Topic 3: Genetics

3.1 Genes

∑ Understandings:

∑ - A gene is a heritable factor that consists of a length of DNA and influences a specific characteristic.

- Gene: The basic unit of heredity or a heritable factor that controls a specific characteristic.
- DNA consists of the base pairs adenine, guanine, cytosine and thymine
- 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, as indicated by the table below.

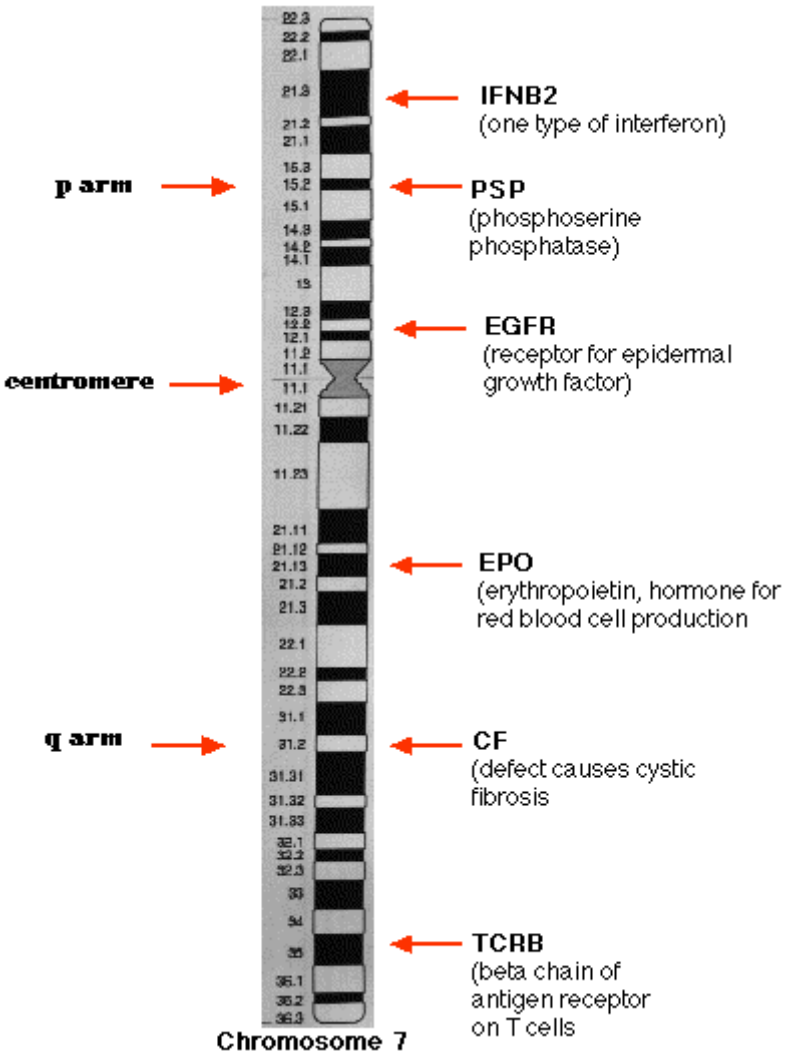
How many genes do other organisms have?

	chromosomes -- diploid	**base pairs**	**genome size (#genes)**	**Reference**
fruit fly	8	$1.65x10^8$	13,600	ref
Budding yeast	16	12,462,637	6,275	ref
human	46	3.3×10^9	~21,000	ref
human mitochondria		16,569	13	ref
rice	24	4.66×10^8	46,022 -55,615	ref
dog	78	2.4×10^9	~25,000	ref
mouse	40	3.4×10^9	~23,000	ref

http://www.edinformatics.com/math_science/human_genome.htm

∑ - A gene occupies a specific position on a chromosome.

- Each gene occupies a specific location or position on a chromosome called a locus (plural loci)
- Since there are only 46 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.



Example of human chromosome 7 fromhttp://users.rcn.com/jkimball.ma.ultranet/BiologyPages/C/Chromo7.gif

∑ **- The various specific forms of a gene are alleles.**

- **Allele**: 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)

∑ **- Alleles differ from each other by one or only a few bases.**

- Genes consist of a certain sequence of DNA bases which can be 100's to 1000's bases in length
- Usually different alleles of the gene vary by only one to a couple of different bases.
- For example, the allele for Sickle Cell Anemia is created by a mutation of a single nucleotide.
- Adenine is switched to Thymine (GAG to GTG) which results in glutamic acid being substituted by valine at position 6 in the Haemoglobin polypeptide.
- This variation when one nucleotide is switched for another is called a single nucleotide polymorphism (SNPs for short)

β - Application: Comparison of the number of genes in humans with other species.

	chromosomes -- diploid	**base pairs**	**genome size (#genes)**	**Reference**
fruit fly	8	$1.65x10^8$	13,600	ref
Budding yeast	16	12,462,637	6,275	ref
human	46	3.3×10^9	~21,000	ref
human mitochondria		16,569	13	ref
rice	24	4.66×10^8	46,022 -55,615	ref
dog	78	2.4×10^9	~25,000	ref
mouse	40	3.4×10^9	~23,000	ref

http://www.edinformatics.com/math_science/human_genome.htm

✓ **Skill: Use of a database to determine differences in the base sequence of a gene in two species.**

- Go to http://www.ncbi.nlm.nih.gov/ and follow the procedure of comparing two gene sequences in your textbook on page 144.
- Do that data-based questions on page 145

∑ - New alleles are formed by mutation.

- As stated above, new alleles are created by random changes in the base sequence called mutations.
- There are a variety of different types of mutations that can be either harmful, neutral or beneficial

Applications and skills:

β - Application: 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.

- A mutation that causes the replacement of a single base nucleotide with another nucleotide in DNA.
- 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.
- This change in the mRNA sequence may change the amino acid in the polypeptide coded for by the gene; in the process of translation.
- Sickle-cell anemia 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 because of their abnormal shape and inflexibility caused by crystallization of the abnormal hemoglobin.
- 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.
- The normal triplet when transcribed and translated codes for the amino acid glutamic acid.
- When the base substitution occurs, the amino acid that is translated is now valine.
- Since valine has a different shape and charge, the resulting polypeptide's shape and structure changes.
- As a result, hemoglobin's shape will change, as does the shape of the red blood cell, resulting in the problems associated with sickle cell anemia listed above.

Good link on malaria and sickle cell anemia in Africahttps://www.youtube.com/watch?v=1fN7rOwDyMQ

∑ - The genome is the whole of the genetic information of an organism.

- **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

∑ - The entire base sequence of human genes was sequenced in the Human Genome Project.

- What they found: 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)

- A complete description of the Human genome project can be found athttp://www.genome.gov/10001772

Notes from the IBO

Guidance:

• Students should be able to recall one specific base substitution that causes glutamic acid to be substituted by valine as the sixth amino acid in the hemoglobin polypeptide.

• The number of genes in a species should not be referred to as genome size as this term is used for the total amount of DNA. At least one plant and one bacterium should be included in the comparison and at least one species with more genes and one with fewer genes than a human.

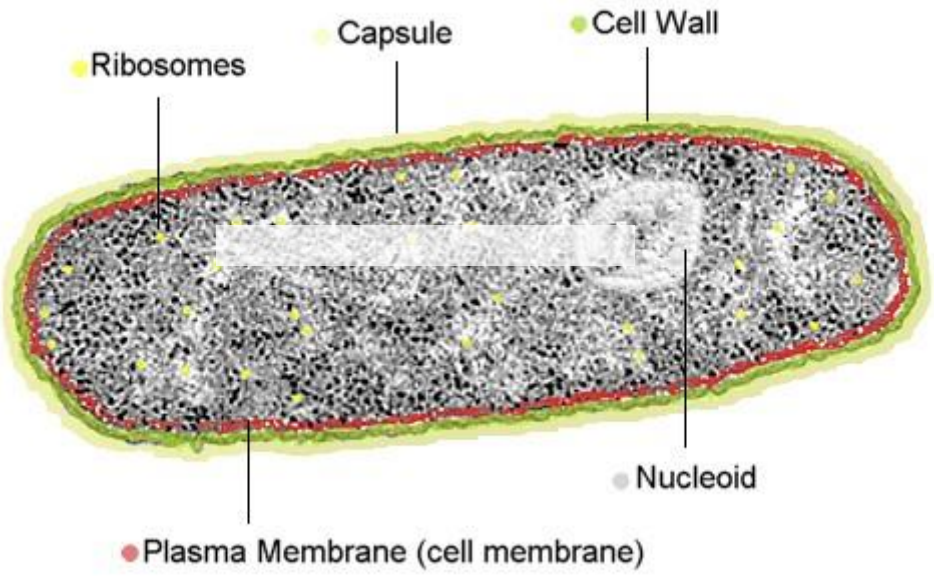
• The Genbank® database can be used to search for DNA base sequences. The cytochrome C gene sequence is available for many different organisms and is of particular interest because of its use in reclassifying organisms into three domains.

• Deletions, insertions and frame shift mutations do not need to be included

3.2 Chromosomes

∑ **Understandings:**

∑ **- Prokaryotes have one chromosome consisting of a circular DNA molecule**.



http://dtc.pima.edu/blc/182/lesson4/prokaryotes/prokaryotespage1.htm

- As you can see in the diagram, the DNA above called the nucleoid region is circular DNA which, unlike eukaryotes, is not associated with any histone proteins
- There is one copy of each gene except when the cell and its DNA are replicating

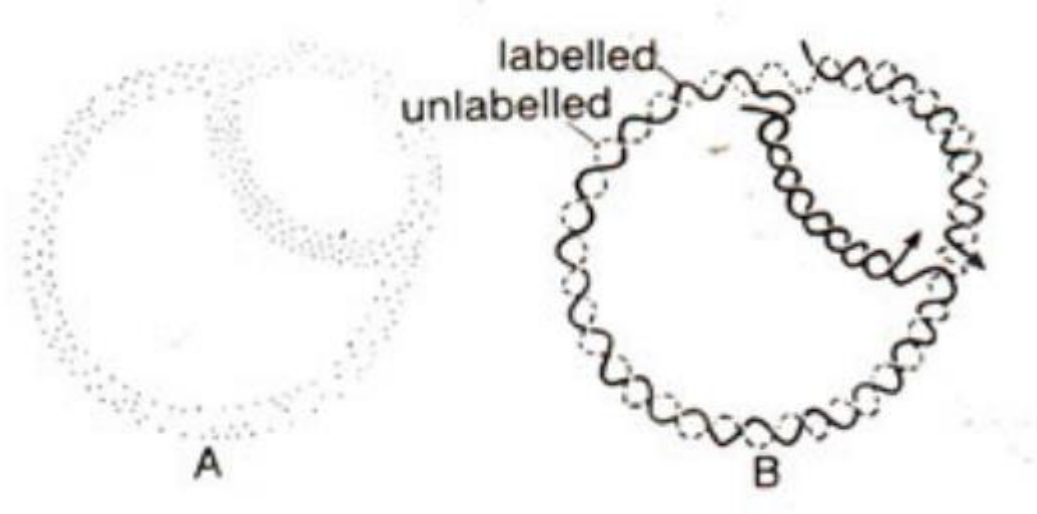
∑ - **Some prokaryotes also have plasmids but eukaryotes do not.**

- 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)

Applications and skills:

β - Application: Cairns' technique for measuring the length of DNA molecules by autoradiography.

