

AQA Biology GCSE

Topic 6: Inheritance, Variation and Evolution

Notes

(Content in bold is for higher tier only)

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Reproduction

Sexual and Asexual Reproduction (6.1.1)

Meiosis is the formation of four non-identical cells from one cell. Mitosis is the formation of two identical cells from one cell.

- 1. Sexual reproduction involves the joining of male and female gametes, each containing genetic information from the mother or father.
 - Sperm and egg cells in animals
 - Pollen and egg cells in flowering plants

Gametes are formed by meiosis, as they are non identical. A normal cell has 46 chromosomes. There are two sets of chromosomes (i.e. 23 pairs). In each pair, one chromosome is from the father and the second set are from the mother. Each gamete has 23 chromosomes and they fuse in fertilisation. The genetic information from each parent is mixed, producing variation in the offspring.

- Asexual reproduction involves one parent with no gametes joining. It happens using the process of mitosis, where two identical cells are formed from one cell. There is no mixing of genetic information.

It leads to clones, which are genetically identical to each other and the parent. Examples of organisms that reproduce this way are bacteria, some plants and some animals.

Meiosis (6.1.2)

Meiosis is the formation of four non-identical cells from one cell. Cells in the reproductive organs divide by meiosis to form gametes. Gametes only have one copy of each chromosome.

- The cell makes copies of its chromosomes, so it has double the amount of genetic information.
- The cell divides into two cells, each with half the amount of chromosomes (46).
- The cell divides again producing four cells, each with a quarter the amount of chromosomes (23).
- These cells are called gametes and they are all genetically different from each other because the chromosomes are shuffled during the process, resulting in random chromosomes ending up in each of the four cells.

These gametes with 23 chromosomes join at fertilisation to produce a cell with 46 chromosomes, the normal number.

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- This cell divides by mitosis to produce many copies.
- More and more cells are produced, and an embryo forms.
- The cells begin to take on different roles after this stage (differentiation).



Advantages and Disadvantages of Sexual and Asexual Reproduction (6.1.3)

The advantages of sexual reproduction are the disadvantages of asexual reproduction, and vice versa.

Advantages of sexual reproduction	Advantages of asexual reproduction
 Produces variation in offspring. This means that if the environment changes it is likely that an organism in the species will have a characteristic that allows them to survive (called a survival advantage). Although some individuals may die, variation decreases the chance of the whole species becoming extinct. 	Only one parent is needed.
 It allows us to use selective breeding. This type of reproduction mixes the genetic information from two organisms Organisms with different desirable characteristics can be bred to produce offspring with even more desirable characteristics. This speeds up natural selection. An example is to increase food production by breeding two animals with lots of meat. 	Uses less energy and is faster as organisms do not need to find a mate.
	In favorable conditions lots of identical offspring can be produced.

Examples of organisms that use both methods to reproduce are below:

- 1. Malarial parasites: causes malaria, spread by mosquitoes and transferred to humans through a bite
 - They reproduce sexually in the mosquito.
 - They reproduce asexually in the human host (in the liver and blood cells).
- 2. Some fungi: many species can undergo both types of reproduction, releasing spores which land and become new fungi
 - Spores produced asexually are genetically identical.
 - Spores are produced sexually when the conditions change, in order to increase variation and avoid extinction.
- 3. Some plants
 - Many reproduce sexually using pollen, which must reach the egg cells in the female parts of another flower. This is called pollination, and it forms seeds.
 - Strawberry plants reproduce asexually, as they produce runners. New identical plants grow off the runner.
 - Daffodils reproduce asexually. They grow from bulbs. New bulbs can grow from the main one, producing a new identical plant.



• It is advantageous in plants as it means they can reproduce even if the flowers have been destroyed by frost or other animals.

DNA and the Genome (6.1.4)

DNA Structure (6.1.5) (Biology Only)

DNA, found in the nucleus, is a chemical that contains genetic material.

DNA stands for deoxyribonucleic acid, and this is a polymer that contains instructions for the body.

