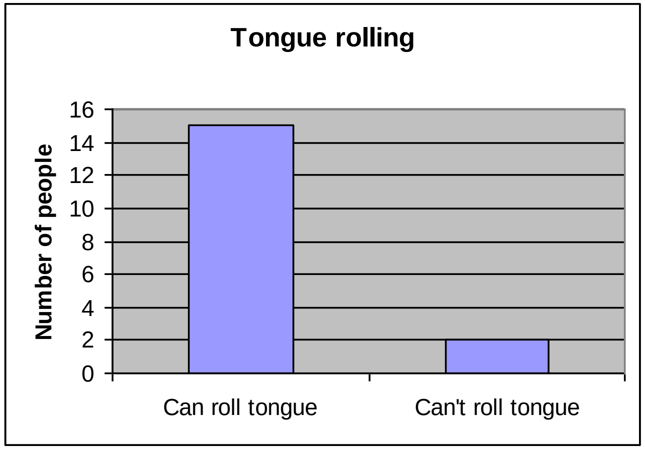


Continuous variation

- This type of distribution is called a bell curve
  - There are few people who are very short or very tall
  - Most people are of average height
- This kind of variation is produced by environmental causes or a mixture of genetic and environmental causes.

Discontinuous Variation

- In this type of variation there are a limited number of values.
- E.g. tongue rolling – you can either do it or not.



Discontinuous Variation

- This type of data is called **discrete** data. It is in separate columns.
- This is usually caused by genetic factors only.

Patterns of Inheritance

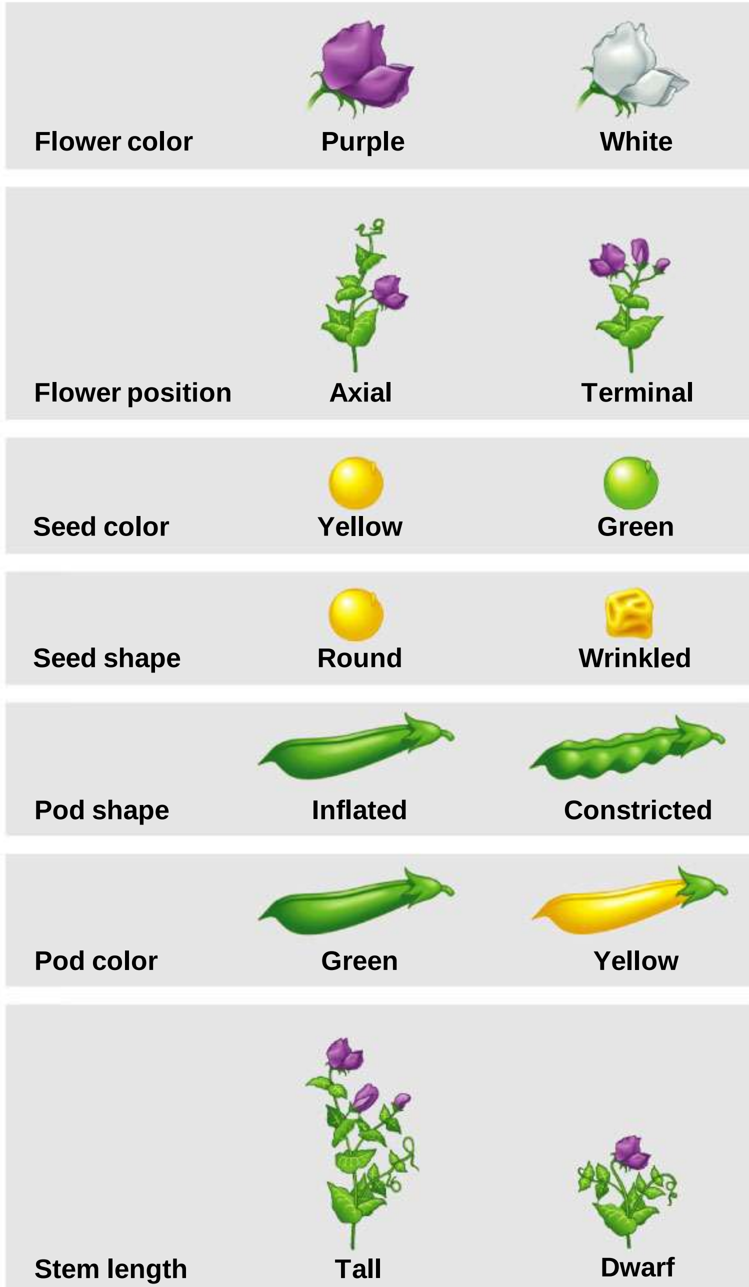
A Difference of Heredity

- Genetics is the science of heredity
- A common genetic background will produce offspring with similar physical and behavioral traits
- Behavioral characteristics are also influenced by environment

Gregory Mendel

- Experimental genetics began in an abbey garden
  - Gregor Mendel hypothesized alternative forms of genes - the units that determine heritable traits
  - Mendel crossed pea plants – 7 traits

LE 9-2d



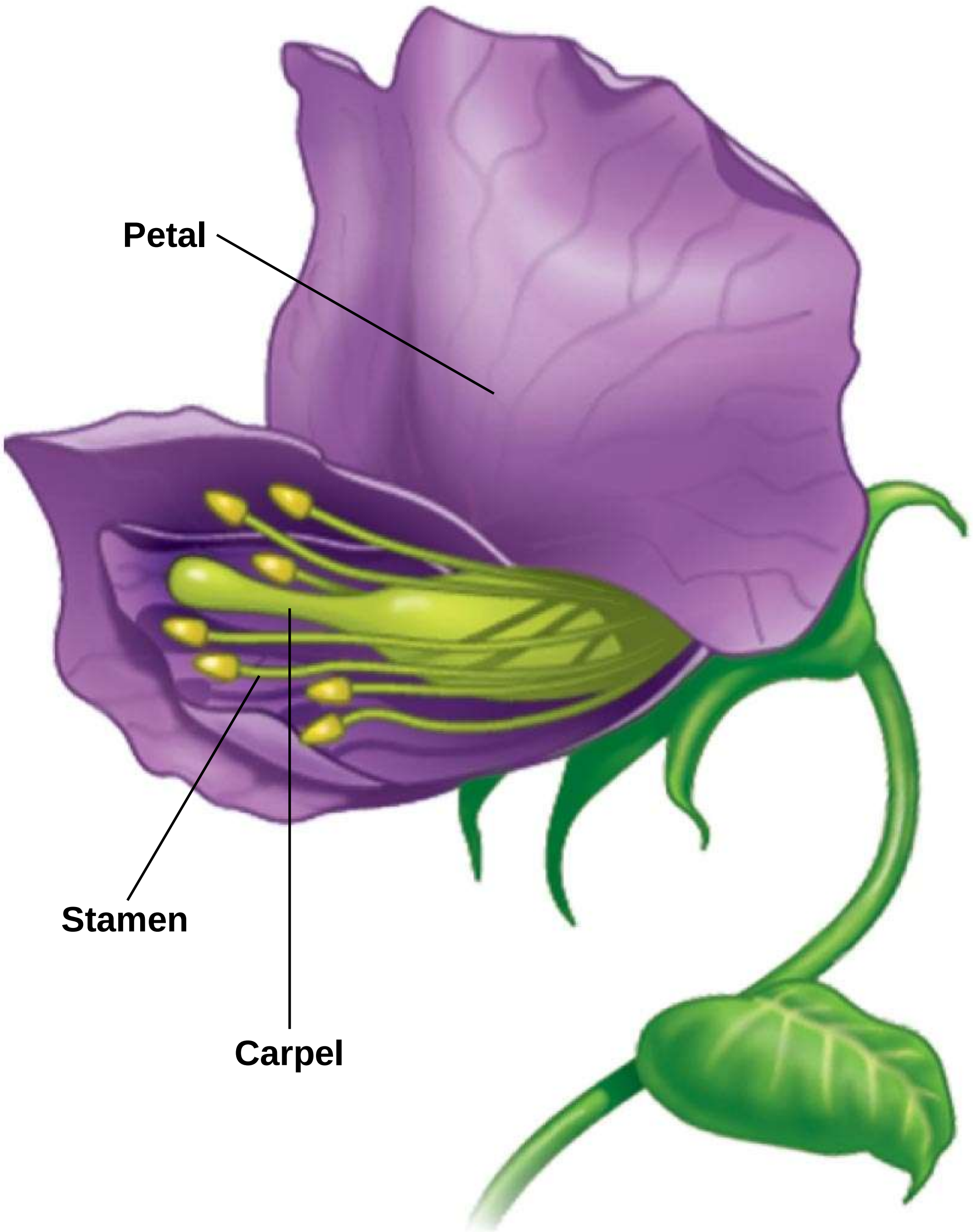
Flower color
Purple
White
Flower position
Axial
Terminal
Seed color
Yellow
Green
Seed shape
Round
Wrinkled
Pod shape
Inflated
Constricted
Pod color
Green
Yellow
Stem length
Tall
Dwarf

