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PhotoMaster Format

Year 12 Biology Module 5

Heredity

Topic Outline

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1. Reproduction

- Sexual v. asexual
- External v. internal fetilisation
- Aspects of reproduction in mammals
- Human manipulation of plant & animal reproduction

2. Cell Replication

- Genes, chromosomes & DNA
- Purposes of cell divisions
- Mitosis & meiosis
- DNA structure & replication
- Cell division & continuity of species

3. Polypeptide Synthesis

- Structure & functions of proteins
- What is meant by a "gene"
- Transcription & translation
- m-RNA & t-RNA
- How proteins create the phenotype

4. Genetic Variation

- Mendelian genetics
- Punnett squares & pedigrees
- Sex-Linkage
- Multiple alleles
- Co-Dominance & incomplete dominance
- Effect of environment
- Genetics & evolution

5. Population Genetics

- Gene frequency & the Hardy-Weinberg Principle
- Human Genome Project
- Single-Nucleotide Polymorphisms
- DNA analysis & human evolution

Attention Teachers & Students

This module might NOT cover all the syllabus content as fully as KISS Resources usually do. This is due to time constraints, as explained by a notice at our website.

What is this topic about?

To keep it as simple as possible, (K.I.S.S. Principle) this topic covers:

1. Reproduction

Sexual & asexual reproduction; advantages & disadvantages. Internal & external reproduction. Human reproduction: structures, processes & hormonal control.

Human manipulation of plant & animal reproduction.

2. Cell Replication

Genes, chromosomes & DNA. Purposes of cell divisions. Mitosis & meiosis. DNA structure & replication. Cell division & continuity of species.

3. DNA & Polypeptide Synthesis

Structure & functions of proteins. What constitutes a "gene". Transcription & translation. Roles of m-RNA and t-RNA. How proteins create the gene phenotype.

4. Genetic Variation

Revision of Mendelian genetics. Punnett squares & pedigrees. Sex-linkage & multiple alleles. Co-dominance & incomplete dominance. Effect of environment. Genetics & evolution.

5. Inheritance Patterns in a Population

Gene frequencies. The Hardy-Weinberg Principle. The Human Genome Project. Single-Nucleotide Polymorphisms. How DNA analysis helps us understand human evolution.

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1. Reproduction

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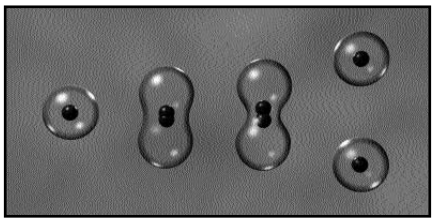
Sexual & Asexual Reproduction

("Sexual" = having male & female sexes. "Asexual"= no sexes.)

All living things reproduce themselves. We are used to the idea that reproduction involves male and female parents who combine their genetic information to produce offspring.

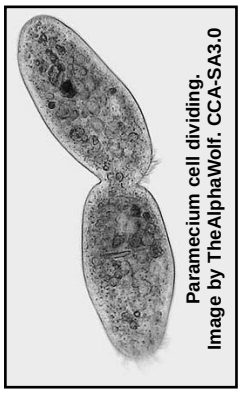
However, many living things do not need male and female parents to reproduce.

Asexual Reproduction



In Unicellular Life

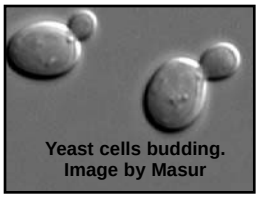
Single-celled organisms such as bacteria reproduce by simply dividing in two by mitosis. (revised later) The offspring cells are genetically identical to each other, and to the "parent cell".



Paramecium cell dividing. Image by TheAlphaWolf. CCA-SA3.0

Among the single-celled, eukaryotic protists such as Amoeba & Paramecium species binary fission (splitting in two) is also common, but is often more complex than simple mitosis division.

In single-celled fungi (yeasts) a process called "budding" is very common. This is a form of binary fission in which a new cell is formed as a small "bud" growing on the parent cell. It separates as a new cell and grows to full size. Each budding cycle doubles the population, so a few cells can become millions very quickly.



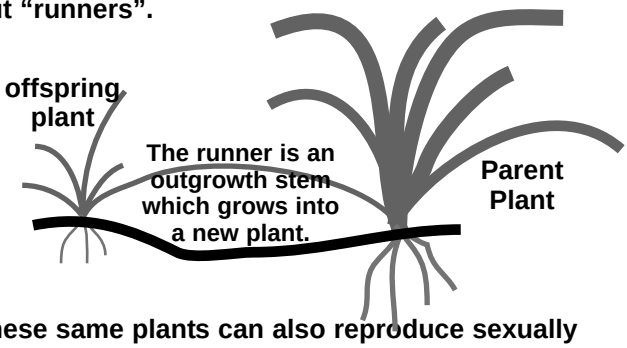
Yeast cells budding. Image by Masur

In Multicellular Life

Many multi-cellular organisms are also able to reproduce asexually.

Fungi, such as mushrooms, reproduce by releasing "spores". Each spore is a single cell which can grow into a new fungus. The spore cells are produced by mitosis, and released from a single "parent".

Many Plants can reproduce asexually by sending out "runners".



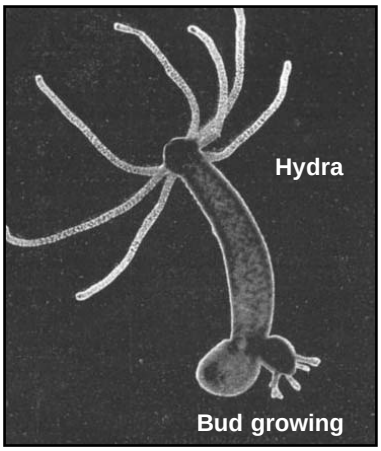
These same plants can also reproduce sexually with their flowers.

Even some animals can reproduce asexually. Perhaps the best-known example is the small aquatic animal Hydra. This is a relative of jellyfish & coral animals.

Hydra can reproduce sexually by releasing eggs or sperm into the water, but can also reproduce asexually by a "budding" process.

A small out-growth appears on its body and grows into a new little hydra.

Eventually this "baby" separates from the parent to live freely as a separate individual.



Regardless of the details, asexual reproduction:

- **requires only one parent.**
- **involves mitosis cell division.**
- **produces offspring which are genetically identical to the parent and to each other.**

Advantage

Asexual reproduction can produce large numbers of offspring quickly, to take advantage of a sudden or temporary increase in some environmental resource such as food.

Disadvantage

By producing genetically identical offspring, there is less variation in the population. If an environmental change occurs, a low-variation species is at risk of extinction.

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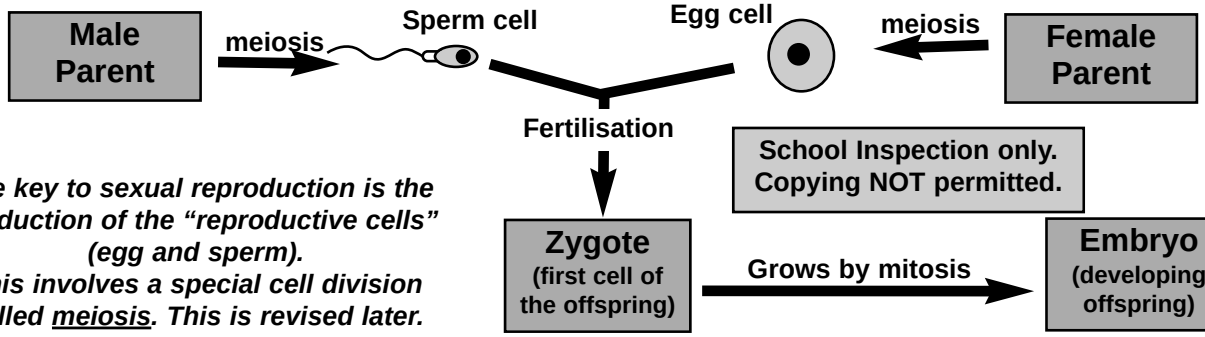
Sexual Reproduction

Sexual reproduction always involves 2 parents who combine part of their genetic information to produce offspring which are different to both parents.



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The key to sexual reproduction is the production of the "reproductive cells" (egg and sperm). This involves a special cell division called meiosis. This is revised later.

Advantage

Sexual reproduction produces more variation in a population, by mixing genes in new combinations. This helps a species survive when environments change.

Disadvantage

Sexual reproduction is more complex, and often takes more time and energy to achieve.

External & Internal Fertilisation

Sexual reproduction always involves the process of fertilisation... when egg and sperm fuse together forming one new cell (the "zygote") which contains genetic information from both parents. For fertilisation to occur, the sperm cells must swim to the egg.

External Fertilisation

For organisms that live in water, fertilisation is generally achieved by both parents simply releasing eggs and sperm into the water environment. Since fertilisation occurs outside the organisms' bodies, this is external fertilisation.

Each species may have some strategy to ensure that male and female parents release their gametes at the same time and in the same place:

Most fish species have "mating rituals" and visual signals which stimulate a mating pair to release gametes together.

Organisms such as corals and sponges release gametes when a certain "trigger" occurs, such as a full Moon, or an especially high tide.

Either way, external fertilisation is to some extent a "hit-and-miss" strategy, often involving millions of gametes, many of which may be wasted.

Internal Fertilisation

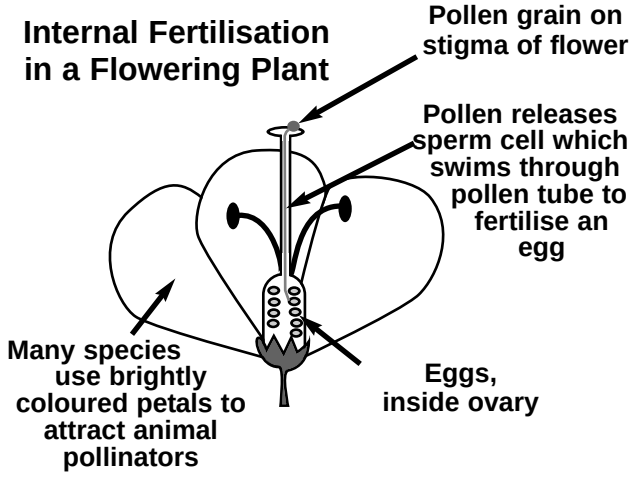
For organisms which live on land, an unprotected egg or sperm cell would rapidly dry out and die. Also, the sperm cells cannot "swim" through the air, or across the ground surface... they need water to swim through. To solve these problems, terrestrial organisms use internal fertilisation.

Terrestrial Plants

• produce their male gametes wrapped in a protective capsule to prevent drying... a pollen grain.

• use either the wind, or animal pollinators (e.g. bees) to carry the pollen to a flower.

• the pollen grain then releases its sperm cell into a fluid-filled tube (the pollen tube). The sperm can swim down to reach the egg, inside the ovary of the flower.



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Internal Fertilisation in Terrestrial Animals

The male uses his penis (or similar structure) to deposit sperm inside the female's reproductive tract. Sperm cells are never exposed to the drying outside environment. The female system is lined with tissue with a film of moisture always present, so the sperm cells can swim to find and fertilise the egg(s) inside the female's body.

There are a number of strategies for development of the zygote after fertilisation. Here are just a few:

Birds & Most Fish & Reptiles

The female lays eggs and the embryos develop outside her body and hatch from the eggs.

Some Fish & Reptiles

The female keeps the fertilised eggs within her body. When they hatch, the babies emerge from her body vent as if being born.

Marsupial Mammals

After a very short gestation, the foetus is born and crawls into a pouch. It feeds on milk, while developing fully in the pouch.

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Placental Mammals

The foetus develops for a relatively long time inside the female's body, nourished via the placenta. When fully developed, the baby is "born".

How Fertilisation Method Relates to Habitat

The great success of sexual reproduction is that it greatly increases the amount of variation in a species. This gives Natural Selection more opportunity when the environment changes, and more chance for species survival and evolution. The big problem with sexual reproduction is achieving fertilisation.

The Evolution of Sex

It is thought that sexual reproduction was "invented" by living things at least 1 billion years ago, in the aquatic environment. The process evolved in a watery environment where the cells could not dry out, and where one gamete cell (defined as "male") could actively swim to seek out the other gamete. The result is that external fertilisation is totally suited to the aquatic environment where it first evolved.

The first land plants to evolve were mosses and later ferns. To this day, both these types rely on very moist conditions for the sexual stage of their complicated reproductive cycles. Both types are confined to relatively wet habitats, or to places where there is a "wet season" during which their male gametes can swim to find the eggs.

The first land vertebrates were the amphibians. They never really mastered the terrestrial environment and 300 million years later, their descendants still return to water to breed so that their external fertilisation will work.

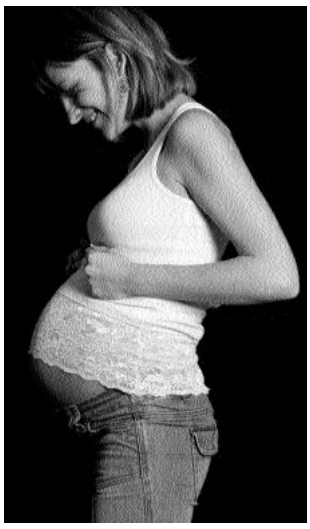
The true colonisation of the terrestrial environment came only when internal fertilisation was first invented:

- **in plants by the cone-bearing "conifers"**
- **in animals by the reptiles, and later birds and mammals.**

Now complete Worksheets 1 & 2

Internal fertilisation is an adaptation to the terrestrial environment

Human Reproduction



Like all placental mammals, humans reproduce sexually and rely on meiosis cell division to make gamete cells (egg & sperm) with half the chromosome number.

Fertilisation occurs inside the female and the foetus develops in the mother's womb, supplied with food and oxygen through the placenta. When fully developed, the baby is born and fed on milk produced by its mother.

The male reproductive system is relatively simple and is really nothing more than a sperm delivery system.

In contrast, the female system is much more complex, since it must be able to produce eggs, support the pregnancy and feed the foetus.

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Male Reproductive System
Structure and Function

The testes (sing: testis) are made of long tubes coiled into balls. The cells in the walls of the tubes carry out meiosis and make millions of sperm cells.