- It is made up of many small parts called nucleotides.
- Each nucleotide is made up of one sugar molecule, one phosphate molecule (which form the backbone) and one of the four types of organic bases.
- The four types of organic bases are A, C, G, T.
- Each DNA molecule is made up of two DNA strands which are twisted together. Each base is connected to another base in the other strand.
- A bases only connect to T bases, and C bases only connect to G bases. This is called complementary base pairing.
- The order of the different bases forms a code- e.g. A, G, T, T, C, A, A etc.
- Each group of three bases codes for an amino acid.
- The amino acids are joined together to make a protein. It is the different types and order of amino acids that determine which type of protein it is.
- Therefore it is the order of bases in DNA that determine which proteins are produced.

DNA is a polymer (long molecule) made up of two strands which are wound around each other to form a structure called a double helix.

Chromosomes are structures made up of long molecules of DNA.

A gene is a short section of DNA. Each gene codes for many amino acids, which are joined together to make a specific protein.

• There are 20 types of amino acid.

There are also non-coding parts of DNA that do not code for proteins. Some of them are responsible for switching genes on or off, i.e. controlling whether the gene is used to form a protein or not.

The word genome describes all the genetic information of that organism. The human genome has been studied, which has improved our understanding of the genes linked to different types of disease, the treatment of inherited disorders and has helped in tracing human migration patterns from the past.

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Protein synthesis: the process of producing a protein from DNA

If a gene is coded to make a protein, it has been expressed.

- 1. DNA contains the genetic code for making a protein, but it cannot move out of the nucleus as it is too big.
- 2. The two strands pull apart from each other, and mRNA nucleotides (messenger RNA: a different type of nucleotide) match to their complementary base on the strand.
- 3. The mRNA nucleotides themselves are then joined together, creating a new strand called the mRNA strand. This is a template of the original DNA.
- 4. The mRNA then moves out of the nucleus to the cytoplasm and onto structures called ribosomes.
- 5. At the ribosomes, the bases on the mRNA are read in threes to code for an amino acid (the first three bases code for one amino acid, the second three bases code for another etc).
- 6. The corresponding amino acids are brought to the ribosomes by carrier molecules.
- 7. These amino acids connect together to form a protein.
- 8. When the chain is complete the protein folds to form a unique 3D structure.

The protein's specific shape helps it to carry out its role. Proteins can be:

- Enzymes biological catalysts that speed up the rate of reaction
- Hormones chemical messengers that send signals around the body
- Structural protein strong proteins in order to form structures, such as collagen

Mutations change the sequences of bases in DNA. Either:

- 1. A base is inserted into the code
 - As they are read in threes, this changes the way it is read.
 - It may change all the amino acids coded for after this insertion.
- 2. A base is deleted from the code
 - Like insertions they change the way it is read.
 - It may change all the amino acids coded for after this deletion.
- 3. A base is substituted
 - This will only change one amino acid in the sequence or it may not change the amino acid (as the new sequence can sometimes still code for the same amino acid)

A change in the type/sequence of amino acids will affect the way it folds and therefore the structure.

Most mutations do not alter the protein or only do so slightly.

Some can have a serious effect and can change the shape

- The substrate will not fit into the active site so it cannot act as a protein.
- A structural protein may lose its shape.

There can also be mutations in the non-coding parts of DNA that control whether the genes are expressed.

We now understand that variation between two organisms arises because of two reasons:

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- 1. The coding DNA that determines the proteins and their activity
- 2. The non-coding DNA that determines which genes are expressed



Genetic Inheritance (6.1.6)

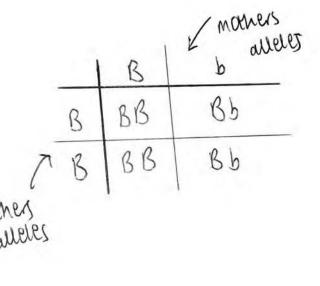
You need to know the definitions for a number of terms.

Gamete	An organism's reproductive cell (egg in female and sperm in males), which has half the number of chromosomes (23).
Chromosome	A structure found in the nucleus which is made up of a long strand of DNA.
Gene	A short section of DNA that codes for a protein, and therefore contribute to a characteristic. Some characteristics are controlled by a single gene, such as fur colour in mice and red-green colour blindness in humans. However, most characteristics are the result of many different genes interacting.
Alleles	The different forms of the gene - humans have two alleles for each gene as they inherit one from each parent.
Dominant allele	Only one (out of the two alleles) is needed for it to be expressed and for the corresponding phenotype to be observed.
Recessive allele	Two copies are needed for it to be expressed and for the corresponding the phenotype to be observed.
Homozygous	When both inherited alleles are the same (i.e. two dominant alleles or two recessive alleles).
Heterozygous	When one of the inherited alleles is dominant and the other is recessive.
Genotype	The combination of alleles an individual has, e.g. Aa
Phenotype	The physical characteristics that are observed in the individual, e.g. eye colour

Family trees show the inheritance of different phenotypes over generations in the same family.

