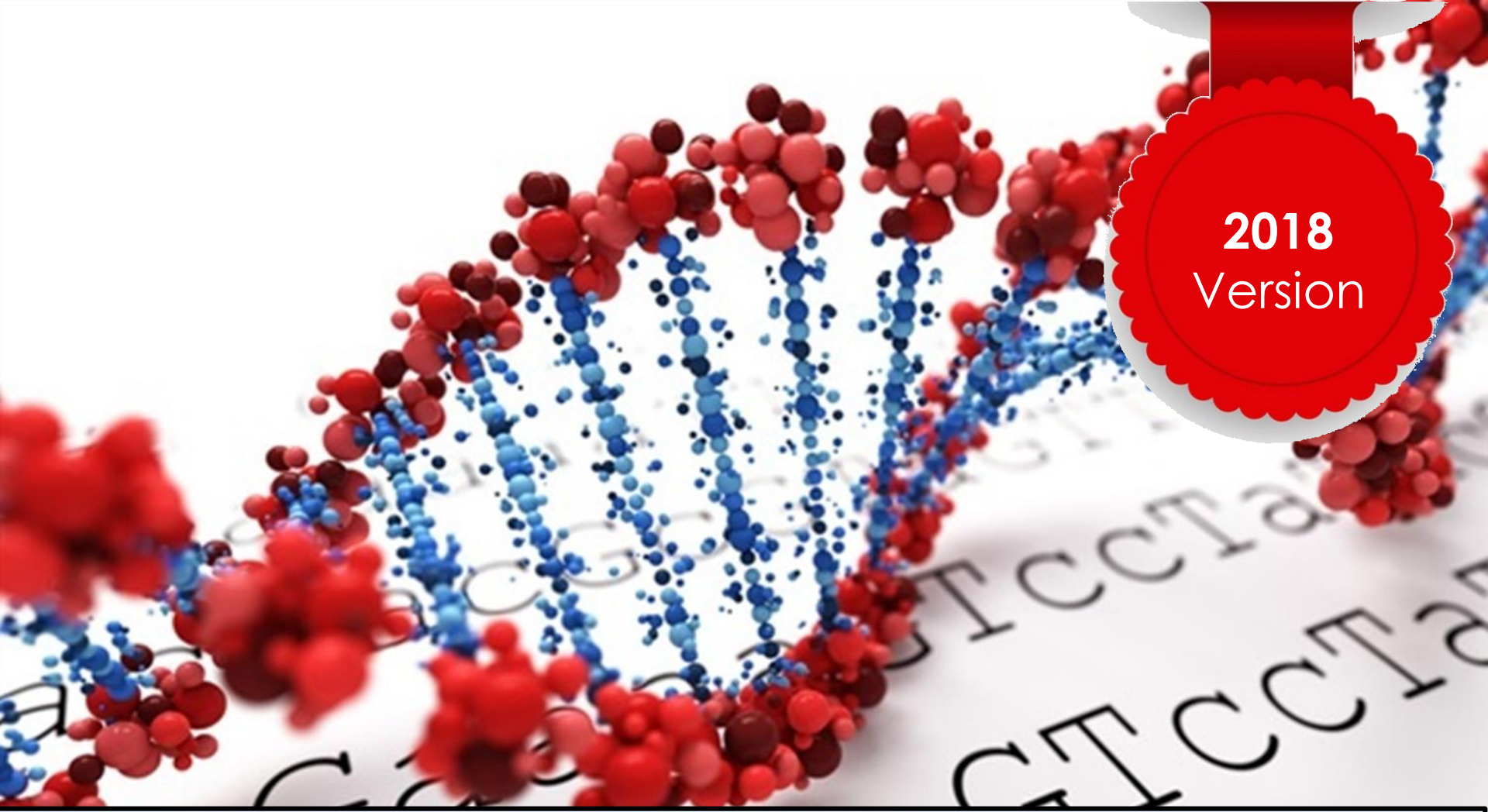


Genetic Variation

Junior Science

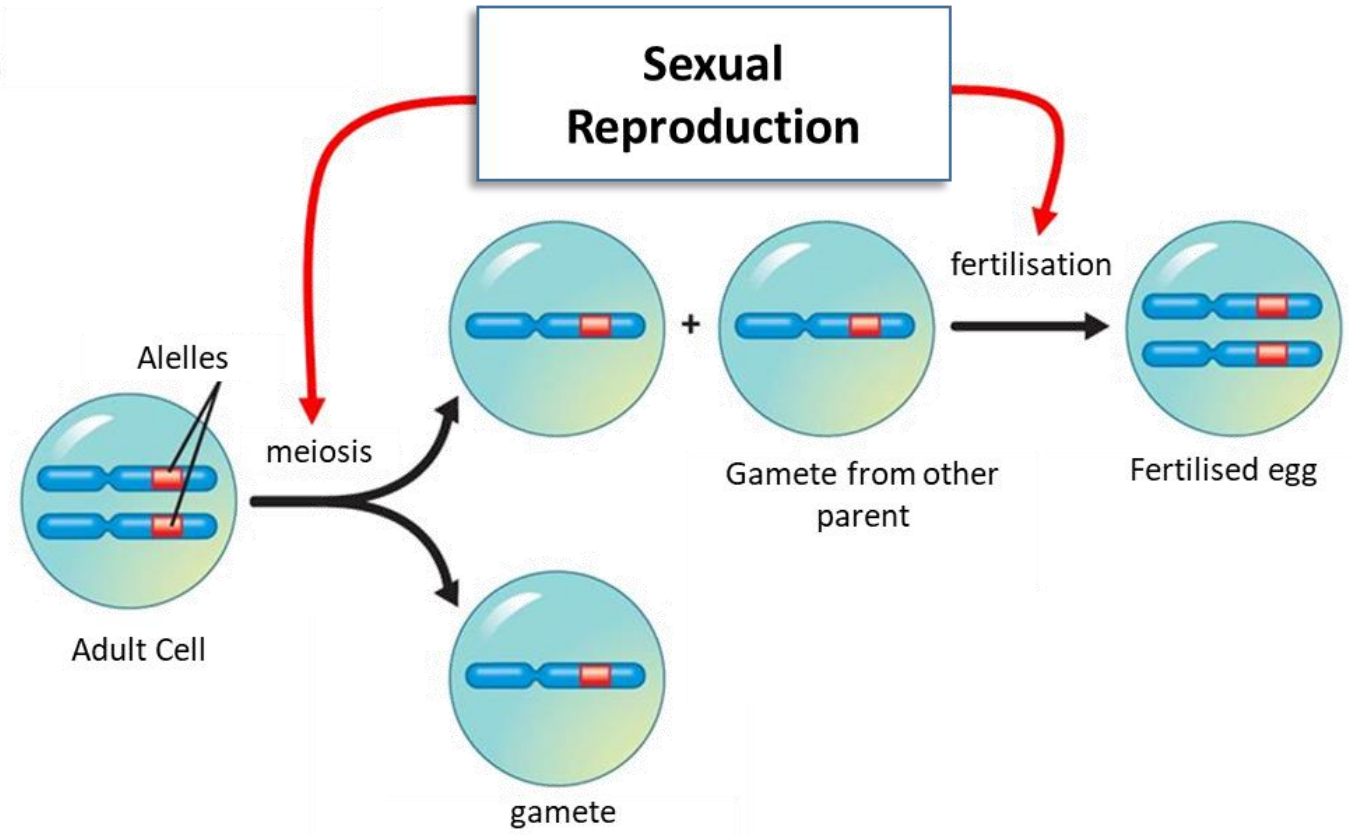
Sexual Reproduction

GZ Science Resources

Organisms of a species that reproduce sexually are not identical therefore they exhibit **variation**. Variation or differences in traits is caused by genetic factors (what genes you are born with) and environmental factors but only genetic variation can be passed onto the next generation by sexual reproduction. Genetic material (DNA) carried in the egg & sperm (gametes) provide the **inherited instructions** for making off-spring. The inheritance of this mixture of genetic material leads to variation in the offspring.

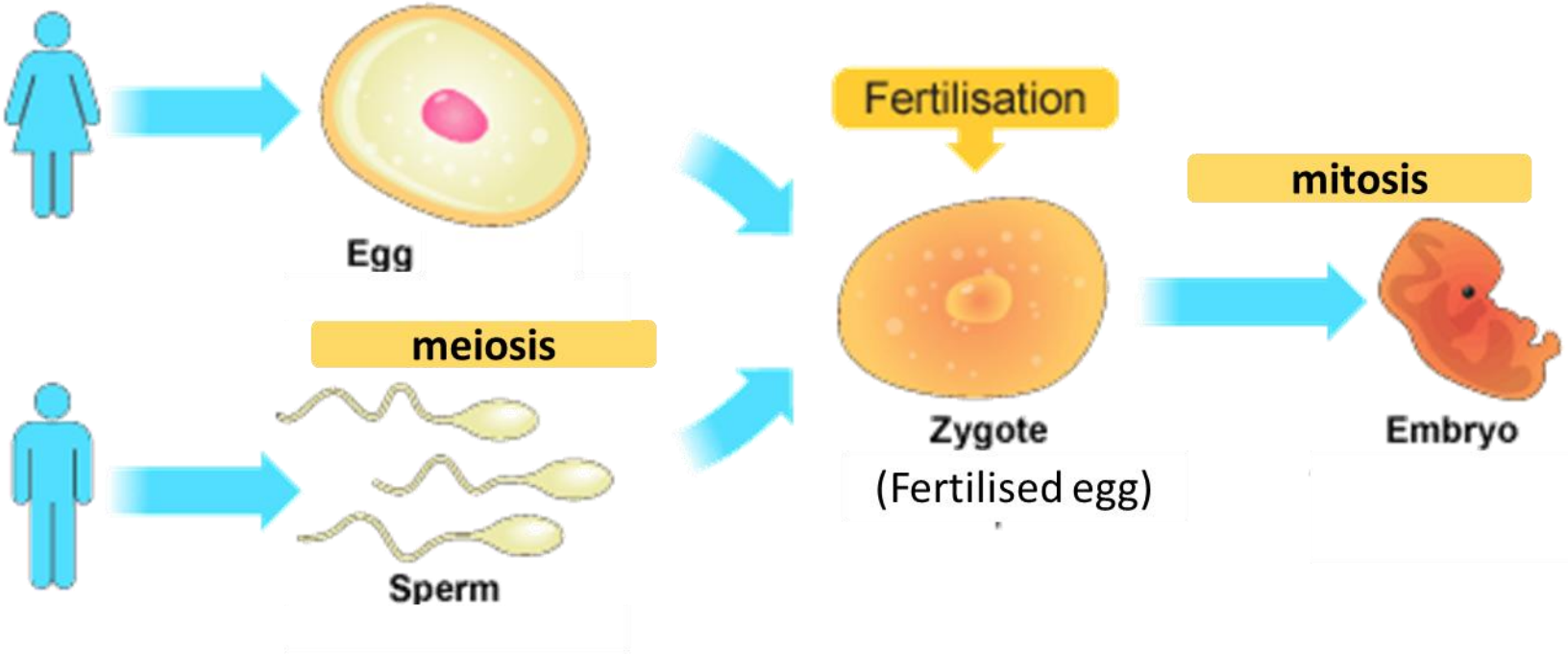
Variation is due to genes being passed on from parents to offspring during sexual reproduction

Genes are passed on from parents when the DNA in each parents **gametes** combine to form an embryo during **fertilisation** which then develops into a baby.
Variation occurs when each parent's gametes are created – sperm in males and eggs in females – through a process of **Meiosis**.



Sexual reproduction involves a mobile male gamete (e.g. sperm) fusing with a stationary female gamete (e.g. egg)

Both males and females only donate half of their chromosomes (one from each **homologous** pair) to form gametes through **meiosis**. (gametes = egg or sperm). When the chromosomes from the egg and sperm rejoin to form a **zygote** (fertilised egg) with the total number of chromosomes **fertilisation** has occurred. Once fertilisation has produced a zygote then **mitosis** occurs throughout the remainder of the babies/persons life time for growth and cell repair.

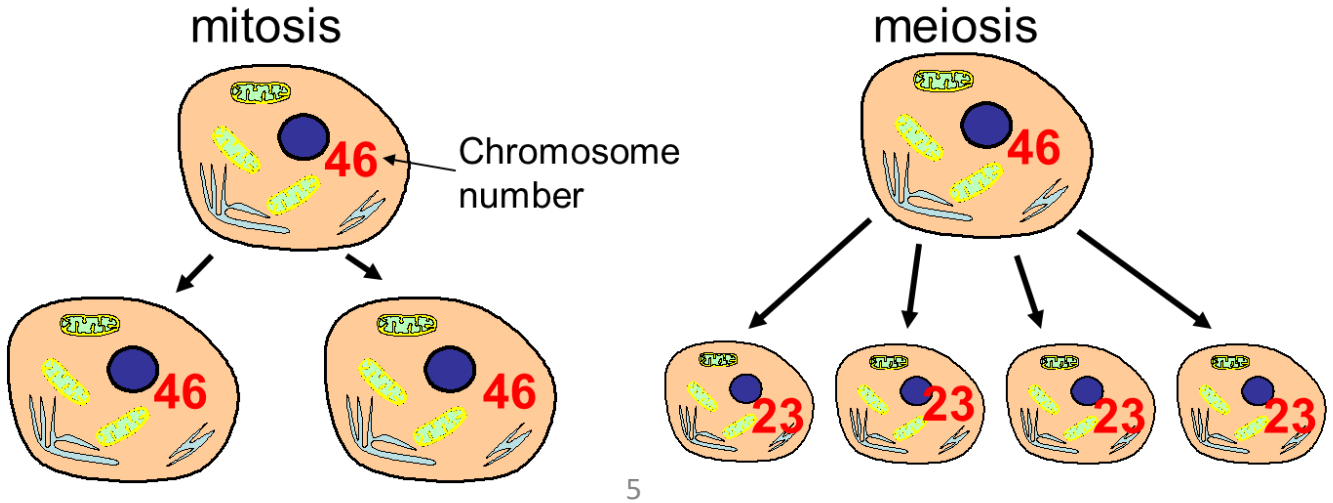


http://www.bbc.co.uk/schools/gcsebitesize/science/edexcel_pre_2011/genes/reproductionandcloningrev1.shtml

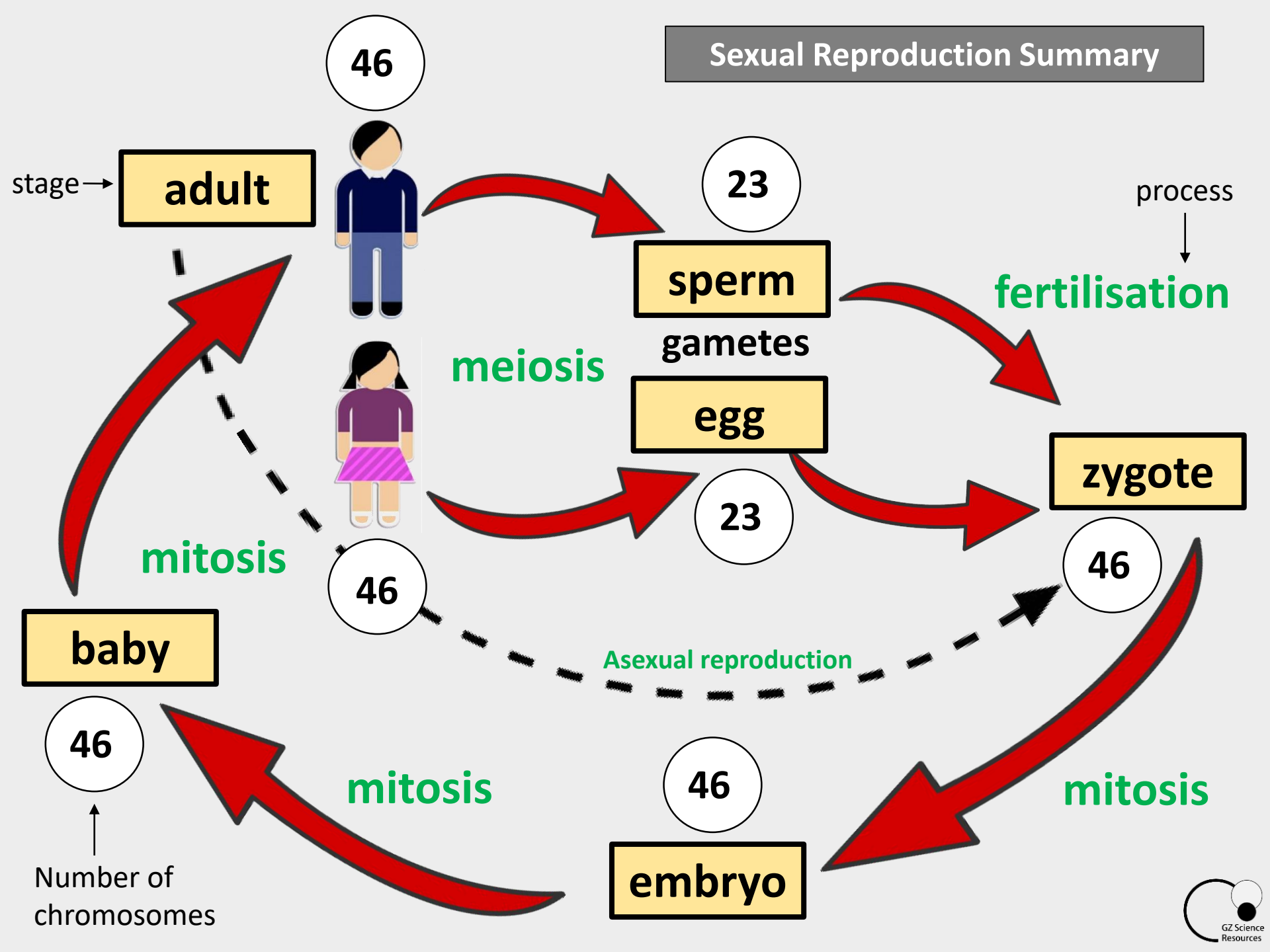
Comparing Mitosis and Meiosis

Meiosis is cell division that occurs in the testes(sperm) and ovaries(eggs) producing unique gametes.
Mitosis is cell division for growth and repair – it makes identical copies of cells to increase number of cells/allows for growth.

