Theoretical Genetics

Variation

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

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

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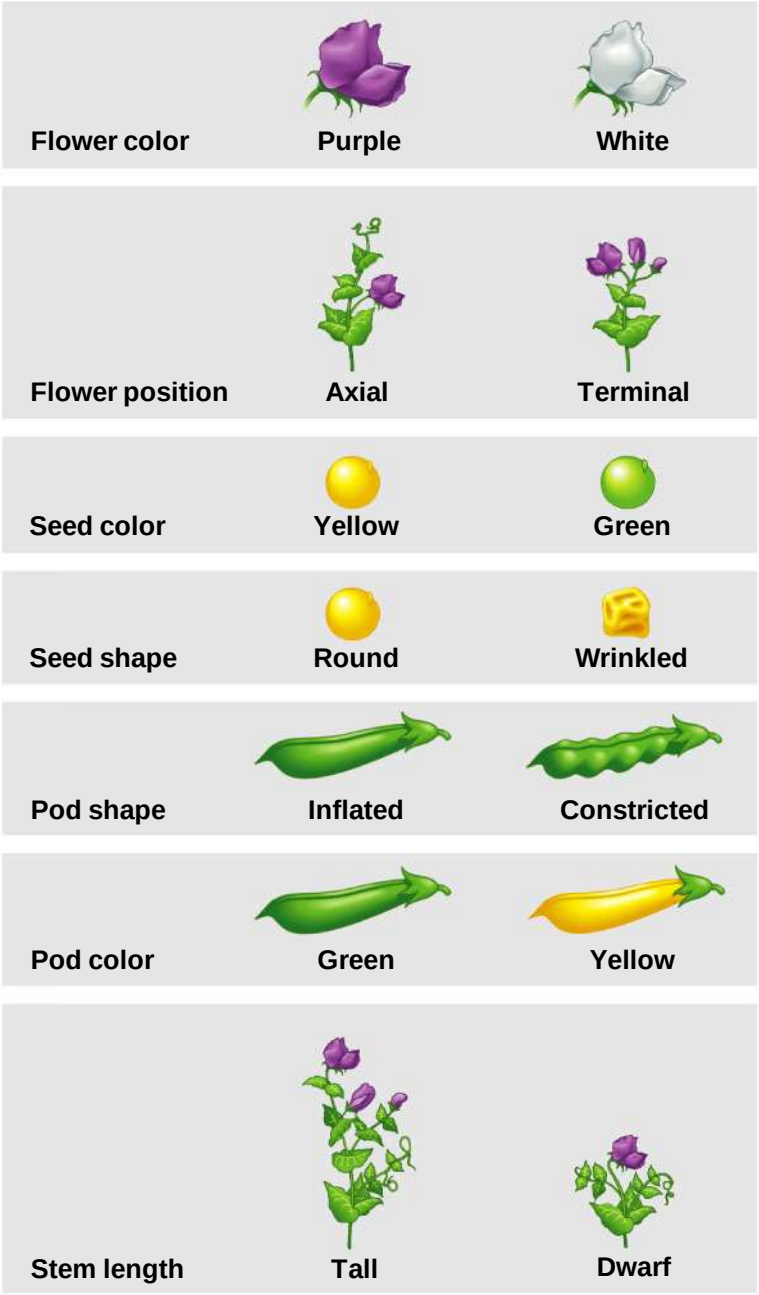

Terminology of Mendelian genetics
Write these in your book and know them

- Genotype
- Phenotype
- Allele
- Dominant allele
- Recessive allele
- Codominant allele
- Locus
- Homozygous
- Heterozygous
- Carrier
- Test cross