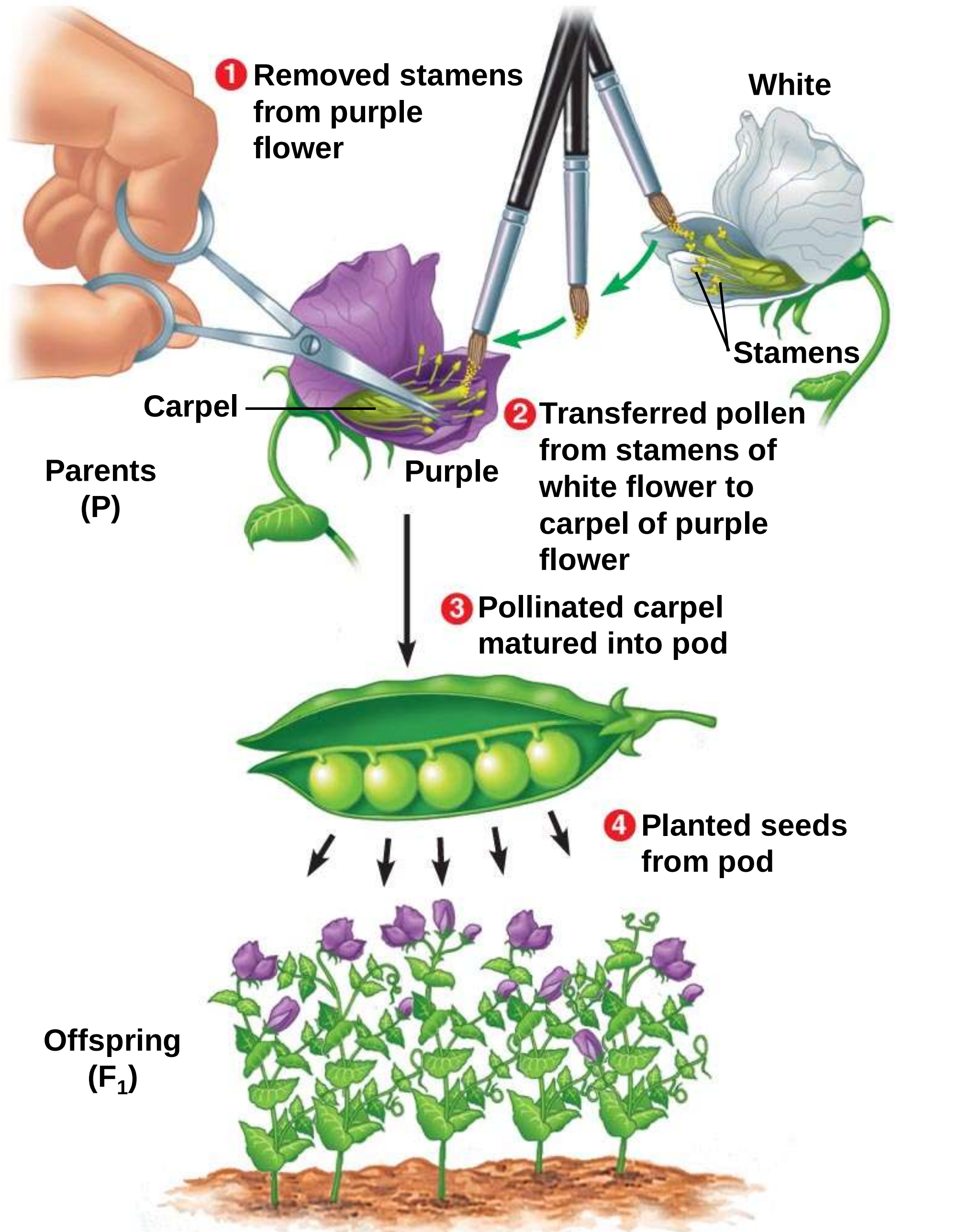
Terminology of Mendelian genetics

- Self-fertilization: fertilization of eggs by sperm-carrying pollen of the same flower
- Cross-fertilization (cross): fertilization of one plant by pollen from a different plant
- True-breeding: identical offspring from self-fertilizing parents
- Hybrid: offspring of two different varieties

Terminology of Mendelian genetics

- P generation: true-breeding parents
- F1 generation: hybrid offspring of true-breeding parents
- F2 generation: offspring of self-fertilizing F1 parents


Petal
Stamen
Carpel



1 Removed stamens from purple flower
White
Stamens
Carpel
Parents (P)
Purple
2 Transferred pollen from stamens of white flower to carpel of purple flower
3 Pollinated carpel matured into pod
4 Planted seeds from pod
Offspring (F_1)

Mendel's Law of Segregation

- Describes the inheritance of a single characteristic

Mendel's Law of Segregation

- Each feature is controlled by a gene
- There are two copies of each chromosome
- The sex cells have only one copy of each chromosome
- There are two forms for each gene

Mendel's Law of Segregation

- One form is dominant over another
- When two different forms are in the same cell only the dominant form is expressed
- An individual can have two dominant forms

Alleles

- Different types of genes that control the same characteristic are called ALLELES.

- So alleles are just different types of the same gene.

Alleles

- Let’s imagine a certain plant can have red flowers or yellow flowers.
- The GENE is flower colour
- The ALLELES are red or yellow

Alleles

- Fill in the table
  - If the gene is hair colour, what are the alleles?
  - If the gene is eye colour, what are the alleles?
  - If the gene is tongue rolling, what are the alleles?

Alleles

- Alleles are usually given a letter:
- The letter is called the **genotype**.
  - e.g. Genotype = **R**
  - Genotype is the letter or term used to describe the allele of an individual gene or pair of genes

Genotype and Phenotype

- How that letter affects the characteristic is the **phenotype**.
  - e.g. Phenotype = **Red** flower
  - Phenotype is how the gene (or pair) shows itself (how it appears).

Dominant and Recessive

- Remember Mendel said that one allele is dominant over the another
- This is expressed by using a capital letter
  - Example, purple flowers are dominant over white flowers
  - The dominant allele will be expressed as P

Dominant and Recessive

- If the allele is recessive it will be expressed as a lowercase letter
  - Example, purple flowers are dominant over white flowers
  - The recessive allele will be expressed as p

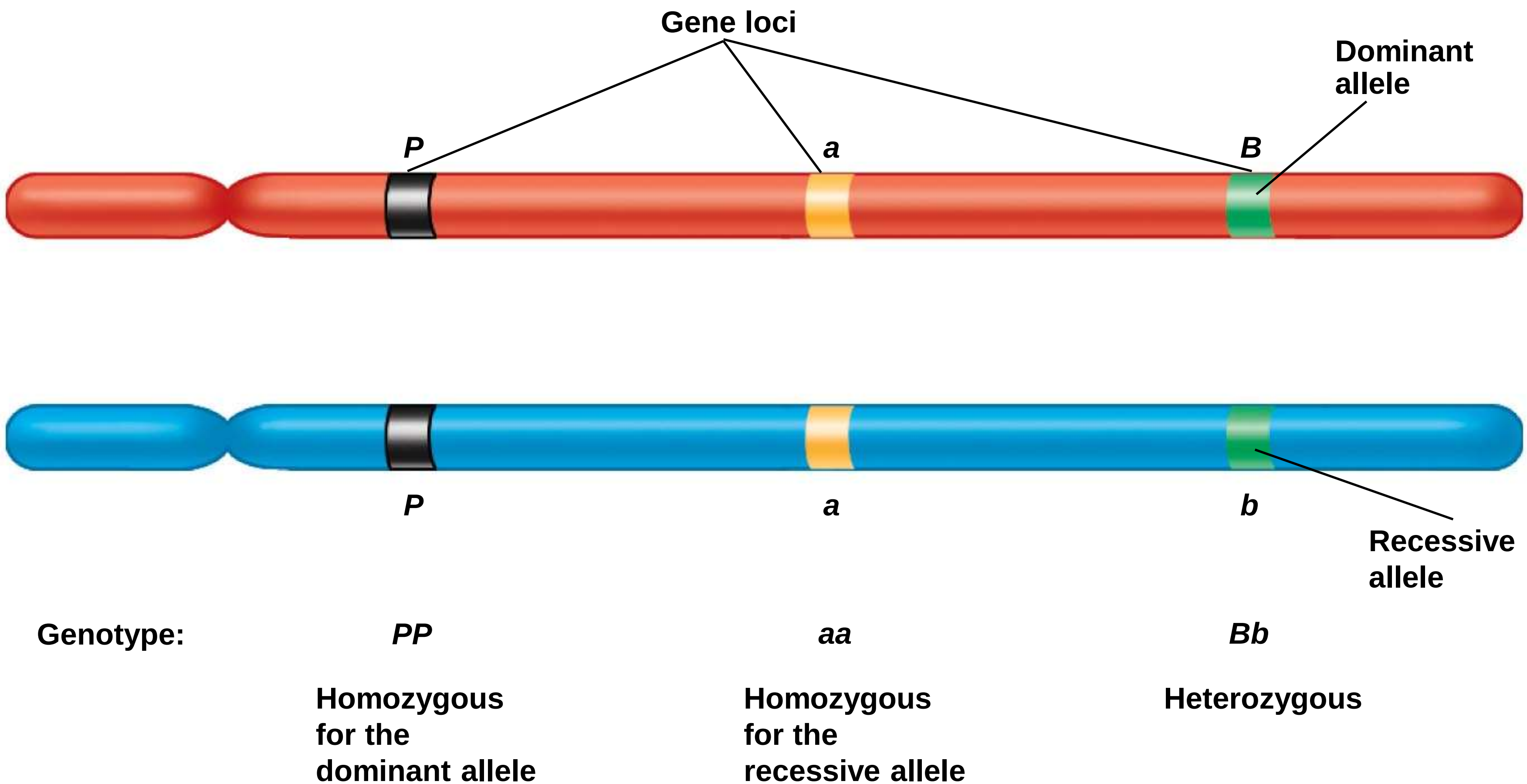
Heterozygous

- If the two alleles are heterozygous;
  - The dominant allele determines the organisms' appearance
  - The recessive allele has no noticeable effect

