Topic 3: Genetics

3.1 Genes

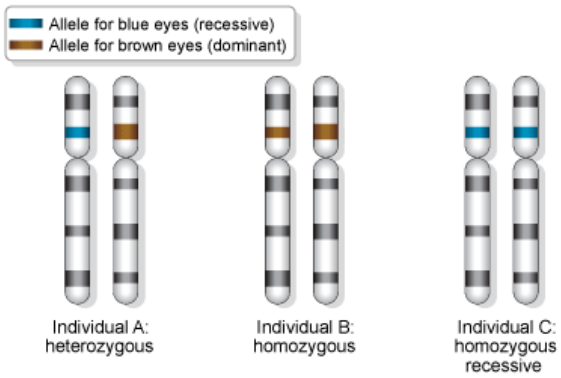
U1	A gene is a heritable factor that consists of a length of DNA and influences a specific characteristic.
U2	A gene occupies a specific position on a chromosome.
U3	The various specific forms of a gene are alleles.
U4	Alleles differ from each other by one or only a few bases.
U5	New alleles are formed by mutation.
U6	The genome is the whole of the genetic information of an organism.
U7	The entire base sequence of human genes was sequenced in the Human Genome Project.
A1	The causes of sickle cell anemia, including a base substitution mutation, a change to the base sequence of mRNA transcribed from it and a change to the sequence of a polypeptide in hemoglobin.
A2	Comparison of the number of genes in humans with other species.

Genes: is a **heritable** factor that controls or influences a specific character consisting of a length of DNA occupying a particular position on chromosome

- Humans have between 21,000-23,000 protein coding genes
- The number of genes in an organism's genome does not indicate how complicated an organism is, for example dogs have larger genome than human
- Each gene occupies a specific location or position on a chromosome called a **locus**.
- Since there are only46 chromosomes in a human diploid cell (23 pairs in females including two X chromosomes and 22 pairs plus and X and a Y chromosome in males). Each chromosome contains many different genes often linked in groups.

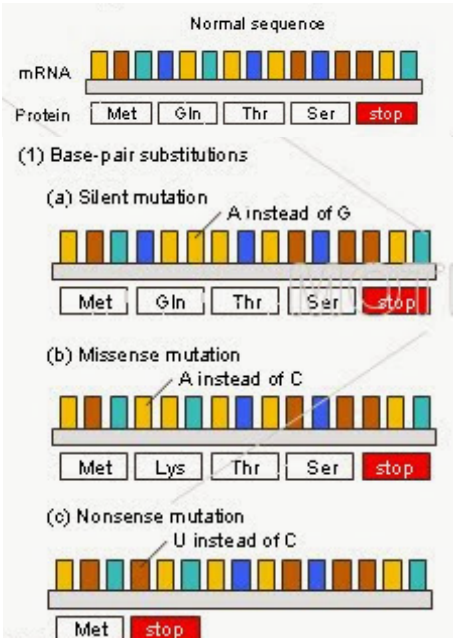
Alleles: one specific form of a gene, differing from other alleles by one or a few bases only and occupying the **same gene locus** as other alleles of the same gene.

- There can be two or more alleles of a specific gene depending on the gene.
- The gene that influences human blood type has three different alleles that code for blood types A, B and O. When there are more than two alleles, this is called multiple alleles.
- Since each human cell consists of **2 copies of each chromosome** (except X and Y), there are two copies of each gene. Sometimes a person can have two of the **same allele (homozygous)** or **two different alleles (heterozygous)**



Gene mutation: is a **permanent change** in the base sequence of DNA. Not all mutation causes disease.

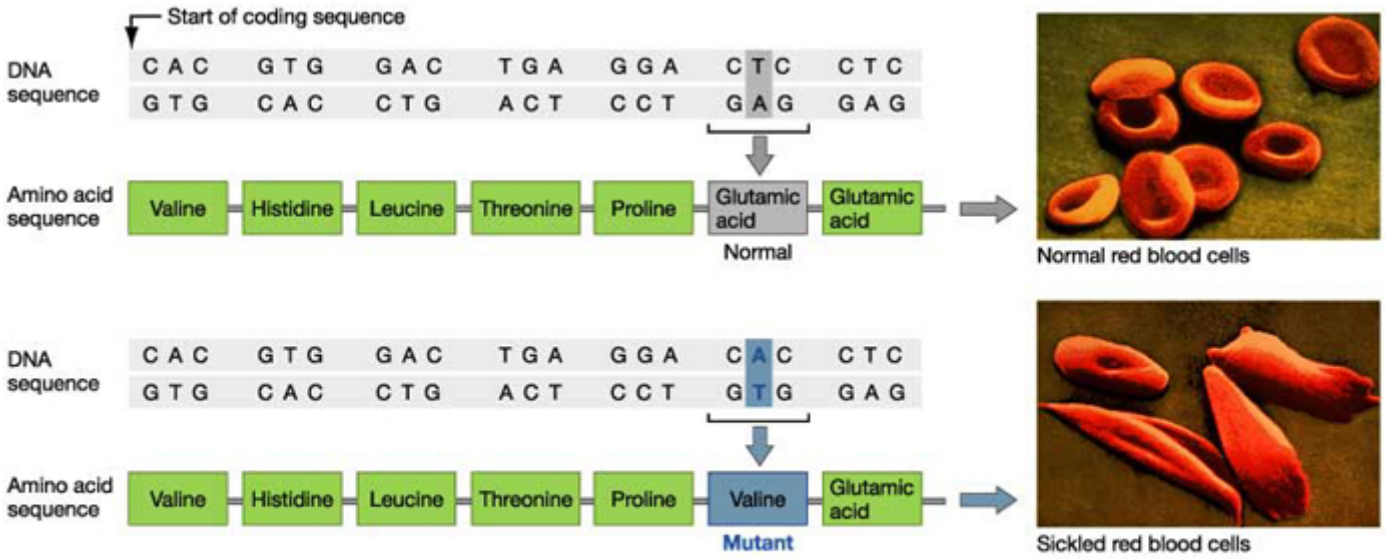
- When one of the bases is changed, this will cause a change in the mRNA sequence when the DNA is copied during **transcription** of the gene.



- **Silence mutation**: a mutation in DNA sequence will not change in protein structure. (multiple codons coded for one amino acid)
- **Missense mutation**: a mutation in DNA sequence leads to a change in protein structure. (change in amino acid sequence)
- **Nonsense mutation**: a mutation in DNA shortens the poly
- peptide chain.(codons mutate to become a STOP signal.)

