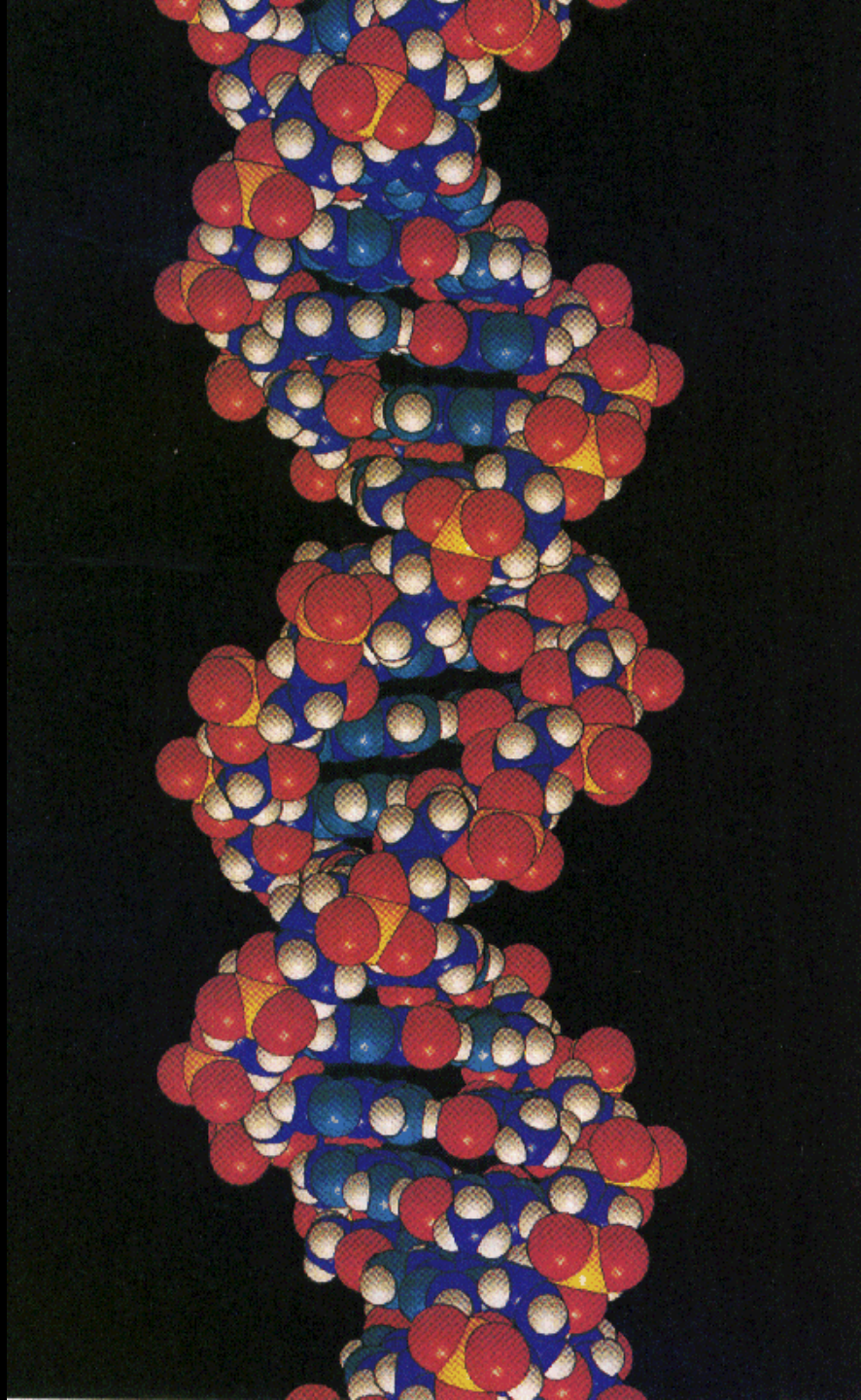
Year 10

Genetics

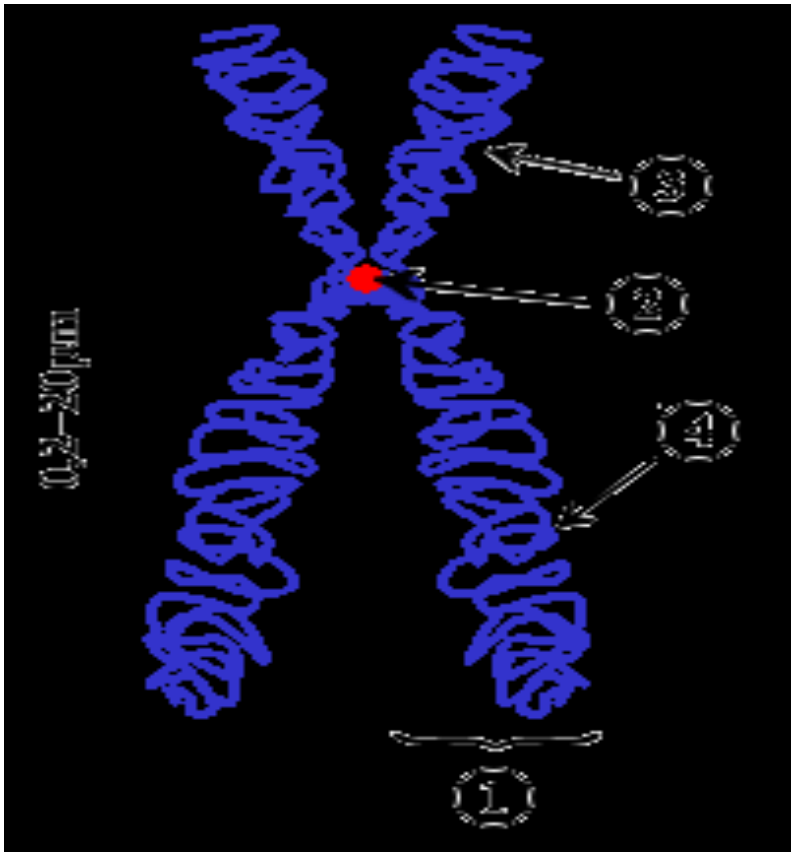


Objectives

- **Describe** the nucleus of cells, DNA, the structure of chromosomes and genes.
- **Explain** the nature of DNA, the genetic code, and how it can go wrong ie mutations.
- **Discuss** the significance and nature of variation, continuous and discontinuous.
- **Discuss** sex determination and the number of human chromosomes in sex cells and body cells.
- **Explain** genes and inheritance, dominant and recessive alleles, the role of chance and what are the possible offspring in a cross.
- **Explain** how to predict crosses, use a Punnet square to predict the outcome of a cross, do some simple genetics problems.
- **Explain** pedigrees and their use.

Chromosomes

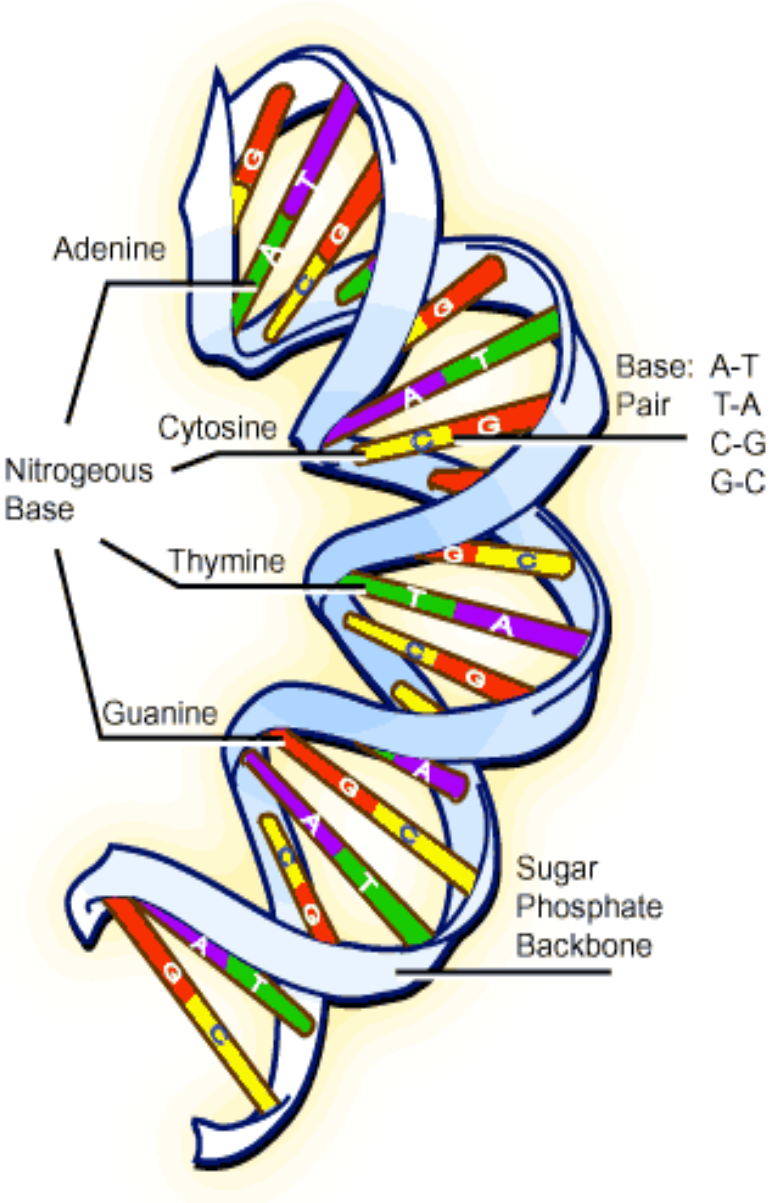
- **Chromosomes** are organized structures of DNA.
- Chromosomes contain a single continuous piece of DNA, which contains many genes.
- Chromosomes are found in the nucleus of the cell.



DNA

- DNA is a double stranded molecule found in the nucleus of the cells of living things.

- It is made up of about 3 billion pairs of 4 different bases, we use letters to represent the 4 bases found in all living things



A = adenine T = thymine

C = cytosine G = guanine

AGTCCGCGAATACAGGCTCGGTAGTCCGCGAATACAGGCTCGGT

- To view an animation of the structure of DNA click here.

http://learn.genetics.utah.edu/content/molecules/dna/

AGTCCGCGAATACAGGCTCGGTAGTCCGCGAATACAGGCTCGGT

DNA Deoxyribonucleic acid

- DNA is made up of four bases.
- These are

  A = adenine

  T = thymine

  C = cytosine

  G = guanine

- These bases form a sequence along the DNA strand.

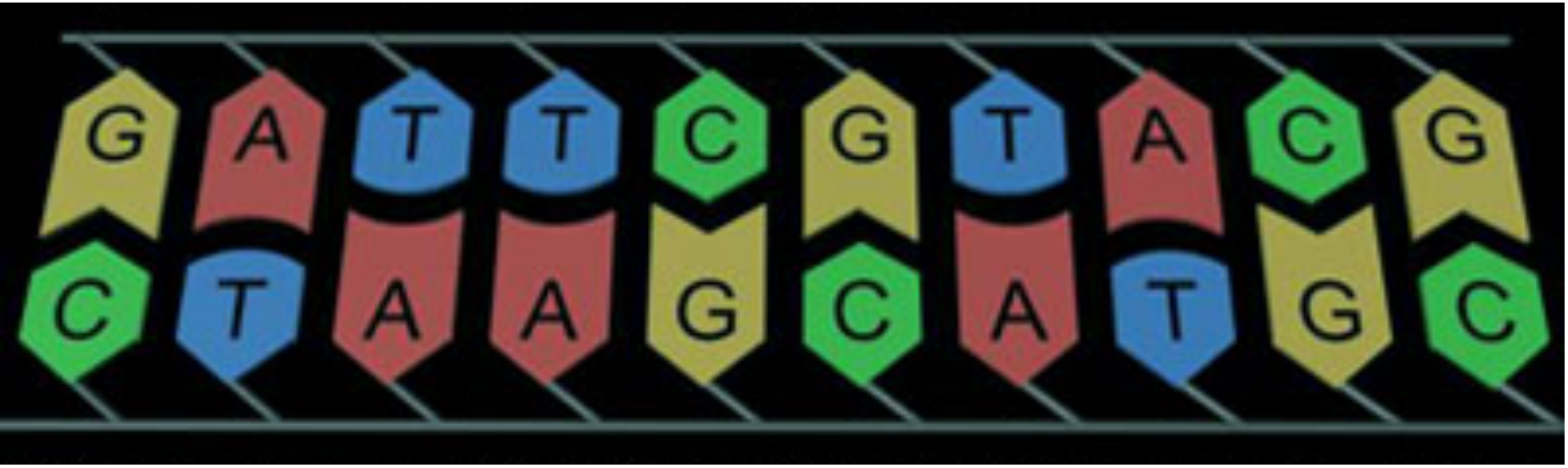
Eg; AGTCCGCGAATACAGGCTCGGT

This forms only one strand of DNA.

DNA is a double helix strand.

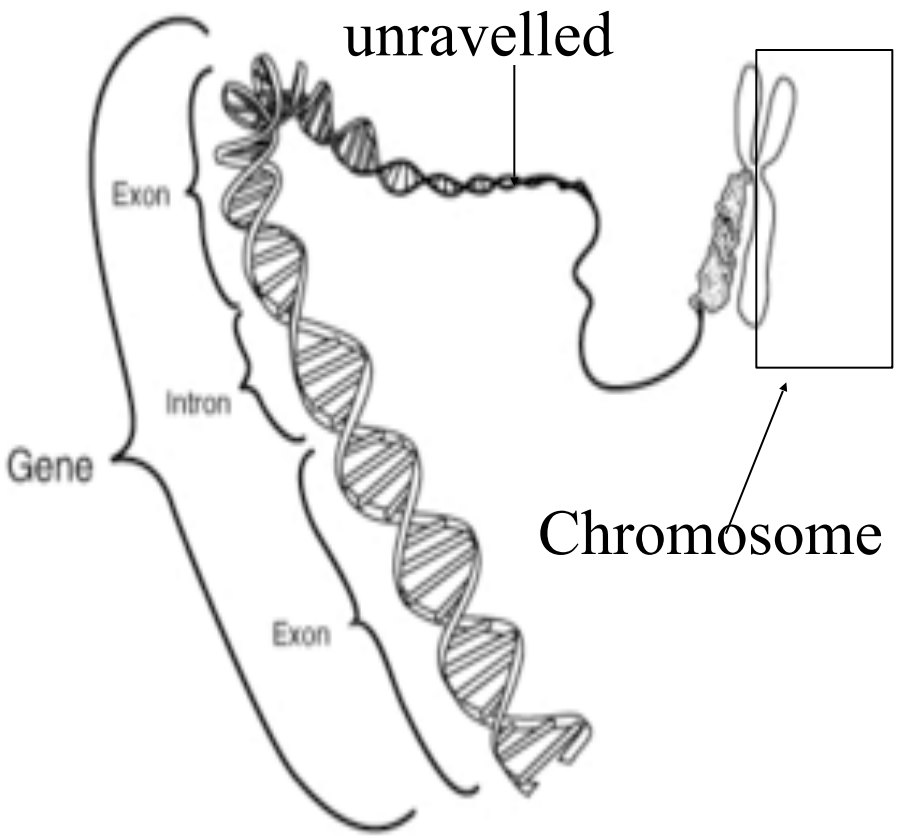
DNA Base pairing rule

- A and T pair together
- C and G pair together



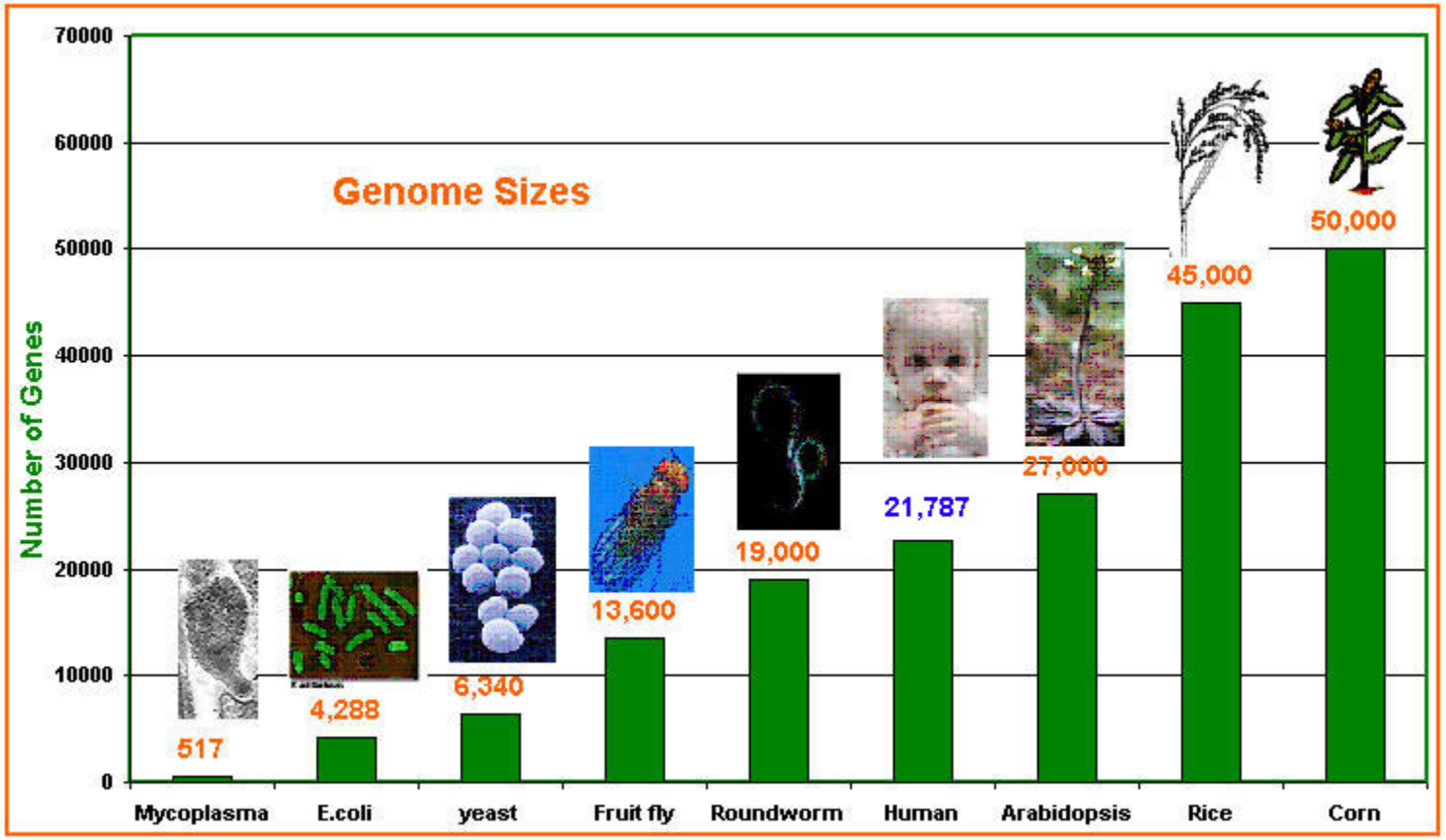
Genes

- A **gene** is a region of a DNA sequence, corresponding to a unit of inheritance.
- There are millions of genes.
- Genes determine our characteristics eg eye colour, height etc…
- Genes are inherited from our parents.