A single gene cross looks at the probability of the offspring of two parents having certain genotypes and phenotypes. This is done using the alleles the two parents have for a gene and a Punnett square diagram. You should be able to draw and use a Punnett square diagram

Uppercase letters are used to represent dominant characteristics. Lowercase letters represent recessive characteristics. You can choose any letter but usually either A or B is used for simplicity.



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Inherited Disorders (6.1.7)

Genetic disorders are caused by inheriting certain alleles.

Polydactyly: having extra fingers or toes

• Caused by a dominant allele

Cystic fibrosis: this is a disorder of the cell membranes, resulting in thick mucus in the airways and pancreas

- Caused by a recessive allele
- Both parents need to either be carriers (have one of the recessive alleles) or one must have CF themselves and the other is a carrier.

Embryonic screening allows scientists to observe whether the child will have a genetic condition or not.

- If the embryo is developed in the lab, cells can be taken from it and analysed
- DNA from embryos in the womb can also be extracted

There are many economic, social and ethical issues surrounding embryonic screening.

Arguments for embryonic screening	Arguments against embryonic screening
Reduces the number of people suffering (ethical).	Could encourage people to pick characteristics - creating designer babies (ethical).
Treating disorders is very expensive (economic).	It is expensive to carry out screening (economic).
There are many regulations in place to stop it getting out of hand (social).	May promote prejudice as it suggests that those with genetic disorders will not live a full life or are unwanted (social).
	Decisions about terminating a pregnancy have to be made (social).
	The procedure can lead to a miscarriage (social).

Sex Determination (6.1.8)

Human body cells have 23 pairs of chromosomes.

- 22 control characteristics, and the chromosomes in each pair look very similar
- The 23rd pair carries sex determining genes, and the two chromosomes can look different to each other (Y chromosomes are much smaller than X chromosomes)

The two possible chromosomes in the 23rd pair are X chromosomes and Y chromosomes. When cells undergo meiosis to form a gamete, one sex chromosome goes into each gamete.

• Females have two X chromosomes, so therefore only pass on X chromosomes in their eggs.



• Males have one X chromosome and one Y chromosome, so therefore can pass on X or Y chromosomes in their sperm.

Punnett squares can be used to show sex inheritance. There is a 50% chance of the child being male, and a 50% chance of the child being female.

Variation and Evolution

Variation (6.2.1)

The phenotype an organism has depends on two things:

- 1. Genotype: the genes it inherited
 - Genes are passed on from the parent in sex cells.
 - The combining of genes from the mother and father creates genetic variation.
 - Only identical twins have the same genotype.
 - There is lots of genetic variation in a population.
- 2. Environment: the place it lives in
 - The conditions the organism grows and develops in also affects its appearance.
 - Examples include scars in animals, or smaller and yellow leaves in plants.

Sometimes characteristics can result from a combination of genetics and the environment, such as weight. Weight depends on the food you eat but also how quickly your body breaks it down and how much it stores as fat depends on your genes.

Genetic variation is introduced by mutations in the sequence of DNA.

- Most have no effect on phenotype.
- Some will influence phenotype but it is unlikely that it will bring about a new phenotype.
- If the mutation does determine the phenotype and it is advantageous, natural selection will mean it becomes the common phenotype relatively quickly.

Evolution (6.2.2)

Evolution is a change in the inherited characteristics of a population over time through a process of natural selection which may result in the formation of a new species.

Theory of Evolution: All species have evolved from simple life forms that first developed more than three billion years ago.

Evolution occurs because of natural selection.

• Mutations occur which provide variation between organisms.

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• If a mutation provides a survival advantage the organism is more likely to survive to breeding age.

• The mutation will then be passed onto offspring.



• Over many generations, the frequency of the mutation will increase within the population.

This may cause one population of a species to become so different that they can no longer interbreed to produce fertile offspring. This means they have become a new species. This is called speciation.