Sickle-cell anaemia:

- is a disease that causes red blood cells to form a **sickle shape** (half-moon). These sickled blood cells cannot carry as much oxygen as normal red blood cells. They can cause **clots in blood vessels (capillaries)** because of their abnormal shape and inflexibility caused by crystallization of the abnormal hemoglobin.
- Sickle cell anaemia occurs on chromosome 11, happens ob gene **HBB**
- Sickle cell is caused by a base-substitution when the adenine base in GAG is replaced by a thymine base, changing the triplet to GTG.
- **Glutamic acid** then changed to **valine**, a negative charged amino acid changed to neutral one.
- Amino acid sequence change will then lead to a change in protein structure.



The change in amino acid sequence causes hemoglobin molecules to crystallize when oxygen levels in the blood are low. As a result, red blood cells sickle and get stuck in small blood vessels.

- Sickle-cell anaemia gives immune to **malaria**, which is a parasite disease carried by mosquitoes.
- Malaria cannot infect sickle cells. So people with sickle cell trait are resistance to the disease.

Genome: the whole of the genetic information of an organism

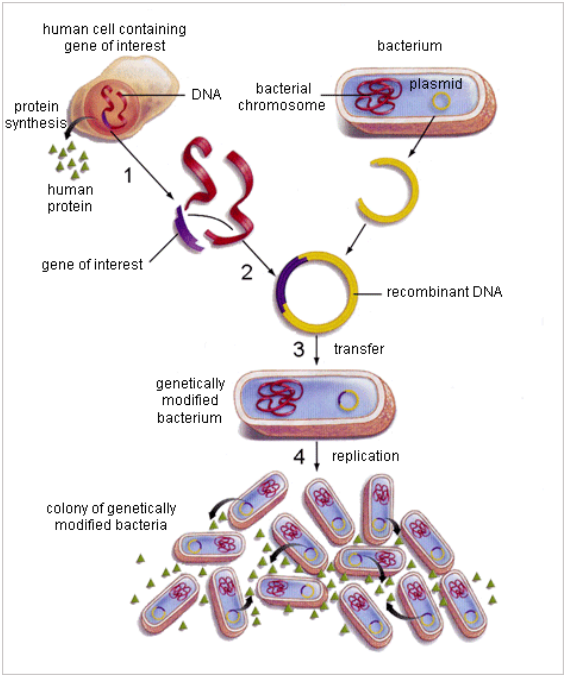
- In humans, the genome consists of 46 chromosomes plus the mitochondrial DNA
- In plants, the genome also consists of chloroplast DNA on top of their chromosomes and mitochondrial DNA
- Prokaryotes have a circular chromosome and plasmids in their genome
- Human Genome Project: **entire base sequence of human genes** was sequenced: Most of the genome does not code for proteins (originally labeled "junk DNA"). Some of these regions consist of areas that can **affect gene expression** or are highly repetitive sequences called satellite DNA. Scientists can now also predict which sequences do code for protein (approximately 21000-23000 sequences)

3.2 Chromosomes

U1	Prokaryotes have one chromosome consisting of a circular DNA molecule.
U2	Some prokaryotes also have plasmids but eukaryotes do not.
U3	Eukaryote chromosomes are linear DNA molecules associated with histoneproteins.
U4	In a eukaryote species there are different chromosomes that carry differentgenes.
U5	Homologous chromosomes carry the same sequence of genes but notnecessarily the same alleles of those genes.
U6	Diploid nuclei have pairs of homologous chromosomes.
U7	Haploid nuclei have one chromosome of each pair.
U8	The number of chromosomes is a characteristic feature of members of aspecies.
U9	A karyogram shows the chromosomes of an organism in homologous pairs ofdecreasing length.
U10	Sex is determined by sex chromosomes and autosomes are chromosomesthat do not determine sex.
A1	Cairns' technique for measuring the length of DNA molecules byautoradiography
A2	Comparison of genome size in T2 phage, *Escherichia coli*,*Drosophila melanogaster*, *Homo sapiens* and *Paris japonica.*
A3	Comparison of diploid chromosome numbers of *Homo sapiens*,*Pan troglodytes*, *Canis familiaris*, *Oryza sativa*, *Parascaris equorum.*
A4	Use of karyograms to deduce sex and diagnose Down syndromein humans.

Prokaryotes:

- Prokaryotes have **circular DNA** without association of protein.
- There is one copy of each gene except when the cell and its DNA are replicating
- Plasmids are small separate (usually circular) DNA molecules located in some prokaryotic cells
- Plasmids are also naked (not associated with proteins) and are not needed for daily life processes in the cell.
- The genes in plasmids are often associated with **antibiotic resistant** and can be **transferred from one bacterial cell to another**.
- Plasmids are readily used by scientists to artificially transfer genes from one species to another (ie. Gene for human insulin)



Insulinproduction in bacteria.

Using DNA ligase and the **same** restriction enzymes.

Plasmid features:

- Naked DNA without association of protein such as histone
- Small circular ring of DNA
- Not responsible for normal life process
- Contain survival characteristics, e.g. antibiotic resistence
- Can be passed on between bacteria
- Can be incorporated into nucleoid chromosomes (save permanently)

Eukaryotes:

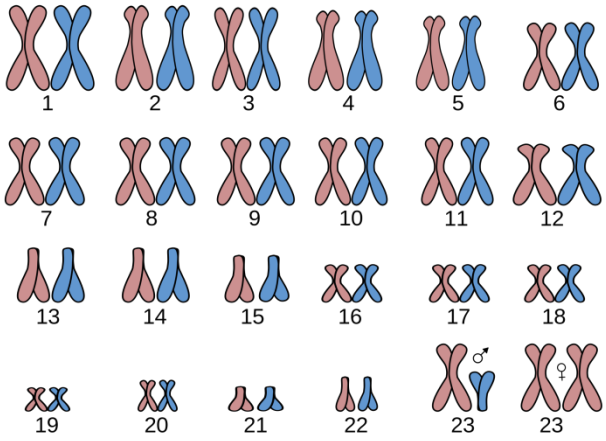
- Eukaryotic chromosomes are linear and are made up of DNA and **histone** proteins.
- Histones are globular shaped protein in which the DNA is wrapped around.
- Linear chromosomes vary in **length**, **centromere** location and **genes** containing
- In humans there are **23** types of chromosomes. There are 22 pairs of **autosomes**. The 23rd pair are the **sex chromosomes**. Males have an X and a Y chromosome and females have two X chromosomes
- Each chromosome carries a specific sequence of genes along the linear DNA molecule. The position where the gene is located is called the **locus**
- The number of chromosomes is known as **N** number.
- Normal cell contains **diploid nucleus – 2N**(two pairs of homologous chromosomes)
- Sex cell contains **haploid nucleus – N**(one pairs of homologous chromosomes)
- The **chromosome number** is an important characteristics of the species

Homologous chromosome:

- Homologous chromosomes are chromosomes within each cell that carry the same genes at the same loci
- One chromosome came from an individual's mother and one from the father
- They have the same **structure and size**
- These chromosomes pair up during meiosis
- Even though these chromosomes carry the same genes, they could have **different alleles**

Sex chromosome:

- The X and Y chromosome determine the sex of an individual
- The X chromosome is quite large in comparison to the Y chromosome and has a centromere that is located near the centre or middle of the chromosome
- The Y chromosome is relatively small with its centromere located near the end of the chromosome
- If an individual has two X chromosomes they will be a female and if they have an X and a Y chromosome they will be a male
- All other chromosomes are called **autosomes** and do not affect the sex of an individual
- **SRY** genes on Y chromosomes lead to male development
- Using a **karyogram**, we distinguish sex, it shows the chromosomes of an organism in homologous pairs of decreasing length.