Mitosis creates **2 identical daughter cells** from each parent cell. Each of these cells maintains a full set of identical chromosomes (diploid). These cells are called somatic cells.
Meiosis divides one parent cell into 4 gamete cells. Each gamete has half the number of chromosome of the parent cell (haploid). A male and a female gamete recombine during fertilisation to form a cell with the complete set of chromosomes.



Sexual Reproduction Summary



Gametes contain half the normal number of chromosomes and fertilisation restores the normal number

Gametes are produced by the process of **Meiosis** – sperm in the males and eggs in the female.
Meiosis randomly sorts one chromosome from each pair of chromosomes (there are 23 pairs or 46 individual chromosomes) contained in a cell and produces a gamete cell which will contain 23 single chromosomes.
When the gametes combine during fertilisation the 23 single chromosomes from each gamete re-join to form 46 or 23 pairs once more in the embryo cell.

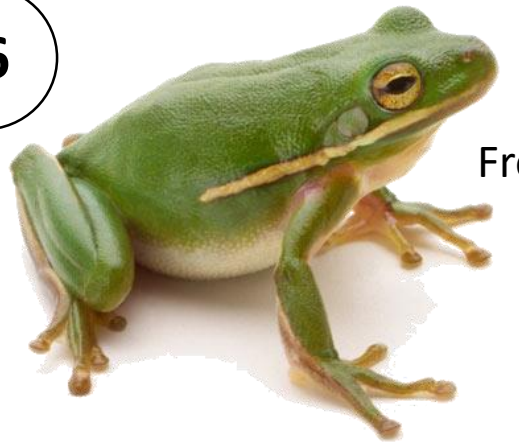
GZ Science Resources

Chromosome numbers of other species

extra info

Other species may have a different number of chromosomes in each cell compared to Humans. The number of chromosomes do not relate to the "complexity" of the organism.

26

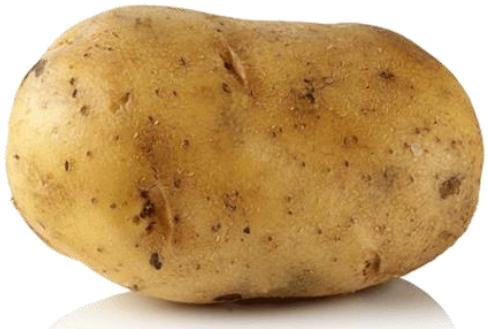


Frog

Pea

14

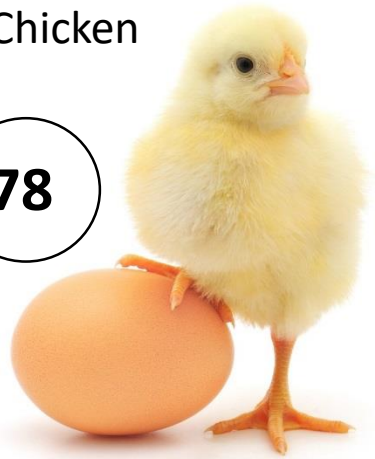
Potato



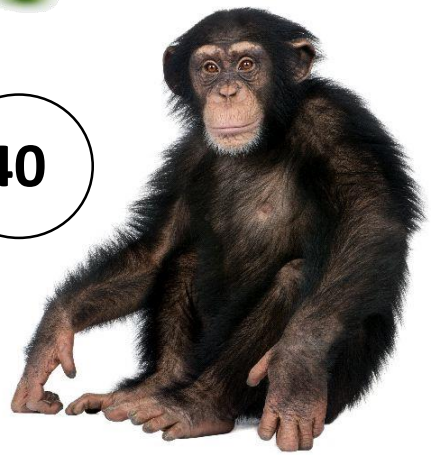
40

Chicken

78



40

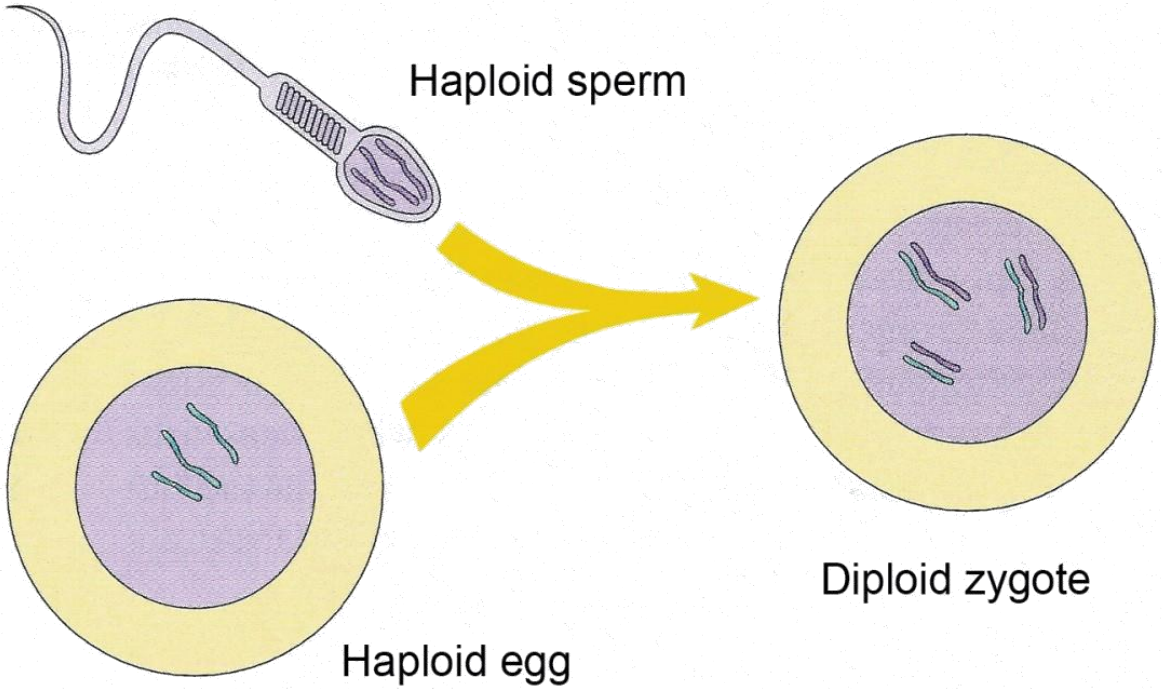


Chimpanzee

GZ Science Resources

Haploid and Diploid

Gametes contain only one set of chromosomes and are known as **haploid** (or half). All other cells in an organism contain the usual two sets of chromosomes inherited from both parents and are known as **diploid** (2 or double).

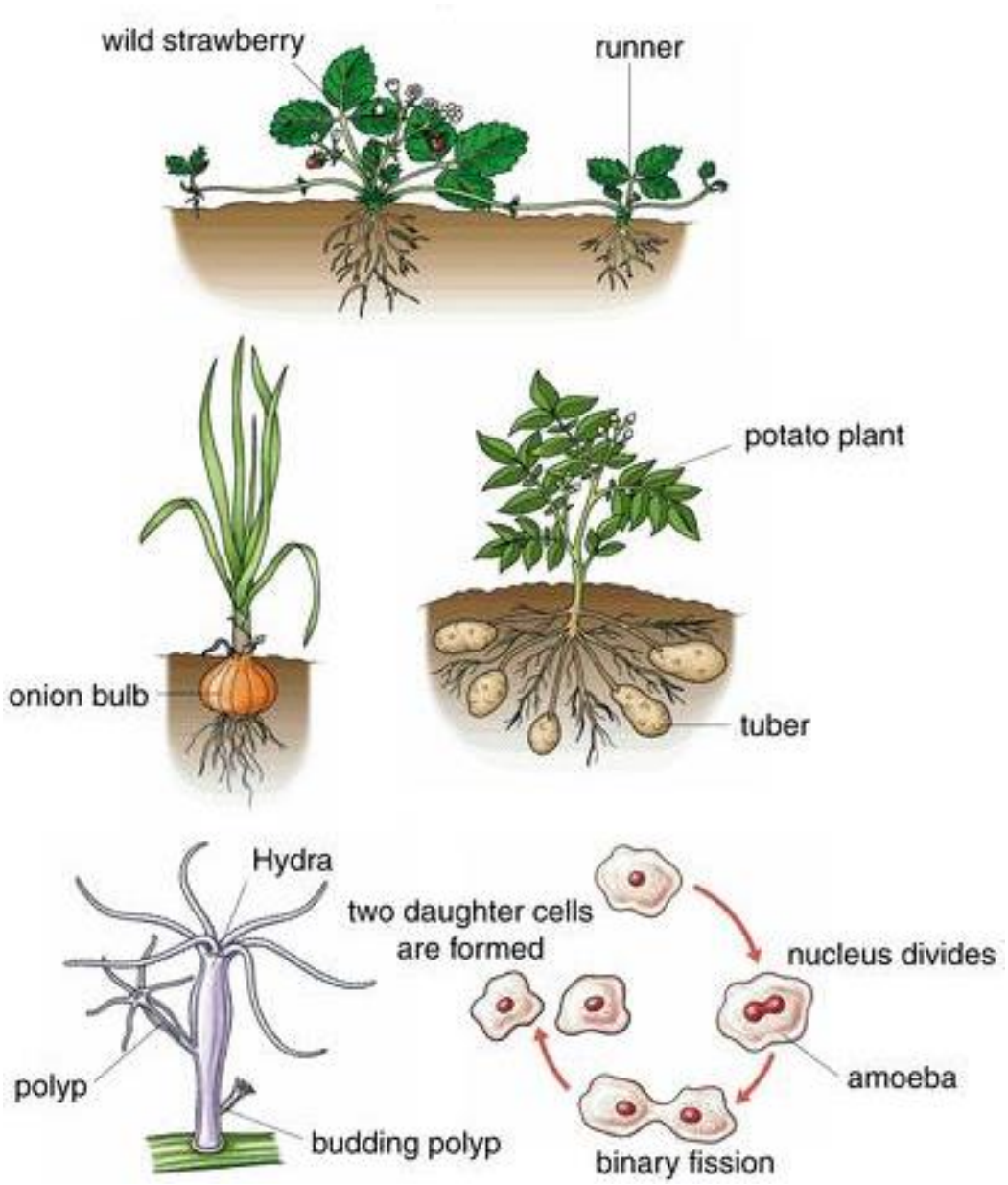


Some species such as bacteria, fungi and some plants consist entirely of haploid cells. These species undergo asexual reproduction to produce identical offspring so the chromosome number remains the same rather than doubled up like during the fertilisation of sexually reproducing species.

extra info

https://www.quora.com/Are-gametes-considered-diploids-or-haploids

Asexual reproduction produces identical offspring



Some organisms, more commonly bacteria and plants but also some animals, reproduce asexually. This type of reproduction does not involve the manufacture of sex cells (gametes) from two parents. Every new organism produced by asexual reproduction is genetically identical to the parent – a **clone**. The advantages are that there is no need to search for a mate. Asexual reproduction can therefore lead to a rapid population build-up.

The disadvantage of asexual reproduction arises from the fact that only identical individuals (clones) are produced – there is **no variation** and an asexual population cannot adapt to a changing environment and is at risk of extinction.

Asexual Reproduction vs Sexual reproduction

These kittens have been produced through the process of **sexual reproduction**. Some organisms use **asexual reproduction** to produce offspring.

Asexual reproduction occurs when only a single individual passes on all its genes to the offspring. The offspring are genetically identical to the parent. Sexual reproduction occurs when two parents create offspring and pass only half their genes to the offspring. The offspring have a different/unique set of genes.

Advantages for asexual reproduction are that only one parent is needed and identical offspring are adapted to same environment as parent. An advantage of Sexual Reproduction is that variation can increase survival chances of a species if the environment changes.

Variation

As a species, Humans all have the same set of genes. However each individual, except identical twins, has a different combination of alleles inherited from both parents and this creates **variation**. Variation of traits causes each individual to look different from another and in many cases behave differently from each other as well.

An individual within an ethnic group tends to have more similar traits in common to others within the same group.

Continuous and discontinuous variation

Variation of a trait in an individual can be **continuous** such as tallness where height can be either very tall or very short as well as any height in between. Offspring will most often show height half way between the two parents as alleles inherited from both parents have a combined effect.

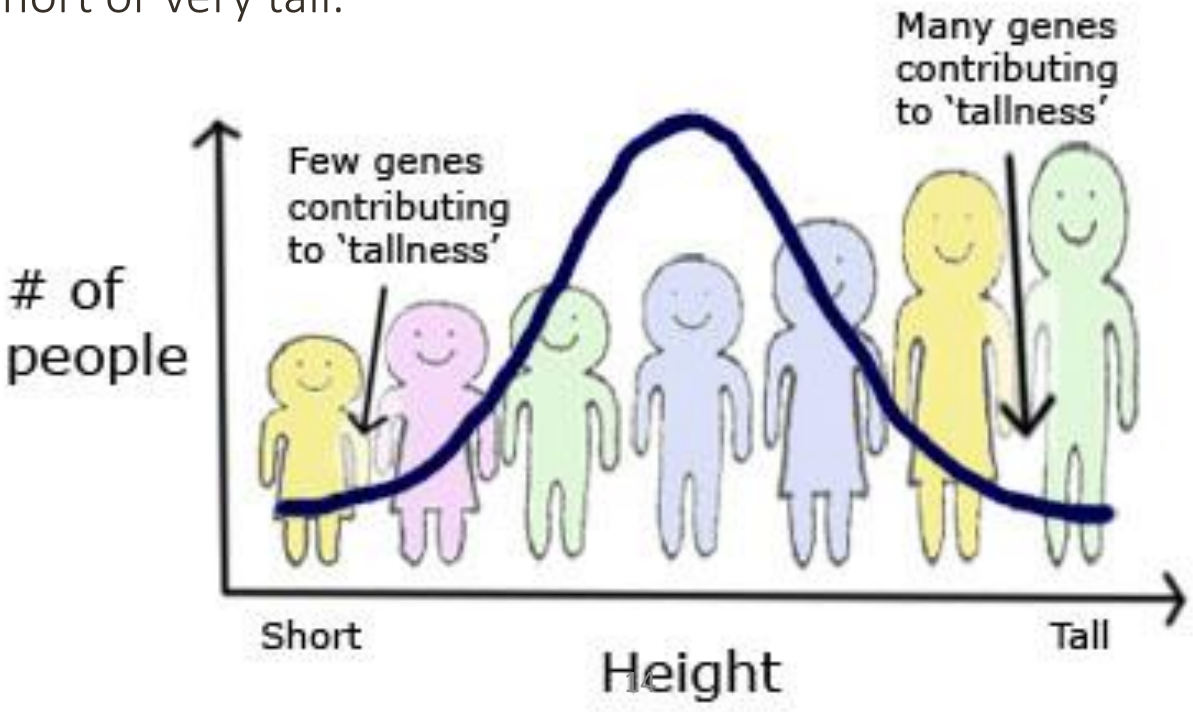
Variation of a trait can also be **discontinuous** such as the ability to roll your tongue. You can either roll it or you can't but you cannot "half roll" it. Offspring will inherit their trait from one parent or the other but not both.

https://www.sciencedaily.com/releases/2010/12/101230123552.htm

Continuous Variation

Traits that cause **continuous variation** are created by a group of genes. When a random group of people are measured for a particular trait the extremes tend to be expressed the least and the mid point tends to be expressed the most. This creates a **bell shaped curve** when graphed.

In the example below, many more people tend to be of average height compared to being very short or very tall.



Discontinuous Variation

Discontinuous Variation produces an "either/or" trait (physical characteristic).

Every person inherits one allele (a version of a gene producing the trait) from each parent. This gives the person **two alleles** for each trait (their genotype)

If a person has one or two dominant alleles then they will also have the dominant phenotype (trait). A person can only have the recessive phenotype (trait) if they have two recessive alleles.