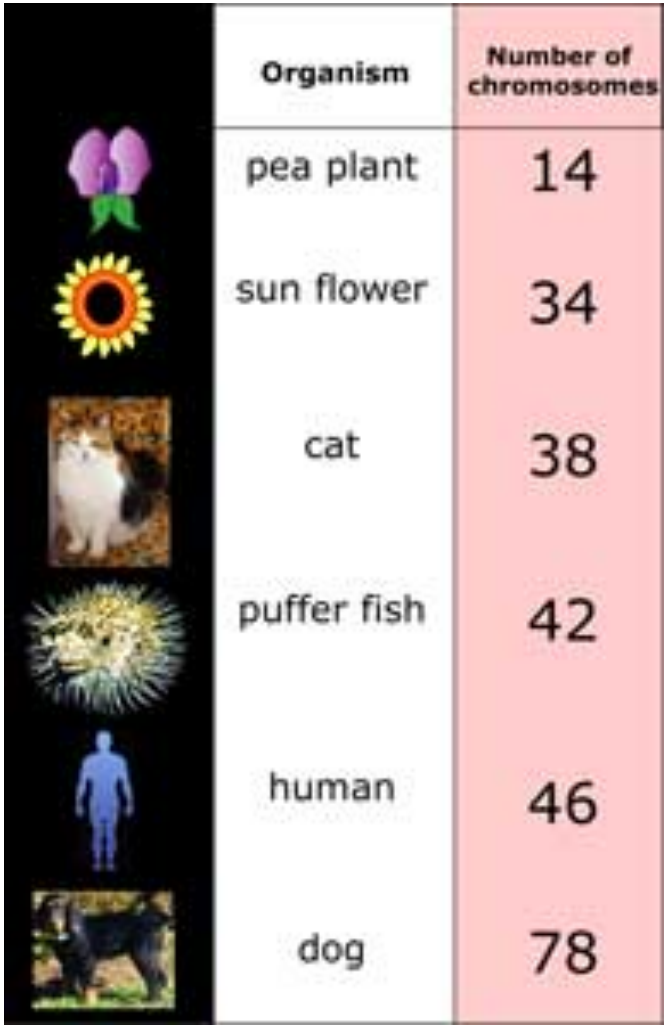


Number of genes

- This is a graph showing the number of genes in different species.



Different living things have different numbers of chromosomes

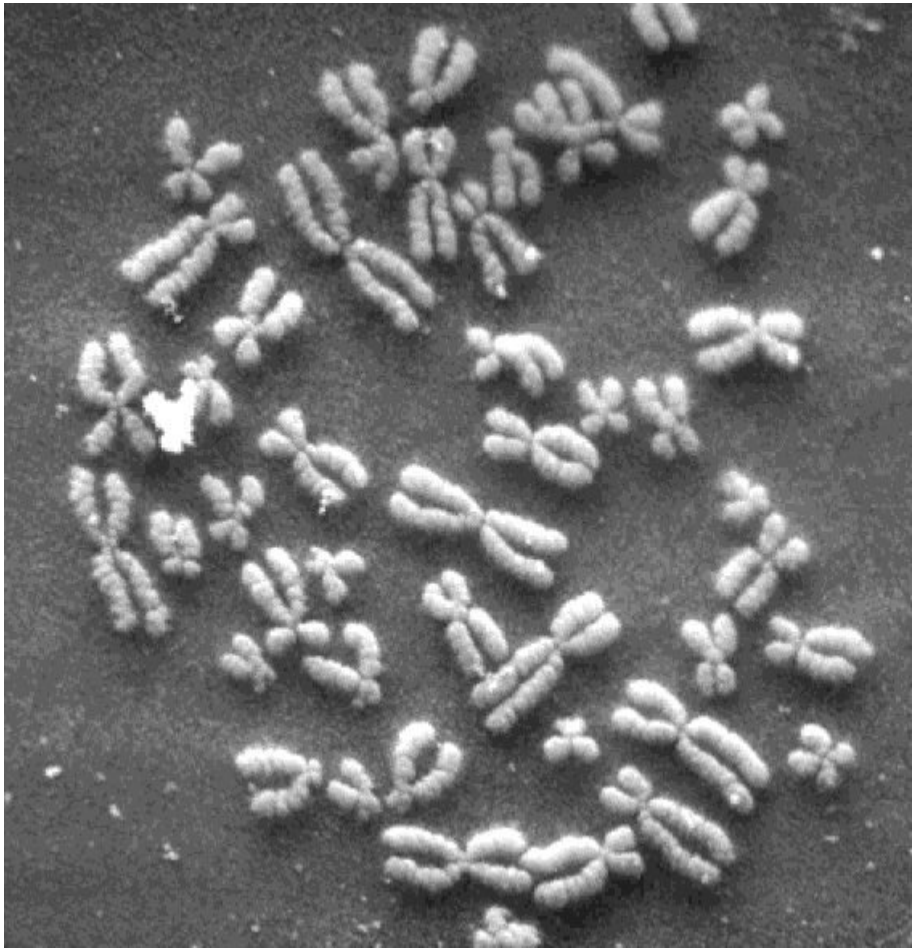


Organism	Number of chromosomes
pea plant	14
sun flower	34
cat	38
puffer fish	42
human	46
dog	78

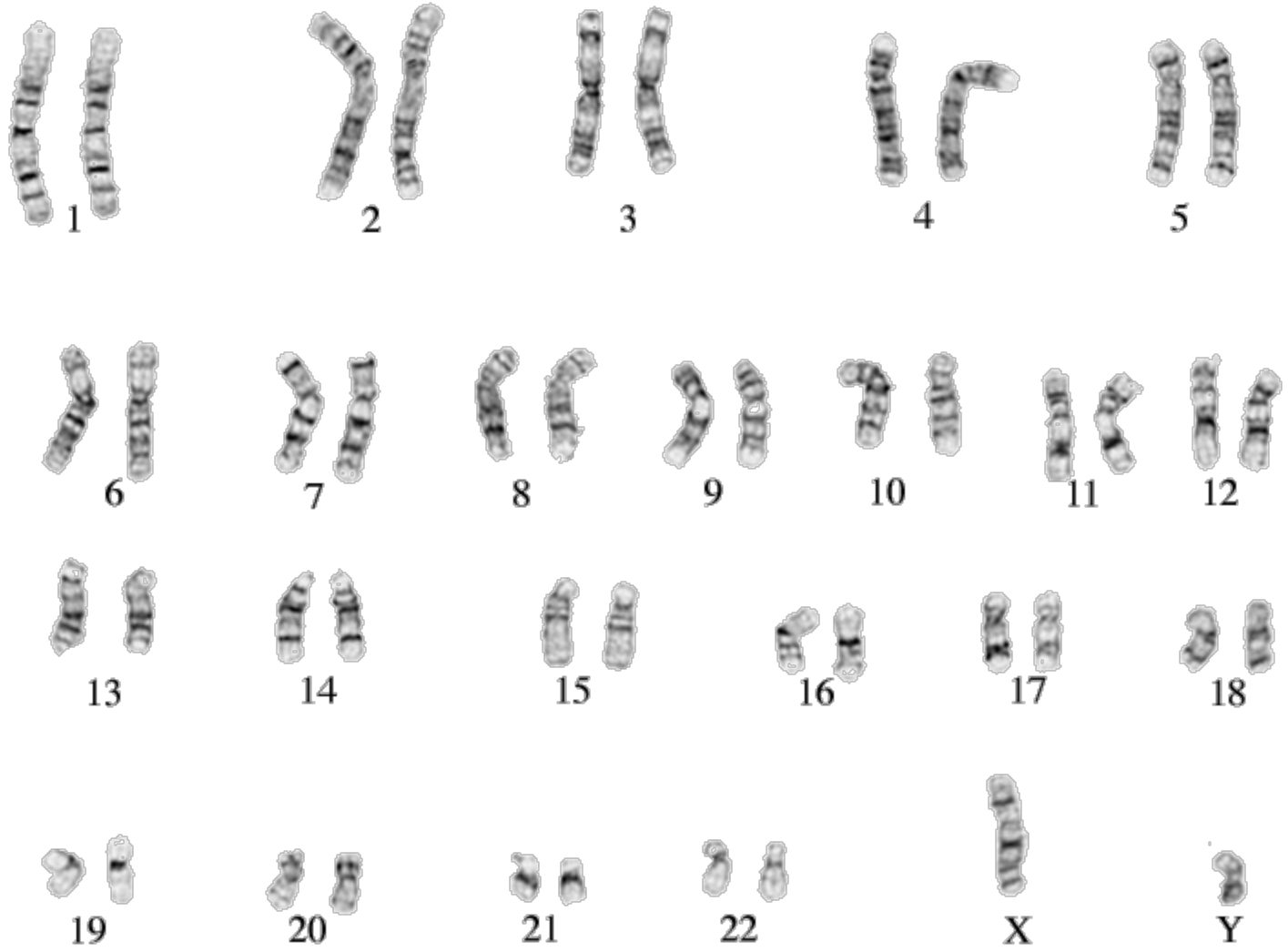
Karyotyping

- Karyotyping is used to detect genetic abnormalities. To do so, the cell containing the chromosomes is put into a hypotonic solution so that it will burst. The chromosomes can then be photographed.

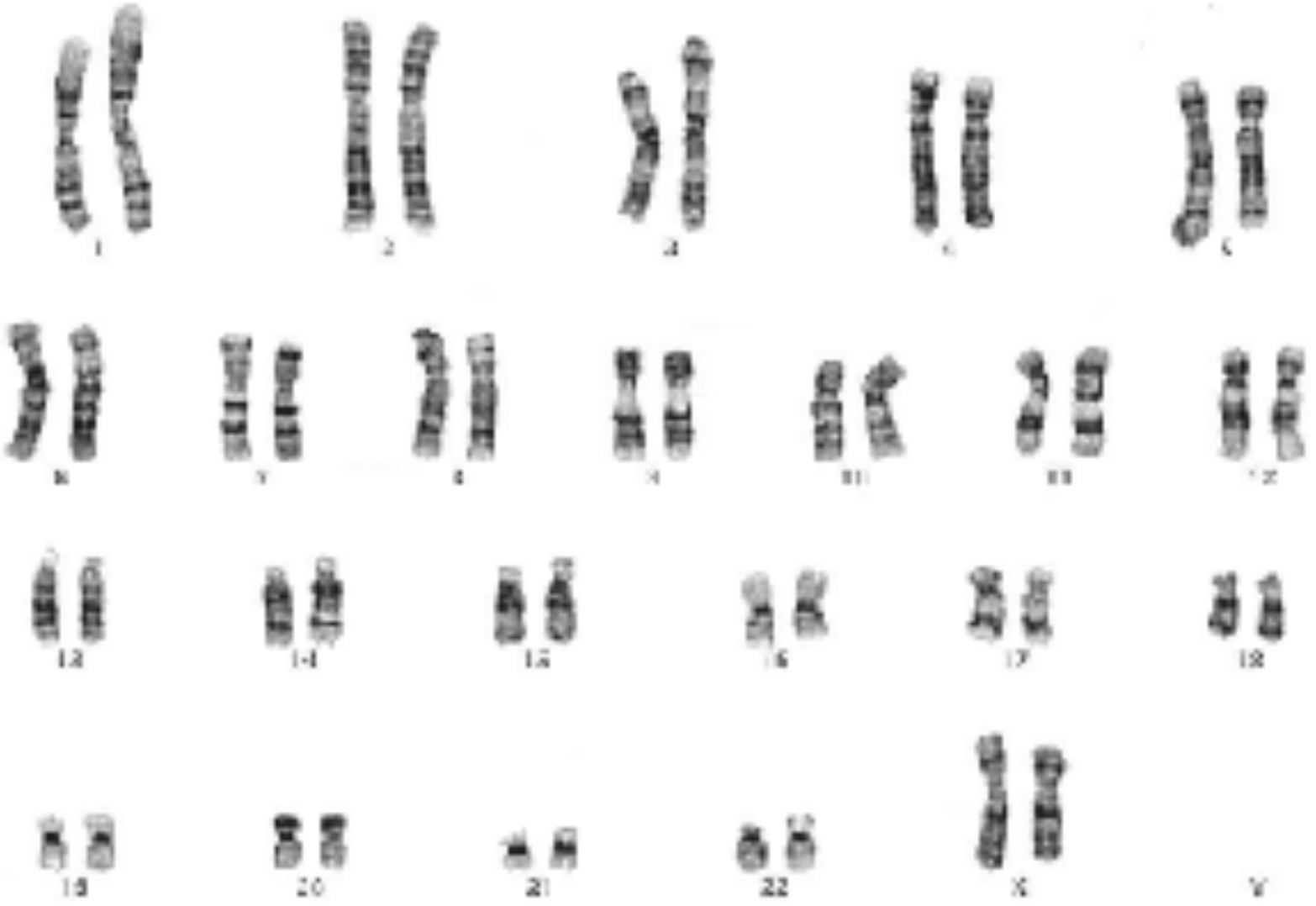
- the photographs are cut up and chromosomes are matched by size.



Human Male Karyotype

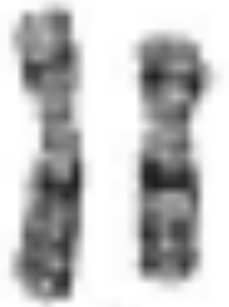


Human Female Karyotype

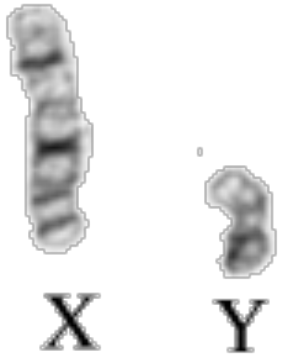


What is the difference ?

- The last pair of chromosomes is different in males and females.
- Females have two X chromosomes (XX)
- Males have one X and a smaller Y (XY)



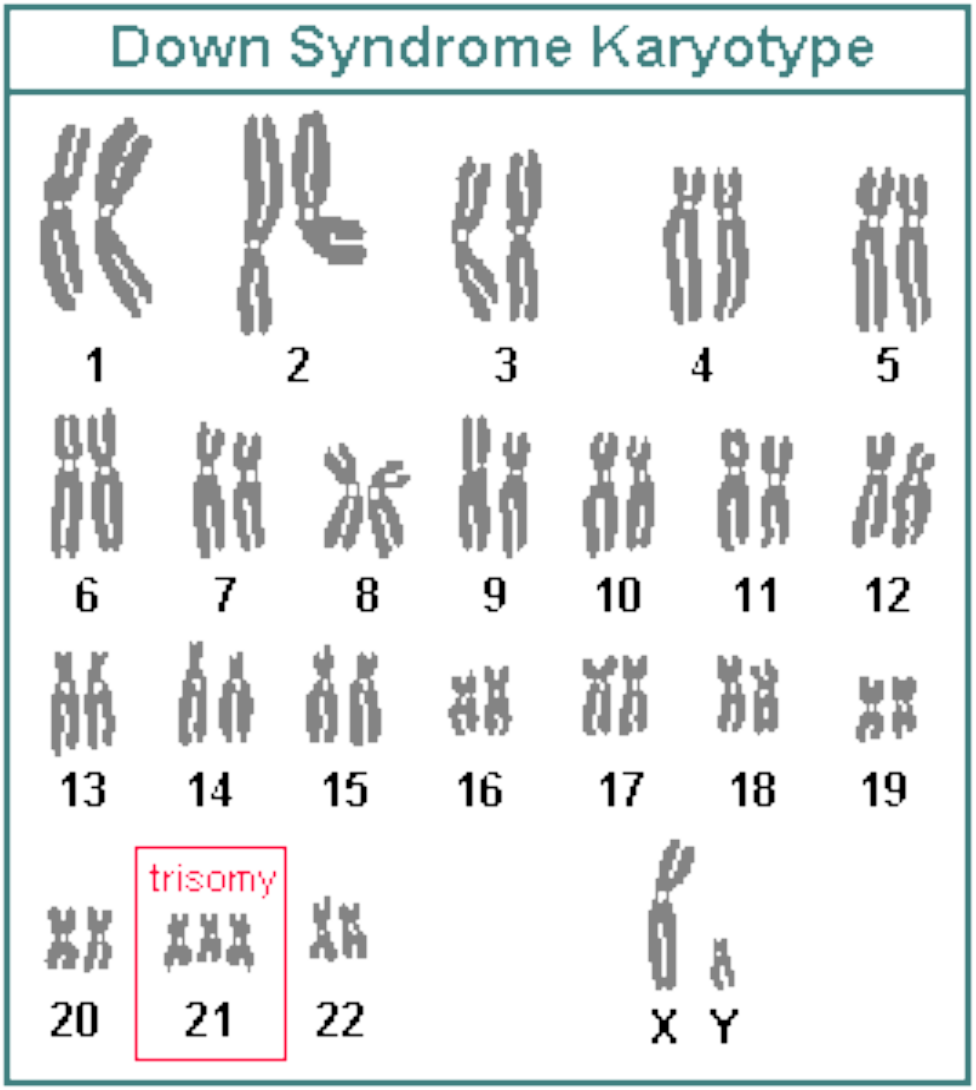
X X



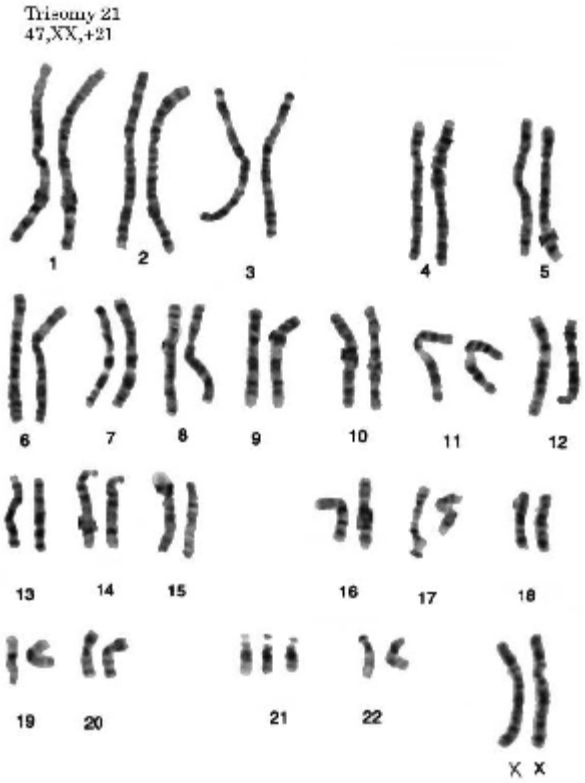
X Y

Complete the karyotype exercise on p95 Sci Pad

Karyotypes are often used to check for abnormalities in the chromosomes. Eg – a person with Down Syndrome has an extra copy of chromosome 21 in each of their cells.