3.3 Meiosis

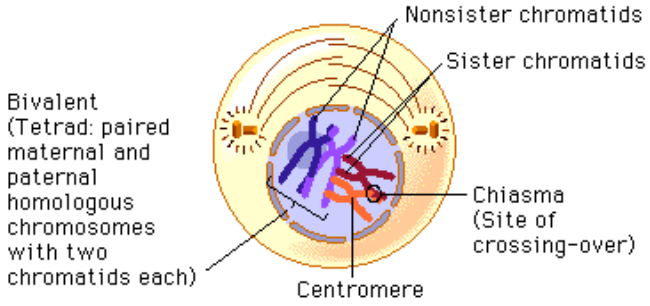
U1	One diploid nucleus divides by meiosis to produce four haploid nuclei.
U2	The halving of the chromosome number allows a sexual life cycle with fusionof gametes.
U3	DNA is replicated before meiosis so that all chromosomes consist of two sister chromatids.
U4	The early stages of meiosis involve pairing of homologous chromosomes and crossing over followed by condensation.
U5	Orientation of pairs of homologous chromosomes prior to separation is random.
U6	Separation of pairs of homologous chromosomes in the first division ofmeiosis halves the chromosome number.
U7	Crossing over and random orientation promotes genetic variation.
U8	Fusion of gametes from different parents promotes genetic variation.
A1	Non-disjunction can cause Down syndrome and other chromosome abnormalities.
A2	Studies showing age of parents influences chances of non disjunction.
A3	Description of methods used to obtain cells for karyotype analysis e.g. chorionic villus sampling and amniocentesis and the associated risks.

Interphase:

- **G1 phase**: increase cytoplasm volume, organelle production and protein synthesis (normal growth)
- **S phase**: DNA replication
- **G2 phase**: increase cytoplasm volume, double the amount of organelle and protein synthesis (prepare for cell division)

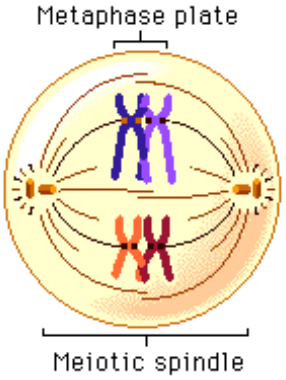
Prophase I:

- DNA supercoils and condenses. Chromosomes are visible under light microscope.
- Nuclear membrane begins to break down and disintegrate.
- The **homologous chromosomes** associate with each other to form **bivalent or tetrads**.
- **Crossing over** occurs: non-sister chromatids exchange genetic information. The crossing over point is called **chaisma** (pl. chaismata)
- **Spindle fiber** begins to form



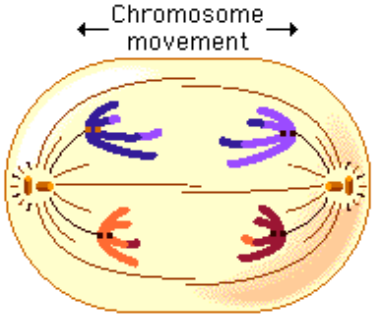
Metaphase I:

- Bivalents line up at the equator
- **Random Orientation** occurs: bivalents (homologous pairs) that come from the mother or the father line up randomly on either side of the cell equator, independently of the other homologous pairs. Hence the daught nuclei get a different mix of chromosomes.
- Spindle fibers (**microtubules**) from each of the centrosomes attach to the centromere of bivalents.



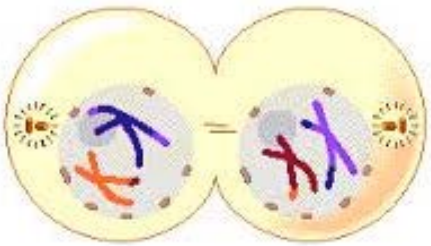
Anaphase I:

- Contraction of the spindle fibers pulls homologous chromosome pair apart.
- Chaismata breaks apart and separate.
- One chromosome of each pair move to opposite poles of the cell.



Telophase I:

- Chromosome begins to uncoil and nuclear envelop reforms.
- Chromosome number reduces from 2n (diploid) to n (haploid); however each chromatid still has the replicated sister chromatid still attached (not **homologous pairs** anymore).
- Cytokinesis occurs and the cell splits into two separate cells.



Prophase II:

- Chromosomes condense again and become visible.
- Spindle fibers again form.
- Nuclear membrane disintegrates again.

Metaphase II

- Chromosomes line up along the equator.
- Spindle fibre attaches to the centromere of the chromosome.