The testes hang outside the body in a pouch called the scrotum. This allows the testes to be kept at a slightly lower temperature. This is important to produce healthy sperm.

The penis is filled with "erectile tissue". This can fill with blood to cause the penis to become hard and erect.

Sperm cells move from the testes to the penis through a tube called the sperm duct. Along the way, fluids are added from several glands. The fluid nourishes the active sperm cells and keep them healthy. This fluid with sperm cells in it is called semen.

During sexual intercourse, semen is ejaculated from the urethra by waves of muscular contractions. Typically, only a few millilitres of semen is released, but it may contain about 200 million sperm cells.

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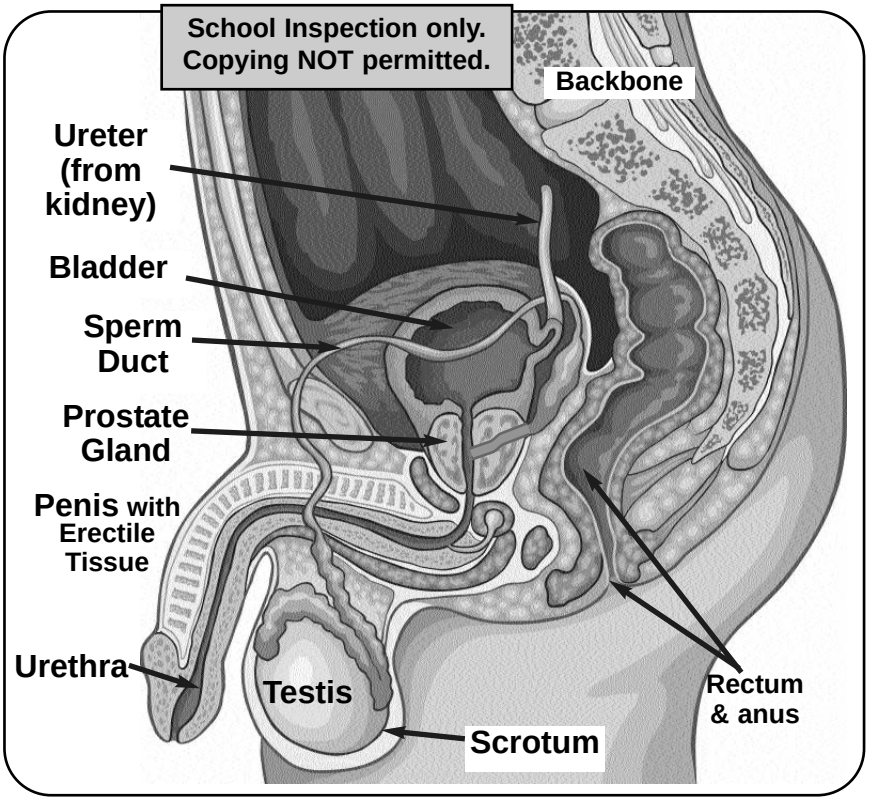
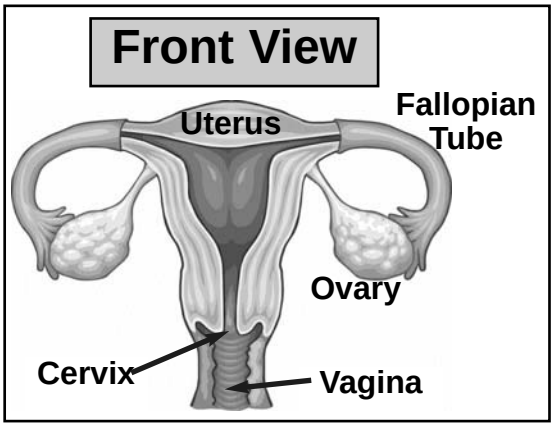


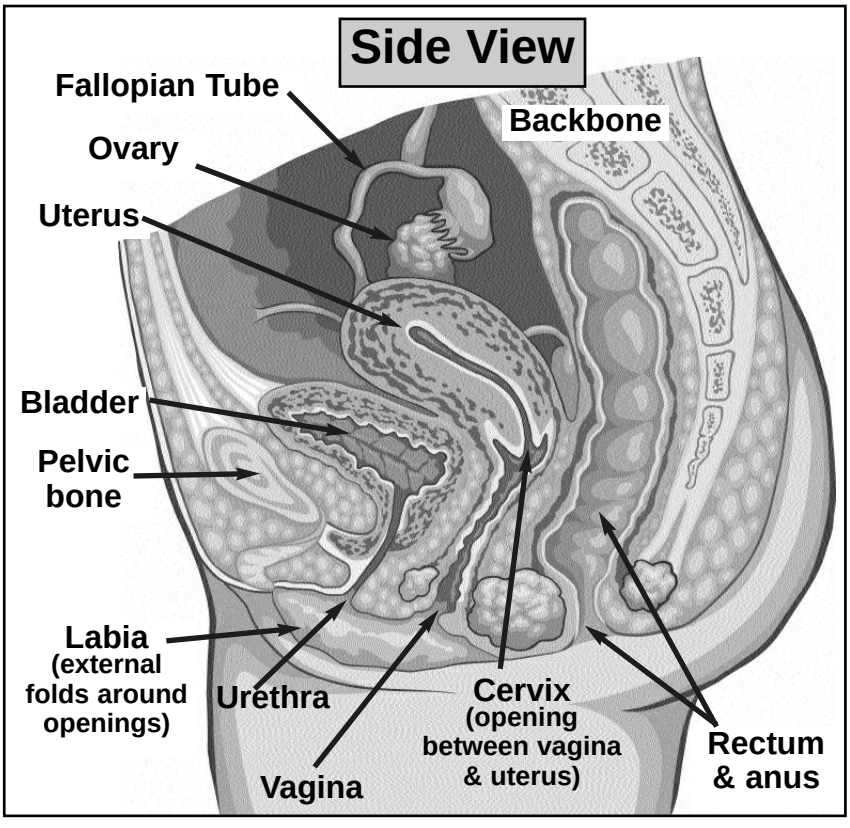
Female Reproductive System



Before a girl is even born, meiosis has occurred in her ovaries and 50,000 immature eggs are present.

After puberty, one egg per month matures and is released into a fallopian tube.

Male sperm cells may swim from the vagina through the cervix, uterus & fallopian tube and fertilise the egg. The zygote (fertilised egg) begins to divide by mitosis and becomes an embryo. Several days after fertilisation, the embryo reaches the uterus.



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Pregnancy & Birth

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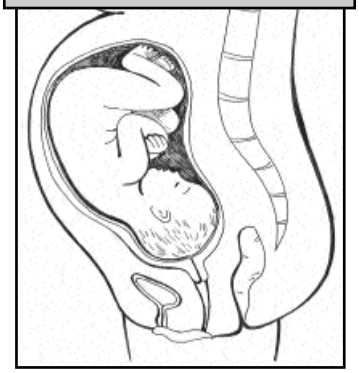


The embryo implants itself into the wall of the uterus and begins to get food and oxygen from the rich blood supply. Gradually a special structure called the placenta grows in the uterus.

The placenta allows exchange of food, oxygen and wastes between the blood of the mother and the developing foetus. Your belly button is where the umbilical chord once connected you to the placenta.

The foetus is enclosed within a flexible bag (the amnion) which is filled with amnionic fluid. This supports the foetus and acts as a "shock absorber".

After about 270 days (9 months) the foetus is fully developed and ready to be born. The birth process is set off by a hormone released from a gland near the brain. The hormone is a chemical which causes the cervix to "dilate" (open wider). The amnion bursts and the amniotic fluid seeps out.

Meanwhile, the hormone causes periodic contractions of the tummy muscles. These get stronger and more frequent until they expel the baby through the cervix and vagina. Later, the contractions expel the placenta as the "after-birth".

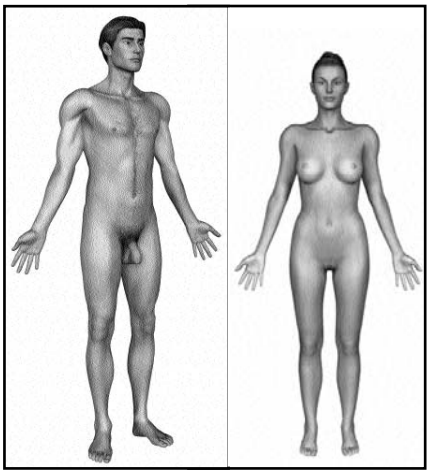
Hormones Control Reproduction

The Endocrine System of hormones controls a number of things from growth, to blood sugar levels to metabolic rate. However, no other body system is so thoroughly controlled by hormones as is the Reproductive System.

Puberty

Except for their external genitals, a little boy or a little girl have exactly the same body shape and pitch of voice.

At puberty this changes dramatically. Hormones from the pituitary gland set off the production of "sex hormones" in the reproductive organs.



From the testes, the male hormone testosterone causes growth changes which deepen the voice, cause facial and body hair to grow and allow for heavier muscle growth.

From the ovaries, the female hormone oestrogen causes development of breasts and changes to the shape of the hips to allow for later child birth.

Pregnancy & Birth

During pregnancy, hormones produced by the placenta suppress any further egg production and maintain the state of the uterus. Hormones cause enlargement of the breasts in preparation for milk production.

The birth process is also set off by a hormone, already described.

Lactation (milk production)

After the baby is born, yet another hormone is produced from the pituitary. This hormone causes the breast tissues to make milk to feed the baby.

Menstrual Cycle

The monthly cycle of egg production and menstrual bleeding is a complex process which is completely controlled by hormones... next page.

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The Menstrual Cycle

The Build-Up

Increasing levels of a pituitary hormone called FSH cause increased release of oestrogen and another hormone called LH. These cause one of the immature eggs in an ovary (the ovaries usually take turns) to begin to mature inside a cyst-like bubble called a follicle.

Approximately 10-14 days into the cycle, the follicle bursts open and releases the egg. (Some women can feel this happen.) The egg now moves slowly along the fallopian tube. The woman is now "fertile" and can become pregnant anytime over the next 3-5 days.

The remains of the egg follicle in the ovary now produces yet another hormone:

Progesterone Hormone

The shattered remnant of the follicle is not finished yet! It pumps out a hormone called progesterone.

Progesterone causes the lining of the uterus to thicken and grow more blood vessels to supply a possible embryo. It also causes changes in the breasts to prepare for possible milk production.

The Break-Down

About 10 days after ovulation (egg release) the follicle remnant finally dies and progesterone suddenly shuts off. The lining of the uterus breaks apart and sloughs away as the menstrual "period". This continues for 3-4 days until FSH production kicks back in and the cycle begins over again.

Please complete Worksheets 3, 4, 5

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Manipulation of Plant & Animal Reproduction

Selective Breeding

Humans have been controlling the reproduction of other species for thousands of years. Early farmers would select the best ram (more wool, more meat, more docile) to mate with the flock, or select seeds from the best corn plants to sow for next years' crop. This has increased yield and quality of plant & animal foods.

Selective breeding has produced all the different varieties of domestic dogs, all the colours of rose flowers, different breeds of cows, sheep, etc. and all of our commercial grains, fruits and vegetables.

The way that the wheat plant has been changed by selective breeding is a good example of <u>why</u> it is done.

Some characteristics of:

<u>Ancestral Wheat</u>	**<u>Modern Wheat</u>**
Few, small seed kernels.	**Many, large seed kernels.**
Seeds are shed when mature.	**Seeds remain attached allowing harvesting.**
Stem bends over easily.	**Stem stronger, remains upright. (easier harvesting)**
Shows many variations in disease resistance, growth rates, etc.	**Selected for uniformity in high disease resistance, fast growth rate, etc.**

Modern wheat is very different to its wild ancestor. Its <u>genome</u> (complete genetic make-up) has been changed almost beyond recognition to give a high yield of food, it is disease resistant and drought tolerant, and has many characteristics to improve its convenience of sowing, harvesting, and so on. Unfortunately, the species now has much less genetic variation, and (in an evolutionary sense of species survival) it is vulnerable to extinction if humans suddenly disappeared. Without humans to sow & harvest it the modern wheat species would probably not survive in the wild for very long.

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Manipulation of Plant & Animal Reproduction (cont.)

Cloning

A "clone" is a group of organisms which are all genetically identical. The simplest form of cloning is asexual reproduction, which has been done artificially with plants since ancient times.

Every time a plant grows from a runner or from a cutting, or by grafting, a clone is being created. Farmers and gardeners have been doing this for thousands of years.

Horticulturalists have always used "cuttings" and plant grafts to make many copies of desirable plants. In more recent times the process was made more efficient with techniques such as "tissue culture" in which many thousands of identical plants can be grown from one parent, by first culturing plant tissue in a laboratory.

Cloning of animals is much more difficult and the first mammal clone was not achieved until the 1990's with the famous "Dolly" the sheep.

Theoretically, we now have the technology to make a whole organism clone of any mammal, including ourselves. This has opened a "can of worms" ethically and morally, prompting most nations (including Australia) to place a ban on human cloning.

Tissue Culture

The process of "Tissue Culture" has allowed plant cloning on a massive scale. This involves taking thousands of small pieces of tissue (each perhaps just a few cells) from a "parent" plant and culturing them in a nutrient liquid or jelly in a test tube or flask in the laboratory.

Plant tissue cultures in laboratory.
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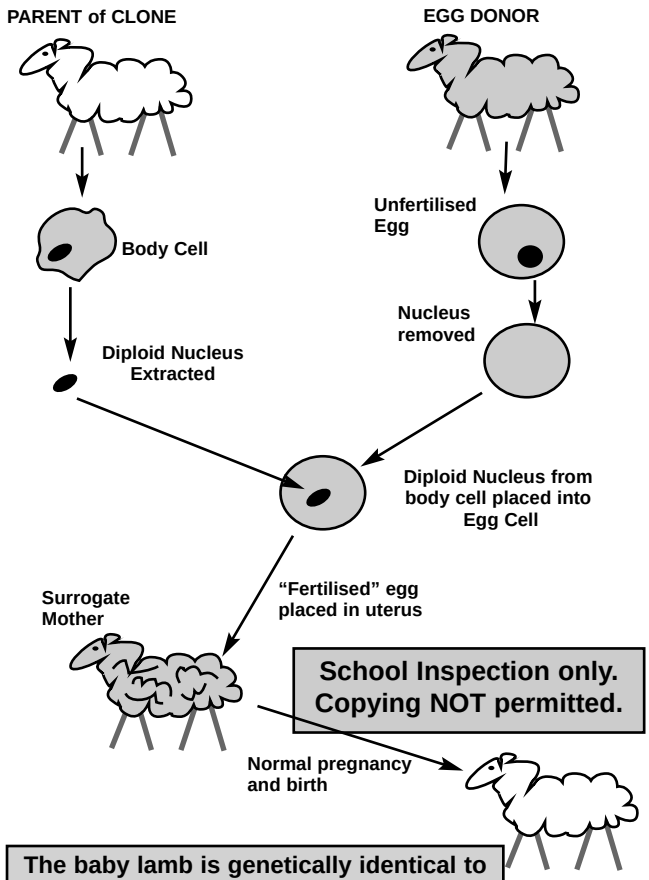
This requires great care & sterile conditions because tissues are vulnerable to disease & decay.