- Semi-conservative mode of replication of bacterial chromosome was also demonstrated by-J. Cairns
- Using the technique of autoradiography Cairns first supplied the cells with suitable radioactive material like tritiated thymidine (H3-TdR)
- H3 is heavy isotope of hydrogen and it replaces normal hydrogen in thymidine to give rise to tritiated thymidine).
- This used because this will selectively label only DNA and will not label RNA, since the thymine base is absent in RNA. The tritiated thymidine gets incorporated into DNA and replaces ordinary thymidine.
- The cellular material is then sectioned or else the cells may be broken down to release the intact bacterial chromosomes on slides. These slides are then covered by photographic emulsion and stored in dark.
- During this storage the particles emitted by tritiated thymidine will expose the film, which can be developed. This photograph will then show the regions of the presence of tritium and thus indirectly show the presence of labelled DNA.
- The results showed that autoradiographs from this replicating material prepared at regular known intervals demonstrated **semi-conservative mode of replication.**
- In the figure below, one of the two strands in the daughter DNA molecules is derived from the parent molecule and the other is newly synthesized. In θ shaped figure, which is obtained in the second cycle of replication in presence of label, two arcs in the split region would never be equally labelled. For instance, one arc would be twice as heavily labelled as the other arc. This is what was actually observed by Cairns. The observations thus clearly supported the semi-conservative nature of replication.



http://www.eplantscience.com/index/genetics/chemistry_of_the_gene_synthesis_modification_and_repair_of_dna/semiconservative_dna_replication_in_e_coli.php

∑ - Eukaryote chromosomes are linear DNA molecules associated with histone proteins.

- 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.
- DNA wrapped around 8 histone proteins is called a nucleosome.
- The DNA wraps twice around the histone protein core.
- Another histone protein is attached to the outside of the DNA strand. This helps maintain the colloidal structure of the nucleosome.
- DNA, because of its negative charge is attracted to the positive charge on the amino acids of the histone proteins.

∑ - In a eukaryote species there are different chromosomes that carry different genes.

- Eukaryotic chromosomes are linear chromosomes that vary in length and in position of the centromere that holds the sister chromatids together
- 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
- All eukaryotic species contain at least two different chromosomes, but most contain more than only two

∑ - Homologous chromosomes carry the same sequence of genes but not necessarily the same alleles of those genes.

- Homologous chromosomes are chromosomes within each cell that carry the same genes

- One chromosome came from an individual's mother and one from the father
- They have the same shape and size
- These chromosomes pair up during meiosis
- Even though these chromosomes carry the same genes, they could have different alleles

Do the data based questions on page 153 on comparing the chromosomes of mice and humans

β - Application: Comparison of genome size in T2 phage, *Escherichia coli*,

- Drosophila melanogaster, Homo sapiens and Paris japonica.

T2 Phage		
E. coli		
D. melanogaster		
H. sapiens		
P. japonica		

✓ Use of online databases to identify the locus of a human gene and its protein product.

- Follow the instructions on page 154 of your text to find the location and the descriptions of the 5 genes suggested. Also choose 3 other genes (can find on the internet) and find their location and description as well.
- http://www.ncbi.nlm.nih.gov/gene/?term=drd4

Gene	Description	Location
DRD4		
CFTR		
HBB		
F8		

TDF		

∑ - Diploid nuclei have pairs of homologous chromosomes.

- Diploid nuclei have two copies of each type of chromosome. One chromosome comes from the mother and one from the father.
- Haploid gametes (sperm and egg) fuse during sexual reproduction which produces zygote with a diploid nucleus
- This cell will then divide by mitosis to produce numerous cells, all with a diploid nucleus
- Each nucleus has two copies of each gene, accept the sex chromosomes

∑ - Haploid nuclei have one chromosome of each pair.

- Haploid nuclei have one copy of each chromosome or one full set of the chromosomes in that particular species eg. Human 23 chromosomes
- These are called gametes, which are sperm and egg
- Human sperm and eggs each contain 23 chromosomes

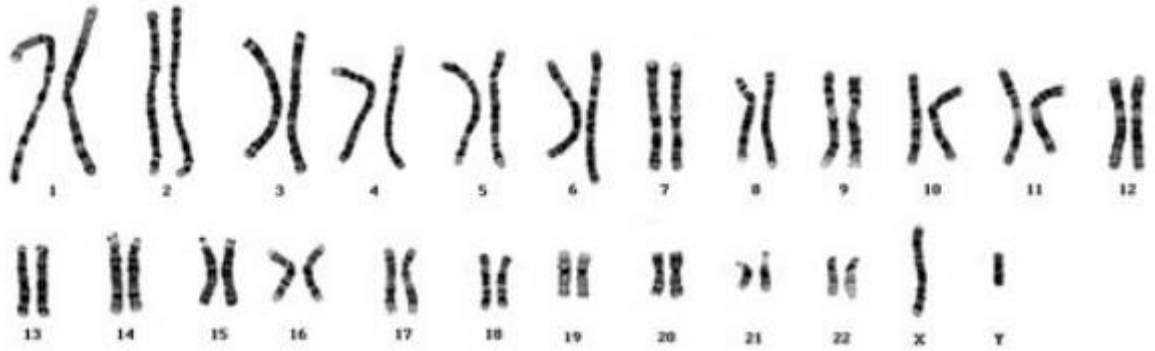
∑ - The number of chromosomes is a characteristic feature of members of a species.

- Chromosome number is a characteristic feature of that species.
- Chromosome number does not indicate how complicated an organism might be
- Organisms with different numbers of chromosomes would unlikely be able to interbreed
- Chromosome number tends to remain unchanged over millions of years of evolution; however, sometimes through evolution chromosomes can fuse together or split to change the number of chromosomes an organism contains

∑ - A karyogram shows the chromosomes of an organism in homologous pairs of decreasing length.

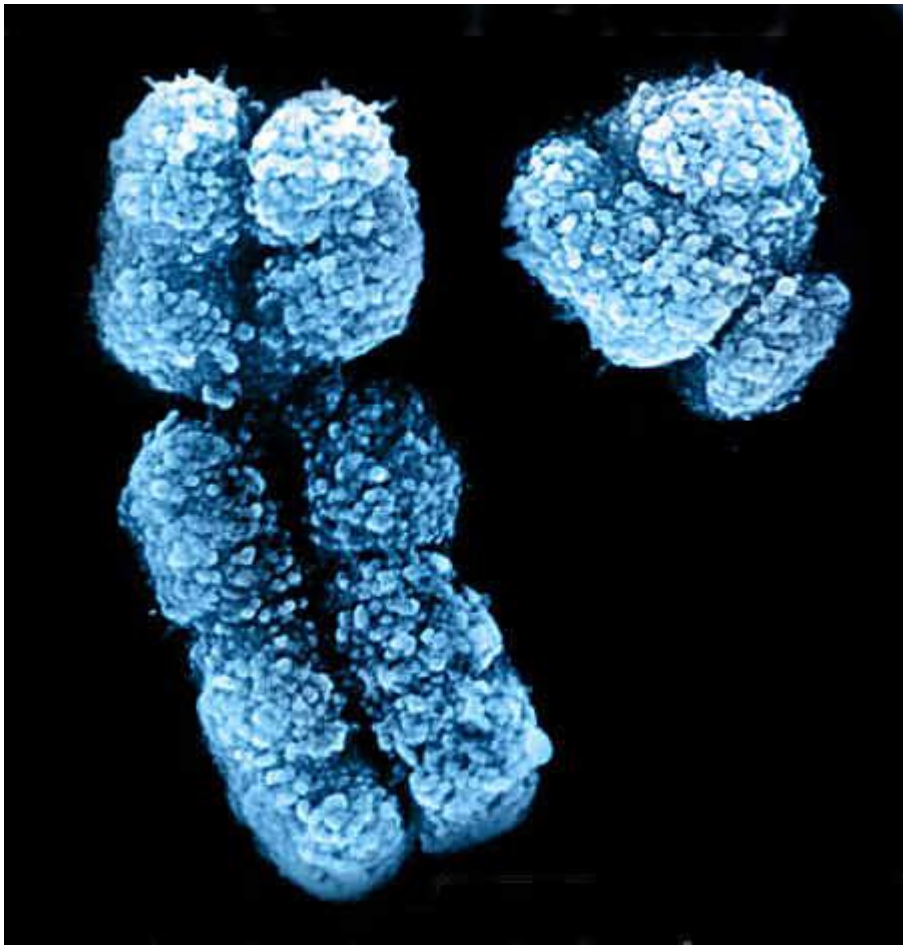
- In karyotyping, chromosomes are arranged in pairs according to their size and structure with the largest at chromosome pair 1 and the smallest at chromosome 22.

- Chromosomes are stained during mitosis (generally in metaphase) in order to see the chromosomes, and a micrograph is taken of the stained chromosomes
- This stained image of the chromosomes is called a Karyogram
- The 23rd pair are the sex chromosomes. Females have two X chromosomes and males have one X chromosome and one Y chromosome.



∑- Sex is determined by sex chromosomes and autosomes are chromosomes that do not determine sex.

- 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
- The X chromosome has many genes located on it essential to human development, while the Y chromosome has a small number of genes (some of these are shared with the X chromosome). The rest of the genes on the Y chromosome are only necessary for male development



X and Y Chromosome

http://blogs.ucl.ac.uk/clinical-molecular-genetics/files/2014/05/chromosome_sem.jpg

- A specific gene only on the Y chromosome called the SRY gene codes for a protein called the testis-determining factor (TDF). The TDF is a DNA-binding protein or regulatory protein that is responsible for the initiation of male sex determination in humans

3.3 Meiosis

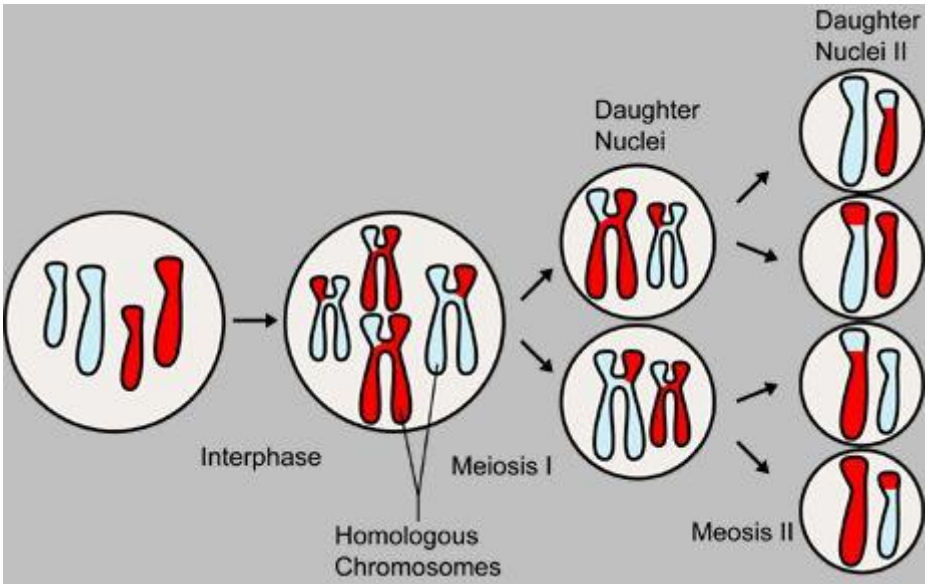
Essential idea: Alleles segregate during meiosis allowing new combinations to be formed by the fusion of gametes.