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

- 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

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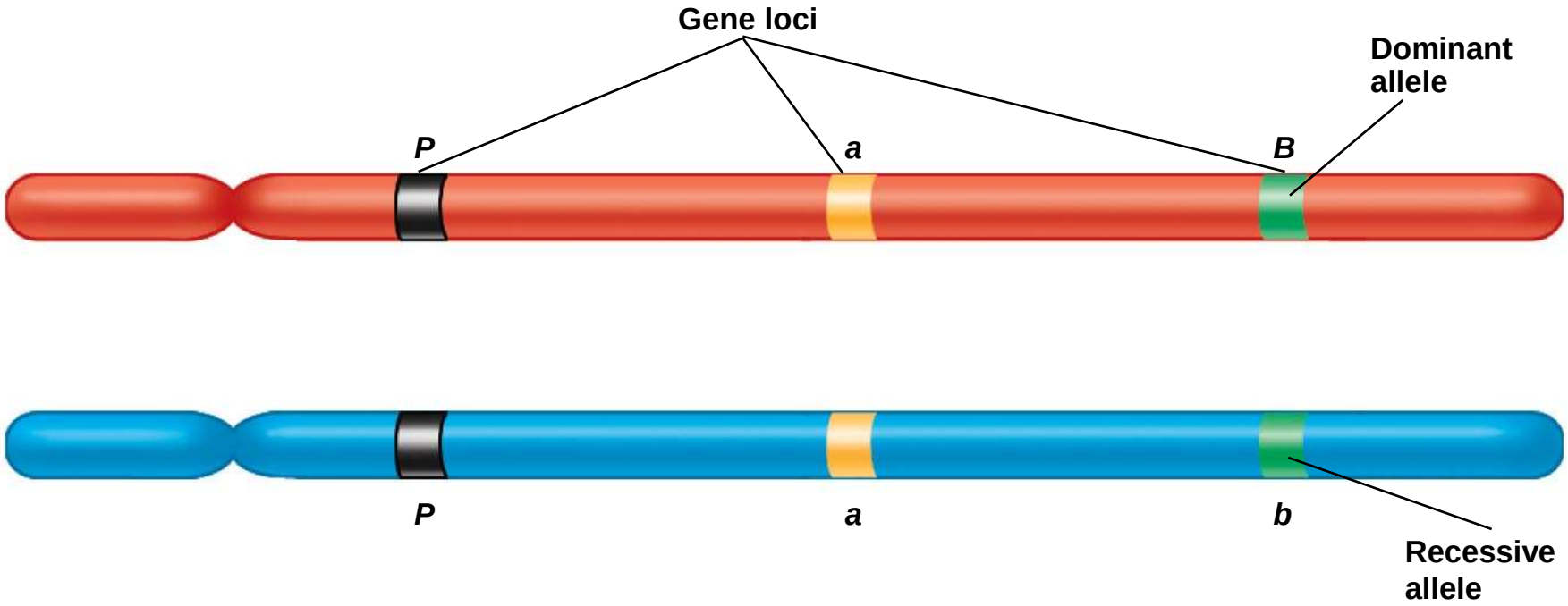


Genotype:	***PP***	***aa***	***Bb***
	Homozygous for the dominant allele	**Homozygous for the recessive allele**	**Heterozygous**

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:

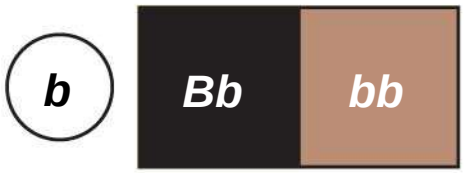
×

Genotypes ***B_*** ***bb***

Two possibilities for the black dog:

BB **or** ***Bb***

Gametes ***B*** ***B*** ***b***



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

Punnett Squares

- Primary 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

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

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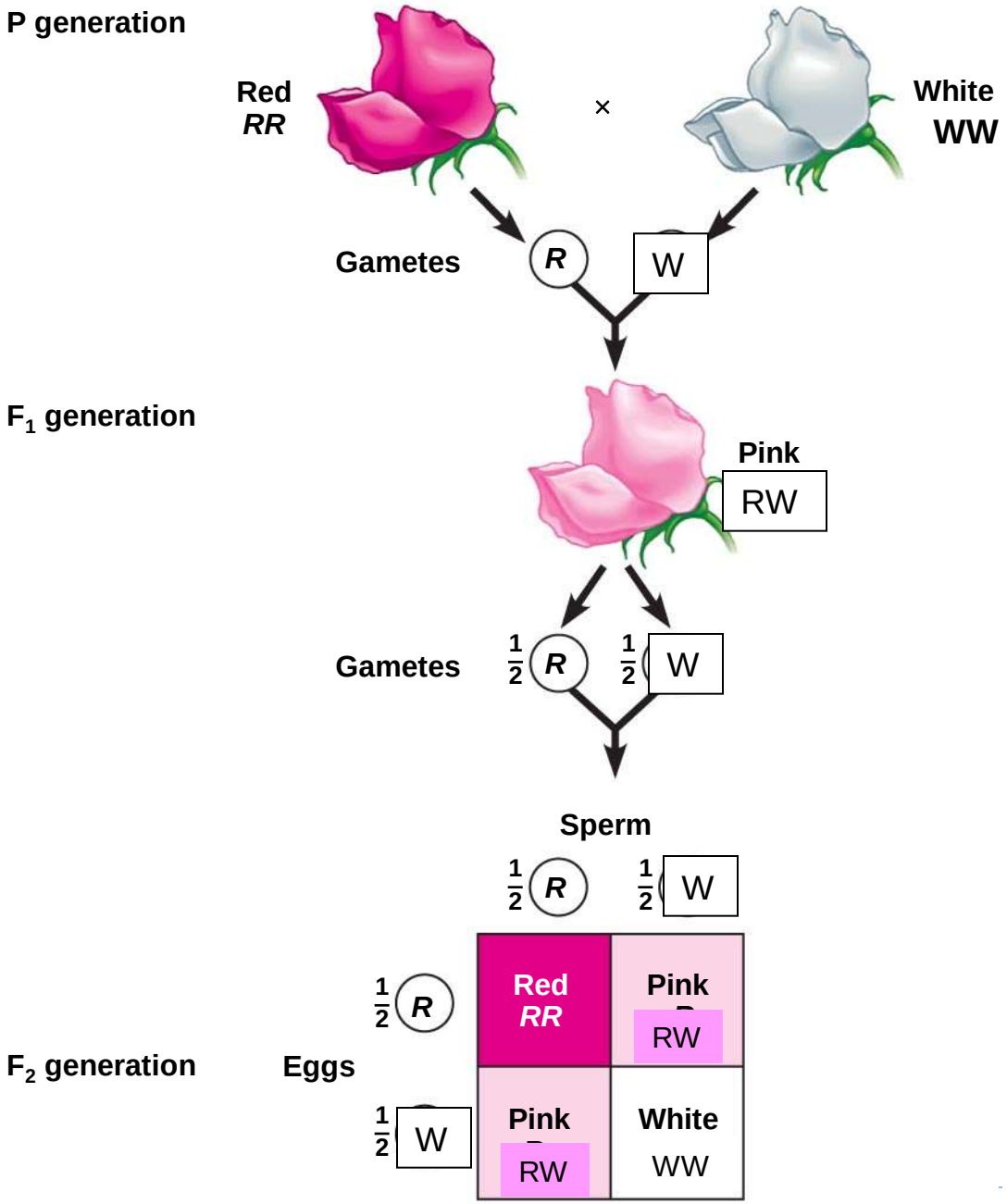