The growth medium contains water, mineral nutients & a few vitamins. The amount of light & the temperature are carefully controlled. Growth is promoted by adding plant hormones. When large enough, other hormones stimulate differentiation of root & shoot tissues.

Eventually, each cultured plant develops enough to be planted out into soil to grow to be an "adult" plant. This technique allows a clone of many thousands to be grown from just one parent plant.

HOW TO CLONE A MAMMAL



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The baby lamb is genetically identical to the single parent at top left. It is an "identical twin", except younger

Why Do This?

The purposes of using tissue culture (or any other cloning technique) are:

• to produce many exact copies of a plant that has some highly desirable characteristic; particularly good flowers or fruit, disease resistance, particular growth habit such as dwarfism, etc.

• to produce many valuable plants of a species which has a very low rate of seed production and/or germination. eg orchids.

• to quickly make copies of a very rare & endangered species as a way to conserve its existence. eg Wollemi Pine.

• to produce a "clean" disease-free population of a plant in which a virus or parasite is endemic in virtually every individual in the wild. This can be to conserve an endangered species, or for purely commercial reasons.

• to quickly increase the population of a genetically-modified species prior to growing enough to produce seed for commercial farming.

• for scientific research on new hybrid varieties, or to search for new plant products for pharmaceutical uses, etc.

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Cloning & Genetic Diversity

Cloning creates genetically identical offspring. If used on a large scale, the result is many offspring being produced from just a few, or even just one, parent. This means that all these offspring are closely related to each other and have less genetic diversity.

Already, many of our food crops, such as wheat and rice, are "monocultures" of millions of individual plants who are "siblings" from relatively few parents. In a banana plantation, the entire population may be a clone of genetically identical plants grown from cuttings and "suckers" (asexual runners) from a single parent.

There are many benefits to this, (eg improved yields of food, consistent high quality) but there may be serious consequences as well. Remember that genetic diversity ("variation") is essential for the long-term survival and evolution of a species. If a species has little variation, then any change in the environment (eg a new disease, climate change) might adversely affect ALL the population, and leave no survivors.

The Irish Potato Famine

In Ireland in the 19th century, many thousands of poor families lived by subsistence farming on fields barely large enough to grow food for a family.

They relied totally on potatoes, the only crop which grew well in the climate and produced enough food to live on.

Almost every field was planted with the same variety of high-yield potato. The plants were grown from "seed-potatoes" saved from the best plants of the previous year's crop. This was asexual reproduction, so there was little genetic variety, and everyone was growing the same crop!

In 1847, a fungal disease struck. Its spores spread on the breeze and it destroyed a crop field within days of infection. This "Potato Blight" totally destroyed the crops for 3 successive years until different, resistant varieties were slowly introduced.

By then, an estimated 1 million people had starved to death. Millions more fled Ireland, settling in USA, Canada and Australia.

Now complete Worksheet 6

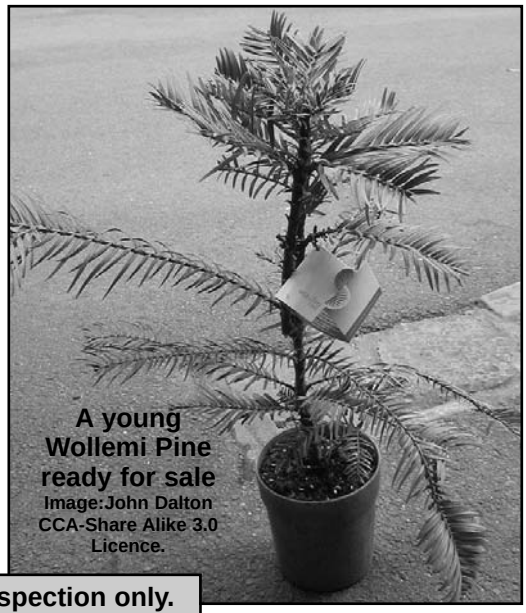
Case Study: Saving the Wollemi Pine

The Wollemi Pine is listed as critically endangered & is legally protected in Australia. Less than 100 specimens exist in the wild. This tree is not technically a pine, but a related type of conifer which can grow to about 40m in height.

The tree (or its closely related ancestor-cousins) was known only from 200 million year-old fossils until 1994. Then a bush-walking National Parks officer accidentally discovered a small population of them in the Wollemi National Park north west of Sydney. Its official scientific name *Wollemia nobilis* honours both the place of discovery & its discoverer, David Noble.

The exact site of this fragile remnant population remains a secret to protect the survivors. The species is variously described as a "living fossil" & as a "dinosaur tree".

Tissue samples of the population have shown that there is virtually no genetic variation within the population & most trees are infected by a virulent fungal disease. It is almost certain that, without help, the species would soon be extinct.



A young Wollemi Pine ready for sale
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Preservation Efforts

Soon after its discovery, a plan for preservation of the Wollemi Pine began. Tissue culture soon produced many young trees. They are commercially available as potted "Xmas trees". Many local council areas have at least one growing in a botanical garden (usually in a metal cage to prevent theft). Many trees are growing in USA, Japan & UK where the trees are thriving in cooler climates than their natural home.

Tissue culture has allowed the population to expand hugely AND has freed the cultured specimens from the fungal disease which threatens the wild survivors. However, the low genetic diversity remains a long-term threat to the species. This is a challenge facing future scientists.

The Australian Botanic Garden at Mount Annan, NSW, has a stand of about 60 plants. The "SeedBank" section of the Botanic Gardens is heavily involved in tissue culture & seed production to save the species.

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2. Cell Replication

Genes, Chromosomes & DNA

We begin with a quick revision of some basic ideas.

What is a "Gene"?

A gene is a unit of inheritance. What colour eyes you have is determined by which "eye-colour genes" you inherited from your parents. Whether your hair is naturally straight, wavy or curly depends on the genes you inherited.

In some plants, the colour of the flowers depends on the genes inherited from its parents. In flies there is a gene which causes "hairy body" and another gene for "hairless". Other genes control wing shape and eye colour, etc.

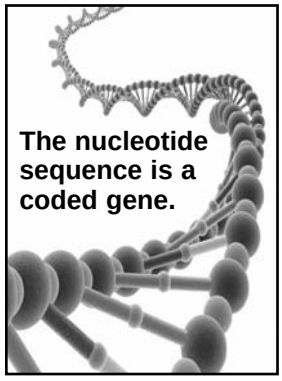
In some cases the situation is much more complicated. Human height is determined by dozens of genes as well as by childhood health and nutrition.

However, to keep it simple (K.I.S.S.) the following principle is often true.

one gene* → *one characteristic

Genes & DNA

DNA is a chemical. Its molecules are the largest known; 1 molecule of DNA may contain millions of atoms bonded in a precise, helix-shaped arrangement.



The nucleotide sequence is a coded gene.

The sequence of "nucleotides" along the DNA molecule is a chemical code.

This tells the cell how to build particular proteins and structures, or how to develop in a particular way. Each gene is specified by the code in a different DNA molecule.

DNA molecule = a gene

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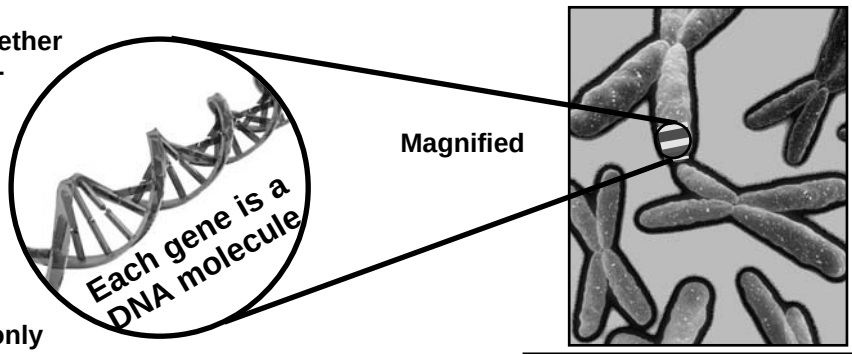
Whether your hair is straight or curly is due to just a slight difference in the "code" sequence of a DNA molecule in the nucleus of your cells.

Chromosomes in Eukaryotic Cells

The DNA molecules which are your genes are not just rattling around loose in the cell nucleus.

Thousands of genes are wrapped up together with protective proteins to form a thread-like structure called a chromosome. Many are roughly "X-shaped" as in the diagram.

Chromosomes are only visible (by microscope) during cell division.



Each chromosome may have 1000's of genes.

In a human body cell there are 46 chromosomes. A sperm or egg cell has only half that number. Chromosomes come in matching pairs. The first 22 pairs are the same size and shape in every human. Other species have different numbers of chromosomes, but always in pairs.

The 23rd pair are different in males & females. This pair of chromosomes are the "sex chromosomes" and determine if you are male or female. More on that later...

Chromosomes in Prokaryotic Cells

In the Bacteria and Archaea, there is usually only one chromosome, called a "genophore". Rather than being rod-shaped or X-shaped, it forms a closed loop which is then "super-coiled" and tethered to the inside of the cell membrane when not actually involved in cell division.

During cell division, the genophore is unravelled from its super-coiled arrangement so that the DNA genes it contains become accessible to the cellular processes involved in duplicating and "reading" the genetic information.

Most prokaryotic cells also contain "plasmids". These are very small loops of DNA, containing only a few genes, which are floating free in the cell cytoplasm.

Plasmids replicate themselves independently and can be transferred from one bacterial cell to another when the cells join together in "conjugation". Plasmid transfer is involved in the rapid spreading of (for example) antibiotic resistance among disease "germs". This "gene sharing" or "horizontal gene transfer" is (sort of) a primitive form of sex.

From here on we will only deal with chromosomes in eukaryotic organisms

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Chromosomes & Cell Divisions

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Purposes of Mitosis

• In unicellular organisms, mitosis is the main method of reproduction by "binary fission".

• In multicellular organisms, mitosis is used mainly for growth and repair.

Remember that individual cells cannot grow large because of SA/Vol ratio limitations. So, the only way to grow larger is to produce many small cells.

Mitosis is also used to replace damaged or worn out cells in the body. For example, your body is constantly producing new blood cells to replace those that wear out.

Purpose of Meiosis

Meiosis is the cell division for sexual reproduction. If 2 parents are to combine their genes in their offspring, it is essential that they firstly halve the genetic information in their reproductive cells ("gametes") so that when fertilisation occurs the offspring receive the correct amount.

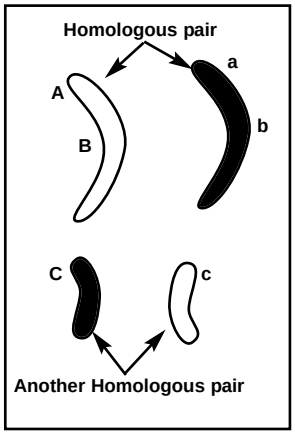
Chromosomes & Genes

• Chromosomes have genes along their length. There may be 1000's of genes on one chromosome.

• Chromosomes occur in pairs. Chromosomes in a pair are called "homologous". Homologous chromosomes correspond with each other by carrying allelic genes in corresponding locations.

Consider 3 sets of genes. Genotype is AaBbCc.
(heterozygous for each characteristic)

This is how the genes could be located on some chromosomes.



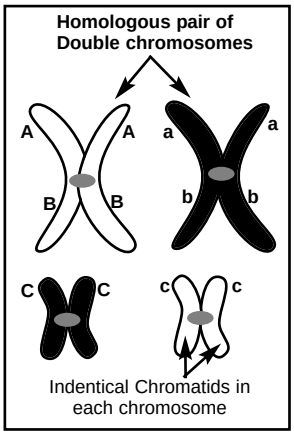
Notice that homologous pairs correspond with each other, but are NOT identical.

• Before cell division, each chromosome is duplicated.

The original and its exact copy remain attached to each other, as a double chromosome. Each part of a double chromosome is called a "chromatid".

This is why they often appear X-shaped. Each chromosome has 2 chromatids and they are often joined in the middle.