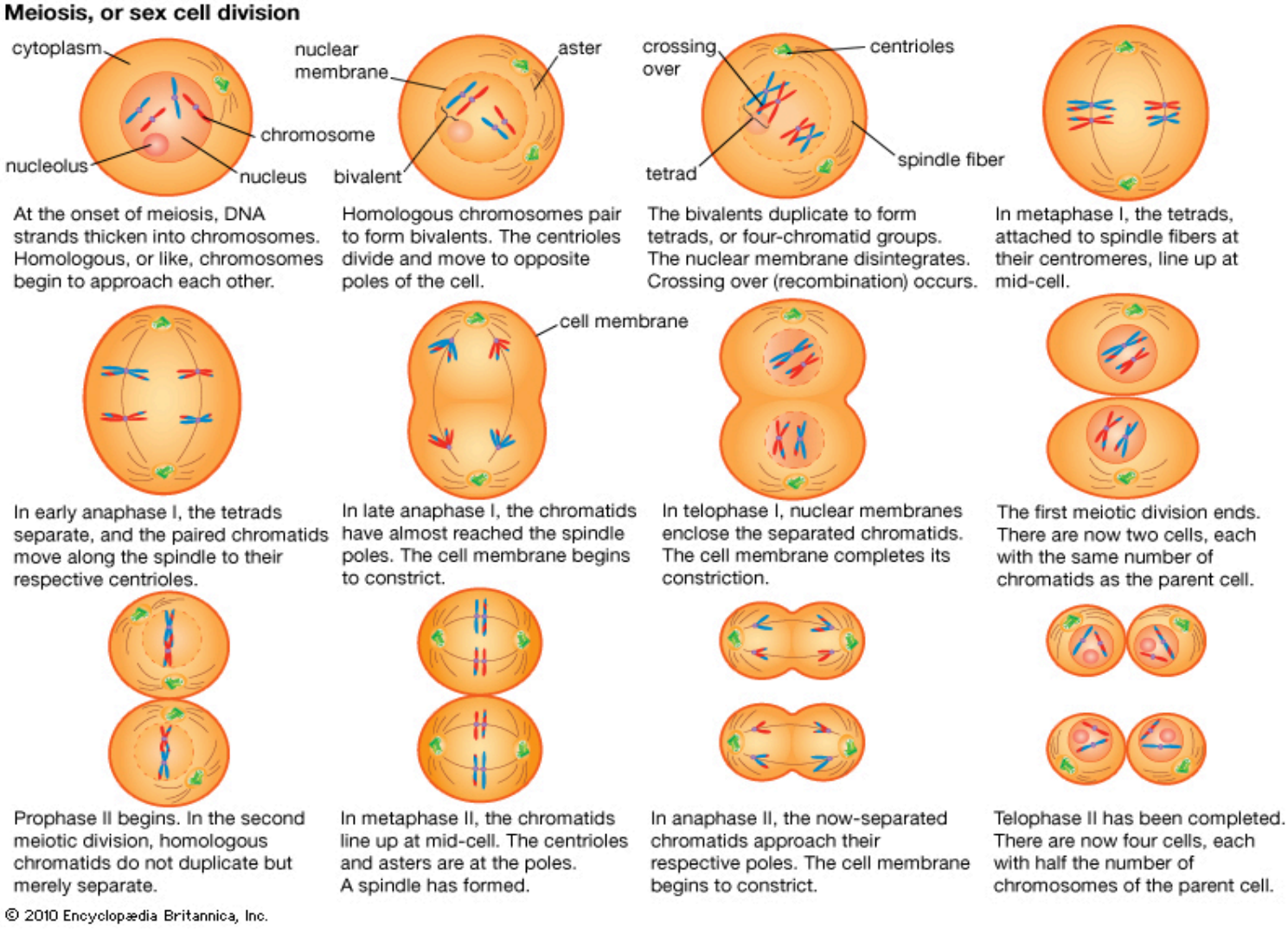
Anaphase II:

- Spindle fibers pull apart the centromeres and sister chromatids are pulled towards the opposite poles.

Telophase II:

- Chromosomes arrive at opposite poles.
- Nuclear envelope begins to develop around each of the four haploid cells.
- Chromosomes begin to unwind to form chromatin.
- Cytokinesis occurs and cells are split apart.

Meiosis, or sex cell division



At the onset of meiosis, DNA strands thicken into chromosomes. Homologous, or like, chromosomes begin to approach each other.

Homologous chromosomes pair to form bivalents. The centrioles divide and move to opposite poles of the cell.

The bivalents duplicate to form tetrads, or four-chromatid groups. The nuclear membrane disintegrates. Crossing over (recombination) occurs.

In metaphase I, the tetrads, attached to spindle fibers at their centromeres, line up at mid-cell.

In early anaphase I, the tetrads separate, and the paired chromatids move along the spindle to their respective centrioles.

In late anaphase I, the chromatids have almost reached the spindle poles. The cell membrane begins to constrict.

In telophase I, nuclear membranes enclose the separated chromatids. The cell membrane completes its constriction.

The first meiotic division ends. There are now two cells, each with the same number of chromatids as the parent cell.

Prophase II begins. In the second meiotic division, homologous chromatids do not duplicate but merely separate.

In metaphase II, the chromatids line up at mid-cell. The centrioles and asters are at the poles. A spindle has formed.

In anaphase II, the now-separated chromatids approach their respective poles. The cell membrane begins to constrict.

Telophase II has been completed. There are now four cells, each with half the number of chromosomes of the parent cell.

Genetic variation:

Crossing over:

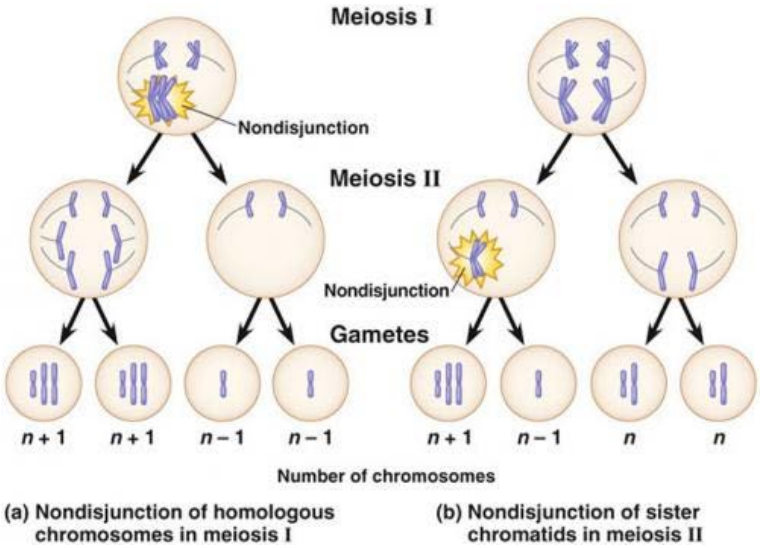
- Occurs in **prophase I** of meiosis.
- Crossing over occurs between non-sister chromatids of a particular chromosome.
- Chiasmata are points where two homologous non-sister chromatids exchange genetic material during crossing over in meiosis.
- Chromosomes intertwine and break at the exact same positions in non-sister chromatids.
- Segments of the adjacent homologues are exchanged during crossing over, therefore the two sister chromatids are no longer identical.
- Crossing over creates new **combinations of linked genes** (genes on the same chromosome) from the mother and the father.
- When the chromatids are separated into different gametes after anaphase II, the gametes produced will not contain the same combination of alleles as the parental chromosomes.
- This creates variation in the offspring regardless of random orientation.

Random Orientation:

- Occurs in metaphase I of meiosis.
- When homologues line up along the equatorial plate in metaphase I, the **orientation of each pair is random**; meaning the maternal or paternal homologue can orient toward either pole.
- This means the number of combinations that can occur in the gamete is $\mathbf{2^n}$(n=number of chromosome pairs).
- Therefore, in a female or male gamete there can be 2^{23} or 8,388,608 different possible combinations.
- Now when you consider there is the same number of possible combinations in the other gamete that it will combine with to form a zygote (random fertilization); the genetic possibilities are staggering.
- If one takes into consideration crossing over, which was explained above, the genetic variation possibilities in the offspring is immeasurable.