GZ Science Resources

Examples of inherited Traits for Discontinuous Variation

Dominant phenotype (trait)	Recessive phenotype (trait)
Cleft Chin 	No Cleft 
Widow's Peak 	No Widow's Peak 
Dimples 	No Dimples 
Brown/Black Hair 	Blonde Hair 
Freckles 	No Freckles 
Brown Eyes 	Gray/Blue Eyes 
Free Earlobe 	Attached Earlobe 

Inherited and Environmental Variation

Many traits that determine our appearance have been **inherited** from our parents. Every single cell in our bodies will contain a copy of the alleles that are responsible for these inherited traits and these can be passed down to our children.

But some variation can be acquired during our lifetime from **environmental effects** such as smaller size due to lack of food while growing or loss of sight due to injury.

This variation will not be passed on to offspring.

This Lion has scars on his face due to environmental effects, and these will not be passed down to his offspring.

Environmental Variation – Hydrangea Case study

extra info

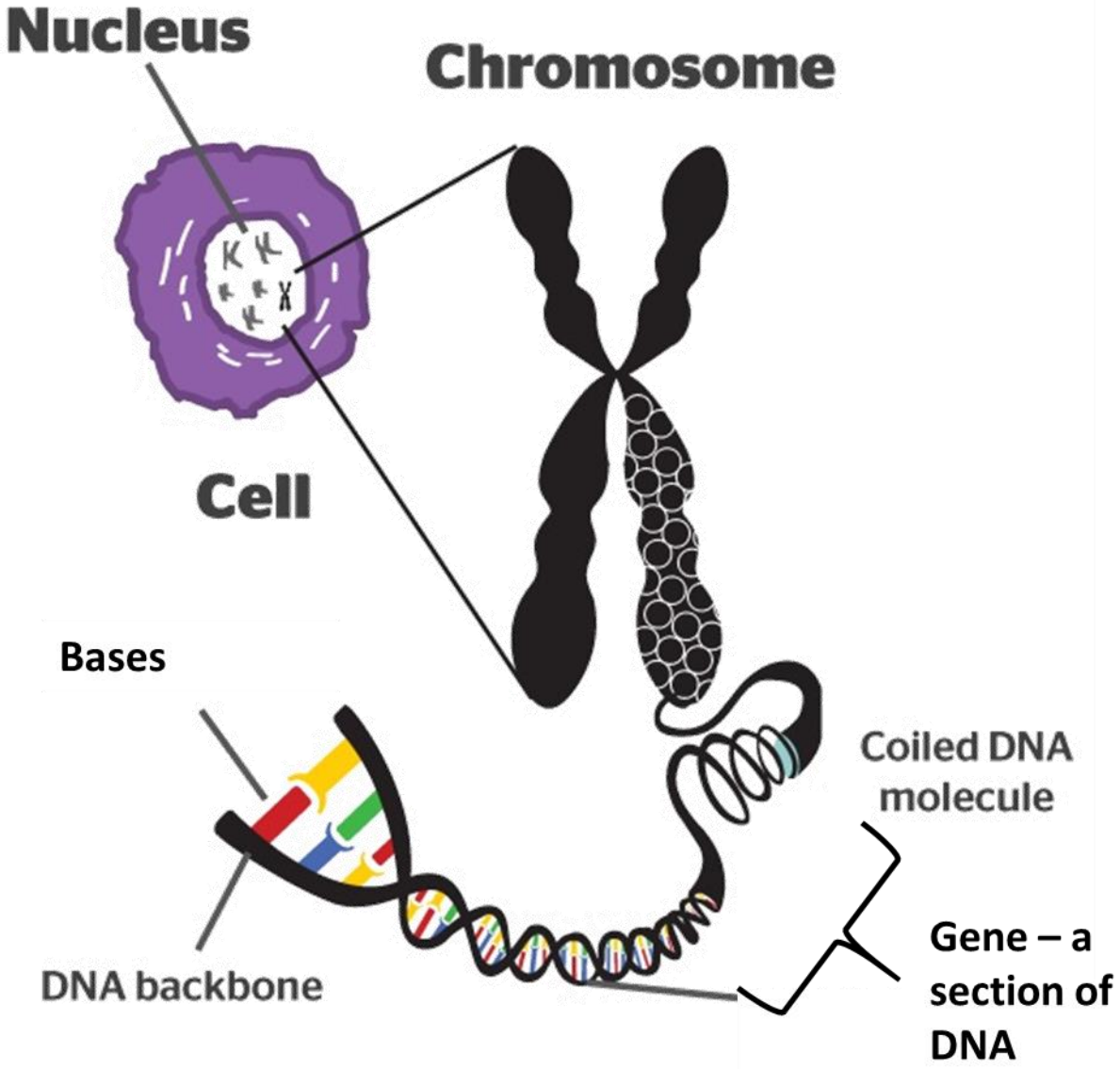
Colour variation in hydrangeas is determined by the **environment** and is due to the presence or absence of **aluminium** compounds in the flowers. If aluminium is present, the colour is blue. If it is present in small quantities, the colour is variable between pink and blue. If aluminium is absent, the flowers are pink.

Soil pH (acid or base) indirectly changes flower colour by affecting the availability of aluminium in the soil. When the soil is acidic (pH 5.5 or lower), aluminium is more available to the roots, resulting in blue flowers.

GZ Science Resources

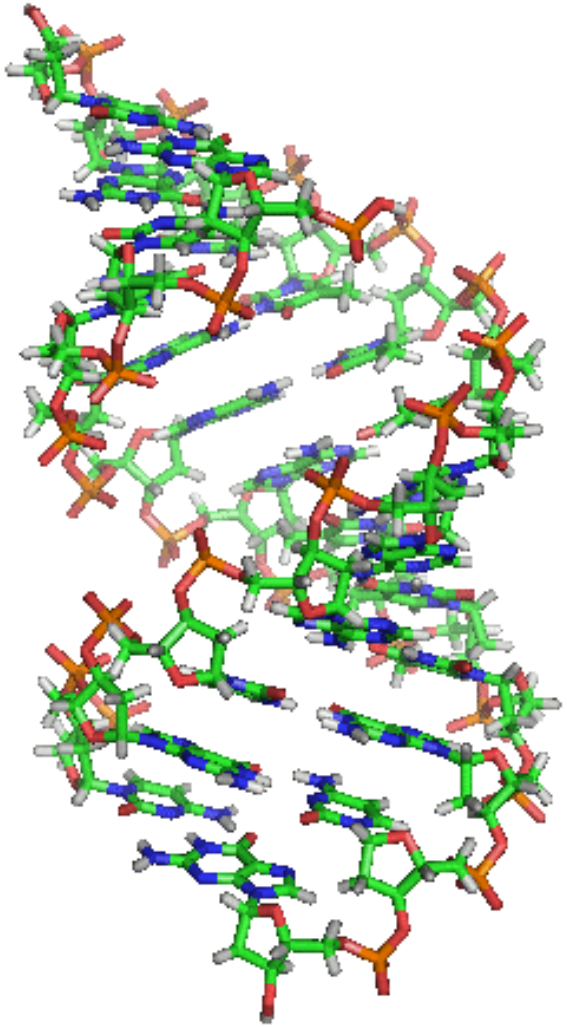
Genes are the sources of inherited information

All living things are made of cells. The **nucleus** of a cell contains **chromosomes** which carry instructions for the physical characteristics of an organism. The chromosomes are made of long strands of **DNA.** The order of molecules on the DNA strand code for protein. The instructions are called the **genetic code**. A segment of the DNA that codes for a specific protein is called a **gene**.



https://byjus.com/biology/chromosomes/

DNA forms a Double Helix shape



DNA is arranged in a **double helix** shape. The up rights of the "ladder" consist of alternating sugar and phosphate molecules bonded together. Making up the "rungs" are **two base molecules** bonded to each other.

The base molecules are held together by hydrogen bonding which can be broken and then later reformed when the DNA molecule splits to make a copy for protein manufacture or DNA replication.

extra info

http://bestanimations.com/Science/Biology/DNA/DNA.html

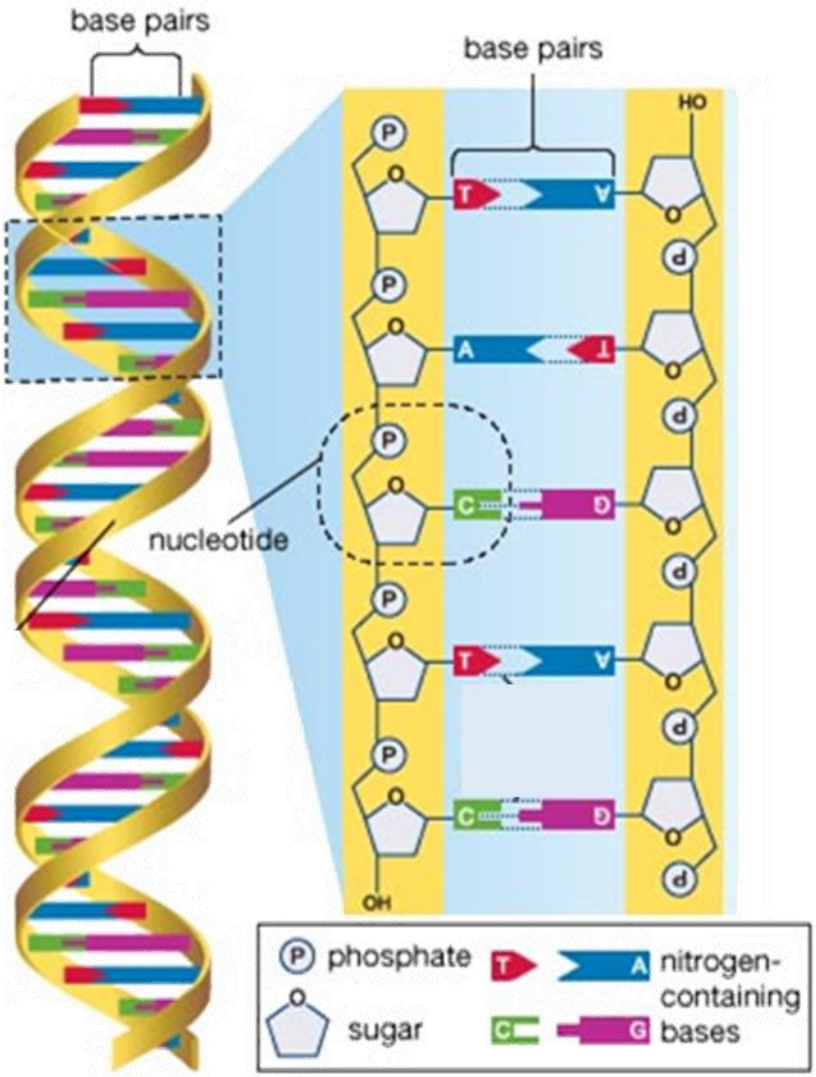
A nucleotide is one unit of DNA

DNA (deoxyribonucleic acid) is made from smaller repeating units called nucleotides which consist of a sugar, a triphosphate and a base.
There are 4 bases
A, T, C, G

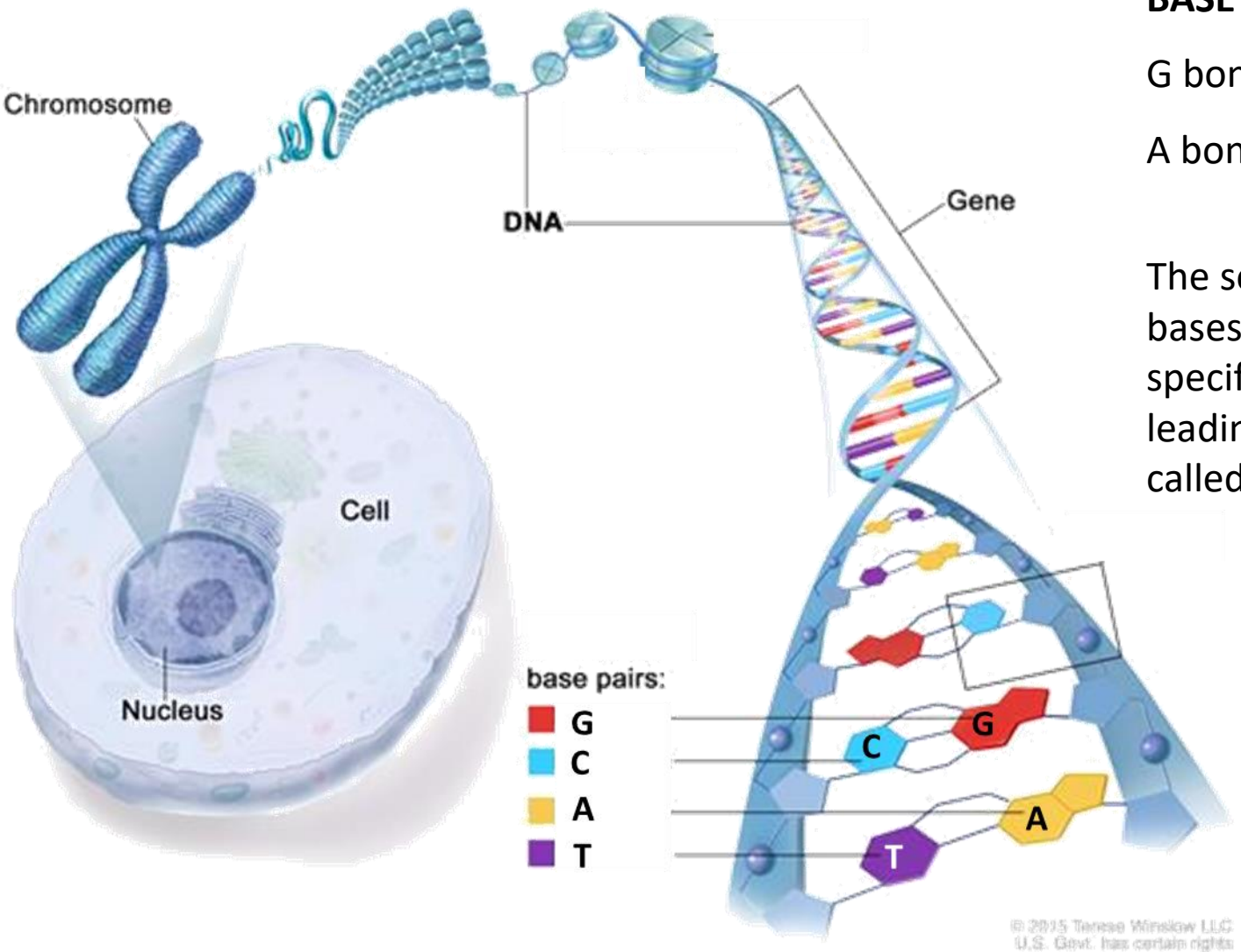
The bases have full names but we only need to know the letters
A – Adenine
C – Cytosine
G - Guanine
T – Thymine

extra info

The base pairing rule makes DNA a stable molecule with less chance of mutation



Bases form pairs with each other



BASE PAIRING RULE:

G bonds with C

A bonds with T

The sequence of bases coding for a specific proteins, leading onto a trait, is called a gene.

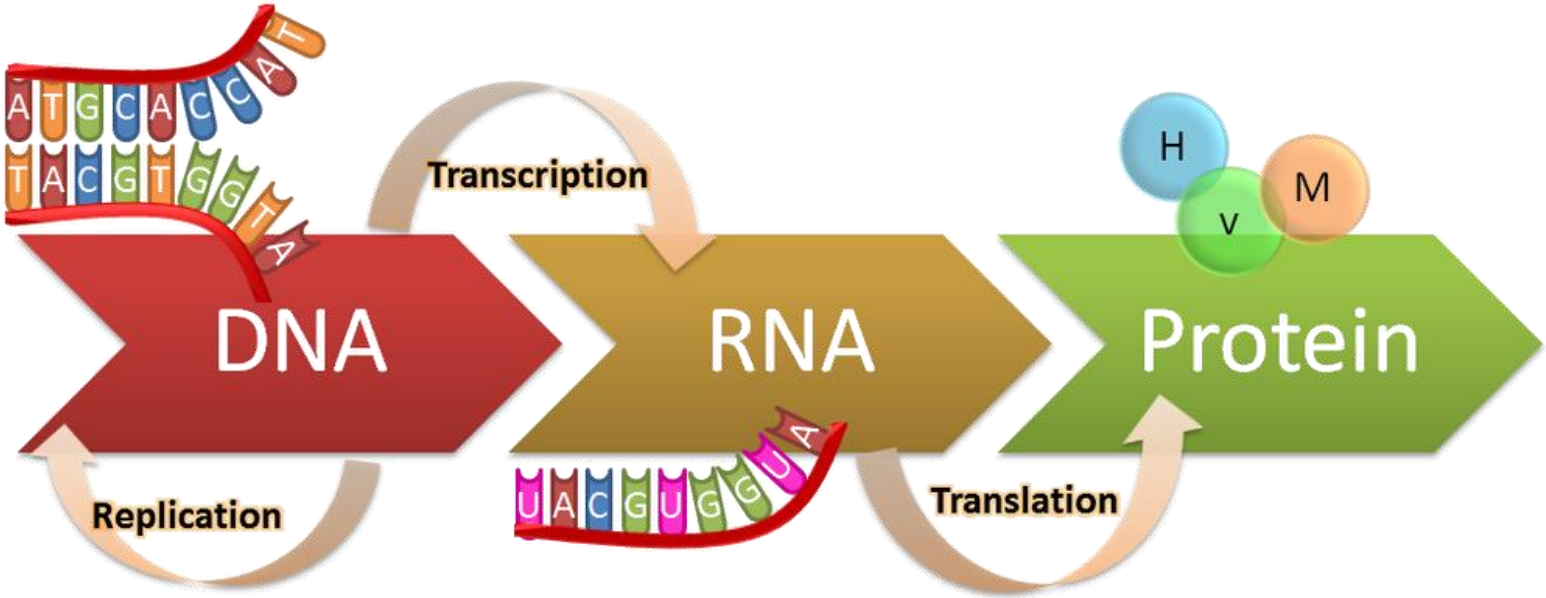
https://www.cancer.gov/publications/dictionaries/genetics-dictionary?cdrid=460201

GZ Science Resources

How to make protein

extra info

The order that the bases are arranged in the DNA segment of a gene determine the type of protein that will be formed and therefore which characteristic will be expressed.