∑ **- Understandings:**

∑ **- One diploid nucleus divides by meiosis to produce four haploid nuclei.**

- Meiosis is the process in which the diploid (2n) nucleus divides to form four haploid (n) nuclei
- Meiosis has two divisions called Meiosis I and Meiosis II

- In the first division the diploid nucleus 2n, which consists of homologous pairs of chromosomes (half maternal and half paternal chromosomes), divides to form two haploid cells (n). These cells after the first division are considered haploid because the homologous pairs of the nucleus are separated into the two new cells.
- In meiosis II, the haploid chromosomes in the two cells (each have 2 chromatids because replication occurs before meiosis takes place) divide to form four haploid cells each with one set of chromosomes
- This is called reduction division because the chromosome number is halved



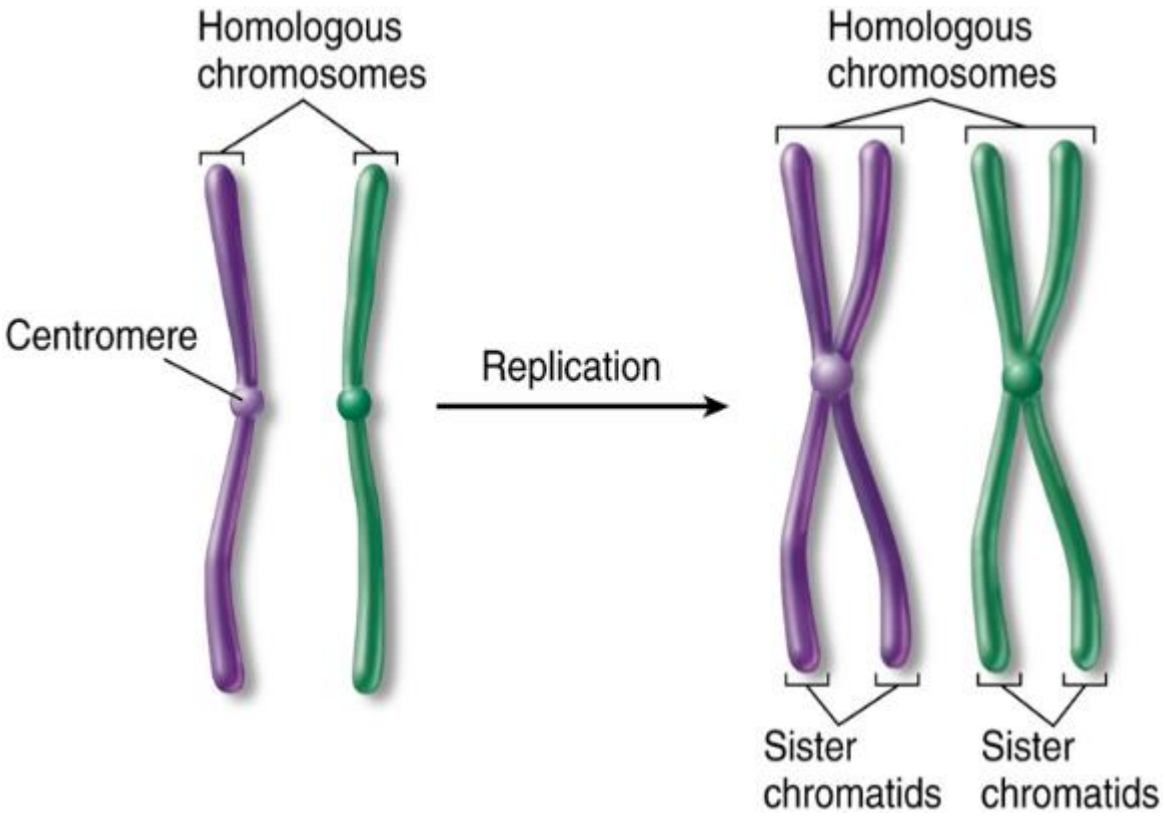
∑ - The halving of the chromosome number allows a sexual life cycle with fusion of gametes.

- During sexual reproduction there is the fusion of two gametes to form a new cell with double the number of chromosomes. The fusion of the gametes takes place during fertilization.
- If an organism did not reduce or half the number of chromosomes during meiosis before fertilisation took place, the new cell would contain double the number of chromosomes in comparison to the original cell.
- This means there would be a doubling of chromosomes with each new generation or sexual life cycle.
- This is why reduction division during meiosis is essential for the sexual life cycle to occur in eukaryotes.
- This also creates genetic diversity as the alleles on the chromosomes from each parent might be different.
- In prokaryotes asexual division occurs given rise to offspring that are genetically identical to their parents.

Do data based question on page 161.

∑ - DNA is replicated before meiosis so that all chromosomes consist of two sister chromatids.

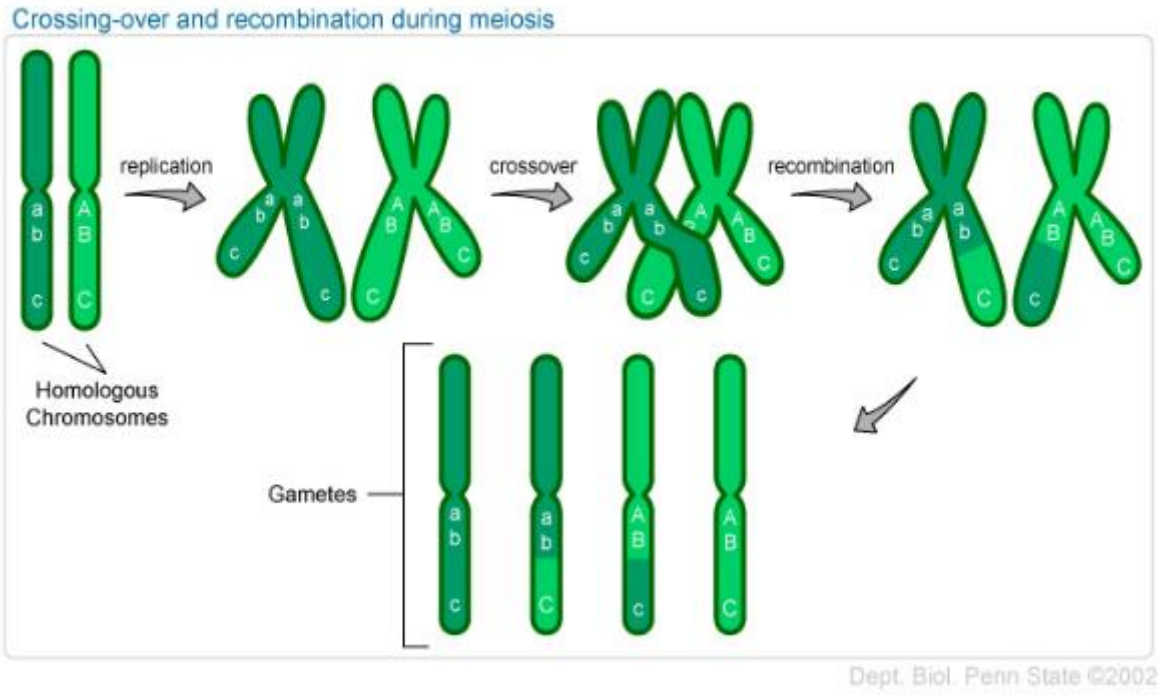
- Chromosomes are replicated in the synthesis (S) phase during interphase
- This means that each chromosome will have an attached identical copy before meiosis occurs
- These are called sister chromatids



www.zo.utexas.edu

∑ - The early stages of meiosis involve pairing of homologous chromosomes and crossing over followed by condensation.

- At the start of meiosis (prophase I), the replicated chromosomes begin to condense and become visible.
- Homologous chromosomes synapse (pair up) to form **bivalents** or tetrads.
- Crossing over occurs between non-sister chromatids. Crossing over occurs when two of the *non-sister chromatids* exchange a segment of their chromosome with each other. Since the genes between the two chromosomes are the same, but the alleles may differ between the maternal and paternal chromosome, a new combination of alleles will be present when the chromosomes separate.
- These crossover points are random and lead to genetic variation in the gametes



∑ - Orientation of pairs of homologous chromosomes prior to separation is random.

- 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 towards either pole.
- The two homologous chromosomes in each bivalent is attached to a different spindle fibre, randomly attaching them to either pole
- The orientation of how one set of chromosomes lines up has no effect on the other bivalents (i.e. The bivalent formed for chromosome 1, does not affect how the bivalent for chromosome 2 will orient)
- This means the number of combinations that can occur in the gamete is 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

∑ - Separation of pairs of homologous chromosomes in the first division of meiosis halves the chromosome number.

- In meiosis I, homologous chromosomes split, but the centromeres do not divide since the sister chromatids do not separate
- One chromosome from each pair separate and migrate towards separate poles. This separation is called a disjunction.

- This halves the chromosome number of each cell and is therefore called reduction division. The two new cells formed after the first division are haploid (n)

β - Applications and skills:

β - Application: Description of methods used to obtain cells for karyotype analysis e.g. chorionic villus sampling and amniocentesis and the associated risks.

- Karyotyping is performed by collecting cells using one of two methods;chorionic villus sampling or amniocentesis.
- Karyotyping is used for pre-natal diagnosis of chromosome abnormalities such as Down syndrome (Trisomy 21), Turner syndrome (XO), and Klinefelter syndrome (XXY).
- The cells obtained by chorionic villus sampling and amniocentesis come from the embryo and not the mother, allowing doctors to analyze the DNA genome of the embryo.
- Amniocentesis procedure involves the extraction of a small amount of amniotic fluid (contains fetal tissues) with a needle, from the amnion or amniotic sacsurrounding a developing fetus. The fetal DNA is examined for genetic abnormalities through karyotyping.
- Chorionic villus sampling involves removing a sample of the chorionic villus(placental tissue) to test for genetic abnormalities through karyotyping. CVS can be carried out 8-12 weeks into the pregnancy.

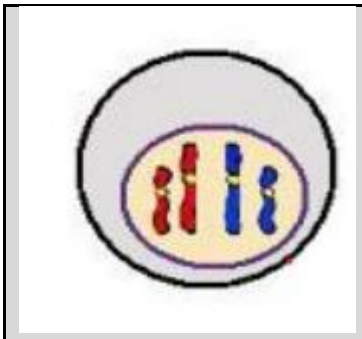
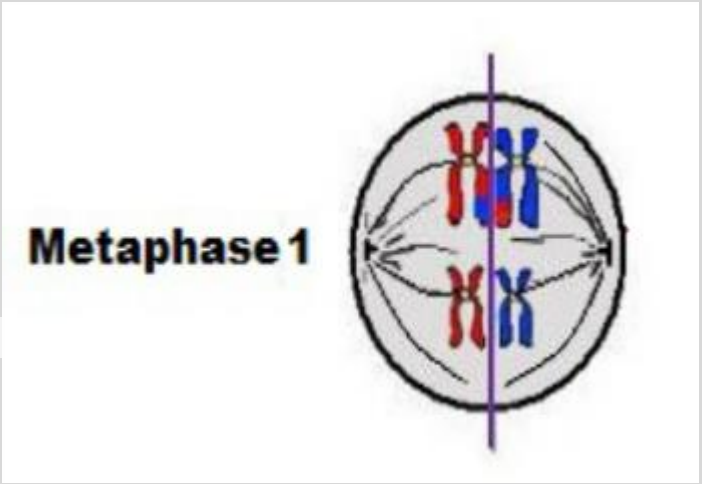
β - Skill: Drawing diagrams to show the stages of meiosis resulting in the formation of four haploid cells.

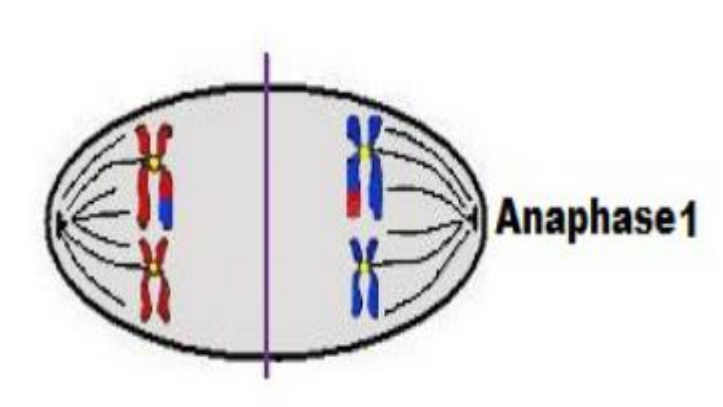
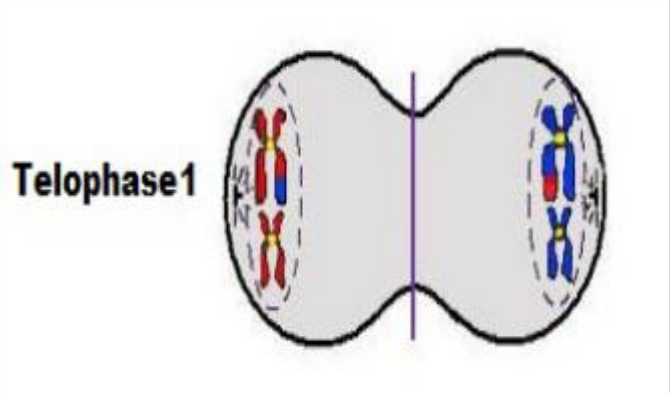
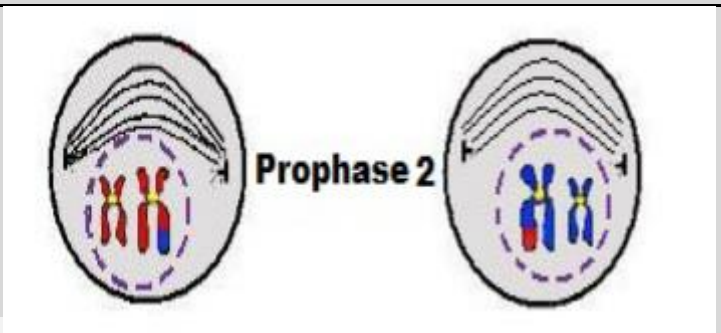
Good Animation - https://www.youtube.com/watch?v=rqPMp0U0HOA

Meiosis

- Meiosis is the process of reduction division in which the number of chromosomes per cell is halved.
- Meiosis results in the production of haploid gametes and is essential for reproduction.
- Meiosis occurs in two stages; meiosis I and meiosis II.