Non-disjunction:

- A non-disjunction is an error in meiosis, where the chromosome pairs **fail to split** during cell division.
- Non-disjunction can occur in **anaphase I** where the homologous pairs fail to split, or it can occur in **anaphase II**, where the sister chromatids fail to split.
- The result of this error is too many chromosomes in a gamete cell or too few chromosomes in the final gamete cell.
- One of the gamete cells could have 22 chromosomes and one could have 24 chromosomes. The resulting zygote will therefore have 47 or 45 chromosomes.
- An example of a non-disjunction is **Down's syndrome**.
- Down syndrome occurs when **chromosome 21** fails to separate, and one of the gametes ends up with an extra chromosome 21. Therefore, a child that receives that gamete with an extra chromosome 21 will have 47 chromosomes in every cell.



- Down syndrome is also called **Trisomy 21**.
- Some Down syndrome symptoms include impairment in cognitive ability and physical growth, hearing loss, oversized tongue, shorter limbs and social difficulties.

Karyogram:

- A diagram or photograph of the chromosomes present in a nucleus arranged in homologous pairs of descending length.
- It can be used to make diagnosis of non-disjunction genetic disorder, such as Down's Syndrome.
- **Amniocentesis**: a sample of the amniotic fluid surrounding the baby is removed using a syringe.
- The sample contains skin cell from the baby, so we can use that to make a karyogram, in order to check for genetic disorder.

3.4 Inheritance

U1	Mendel discovered the principles of inheritance with experiments in whichlarge numbers of pea plants were crossed.
U2	Gametes are haploid so contain only one allele of each gene.
U3	The two alleles of each gene separate into different haploid daughter nucleiduring meiosis.
U4	Fusion of gametes results in diploid zygotes with two alleles of each genethat may be the same allele or different alleles.
U5	Dominant alleles mask the effects of recessive alleles but co-dominant alleleshave joint effects.
U6	Many genetic diseases in humans are due to recessive alleles of autosomal genes, although some genetic diseases are due to dominant or co-dominant alleles.
U7	Some genetic diseases are sex-linked. The pattern of inheritance is differentwith sex-linked genes due to their location on sex chromosomes.
U8	Many genetic diseases have been identified in humans but most are veryrare.
U9	Radiation and mutagenic chemicals increase the mutation rate and can causegenetic diseases and cancer.
A1	Inheritance of ABO blood groups.
A2	Red-green colour blindness and hemophilia as examples of sexlinkedinheritance.
A3	Inheritance of cystic fibrosis and Huntington's disease.
A4	Consequences of radiation after nuclear bombing of Hiroshimaand accident at Chernobyl.

Definitions

Genotype:the **combination** of alleles of a gene carried by an organism

Phenotype:the **expression** of alleles of a gene carried by an organism

Homozygous dominant: two copies of the same dominant gene (capital letter AA)

Homozygous recessive: two copies of the same recessive gene (lowercase aa)

Heterozyous: two different alleles (one dominant, one recessive) (Aa)

Codominant: pairs of alleles which are both expressed when present

Carrier: an individual that has one copy of a recessive allele that causes a genetic disease in individuals that are homozygous for this allele.

Test cross: testing a suspected heterozygote by crossing it with a known **homozygous recessive**.

Mendel's pea plants:

- Mendel was known as the father of genetics
- Mendel performed experiments on a variety of different **pea plants**, crossing these varieties by using the male pollen from one variety and transferring it to the female part of another variety
- He collected the seeds and grew them to determine their characteristics
- He then crossed these offspring with each other and also grew their seeds to determine their characteristics
- He continued performing many crosses and recorded his results.

Gametes

- Gametes which are sex cells such as sperm and eggs
- Gametes contain **one set of chromosomes** or one chromosome of each type and are therefore haploid **(n)**
- Since they have only one chromosome of each type, gametes also only contain one allele of each gene
- Together the two gametes form a **zygote**
- When the gametes (n) fuse to form a zygote (2n), **two copies of each gene** exist in the diploid zygote
- The zygote may contain two of the same allele AA or aa or two different alleles such as Aa

mono-hybrid

Aa x Aa

	A	a
A	AA	Aa
a	aA	aa

Phenotype - 3:1
(normal : albino)

Genotype - 1:2:1
(normal : het for albino : albino)

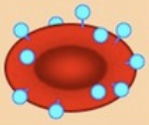
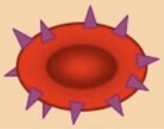
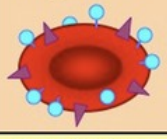
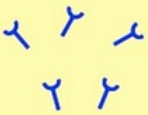
Monohybrid crossing:

- Cross using a **Punnett square**
- F1 generation genotype ratio is 1:2:1 and phenotype ratio is 3:1

ABO Blood Group:

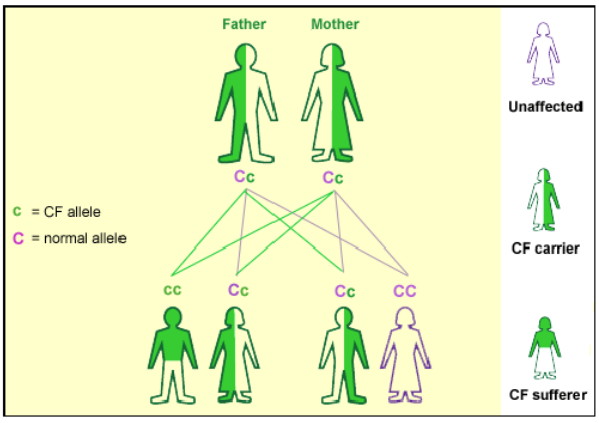
- Human blood types are an example of both multiple alleles (A, B, O) and co-dominance (A and B are co-dominant).
- Co-dominant alleles such as A and B are written as a superscript **(I^A and I^B)**. Blood type O is represented by **(i).**
- Both I^A and I^B are dominant over the allele (i).
- A, B and O alleles all produce a basic **antigen** (glycoprotein) on the surface of the red blood cells
- People with **A** blood group will possess **anti-B antibodies** and **antigen A**, which are able to kill B and AB type blood. A blood group people can accept **A and O** type blood but not the **AB and B** type.
- People with **B** blood group will possess **anti-A antibodies** and **antigen B**, which are able to kill A and AB type blood. B blood group people can accept **B and O** type blood but not the **AB and A** type.
- People with **AB** blood group will possess **NO antibodies** and **antigen A&B**. AB blood group people can accept **ALL type of blood.**
- People with **O** blood group will possess **anti-A & anti-B antibodies** and **NO antigens**, which are able to kill A, B and AB type blood. O blood group people can accept **O** type blood but not the **A, AB and B** type.
- AB blood group is universal receiver; O blood group is universal donor.