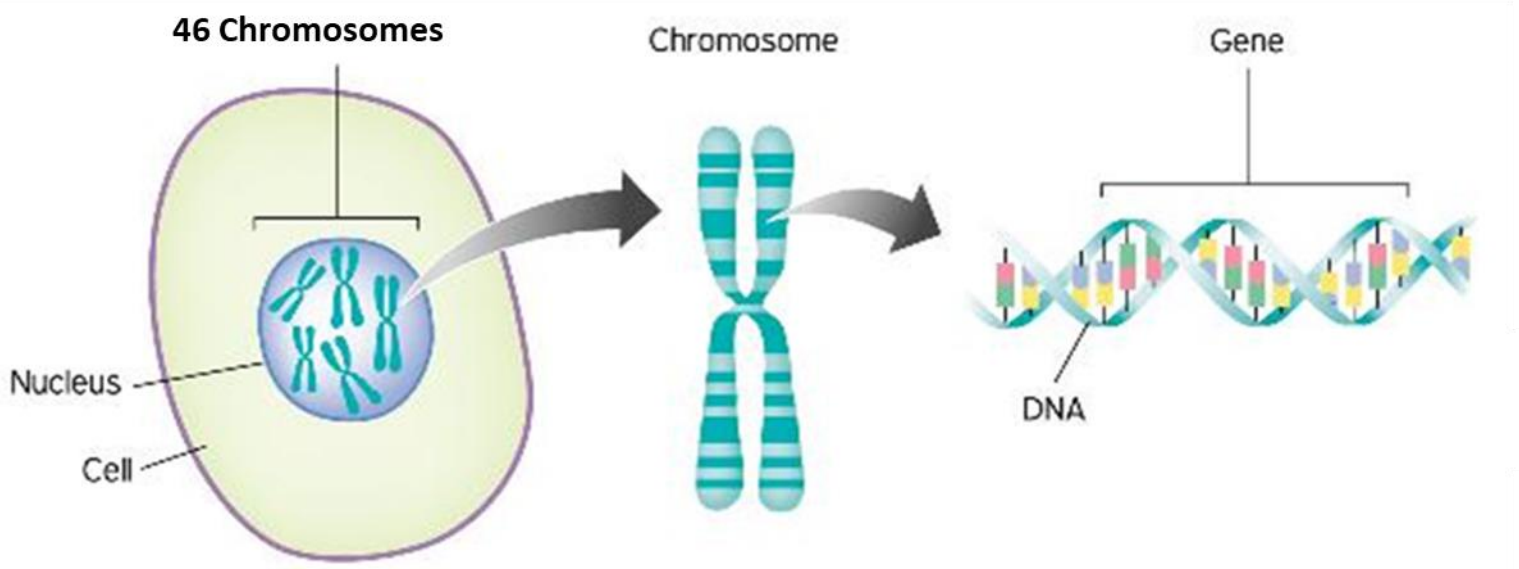
GZ Science Resources

The bases are “read” in sets of three called codons. Codons are matched to molecules called amino acids which are the building blocks of proteins.
A **single stranded copy** of DNA (called RNA) from a gene moves out of the nucleus into a part of the cell where protein is made (Transcription) and the codons (3 bases) code for different amino acids which link together to form a protein molecule. (Translation)

DNA is organised into **chromosomes and genes**

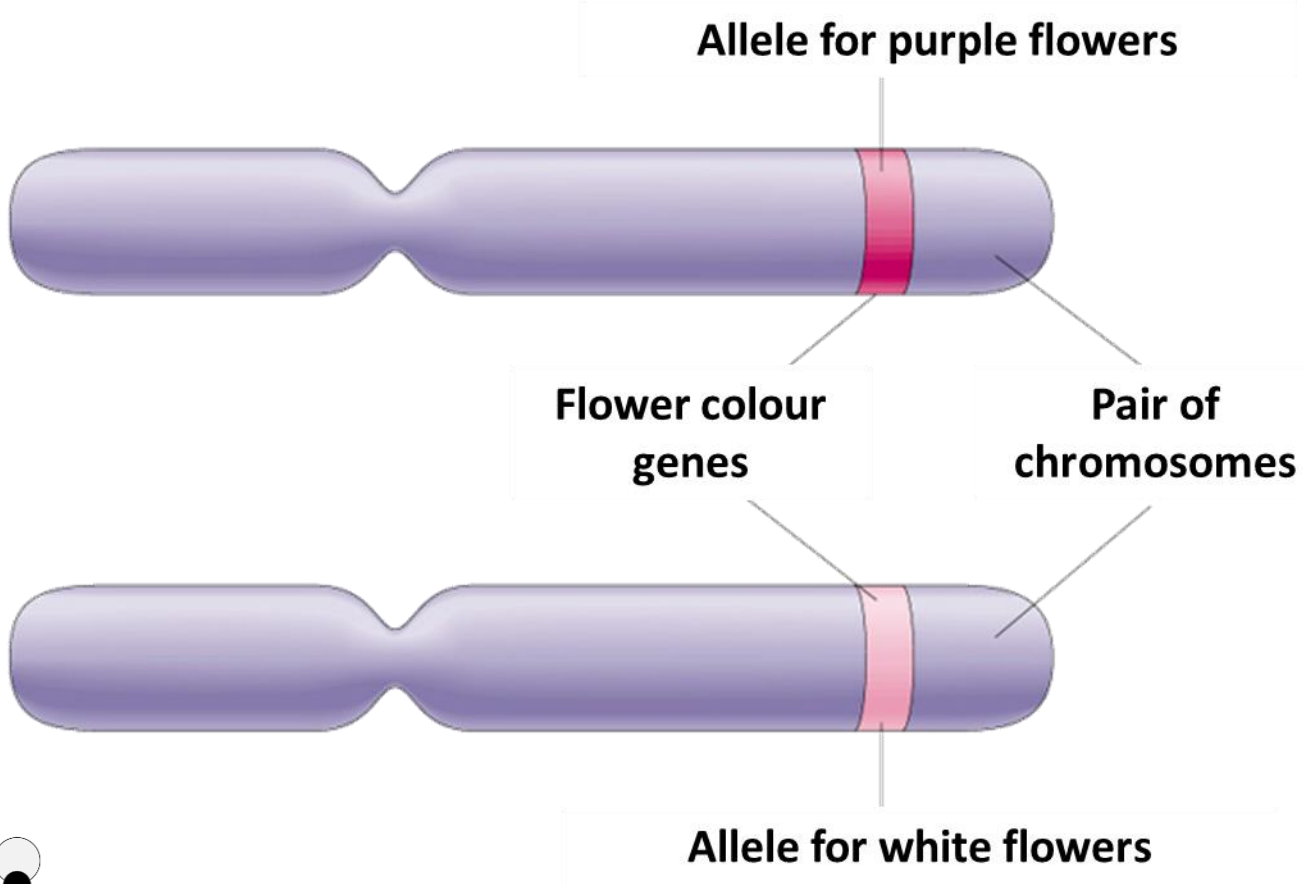
The human cell has **46 chromosomes** arranged into 23 pairs of chromosomes. Each chromosome in a pair has the same genes, (called **homologous** pairs) although there may be variation between the genes of each pair, as one comes from the father and one comes from the mother.

Each gene is represented by two **alleles**, which are different varieties. The alleles can be the same or different but the body only uses one.



Alleles are different forms of the same gene

Chromosomes occur in **pairs**. These pairs of chromosomes have the same genes in them at the same place. The versions of genes are called **alleles** and they may be different from each other.

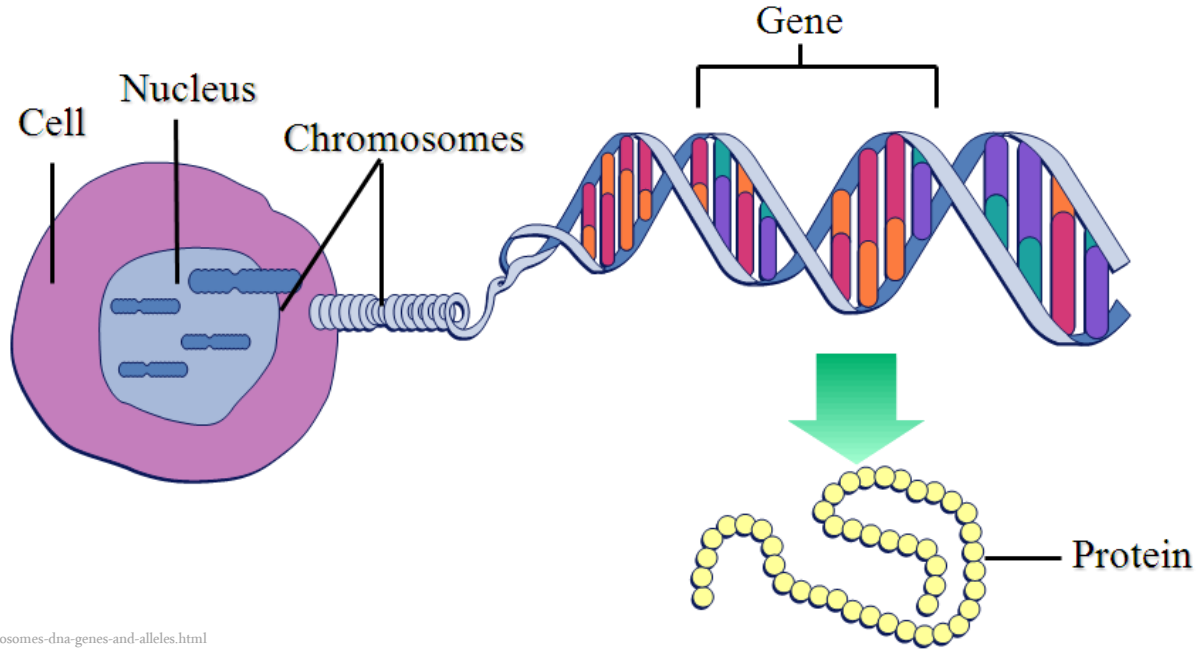


extra info

When the genes are being used only one set of instructions from the alleles is needed and the other allele is "switched off"

Summary: Cell, nucleus, chromosome, DNA, gene and protein

Chromosomes are found in the **nucleus** of each **cell** and are made up of **DNA**. DNA is a large molecule that is coiled into a double helix (twisted ladder structure). Along this molecule are **bases**. These bases pair up; A always pairs with T, and G with C.
A sequence of bases which codes for a particular **trait/characteristic** (eg, eye colour) is called a **gene**. Genes code for **proteins** - the **genetic code/ base order** determines which particular **protein** is made and therefore which **characteristic** is coded for.
The different versions of each gene are called **alleles**, and these show the different **variations** of each characteristic, eg brown / blue eyes. Because chromosomes come in pairs for each trait, there will be two possible alleles. These different versions of genes (alleles) occur because the DNA base sequence is different.

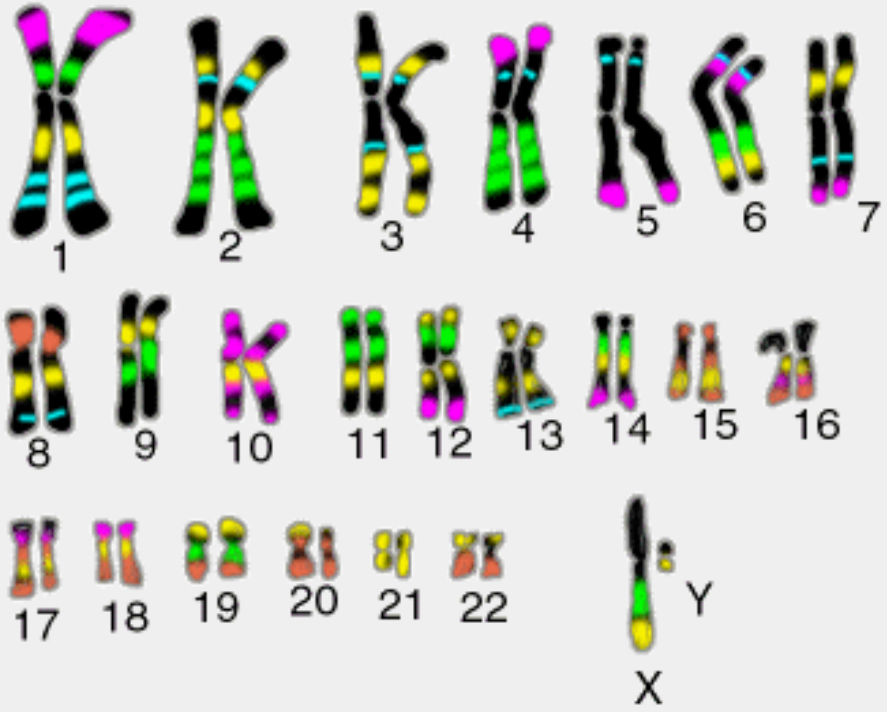


Background Knowledge

Chromosomes come in pairs

extra info

Chromosomes come in pairs. One pair is the sex chromosomes – XX in females and XY in males. A complete set of chromosomes of an organism placed into pairs of matching chromosomes is called a **karyotype**. The human karyotype consists of 23 pairs of chromosomes.