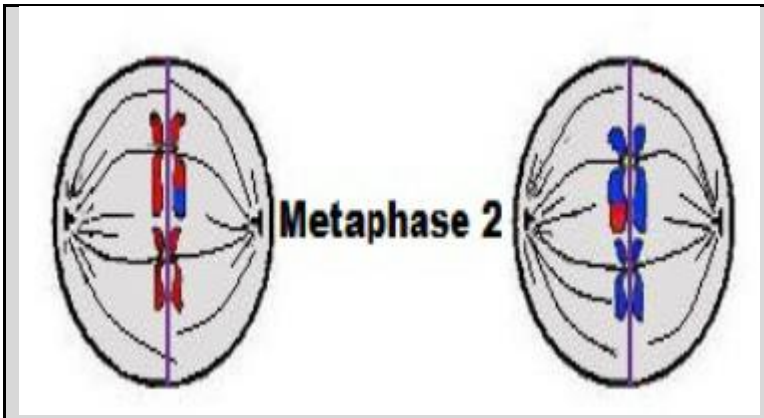
Meiosis

Interphase (interphase is not a part of meiosis • Replication occurs before meiosis during interphase.	
Prophase 1: • The nuclear membrane begins to break down and disintegrate. • The replicatedchromosomes begin to condense and become visible. • Homologous chromosomes synapse (pair up) to form bivalents or tetrads. • Crossing over occursbetween non-sister chromatids. • The crossover pointsbetween chromosomes are called chiasmata (plural) and chiasma (singular). • Spindle microtubulesbegin to form.	
Metaphase 1 • The homologouschromosome pairs line up along the cell's equator (metaphase plate). • 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. • Each bivalent has aspecial protein structure called a kinetochorewhere spindle fibersattach during division to pull the chromosomes apart. These kinetochores areattached to spindle microtubules that areattached to the opposite poles.	

Anaphase 1 • Spindle fibers attached to the kinetochore of the homologous pairs,shorten and pull thehomologous pairs apart. • The chiasmata also break down and separate. • One chromosome of each pair move to opposite poles of the cell.	
Telophase 1: • Chromosomes begin touncoil and nuclear envelope reforms. • Spindle fibers and microtubules break downand disintegrate. • **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. • No more replication is needed.	
Prophase II: • Chromosomes condense again and become visible. • Spindle fibers again form. • Nuclear membrane disintegrates again.	

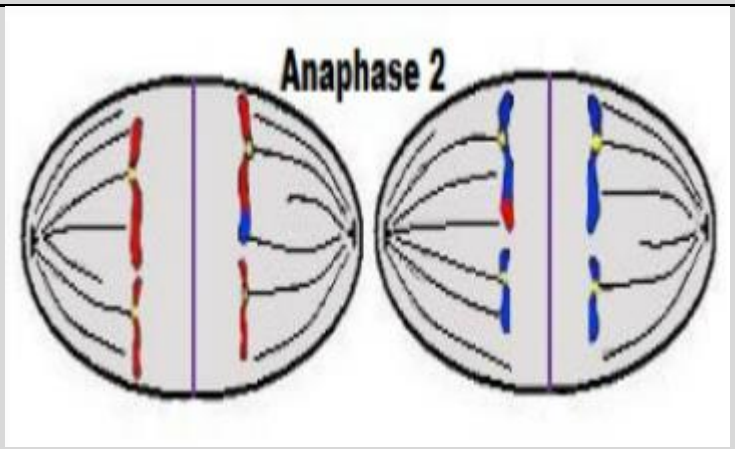
Metaphase II:

- Chromosomes line up along the equator.
- Centromeres contain two kinetochores that attach to spindle fibers from the centrosomes at each pole.
- Resembles metaphase from mitosis.



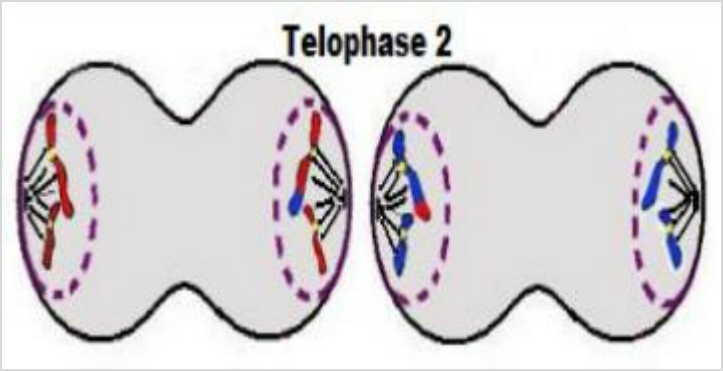
Anaphase II:

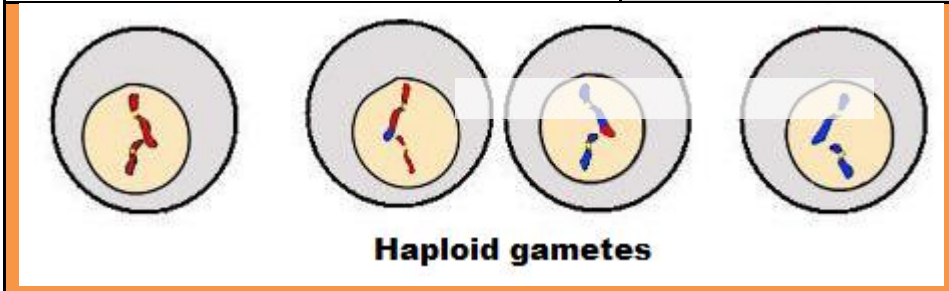
- Spindle fibers pull apart the centromeres andsister chromatids are pulled towards the opposite poles.
- At this point thechromatids are considered chromosomes again.



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.





∑ **- Crossing over and random orientation promotes genetic variation.**

- Meiosis is the formation of gametes that produce offspring that are genetically different than their parents.
- The two main ways variation is created in the offspring is through crossing-over and through random orientation of the chromosomes.

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 Variation

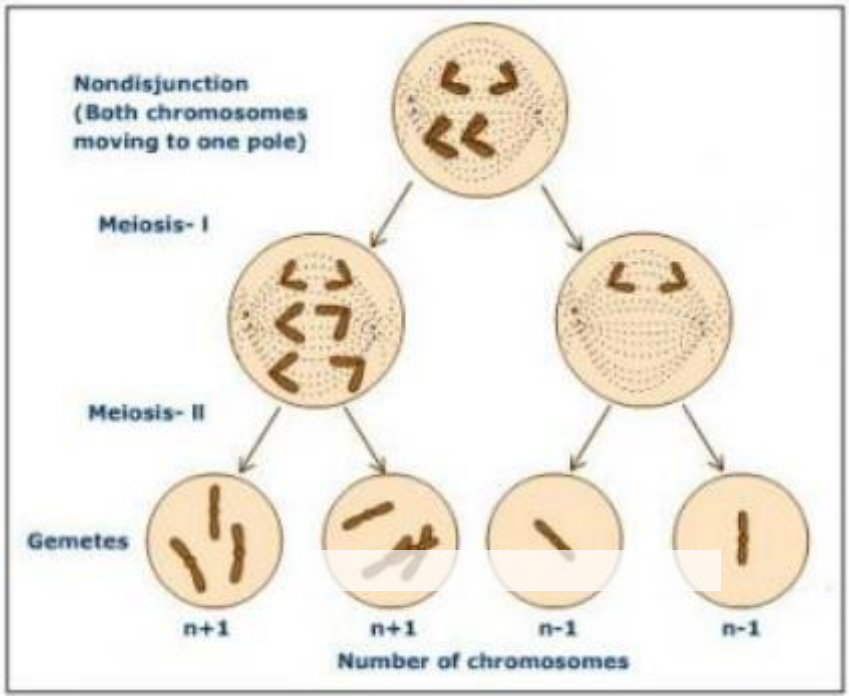
- Occurs in metaphase I of meiosis.
- When homologues line up along the equatorial plate in metaphase I, the orientation of each pair is random; meaning the maternal or paternal homologue can orient toward either pole.
- This means the number of combinations that can occur in the gamete is 2^n(n=number 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.

∑ - Fusion of gametes from different parents promotes genetic variation.

- The fusion of two gametes to form a zygote is the start of a new organism and new life
- It combines genetic information from two different individuals
- As explained above in the section on random variation. When considering the different combinations that could exist in each gamete, the number of male gametes released at one time (millions), and because of crossing over, the possible different combinations of alleles in the zygote is immeasurable
- Fusion of gametes from different parents therefore, promotes genetic variation

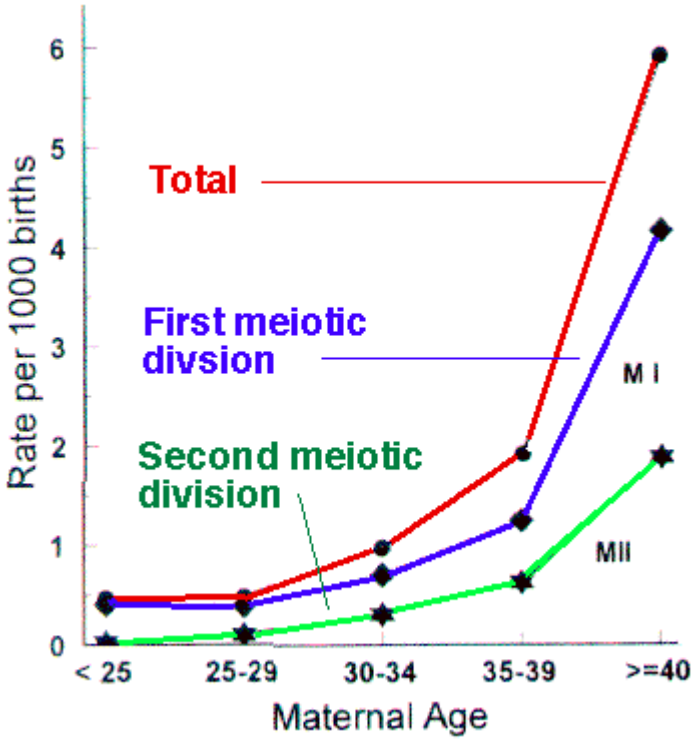
β - Application: Non-disjunction can cause Down syndrome and other chromosome abnormalities.

- 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 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.
- Other types of non-disjunctions are trisomy 18 (Edwards Syndrome - many of these fetuses die before birth), trisomy 13 (Patau's syndrome – causes multiple and complex organ defects and highly effects normal development).