Selective Breeding (6.2.3)

Selective breeding is when humans choose which organisms to breed in order to produce offspring with a certain desirable characteristic (e.g animals with more meat, plants with disease resistance or big flowers).

This has been happening for many years since animals were domesticated and plants were grown for food.

- Parents with desired characteristics are chosen.
- They are bred together.
- From the offspring those with desired characteristics are bred together.
- The process is repeated many times until all the offspring have the desired characteristic.

The problem is that it can lead to inbreeding.

- Breeding those with similar desirable characteristics means it is likely you are breeding closely related individuals.
- This results in the reduction of the gene pool, as the number of different alleles reduce (as they mostly have the same alleles).
- This means if the environment changes or there is a new disease, the species could become extinct as they all have the same genetic make-up (so the chance of a few organisms having a survival advantage and not dying is reduced).
- Another problem is that the small gene pool leads to a greater chance of genetic defects being present in offspring, as recessive characteristics are more likely to present.

Genetic Engineering (6.2.4)

Genetic engineering: Modifying the genome of an organism by introducing a gene from another organism to give a desired characteristic.

- Plant cells have been engineered for disease resistance or to have larger fruits
- Bacterial cells have been engineered to produce substances useful to humans, such as human insulin to treat diabetes.

The process:

- 1. Genes from chromosomes are 'cut out' using restriction enzymes leaving 'sticky ends' (short sections of exposed, unpaired bases)
- 2. A virus or bacterial plasmid is cut using the same restriction enzyme to also create sticky ends. This also contains an antibiotic marker gene.
- 3. The loop and gene sticky ends are then joined together by DNA ligase enzymes
- 4. The combined loop is placed in a vector, such as a bacterial cell, and then allowed to multiply as it will now contain the modified gene. As the bacteria grows we can see

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which ones are resistant to antibiotics. The colonies that are will be the bacteria that are also producing the modified gene, as they were inserted together. In plants the vector is put into meristematic cells (unspecialised cells) which can then produce identical copies of the modified plant.

Genetically modified crops

- They are engineered to be resistant to insects and to herbicides.
- This will result in increased yields as less crops will die.

Genetic modification in medicine

- It may be possible to use genetic engineering to cure inherited disorders.
- It is called gene therapy and involves transferring normal genes (not faulty) into patients so the correct proteins are produced.

Perceived benefits of genetic engineering	Perceived risks of genetic engineering
It can be very useful in medicine to mass produce certain hormones in microorganisms (bacteria and fungi).	 GM crops might have an effect on wild flowers and therefore insects. GM crops are infertile and these genes could spread into wild plants, leading to infertility in other species, which affects the entire environment. Growing with herbicides and pesticides can kill insects and other plants, which would reduce biodiversity.
 In agriculture it can be used to improve yields by: Improving growth rates Introducing modifications that allow the crops to grow in different conditions, e.g. hotter or drier climates Introducing modifications that allow plants to make their own pesticide or herbicide 	People are worried that we do not completely understand the effects of GM crops on human health.
Crops with extra vitamins can be produced in areas where they are difficult to obtain.	Genetic engineering in agriculture could lead to genetic engineering in humans. This may result in people using the technology to have designer babies.
Greater yields can help solve world hunger, which is becoming an increasingly bigger issue due to population growth.	They pose a selection pressure, which could lead to increased resistance in other species, creating super weeds and pests.

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Cloning (6.2.5) (Biology Only)

Cloning is creating genetically identical copies of an organism.

Methods of plant cloning:

- 1. Tissue culture: Important to preserve rare plant species or commercially in plant nurseries.
 - Plant cells are taken
 - They are placed in a growth medium with nutrients and hormones
 - They grow into new plants, and are clones as they are genetically identical to the parent
- 2. Cuttings: An older, easier method to produce clones
 - Cuttings, such as a section of the stem, are taken from a plant with a desirable feature
 - They are planted and produce clones as they are genetically identical to the parent

Cloning in animals:

- 1. Embryo transplants
 - Sperm cells and egg cells from parents with desirable features are obtained.
 - In the lab, they are fertilised to form an embryo.
 - The embryo divides many times and is then inserted into a host mother.
 - The offspring which is eventually born is genetically identical (with the desirable feature) as they have genetic information from the same mother and father.
- 2. Adult cell cloning
 - The nucleus is removed from an unfertilised egg cell.
 - The nucleus is removed from an adult body cell and placed in the denucleated egg cell.
 - Through the stimulation of an electric shock, the egg cell begins to divide to form an embryo.
 - The embryo is implanted into the womb of a female.
 - The offspring born is a clone of the adult body cell.