    - Example, Pp

Homologous

- Homologous chromosomes bear the two alleles for each characteristic
  - Alternative forms of a gene reside at the same locus on homologous chromosomes
  - Example, PP or pp


Gene loci
Dominant allele
P
a
B
P
a
b
Recessive allele
Genotype:
PP
aa
Bb
Homozygous for the dominant allele
Homozygous for the recessive allele
Heterozygous

Alleles

- Complete the paragraph

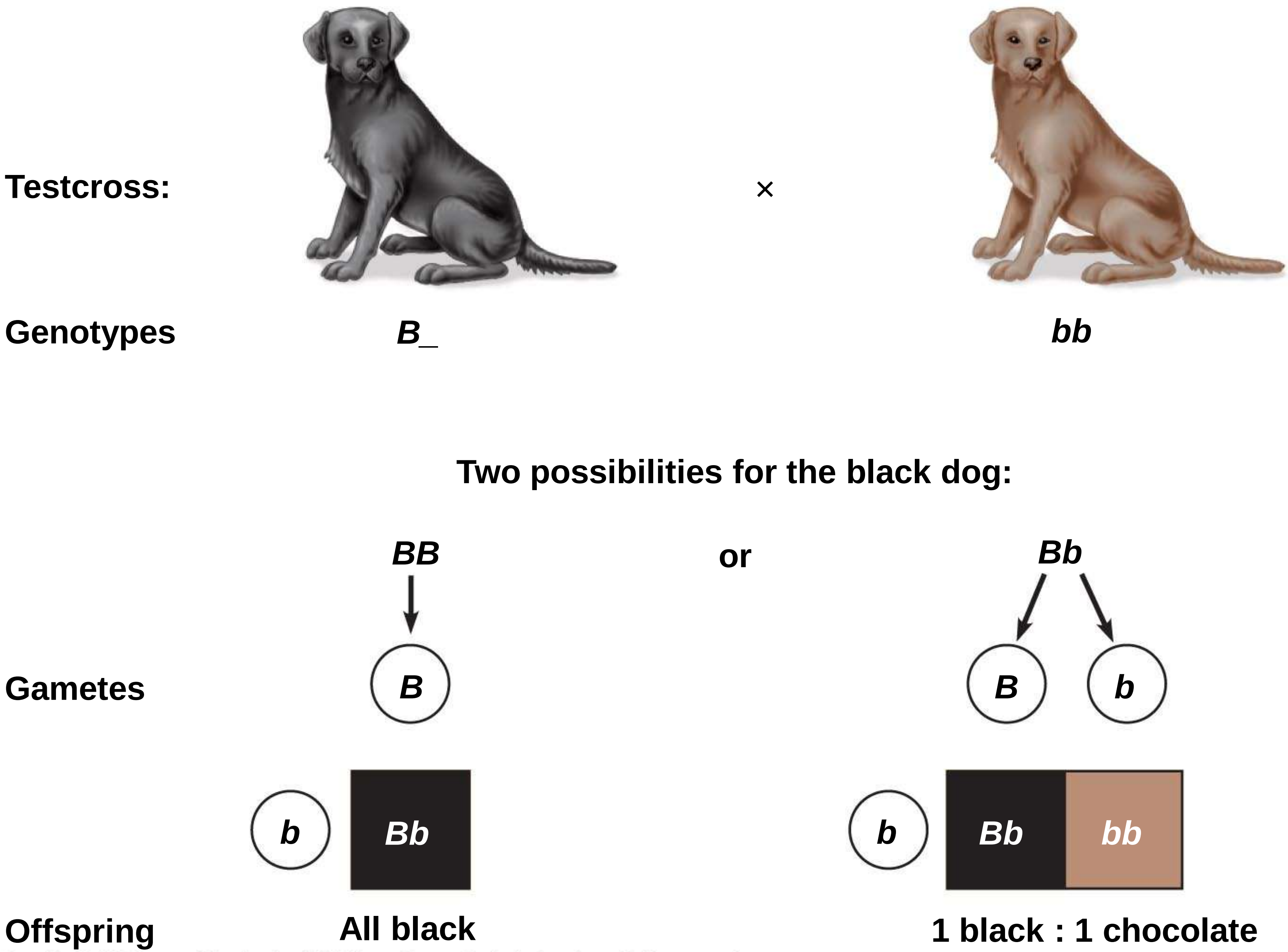
Working Out Genotypes

Testcross

- Geneticists use the testcross to determine unknown genotypes
  - A testcross can reveal an unknown genotype
    - Mate an individual of unknown genotype and a homozygous-recessive individual
    - Each of the two possible genotypes (homozygous or heterozygous) gives a different phenotypic ratio in the F1 generation

Test Cross

- In a test cross you breed an organism showing the dominant features with one showing the recessive feature


Testcross:
×
Genotypes
B_
bb
Two possibilities for the black dog:
BB
or
Bb
Gametes
B
B
b
b
Bb
b
Bb
bb
Offspring
All black
1 black : 1 chocolate

Flower colour

- Genotype of alleles- R = red flower
r = yellow flower

Genotype "R" means the phenotype "Red" is dominant

Genotype "r" means the phenotype "Yellow" is recessive.
The same letter is used to show it is the same gene.

Flower colour

- Genotype of alleles- R = red flower
  r = yellow flower
- Possible combinations of alleles are:

Genotype	RR	Rr	rr
Phenotype	Red	Red	Yellow

Flower colour

Genotype	RR	Rr	rr
Phenotype	Red	Red	Yellow

- The dominant genotype masks the recessive genotype.
- The phenotype is the same as the dominant one – not a mixture!
- The flower looks **RED**.

Example

- A plant can be tall or short.
- The gene for height is represented by the letter H.
- The dominant characteristic is tall.
- What are the possible genotypes and phenotypes?

Answer

- H =
- h =

Genotypes:

Phenotypes:

This is just a clue...

Have a go at doing it yourself!!

Answer

- H = tall
- h = short

Genotypes:

Phenotypes:

HH	Hh	hh
Tall	Tall	Short

Genetic Crosses

- Useful way of showing how genes are passed through one or two generations, starting with the parents

Genetic Crosses

- To do this we need to do a Genetic Cross. There are two ways of doing this:
  1. Gene Cross diagram
  2. Punnett Square diagram

- Let's think again about our flowers:

R = Red r = Yellow

Gene cross diagram

1. Insert the parents and their phenotype and genotype.

Gene Cross diagram 1

Parent	Male	female
Phenotype	**Red**	**Yellow**
Genotype	**RR**	**rr**

Gene cross diagram

1. Insert the parents and their phenotype and genotype.
2. Show what gametes could be formed (just separate the genotype)

Gene Cross diagram 1

Parent	Male	female
Phenotype	**Red**	**Yellow**
Genotype	**RR**	**rr**
gametes	**R** **R**	**r** **r**

Gene cross diagram

1. Insert the parents and their phenotype and genotype.
2. Show what gametes could be formed (just separate the genotype).
3. Either of the male gametes could join with either of the female gametes. Show this with lines and write the offspring's possible genotypes.

Gene Cross diagram 1

Parent	Male		female	
Phenotype	Red		Yellow	
Genotype	RR		rr	
gametes	R	R	r	r
Offspring genotype	Rr	Rr	Rr	Rr

Gene cross diagram

1. Insert the parents and their phenotype and genotype.
2. Show what gametes could be formed (just separate the genotype).
3. Either of the male gametes could join with either of the female gametes. Show this with lines and write the offspring's possible genotypes.
4. Now add the phenotypes and ratio of the different phenotypes.

Gene Cross diagram 1

Parent	Male		female	
Phenotype	Red		Yellow	
Genotype	RR		rr	
gametes	R	R	r	r
Offspring genotype	Rr	Rr	Rr	Rr
Offspring Phenotype	Red	Red	Red	Red

Ratio of Red : Yellow = 4:0

Gene Cross diagram 2

Heterozygous (Rr)

with

Heterozygous (Rr)

Gene Cross diagram 2

Parent	Male		female	
Phenotype	Red		Red	
Genotype	Rr		Rr	
gametes	R	r	R	r
Offspring genotype	RR	rr	Rr	Rr
Offspring Phenotype	Red	Yellow	Red	Red

Ratio of Red : Yellow = 3:1

Gene Cross diagram 3

Heterozygous (Rr)

with

Homozygous recessive (rr)

(Try this one on your own!)

Gene Cross diagram 3

Parent	Male	female
Phenotype	Red	Yellow
Genotype	Rr	rr
gametes		

Ratio of Red : Yellow = 1:1

Offspring genotype	Rr	rr	rr	Rr
Offspring Phenotype	Red	Yellow	Yellow	Red

Punnett Squares

- Another method of showing crosses

Punnett Squares

- Add the parents genotype and phenotype:

Rr male Red

rr
female
yellow

Punnett Squares

- Add the gametes:

Rr male Red

rr female yellow

	R	r
r		
r		

Punnett Squares

- Do the cross and add the phenotypes:

Rr male Red

rr female yellow

	R	r
r	Rr Red	rr Yellow
r	Rr Red	rr Yellow

Punnett Squares

- Work out the ratios:

Rr male Red

rr female yellow

	R	r
r	Rr Red	rr Yellow
r	Rr Red	rr Yellow

2 yellow and 2 red offspring

1:1 chance with these parents

Question for you!

- Rats have 2 coat colours:

Black = B

White = b

Which colour is dominant?