β - Application: Studies showing age of parents influences chances of non-disjunction

- Studies showing the how the age of parents affects the chances of a non-disjunction occurring



http://10e.devbio.com/article.php?ch=19&id=189

- The study of Yoon and colleagues (1996) concluded that 86% of the trisomy 21 cases from 1989-1993 in Atlanta were maternal in origin, 9% were paternal in origin, and 5% occurred during the mitotic divisions of the embryo. They also showed that 75% of the maternally originated Down syndrome cases arose from non-disjunction during the first meiotic division, and 25% originated in the second meiotic division.

Possible discussion
Living with Down's syndrome https://www.youtube.com/watch?v=-jz5znsygn0
Another Video - https://www.youtube.com/watch?v=eBVtcvj8rN8
Article on parents living with a son for 47 years with Down syndrome http://www.dailymail.co.uk/femail/article-2803834/I-wish-d-aborted-son-ve-spent-47-years-caring-s-shocking-admission-read-judge.html
What would you do if you had that choice?

Do the questions on incidences of non-disjunctions on page 167 of your text.

Guidance:

• Preparation of microscope slides showing meiosis is challenging and permanent slides should be available in case no cells in meiosis are visible in temporary mounts.

• Drawings of the stages of meiosis do not need to include chiasmata.

• The process of chiasmata formation need not be explained.

Theory of knowledge:

In 1922 the number of chromosomes counted in a human cell was 48. This remained the established number for 30 years, even though a review of photographic evidence from the time clearly showed that there were 46. For what reasons do existing beliefs carry a certain inertia?

3.4 Inheritance

Nature of science:

Making quantitative measurements with replicates to ensure reliability. Mendel's genetic crosses with pea plants generated numerical data. (3.2)

Some definitions

Genotype: the specific alleles of an organism.

Phenotype: the observable characteristics or traits of an organism.

Dominant allele: an allele that has the same effect on the phenotype whether it is present in the homozygous or heterozygous state. The dominant allele masks the recessive in the heterozygous state.

Recessive allele: an allele that only has an effect on the phenotype when present in the homozygous state.

Co-dominant alleles: pairs of alleles that both affect the phenotype when present in a heterozygote.

(The terms incomplete and partial dominance are no longer used.)

Locus: the particular position on homologous chromosomes of a gene.

Homozygous: having two identical alleles of a gene.

Heterozygous: having two different alleles of a gene.

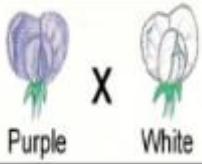
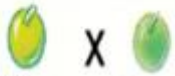
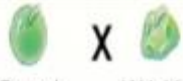
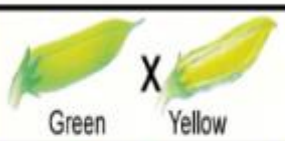
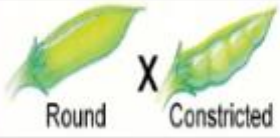
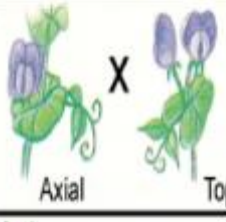
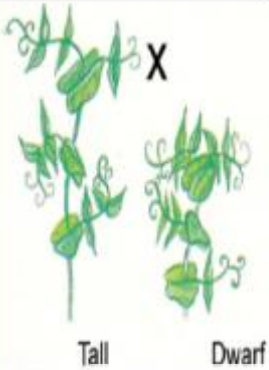
Carrier: an individual that has one copy of a recessive allele that causes a genetic disease in individuals that are homozygous for this allele.

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

∑ **- Understandings:**

∑ **- Mendel discovered the principles of inheritance with experiments in which large numbers of pea plants were crossed.**

- 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.
- The large number of crosses and replicates he performed were essential in ensuring reliability in his test results and determining the ratios from the crosses

Trait	Dominant vs. recessive	F_2 generations Dominant form	F_2 generations Recessive form	Ratio
Flower color	 Purple X White	705	224	3.15:1
Seed color	 Yellow X Green	6022	2001	3.01:1
Seed shape	 Round X Wrinkled	5474	1850	2.96:1
Pod color	 Green X Yellow	428	152	2.82:1
Pod shape	 Round X Constricted	882	299	2.95:1
Flower position	 Axial X Top	651	207	3.14:1
Plant height	 Tall X Dwarf	787	277	2.84:1

∑ - Watch this video on Gregor Mendel and write down a couple of interesting facts https://www.youtube.com/watch?v=GTiOETaZg4w

-
-
-
-

∑ - Gametes are haploid so contain only one allele of each gene.

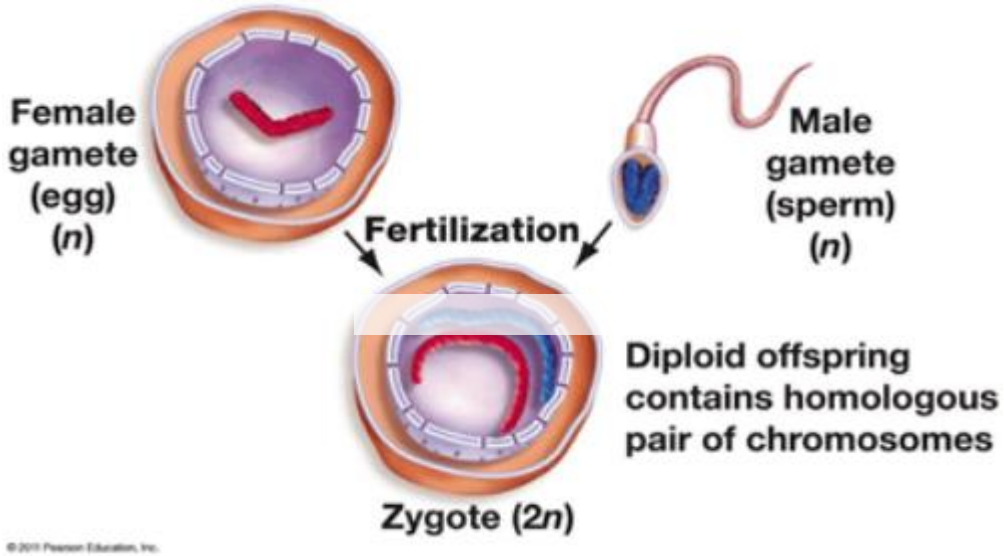
- Gametes which are sex cells such as sperm and eggs
- Gametes contain one set of chromosomes or one chromosome of each type and are therefore haploid (n)
- Since they have only one chromosome of each type, gametes also only contain one allele of each gene
- The specific allele depends upon if that particular chromosome came from the mother or father and if crossing over occurred during prophase 1
- Together the two gametes form a zygote

∑ - The two alleles of each gene separate into different haploid daughter nuclei during meiosis.

- During meiosis a diploid nuclei in a germ cell divides to produce 4 haploid nuclei
- If an individual has two of the same allele AA for a particular gene, all 4 haploid cells will contain the allele A. This is the same if the alleles for the gene are aa
- If an individual has two different alleles for a particular gene such as Aa, the haploid gametes will contain 50% A and 50% a for that specific gene
- The separation of the alleles into different nuclei is called segregation

∑ - Fusion of gametes results in diploid zygotes with two alleles of each gene that may be the same allele or different alleles.

- 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



∑ - Dominant alleles mask the effects of recessive alleles but co-dominant alleles have joint effects.

- Dominant alleles mask the effects of recessive alleles and are expressed in the phenotype
- For example, if B is dominant for brown hair color and little b is recessive for blonde hair colour, an individual that is BB (homozygous dominant) will have brown hair.
- If the individual has the genotype Bb (heterozygous), they will also have brown hair, as the dominant B is masking the expression of b
- If the individual has the genotype bb (homozygous recessive), that person will have blonde hair

Video on Inheritance: https://www.youtube.com/watch?v=CBezq1fFUEA

β - Application: Inheritance of ABO blood groups.

- 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). The (I) represents immunoglobulin. 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
- The allele for blood **type B** alters the basic antigen by adding a **galactose**to the glycoprotein. Individuals that do not have this allele and are exposed to blood type B, will produce Anti-B antibodies that will attack and destroy these red blood cells (RBC)
- The allele for blood **type A** alters the basic antigen by adding an**acetylgalactosamine**. So individuals that do not have the A allele will produce Anti-A antibodies that will attack and destroy these RBC's
- The allele for blood **type O** produces the **basic antigen** that will be present on the cell membrane of these RBC's. Individuals with blood type O will produce both Anti-A and Anti B antibodies if exposed to either A or B blood cells
- Individuals that have both A and B alleles will have both of the antigen modifications. Hence, the alleles for A and B are co-dominant. If exposed to blood type A or B, no Anti-A or Anti-B antibodies will be produced.
- If individuals with blood type A, B or AB are exposed to blood type O, no immune response will occur because blood type O only contains the basic antigen

Phenotype	Genotype

O	ii
A	I^AI^A or I^Ai
B	I^BI^B or I^Bi
AB	I^AI^B

β - Skill: Construction of Punnett grids for predicting the outcomes of monohybrid genetic crosses.

- Monohybrid inheritance is the inheritance of a single gene.
- The trait coded for by the gene is controlled by different forms of the gene called alleles.
- A Punnett square or grid is a tool used to solve genetic problems.

For example:

- Mendel studied many different traits related to pea plants.

- One example is seed color. In pea plants, yellow seeds are dominant over green peas.
- If a pea plant that is heterozygous for yellow peas is crossed with a plant with green peas, what are the genotypes and phenotypes of the first generation (F1) of pea plants?

The following are steps to solve the above problem.

1) Create a key for the pea plants using the uppercase letter for the dominant allele and the lower case letter for the recessive allele. In this case yellow peas can be represented as Y and green peas can be represented as y (yellow peas = Y and green peas = y).

2) Write out the parental cross using the key you created. In this case the cross would be Yy x yy. This cross is a heterozygous x homozygous recessive cross.

3) Write down the possible genotypes of the gametes. In this case they would be Y and y from the yellow plant and only little y from the green pea plant as that is the only type of allele.

4) Draw a Punnett square and insert the possible gametes along the top and side. Fill in the possible genotype combinations.

	y	**y**
Y	Yy	Yy
y	yy	yy

5) Write out the possible genotypes and the genotypic ratio. For this example the genotypes and genotypic ratio is 2:2 or 1:1 Yy:yy

6) Write out the possible phenotypes and phenotypic ratio. For this example the phenotypes and phenotypic ratio is also 2:2 or 1:1 yellow: green.

Quick Practice

1) In dogs, short hair is dominant over long hair. Two heterozygous short-haired dogs are mated. What are the genotypic and phenotypic ratios of the puppies? What is the probability that the fourth puppy has long hair? Show all your work using a Punnett square.

Co-Dominance

2) A woman that has blood type AB and a man that is homozygous for blood type A, have 4 children. What are the possible genotypes and phenotypes of the offspring? What is the probability they will have a child with the blood type O? Show all work using a Punnett square.

Come back and do this question after we do sex-linked inheritance.

3) A man with hemophilia (a recessive, sex linked trait condition) has a daughter with a normal phenotype (*hint she must be heterozygous)*. She marries a man who is normal for the trait. What is the probability that a daughter of this mating will be a hemophiliac? That a son will be a hemophiliac? List the possible genotypic and phenotypic ratios. Show all your work using a Punnett square.

*****Do the Data-based question on page 173-174 of your textbook*****

β - Skill: Comparison of predicted and actual outcomes of genetic crosses using real data.