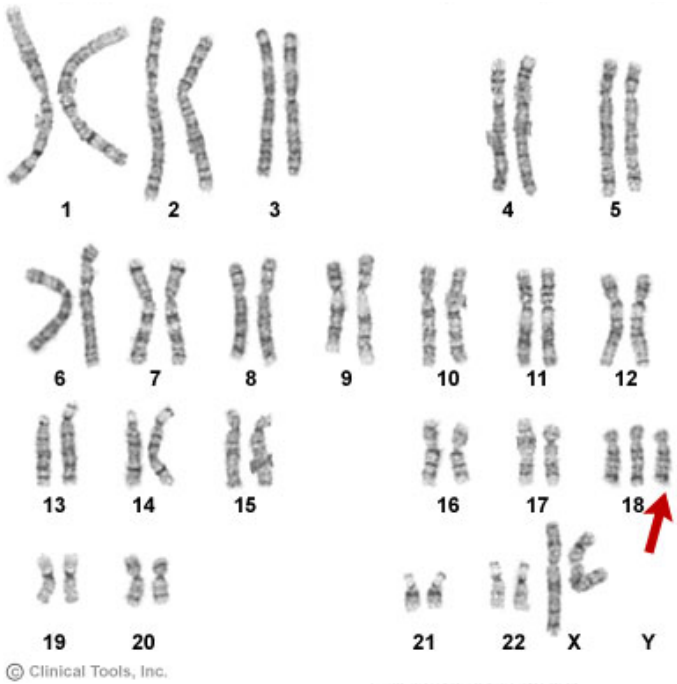
Down syndrome

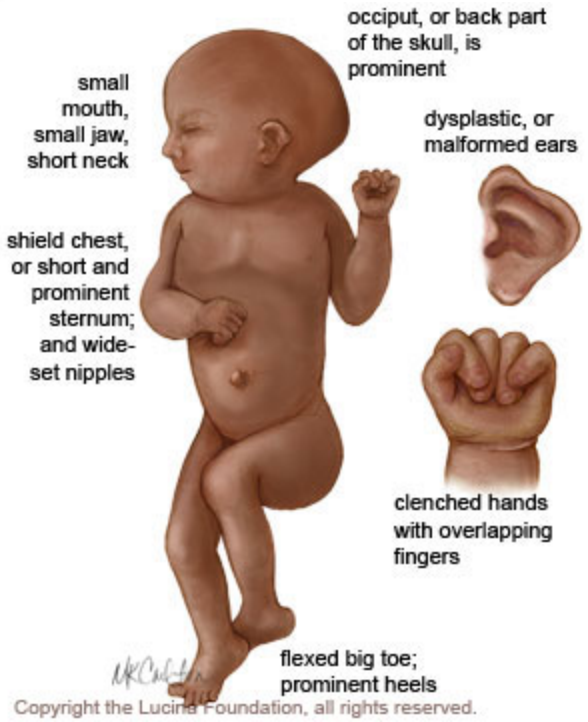


Trisomy 21

1 in 800 births in women of 30 to 31 (the rate increases with maternal age effect)

Features – growth failure, mental retardation, slanting eyes, broad flat face, short nose, abnormal ears, poor muscle tone, enlarged colon, congenital heart disease, one rib missing one on both sides, short broad hands.





Edward Syndrome

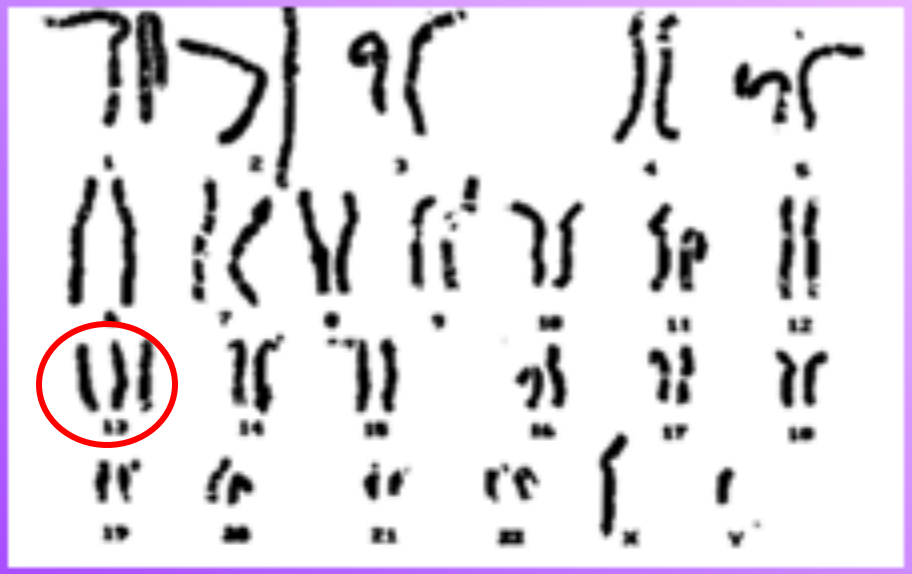
Trisomy 18

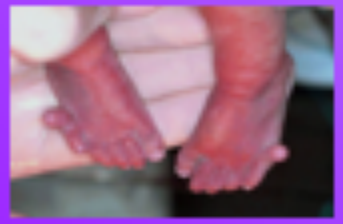
1 in 5000 live births (with a maternal age effect)

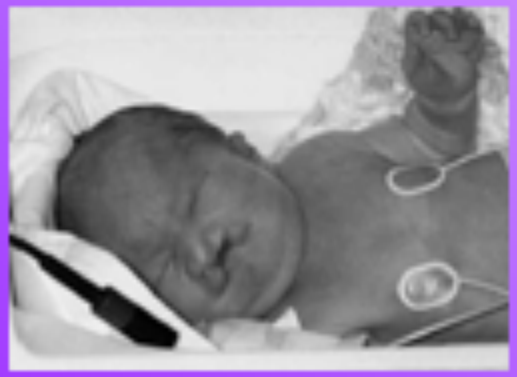
Features – severe mental retardation, low set malformed ears, heart defects, small mouth and rocker-bottom feet.

½ die by two months only a few have survived beyond a few years. 95% of foetuses are miscarried.

Patau syndrome







Trisomy 13

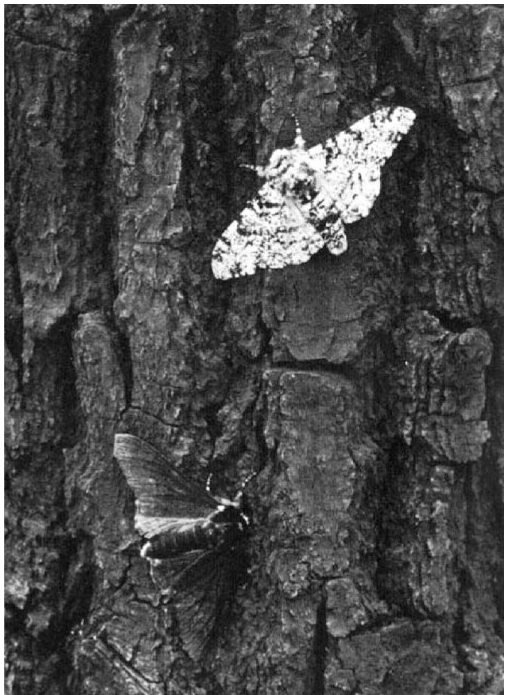
1 in 3000 live births (with a maternal age effect)

Features – retarded mental and physical development, eye defects, cleft palate and lip, extra fingers & toes, low set malformed ears, heart and spinal defects.

½ babies die by 1 month, one patient was known to survive to 10 years old.

Variation

- The difference between individuals within a species. Eg Pepper moth.
- Slight differences in the genetic code (the ordering of the bases A,C,G,T) lead to different traits/ characteristics.
- These differences cause variation.



Continuous and Discontinuous Variation

- **Discontinuous** variation refers to differences in characteristics that have a defined form.
- You can think of it as being either/or.

  Your earlobes are either attached or they are not.

Eg.

Tongue roller
Hitchhikers thumb
Attached lobes

- **Continuous** variation comes in a range of forms.

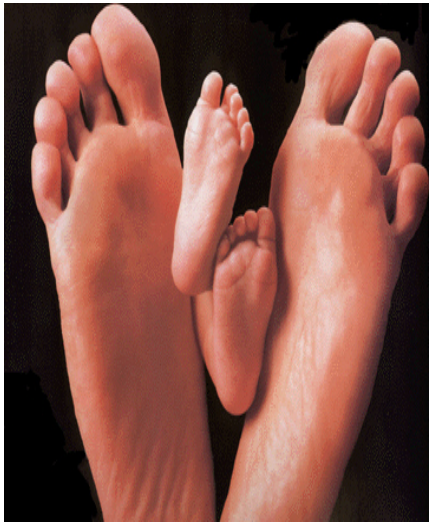
  Height - is not a set number or another.

Eg.

Hair Colour
Skin colour
Foot size

Discrete

Continuous



1. Dark Hair
LL, Ll

light hair
ll

2. Curly hair
TT, Tt

straight hair
tt

3. Curl's tongue
CC, Cc

can't curl tongue
cc

4. Mid-digital
hair present
MM, Mm

mid-digital
hair absent
mm

5. Eyes not blue
EE, Ee

blue eyes
ee

6. Widow's peak
WW, Ww

no peak
ww

7. bent little finger
BB, Bb

straight little finger
bb

How does genetic variation occur?

Changes in the genetic code cause genetic mutations.

Sometimes these mutations can be bad causing death or illnesses.

However sometimes these mutations can be beneficial. Eg longer neck in a giraffe.

Genetic Mutations

- The genetic code is altered
- Eg CCTAGCTGCTACGTAA
  becomes
  CCTAGCTGCAACGTAAACTGTCATC
- Only one base needs to be changed to give rise to a mutation.

Genetic Mutations

- Genetic mutations are caused by;

➢Errors in DNA copying(when new cells are formed)

➢DNA damage.

These errors are called mutations.

- Mutations create <u>new genes</u>

What causes DNA damage?

Mutagens

- **Chemical Mutagens** change the sequence of bases in a DNA strand. E.g. chemicals in tobacco smoke.
- **Radiation** High energy radiation from a radioactive material or from X-rays is absorbed by the atoms in water molecules surrounding the DNA, heating it and altering it.
- **Sunlight** contains ultraviolet radiation. This radiation can damage the DNA structure.
- **Spontaneous** mutations occur without exposure to any obvious mutagenic agent.

Mutation

Harmful mutations
(Make you worse)

Beneficial mutations
(Make you better)

Silent Mutations
(Don’t make you better or worse)

What does a genetic mutation do?

- **mutations** can affect the outward appearance of an individual. Mutations can change the height of a plant or change it from smooth to rough seeds.
- Mutations often cause diseases such as

❖ Muscular Dystrophy
❖ Huntington's Disease
❖ Cystic fibrosis

Horse mutation

- In horse racing, the difference between a trot and a gallop is crucial knowledge to have.
- As a horse increases its speed it will normally switch from trot to gallop, which is the natural gait at high speed, but this leads to disqualification for trotters.
- Now researchers at Uppsala University in Sweden have discovered a mutation in a single gene in horses that inhibits the transition from trot to gallop, allowing a horse to trot at very high speed.
- The tweak to gene 'DMRT3' controls a horse's skills in ambling gaits, pacing and galloping.

What can genetic mutations look like

- Scottish Fold is a breed of cat with a slight genetic mutation.

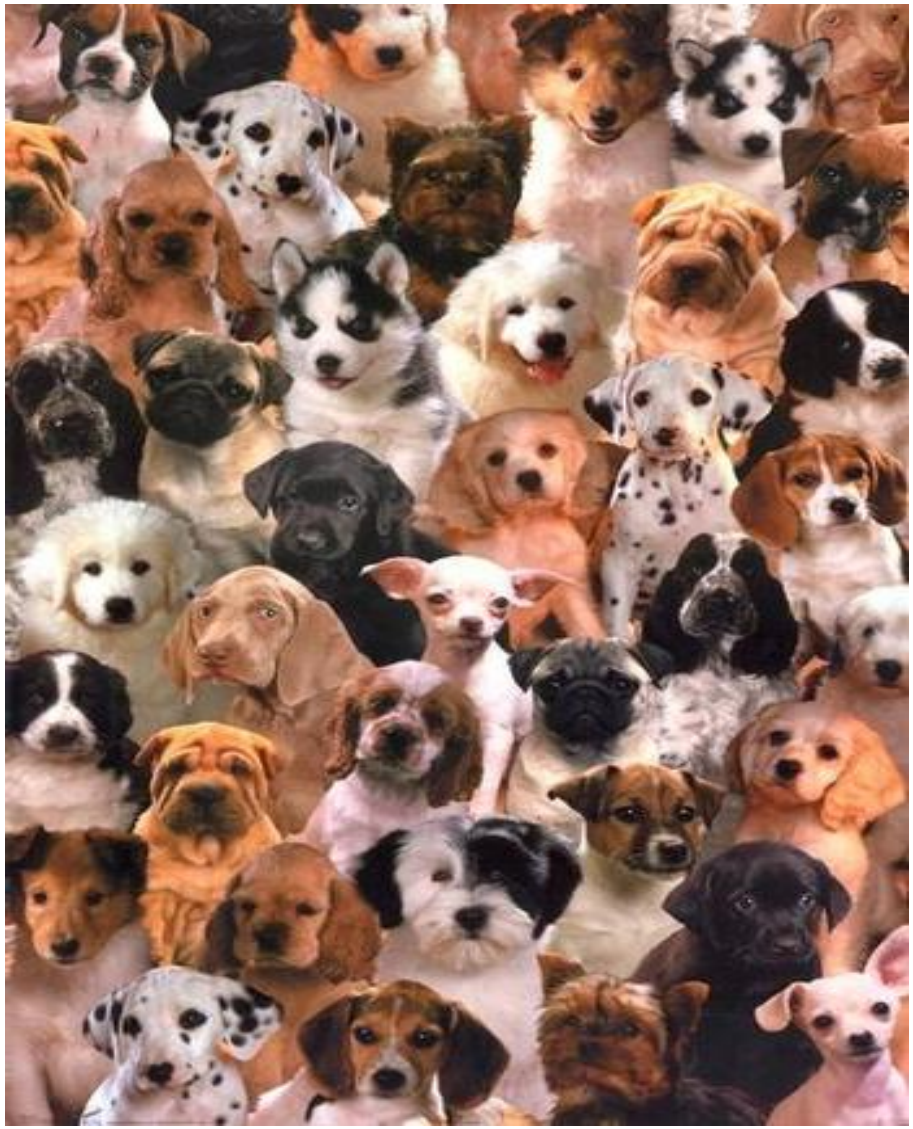








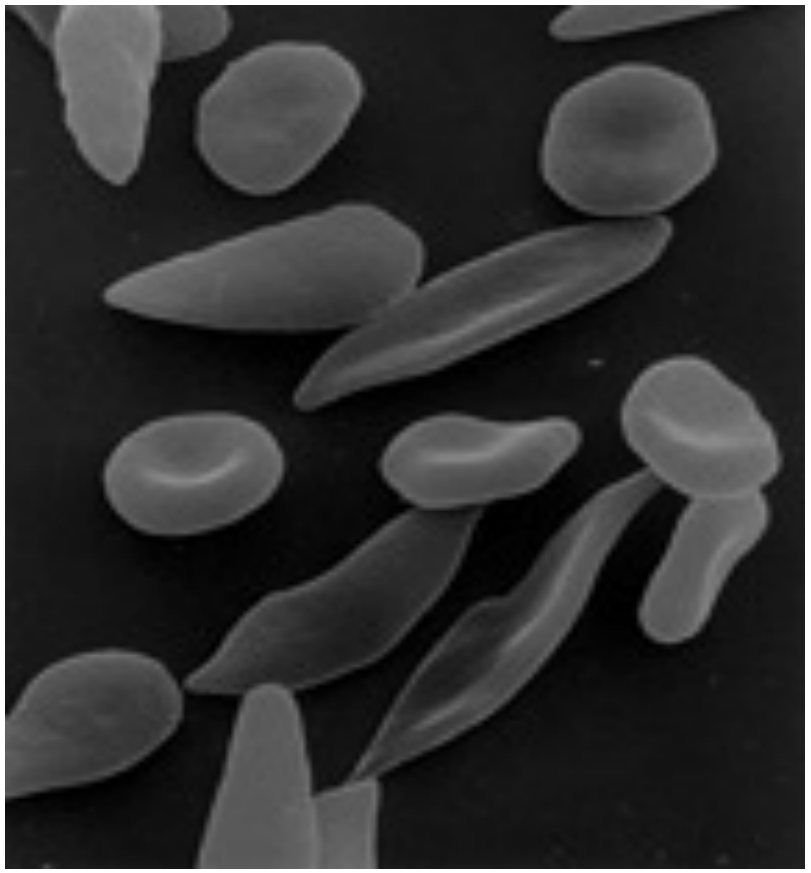

Why is genetic variation so important.