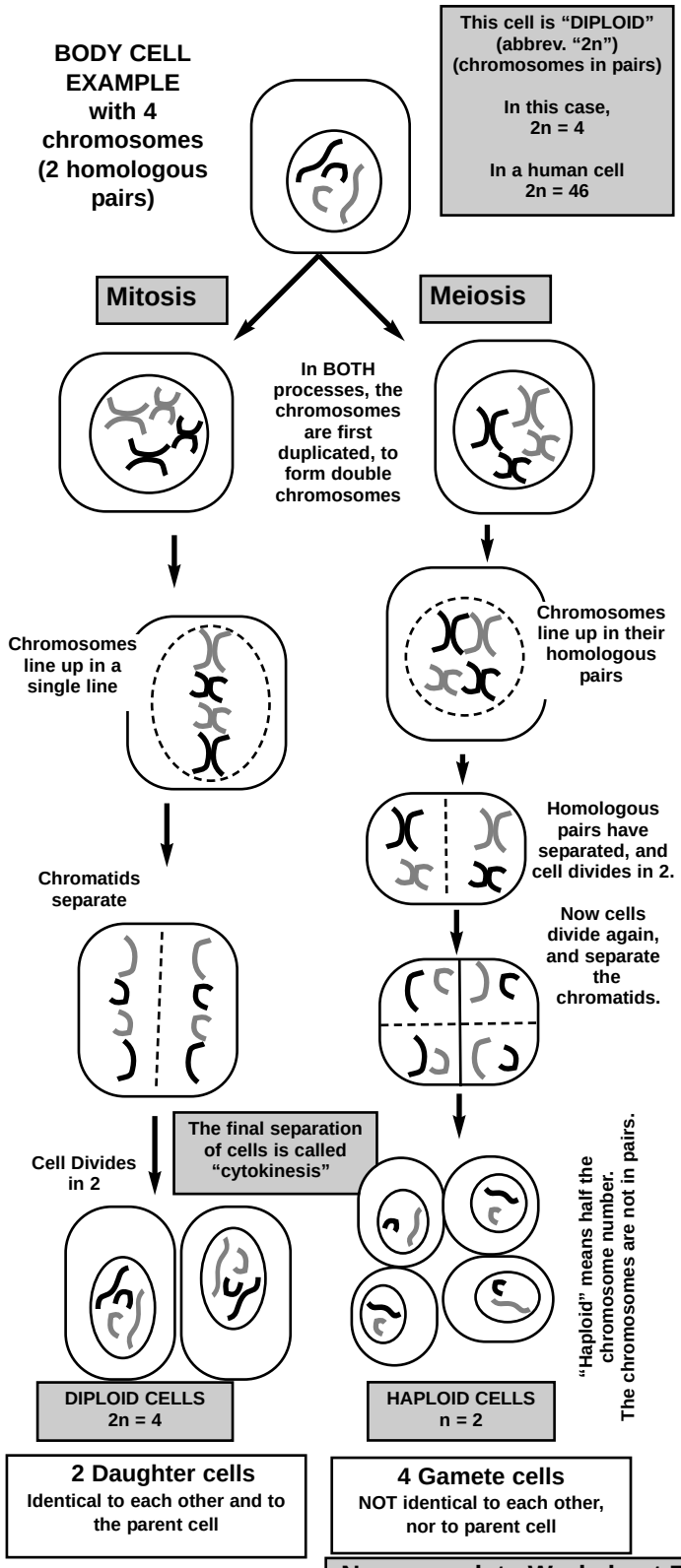
The chromatids in a double chromosome are identical... (look at the genes in the diagram) but the homologous partners are not identical... merely corresponding.



Mitosis & Meiosis

You should already be familiar with the difference between these cell divisions in terms of their outcomes.

Now look more carefully at what happens to the chromosomes during each process.



Now complete Worksheet 7

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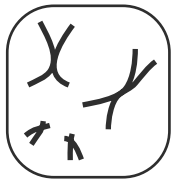
Mitosis Through the Microscope

You may have done a laboratory practical in which you used a microscope to examine a prepared slide of cells undergoing cell division. Commonly the root tip of a plant seedling is viewed.

Most cells look like this one. No chromosomes visible, but DNA is being copied ready for mitosis to start.

Chromosomes just becoming visible. Mitosis is under way.

These 3 diagrams (below right) show how mitosis is often drawn in textbooks, in a stylised way. If you need to sketch them, do it like this for greater clarity.



Chromosomes become visible after copying.

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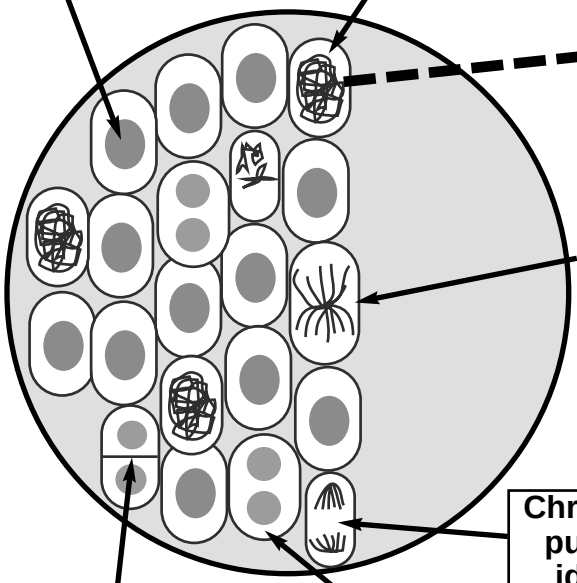
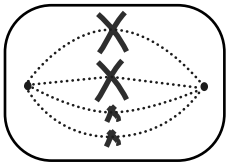
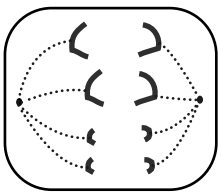


Chromosomes have lined up in the middle of the cell.



Chromosomes lined up in middle of cell.

Chromosomes being pulled apart into 2 identical groups.

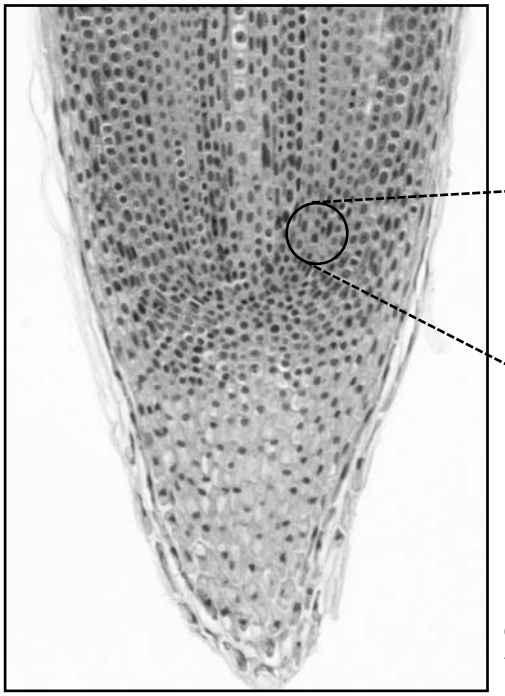


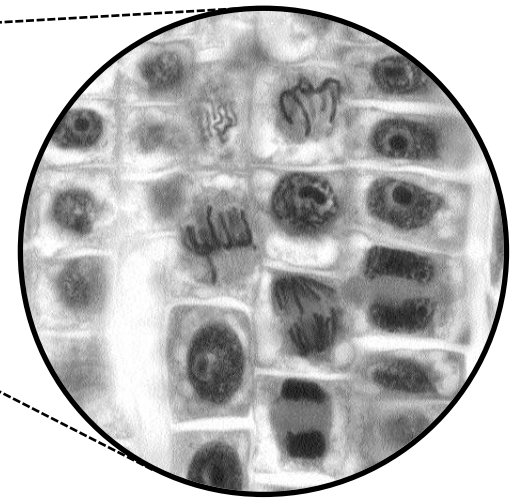
Chromosomes being pulled apart.

These have just finished Cytokinesis.

These have just finished dividing the nucleus.

The colours you may see though a microscope, are due to dyes used to give greater contrast.





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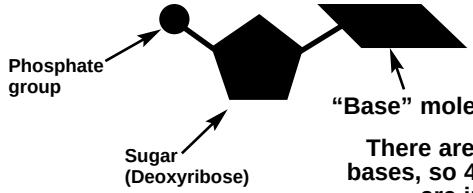
DNA Structure & Replication

The Structure of DNA

Like many biological molecules, DNA is a <u>polymer</u>, made of many smaller units which are joined in long chains. The basic unit of DNA is a "Nucleotide". (named for nucleus)

A NUCLEOTIDE



There are 4 different bases, so 4 nucleotides are in DNA

The 4 different bases are usually known just by the first letter of each name:

A	Adenine
C	Cytosine
G	Guanine
T	Thymine

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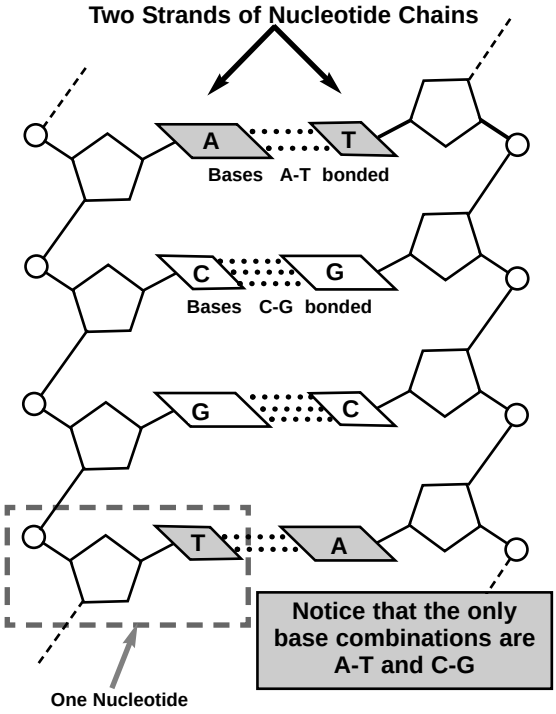
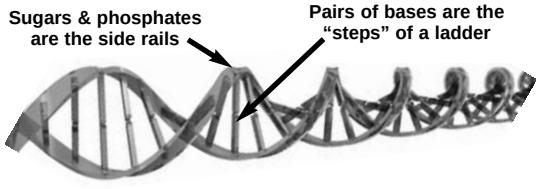
DNA molecules are composed of 2 strands of nucleotides (one running "upside-down" compared to the other) which are joined by the bonding between "base" molecules.



Notice that the only base combinations are A-T and C-G

Finally, the entire molecule is corkscrewed into a "double helix", rather like a spiral staircase or twisted ladder.



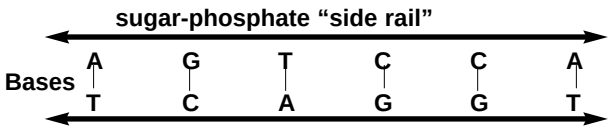
DNA Replication

So how does the structure of the DNA molecule lend itself to replication?

The key is the way the <u>complementary bases</u> bond together in the double stranded structure.

This means that if you have ONE STRAND of a DNA molecule it is a "mirror-image" template for the other. If you split a DNA molecule into 2 separate strands, each strand can be used to build a new, complementary strand.

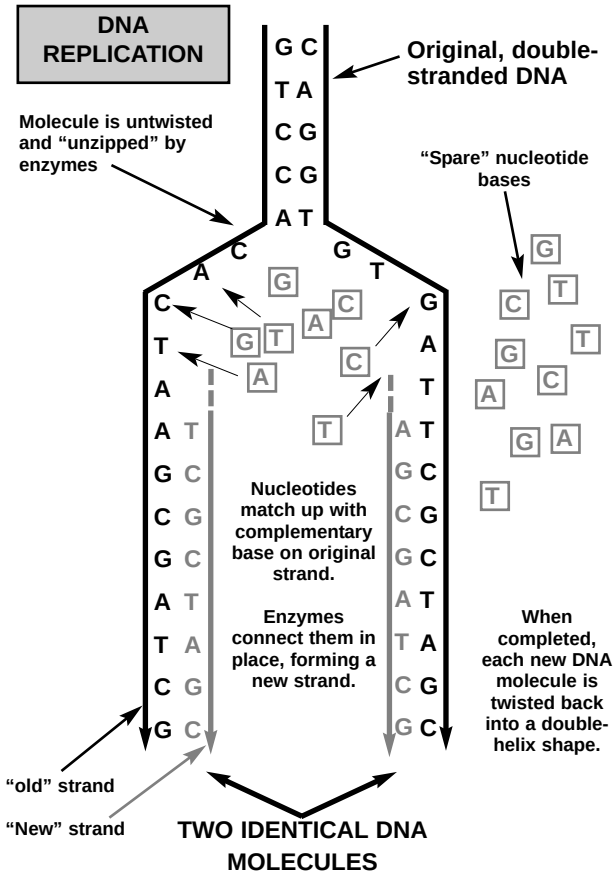
That's exactly what happens to all the DNA in each chromosome, before a cell division occurs.



The only combinations that will bond are

A-T and C-G

DNA REPLICATION



This is how the DNA molecules (which are the genes on the chromosomes) are replicated in preparation for cell division.

Now complete Worksheet 8

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How DNA Structure Was Discovered

By the middle of the 20th century it was suspected that DNA was probably the "genetic chemical" and it was known that it contained sugar, phosphate and the 4 bases A,C,G and T. What no-one could understand was, if DNA was the genes, how could it:

• Replicate (duplicate) itself for cell division? and
• Control the phenotype of an organism?

It seemed likely that the key to this problem was to find out the structure of the DNA molecule. The story of what happened is a classic example of how Science and scientists make progress using collaboration and communication.

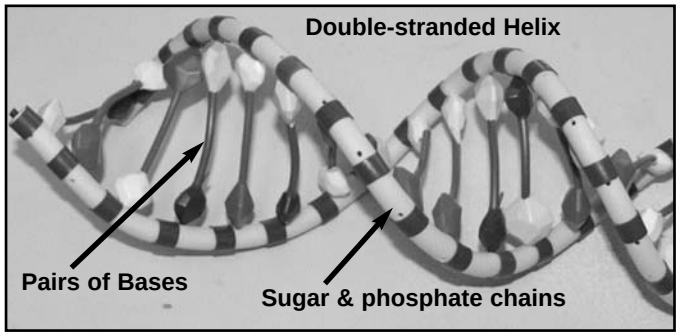
In 1953, English scientist Francis Crick had become an expert at interpreting the shapes of molecules using the (then new) technique of "X-ray Diffraction".

Meanwhile, at another laboratory, Maurice Wilkins (New Zealand) managed to prepare a pure crystal of DNA, and Rosalind Franklin (English) was able to get an X-Ray Diffraction image of it, but neither understood how to make any sense of the pattern it produced.

Then a young American, James Watson, who was interested in understanding the DNA mystery, visited the Wilkins-Franklin laboratory. With their collaboration, he took their data to Crick for his expert interpretation. Between them, Watson and Crick made one of the most notable scientific breakthroughs in the history of Biology... they figured out the base-pairing, double-helix structure of DNA and realised immediately how this structure could lend itself to replication... an essential feature of a gene.

No one of these scientists could have made progress alone. Each had certain data, or skills or expertise, but only by bringing it all together was the great discovery possible. Success came from different people communicating and unselfishly sharing their knowledge and talents.