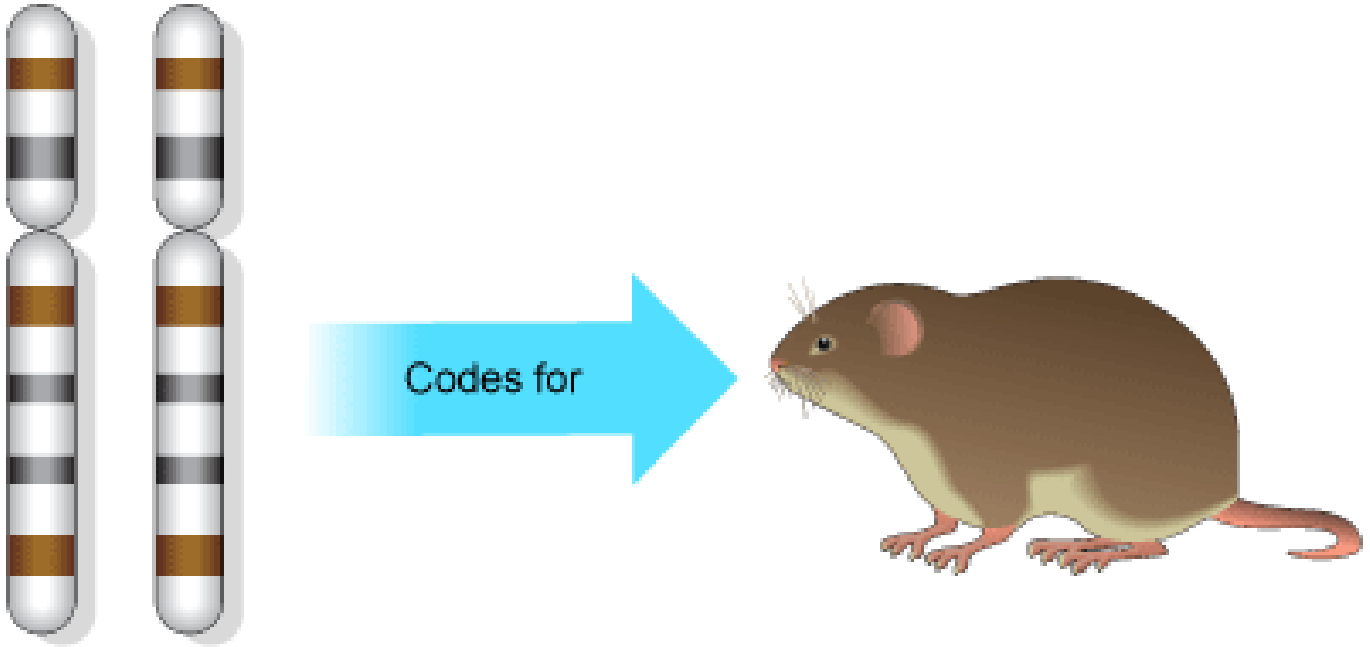


GZ Science Resources

Phenotype and Genotype

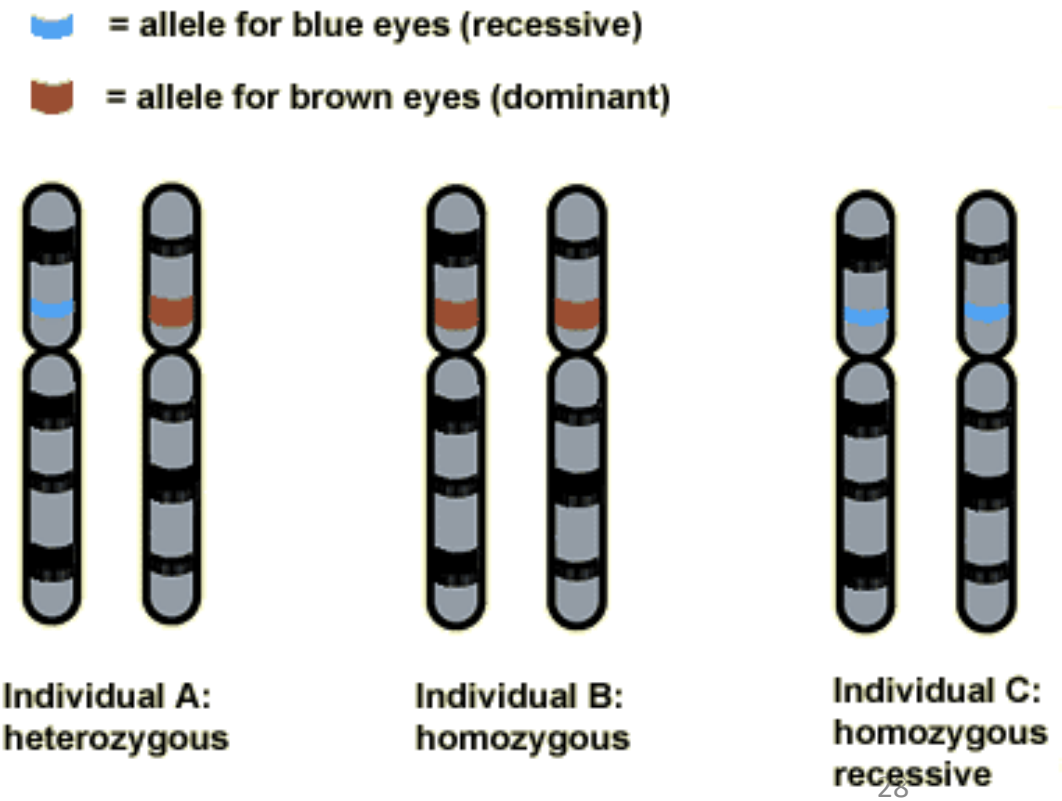
The combination of alleles for each trait is called the **genotype**; this can be any combination of two of the available alleles.

The **phenotype** is the physical trait that is expressed because of the alleles.
The genotype determines the phenotype (the physical appearance) of the organism.



Dominant and recessive alleles

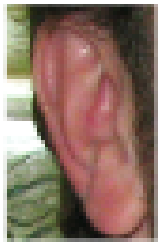
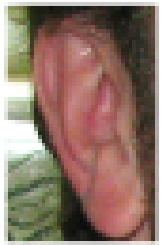
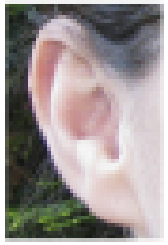
Alleles that are present in the pair are expressed. **Dominant** alleles (B) if present will be expressed over **recessive** alleles (b).
Only if there are no dominant alleles present in the pair will the recessive allele be expressed. Expressed means the protein is produced which determines the characteristic.



When there are two of the same allele this is called **homozygous** and the cell could randomly use either allele. When there is two different alleles this is called **heterozygous** and the cell always uses the dominant allele. **Pure Breeding** is another term for homozygous.

Dominant and recessive genes

Recessive alleles need 2 copies present to be expressed in the phenotype
Dominant allele needs only 1 copy present to be expressed and is always expressed when present

Genotype	Phenotype
EE Homozygous dominant	Detached Earlobes 
Ee Heterozygous	Detached Earlobes 
ee Homozygous recessive	Attached Earlobes 

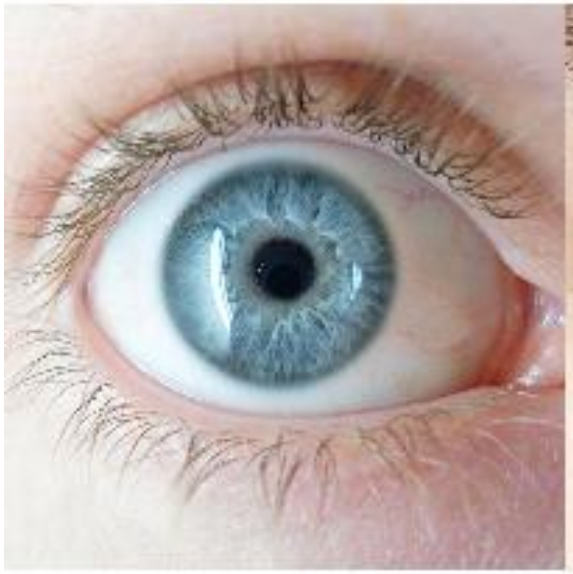
Many letters can be used to represent dominant or recessive, such as Bb, Ee, Ff as long as the upper and lower case letters look different.

The genotype determines the phenotype.

Phenotype, genotype and dominance

When the **phenotype** is recessive then the **genotype** can only be homozygous recessive. If the phenotype is dominant then the genotype can either be heterozygous or homozygous dominant, as long as one dominant allele is present in the genotype.

Phenotype= Blue Eyes



Genotype=bb

Recessive=b

Phenotype=Brown Eyes

Genotype = Bb or BB

Dominant =B

Lop Eared rabbits – an example

Rabbit ears normally point straight up. Some rabbits have an allele for lop ears that cause the ears fold down. The allele that produces lop ears is recessive and was created by a **mutation**.

lop eared rabbit

straight eared rabbit

We can use the symbol R to show the dominant allele and r to show the recessive allele. The genotype of the two rabbits if both are **pure breeding** are:

Lop eared rabbit genotype
rr

Normal eared rabbit genotype
RR

Gregor Mendel

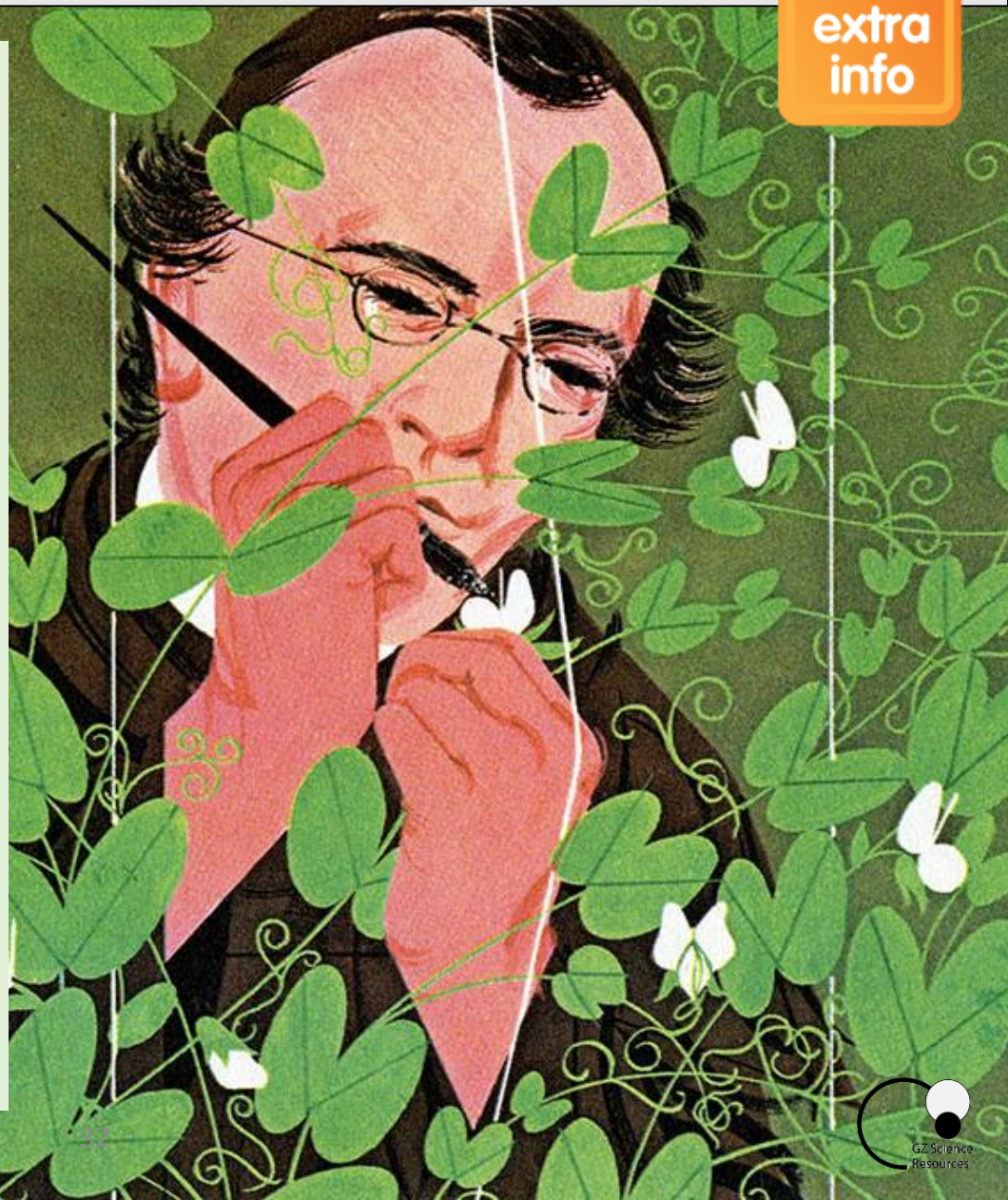
extra info

Gregor Mendel was a German Friar who lived from 1822-1884. Mendel is called the "Father of Genetics" because through his work on over 10,000 pea plants he discovered the main laws of inheritance.

1) **The Law of Segregation**: Each inherited trait is created by a gene pair. Adult genes are randomly separated to the gametes and gametes contain only one gene of the pair. Offspring inherit one genetic allele from each parent when gametes combine in fertilization.

2) **The Law of Independent Assortment**: Genes for different traits are sorted separately from one another

3) **The Law of Dominance**: An organism with different alleles of a gene will use the allele that is dominant.



Using Punnett squares to predict offspring

We use Punnett squares to predict the frequency of the genotypes of any offspring created when two organisms are mated, and therefore phenotypes.

Agouti Rabbit

Parents genotypes

Black Rabbit

	B	b
b	Bb	bb
b	Bb	bb

B is the dominant allele for Agouti colour.

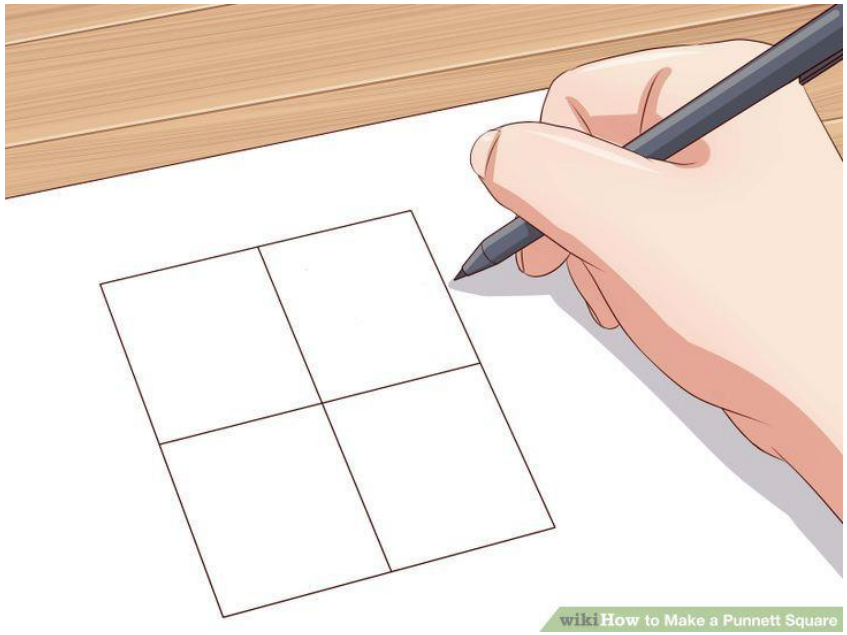
b is the recessive allele for Black colour

Each adult gives one allele from its pair to each offspring.

Possible offspring genotypes

GZ Science Resources

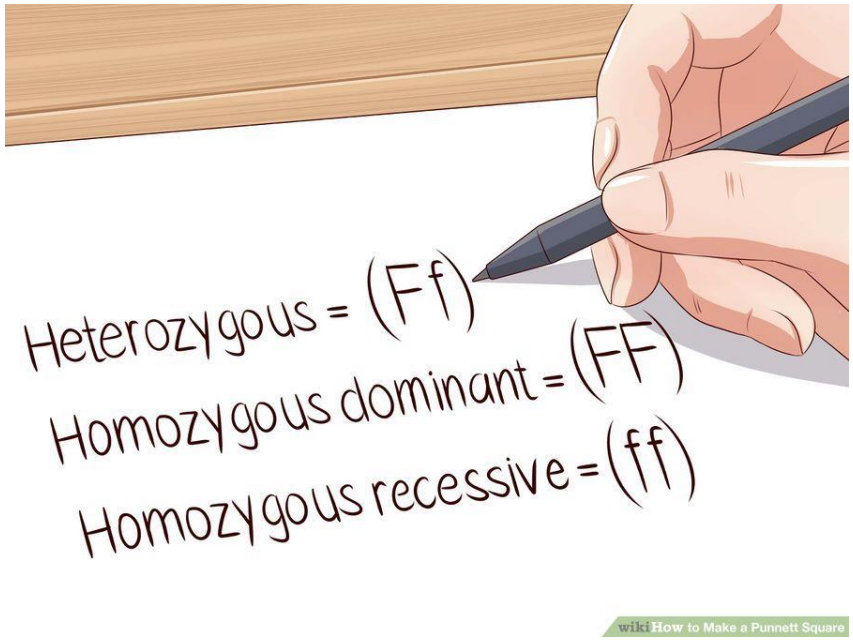
How to use a Punnett squares to predict offspring



1. Draw a grid with 4 squares. Each square will represent 1 out of 4 offspring or 25% of offspring out of 100%

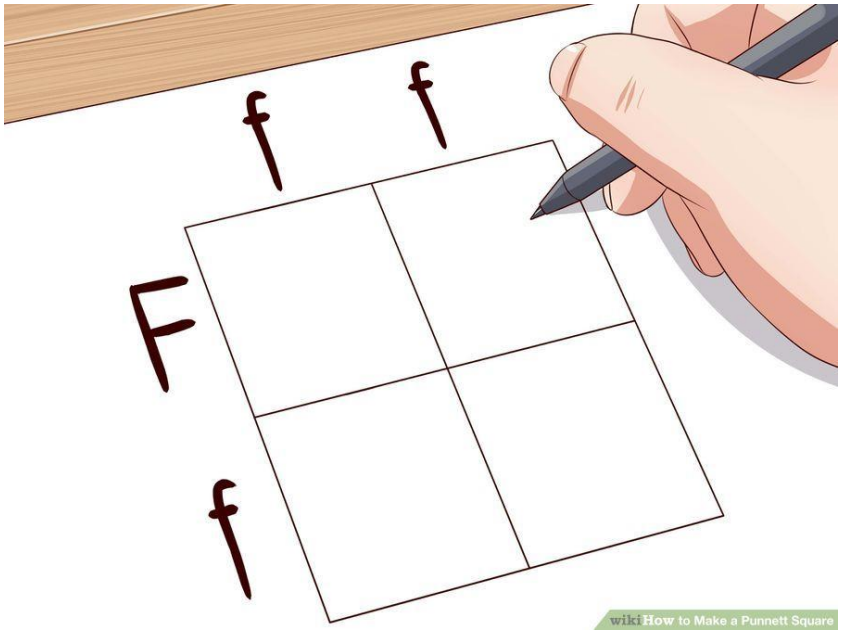
2. Write down the possible 2 phenotypes (physical traits) and label them with a capital letter for the dominant trait and lower case letter for the recessive trait.