- In breeding of dogs and other animals reduces the variation of the individual and makes it more susceptible to disease and illness

Sickle-cell disease

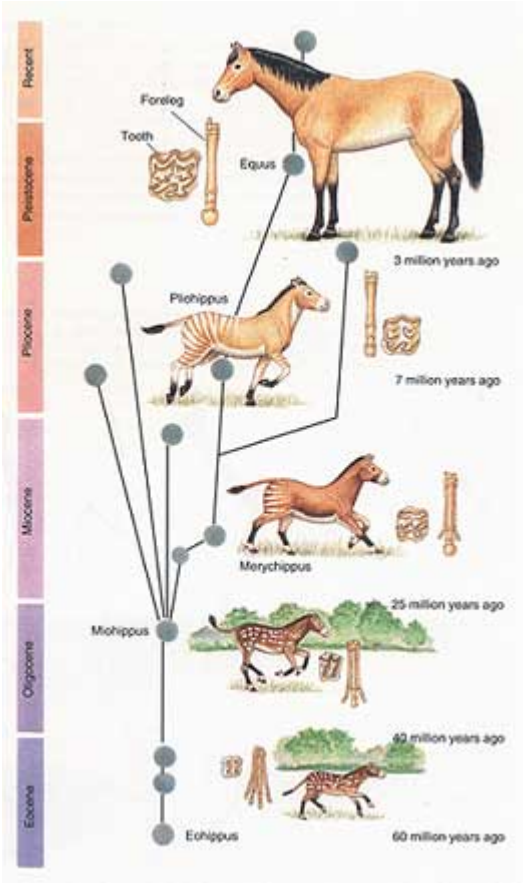
- **Sickle-cell disease** is a blood disorder characterised by red blood cells that assume an abnormal, rigid, sickle shape.
- The disease is chronic and lifelong: individuals are most often well, but their lives are punctuated by periodic painful attacks.
- Sickle-cell anaemia is caused by a mutation in the haemoglobin gene.

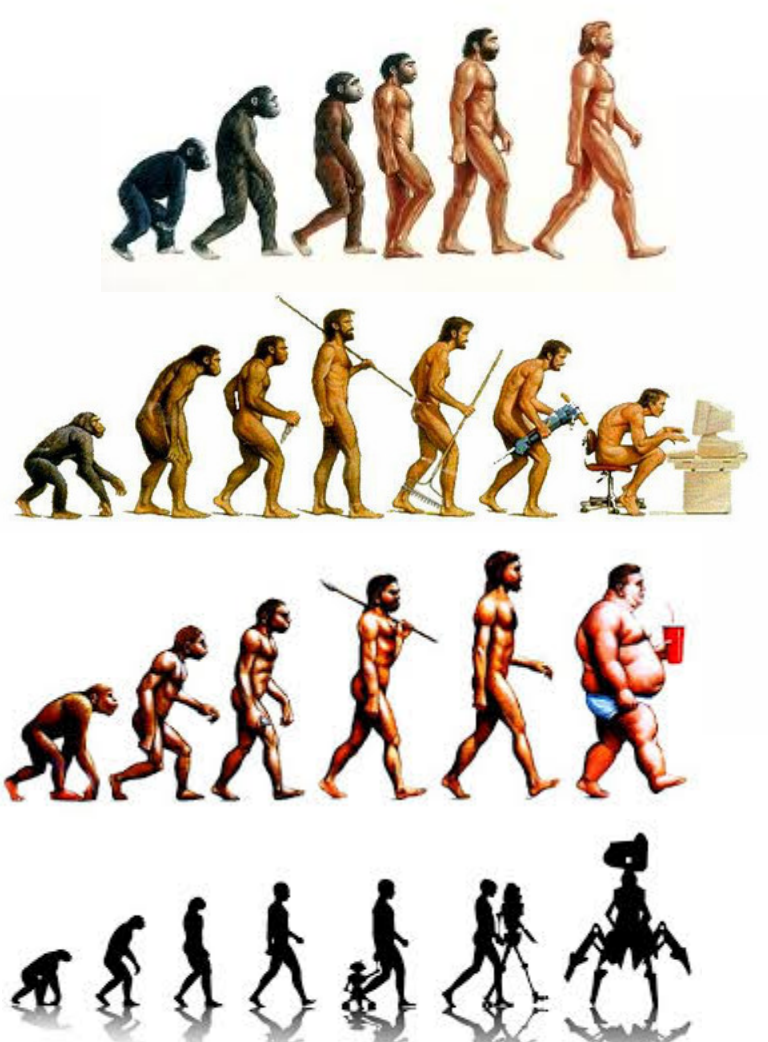


Achondroplasia/Dwarfism.

- Achondroplasia is a result of an mutation in the growth factor receptor gene.

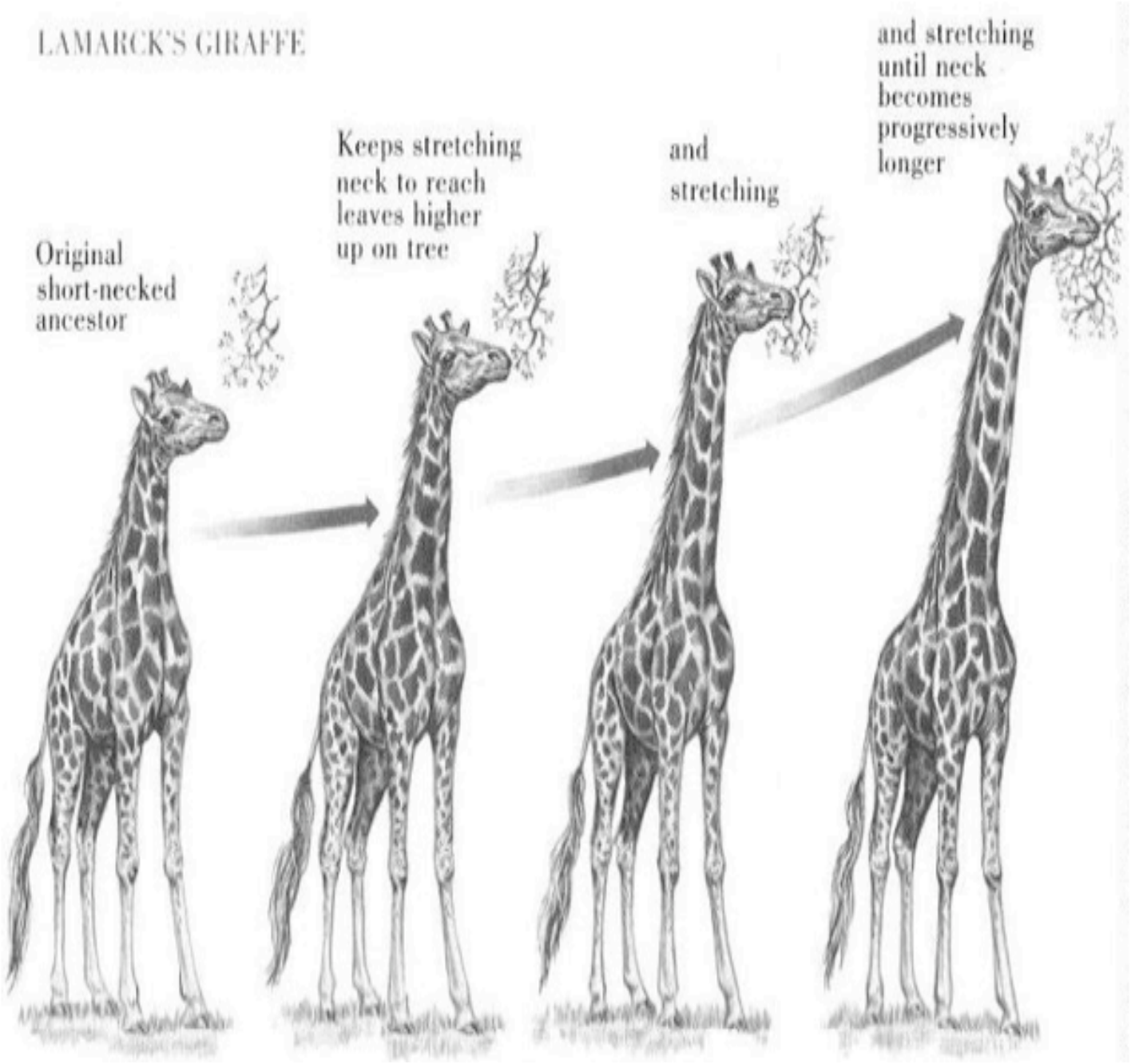
Evolution

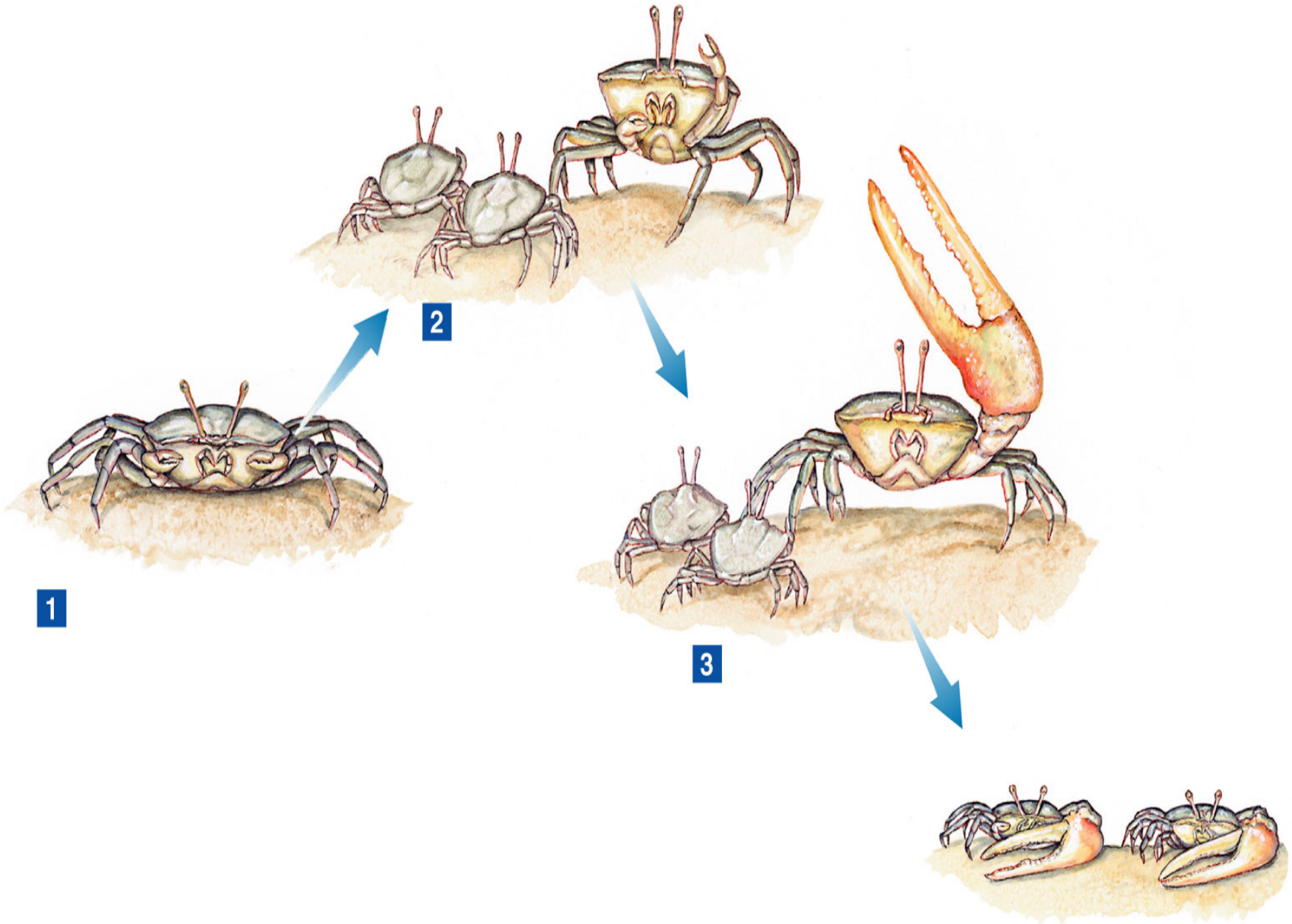

Recent
Pleistocene
Pliocene
Miocene
Oligocene
Eocene
Forefog
Tooth
Equus
3 million years ago
Pliohippus
7 million years ago
Merychippus
25 million years ago
Miohippus
40 million years ago
Eohippus
60 million years ago



Lamarck's Theory of Evolution

- Tendency toward Perfection(Giraffe necks)
- Use and Disuse (bird's using forearms)


LAMARCK'S GIRAFFE
Original short-necked ancestor
Keeps stretching neck to reach leaves higher up on tree
and stretching
and stretching until neck becomes progressively longer


1
2
3

Natural Selection

Inference: Those organisms that are better adapted to their environment have a greater likelihood of surviving to adulthood and passing these characteristics on to their offspring.

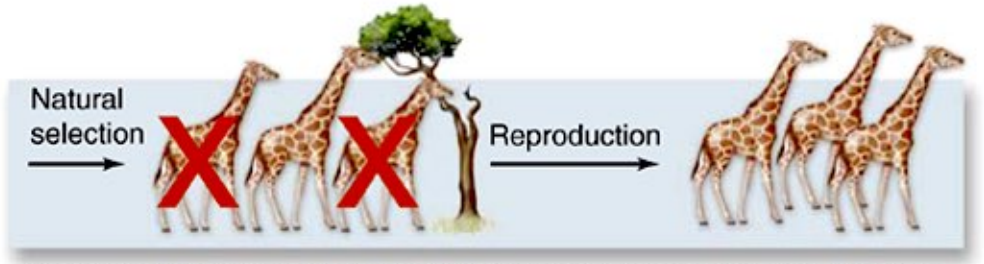
Survival of the “fittest.”

Survival of the "fittest."

Darwin's theory for how long necks evolved in giraffes

Some individuals born happen to have longer necks due to genetic differences.

Individuals pass on their traits to next generation.



Over many generations, longer-necked individuals are more successful, perhaps because they can feed on taller trees, and pass the long-neck trait on to their offspring.

b. Darwin's theory: natural selection or genetically-based variation leads to evolutionary change.

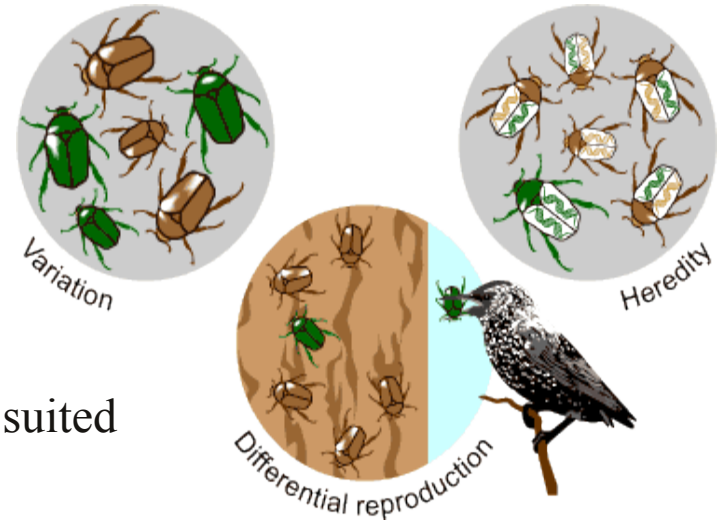
Natural selection

Lots of babies
Population produces too many offspring, many will die

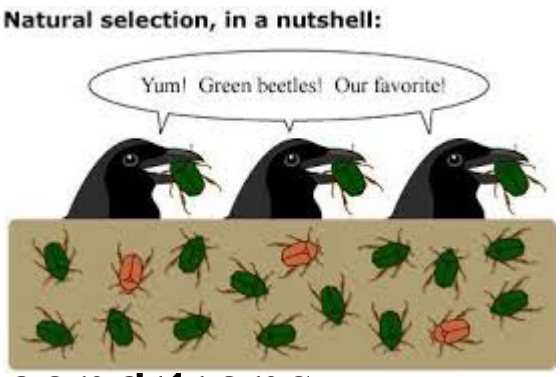
Variation
Individuals show variation: some variations are more favorable than others

Natural Selection
Natural selection favors the **best** suited at the time

Inheritance
Variations are inherited. The best suited variants leave more offspring.