Benefits of cloning	<u>Risks of cloning</u>
Produces lots of offspring with a specific desirable feature.	The gene pool is reduced through producing clones, meaning it is less likely that the population will survive if a disease arises with low diversity in the population.
The study of clones could help research into embryo development.	Clones have a low survival rate, and tend to have some genetic problems.
Can help extremely endangered species, or even bring back species that have become extinct.	It may lead to human cloning.

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The Development of Understanding of Genetics and Evolution Theory of Evolution (6.3.1) (Biology Only)

Charles Darwin

- Scientist and naturalist
- Put forward the theory of evolution
- This was supported by experimentation and his knowledge of geology and fossils that he discovered on a round the world expedition
- Published 'On the Origin of Species' in 1859

Theory of Evolution:

- Variation exists within species as a result of mutations in DNA
- Organisms with characteristics most suited to the environment are more likely to survive to reproductive age and breed successfully called survival of the fittest.
- The beneficial characteristics are then passed on to the next generation
- Over many generations the frequency of alleles for this advantageous characteristic increase within the population

There was lots of controversy surrounding his ideas for many reasons:

- 1. It contradicted the idea that God was the creator of all species on Earth.
- 2. There was not enough evidence at the time as few studies had been done on how organisms change over time.
- 3. The mechanism of inheritance and variation were not known at the time.

There were other opinions, such as that of Jean-Baptiste Lamarck. He thought that:

- Changes that occurred during the lifetime of an organism were passed onto offspring
- If an individual continually repeated an action, the characteristic that allowed it to do this would develop further
- For example, if a giraffe stretched to reach leaves on a tree high up, its neck would become longer allowing it to do this more easily. This characteristic would then be passed on to its offspring.

Lamarck's theory was proven wrong when it was understood that changes caused by the environment were not passed on in the sex cells.

Darwin's theory was supported by genetics as it provided a mechanism for beneficial characteristics caused by mutations to be passed on. Fossil evidence showed how developments in organisms arose slowly.

Speciation (6.3.2) (Biology Only)

Speciation is the process of a new species developing through the selection of different alleles. This increases the genetic variation until the new population cannot breed with those in the old population to produce fertile offspring.

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Alfred Russel Wallace developed the theory of speciation, and therefore evolution by natural selection.

- On his travels, he had the idea that the individuals who did not have characteristics to help them survive a change in the environment would die out.
- He published joint studies with Darwin.
- The publication of 'On the Origin of Species' meant Darwin received the credit for the theory.
- He continued to work across the world to collect evidence one of his most important works was on warning colouration in animals
- Much more evidence over time has resulted in our current understanding.

The process of speciation:

- 1. Variation exists within a population as a result of genetic mutations.
- 2. Alleles which provide a survival advantage are selected for through natural selection.
- 3. Populations of a species can become isolated, for example through physical barriers such as a rock fall preventing them from breeding together.
- 4. Different alleles may be advantageous in the new environment, leading to them being selected for.
- 5. Over time the selection of different alleles will increase the genetic variation between the two populations.
- 6. When they are no longer able to breed together to produce fertile offspring, a new species has formed.

The Understanding of Genetics (6.3.3) (Biology Only)

Gregor Mendel

- Trained in mathematics and natural history in Vienna
- Worked in the monastery gardens and observed the characteristics passed on to the next generations in plants
- He carried out breeding experiments on pea plants.
- He used smooth peas, wrinkled peas, green peas and yellow peas and observed the offspring to see which characteristics they had inherited
- Through keeping a record of everything he did and eventually publishing his work in 1866, he came to the conclusions that:
 - Offspring have some characteristics that their parents have because they inherit 'hereditary units' from each.
 - One unit is received from each parent.
 - Units can be dominant or recessive, and cannot be mixed together.

Mendel was not recognised till after his death as genes and chromosomes were not yet discovered, so people could not understand.

- In the late 19th century chromosomes as a part of cell division were observed
- In the 20th century, it was understood that chromosomes and units had similar behaviours. It was decided that units (now known as genes) were on the chromosomes.
- The structure of DNA was determined in 1953, which meant we were able to understand how genes worked.