Co Dominance

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

Multiple Allels

Multiple Alleles

- Many times you may have three or more alleles for the same gene
- ABO Blood Typing
  - Four possible phenotypes A, B, AB and O
  - Three alleles of the gene
  - Produce six different genotypes

Blood Typing

- Blood typing represented by the letter I (I or i)
  - I^A = allele for type A
  - I^B = allele for type B
  - I = allele for type O

Blood Typing

- Possible combinations
  - $I^A I^A$ or $I^a i$ = type A
  - $I^B I^B$ or $I^b i$ = type B
  - $I^A I^B$ = type AB
  - ii = type O

Note a codominance??

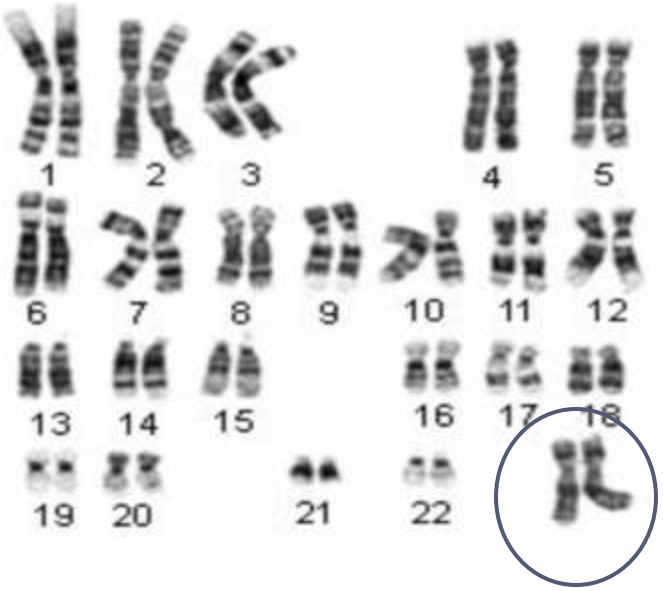
Sex Chromosomes

SEX CHROMOSOMES AND SEX-LINKED GENES

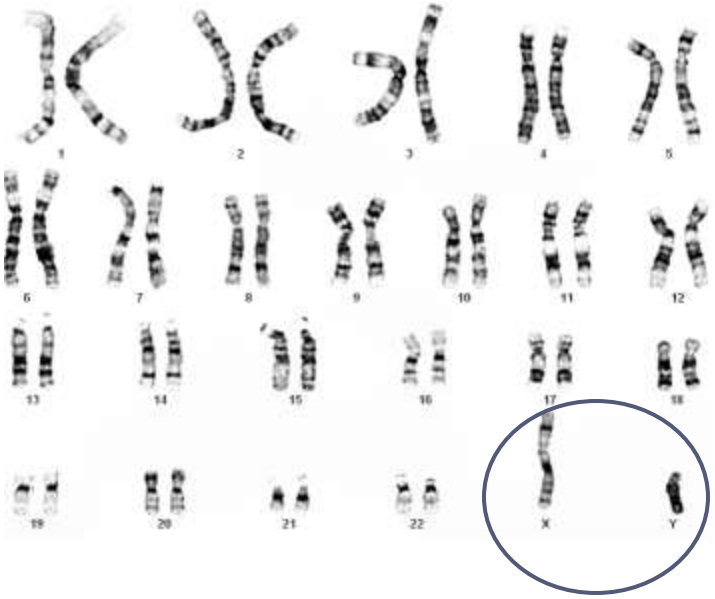
- Chromosomes determine sex in many species
  - Many animals have a pair of chromosomes that determine sex
    - Humans: X-Y system
      - Male is XY; the Y chromosome has genes for the development of testes
      - Female is XX; absence of a Y chromosome allows ovaries to develop