ABO Blood Groups

Antigen (on RBC)	Antigen A 	Antigen B 	Antigens A + B 	Neither A or B 
Antibody (in plasma)	Anti-B Antibody 	Anti-A Antibody 	Neither Antibody	Both Antibodies 
Blood Type	**Type A** Cannot have B or AB blood Can have A or O blood	**Type B** Cannot have A or AB blood Can have B or O blood	**Type AB** Can have any type of blood Is the universal recipient	**Type O** Can only have O blood Is the universal donor

Cystic fibrosis:

- Cystic fibrosis is a **autosomal recessive disease** caused by an allele of the **CFTR gene** on **chromosome 7**
- Mutation in the CFTR gene causes secretion of **mucus** to become very thick. Thick mucus **blocks the airway** tubes especially in lungs
- Cystic fibrosis patient dies young, around the age of 35-50



Huntington's disease:

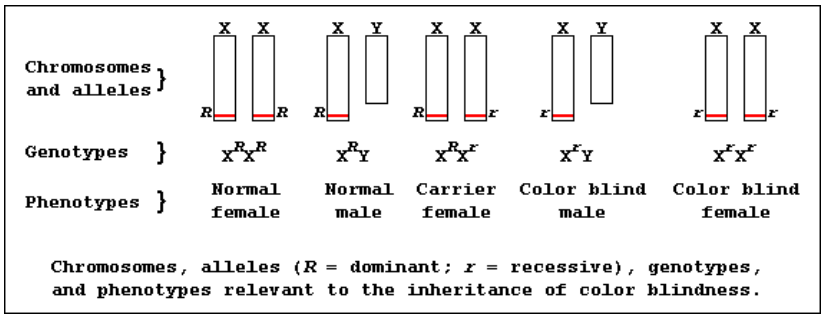
- Humans have two copies of the Huntingtin gene (HTT) on **chromosome 4**, which codes for the protein Huntingtin (Htt)
- Huntington's disease is **dominantly inherited**. Meaning only one bad copy of the gene from either the mother or father will result in Huntington's disease.
- Huntington's disease is a **neurodegenerative genetic disorder** that affects muscle coordination and leads to mental decline and behavioral symptoms
- **Neuron degeneration** will lead to brain disorder, affecting the ability to think, talk and move.

Sex linkage:

- These are patterns of inheritance where the ratios are different in males and females because the **gene is located on the sex chromosomes**
- Generally, sex-linked diseases are on the **X chromosome**
- Sex-linked trait are those which carried on the X chromosomes **non-homologous region**.
- Normal genes are expressed in $\mathbf{X^N}$(dominant)
- Abnormal genes are expressed in $\mathbf{X^n}$ (recessive)
- X-linked recessive diseases such as color blindness and hemophilia are more common in males because males only carry one X chromosome, therefore if they inherit the X chromosome with the disease, they will have the disease.
- Males that have the disease can only pass the colorblind or hemophilia allele onto **their daughters**. Their sons will receive the Y chromosome.

Red-green colour blindness:

- Red-green blindness genes are **recessive** on the non-homologous region of X chromosome Xq28. So it is a **sex-linked disease**.
- Patient's retinal pigment will lose certain frequency so the cannot distinguish between red and green.



Chromosomes, alleles (*R* = dominant; *r* = recessive), genotypes, and phenotypes relevant to the inheritance of color blindness.

Hemophilia:

- Globular protein called **clotting factor** is needed to clot the blood. A mutation will cause clotting factor not to work.
- Clotting response to injury does not work: patient may bleed to death.
- Hemophilia is a **recessive sex-linked disease**.

Genetic disease summary table

Genetic Disease	Allele nature	Location of mutation	Sex-linked	Symptom
Sickle-cell anaemia	**Co-dominant**	HBB genes on chromosome 11 GAG mutate to GUG	Not	clots in blood vessels (capillaries) because of their abnormal shape Immune to malaria
Cystic fibrosis	recessive	CFTR gene on chromosome 7	Not	Thick secretion of mucus causes block of airway in lungs, leading to premature death
Huntington's disease	**dominant**	HTT gene on chromosome 4	Not	Neuron degeneration will lead to brain disorder, affecting the ability to think, talk and move
Red-green colour blindness	recessive	Xq28 gene on X chromosome	Yes	Failure to distinguish between red and blue. Lose certain frequceies of light
Hemophilia	recessive	X chromosome	Yes	Clotting response to injury does not work. Patient may bleed to death

Radiation effects:

- A mutation is a random change to the base sequence of a gene
- Both radiation and certain chemicals can cause **genetic diseases and cancer**
- Radiation can cause mutations if it has enough energy to chemical change one's DNA. Gamma rays and alpha particles from **radioactive** decay, UV radiation and x-rays are all considered to be mutagenic
- Nuclear bombing of Hiroshima and accident at Chernobyl leads to high cancer rate

Mutagens and oncogenes:

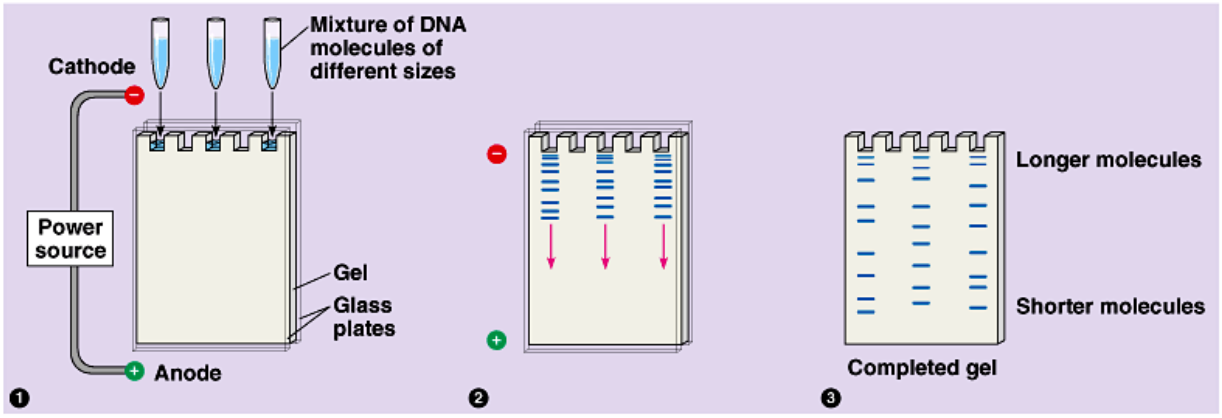
- **Mutagens**: are agents that cause gene mutation such as chemicals, high energy radiation, ultraviolet light and some virus
- **Oncogenes**: genes that control the cell cycle and **cell division**
- If a mutation occurs on oncogenes, it may lead to cancer.
- **Mutation in oncogenes** will lead to malfunction in control of the cell cycle, leading to uncontrolled cell division and cancer.
- Mutation occurs in somatic cells will remain in the organism, but if it occurs in gametes, it will be passed on for generations