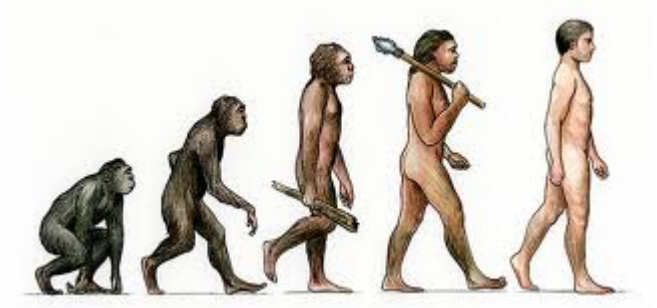
Natural Selection

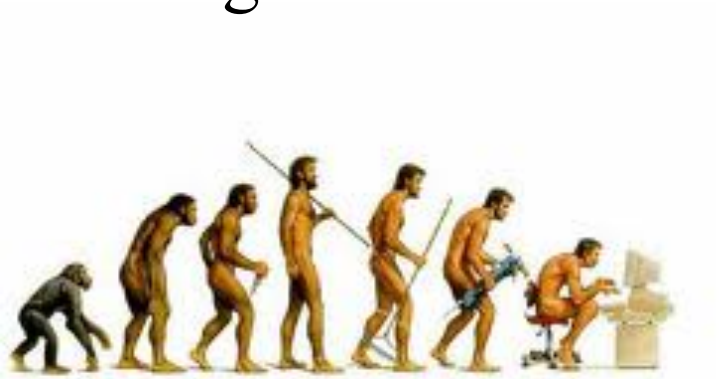


- **Natural selection:** environmental conditions determine which individuals in a population survive to produce the most offspring
- 3 conditions for natural selection to occur
  - Variation must exist.
  - Variation must result in more offspring surviving.
  - Variation must be genetically inherited.

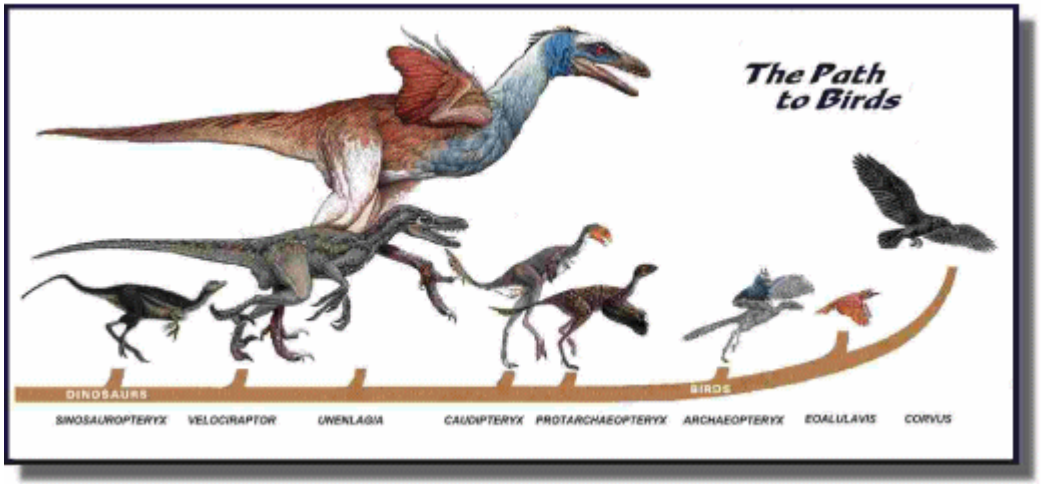
Natural selection causes evolution

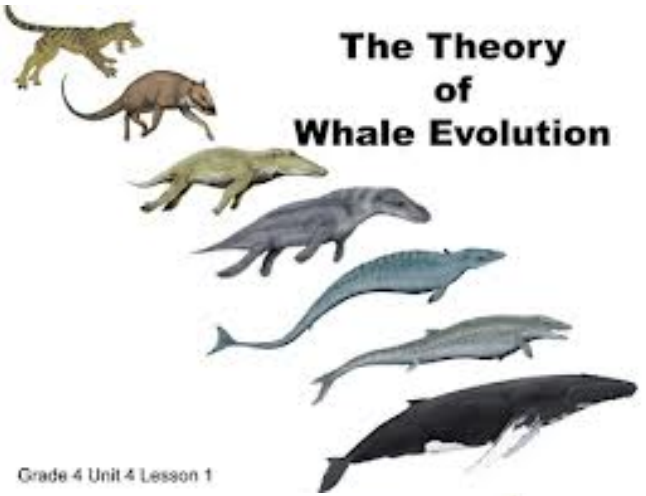
- Evolution is the slow process that changes animals and plants.
- Evolution is driven by changes in environments such as climate change.
- Evolution is the result of natural selection.
- Evolution is constantly occurring.

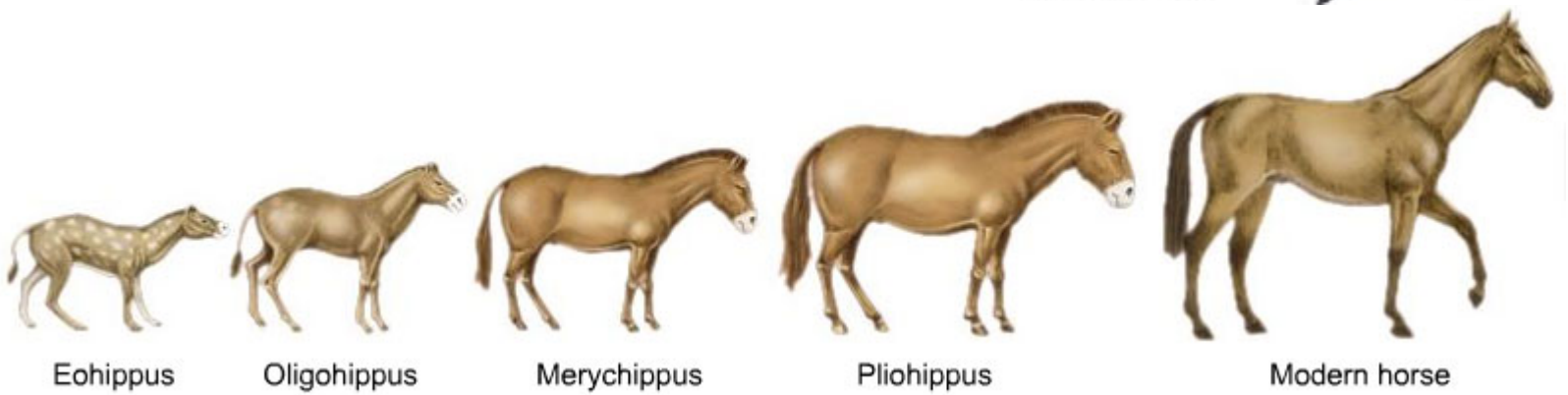




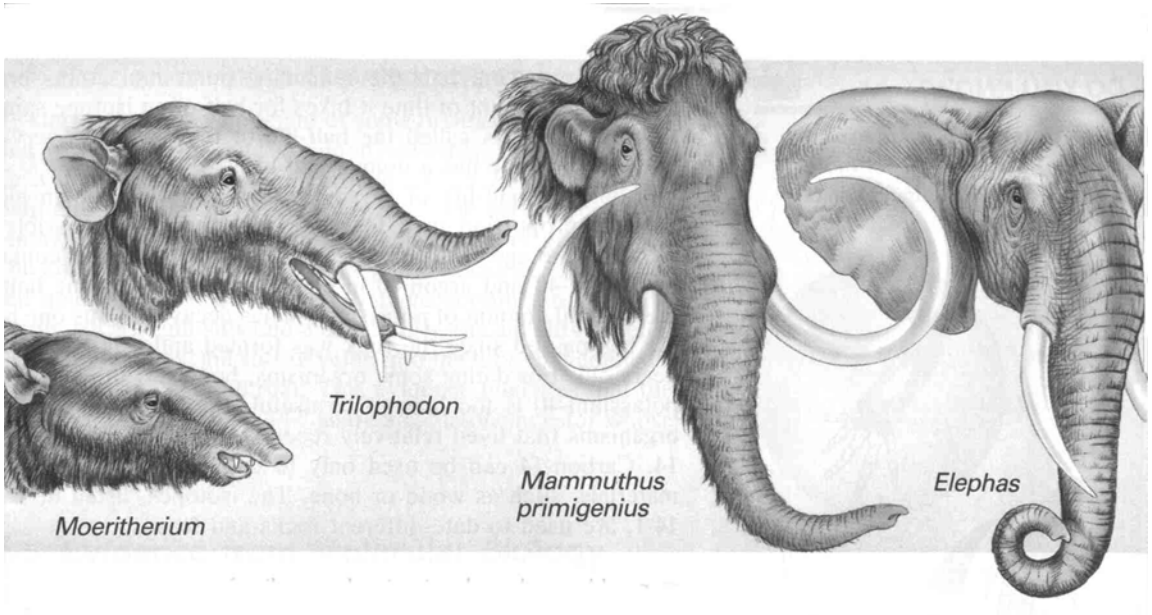
Examples of evolution



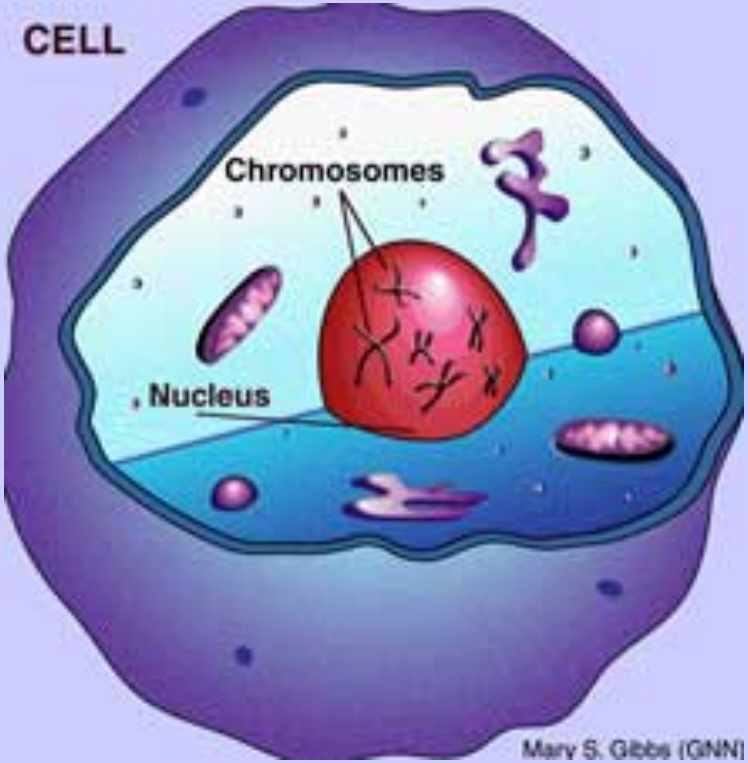




Evolution of elephanrs.



Chromosomes



- Chromosomes are found in the nucleus, they are made of DNA. DNA never leaves the nucleus.
- Humans have 23 pairs of chromosomes (a total of 46) in every cell in our bodies (except our eggs or sperm – they have a half set of only 23 chromosomes.)

Genes

Genes are the basic units of heredity in living cells.

They consist of a length of DNA that contains instructions ("codes") for making a specific protein.

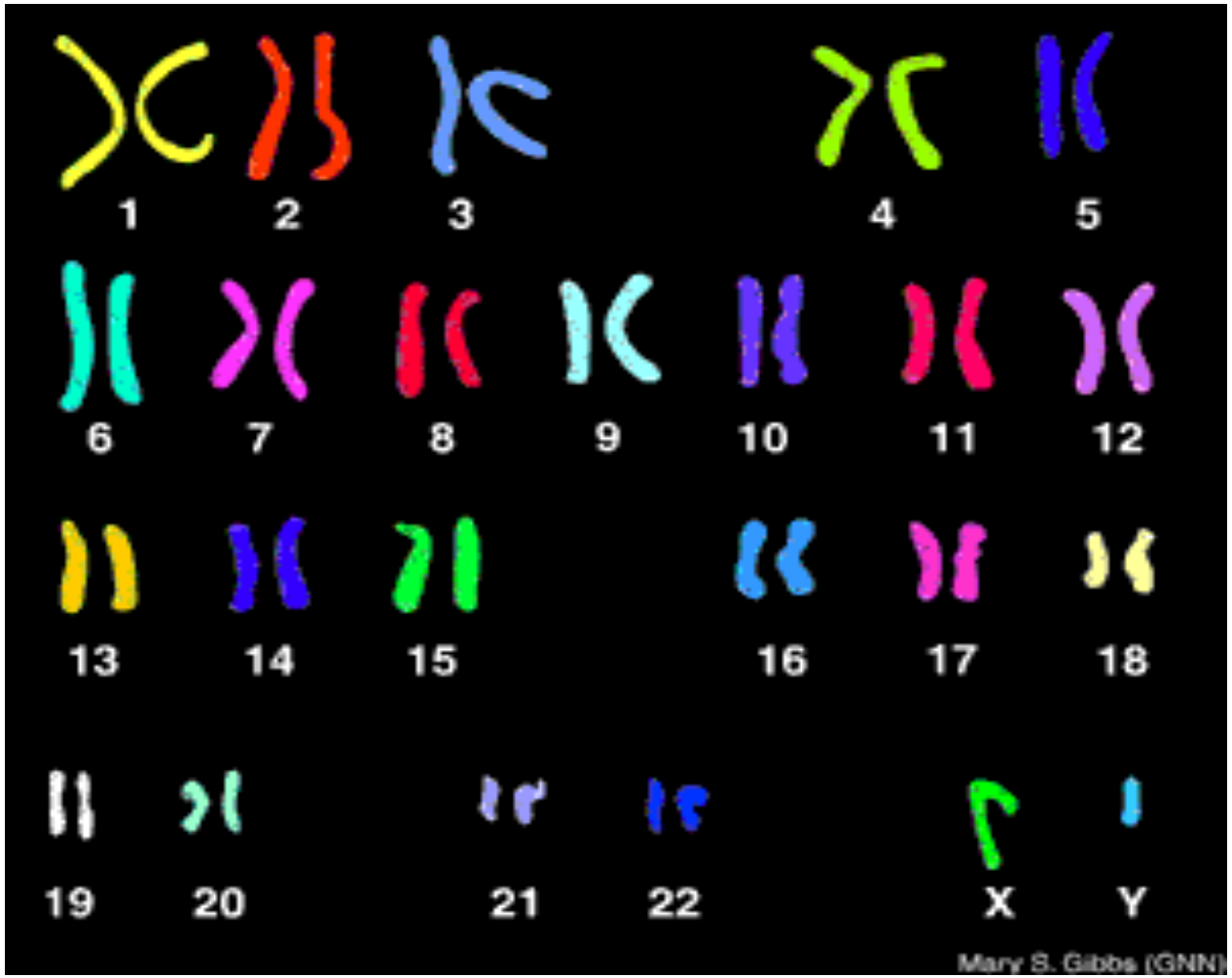
Through these proteins, our genes influence almost everything about us, including how tall we will be, how we process foods, and how we respond to infections and medicines.

Alleles = ½ Gene

- When sex cells form, only one form of each gene (allele) is present.
- For example, you have inherited the eye colour gene from your parents, this could consist of these two parts;
  - a brown eye allele from your mother and
  - a blue eye allele from your father.

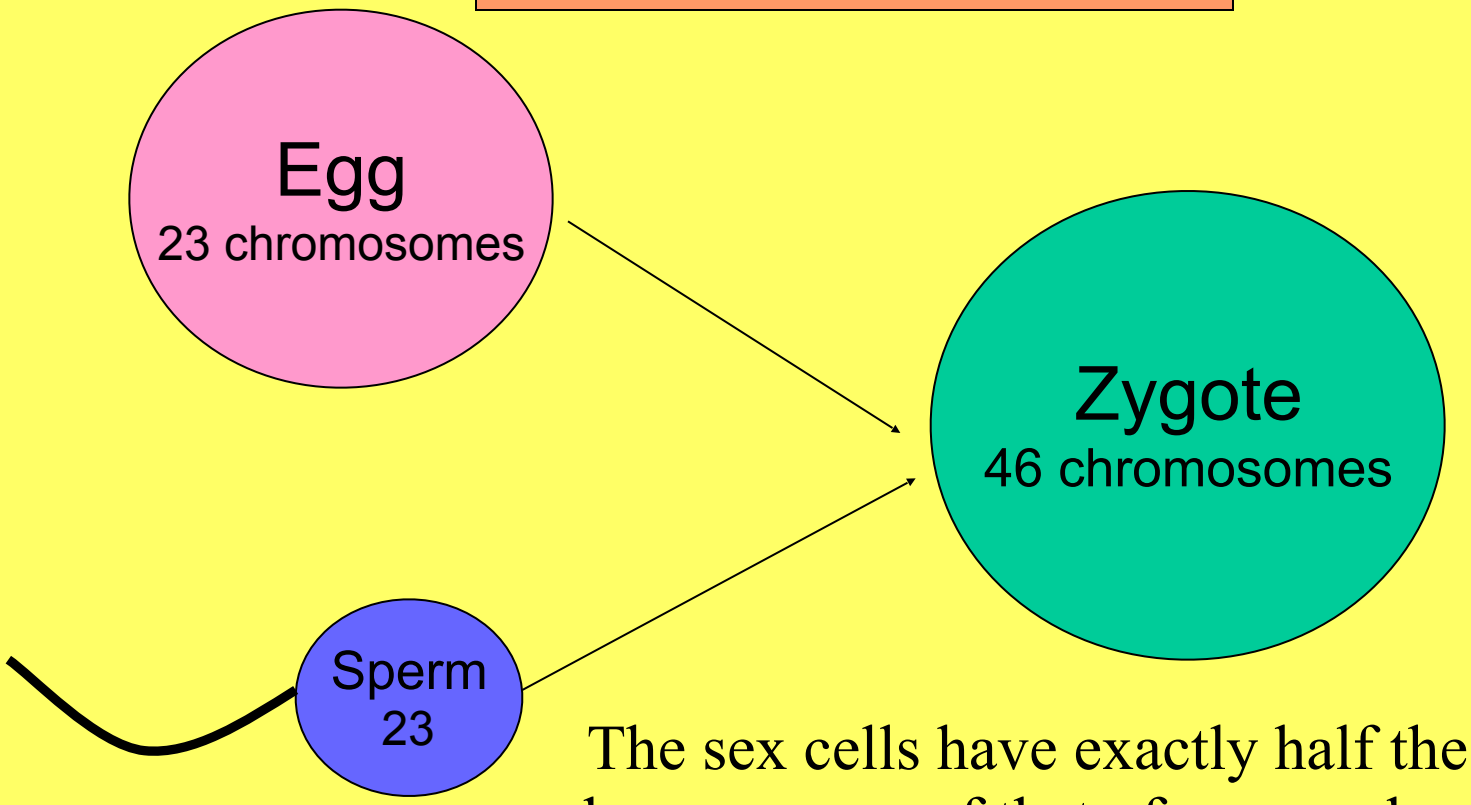
  (each of your parents gametes (sex cells) will have only one of these alleles.)
- When these sex cells combine together they form genes.
- You can only pass on one of your alleles to your off spring (child).