Sex Chromosomes

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



female



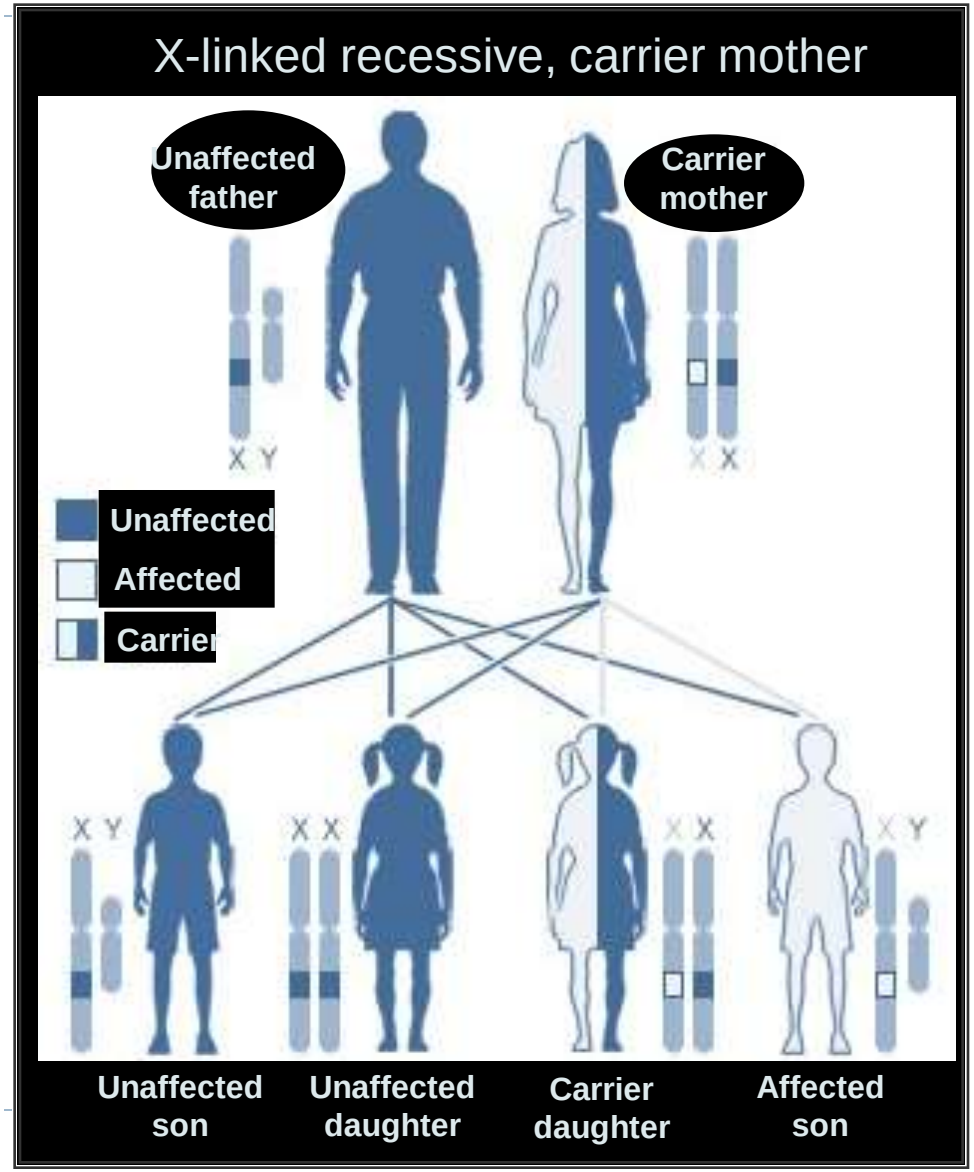
male

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
  - Haemophilia

Sex-Linked Chromosomes

- Since the alleles for colour blindness and haemophilia are found only on the X chromosome, the letter X is used
  - X^b
  - X^B

  - X^bY give the phenotype of an affected male

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