Do the lab on genetic crosses and probabilities using coin flips

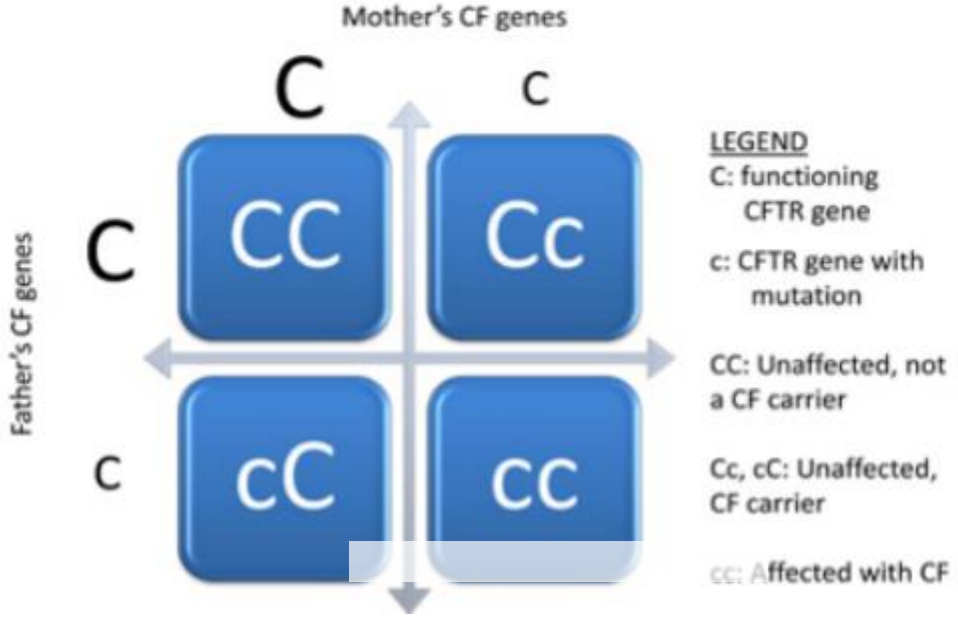
Do data based questions on page 176-177

∑ - 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.

- Many genetic diseases are caused by recessive alleles contained on the autosomal chromosomes (chromosome 1-22)
- Therefore, the disease would only be expressed if an individual has two recessive alleles (i.e. aa)
- If an individual has one of the dominant alleles (i.e. Aa), they will not show symptoms of the disease. These people are known as carriers. They can pass this allele on to their offspring
- If the other parent is also a carrier then their offspring have a 25% chance of getting the disease
- A small number of diseases are co-dominant, such as sickle cell anemia which was studied in 3.1
- H^AH^A – do not have sickle cell anemia, H^AH^S – mild anemia, H^SH^S – severe anemia
- An example of a recessive genetic disease is cystic fibrosis and a dominant disease is Huntington's Disease

β - Application: Inheritance of cystic fibrosis and Huntington's disease.

Cystic Fibrosis

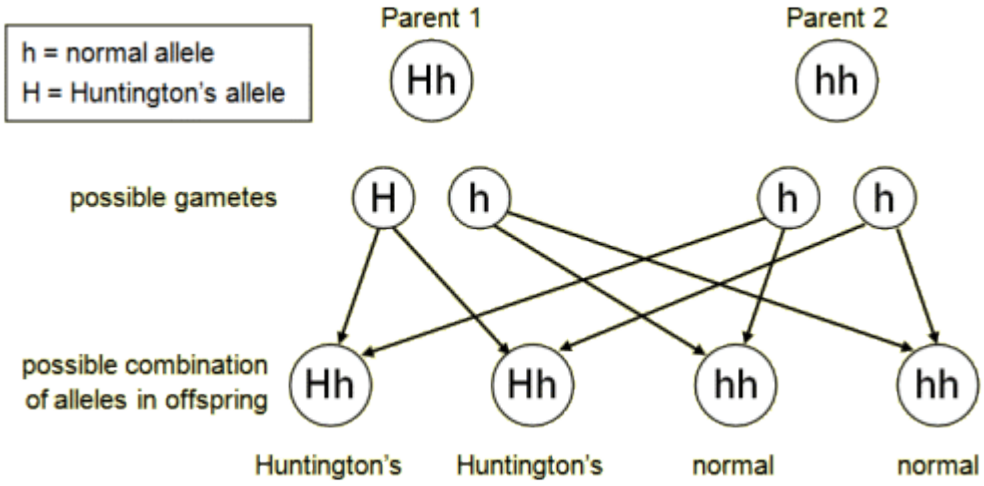


- Cystic fibrosis is a autosomal recessive disease caused by an allele of the**CFTR** gene on chromosome 7
- The CFTR gene codes for a chloride ion channel protein that transports chloride ions into and out of cells. Chloride is a component of sodium chloride, a common salt found in sweat. Chloride also has important functions in cells;

for example, the flow of chloride ions helps control the movement of water in tissues, which is necessary for the production of thin, freely flowing mucus.

- Mutations in the CFTR gene disrupt the function of the chloride channels, preventing them from regulating the flow of chloride ions and water across cell membranes. As a result, cells that line the passageways of the lungs, pancreas, and other organs **produce mucus that is unusually thick and sticky.** This mucus clogs the airways and various ducts, causing the characteristic signs and symptoms of cystic fibrosis

Huntington's disease



- Humans have two copies of the Huntingtin gene (HTT), 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.
- Therefore, children of people affected with the disease have a 50% chance of getting that allele from an affected parent.
- If both parents have Huntington's disease, offspring have a 75% chance of being affected by the disease.
- Huntington's disease is a neurodegenerative genetic disorder that affects muscle coordination and leads to mental decline and behavioral symptoms
- In Huntington's disease, a repetition of a **CAG** sequence in the gene encoding for the protein Huntingtin makes it clump together in our brain cells, ultimately making the brain cell die.
- The exact mechanism of the disease is still being researched; however, this is what is current research suggests.
- The repetitive glutamates (CAG) in the Huntington protein change the shape of the brain cells, affecting their function. The glutamate sends signals that constantly over-excite brain cells. Their overexcitement leads to cell damage, and ultimately cell death.
- https://www.youtube.com/watch?v=TkfVnzavREw

http://www.thetruthaboutgenetics.com/2011/07/huntingtons-disease-explained-simply.html

∑ - Some genetic diseases are sex-linked. The pattern of inheritance is different with sex-linked genes due to their location on sex chromosomes.

- 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 inheritance for eye colour is observed and studied in Drosophila (fruit flies)

β - Application: Red-green colour blindness and hemophilia as examples of sex-linked inheritance.

- Color blindness and hemophilia are both examples of sex linkage.
- Color blindness and hemophilia are produced by a recessive sex-linked allele on the X chromosome.
- 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.
- On the other hand, since females have two X chromosomes, if they inherit one X chromosome with the disease; they have another normal X chromosome to make the correct gene product. These individuals are considered carriers.
- Since male offspring have to receive a Y from their father, they will always inherit the colorblind or hemophilia allele from their mother; not the father.
- Males that have the disease can only pass the colorblind or hemophilia allele onto their daughters. Their sons will receive the Y chromosome.
- Females can only get X-linked recessive diseases if the mother happens to be a carrier of the disease (or has the disease) and the father also has the disease.
- Therefore sex-linked diseases are rare in females.

Punnett square example: Colorblind man X^b y crossed with a woman with normal vision X^B X^B

-	X^b	y
X^B	X^B X^b	X^B y
X^B	X^B X^b	X^B y

- As you can see above both daughters will be carriers and both males will have normal vision. This would be the same Punnett square for Hemophilia (X^h y x X^H X^H).

Punnett square example: Carrier female $X^B X^b$ is crossed with a normal vision male $X^B y$

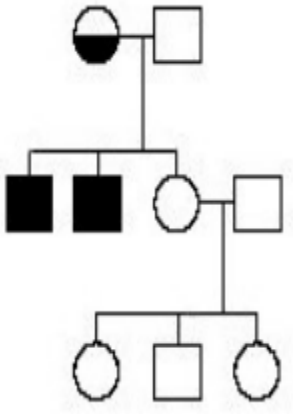
	X^B	y
X^B	$X^B X^B$	$X^B y$
X^b	$X^B X^b$	$X^b y$

- As you can see from the Punnett square above, the following combinations are possible during fertilization: 1 female with normal vision $X^B X^B$, 1 female that is a carrier for the trait $X^B X^b$, 1 male with normal vision $X^B y$ and 1 colorblind male $X^b y$. Again, this would be the same Punnett square for Hemophilia ($X^H y$ x $X^H X^h$).

***** X^b and X^h is the notation for the colorblind and hemophilia alleles. The corresponding dominant alleles are X^B and X^H *****

β - Skill: Analysis of pedigree charts to deduce the pattern of inheritance of genetic diseases.

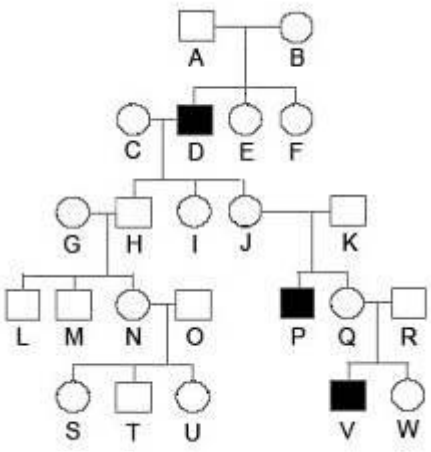
- Pedigree charts or diagrams display all of the known genotypes for an organism such as humans and their ancestors.

Symbol Representation in a Pedigree	Diagram of a Pedigree
• In a pedigree chart, males are represented as squares and females as circles • If the square or circle is filled in black, the individual is affected by the condition • Some pedigree's represent a carrier with a half filled in circle or square (males are only carriers for autosomal diseases). If it is not filled in, you have to figure out if the individual is a carrier from the inheritance pattern. • Mating between two individuals is represented by a horizontal line • Children are represented by a vertical line between two parents that divides out with a horizontal line to connect the offspring. In the example to the right, the two boys affected by the trait being studied and the unaffected girl are the offspring of the two	

individuals (parents) in the first line	

- The pedigree above most likely displays an x-linked recessive disease because the mother is a carrier, and she gives the allele to her two sons but not her daughter. The daughter could have inherited the allele carrying the trait being studied. However, one can see in the 3rd generation, the boy does not have the trait. Since he does not have the trait, she probably did not inherit the affected allele. You would have to look to the next generation to get a better idea if the daughter was a carrier or not.

Here is another more in depth pedigree chart showing Hemophilia:

How to Determine a Pedigree	Diagram of a Pedigree
• For dominant and recessive alleles, upper-case and lower-case letters, respectively, should be used. • If neither parent is affected, the trait cannot be dominant. The trait could be recessive and either parent or both could be heterozygous carriers unless the disease is sex-linked (diagram to the right) • If the father is affected and none of his sons or daughters are affected the trait most likely is recessive. If an affected boy shows up again in the next generation, the trait is sex-linked recessive • If the trait shows up equally between boys and girls and tends to skip generations, the	

trait is most likely autosomal recessive • In the chart to the right D, P, and V would be represented as (X^h Y) while the unaffected women G, N, S, and U would be represented by (X^H X^H). Women represented by B, I, J, and Q have to be carriers X^H X^h. The other women could be either carriers or homozygous dominant. None of the women are homozygous recessive as they don't have the disease.	

- For co-dominance, the main letter should relate to the gene and the suffix to the allele; both upper case. For example, Roan horses that are co-dominant could be represented as R^B and R^w, respectively. For sickle-cell anemia, $Hb^A$is normal and Hb^s is sickle cell.

∑ - Many genetic diseases have been identified in humans but most are very rare.

- There are over 6000 identified genetic disorders, most of these diseases are caused by rare recessive alleles that follow Mendelian genetics
- Even though this might seem like a lot, most of the human population does not suffer from a genetic disorder and since you need both recessive alleles, these diseases are very rare

Some good links

http://www.cbsnews.com/news/genetic-disorders-hit-amish-hard/

http://www.medicaldaily.com/woman-not-afraid-anything-even-danger-due-rare-genetic-disorder-318750

https://www.youtube.com/watch?v=SJ6Xpun3qfA

https://www.youtube.com/watch?v=6zMll7x2WSY

∑ - Radiation and mutagenic chemicals increase the mutation rate and can cause genetic diseases and cancer.