Sadly, Rosalind Franklin died from cancer before the Nobel Prize awards were decided. Under the rules, the prize could not be awarded to her and consequently her contribution is often forgotten. DNA structure is often described as the "Watson-Crick" Model.



Cell Replication & the Continuity of Species

The purposes of cell division were outlined earlier.

At this point the Syllabus asks you to think about and assess the effects of cell division & replication on the continuity of species.

In one sense this is pretty simple... cell divisions are essential steps in the reproduction of every species. Without reproduction there can be no continuity of any species. Draw your own conclusion from that.

Evolution & Continuity of a Species

Your study of Evolution should have impressed upon you the importance of variations within a species. A species without variations could be at grave risk of extinction if the environment changes.

A species with many variations has a greater chance of species survival because at least some individuals may survive environmental change, then breed to continue the species, possibly in a modified form due to Natural Selection.

Therefore, genetic variation is important to the continuity of a species AND to its evolutionary change by development of new characteristics & adaptations.

Sources of Variations

If a species reproduces asexually, the only way that new variations can be created is by genetic mutations. (More on those in a later topic.)

Some of the bacteria achieve variations faster by exchange of plasmids, (even from one species to another) but the point is that the accumulation of variations is quite slow in asexual organisms.

It's the sexual reproducers who win the honours for developing lots of variations. When 2 parents mix their genes together in their offspring, naturally they create many variations and new combinations of characteristics in the next generation.

Not only that, but the cell division meiosis creates even more variations & combinations because of the random way the chromosomes are separated from each other. A process called "crossing-over" mixes up the gene combinations even more. (More later)

The result is that (for example) a male animal produces millions of sperm cells at a time, yet no 2 of them are likely to be identical genetically. The same applies to the female eggs and when the highly varied sperm cells combine randomly with the highly varied eggs, the result is... well, it varies a lot!

Conclusion: Sex is great for species survival because it contributes hugely to the variations within a species.

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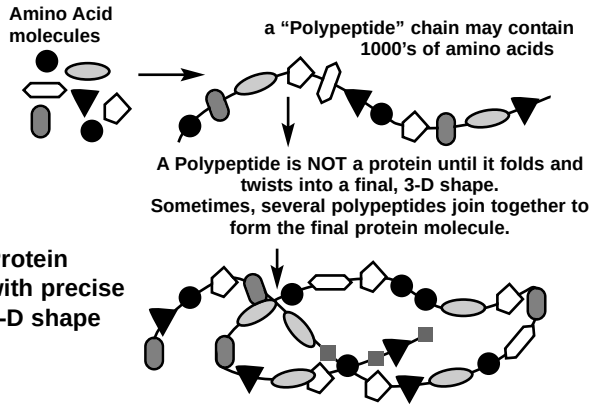
3. DNA & Polypeptide Synthesis

Protein Structure

PROTEINS are polymers of amino acid molucules. A chain of amino acids is called a "polypeptide". However, a polypeptide chain does not become a functioning protein until it twists & folds into a final 3-D shape.



The exact shape of the final protein depends on the sequence of the amino acids in the polypeptide chain. There are about 20 different amino acids, and some of them are attracted to (or repelled from) each other, so how the chain twists and folds upon itself depends on exactly which ones are located where.

Protein Functions

Proteins have many functions within an organism:

- **Enzymes are all protein molecules.**
- **Structural Molecules, such as in muscle fibres, skin, hair and bone matrix are proteins.**
- **Many "Special Molecules" are proteins, such as haemoglobin, (the oxygen carrier in blood) chlorophyll, (absorbs light for photosynthesis) antibodies (which help fight disease)**

... and many more.

In every case, it is the shape of the protein molecule which is essential to its correct functioning.

Enzymes can only connect to their substrate if their shape is right. Haemoglobin can only pick up oxygen if the shape is correct... and so on.

The shape is determined by the precise sequence of amino acids in the polypeptide chain.

This sequence is determined genetically by the "code" in a DNA molecule.

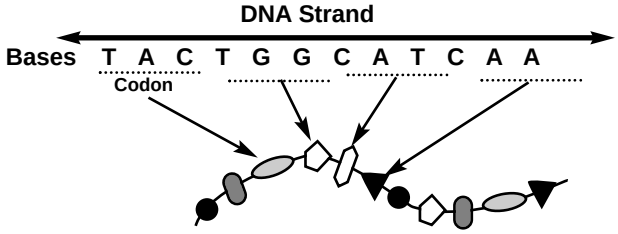
Proteins carry out many vital functions.

Correct functioning depends shape, determined by the sequence of amino acids in the polypeptide chain.

IT IS THE DNA WHICH CONTROLS THE AMINO ACID SEQUENCE.

DNA and Protein Synthesis

The sequence of bases in the DNA molecule is a code. Each 3 bases are a "code word" (called a "codon") which specifies an amino acid to go into the polypeptide chain.



Amino Acids forming a Polypeptide

If a polypeptide containing 1,000 amino acids is needed, then a DNA molecule made up of 3,000 nucleotide bases, will be the gene for this polypeptide.

Only one strand of the double-helix of DNA is the gene. The other "mirror-image" strand is present only to allow the gene to be replicated for cell divisions.

How the DNA base sequence makes a functioning protein which then produces a genetic phenotype is explained by a simple model on the next page.

Changing Definition of a "Gene"

This is what "gene" means in Mendelian Genetics:
"gene" = a hereditary unit which determines one trait in the organism's phenotype.

However, in the 1940's, two American scientists discovered more complexity. Studying a genetic defect in a common fungus, they found that there were 3 different genes that could produce the same defective phenotype.

They realised that the phenotype "S" must result from a chain of reactions each controlled by a protein enzyme.

Normal Chemical Pathway in Fungi Cells

P —Enzyme 1→ **Q** —Enzyme 2→ **R** —Enzyme 3→ **S**

P,Q,R and S are cell chemicals. If any reaction is blocked, then "S" cannot be made, and the organism has a chemical defect

The phenotype "S-defect" could be produced by a defect to the gene for enzyme 1, or the gene for enzyme 2, or the gene for enzyme 3. So, the new definition for a gene became:

"gene" = a unit of heredity that specifies a protein

But now that we know about DNA, and that some proteins require more than one polypeptide chain...

A "GENE" IS A DNA MOLECULE WHICH SPECIFIES ONE POLYPEPTIDE

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How DNA Causes a Phenotype

Part 1. From DNA to Polypeptide

The initial step is called TRANSCRIPTION

This occurs in the nucleus

DNA

m-RNA

RNA is a polymer of nucleotides, similar to DNA, but
- single strand only
- different sugar
- one different nucleotide

m-RNA moves from the nucleus out to a ribosome

One strand of the DNA (the gene) acts as a "template" for the production of a single-stranded "messenger RNA" (m-RNA)

Each 3-base "codon" in m-RNA specifies one amino acid in the polypeptide chain

The next step is called TRANSLATION

Amino Acids are carried into position by t-RNA ("Transfer RNA") molecules

t-RNA's

Amino Acids

Polypeptide chain growing by addition of Amino Acids

Ribosome enzymes connect the amino acids to form a polypeptide chain

m-RNA

Ribosome moves along m-RNA

You should recall that the Ribosomes are organelles responsible for protein synthesis in cells

then, Part 2.

From Polypeptide to Phenotype

The polypeptide chain twists and folds to form a protein

Possibly it is combined with other polypeptides or special chemical groups to form...

a functional PROTEIN molecule

Perhaps it's a structural protein, used to build muscle or skin

Maybe it becomes a molecule of haemoglobin, used to carry oxygen in the blood

It could be a hormone, which controls some body function, such as growth.

It could be an ENZYME, able to catalyse a chemical reaction.

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Example: Eye Colour

The colour of your eyes is a genetically determined phenotype. The coloured pigment is made by enzymes, which are proteins, coded in your DNA

Substrates

Enzyme

STARTING CHEMICALS (Substrates)

Enzyme catalyses reaction(s) to produce coloured pigment in cells of the iris of the eye

Product; a Coloured Pigment

Now complete Worksheet 9

The gene(s) for eye colour are present in every cell of your body, but are only expressed in your iris cells. Similarly, the genes for liver functions are only expressed in your liver cells, and genes to make taste buds are only expressed in your tongue. What controls which genes are expressed is still unknown in most cases.

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m-RNA & t-RNA

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In between the DNA "gene" and the protein it produces there are 2 vital molecules you need to know about.

Messenger RNA (m-RNA)

RNA stands for "ribonucleic acid".
m-RNA molecules are very similar to a single-strand of DNA. The sugar in the "backbone" is slightly different and one of the "bases" in its 4 nucleotides is different. The DNA base "Thymine" (T) is replaced in RNA with "Uracil" (U). Uracil is chemically similar to thymine and will bond with Adenine in the same way.

Transcription

Imagine you had a document written in (say) Russian and some multi-lingual computer transcribed the words into (say) Spanish.

We're sure that there are many people who understand both those languages, but for English-only morons like us, re-writing a Russian document in Spanish does NOT help us know what it says.

This is similar to what happens in the nucleus when the DNA code is transcribed onto an m-RNA molecule. The cell does NOT understand DNA code, nor does it understand m-RNA code.
(Cells only understand protein-language)

The genetic code has been transcribed from one code into another, but it doesn't mean anything yet.

Example Transcription

DNA Code Sequence (3 bases = "codon")	m-RNA Code Sequence (3 bases = "anti-codon")
A	U
C	G
T	A
G	C
G	C
A	U
T	A
C	G
A	U

Once constructed, the m-RNA molecule moves out of the nucleus and attaches to a ribosome. This organelle is the "translator" which uses the m-RNA code to construct a polypeptide chain of amino acids. This eventually becomes a protein which has a meaning within a cell, as a structural molecule or an enzyme, etc.

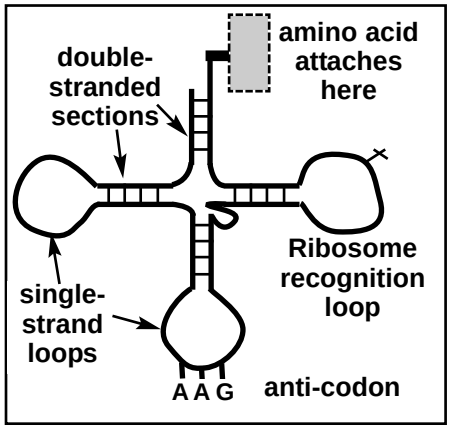
To carry out the "translation", the ribosome needs the help many enzymes (which it carries) AND it needs another form of RNA to carry amino acids and place them correctly into the chain.

Transfer RNA (t-RNA)

There are several hundred different t-RNA molecules. They all have the same basic structure, sometimes described as a "clover leaf".

At one end (top in this diag) each t-RNA molecule can attach to a specific amino acid and carry it.

At the opposite end is a loop with 3 nucleotide bases turned outwards forming an exposed "anti-codon"

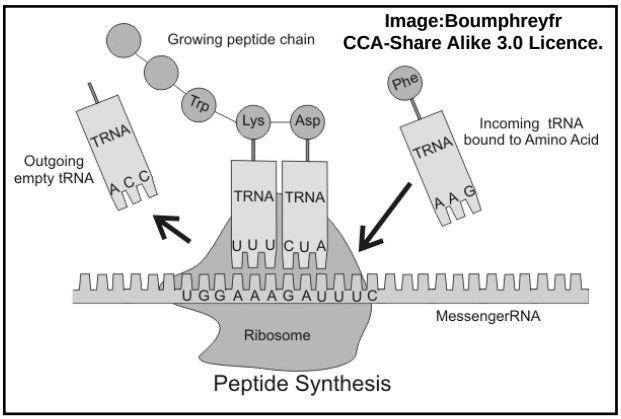


If that anti-codon says "AAG", then that molecule will only attach to the amino acid "phenylalanine". If it says "UUU" it will only attach to amino acid "lysine", and so on.

Another of its "loops" is the correct shape to "lock-onto" a ribosome and activate the enzymes which join the amino acid onto the growing polypeptide chain and dis-engage the t-RNA molecule. The t-RNA can migrate away and carry another amino acid to repeat its transfer role.

However, the t-RNA cannot just lock onto a ribosome in any old way. It can only engage if its anti-codon is complementary to the next available triplet of bases on the m-RNA which the ribosome is rolling along like a little train along a track.

If the next m-RNA triplet says "UUC", then the t-RNA with anti-codon "AAG" (carrying amino acid phenylalanine") can lock-on and place its payload in position to add onto the growing polypeptide chain.



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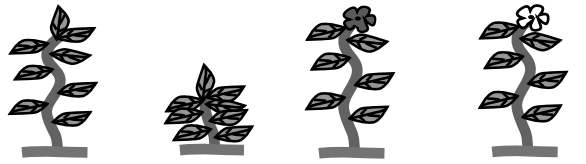
4. Genetic Variation

Revision of Mendelian Genetics

Gregor Mendel's Experiments

Mendel was the Abbot of a monastery in what today is the Czech Republic. He was trained as a teacher and was not a professional scientist, but became interested in discovering how inheritance works. The monks grew most of their own food, so Mendel worked his investigations into the day-to-day vegetable gardening by choosing to experiment with garden peas.

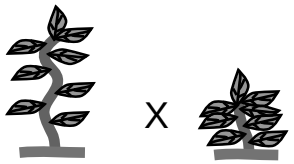
First he bred his pea plants over several generations to select plants that were "pure breeding" for certain contrasting characteristics, such as...



TALL **DWARF** **PURPLE FLOWER** **WHITE FLOWER**

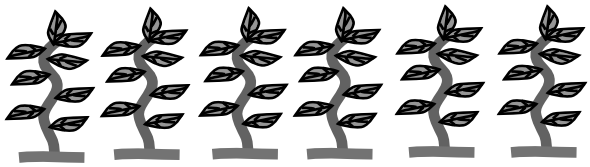
Each type was "pure breeding", meaning that if they were self-pollinated they always produced offspring of exactly the same type as themselves.