How to use a Punnett squares to predict offspring

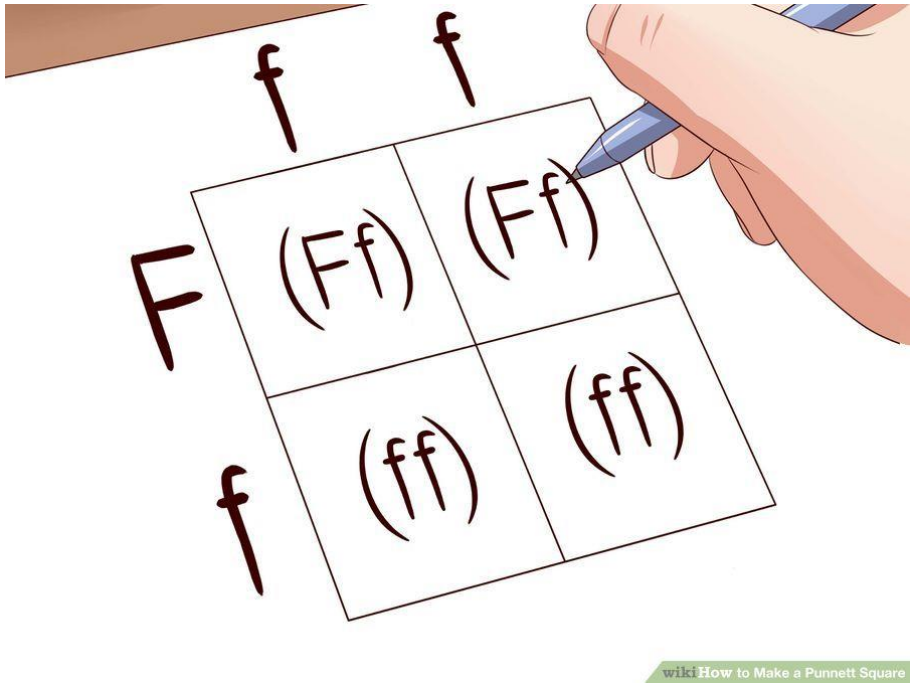


3. Write down all 3 possible genotypes and their letter combinations.

4. Select the correct genotypes for each parent (they may be the same or different) and write them with one parent on the top and one parent to the left side.



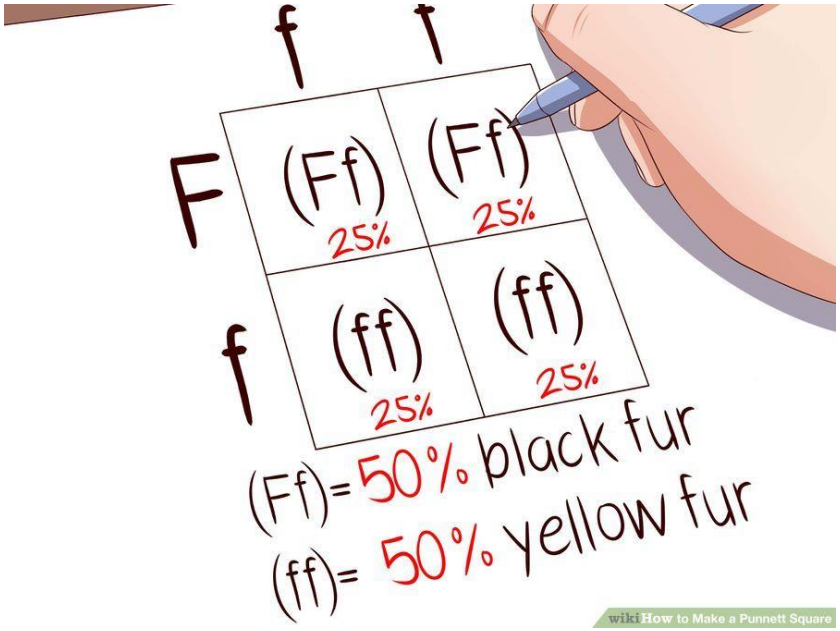
How to use a Punnett squares to predict offspring



5. Write each letter in the square below for one parent and squares to the right for the other parent. Each offspring will have a genotype of 2 letters.

6. Calculate the genotype ratio – the total percentage each genotype occurs (in this order FF : Ff : ff) or total out of 4 squares.

Then calculate phenotype ratio – FF and Ff count as dominant and only ff counts as recessive (in this order dominant : recessive) can be % or out of 4



Using Punnett squares to predict offspring

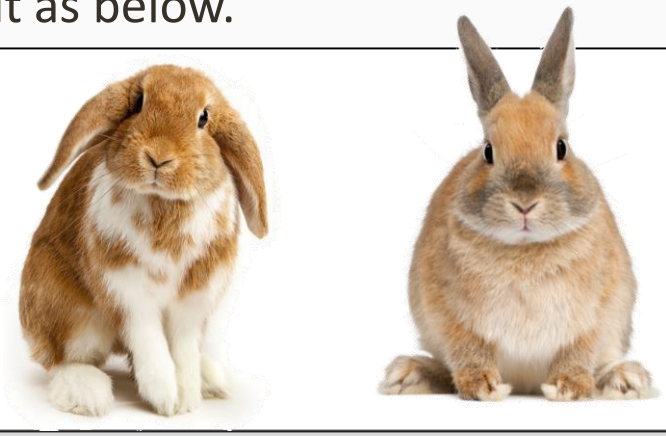
The Punnett square is used to predict the **probability** of what the offspring's phenotype and genotype will be, which may or may not match up to the actual results due to the random nature of each fertilisation.

The phenotype and genotype ratios are only **predictions.** Each time a new offspring is created through fertilisation it is a new event and the same probabilities apply regardless of the phenotype or genotype of previous offspring.

Calculating Phenotype and genotype ratios example

We can use the example of our straight eared and lop eared rabbit again when they breed, and all their offspring will have the genotype of Rr and phenotype of straight ears. If we cross two of their offspring (Rr) the genotype and phenotype ratios of **their offspring** (second generation) can be set out as below.

	R	r
R	RR	Rr
r	Rr	rr



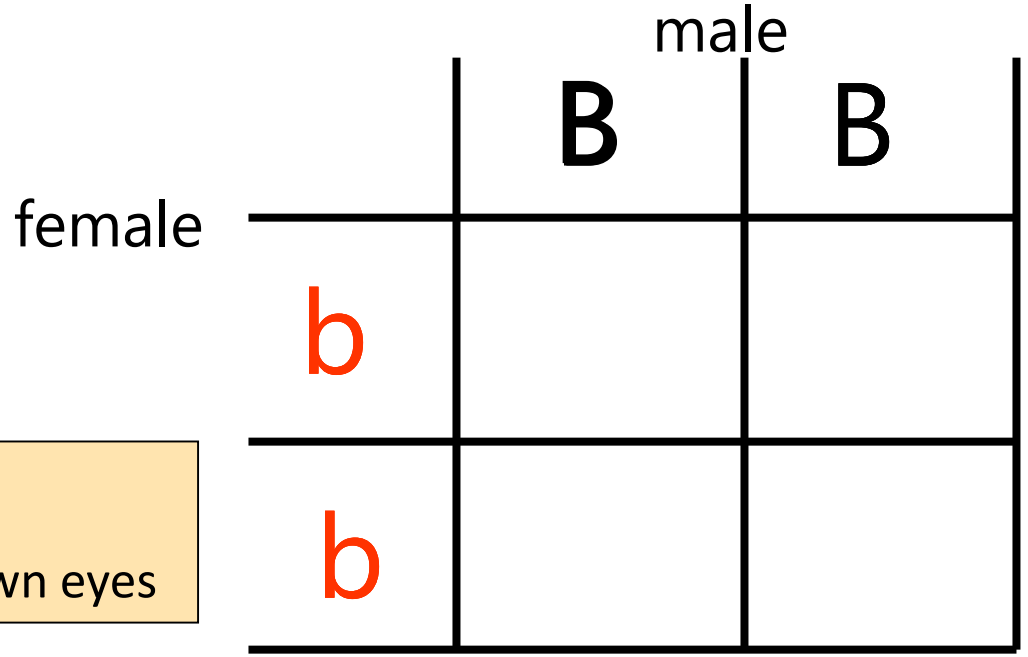
Genotype Ratios		
RR	Rr	rr
1	2	1
Phenotype Ratios		
Straight Ears	Lop ears	
3	**1**	

Using Punnett squares to predict offspring

1. **Determine** the **genotypes** of the parents or whatever is given in problem.
2. Set up your **Punnett square** as follows:

Male – BB genotype
brown eyes - phenotype

Female – bb genotype
blue eyes - phenotype



Genotypic ratio = 100% Bb

Phenotypic ratio = 100% Brown eyes

3. **Fill in** the squares. This represents the possible combinations that could occur during fertilization.
4. **Write out** the possible **genotypic ratio** of the offspring.
5. **Using** the **genotypic** ratio determine the phenotypic ratio for the offspring.

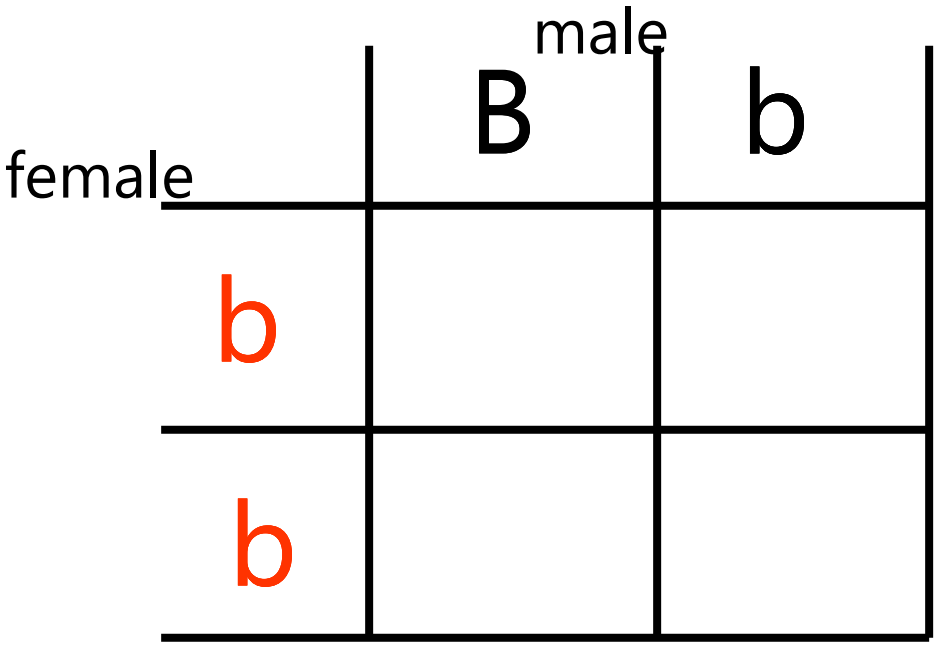
Using Punnett squares to predict offspring

A heterozygous male, black eyed mouse is crossed with a red eyed, female mouse. Predict the possible offspring!

Male – Bb genotype
black eyes - phenotype

Female – bb genotype
red eyes - phenotype

Black must be dominant (B) as phenotype is black when mouse is heterozygous (has both B and b)



Genotypic ratio = 50% Bb 50% bb
Phenotypic ratio = 50% Black eyes 50% red eyes

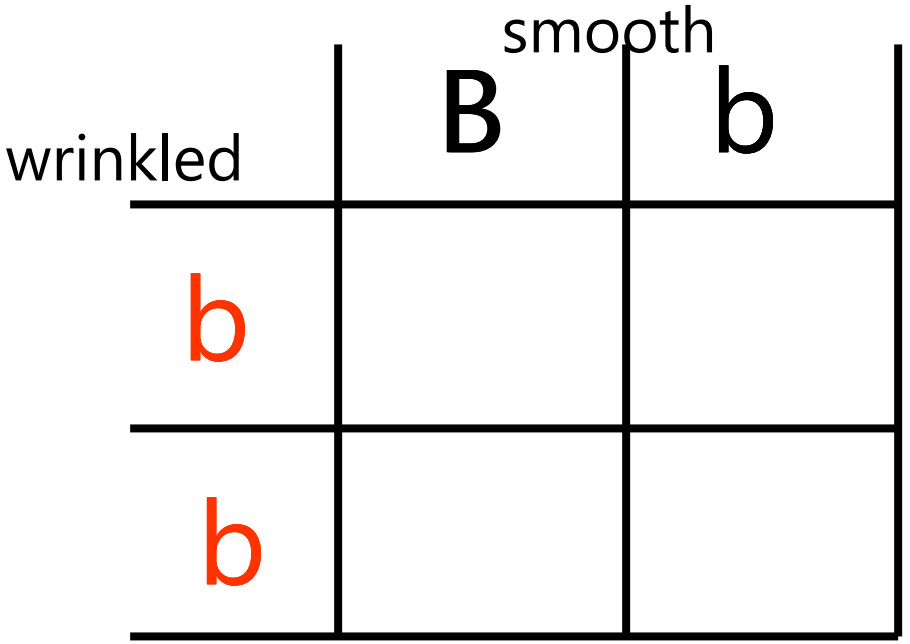
Using Punnett squares to predict offspring

A heterozygous, smooth pea pod, plant is crossed with a wrinkled pea pod plant. *There are two alleles for pea pod, smooth and wrinkled.* Predict the offspring from this cross.

heterozygous Bb genotype
smooth - phenotype

homozygous bb genotype
wrinkled - phenotype

Smooth must be dominant (B) as phenotype is smooth when plant is heterozygous (has both B and b)



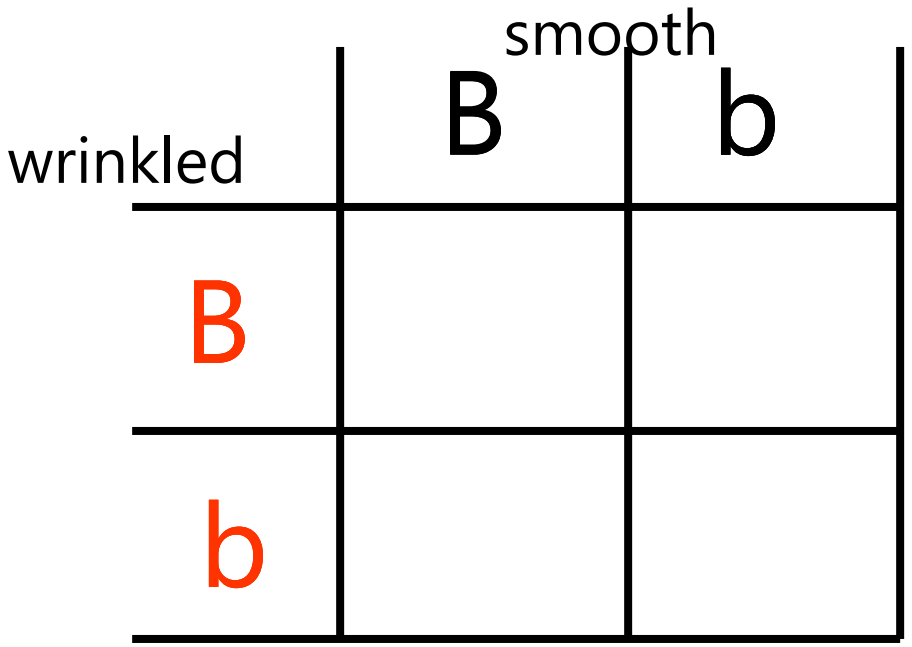
Genotypic ratio = 50% Bb 50% bb
Phenotypic ratio = 50% smooth 50% wrinkled

Using Punnett squares to predict offspring

Two heterozygous, smooth pea pod, plants are crossed. *There are two alleles for pea pods, smooth and wrinkled.* Predict the offspring from this cross.

heterozygous Bb genotype
smooth - phenotype

Smooth must be dominant (B) as phenotype is smooth when plant is heterozygous (has both B and b)



Genotypic ratio = 25% BB 50% Bb 25% bb
Phenotypic ratio = 75% smooth 25% wrinkled

Using Punnett squares to predict parents genotype

extra info

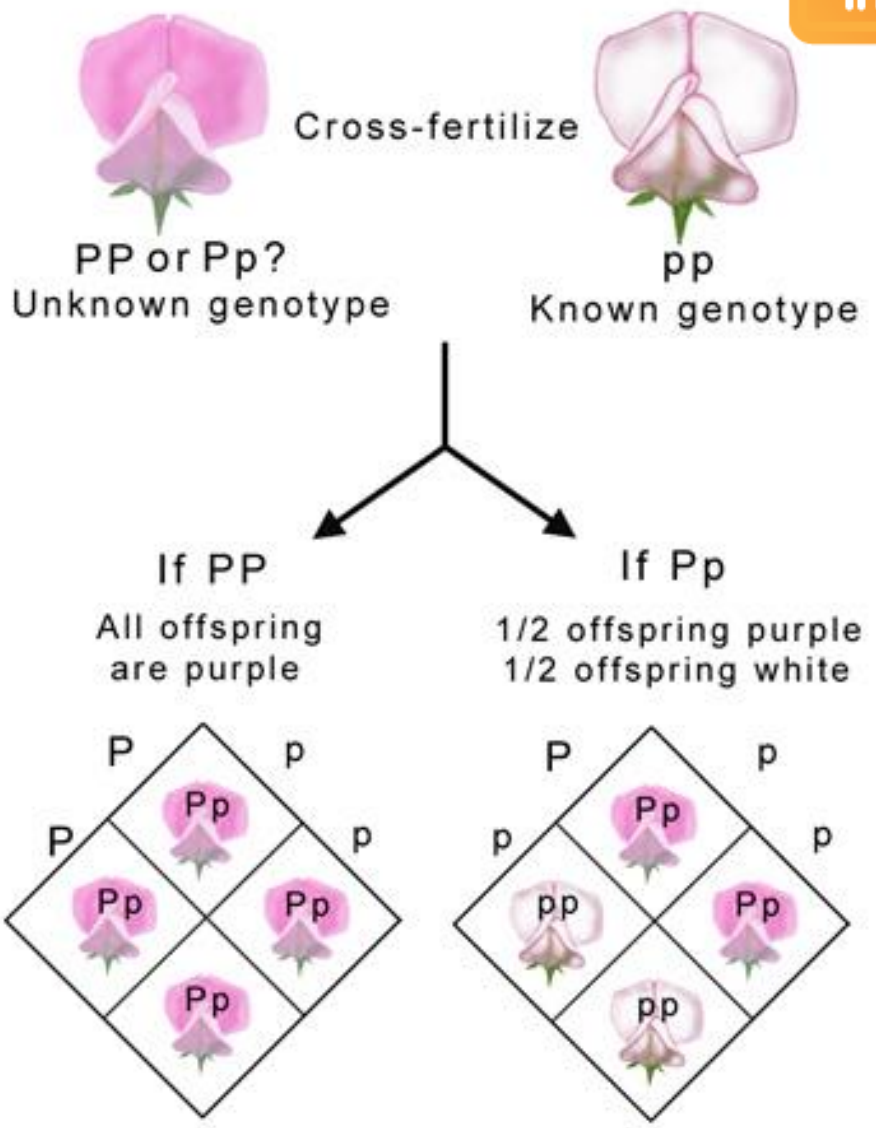
Parent's genotypes can be predicted by the phenotype of the offspring. If 100% of the offspring show the dominant phenotype then at least one of the parents must be homozygous dominant.
If any of the offspring show the recessive phenotype then each parent must contain at least 1 recessive allele each in order to have offspring that has a recessive allele donated from each parent. If the parents show the dominant phenotype then they must be heterozygous.