Answer = Black

B = Black; b = white

- Two rats with black coats are mated. All their offspring are black.

What are the possible genotypes of the parents?

Answer = both BB or

one parent could be Bb

(see next slide)

B = Black; b = white

BB male Black

BB female black

	B	B
B	BB Black	BB Black
B	BB Black	BB Black

All offspring black

OR

B male Black

BB female black

	B	b
B	BB Black	Bb Black
B	BB Black	Bb Black

All offspring black

B = Black; b = white

- The male is then mated with a white female. They have 6 black mice and 7 white mice.

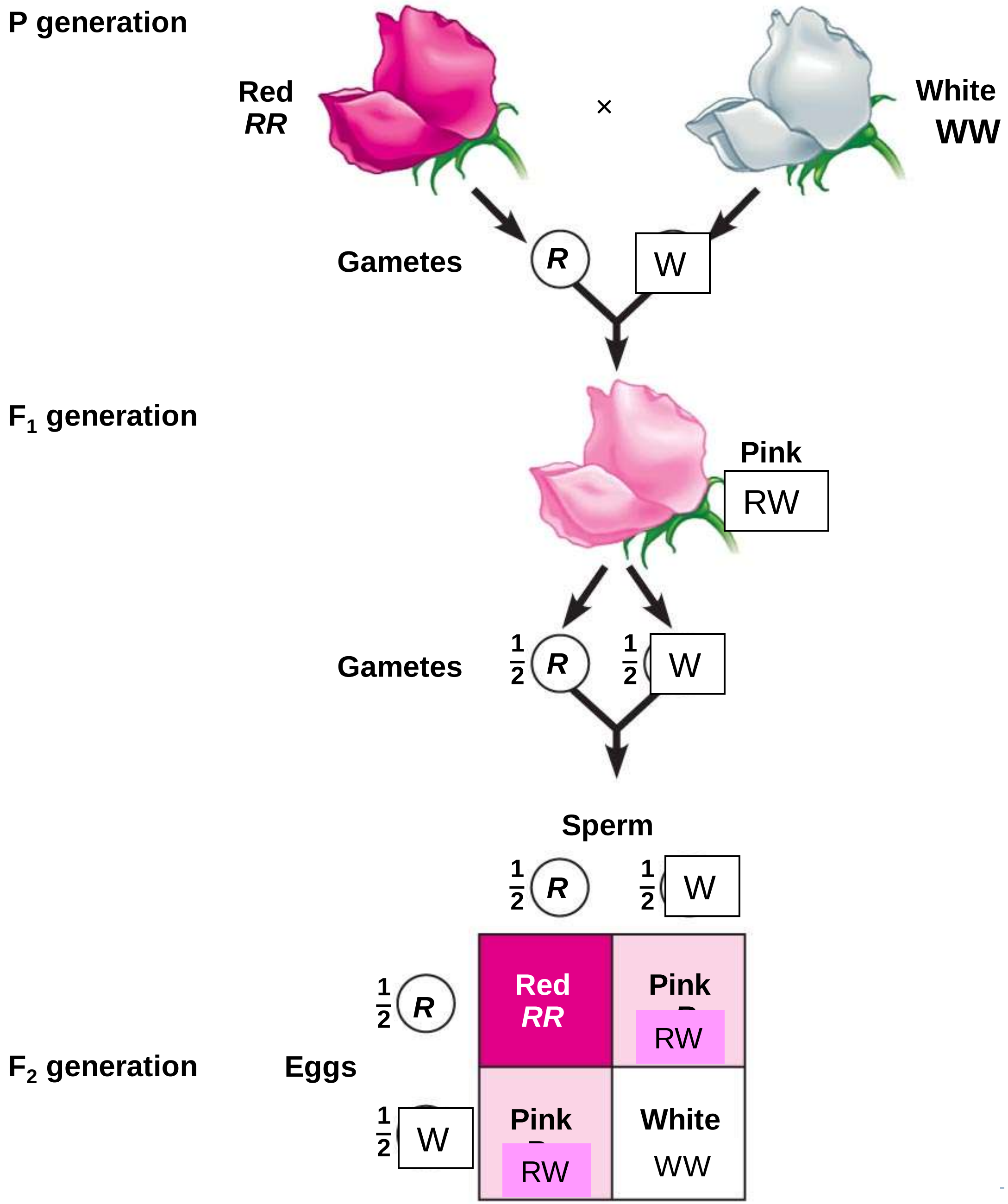
- What was the genotype of the original parents?
- Draw a genetic diagram to illustrate your answer.

Co Dominance

Codominance

- Codominance results when two alleles are expressed in the same phenotype
  - The resulting cross will be a mixture of the two parents

LE 9-12a


P generation
Red
RR
×
White
WW
Gametes
R
W
F1 generation
Pink
RW
Gametes
1/2 R
1/2 W
Sperm
1/2 R
1/2 W
F2 generation
Eggs
1/2 R
1/2 W
Red
RR
Pink
RW
Pink
RW
White
WW

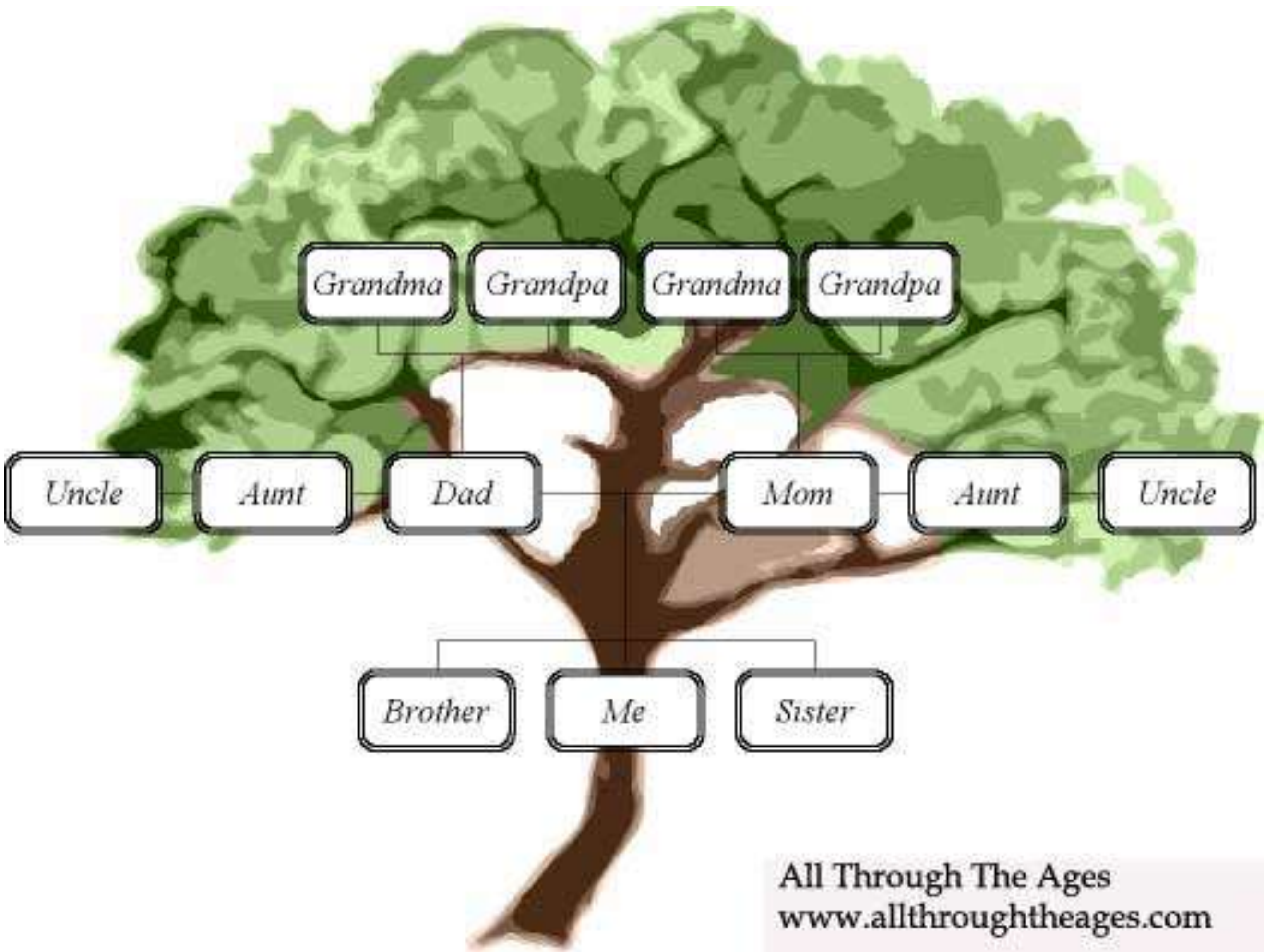
Co Dominance

- Most genes don’t show complete dominance
- Genes can show a range of dominance

Pedigree

- Tracking Family History

Pedigrees

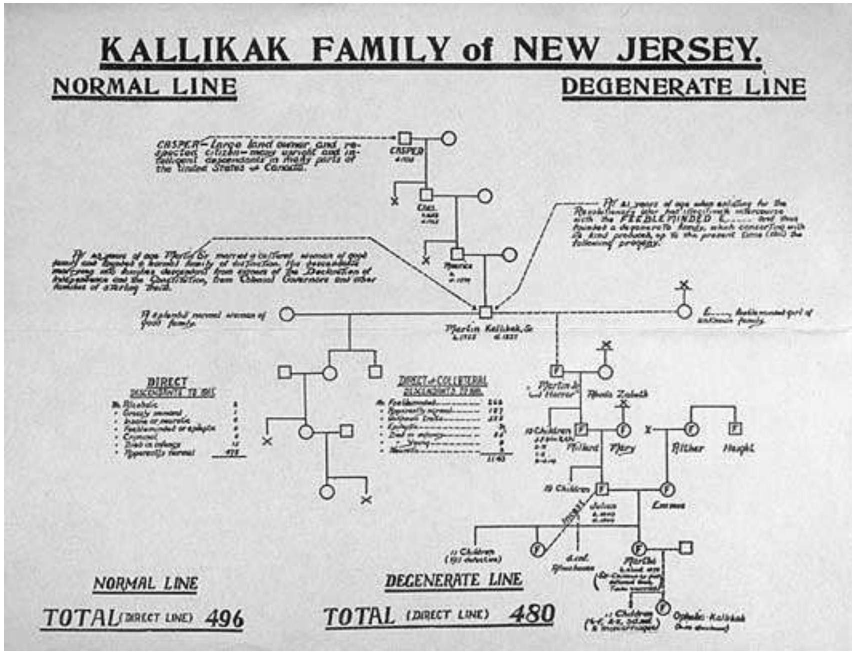


Pedigrees

- Can be used to determine genotypes of family members.
- Can be used to help predict probability of future generations expressing certain traits.
- Important tool for genetic counselors