Then he cross-pollinated 2 contrasting types to obtain "hybrid" (cross-breed) offspring. The result was that all the offspring showed the characteristic of one parent and none took after the other. For example, when TALL plants were crossed with DWARF:



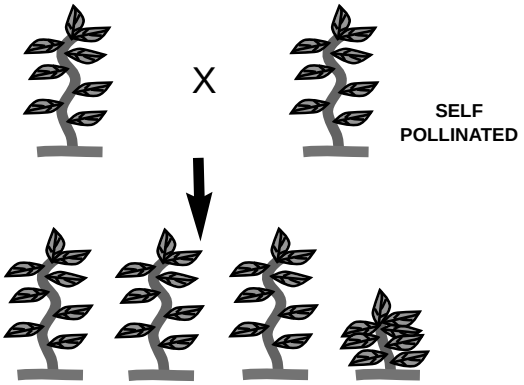
"X" is short-hand for "crossed with"

F_1 (1st Generation) Offspring were ALL TALL



Next, he self-pollinated the plants of the F_1.

TALL F_1 plant



SELF POLLINATED

F_2 (2nd Generation) Offspring occurred in a ratio of 3:1

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Mendel didn't do this with one or two plants, but with hundreds. His 2nd generation totalled thousands of plants, not just a few.

He got the same result with flower colours, seed shapes, seed pod colours, and so on. In every case the first generation always took after one parent completely, and the second generation always appeared (in their thousands) in a ratio of very close to 3:1.

Mendel's Explanation of His Results

(Using Modern Terminology)

Each characteristic is produced by "factors" (we now call them genes) carried by the plants. For example, there is a gene for tallness of stem, and a corresponding gene for dwarf stem. There is a gene for purple flower and another for white flowers, and so on for other characteristics.

The genes which control "opposite" forms of the same characteristic are called "alleles", or "allelic genes". Genes for "tall" and "dwarf" are alleles. Genes for "purple flower" and "white flower" are alleles.

One of the alleles is "Dominant" over the other, which is said to be "Recessive". We usually use letters of the alphabet to designate this:
e.g. Tall (T) is dominant to Dwarf (t)
Purple flower (P) is dominant to white (p)

Each plant carries 2 genes for a characteristic. The 2 genes may be the same as each other ("homozygous") or different to each other ("heterozygous").

Example: for the height characteristic, the possibilities are:
TT = homozygous, Tall plant
Tt = heterozygous, Tall plant (T dominant)
tt = homozygous, Dwarf plant

Although each plant carries 2 genes for each characteristic, only one gene is passed into the gametes (pollen or ovules). Each parent passes on one gene, so the offspring gets one from each parent and gets back to having 2 genes for the characteristic.

Which one of the 2 genes for each characteristic is passed on is completely at random.

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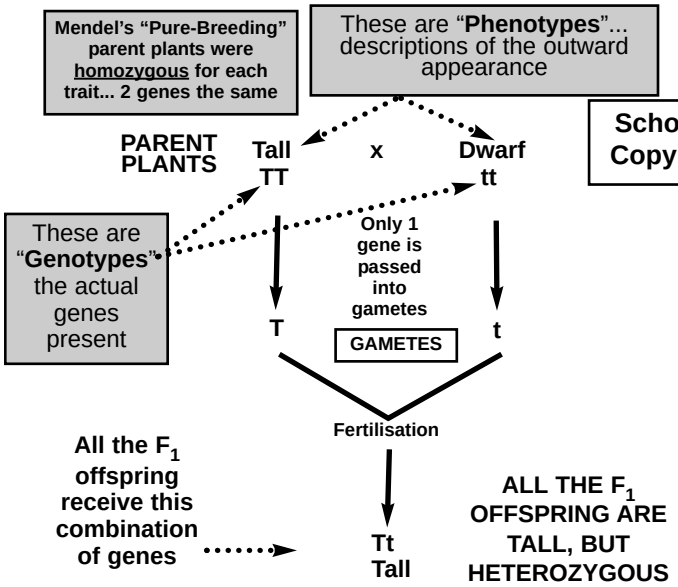
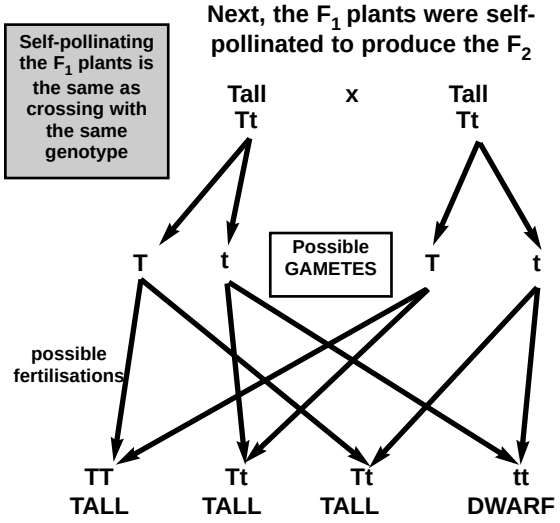
Revision of Mendelian Genetics *(cont.)*

This diagram explains why Mendel observed a ratio of about 3:1 in the plants of his F_2 offspring.

His experimental ratios were approximately 3:1, but not exactly 3:1. This is because the actual combinations of gametes at fertilisation occur at random. He bred large numbers of plants and so his actual ratio was very close to theoretical.



Next, the F_1 plants were self-pollinated to produce the F_2



Ratio of Phenotypes 3 Tall : 1 Dwarf

You will soon come to realise that in crosses of this type only 3 outcomes are possible:

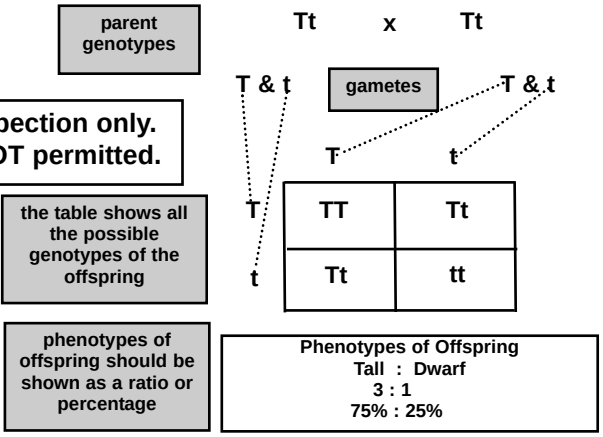
100% : zero
50% : 50% = 1 : 1 ratio
75% : 25% = 3 : 1 ratio

Now complete Worksheet 10

The Punnett Square

The "working out" of a cross by a diagram can be a bit messy and confusing. A scientist called Punnett invented a simpler method which you must learn to use.

The "Punnett Square" working below shows the formation of the F_2 plants in Mendel's experiment.



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Monohybrid Crosses

"mono" = 1 (referring to just one characteristic)
"hybrid" = cross-breed

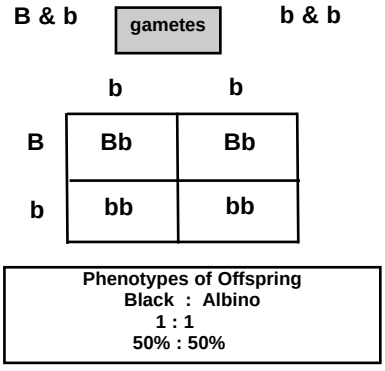
It has been found that there are many characteristics, in all sexually-reproducing organisms, which are inherited just the way Mendel discovered...
these are cases of "Mendelian Genetics" and you need to be able to predict the outcomes of simple crosses.

Study the following examples to help you work through the next worksheet.

Sample Problem

In mice, black coat (B) is dominant to albino (b). Predict the outcome of mating a <u>heterozygous</u> black mouse with an albino.

<u>Solution</u>
Parents are Bb x bb



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Pedigrees (Family Trees)

Another skill you must learn is how to read, interpret and construct a pedigree diagram.

This is a diagram which shows the inheritance of a trait through a family. It is used particularly with human families to trace some characteristic over a number of generations.

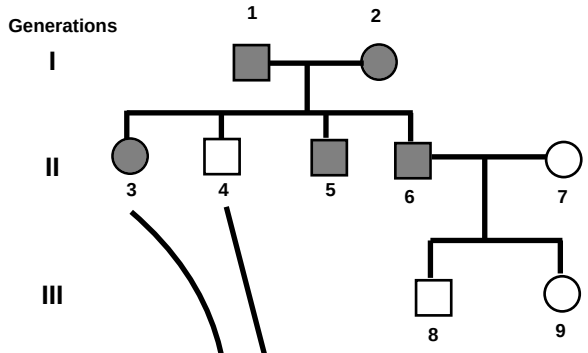
Symbols Used in Pedigree Diagrams

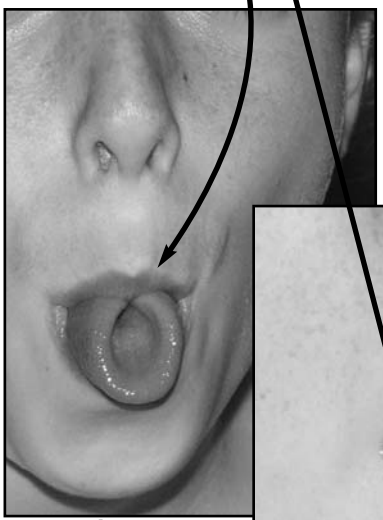
Male with trait being studied — **Male without trait**

Female with trait — **Female without trait**

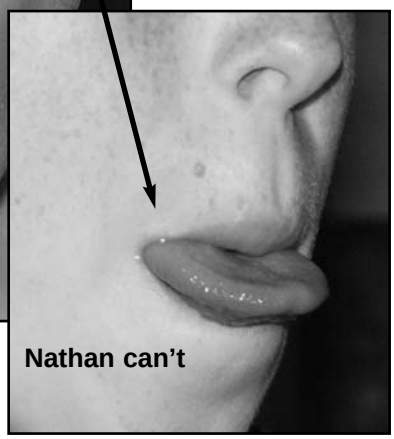
Horizontal connections are "marriage lines".
Vertical lines lead to children of that couple.
Each generation is numbered by Roman Numerals.
Individuals may be numbered for identification.

Example
In humans, some people can "roll their tongue" while others cannot. This is passed on by simple Mendelian Inheritance. Here is a pedigree of a family showing how this trait was passed on.





Sarah can...



Nathan can't

Complete Worksheet 11

Interpretation of this Pedigree

In Generation I, individuals 1 & 2 were both tongue-rollers. They had 4 children, a daughter and 3 sons. Most of the kids can roll their tongues, but son "4" cannot.

This means the <u>inability</u> to tongue roll must be recessive.
(Whenever a child shows a trait different to both parents, the child's phenotype must be recessive.)

Therefore, <u>tongue-rolling</u> ability must be dominant.

We can now assign symbols...

tongue-rolling = R non-rolling = r

...and work out most people's genotypes:

1	2	3	4	5	6	7	8	9
Rr	Rr	?	rr	?	Rr	rr	rr	rr

(Individuals "3" & "5" might be either "RR" or "Rr".
... more information needed to be sure which)

Questions & Answers

How can we be sure that parents 1 & 2 are both "Rr" (heterozygous)?
A: Since they produced son "4" who is a non-roller (must be genotype "rr") both 1 and 2 must be carrying the recessive gene. Therefore, both must be "Rr".

Can we be sure that son "6" is "Rr" and NOT "RR"?
A: He married a non-roller (rr) and both the children in generation III are non-rollers. Therefore, son "6" must have passed on a recessive gene to his children. He <u>must</u> be heterozygous (Rr) to do this.

If "6" and "7" had another child, could it be a tongue roller? What's the chance?
A: Yes. The cross is Rr x rr.

If you work out a punnett sqare for this, you will see that the expected outcome is 50% rollers and 50% non-rollers. The chance for the next child is 50% either way, and is NOT affected by the fact they have already had 2 non-roller children.

Uses of Pedigree Diagrams

Pedigree diagrams are used to study human inheritance because it is not moral or ethical to carry out breeding experiments on people to discover which traits are dominant / recessive.

Some human disorders are inherited. Examples are <u>haemophilia</u> (in which blood will not clot properly) and <u>colour-blindness</u> (inability to distinguish certain colours). Health professionals can study affected families by compiling a pedigree chart, then advise people about the risks to future children. This allows people to make informed decisions about family planning.

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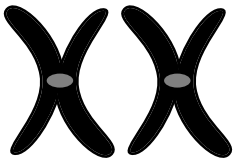
What Determines Sex?

In humans, and in many other species, sex is determined by a special pair of chromosomes... the "sex chromosomes".

In a normal human body cell there are 46 chromosomes arranged in 23 homologous pairs. Of these, 22 pairs are called "autosomes" and are the same size and shape in males as in females.

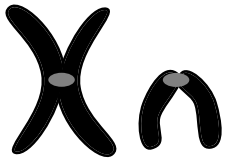
The 23rd pair are the "sex chromosomes":

Females have a matching pair of chromosomes that are known as "X" chromosomes. A female is described as "XX"



Males have one "X" chromosome, and one smaller "y" chromosome.

Males are described as being "Xy".



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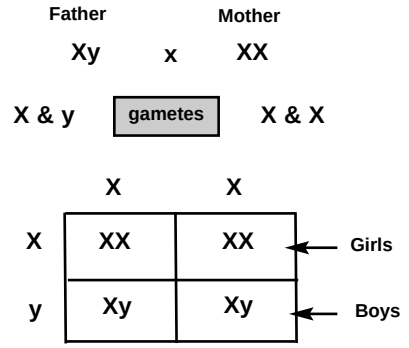
How these chromosomes are passed on to children can be shown using the Punnett Square diagram to track, not genes, but the sex chromosomes:

Notice that females can only pass on an X chromosome in their eggs.

Male's sperm cells can carry an X or a y.

Which type of sperm fertilises the egg determines the sex of the baby.



Phenotypes of Offspring
Girls : Boys
1 : 1
50% : 50%

Non-Mendelian Inheritance

Gregor Mendel discovered the basics of Genetics, but it was found early in the 20th century that genes don't always work in that simple "Mendelian" fashion.

Thomas Morgan began experimenting with *Drosophila* fruit flies and quickly found they were ideal for genetics experiments.

In 1910, in an experiment involving flies with different eye colours, Morgan realised that the way this characteristic was being inherited depended on the sex of the fly... males and females were inheriting eye colour differently.