There are 23 pairs of chromosomes in a human in total 46 chromosomes



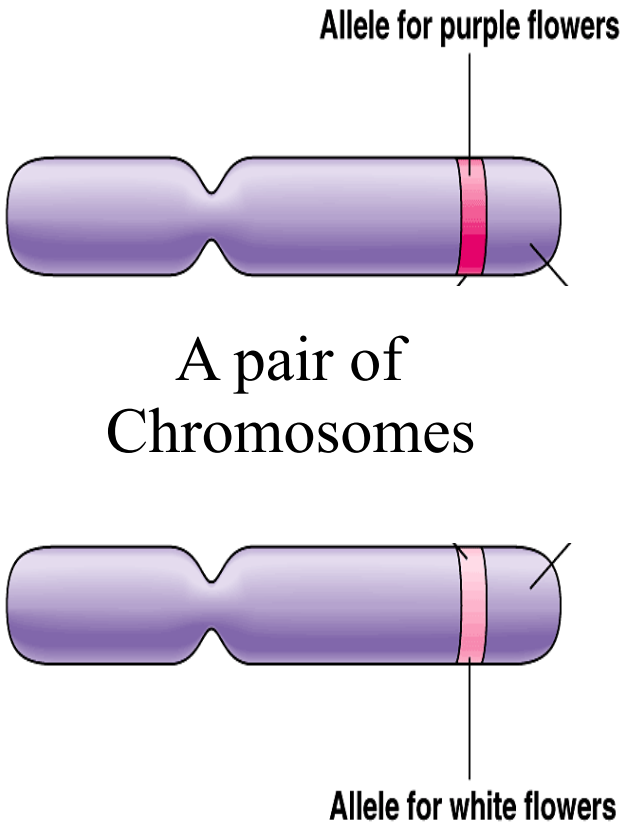
- Each ova (egg) or sperm contain only one of each pair. Therefore they contain only 23 chromosomes.
- Each pair contains one allele on it. The pair of alleles makes a gene.

Fertilisation



The sex cells have exactly half the number of chromosomes of that of a normal cell. There are 23 chromosomes in the human sex cells. When they combine during fertilisation you end up with 46 chromosomes (23 pairs).

Gene and Alleles (½Gene)



A pair of Chromosomes

Allele is an alternative form of a gene

The gene is flower colour

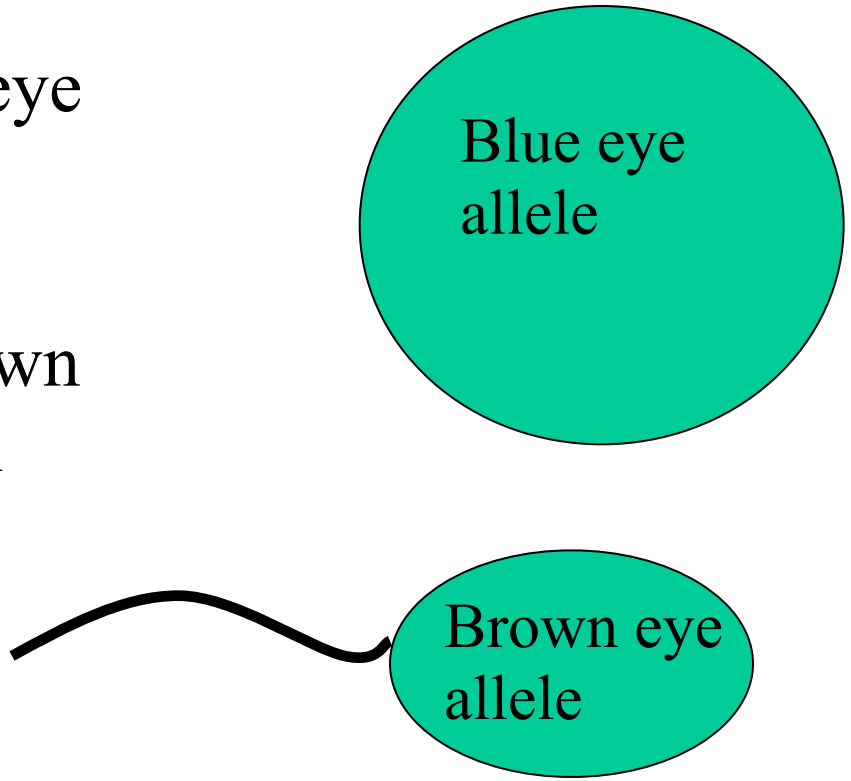
The two alleles are

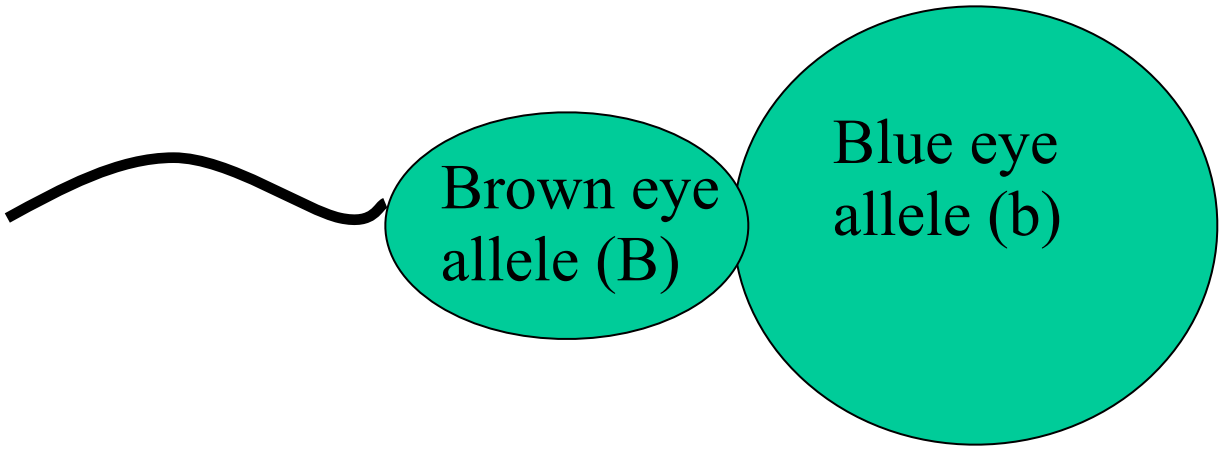
purple or white

You inherit one allele from each parent which combine together to make a gene

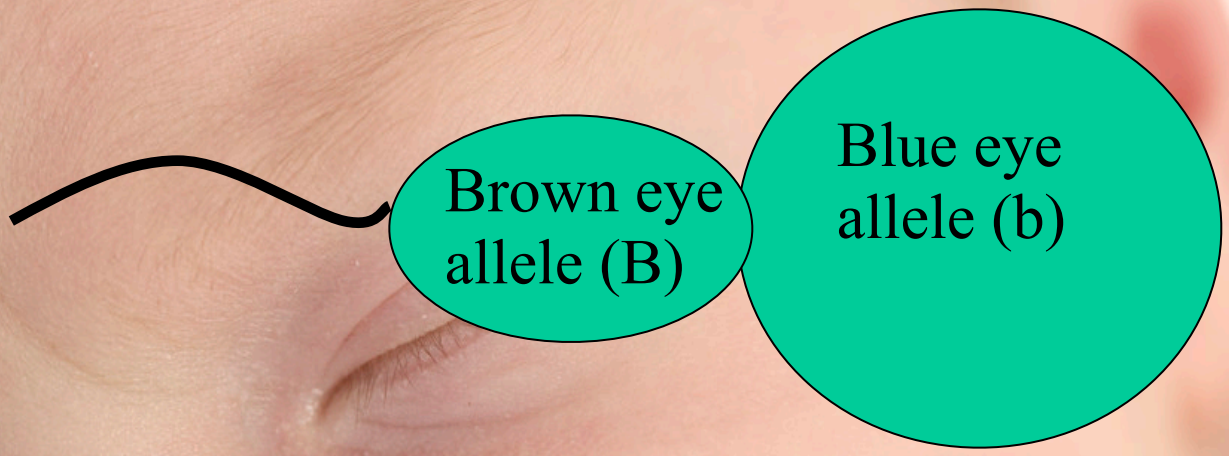
You inherit one half of each gene from each parent, these halves are called alleles.

- You may get a blue eye allele (b) from your mother in the egg.
- You may get the brown eyed Allele (B) from your father.





- When these two gametes (sex cells) combine during the process of fertilization a zygote is formed. A new life is formed.
- The two alleles B= brown eyes and b=blue eyes join.
- These two alleles form the gene for eye colour for this new individual.



- What eye colour will this new individual have???????
- Brown why?
- Because brown is the Dominant allele.
- Therefore you only need one dominant allele for it to be expressed .
- When we express a dominant allele we use a capital letter Eg. B
- When we express a recessive allele we use a lower case letter Eg. b

Monohybrid cross

The study of single-gene inheritance is done through monohybrid crosses.

- Capital letters represent dominant alleles eg.B
- Lower case letters represent recessive alleles eg. b
- (an allele is an alternative form of a gene)

e.g. coat colour in guinea pigs

Genotype	Phenotype
BB	Black
Bb	Black
bb	white

Homozygous = 2 of the same alleles eg BB or bb

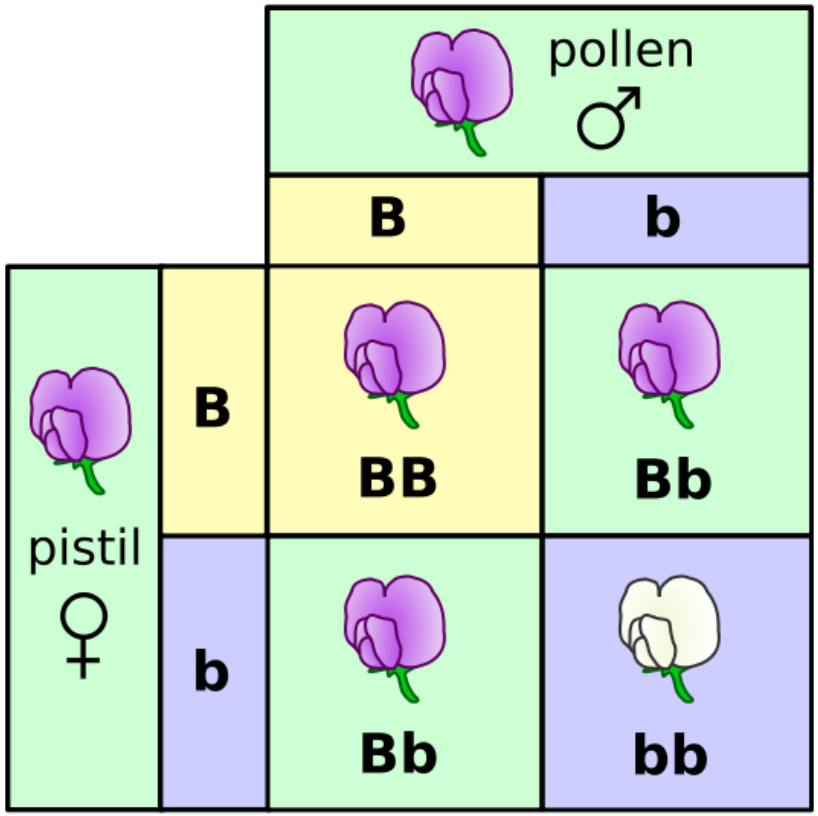
Heterozygous =2 different alleles eg Bb

Dominance

If you have an **allele** for two different traits,
Eg B brown eyes and b for blue eyes your phenotype will be of the **dominant** allele. Brown eyes

That is ALL dominant means.
Dominant genes are **not** always better
Dominant genes are **not** always more common
Dominant does not mean stronger.
It just means you need only one allele to have that trait.

To determine the outcomes of crossing individuals you use a Punnett Square



How to draw a punnett Square

A cross between 2 heterozygous black guinea pigs (Bb x Bb).

Possible fertilisations	BB	b
BB		B b
b	B b	b b

Place parents alleles at the top and side of the punnett square

B is the dominant allele

b is the recessive allele

Draw the Punnettt Square

Next put each allele from each parent in the corresponding box

Do the same with the rest of the alleles

Determining offspring

You can use a ratio or a percentage%

Possible fertilisations	B	b
B	BB	Bb
b	Bb	bb

Black

White

Percentage

75%Black and 25%White

Ratio

3:1 Black to white

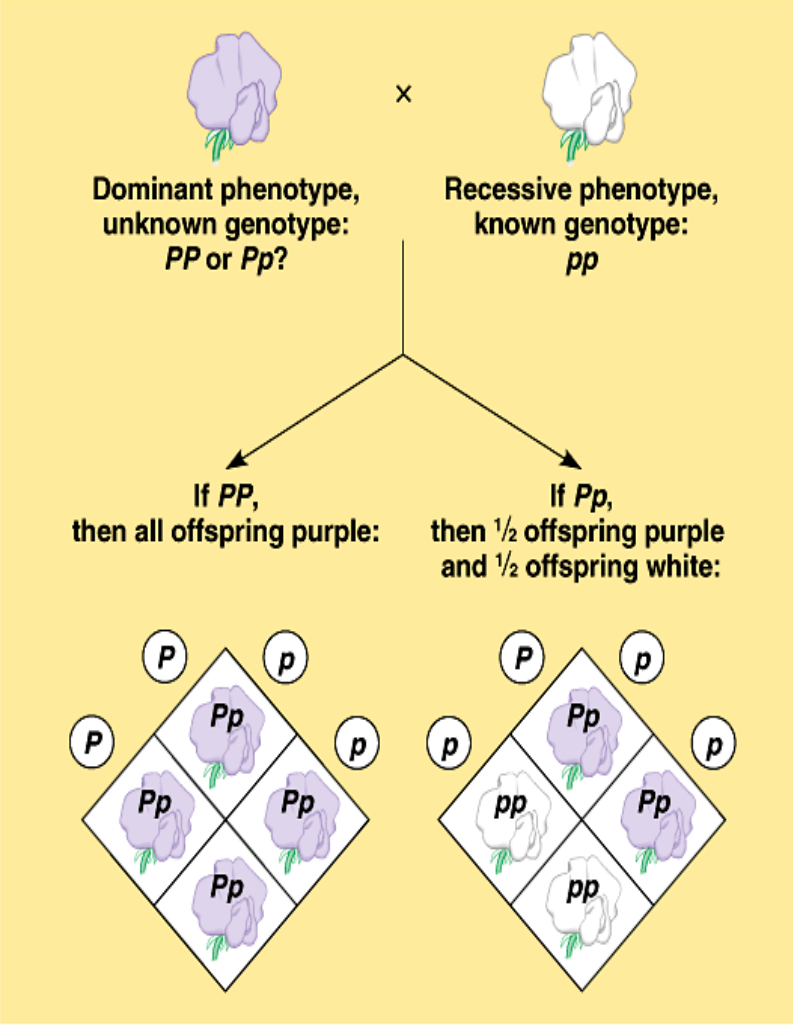
Punnet Square Practice

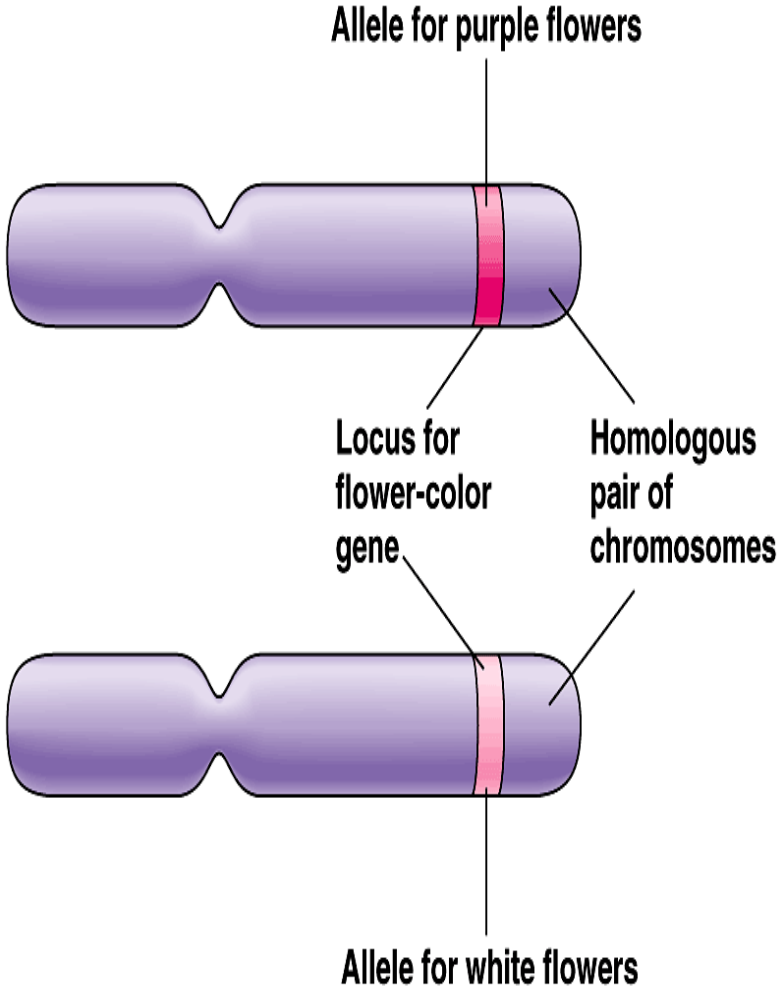
1. When a grey rabbit mates with a black domestic rabbit the off spring is often grey. If G for grey coat is the dominant allele and g is for a black coat is the recessive allele. Do the following crosses using the punnet square and predict the expected ratio of grey and black offspring.