Pedigrees

- A chart that shows multiple family generations and relationships to track the inheritance of genetic traits.



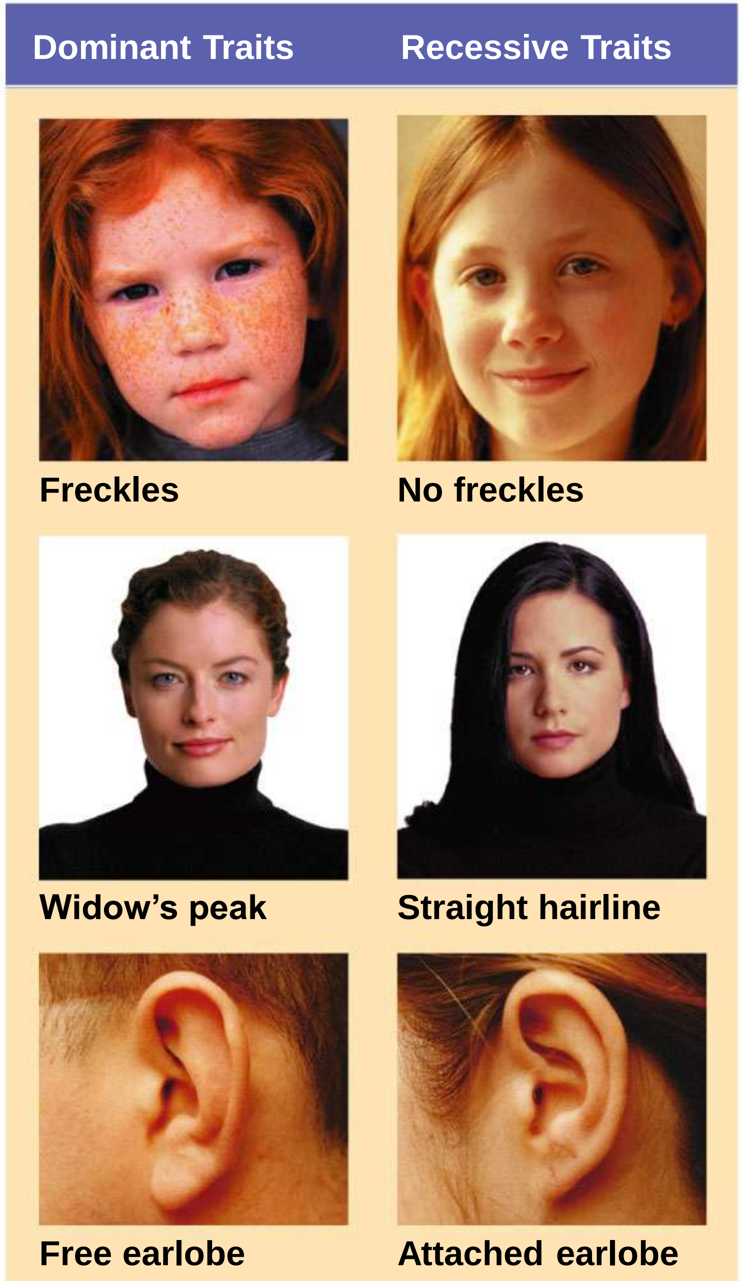
Pedigrees

- It is possible to work out which allele is dominant from pedigrees
- Look for a situation where two parents show the same feature and at least one child shows the contrasting feature

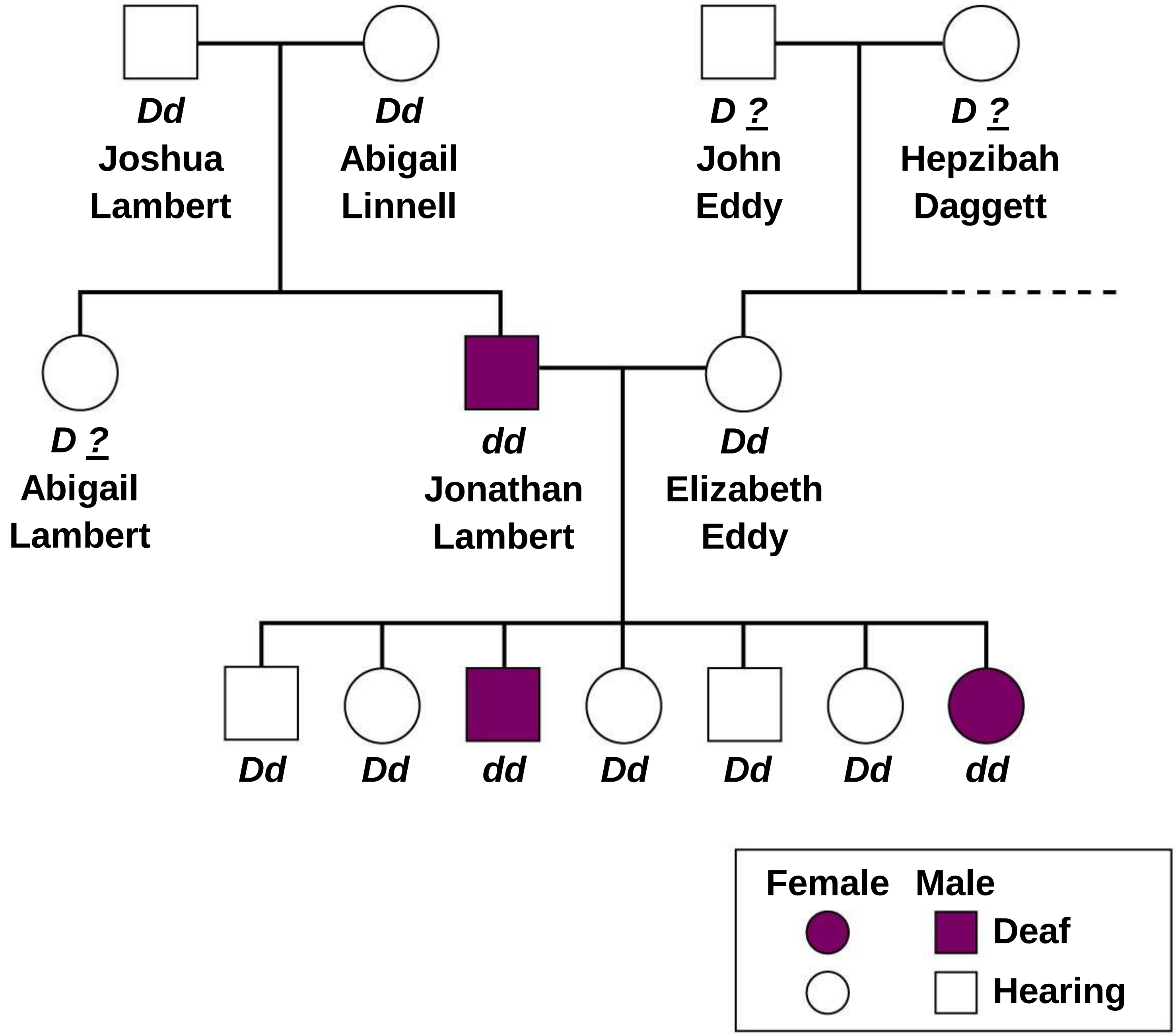
Pedigrees

- Family pedigrees can be used to determine individual genotypes

LE 9-8a


Dominant Traits
Recessive Traits
Freckles
No freckles
Widow's peak
Straight hairline
Free earlobe
Attached earlobe