- 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
- Certain **chemical substances** can all cause chemical change in DNA and are therefore considered mutagenic. Some examples are **Benzene**(industrial solvent and precursor in the production of drugs, plastics, synthetic rubber and dye), **Nitrosamines** (an important group of mutagens found in tobacco), and **Aromatic amines** and amides (which have been associated with carcinogenesis since 1895 when German physician Ludwig Rehn observed high incidence of bladder cancer among workers in German synthetic aromatic amine dye industry).

β - Application: Consequences of radiation after nuclear bombing of Hiroshima and accident at Chernobyl.

https://www.youtube.com/watch?v=5WGUbfzr31s (watch from 26:12 - 47:08)

https://www.youtube.com/watch?v=YfulqRdDbsg Called Inside Chernobyl – quite a good film by an amateur filmmaker. There are some disturbing images.

https://www.youtube.com/watch?v=b8QY5gt1weE (watch 35:00 to 49:00 – study on the effects of radiation on Hiroshima survivors)

******Write 10 facts regarding the consequences of the nuclear bombs in Japan and the nuclear accident in Chernobyl. ******

*****Do the Data-Based questions on page 186*****

3.5 Genetic Modification and Biotechnology

Understandings:

∑ - Gel electrophoresis is used to separate proteins or fragments of DNA according to size.

- Before gel electrophoresis takes place, enzymes are used to cut DNA into fragments of various lengths and different charges.
- 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 thelarge non-charged particles fall out and embed in the gel the quickest.

∑ - PCR can be used to amplify small amounts of DNA.

- 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 copiesof 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.

*****Do data based questions on page 188*****

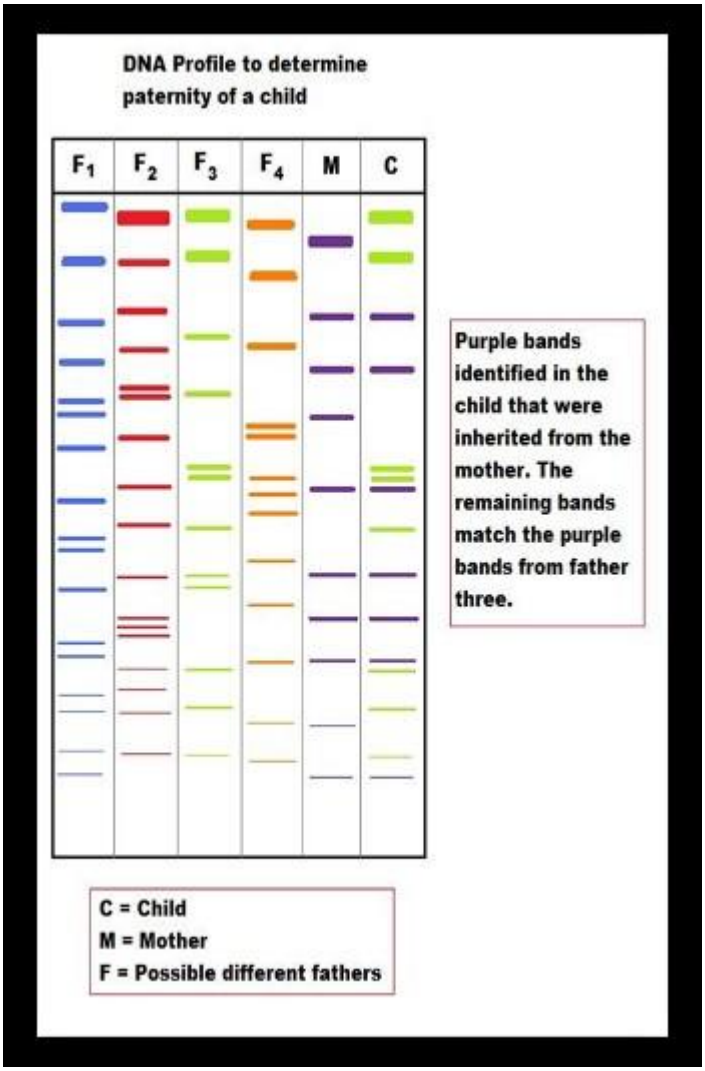
∑ - DNA profiling involves comparison of DNA.
β - Application: Use of DNA profiling in paternity and forensic investigations.

- 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 suits 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 endonucleases 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.

β - Skill: Analysis of examples of DNA profiles.

Steps to identifying unknown individual	DNA Profile

1. The first step in identifying an unknown child is to first match the bands (size and location) from the mother that appear in the child. A good way to do this is to mark the child with the same color as the mother for the bands that match or to put a small M next to the matching bands.
2. The next step is to match the remaining bands with one of the unknown samples from the different father possibilities. As 50% of the DNA inherited in the child will come from the mother and 50% of the DNA will come from the father, the remaining bands should match with the unknown father's sample.
3. Once again, colors can be used to match the child's remaining bands to the correct father's bands or by using other notation such as F3 to mark the matching bands.
4. A similar technique can be used in criminal investigations using the victim's blood and the possible suspect's blood to match an unknown sample found at a crime scene



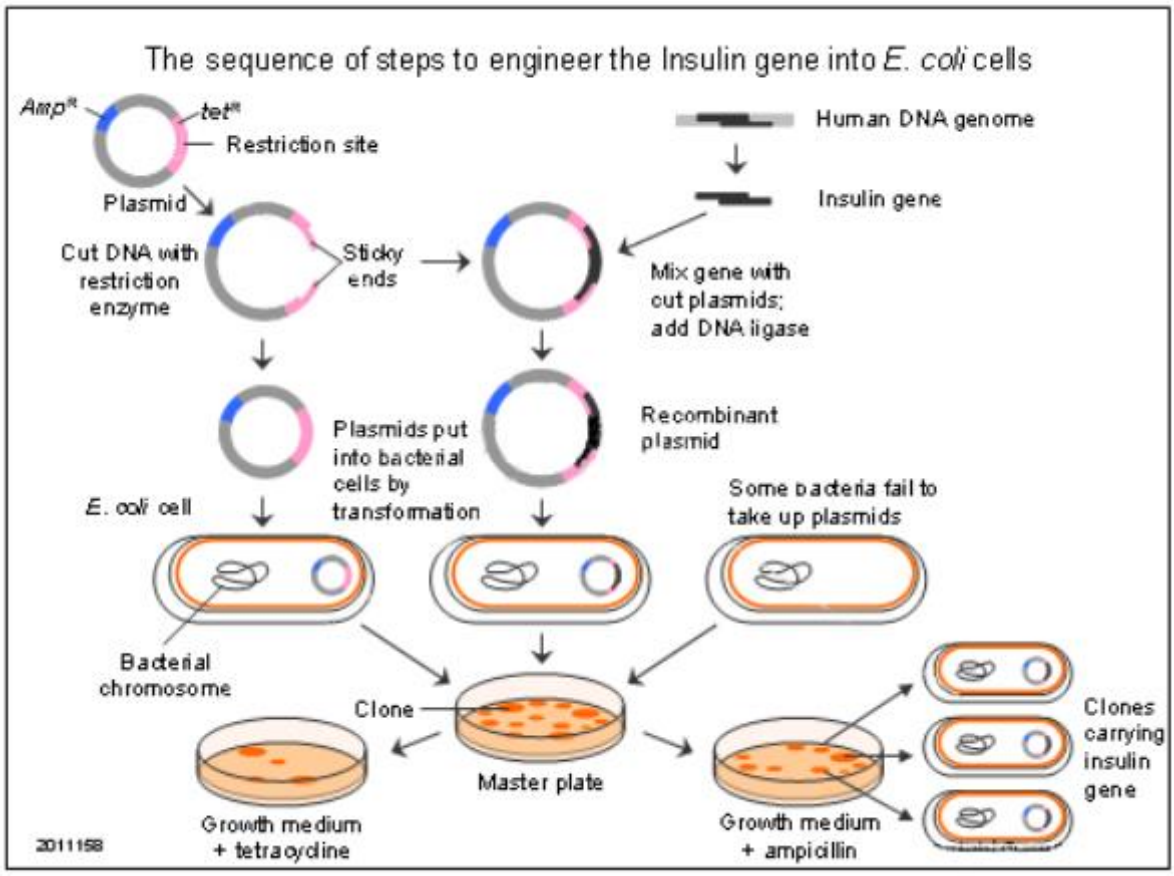
∑ - Genetic modification is carried out by gene transfer between species.

- 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 modification has been used to introduce new characteristics to certain animal species. For example goats that produce milk containing spider silkand

bacteria that produce human insulin. A plant example is the production ofgolden rice that contains beta-carotene .

β - Application: Gene transfer to bacteria using plasmids makes use of restriction endonucleases and DNA ligase.

- 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, mRNA that codes for insulin produced in the pancreatic cells isextracted.
- The enzyme reverse transcriptase is mixed with the mRNA. This enzymeproduces a strand of coding DNA called cDNA.
- Plasmids are small circles of DNA found in bacteria cells. These plasmids arecut with a restriction enzyme, leaving sticky ends to which the cDNA (cDNA is cut with the same restriction enzyme) can attach.
- DNA ligase is used to seal the nicks between the cDNA and the plasmid.
- Linking sequences are added to the cDNA allowing them to be inserted into the plasmid.
- The bacterial plasmid carrying the insulin gene is now inserted into plasmid free bacterial cell such as E.coli bacteria (with plasmid removed). This is known as the host cell.
- These insulin producing bacterial cells will now reproduce rapidly during fermentation, creating millions of insulin producing bacteria cells.
- Finally, the insulin produced is extracted from the cell and purified to be used by diabetics.



Nature of science:

Assessing risks associated with scientific research—scientists attempt to assess the risks associated with genetically modified crops or livestock. (4.8)

- Read the short paragraph on assessing the risks of genetically modified crops or livestock on page 192. Write a very brief summary below

β - Application: Assessment of the potential risks and benefits associated with genetic modification of crops.

Benefits	Possible Harmful Effects
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	Cross-pollination could occur when seeds from the GM crop pollinate neighboring farmer's crops which are made from locally adapted seeds thathave adapted over time to the specific microclimates, soils, other environmental conditions.
Can use pest resistant crops or modified crops in areas where water availability is limited	Cross-pollination could occur with wild species giving them a competitive advantage. This could allow these plants to outcompete and eliminate other plants (decrease biodiversity).
Could add genes for certain proteins, vitamins or possible vaccines (less cost than producing in a lab)	Patent protection given to companies that develop new types of seeds using genetic engineering. Since companies own intellectual ownership of their seeds, they have the power to dictate terms and conditions of their patented product. They could charge large amounts of money for seeds and the people that need them the most in the 3rd world countries, couldn't afford to grow these crops.
Crops last longer or don't spoil during storage	Crops which produce toxins to kill insect (pest resistant) could be harmful to humans.
Varieties of crops lacking certain allergens or toxins	Some people or livestock might have allergic reactions to certain proteins produced by transferred genes
	Use of GMO crops that contain a toxin to kill a pest can lead to resistance to the toxin in the target pest and secondary pests that are resistance to the toxin but were previously scarce

β - Skill: Analysis of data on risks to monarch butterflies of Bt crops.

http://www.inspection.gc.ca/plants/plants-with-novel-traits/general-public/monarch-butterflies/eng/1338140112942/1338140224895

http://cls.casa.colostate.edu/transgeniccrops/hotmonarch.html

http://www.ars.usda.gov/is/br/btcorn/

*****Data Based question page 195*****

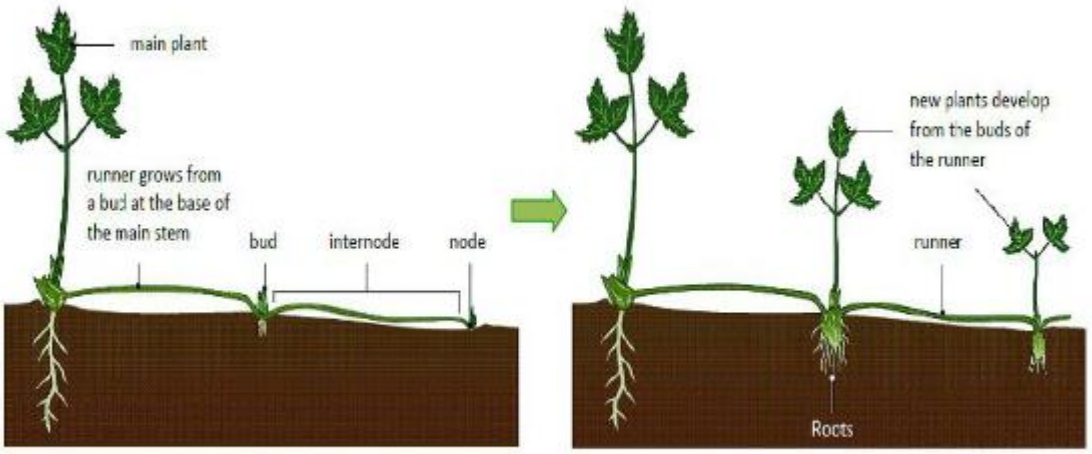
∑ - Clones are groups of genetically identical organisms, derived from a single original parent cell.