A. GG with gg.
B. Gg with gg
C. Gg with Gg

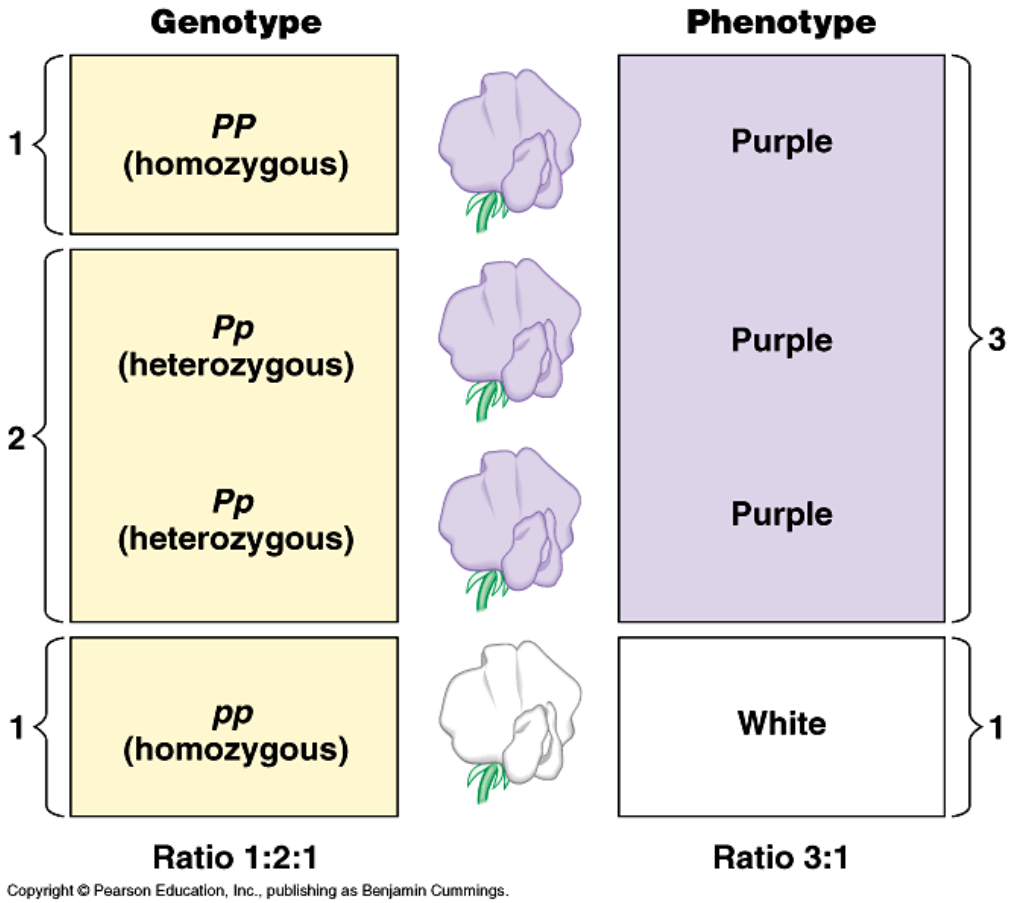
2. A wild grey rabbit crossed with domestic black rabbit. It had 12 offspring. Out of these 12 offspring 6 were black.

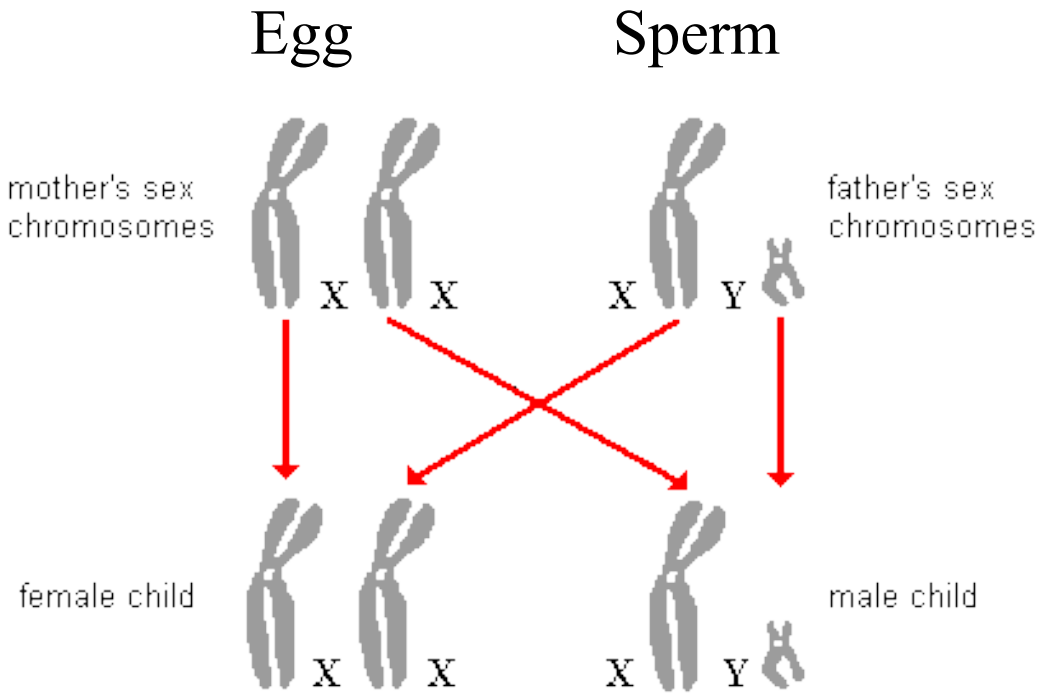
Explain this by describing the genotype of the wild and domestic rabbits genotypes. And do a punnet square to prove your answer.





Genotype and phenotype





You receive one sex chromosome from your mother and one from your father

Sex determination

What are the chances of having a boy or a girl???

Everyone has a pair of chromosomes that determine our sex

XX = female XY = male

	X	X
X	XX	XX
Y	XY	XY

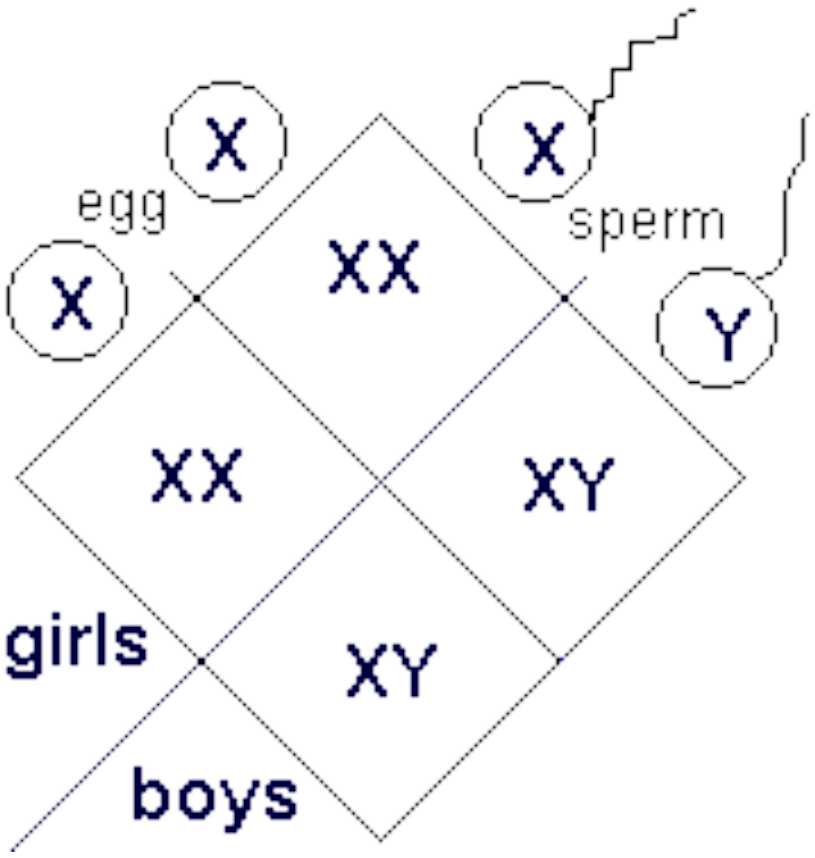
The punnett square on the left shows us the probabilities that a man and woman are faced with each time they have a child.

XX = 50%

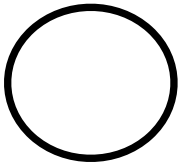
XY = 50%

If you have had two boys it does not mean you will have a girl next, each time a new zygote is formed the chances of it being male or female are 50 : 50.

- Notice how you can only receive an X from your mother and either an X or Y from your father!
- Therefore it is the father that determines the sex of the baby.
- If you receive an X from your father you will become a female and if you receive a Y from your father you will become a male.



How to determine the percentage of each gender using a punnett square



	X	Y
X	X X	X Y
X	X X	X Y

Therefore the percentage is 50% Female

And 50% males


THE WAR BETWEEN THE SEX CHROMOSOMES
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THE PROBLEM WITH YOU IS... YOU DON'T HAVE TWO LEGS TO STAND ON!
WHY WOULD I WANT TO STAND?
MICHAEL FRY
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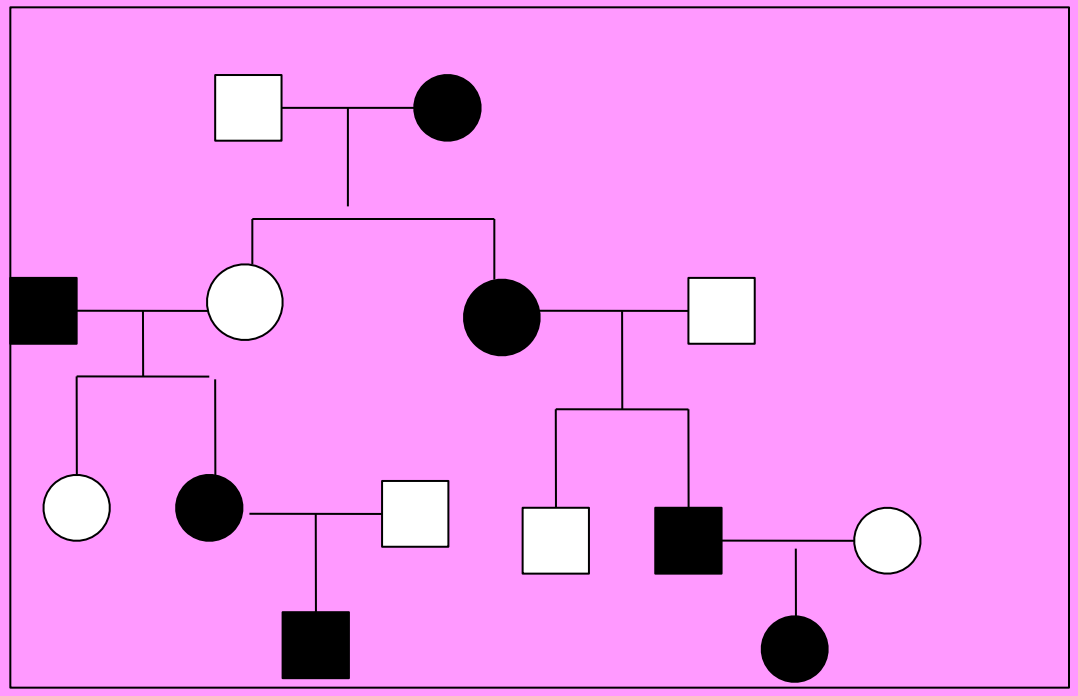
Pedigree Charts



= affected female

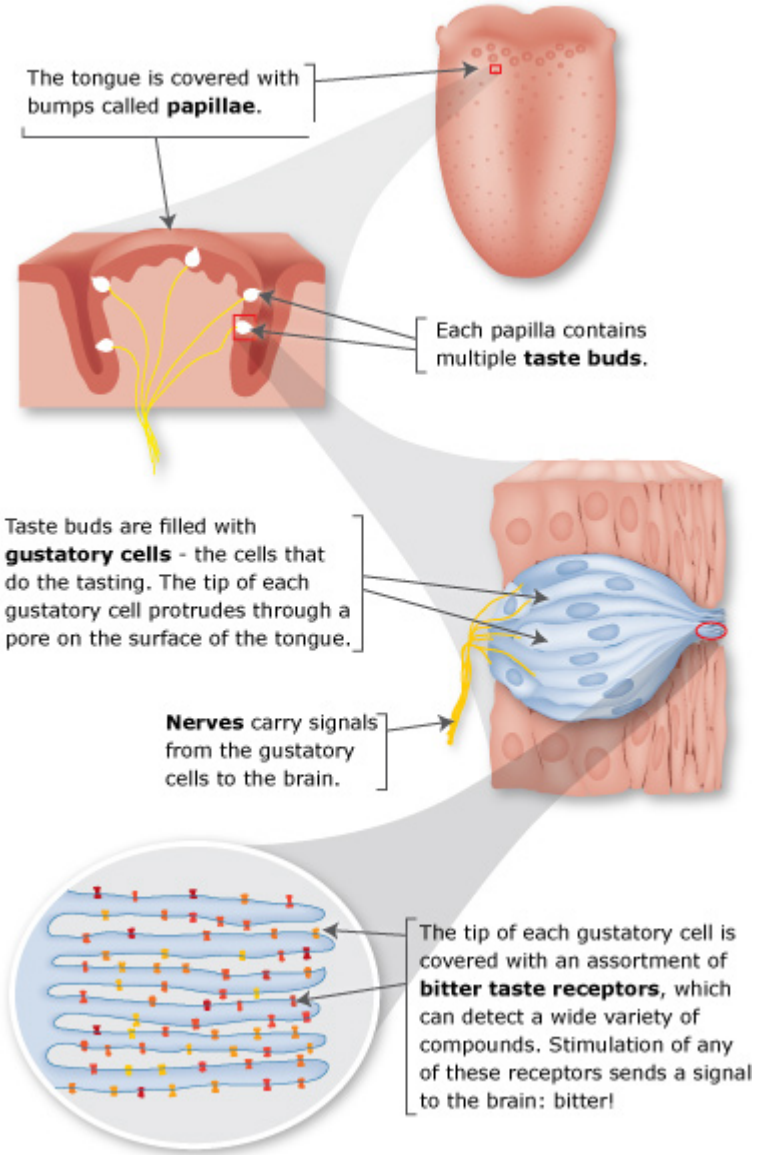
= unaffected female

= affected male

= unaffected male

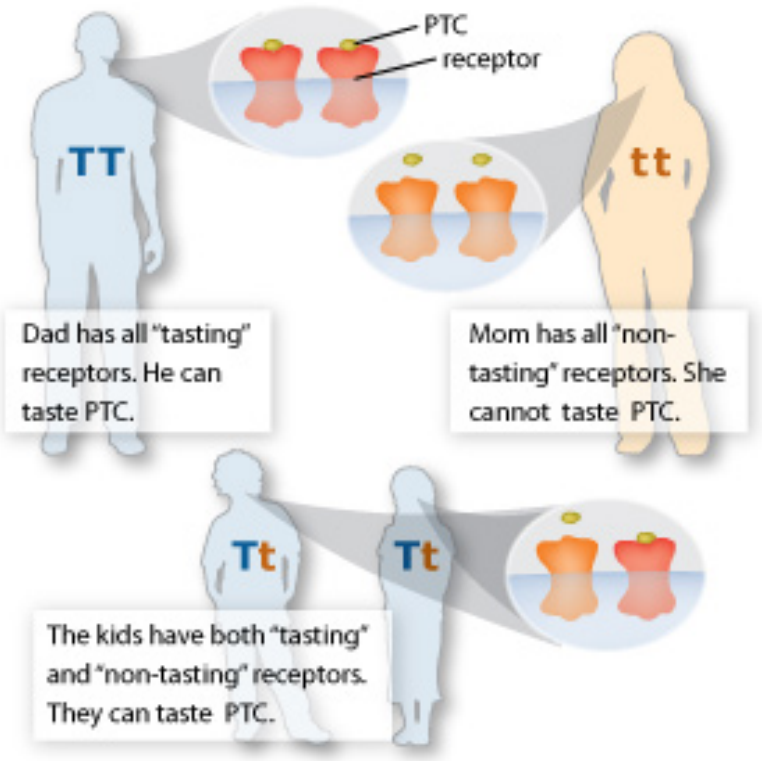
Genetics of taste

- In 1931, a chemist named Arthur Fox was pouring some powdered PTC into a bottle. When some of the powder accidentally blew into the air, a colleague standing nearby complained that the dust tasted bitter. Fox tasted nothing at all. Curious how they could be tasting the chemical differently, they tasted it again. The results were the same. Fox had his friends and family try the chemical then describe how it tasted. Some people tasted nothing. Some found it intensely bitter, and still others thought it tasted only slightly bitter.