LE 9-8b

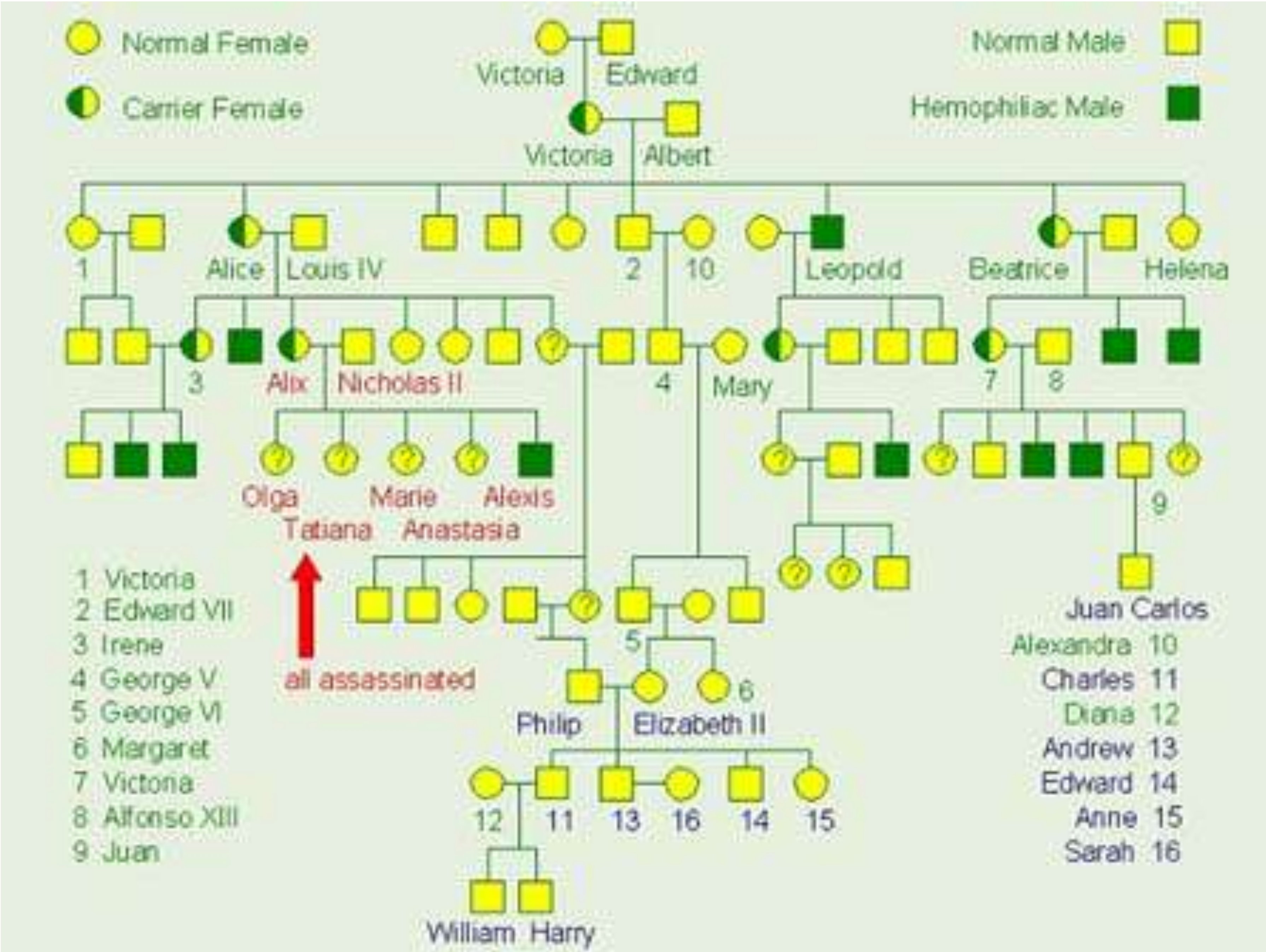

Dd
Joshua
Lambert
Dd
Abigail
Linnell
D ?
John
Eddy
D ?
Hepzibah
Daggett
D ?
Abigail
Lambert
dd
Jonathan
Lambert
Dd
Elizabeth
Eddy
Dd
Dd
dd
Dd
Dd
Dd
dd
Female
Male
Deaf
Hearing

LE 9-9a

Parents Normal *Dd* × Normal *Dd*

Offspring

		Sperm	
		D	*d*
Eggs	*D*	*DD* Normal	*Dd* Normal (carrier)
	d	*Dd* Normal (carrier)	*dd* Deaf



Normal Female
Carrier Female
Normal Male
Hemophiliac Male
Victoria
Edward
Victoria
Albert
1
Alice
Louis IV
2
10
Leopold
Beatrice
Helena
3
Alix
Nicholas II
4
Mary
7
8
Olga
Tatiana
Marie
Anastasia
Alexis
9
all assassinated
5
6
Philip
Elizabeth II
12
11
13
16
14
15
William
Harry
Juan Carlos
1 Victoria
2 Edward VII
3 Irene
4 George V
5 George VI
6 Margaret
7 Victoria
8 Alfonso XIII
9 Juan
Alexandra 10
Charles 11
Diana 12
Andrew 13
Edward 14
Anne 15
Sarah 16

Review tables

TABLE 9.9 SOME AUTOSOMAL DISORDERS IN HUMANS

Disorder	Major Symptoms	Incidence
Recessive disorders		
Albinism	Lack of pigment in skin, hair, and eyes	$\frac{1}{22,000}$
Cystic fibrosis	Excess mucus in lungs, digestive tract, liver; increased susceptibility to infections; death in early childhood unless treated	$\frac{1}{2,500}$ Caucasians
Galactosemia	Accumulation of galactose in tissues; mental retardation; eye and liver damage	$\frac{1}{100,000}$
Phenylketonuria (PKU)	Accumulation of phenylalanine in blood; lack of normal skin pigment; mental retardation	$\frac{1}{10,000}$ in U.S. and Europe
Sickle-cell disease (homozygous)	Sickled red blood cells; damage to many tissues	$\frac{1}{400}$ African-Americans
Tay-Sachs disease	Lipid accumulation in brain cells; mental deficiency; blindness; death in childhood	$\frac{1}{3,500}$ Jews from central Europe

Review tables

TABLE 9.9 SOME AUTOSOMAL DISORDERS IN HUMANS

Disorder	Major Symptoms	Incidence
Dominant disorders		
Achondroplasia	Dwarfism	$\frac{1}{25,000}$
Alzheimer's disease (one type)	Mental deterioration; usually strikes late in life	Not known
Huntington's disease	Mental deterioration and uncontrollable movements; strikes in middle age	$\frac{1}{25,000}$
Hypercholesterolemia	Excess cholesterol in blood; heart disease	$\frac{1}{500}$ are heterozygous

Genetic testing

- Genetic testing can detect disease-causing alleles
  - Predictive genetic testing may inform people of their risk for developing genetic diseases
    - Used when there is a family history but no symptoms
    - Increased use of genetic testing raises ethical, moral, and medical issues

Sex Chromosomes

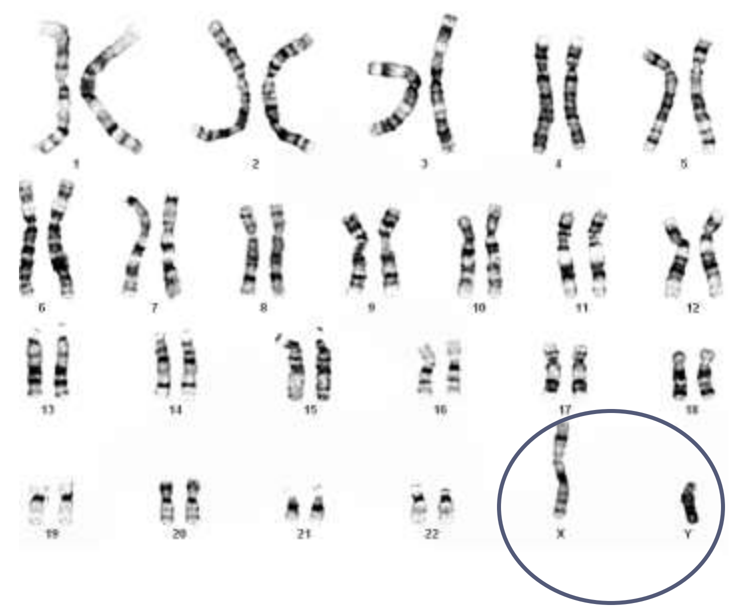
SEX CHROMOSOMES AND SEX-LINKED GENES

- Chromosomes determine sex in many species
  - Many animals have a pair of chromosomes that determine sex
    - Humans: X-Y system
      - Male is XY; the Y chromosome has genes for the development of testes
      - Female is XX; absence of a Y chromosome allows ovaries to develop