- 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

∑ - Many plant species and some animal species have natural methods of cloning.

- Plants use a variety of natural methods of cloning involving stems, roots, leaves or bulbs.
- Garlic bulbs are modified plant leaves. All the bulbs in the group are genetically identical to each other.
- Strawberry plants grow horizontal stems called runners that grow roots into the soil. These small plants develop into independent cloned strawberry plants

Another method of vegetative propagation used by some plants is to use runners, small shoots which run along the ground and cause buds to grow which develop roots. The diagram shows how runners can produce offspring.



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.

www.a2biology101.wordpress.com

Strawberry plant

- Underground stems called tubers in potatoes can form new potato plants which are clones of the original parent potato plant
- Asexual reproduction in animals is less common than sexual reproduction. For example, it can happen in hydra (budding), sea anemones (budding) and starfish (autonomy or fission).

β - Skill: Design of an experiment to assess one factor affecting the rooting of stem-cuttings.

Design you own experiment to investigate a factor that effects the rooting of stem-cuttings. The lab will be graded according to the IB internal assessment rubric

∑ - Animals can be cloned at the embryo stage by breaking up the embryo into more than one group of cells.

- 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
- This ability was first discovered by trying on Sea anemones

∑ - Methods have been developed for cloning adult animals using differentiated cells.

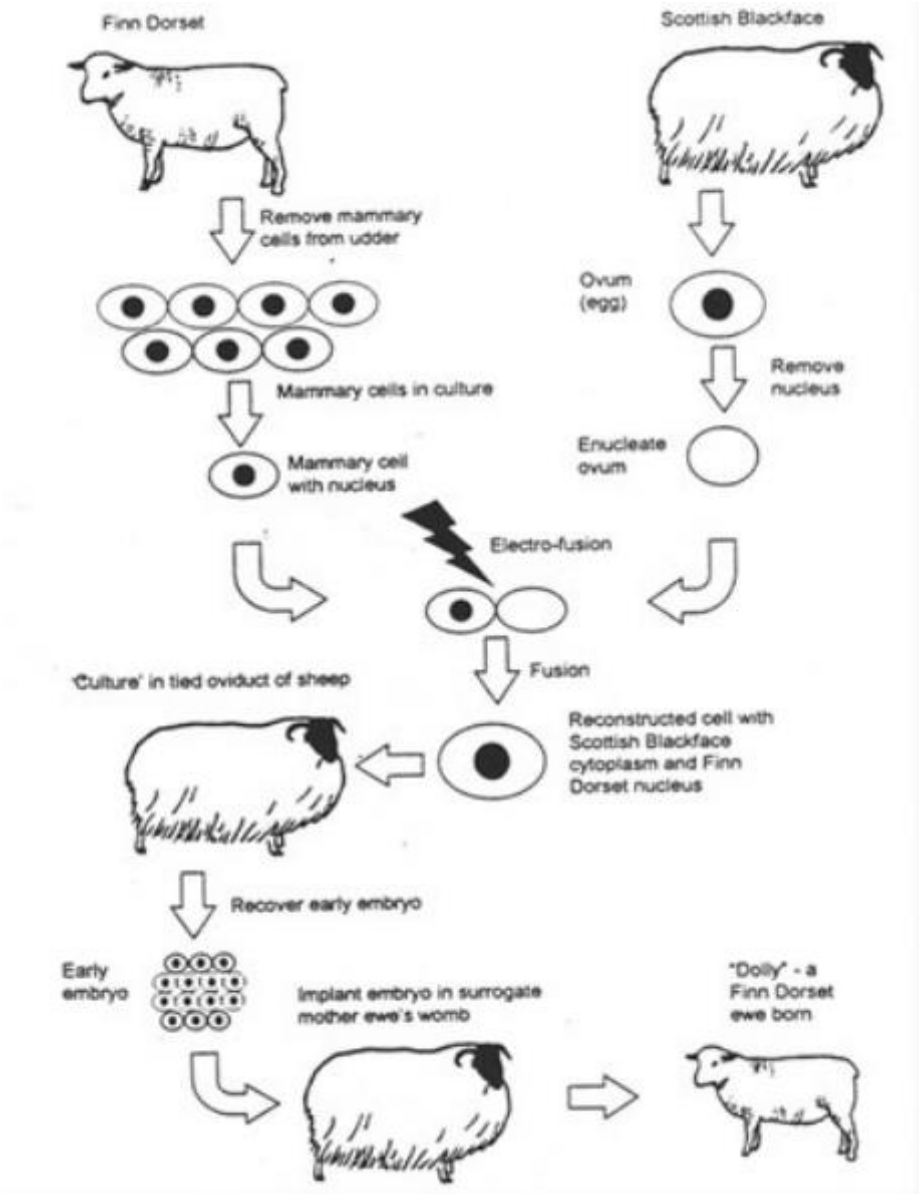
- Once cells start to differentiate and embryos develop into a fetus and eventually an adult cloning becomes much more difficult
- Therapeutic cloning is an example of cloning using differentiated cells
- This type of cloning can be used to create a specific tissue or organ

- Cloning using differentiated cells can also be used to reproduce organisms like dolly the sheep. This is done through somatic-cell nuclear transfer.

∑ **- Application: Production of cloned embryos produced by somatic-cell nuclear transfer.**

Reproductive Cloning – Method used to create Dolly (first cloned sheep)

- Somatic cell removed from the donor and cultured (removed from the udder in Dolly).
- Unfertilized egg is removed from another sheep and its nucleus is removed.
- The unfertilized egg is fused with the cultured somatic cell from the first sheepusing an electric current.
- The embryo created now contains all the genetic information from only the first sheep.
- The embryo divides by mitosis "in vitro" until it reaches the blastocyst stage(hollow ball of about 16 cells).
- Next, the blastocyst is transferred and embedded into the womb of a third surrogate sheep.
- The cloned embryo will continue to grow, until a baby lamb is born that isgenetically identical to the first sheep that donated the somatic cell.



Guidance:

• Students should be able to deduce whether or not a man could be the father of a child from the pattern of bands on a DNA profile.

• Dolly can be used as an example of somatic-cell transfer.

• A plant species should be chosen for rooting experiments that forms roots readily in water or a solid medium.

4.1.4 Explain the consequence of a base substitution mutation in relation to the processes of transcription and translation, using the example of sickle-cell anemia (objective 3)

- A mutation that causes the replacement of a single base nucleotide with another nucleotide in DNA.
- 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.
- This change in the mRNA sequence may change the amino acid in the polypeptide coded for by the gene; in the process of translation.
- Sickle-cell anemia 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 because of their abnormal shape and inflexibility caused by crystallization of the abnormal hemoglobin.
- 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.
- The normal triplet when transcribed and translated codes for the amino acid glutamic acid.
- When the base substitution occurs, the amino acid that is translated is now valine.
- Since valine has a different shape and charge, the resulting polypeptide's shape and structure changes.
- As a result, hemoglobin's shape will change, as does the shape of the red blood cell, resulting in the problems associated with sickle cell anemia listed above.

Discussion Topics: One could discuss the ethical issues relating to screening of fetuses for genetic diseases? If a genetic disease is found in the fetus, is it ok to abort the fetus? Should parents be allowed to screen for genetic diseases if there is a possibility that they will abort the fetus?

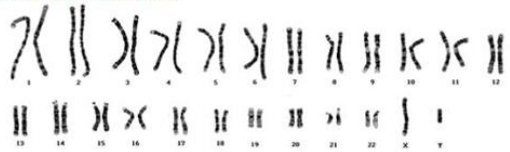
Another topic of discussion is the occurrence of sickle-cell allele in areas that are prone to malaria, such as regions of Africa. Is this a correlation or is there a causal link between the occurrences of the Hb^s (sickle-cell) allele and the frequency of malaria in a particular region. In this particular case, there is a clear causal link because the malaria parasite has a difficult time living inside the sickled red blood cells. This makes the carrier of the allele resistant to malaria; therefore natural selection favors the carrier of the sickle cell trait. One could look at other examples that show correlation, to discuss the differences between correlations and causal links.

4.2.4 Explain that non-disjunction can lead to changes in chromosome number, illustrated by reference to Down syndrome (trisomy 21) (objective 3)

- 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.
- An example of a non-disjunction is Down 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.

4.2.5 State that, in karyotyping, chromosomes are arranged in pairs according to their size and structure (objective 1)

- In karyotyping, chromosomes are arranged in pairs according to their size and structure with the largest at chromosome pair 1 and the smallest at chromosome 22.
- The 23rd pair is the sex chromosomes. Females have two X chromosomes and males have one X chromosome and one Y chromosome.



4.3.8 Describe the inheritance of colour blindness and hemophilia as examples of sex linkage (objective 2)

- Color blindness and hemophilia are both examples of sex linkage.
- Color blindness and hemophilia are produced by a recessive sex-linked allele on the X chromosome.
- 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.
- On the other hand, since females have two X chromosomes, if they inherit one X chromosome with the disease; they have another normal X chromosome to make the correct gene product. These individuals are considered carriers.
- Since male offspring have to receive a Y from their father, they will always inherit the colorblind or hemophilia allele from their mother; not the father.
- Males that have the disease can only pass the colorblind or hemophilia allele onto their daughters. Their sons will receive the Y chromosome.
- Females can only get X-linked recessive diseases if the mother happens to be a carrier of the disease (or has the disease) and the father also has the disease.
- Therefore sex-linked diseases are rare in females.

Punnett square example: Colorblind man X_b y crossed with a woman with normal vision X_B X_B

	X_b	y
X_B	X_B X_b	X_B y
X_B	X_B X_b	X_B y

- As you can see above both daughters will be carriers and both males will have normal vision. This would be the same Punnett square for Hemophilia (X_b y x X_H X_H).

Punnett square example: Carrier female X_B X_b is crossed with a normal vision male X_B y

	X_B	y
X_B	X_B X_B	X_B y
X_b	X_B X_b	X_b y

Here is another more in depth pedigree chart showing Hemophilia:

How to Determine a Pedigree	Diagram of a Pedigree
• For dominant and recessive alleles, upper-case and lower-case letters, respectively, should be used. • If neither parent is affected, the trait cannot be dominant. The trait could be recessive and either parent or both could be heterozygous carriers unless the disease is sex-linked (diagram to the right) • If the father is affected and none of his sons or daughters are affected the trait most likely is recessive. If an affected boy shows up again in the next generation, the trait is sex-linked recessive • If the trait shows up equally between boys and girls and tends to skip generations, the trait is most likely autosomal recessive • In the chart to the right D, P, and V would be represented as (X_h Y) while the unaffected women G, N, S, and U would be represented by (X_H X_H). Women represented by B, I, J, and Q have to be carriers $X_H X_h$. The other women could be either carriers or homozygous dominant. None of the women are homozygous recessive as they don't have the disease.