How does this gene assist survival.

Plants produce a variety of toxic compounds in order to protect themselves from being eaten. The ability to discern bitter tastes evolved as a mechanism to prevent early humans from eating poisonous plants. Humans have about 30 genes that code for bitter taste receptors. Each receptor can interact with several compounds, allowing people to taste a wide variety of bitter substances.



= Taster female

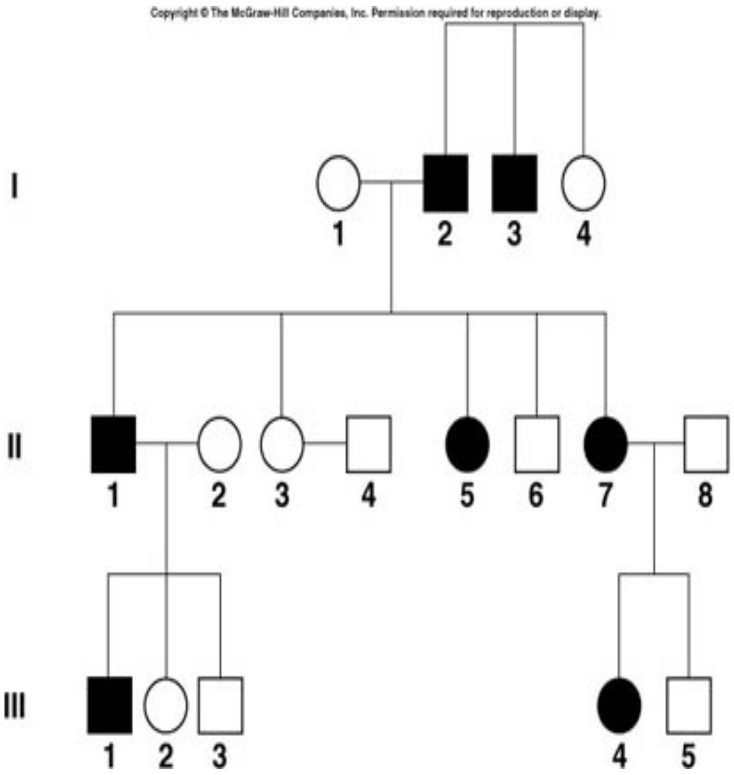
= Non taster female

= Taster male

= Non taster male

T = Taster

t= non Taster



- To find out if a taster individual is homozygous dominant TT or heterozygous Tt you will need to do a <u>test cross</u>.
- In a test cross you cross the unknown individual with a homozygous recessive non taster individual tt and look at the offspring.
- If an offspring shows the recessive trait the unknown must be Heterozygous Tt.
- If none of the offspring show the recessive gene the unknown must be homozygous dominant TT

Use the following information to answer the next two question.

= Red flower

= White flower

Red = R

White = r

11. The genotype of individual **II-4** is

A.rr

B.Rr

C.Rr or RR

D.RR or rr

12. An individual on the pedigree who has a homozygous genotype is individual

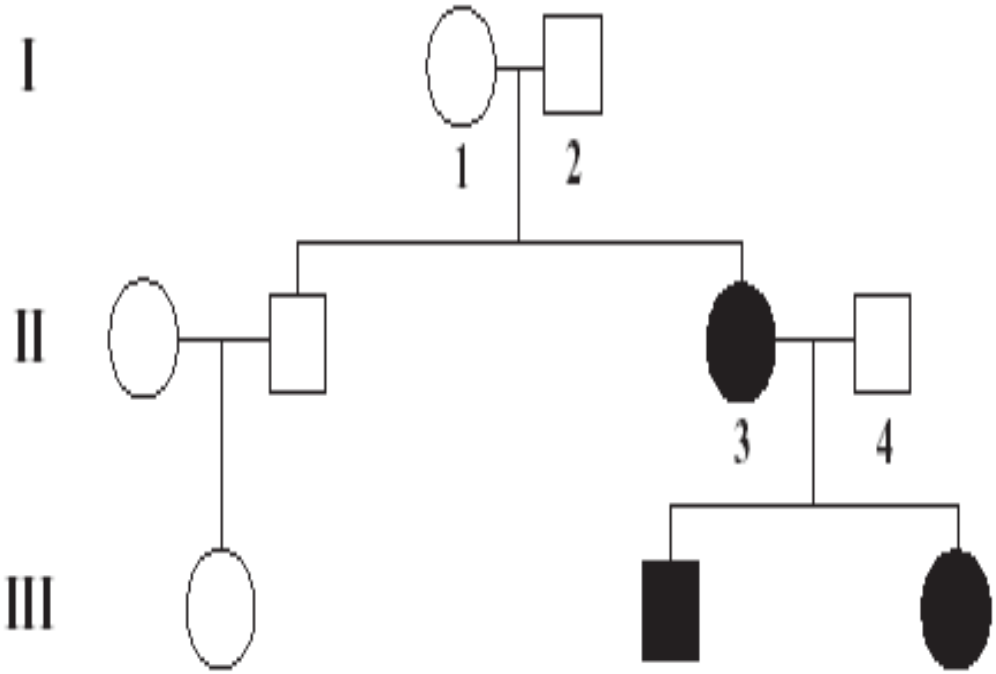
A.I-1

B.I-2

C.II-3

D.II-4

Family Pedigree



Case Study- Pedigree Charts

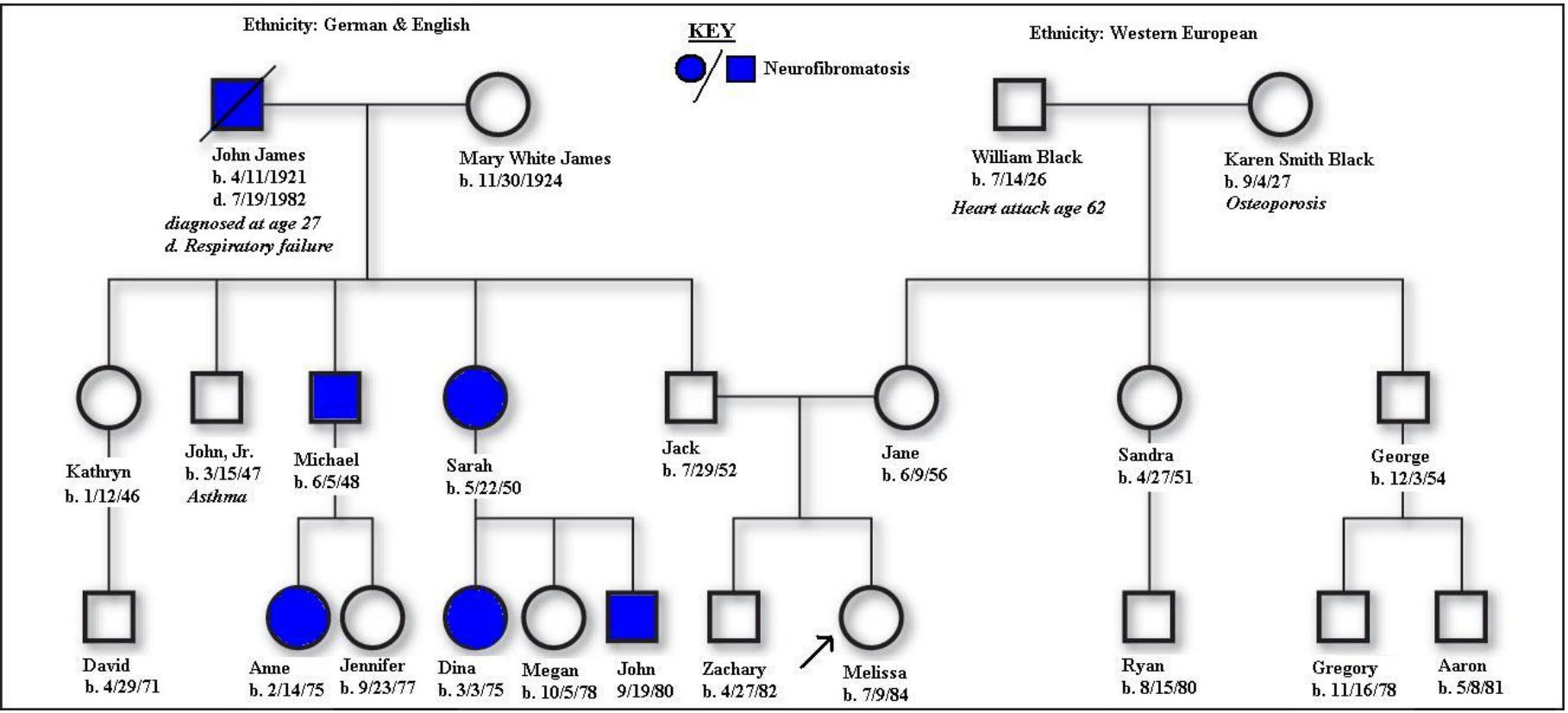
- **Neurofibromatosis** is a genetically inherited disorder in which the nerve tissue grows tumours that may be benign or may cause serious damage by compressing nerves and other tissues.

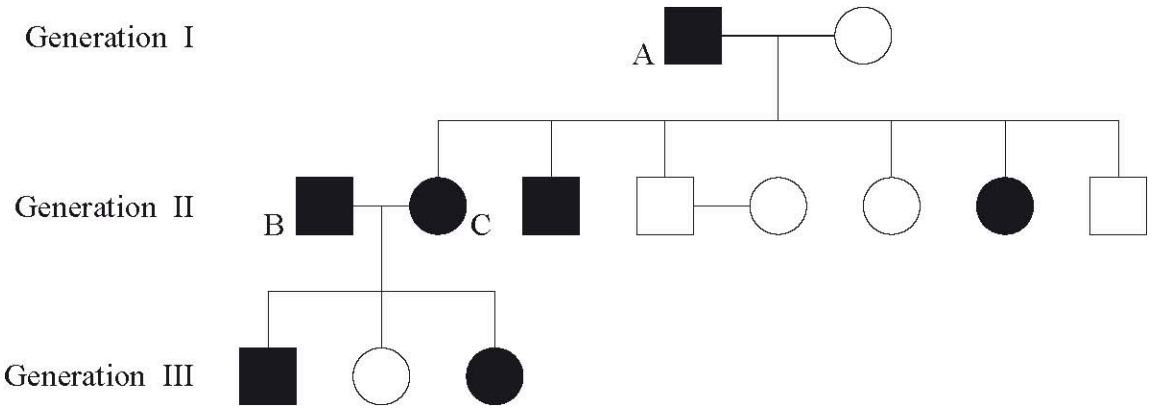
To yucky to show a photo!

A bit of information about the genetics.

- Neurofibromatosis is an autosomal dominant disorder, which means only one copy of the affected gene is needed for the disorder to develop.
- Therefore, if only one parent has neurofibromatosis, his or her children have a 50% chance of developing the condition as well.
- The severity in affected individuals can vary, this may be due to variable expressivity.
- Approximately half of cases are due to *de novo* mutations and no other affected family members are seen. It affects males and females equally.

An example of a pedigree chart.





Key: Black = at least 1 dominant allele (normal or carrier)
White = no dominant alleles (CF sufferer)
Squares = Males
Circles = Females

Cystic Fibrosis (CF) is a disease that affects the lungs and the digestive system. CF is controlled by the CFTR gene. Everyone has two copies of the CFTR gene; one inherited from each parent. A person with two recessive forms of the CFTR gene is affected by CF. A person with only one recessive form of the gene is unaffected, but is a carrier.

The pedigree diagram above, shows three generations of a family with CF sufferers. Use the information in the diagram to answer the following questions:

(a) Male A has six generation II children. What fraction of his **male** children have CF?

(b) **Explain** the genotype of female C.

- **Discuss** the phenotype of female C, and the phenotype of any children she has with:
  (i) male B; or
  (ii) a normal male(non carrier).

Achieved	Merit	Excellence
4(a) 2/3rds or (66.66%) or 2 out of 3		
4(b) She is heterozygous. **OR** She is the carrier of the recessive allele	She is the carrier of the recessive allele She must be heterozygous because some of her offspring are recessive (CF sufferers). **OR** If she was homozygous, her offspring would carry the dominant allele (be carriers NOT sufferers).	
4(c) The phenotype of female C is normal / not affected by CF.	The phenotype of female C is normal / not affected by CF **PLUS** Explanation for the appearance of ONE set of children (from normal or carrier male) is correct	If she has children with B who must be a carrier, then 75% of them would appear normal whilst 25% could show signs of CF. 50% of her children would be carriers but we couldn't tell this from their phenotype. If she has children with a normal male, then all her offspring would appear normal, but 50% of them would be carriers. For male B instead of ratios may use information from Punnett or pedigree.

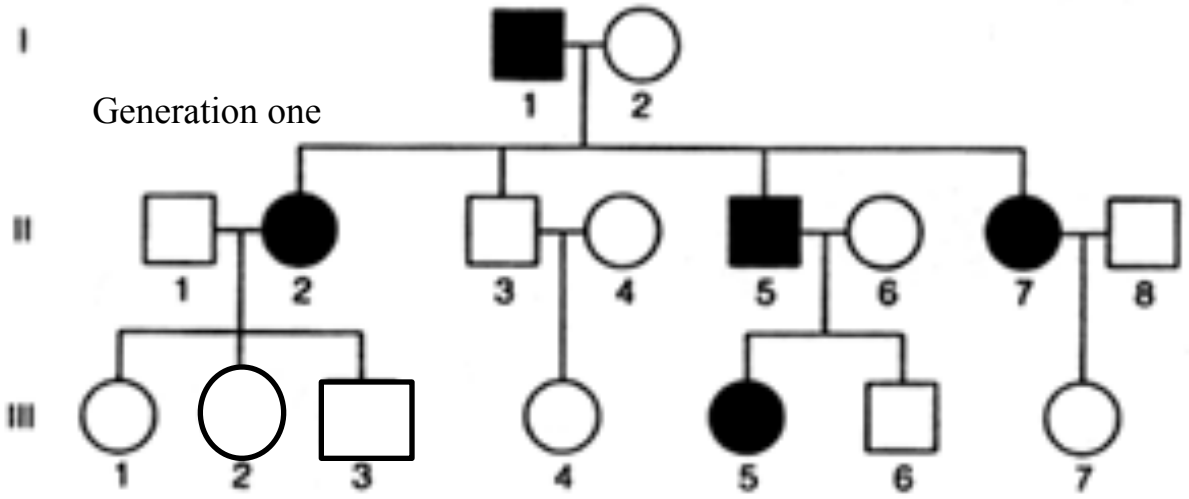









Albinism – Recessive genetic disorder.



A=Normal

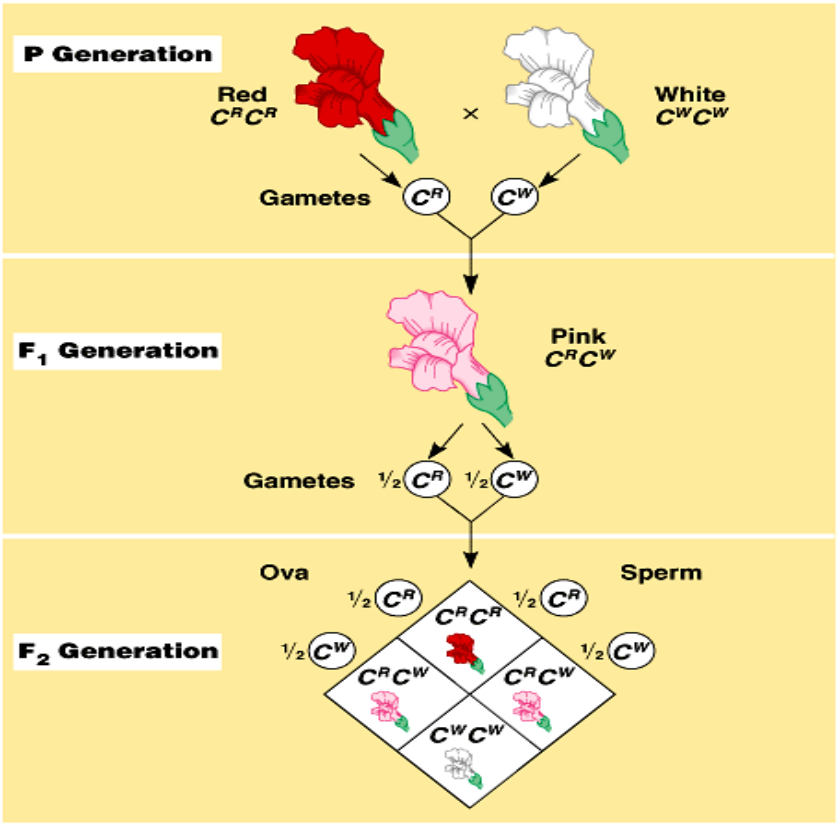
a= albino

Affected

Not affected

1. What is the genotype of individual 2 generation one?
   Explain your answer
1. What is the genotype of individual 1 generation 3. Explain your answer.

Incomplete dominance



AGTCCGCGAATACAGGCTCGGT AGTCCGCGAATACAGGCTCGGT

DNA

DNA is a double stranded molecule found in the nucleus of the cells of living things.

It is made up of about 3 billion pairs of 4 different bases, we use letters to represent the 4 bases found in all living things

A = adenine

T = thymine

C = cytosine

G = guanine