Sex Chromosomes

- 46 total chromosomes
- 44 non-sex
- 2 sex

female



male

Sex Chromosomes

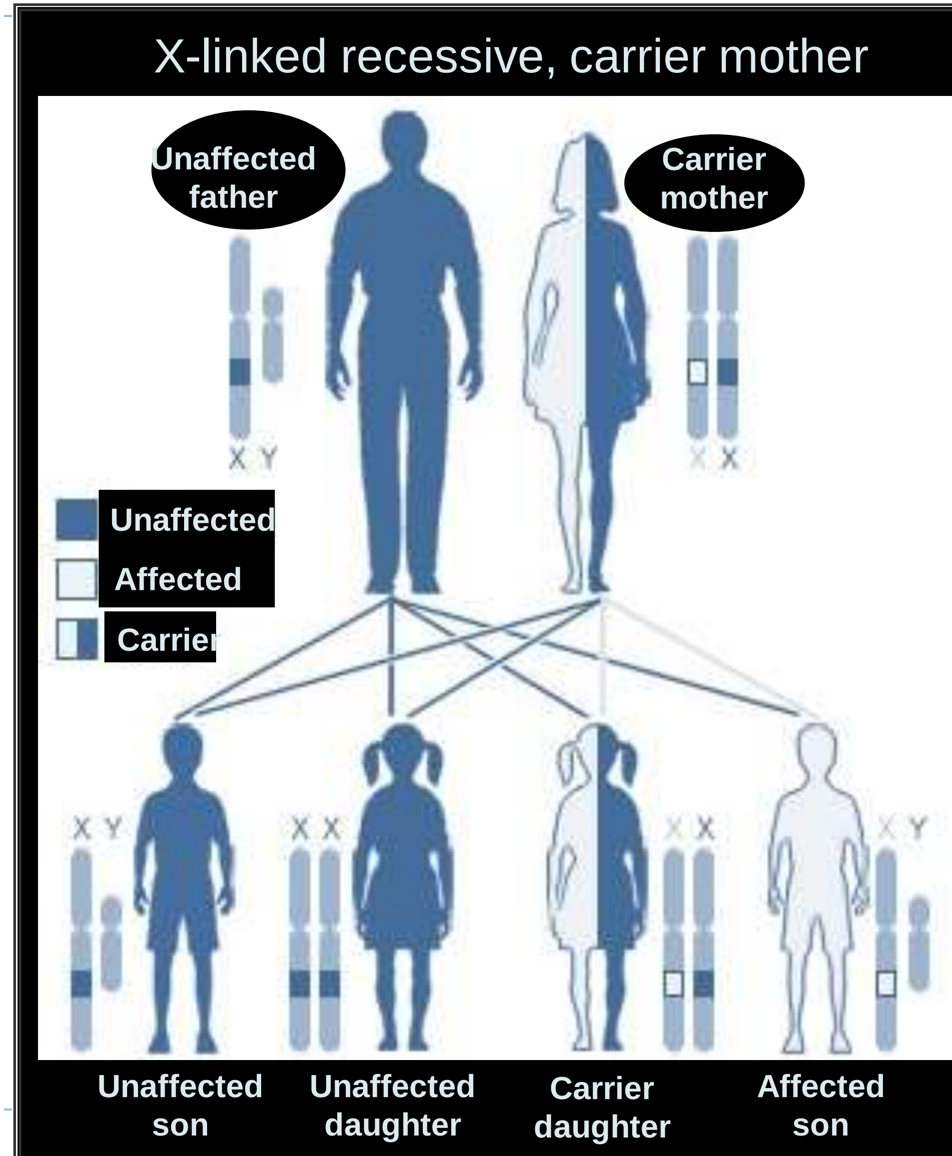
- Our sex is determined by the presence or absence of the Y chromosome

- Who determines the sex of the off spring?

- The male!!

Sex-Linked Traits

- It is possible for a female to be a carrier of an X-linked trait, but not express it
- Men will express all X-linked traits they inherit



Sex-Linked Chromosomes

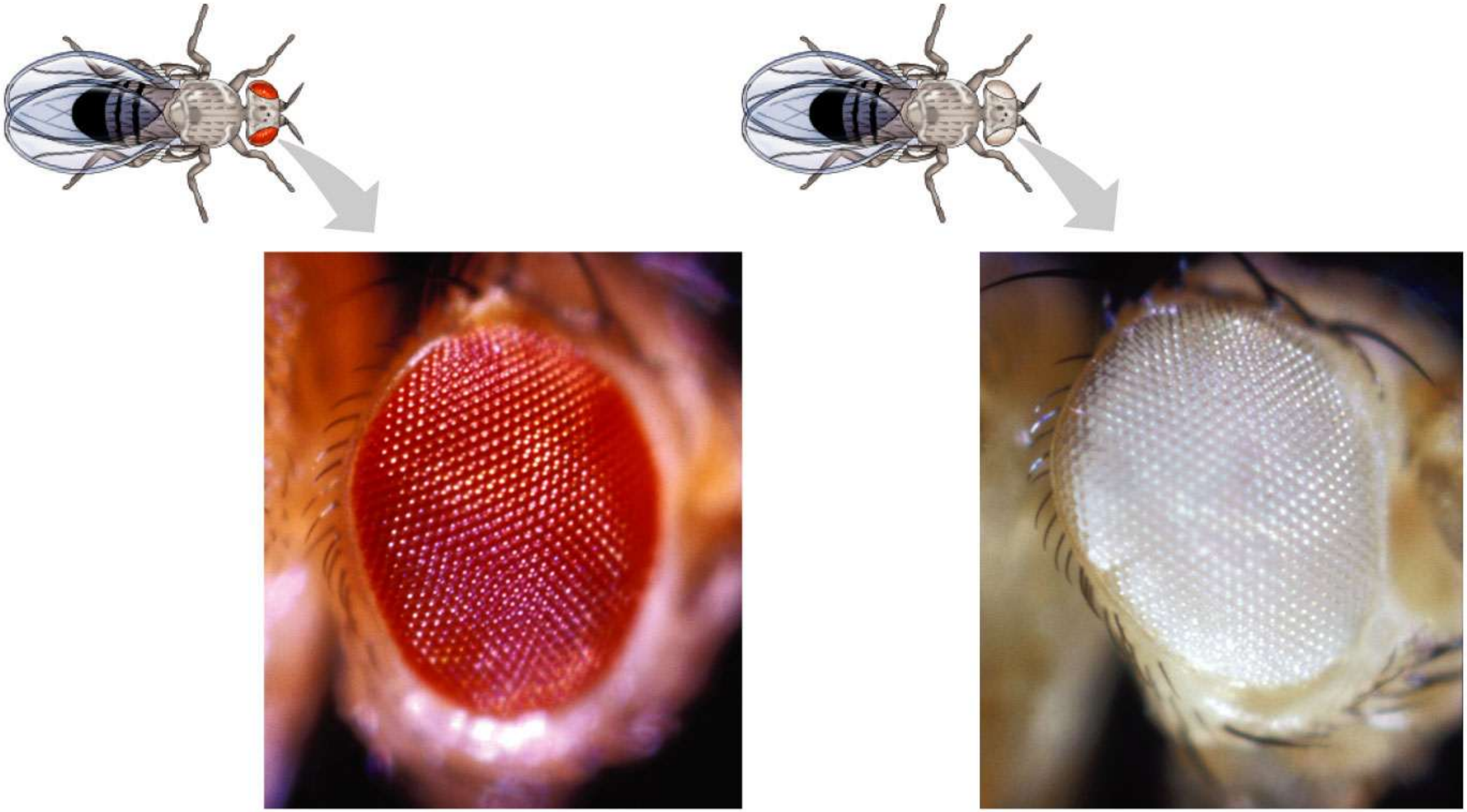
- Sex-linked genes exhibit a unique pattern of inheritance

Sex-Linked Chromosomes

- Sex-linked genes are genes for characteristics unrelated to sex that are located on either sex chromosome
  - In humans, refers to a gene on the X chromosome
  - Colour vision
  - Baldness

Sex-Linked Chromosomes

- Because of linkage and location, the inheritance of these characteristics follows peculiar patterns
  - Example: eye color inheritance in fruit flies follows three possible patterns, depending on the genotype of the parents