Sex-Linkage Inheritance

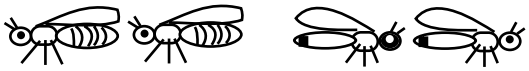
The common and normal eye colour in the flies is red. Morgan discovered a male fly with white eyes. He set out to do a "Mendel-type" breeding experiment:

PARENTS White-eyed Male x Red-eyed Female

F_1 generation

All have red eyes. Morgan concluded (correctly) Red is DOMINANT, White RECESSIVE

F_1 flies were allowed to mate among themselves



F_2 generation

Females 100% red eyed. **Males 50%: 50%**

What's going on?

Morgan produced hundreds of flies in the experiment so, like Mendel, his results were statistically significant.

The Explanation:

The genes for eye colour are carried on the X chromosome.

The dominant (Red) gene can be designated as "X^R". The recessive (White) gene is "X^r" The male "y" chromosome does NOT carry one of these alleles at all.

The possible female (XX) genotypes & phenotypes are:

X^RX^R	Red eye female (homozygous)
X^RX^r	Red eye female (heterozygous)
X^rX^r	White eye female

The possible male (Xy) genotypes & phenotypes are:

X^Ry	Red eye male
X^ry	White eye male

Note that females get 2 genes, but males only get one because their "y" chromosome lacks this allele totally. A male cannot be heterozygous for this trait and cannot have a "hidden" recessive gene.

WHEN DOING PUNNETT SQUARES WITH SEX-LINKAGE, YOU MUST TRACK THE "X" AND "y" CHROMOSOMES...

see next page.

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Morgans's Sex-Linkage Experiment (cont.)

Here is the experiment explained by Punnett Square.

X^ry x X^RX^R

X^r & y gametes X^R & X^R

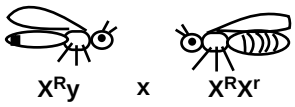
	X^R	X^R
X^r	X^RX^r	X^RX^r
y	X^Ry	X^Ry

The F_1 are all red-eyed. Note that females are all heterozygous.

Phenotypes of F_1 Offspring (both sexes)
Red:White
100% : 0

Then, the F_1 generation were allowed to breed among themselves. Notice that the F_1 females are all heterozygous red eyed (X^RX^r) and the males are all red eyed (X^Ry).

F_2



X^Ry x X^RX^r

The F_2 are red : white 3 : 1 just like Mendel's results, but the sexes are different.

X^R & y gametes X^R & X^r

	X^R	X^r
X^R	X^RX^R	X^RX^r
y	X^Ry	X^ry

Phenotypes of F_2 Offspring	
FEMALES	MALES
Red : White	Red : White
100% : 0	50% : 50%

There are a number of genes in humans that are sex-linked. The best-known are 2 genetic disorders:
- **<u>Red-Green Colour Blindness</u>, is where a person cannot distinguish between certain colours.**
- **<u>Haemophilia</u>, is a condition in which the blood does not clot properly, putting the person at constant risk of internal bleeding. In the past it was always a fatal condition, but in modern times people with haemophilia are treated with "clotting factors" from donated blood.**

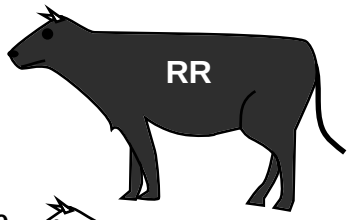
Both conditions are sex-linked, inherited as recessive genes on the "X" chromosome. They occur much more often in males than females.

Co-Dominance & Incomplete Dominance

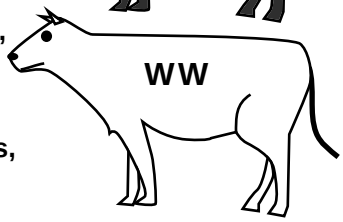
This is a fairly common situation in which the 2 alleles for a characteristic do not show a Dominant-Recessive pattern, but when both genes are present (heterzygous) they are both expressed, or their effects blend together resulting in an "in-between" phenotype. Collectively, these can be called "<u>Intermediate Inheritance</u>".

An Example of Co-Dominance

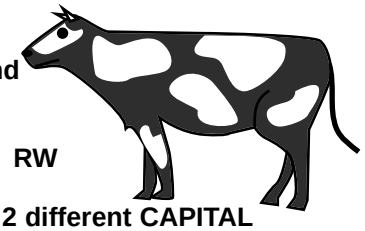
<u>Example</u>: In Shorthorn cattle, there is gene (R) that causes the hair of the animal's coat to be "red" (actually a rusty-brown colour). To be red, the animal must be homozygous for this gene.



The other allelic gene (W) causes the coat to be white, if the animal is homozygous.



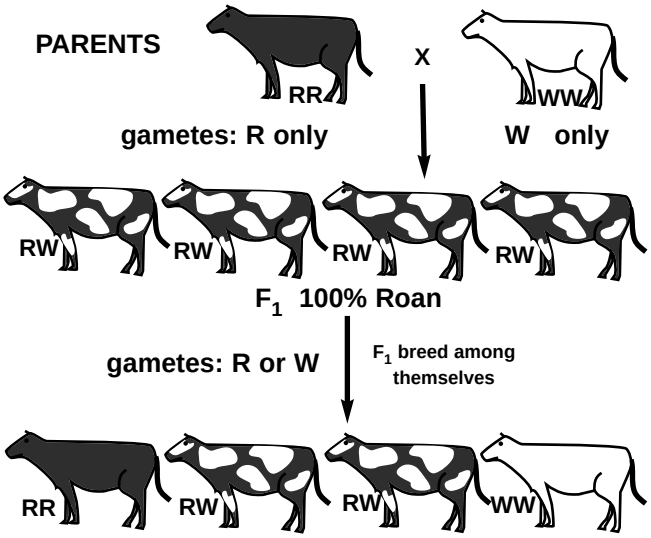
If an animal is heterozygous, with both alleles present, neither gene dominates the other. Both genes are expressed, producing mottled patterns of red and white hair which is called "roan".



Genotype RW

Note that it is best to use 2 different CAPITAL letters as symbols in this case, since neither gene is recessive.

Here is what happens in a "Mendel-type" cross:



Phenotypes: RED : ROAN : WHITE
1 : 2 : 1
25% : 50% : 25%

Try to verify for yourself the F_2 result by using a Punnett Square.

Now complete Worksheet 12

How is "Incomplete Dominance" Different?

Co-Dominance (above) is when the 2 alleles are <u>both</u> expressed. Incomplete dominance is when the alleles result in a "blending" to produce an "in-between" phenotype. For example, in snapdragon plants, a gene "R" produces red flowers if the genotype is RR. The allelic gene "W" produces white flowers (genotype WW). If a plant is heterozygous for this trait (RW) the flowers will be PINK.

<u>Co-Dominance = patchy, spotty, both things at once</u>. <u>Incomplete Dominance = blended, combined trait</u>.

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Multiple Alleles

Mendel studied situations where there were only 2 alternative forms of each trait, controlled by only 2 allelic genes.
However, it's not always so simple...

Some characteristics have more than 2 alternative forms and more than 2 alleles.

In clover plants, the pattern of "chevrons" on the leaves is controlled by 7 different allelic genes. Each plant inherits one gene from each parent (as usual) for leaf pattern, but with 7 alleles to choose from, there are many possible genotypes and phenotypes.

To get the idea, let the 7 alleles be labelled
a,b,c,d,e,f,g.
Then the possible genotypes are:

aa, ab, ac, ad, ae, af, ag
bb, bc, bd, be, bf, bg
cc, cd, ce, cf, cg
dd, de, df, dg
ee, ef, eg
ff, fg
gg

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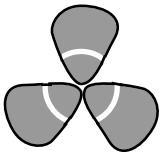
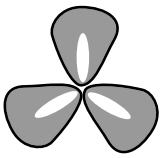
... a total of 28 gene combinations, each giving a different phenotype.





2 of the possible phenotypes of leaf pattern

Human ABO Blood Groups

The classic example of multiple alleles in human genetics is the way that we inherit our "blood type".

Each of us inherit 2 genes for blood type, but there are 3 allelic genes available. The genes are usually given the symbols I^A, I^B & i.

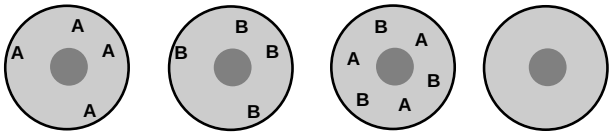
Genes I^A and I^B show co-dominance with each other, and are both dominant to allele "i".

This results in 6 possible genotypes, and 4 different blood group phenotypes.

Possible Genotypes	Phenotype
I^AI^A	blood type A
I^Ai	A
I^BI^B	B
I^Bi	B
I^AI^B	AB
ii	O

What are the Blood Groups?

When expressed, these genes cause the production of antigen molecules on the surface of the red blood cells. These antigens have no effect on how the blood cells work, but are part of the system by which your immune system recognises cells which are "self" or "not-self". When foreign cells are detected, you produce antibodies to destroy them.



Type A | **Type B** | **Type AB** | **Type O**

carry "A" antigen | **carry "B" antigen** | **carry both antigens** | **have neither**

Blood group antigens become a matter of life-or-death during a blood transfusion. If a patient is given blood which carries an antigen which their own cells do not have, their immune system will react with antibodies against the foreign cells. This can result in fatal blood clots forming inside the blood vessels.

It follows that:
- **type O blood can be given to anyone.**
- **type AB patients can receive any type.**
- **type A or type B must be given their own type, or O.**

In practice, every effort is made to only give blood of the exact same type as the patient.

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Multiple Alleles *(cont.)*

Examples of Blood Type Inheritance

Example 1

Predict the genotype and phenotype probabilities in the children of a couple with genotypes I^Ai and I^Bi. (i.e. one is heterozygous type A and the other is heterozygous type B)

Solution

parent genotypes I^Ai x I^Bi

genes passed on in gametes I^A or i x I^B or i

Punnett Square of possible children's genotypes

	I^B	i
I^A	I^AI^B	I^Ai
i	I^Bi	ii

Possible Children's Genotypes & Phenotypes

I^AI^B, Type AB 25% chance
I^Ai, Type A 25%
I^Bi, Type B 25%
ii, Type O 25%

Example 2

Bill and Mary are both blood type B. They have 2 children; Freddy has blood type O and Susie is type B. Deduce the genotypes of each person, and predict the probable blood types if Bill and Mary have more children.

Solution

Since they have produced a child with blood type O (genotype ii) both parents must be heterozygous and carry the recessive "i" gene.

parent genotypes I^Bi x I^Bi

genes passed on in gametes I^B or i x I^B or i

Punnett Square of possible children's genotypes

	I^B	i
I^B	I^BI^B	I^Bi
i	I^Bi	ii

Possible Children's Phenotypes

Type B 75% chance
Type O 25%

Answer:

Bill & Mary are both I^Bi. Freddy is ii.

Susie might be I^BI^B or I^Bi.

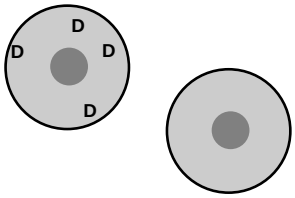
If they have more children, there is a 75% chance of type B, and 25% of type O.

The Rhesus Factor

The ABO blood groupings are not the end of the story. Human red blood cells can also carry another antigen "D" known as the "rhesus factor" because it was first discovered in the blood of Rhesus monkeys.

The inheritance of the "D" rhesus factor is entirely independent of the ABO antigens and is controlled by 2 alleles which show a simple dominant-recessive pattern.

If you have antigen "D" on your blood cells you are said to be "Rh+".



If you lack this antigen you are said to be "Rh-".

If a patient with Rh- blood is given a transfusion containing Rh+ cells, the resulting immune system response could be fatal, so (as with the ABO grouping) it is important to match Rh blood types for transfusions.

When describing a person's blood type, both the ABO and Rh classifications are described. For example, "blood type AB+" means the cells carry antigens A, B and D. "Blood type A-" means the cells carry antigen A only.

Various symbols are used to denote the rhesus factor alleles, but the simplest system is to use "D" for Rh+, and "d" for Rh-.

Comparison of Rhesus to ABO Inheritance

The rhesus factor is inherited in a mendelian pattern: ...two alleles in dominant-recessive pattern.

The ABO blood types are inherited by a multiple alleles system; 3 alleles giving 6 genotypes and 4 phenotypes.

Example Problem

An Rh- man married a woman who is Rh+. Her father was Rh-, so she must be heterozygous.
Predict the Rh phenotype probabilities of their children.

Solution

parent genotypes Dd x dd

genes passed on in gametes D or d x d or d

Punnett Square of possible children's genotypes

	d	d
D	Dd	Dd
d	dd	dd

Possible Children's Phenotypes

Rh+ 50 % chance
Rh- 50%

Now complete Worksheet 13

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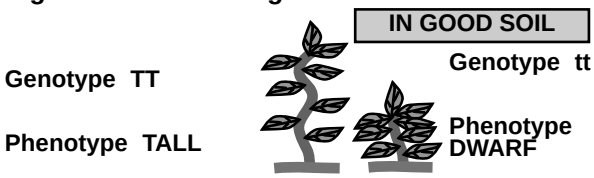
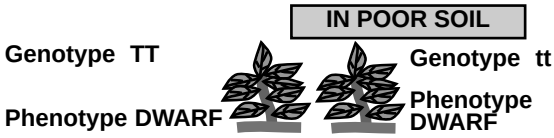
The Effect of Environment

It's not just an organism's genes that produce its phenotype; the environment has an effect as well.

For example, consider some of Mendel's pea plants with different genes for stem height.



Now, imagine planting them (as baby seedlings) in very poor soil, so that normal growth was not possible.



The point is that the genes may control what the organism COULD grow up to be, but the environment may influence this, possibly altering the final appearance (phenotype).

In Hydrangea plants, if cuttings are taken from a single individual (the cuttings would be genetically identical) and grown in different soils, the flowers on each cutting can be different colours. If the soil is slightly acidic the flowers will be blue, but in slightly basic soil they'll be pink.

Identical twins have inherited exactly the same genes. In the USA about 40 years ago, a famous study was done on identical twins who had been adopted into separate families and raised in different environments. The study found quite large differences between the twins in intelligence, personality, and even appearance. Presumably these differences were due to different foods, education and upbringing, etc.

Genetics, Sex & Evolution

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During his lifetime, Charles Darwin freely admitted that there were 2 big gaps in his (then) controversial Theory of Evolution...