3.5 Genetic modification and biotechnology

U1	Gel electrophoresis is used to separate proteins or fragments of DNA according to size.
U2	PCR can be used to amplify small amounts of DNA.
U3	DNA profiling involves comparison of DNA.
U4	Genetic modification is carried out by gene transfer between species.
U5	Clones are groups of genetically identical organisms, derived from a single original parent cell.
U6	Many plant species and some animal species have natural methods ofcloning.
U7	Animals can be cloned at the embryo stage by breaking up the embryo into more than one group of cells.
U8	Methods have been developed for cloning adult animals using differentiated cells.
A1	Use of DNA profiling in paternity and forensic investigations.
A2	Gene transfer to bacteria using plasmids makes use of restriction endonucleases and DNA ligase.
A3	Assessment of the potential risks and benefits associated with genetic modification of crops.
A4	Production of cloned embryos produced by somatic-cell nuclear transfer.

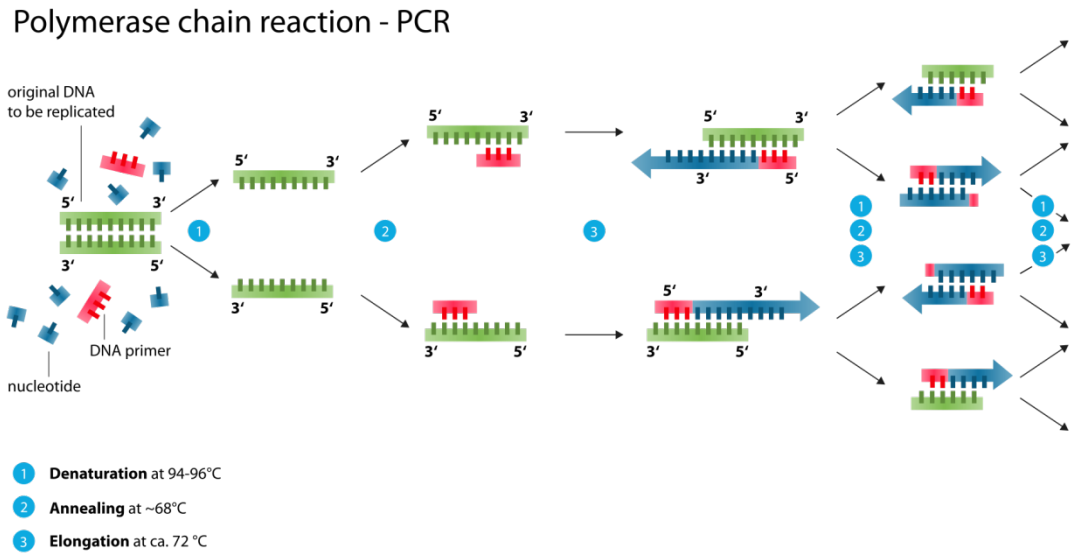
Gel electrophoresis:

- Before gel electrophoresis takes place, **restriction enzymes** are used to cut DNA into fragments of various lengths and different charges.
- **Restriction enzyme**: cut DNA into fragments at specific base sequences in each sample.
- These fragments are placed into small depression or wells at one end of the gel.
- An electrical current is applied to the gel (positive on one side and negative on the other).
- The fragments of DNA will fall out and embed in the gel based on **their size and charge**.
- The smallest particles that are charged go the farthest in the gel, while the large non-charged particles fall out and embed in the gel the quickest.



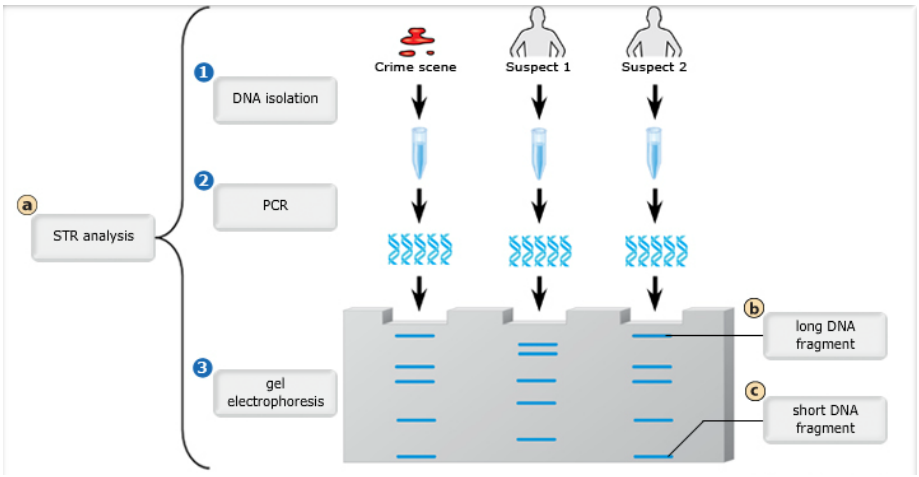
PCR (polymerase chain reaction):

- **PCR (polymerase chain reaction)** is a laboratory technique that takes a single or few copies of DNA and **amplifies** them to generate millions or more copies of a particular DNA sequence.
- When you collect DNA from different sources such as sperm samples or small drops of blood, there are usually very little usable cells to collect DNA.
- Therefore, PCR is used to create enough DNA to be analyzed for investigations such as forensics or custody cases.
- Once large quantities of the DNA have been created, other methods such as gel electrophoresis are used to analyze the DNA.
- **Denaturation**: DNA sample is heated to separate it into two strands
- **Annealing:** DNA primers attach to the opposite ends of the **target gene sequence**
- **Elongation:** A heat-tolerant DNA polymerase (**Taq polymerase**) copies the strand



DNA profiling:

- DNA profiling is a method or technique used to **identify individuals on the basis of their DNA profiles** in comparison to an unknown sample of DNA.
- DNA profiling can be used in **paternity test** to identify the biological father of a child. Scientists can take a blood sample which contains a father's DNA and a blood sample from a child which contains the child's DNA. They can then run a gel electrophoresis to compare the banding patterns between the father and the child.
- DNA profiling can also be used in **criminal investigations** where a small sample of blood, semen, hair or other cells where DNA is present is collected.
- **PCR** can be applied to these small samples of DNA to amplify the DNA into millions of copies to create enough DNA to be analyzed for the investigation.
- Using **restriction enzyme** to cut the DNA into fragments that are separated through **gel electrophoresis** and DNA profiling, the DNA sample can be compared to a suspect's DNA to prove if they are innocent or guilty.
- DNA profiling can also be used to support **ancestral relationships** between organisms for evolutionary studies.
- **Fluorescent marker** may be added to show the colour.