GZ Science Resources

Pure Breeding and test Crosses

extra info

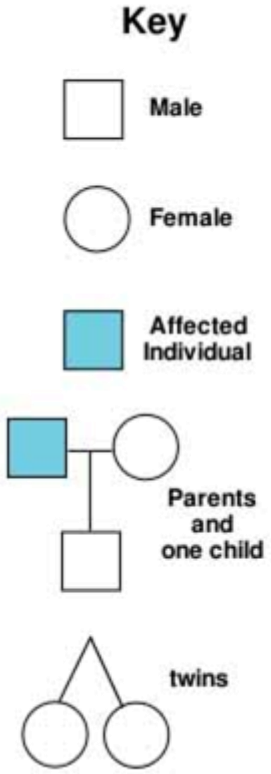
An individual that is **pure breeding** has a homozygous (two of the same alleles) genotype. The pure breeding recessive homozygous can be seen by its phenotype – it will show the recessive phenotype. However, a pure breeding dominant homozygous has the same phenotype as a heterozygous individual. The pure breeding individual can be found by a **test cross** which it mates with a recessive phenotype individual. If any of the offspring have the recessive phenotype then the individual is not pure breeding.

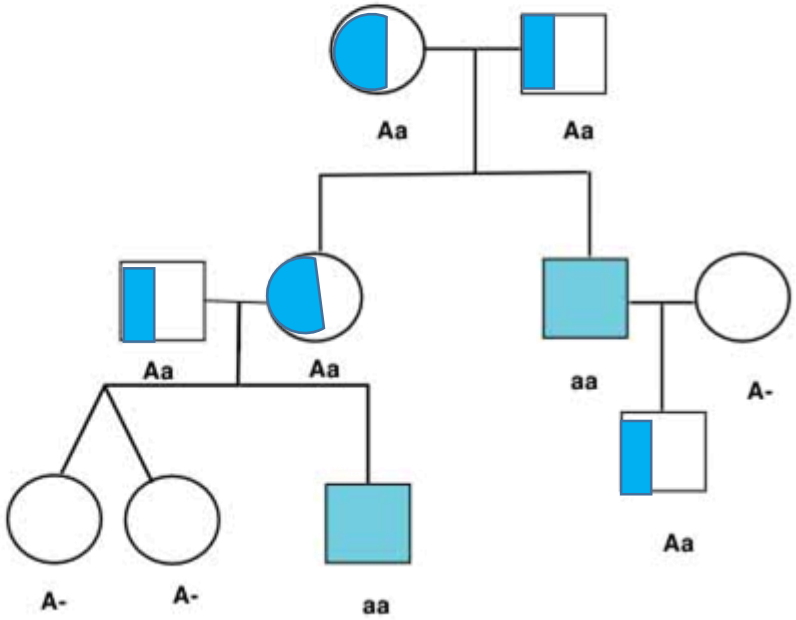


GZ Science Resources

Using Pedigree charts to predict offspring

A pedigree chart is a diagram that shows **inheritance** patterns of a certain allele. A square represents a male and a circle represents a female. If a person's symbol is shaded in, this means that they have the phenotype. It if is half-shaded, then they are heterozygous but do not have the phenotype. If they are not shaded at all, they don't have the allele. Pedigree charts are good for showing the patterns of a recessive or dominant gene.



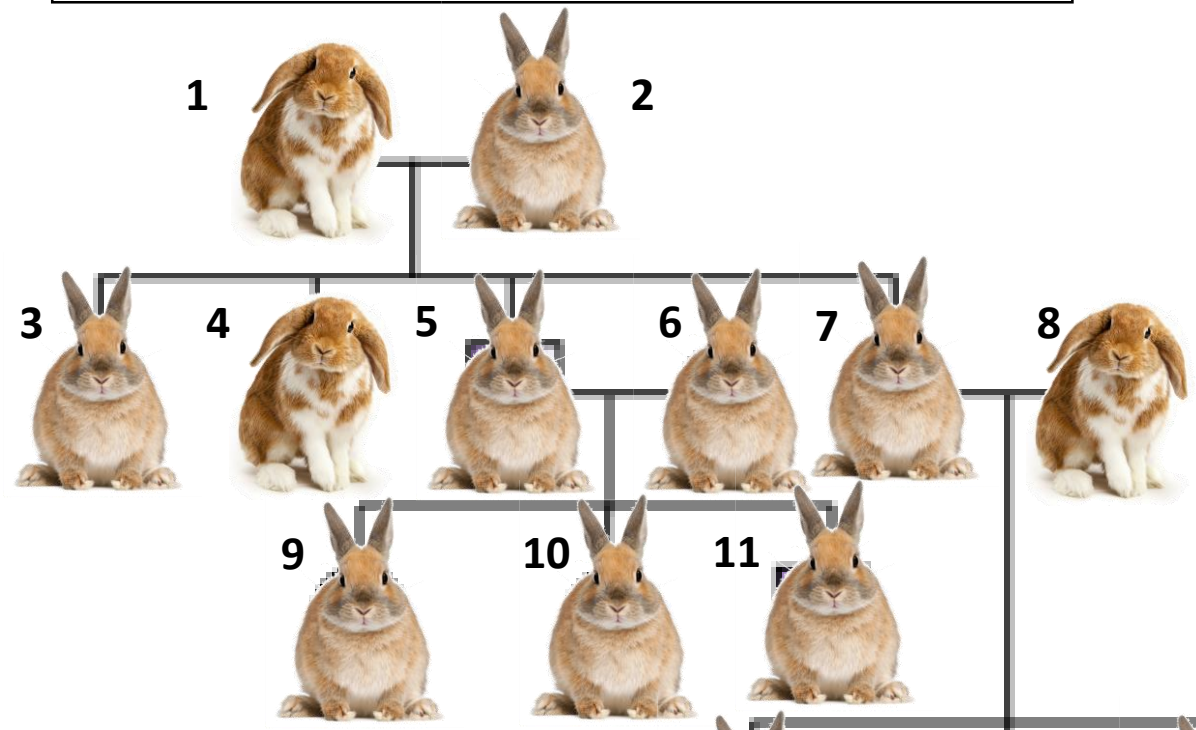


GZ Science Resources

Using Pedigree charts to predict genotype

Lop-eared
rr

straight-eared
RR or Rr



The genotypes of rabbits 9,10 and 11 cannot be confirmed as RR or Rr without more information

When an individual has a recessive phenotype on a pedigree chart then the **genotype must always be homozygous recessive** (i.e. rr for the lop-eared rabbit 1)

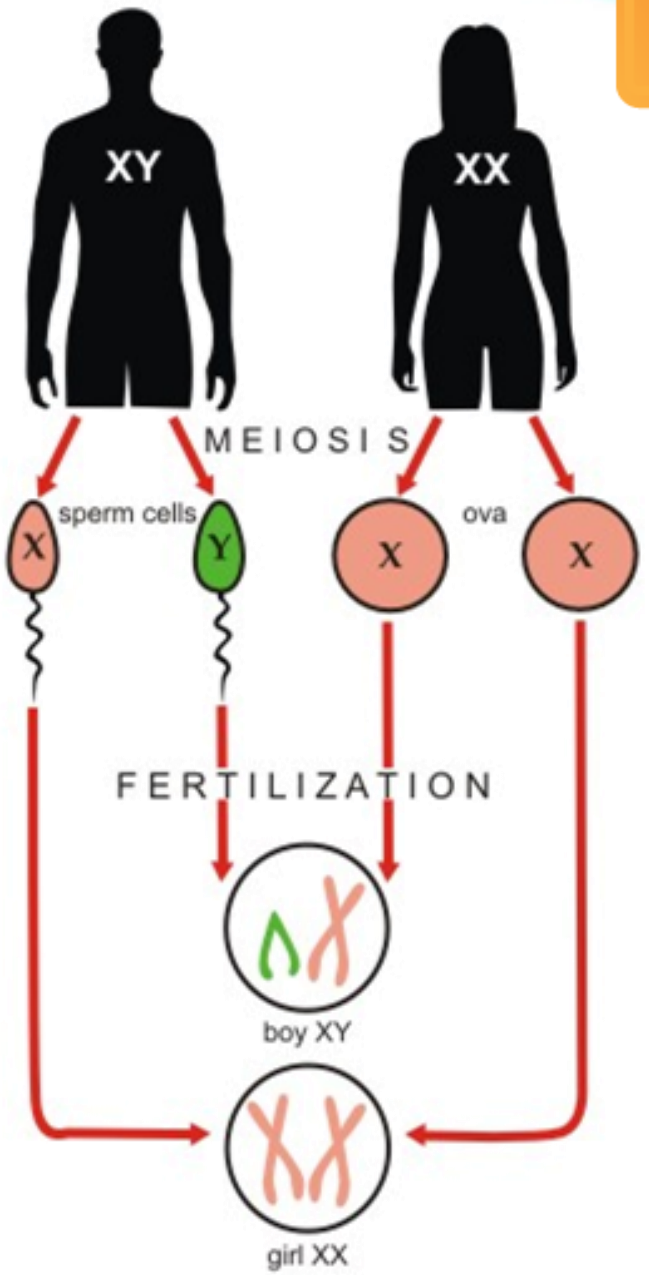
To find out the genotype of a dominant phenotype you can look at either the parents of the offspring. (i.e. rabbit 7 is straight eared but had one parent who was a lop-eared rr so must have one r allele – genotype Rr. Also 13 is an rr (see above) so rabbit 7 must have at least one r allele to pass onto 13.

Sex determination

extra info

A pair of chromosomes are called the **sex chromosomes**. The female always has a homologous (same) pair of two x chromosomes. The female can only give a x chromosome to a gamete. The male has a x and y chromosome. He can give either an x or y chromosome to form a gamete. **The male determines the sex of any children.**

Male genotype = XY
Female genotype = XX



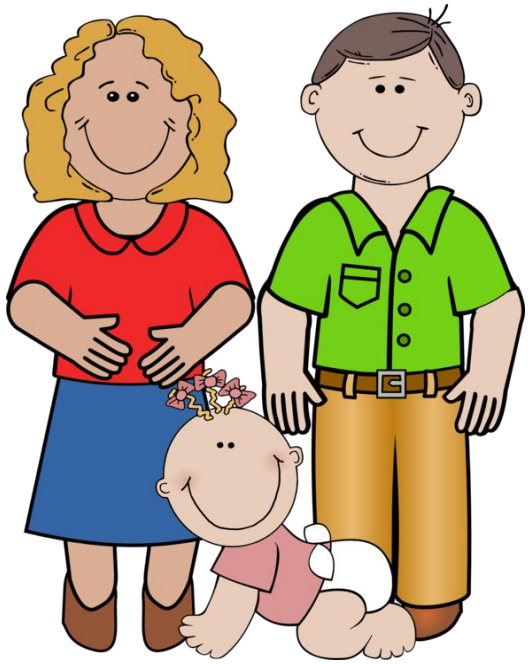
GZ Science Resources

Sex determination

extra info

A Punnett square can be used to demonstrate that in any fertilisation there will be a 50% chance of either a boy or a girl.

If there are a small number of offspring then there is less chance that the actual ratio of male to female offspring will be the same as the predicted ratio.
Each new fertilisation is independent of any previous fertilisation before hand.



	Male gametes: X	Male gametes: Y
Female gametes: X	XX	XY
Female gametes: X	XX	XY

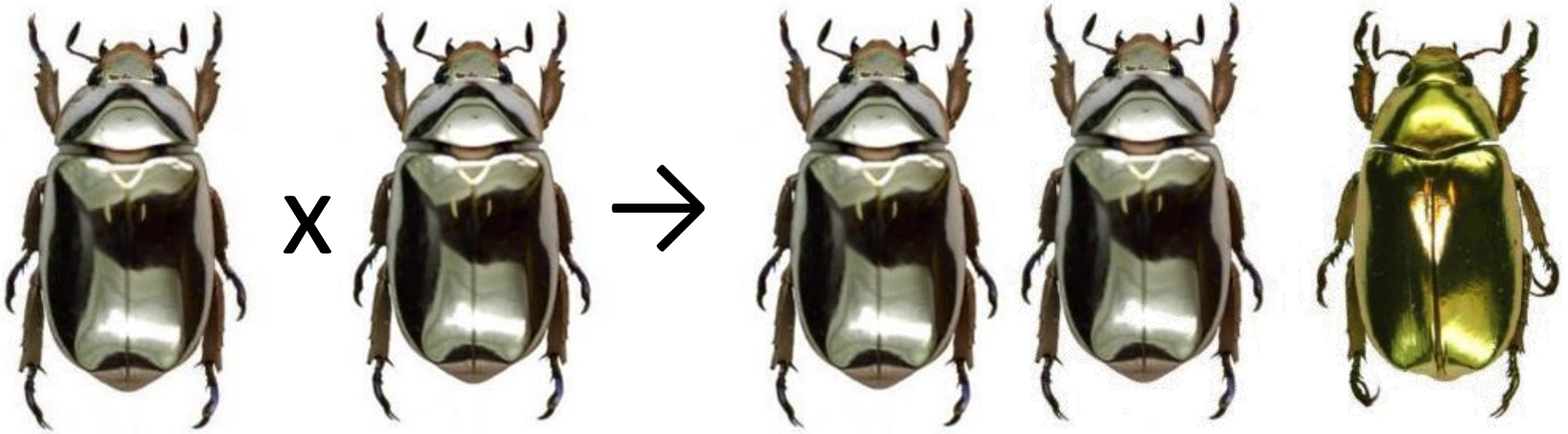
GZ Science Resources

Mutations introduce new alleles into a population

extra info

Most mutations cause death because the gene in which the mutation occurs creates an incorrect protein. Very occasionally mutations produce a new type of protein which gives the organism an advantage over others in its species in adapting to its environment. The organism containing the mutation will have more chance of surviving than those individuals without it and it will pass the mutated gene on to the next generation more successfully.

Mutations increase variation in a population by adding new types of alleles.