1. <u>How are characteristics inherited?</u>
When the "fittest" survive and breed, how do they pass on their "survival traits" to their offspring?

The Science of Genetics can now explain that

2. <u>Where does variation come from?</u>
Natural Selection needs differences between individuals to choose the survivors. Why is there variation anyway?

We now know that a lot of variation comes from MEIOSIS and Sexual Reproduction

Variation Caused by Meiosis

The process of meiosis to produce the sperm and egg cells increases variation, even before fertilisation occurs.

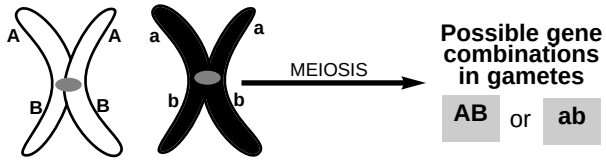
Study the diagram of meiosis on page 11. Remember that homologous chromosomes are NOT identical.

Each pair of homologous chromosomes line up and separate at random, and <u>independently of all other pairs</u>, so the number of different possible gametes is very large. In humans, with 23 pairs of chromosomes, it is possible for meiosis to produce about 8 million different combinations of chromosomes in the gametes of each person!

Crossing-Over

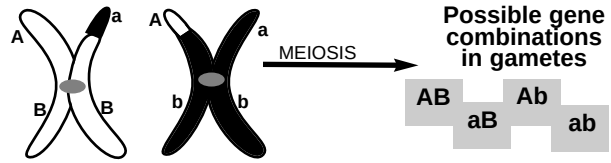
During meiosis homologous chromosomes also <u>swap pieces of chromatid with each other</u>, further mixing up the possible gene combinations:

GAMETE FORMATION WITHOUT CROSSING-OVER



Remember, each gamete gets just one of these 4 chromatids

WITH CROSSING-OVER



These chromosomes have exchanged pieces of chromatid with each other. This has mixed up the combinations of genes "A", "a", "B" and "b".

Variation Caused by Sexual Reproduction

The simple fact that sexual reproduction involves TWO parents, creates a lot of variation. The offspring receives genes from 2 different individuals, thereby getting a new "mix" of traits.

Summary

Meiosis creates variations in the way <u>homologous chromosomes separate</u>, AND in the process of <u>Crossing-Over</u>. Further variation comes from <u>combining genes</u> from 2 parents.

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5. Inheritance Patterns in a Population = Population Genetics

Population Genetics **is the branch of Biology where Evolutionary Theory meets Genetics.**

A starting point to understand what's involved is to realise that the evolution of any species must necessarily change the gene frequencies present in the species population.

"Gene frequency" is a measure of how common a particular gene is within a population. If a certain gene produces a phenotype characteristic which helps survival, one that is favoured by Natural Selection, then the frequency of that gene in the population will increase over generations, as the "favourable" characteristic becomes more common.

Dominant Genes & Gene Frequency

Won't dominant genes automatically increase in frequency anyway?

No!!

This is a common mis-conception. You must NOT confuse the dominance or recessiveness of a gene with its occurrence across a population.

Just because a dominant gene causes a higher frequency of its phenotype, this does not mean its gene frequency has risen.

Look at the results of Mendel's classic breeding experiment with pea plants. Starting with pure-breeding tall (TT) and dwarf (tt) parents, the F_2 offspring are:

Phenotypes: Tall : dwarf = 3 : 1 = 75% : 25%
It looks like the tall gene (T) must be more common.

Wrong! Examine the genotypes in the F_2.

Genotypes:	**TT**	**Tt**	**tt**
	25%	**50%**	**25%**

Gene Frequency: **Occurrence of T gene = 50%**
Occurrence of t gene = 50%

Despite the more frequent occurrence of the "Tall" phenotype, the gene frequencies are in fact equal, just as they were at the beginning of the breeding experiment. (TT x tt)

One way to think about it is that the recessive genes can "hide" in the heterozygous members of a population.

This concept was proven mathematically about 100 years ago. We are not going into the maths, but you need to be aware of...

The Hardy-Weinberg Principle

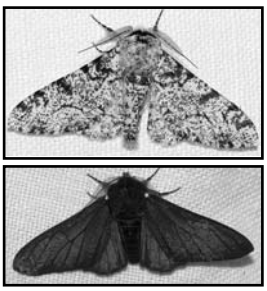
This principle says that, in any sexually-reproducing population where the matings are at random, there will be NO CHANGE in gene frequencies, UNLESS certain things are occurring.

Sounds pretty boring! "Nothing happens unless something happens"? Well, DUHH!

What are the "certain things" that can occur which can cause gene frequencies to change?

It might be migration in or out of the area studied. It includes the possibility of random changes due to chance events, especially in small populations where the statistical likelihoods don't occur as expected by the mathematics. (This is called "Genetic Drift")

There are other possibilities as well, but the big one is Natural Selection. If a species is evolving, some characteristics are being favoured for survival while others are selected against. This can change the frequencies of the genes which contribute to these characteristics.



Light and dark forms of the Pepper Moth.
Image by Olaf Leillinger CCA-SA 2.5

Remember the case of observed "micro-evolution" of the English Pepper Moth? When it was studied, scientists went to some pains to measure gene frequencies.

Then they looked for evidence of migration, non-random mating and other factors.

Finally, it was concluded that:
• the relative abundance of genes (gene frequencies) for colouration were changing over time.
and
• the impacts of all other reasons for gene frequency change were eliminated or accounted for. These could NOT explain the genetic changes. Therefore, it had to be Natural Selection at work.

That's why we are quite sure that micro-evolution was and is occurring... it's not just random chance or guesswork.

Population Genetics, with the Hardy-Weinberg Principle as one of its foundation stones, has become a major branch of modern Biology.

But how is it being used? ...read on...

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The Human Genome Project (HGP)

The HGP was an international scientific project carried out collaboratively by twenty universities in 7 countries between 1990 to 2003. Its target was to find the sequence of nucleotide base pairs in the total human genome.

("Genome" = the total genetic material of a species)

When first conceived, the HGP was predicted to need 25-30 years to complete. However, as new automatic base-sequencing machines (plus new computer software for analysis) were developed, the progress accelerated so that it was completed much faster.

Outcomes

The key findings of the HGP were:

• the complete human genome contains about 3.3 billion nucleotide base pairs in all the DNA in our chromosomes.
(More DNA is located in other cell organelles such as mitochondria, but this was not included in the HGP.)

• humans have about 22,000 genes.
ie there are 22,000 separate DNA molecules in our chromosomes (present in every cell in our bodies) which cause the production of a polypeptide and ultimately produce a genetic phenotype.

• less than 2% of our DNA is actually used to express genes by coding for a polypeptide.

Some of the other 98% of DNA is to code for the many t-RNA's needed for gene translation. Some is involved in regulating gene expression. A lot of it seems to be duplications. Some might be corrupted ancestral genes which are no longer in use. Some is "telomere" DNA which seems to be important for protecting chromosomes from damage and may be involved in the aging process. A lot of it we simply cannot yet explain.

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Possible Benefits of HGP

The HGP promises to be one of the most important scientific achievements in history, ranking beside the control of electricity or the invention of computers, in its benefits to society.

Some of these potential benefits include:

• understanding (and curing) many forms of cancer.

• treatment for many infectious diseases which work by interacting with our genome.

• designing better drugs to treat many conditions.

In addition, the "spin-off" of DNA-sequencing technology will have many applications in food-production and developing renewable fuels, as well as in the biological sciences in understanding (for example) human evolution.

Single-Nucleotide Polymorphisms (SNiP's)

"Polymorphism" literally means "having many shapes", but in Biology is used to refer to variations or different characteristics found within a species.

A "single-nucleotide polymorphism" refers to a variation in a single nucleotide base-pair at a specific location within a person's DNA. An SNP is often spoken of as a "SNiP".

One of the "spin-offs" from the HGP and its technology has been the discovery of, and ability to detect, "SNiPs" in the DNA of an individual person.

Effects of a SNiP

If a SNiP occurs in a DNA sequence which codes for a polypeptide, this can cause one amino acid to be different, because a DNA codon is different. In some cases this makes no difference to the final protein; in other cases it can make the final protein perform differently or not work at all. This is rarely beneficial and can be lethal.

In some cases, a SNiP in a coding DNA gene does NOT change the amino acid. This is because there are multiple codons for each amino acid. For example, the DNA codons GGA, GGT, GGC & GGG all code for the amino acid Glycine. A SNiP in the 3rd nucleotide of the codon makes no difference whatsoever.

Effects of a SNiP (cont.)

If a SNiP occurs in a non-coding portion of DNA it can still have an effect by changing (for example) the structure of t-RNA molecules, or the way that genes are regulated.

A single SNiP in a gene for a membrane protein which regulates the movement of substances across the membrane is known to be the cause of the genetic disease cystic fibrosis. In other cases, conditions like osteoporosis are known to be associated with multiple SNiPs at specific locations.

Applications of SNiP Technology

How a disease (such as certain cancers) develops and progresses in each person and how he/she responds to drug treatments can be dependant on certain specific SNiPs. Therefore, analysis of a patient's SNiPs opens up new possibilities for personalised treatment.

Some SNiPs are known to occur commonly in some geographical racial groups compared to others. Analysing a person's DNA for these SNiPs leads to "DNA Profiling" which can be used in forensics, or in the commercial service of analysing a person's possible ethnic origins.

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Understanding Human Evolution

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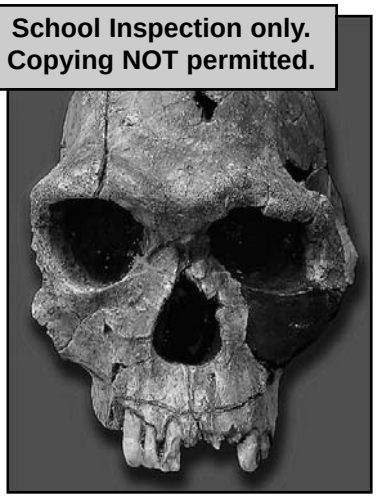
Fossil Evidence

Fossils of possible human ancestors began to be discovered in the 19th century, but these were generally considered to be modern-type humans from a few thousand years ago, who had suffered some horrible disfiguring disease.

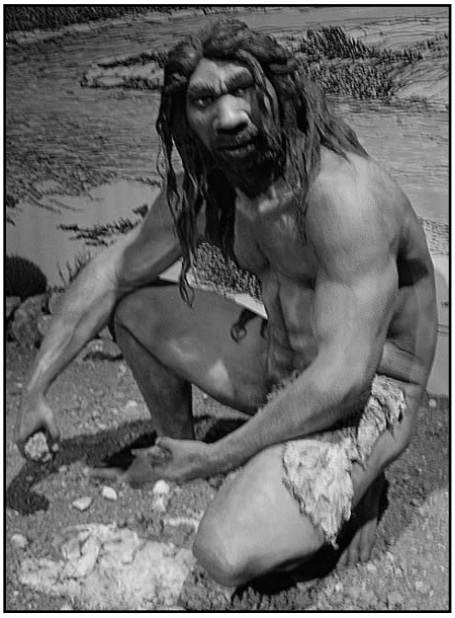
By the 1950's sufficient fossils had been discovered for scientists to begin describing the possible course of human evolution. By that time it was possible to estimate the age of fossils with some certainty, so the different "types" could be placed in chronological order.

The general concensus was that humans and chimpanzees had shared a common ancestor some 10 million years (or more) ago in Africa, where most of the fossils had been found.

To confuse the issue, for most of our prehistoric existence there was more than one distinct genetic type of human co-existing. The best known were the Neanderthal people who disappeared only about 40,000 years ago.



Fossil skull of *Homo habilis*. a probable ancestor from 3.5 MYa



Reconstruction of a Neanderthal.
Image by Jose Luis Martinez Alvarez CCA-SA 2.0

Genetic Evidence

The Human Genome Project and its technology changed our understanding dramatically. When the complete chimpanzee genome was compared to that of humans it was found to be between 95-99% identical, despite a difference in chromosome number. (It is thought that one of our chromosomes was originally 2 chimp chromosomes which fused together.)

Using the known average rate of mutation gives us a "genetic clock". When the human & chimp genomes are compared, the accumulated differences can be used to calculate how long ago the 2 species separated. We now think that proto-humans must have separated genetically (ceased inter-breeding) from proto-chimps between 5-7 MYa.

Dispersal of Modern Humans

Genetic studies, including the occurrence of specific SNiPs, has also given us clues about the spread of modern humans. By comparing SNiPs in the indigenous people in different geographical locations, we get clues about the patterns of migration & inter-breeding in ages past.

One of the most interesting findings is that most people of European or Asian descent possess between 2-4% Neanderthal genes. (The Neanderthal genome has been sequenced from DNA in teeth & bones preserved in cave deposits.) There were always questions about why the Neanderthals disappeared and some suggested that genocide had been committed by our ancestors. It may be more likely that they were simply absorbed into the more numerous modern human population and disappeared by inter-breeding.

In SE Asia, Tibet and especially the islands of Melanesia, modern humans also possess 4-6% of genes from the mysterious Denisovan people. Little is known about the Denisovans except they were a human sub-group with a distinct genome who lived in Asia until about 50,000 years ago.

Now complete Worksheet 14

Mitochondrial DNA & Y-Chromosomal DNA

It seems certain that humans originated in Africa and that there were 4 or 5 waves of migration outwards from there starting as early as about 1.8 Myr BP. The first "out of Africa" migrants were not modern humans, but an ancestral species known as *Homo erectus*. Modern humans are all descended from the wave of migration which began only about 60,000 years ago. We know this from more DNA studies.

As well as DNA in our cell nuclei, there is DNA in the mitochondria. Because your mitochondria are all derived from those in your mother's egg, mt-DNA is passed on through the female line only. Studies of rates of mutation & SNiPs in mt-DNA have allowed scientists to establish that every human on the planet is descended directly (female to female) from a single woman (dubbed by the media "Mitochondrial Eve") who lived in Africa about 150,000 years ago.
In a similar way, the DNA in the Y-chromosome is passed only from father to son. DNA analysis of Y-chromosomes shows that all males descend from a single "Adam" who lived approx. 130,000 years ago, probably in what is now SW Africa, around Namibia.

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