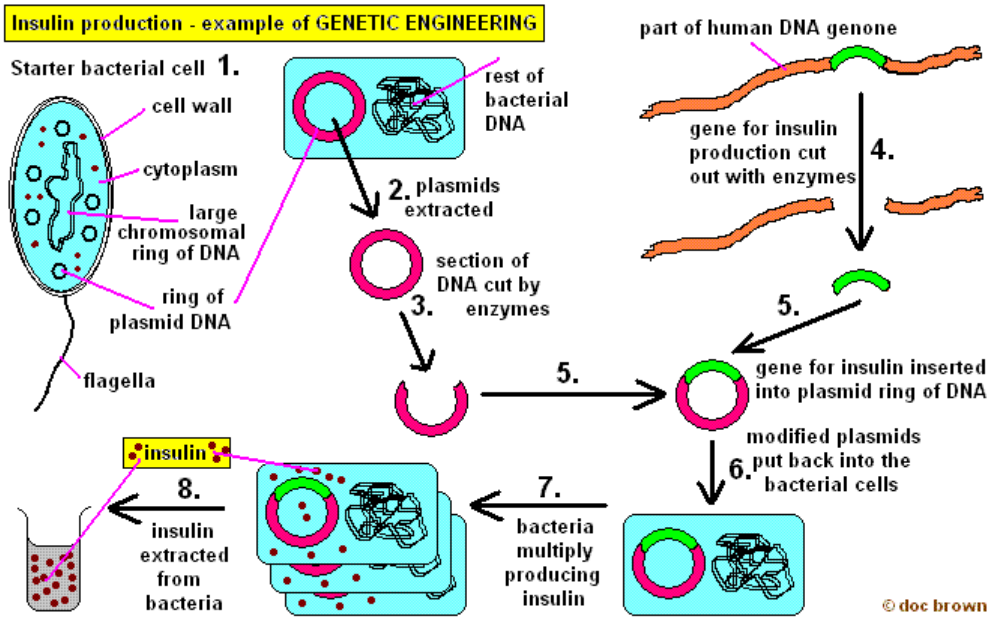
Genetic modification:

- A gene produces a certain polypeptide in an organism.
- Since the genetic code is **universal**, when a gene is removed from one species and transferred to another the sequence of amino acids in the polypeptide produced remains unchanged.

Gene transfer:

- Gene transfer is taking one gene from an organism and inserting it into another organism.
- An example of gene transfer is for the production of **human insulin** produced by the pancreatic cells.
- First, insulin production genes are cut off using **restriction enzyme**.
- Use the **same restriction enzyme** to cut the bacteria plasmid open

- Place the gene into the plasmid using **DNA ligase**. (antibiotic resistance may also be put in to make the plasmid attractive)
- Put the plasmid back in the bacteria.
- Bacteria go through replication and production of human insulin.
- Harvest and purify the insulin.



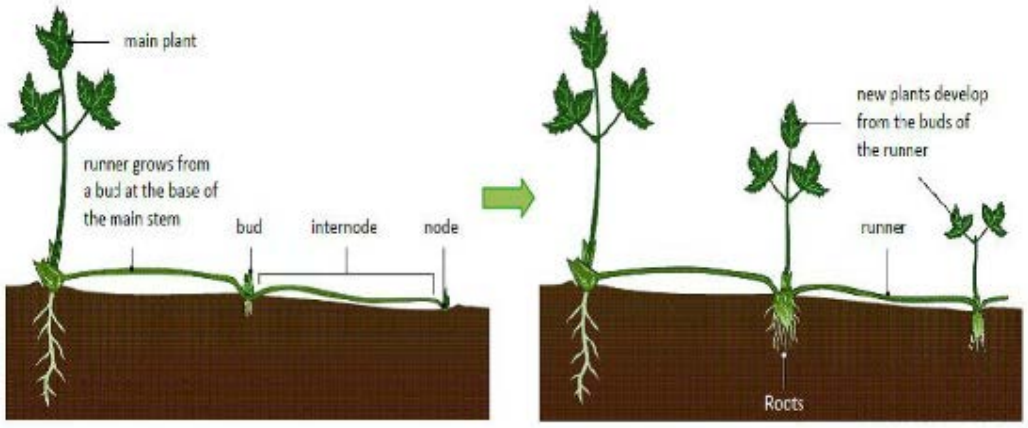
Potential risks and benefits of genetic modified crops:

- **Bt corn**: combine with soil bacterium that produces insecticidal toxin – give resistance to insects

Potential Benefits	Potential Risks
Higher crop **yield** (more production = more money) - crop yield is a debatable benefit	Long term effects on humans are unknown
Less or **no pesticides** used because already resistant to harmful pests	Non-targeted organism will be affected by the toxin
Can use pest resistant crops or modified crops in areas where water availability is limited	Transfer gene could **mutate**
Could add genes for certain proteins, vitamins or possible vaccines (less cost than producing in a lab)	Increased resistance to toxin **eolves** in pest
Crops last longer or don't spoil during storage	Accidental release may result in competition with native species
Increased disease resistance	Biodiversity reduced
Increased hardiness: grow in more locations/seasons	Super weed may appear

Clone:

- Clone: **a group of genetically identical organisms** or a group of cells derived from a single parent cell.
- Organisms that reproduce asexually, produce genetically identical offspring
- Identical twins in humans are also clones (monozygotic twin)
- Bacteria uses binary fission to clone itself
- Underground stems called **tubers** in potatoes can form new potato plants which are clones of the original parent potato plant
- **Runner**: growing stems used to reproduce asexually



Other natural methods of vegetative propagation include specialised underground stems forming **tubers** – stems which become swollen full of nutrient molecules – from which new plants can grow. Potatoes grow in this way.

Embryonic stage cloning:

- At the very early embryo stage, cells are still **pluripotent** (meaning they can become any type of tissue)
- These cells can be separated artificially in a laboratory in order to create **more than one of the same organism**
- The separated pluripotent cells can then be inserted into the uterus of a surrogate mother or mothers in order to produce genetically identical offspring
- The separation of cells has to happen early in development, preferably the 8 cell stage

Cloning differentiated cells:

- Once cells start to differentiate and embryos develop into a fetus and eventually an adult cloning becomes much more difficult
- Therapeutic cloning is an example of cloning using differentiated cells
- This type of cloning can be used to **create a specific tissue or organ**
- Cloning using differentiated cells can also be used to reproduce organisms like dolly the sheep. This is done through **somatic-cell nuclear transfer.**

1. Remove a differentiated cell **nucleus**
2. **Enucleate** a donor egg cell
3. Insert nucleus into the cell
4. Treat with electricity and put in back to the womb
5. Produce genetically indentical organism