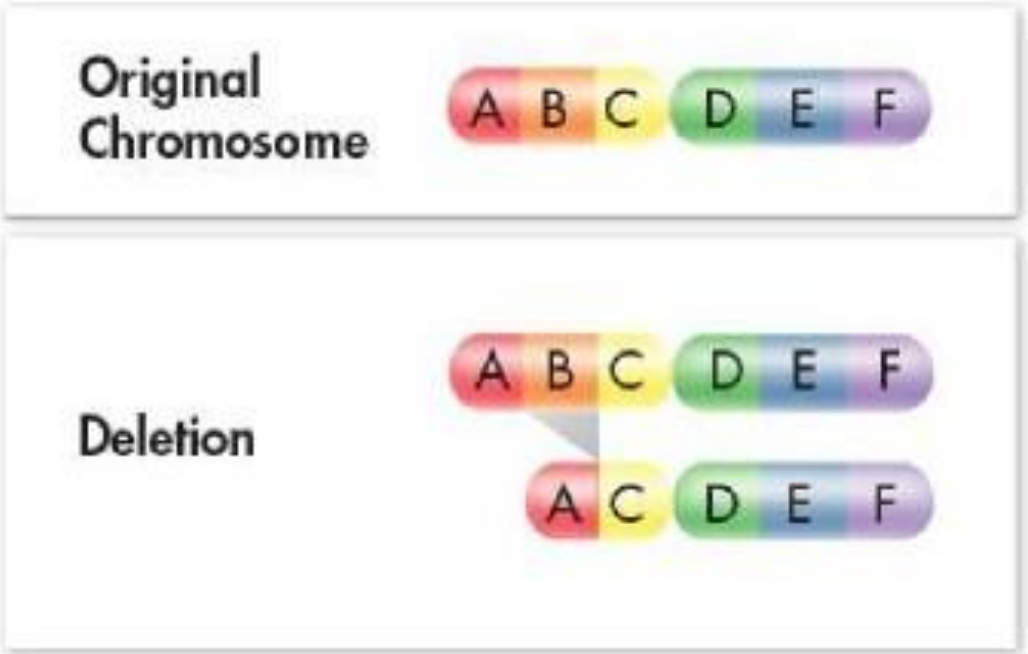
Normal parents **Normal offspring** **Mutant offspring**

Mutations are caused by a random change in the sequence of bases in the DNA.

extra info

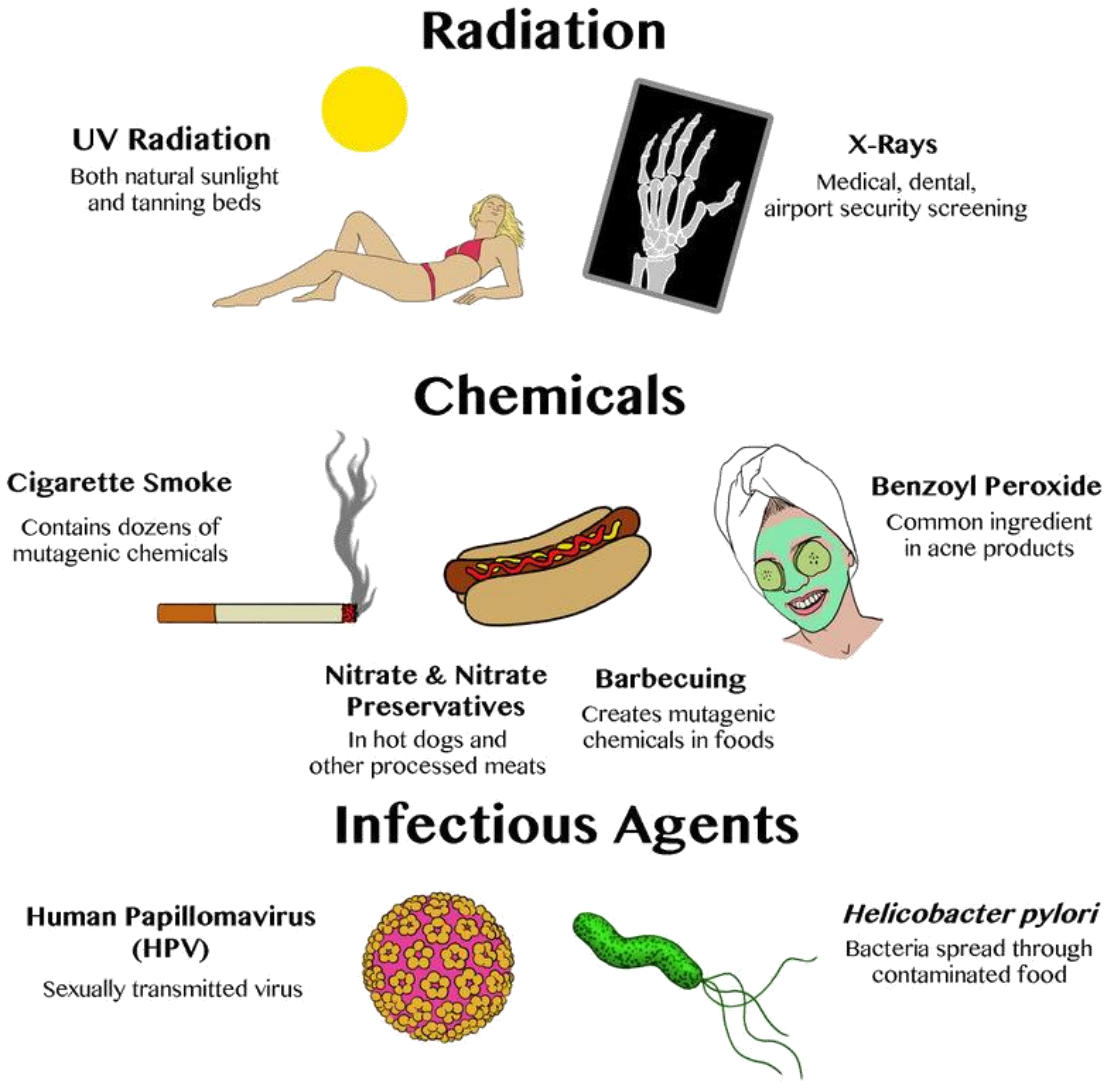
Mutations can either occur in individual cells of an organism such as cancer or during the process of Meiosis to form the gametes (egg or sperm cells) which causes every cell in the fertilised developing organism to contain the mutation.

Mutations can be cause by a single change in one base pair – either deleted, an extra added or a base changed, one segment of DNA or gene, or a whole chromosome added or deleted.



GZ Science Resources

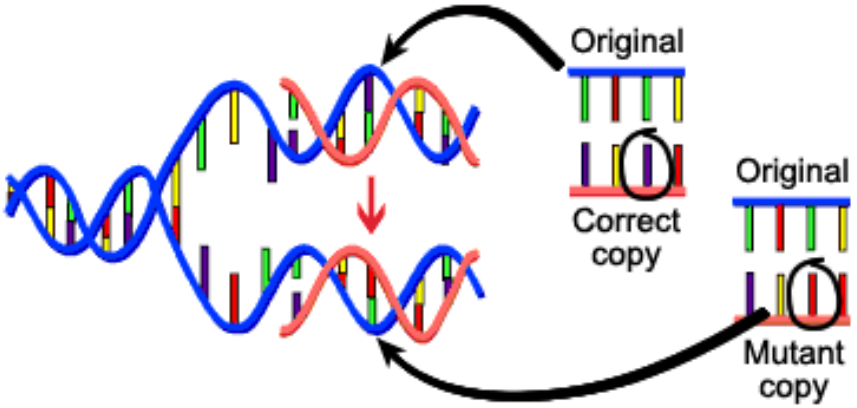
Humans can have specific genetic disorders caused by a mutation during meiosis



Inheritable Genetic disorders in humans are caused by one or more mutations in the genome (full set of genes), and is present from birth. Most genetic disorders are very rare. Mutations can be caused by random mistakes during DNA replication and meiosis or by environmental effects like chemical or radiation exposure during meiosis.

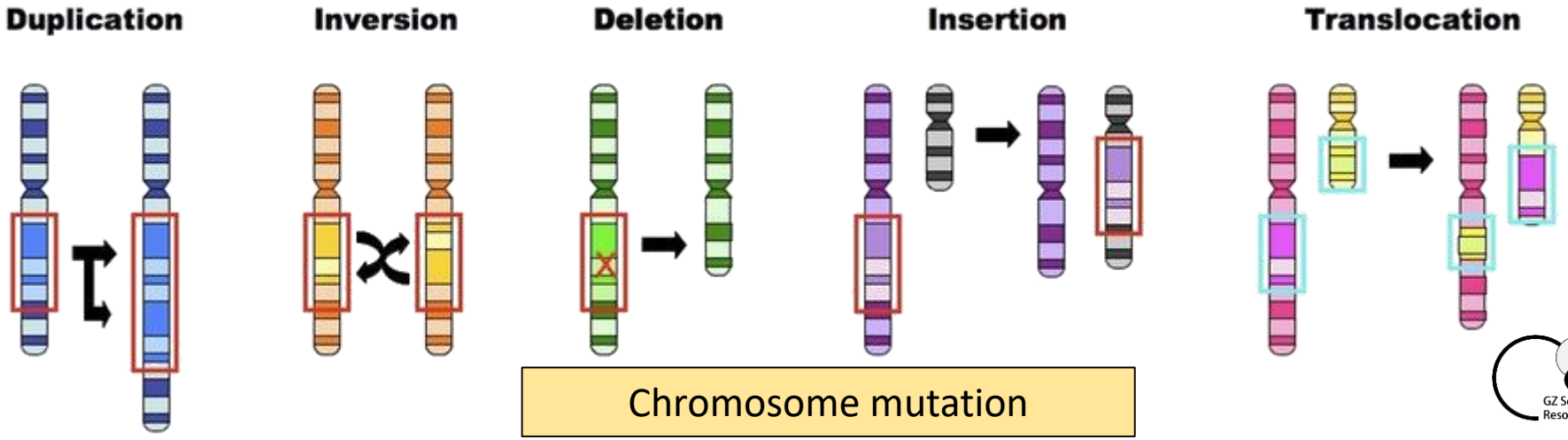
Humans can have specific genetic disorders caused by a mutation during meiosis

extra info



Single base mutation

Genetic disorders are passed down from the parent's genes. Some disorders occur through a mutation in an entire chromosome (or the addition of an extra chromosome) and cause a syndrome with many different parts of the body effected, such as down syndrome. Other disorders are a mutation to only one part of a gene and cause a single change like sickle-cell anaemia does to the shape of red blood cells.



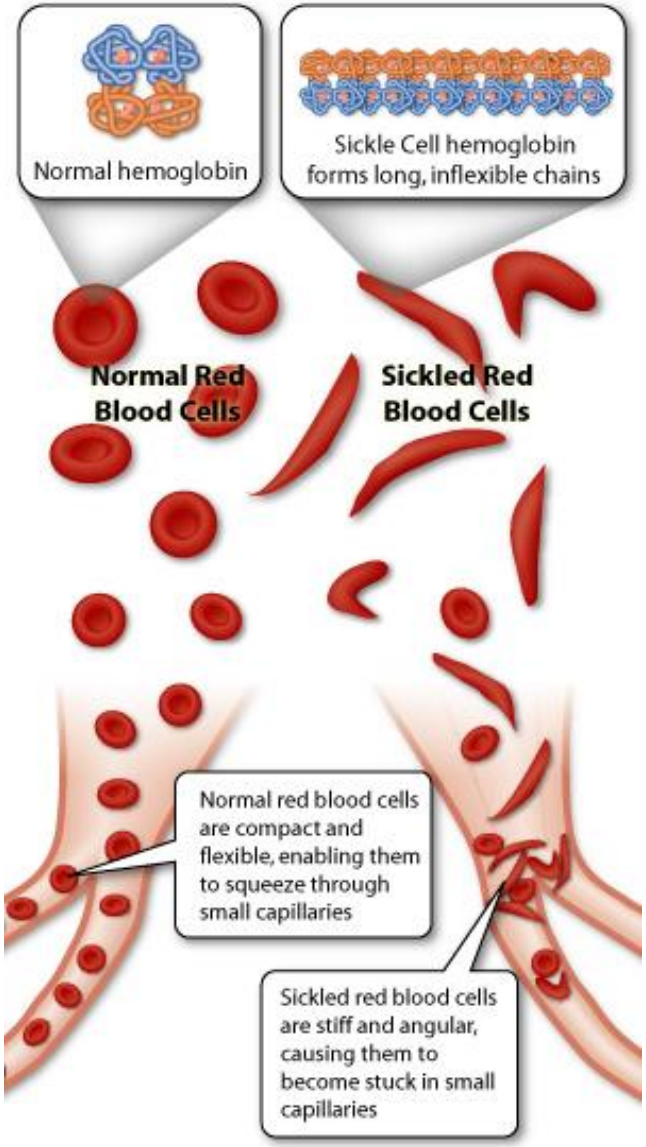
Chromosome mutation

GZ Science Resources

Humans can have specific genetic disorders caused by a mutation during meiosis

extra info

Most disorders are recessive and require both parents to carry a copy of the mutation to show up in the offspring.
Some types of recessive gene disorders confer an advantage in certain conditions when only one copy of the gene is present, such as one copy of sickle-cell anaemia mutation gives resistance against malaria – a common disease in tropical countries.

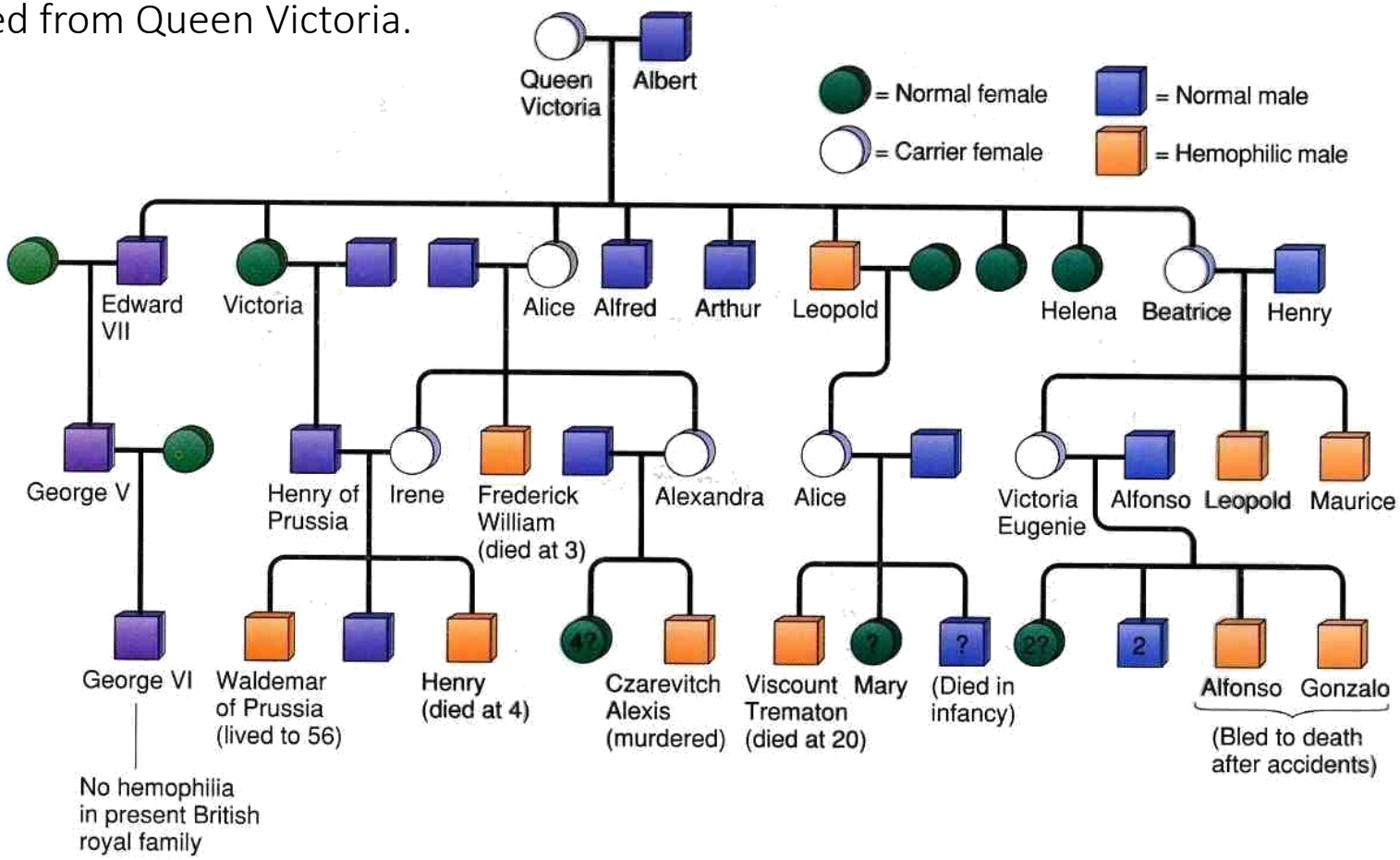


Humans can also have mutations occurring in either their X or Y chromosome (sex chromosomes) and therefore only passed on to Male offspring - if on the Y, or to female offspring – if on the X and is recessive.
If the mutation is recessive and on the X chromosome a male parent will only be a carrier.
This are called sex-linked disorders. Haemophilia is an example and carried on the X chromosomes. Males without an extra X chromosome (a healthy chromosome to "block") show the mutation.

Using Pedigree charts to predict genetic disorders in offspring

extra info

A pedigree chart can be used to predict the likelihood of offspring receiving a mutation from parents as well as look for where the mutation has come from.
A particularly famous pedigree of a mutation in a family comes from the European royal family. The mutation is haemophilia (which prevents clotting of the blood) and can be traced from Queen Victoria.



GZ Science Resources