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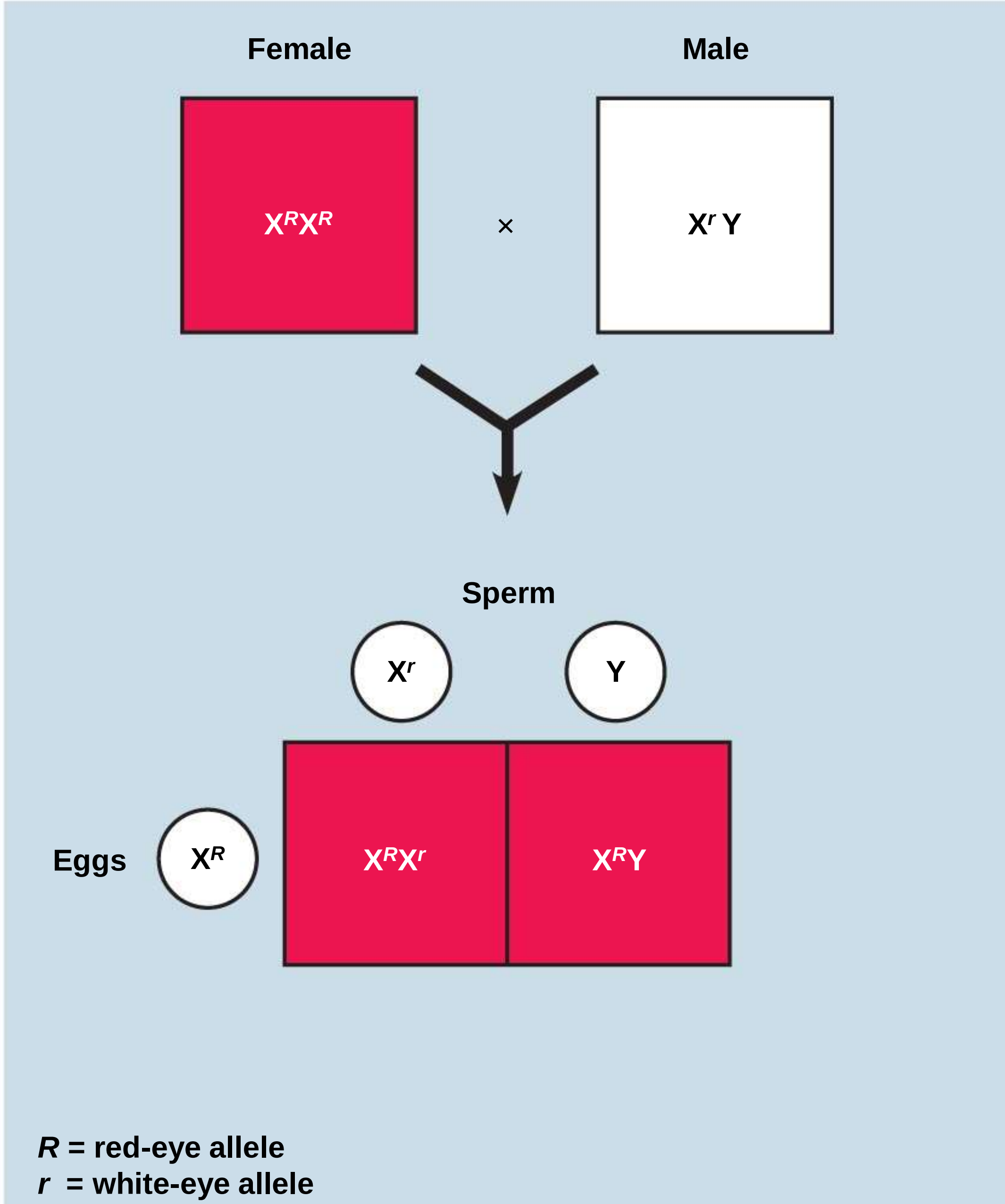
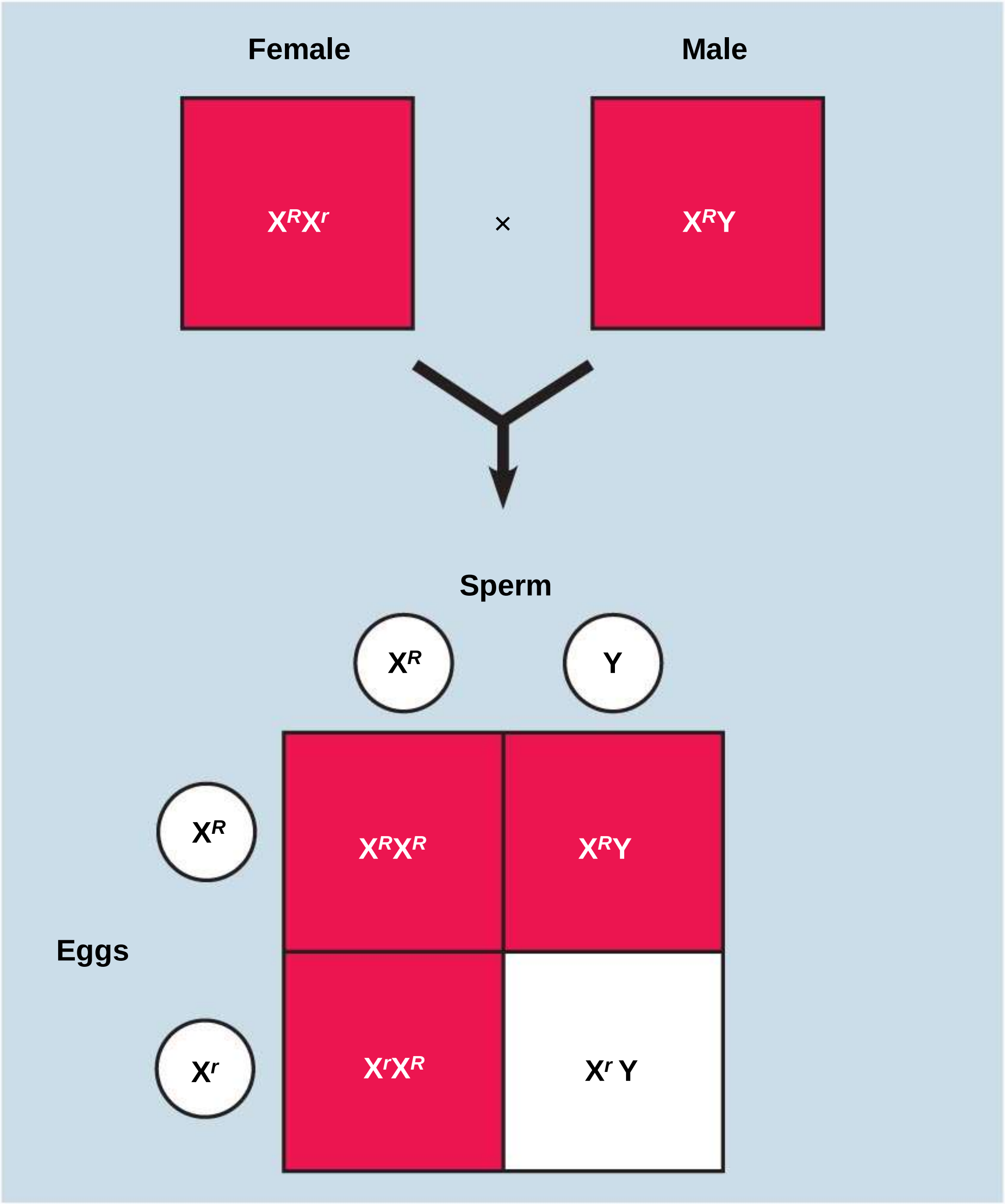
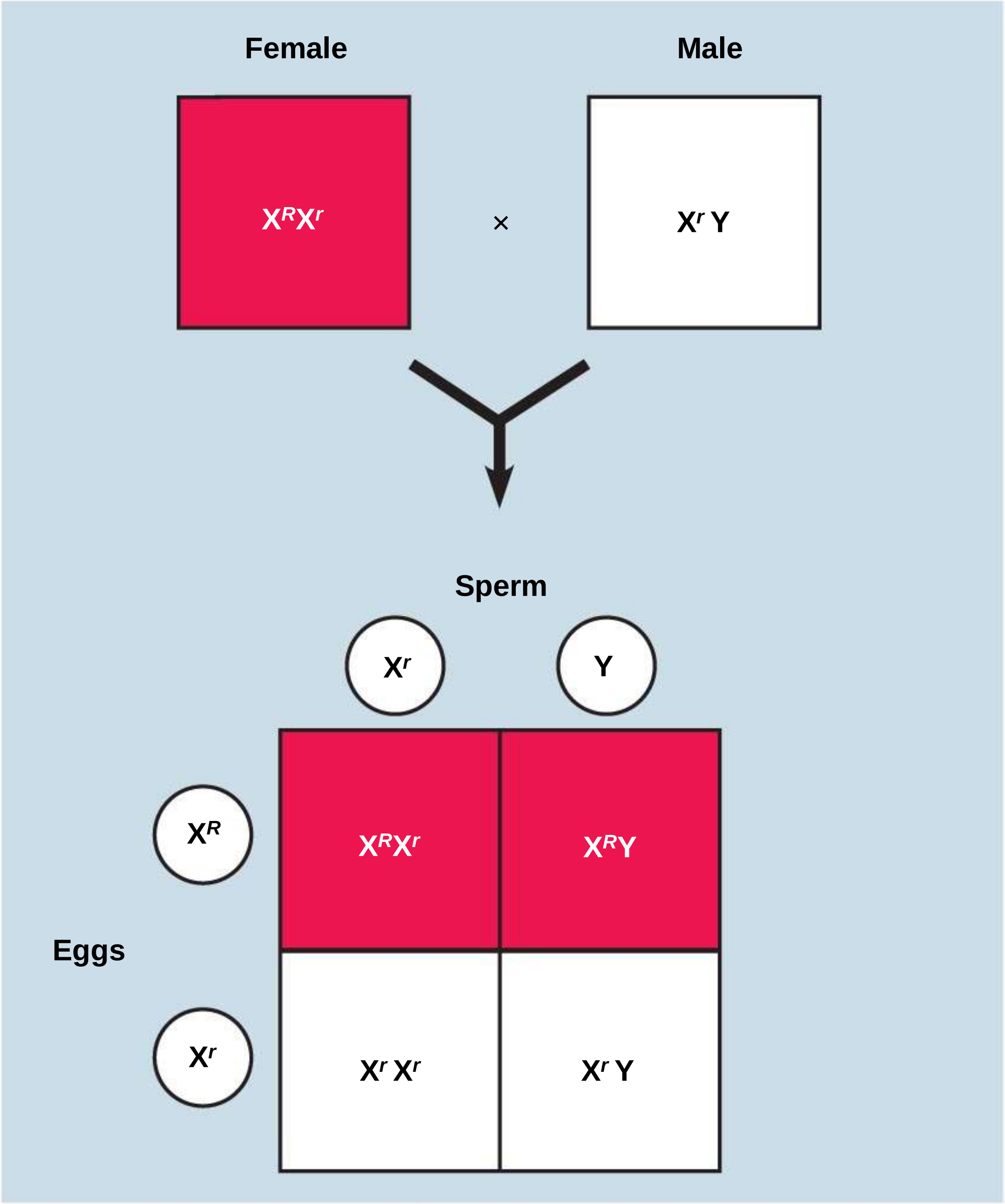


Female
Male
$X^R X^R$
×
$X^r Y$
Sperm
X^r
Y
Eggs
X^R
$X^R X^r$
$X^R Y$
R = red-eye allele
r = white-eye allele

LE 9-23c




Female
Male
X^RX^r
×
X^rY
Sperm
X^r
Y
Eggs
X^R
X^r
X^RX^r
X^RY
X^rX^r
X^rY

Sex-Link Disorders

- Sex-linked disorders affect mostly males
  - Most known sex-linked traits are caused by genes (alleles) on the X chromosome
  - Females with the allele are normally carriers and will exhibit the condition only if they are homozygous
  - Examples: red-green color blindness, hemophilia, Duchenne muscular dystrophy