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Evidence for Evolution (6.3.4)

The theory of evolution by natural selection is now widely accepted.

Evidence for evolution is seen in:

- 1. Fossils: the remains of organisms from many years ago, which are found in rocks
- 2. Antibiotic resistance in bacteria: the selection pressure of antibiotics leads to advantageous mutations being selected for in bacteria populations so they are no longer killed when exposed to antibiotics.

Fossils (6.3.5)

Fossils are the remains of organisms from millions of years ago, which are found in rocks.

They are formed from:

- 1. Parts of organisms that have not decayed because oxygen or moisture were not present, meaning that the microbes that cause decay cannot survive.
- 2. Parts of the organism such as teeth, shells and bones are replaced by minerals as they decay, forming a rock structure of the original part.
- 3. Preserved traces such as footprints, burrows and rootlet traces (the plants roots) remain due to the ground hardening around them and forming a cast.

Fossils are used to show how the anatomy of organisms has changed over time. They can be used to compare how closely related two organisms are, through looking at the number of similarities they have. This information is used to create evolutionary trees.

Fossils cannot be used to tell us how life started on Earth because:

- Most early life forms are soft-bodied and therefore decay completely, so there are few fossils of them.
- Any traces left have been destroyed by geological activity.

Extinction (6.3.6)

Extinction: when no individuals of a species are alive

Factors which can contribute to extinction include:

- 1. Changes in environment which the species cannot adapt fast enough to.
- 2. New predators may have evolved or migrated to the area.
- 3. A new disease arises and there are no resistant alleles to it.
- 4. They have to compete with a species which has advantageous mutations for the same food source.
- 5. A catastrophic event can wipe out a species.
- 6. Destruction of habitat

Resistant Bacteria (6.3.7)

Bacteria are labelled resistant when they are not killed by antibiotics which previously were used as cures against them.

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- Bacteria reproduce at a fast rate.
- Mutations during reproduction can result in new genes, such as the gene for antibiotic resistance. This the creation of a new strain.
- Exposure to antibiotics creates a selection pressure, as those with antibiotic resistant genes survive and those without die.
- As a result those with antibiotic resistance can reproduce and pass on the advantageous gene to their offspring.
- This population of antibiotic resistant bacteria increases.
- Bacterial diseases spreads rapidly because people are not immune to these new resistant bacteria and there is no treatment for it.

An example is MRSA.

- Called a 'superbug' as it is resistant to many different types of antibiotics
- Common in hospitals: spreads when doctors and nurses move to different patients

How to slow the development of resistance in bacteria:

- 1. Antibiotics should not be given for viral or non-serious infections
- 2. Specific antibiotics should be given for specific bacteria
- 3. Patients should complete their course of antibiotics if they do not some bacteria may survive and mutate to become antibiotic resistant.
- 4. Antibiotics should be used less in agriculture farmers currently use them to prevent their livestock dying from disease, but this overuse leads to antibiotic resistant bacteria which are then transferred to humans when they consume the meat.

How to slow the transmission of the bacteria:

- 1. Maintain high standards of hygiene in hospitals
- 2. Medical staff and visitors should wash hands regularly
- 3. Medical staff should wear disposable clothing or clothing that is regularly sterilised

As the development of antibiotics is expensive and slow, it is difficult to keep up with the development of resistant strains.

Classification of Living Organisms

Classification involves putting organisms into groups depending on their structure and characteristics.

The Linnaean system

- Carl Linnaeus put this forward in the 1700s
- Living things were divided into kingdoms, phylum, class, order, family, genus, species.

The binomial system gives each organism a name which is used worldwide (overcomes language barriers). The first part is their genus and the second part is their species. An example is the ladybug, which has the name *Harmonia* (genus) *axyridis* (species).

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Three-domain system



- Developments in science such as the improvement of the microscope and increased knowledge of biochemistry (for example, RNA sequence analysis) found that some species were more distantly related than first thought
- Carl Woese added three large groups called domains above kingdoms
 - Archaea: primitive bacteria which live in extreme environments such as hot springs
 - Bacteria: true bacteria (despite having similar features to archaea)
 - Eukaryota: organisms who have a nucleus enclosed in membranes, includes the kingdoms protists, fungi, plants and animals

Evolutionary trees: used to show how closely related organisms are. To complete this they use classification data and fossils for extinct species.

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