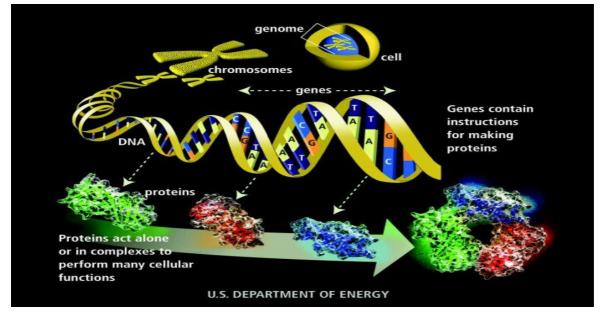
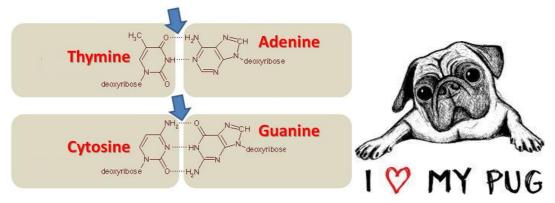
Genetics, DNA & Evolution | Topic Notes

- > Heredity is the passing on of characteristics from one generation to the next.
- > Genes are short regions of chromosomes that contain codes for the production of proteins.
- > Gene expression is the process by which the code in DNA is used to make a protein.



- The proteins associated with DNA are <u>histones</u>. These function in stabilising the DNA and keeping it organised. (analogy = storing Christmas tree lights)
- Chromosomes exist only during mitosis. At all other times it's known as chromatin. Of the 23 pairs of chromosomes in a diploid human cell, the first 22 are called autosomal chromosomes and the 23rd pair are the sex chromosomes (either XX=female or XY=male).
- DNA (deoxyribonucleic acid) is one of 2 types of nucleic acid (the other is RNA). DNA consists of 2 strands attached together by molecules called <u>nitrogenous bases</u> (because they contain Nitrogen). Twisted around into a double helix shape.
- The four nitrogenous bases found in DNA are adenine (A), thymine (T), cytosine (C) and guanine (G). A is always opposite T and C is always opposite G. These <u>complementary base pairs</u> exist due to the *hydrogen bonding* pattern shown below. A and G are known at <u>purines</u> because they contain 2 rings. (Think of PUag dog). C and T are known as **pyrmadines** because they've 1 ring.



The sides of the double helix DNA are mad up <u>deoxyribose</u> sugars (where basses are attached) and <u>phosphate</u> molecules. The sequence of a phosphate bonded to a sugar bonded to a base is called a <u>nucleotide.</u>

> Mechanism of DNA replication:

First, *the double helix unwinds*. Enzymes *opens the 2 strands by breaking the 'H' bonds between the nitrogenous bases*. The exposed bases act as a template for the production of a new strand of DNA. *New nucleotides, which enter from the cytosol are connected to the old bases* (they contain an A, T, C or G base). This assembly is *carried out by the anabolic enzyme, 'DNA polymerase'.* Each new chromosome consists of 1 old strand of bases and 1 new strand. Lastly, the *DNA re-associates itself with histones and recoils* and supercoils into chromosomes. The duplicated *homologous chromosomes are now ready for mitosis*.

DNA replication is *significant* because it allows for cell continuity (the process of cells arising from cells of the same type)

- DNA profiling is a method of producing a unique pattern of bands from the DNA of a person, so that it can be used for identification purposes.
 - 1. **<u>DNA isolation</u>**-the DNA is extracted from the cells using a *detergent* (non-polar solvent dissolves phospholipid bilayer which forms the cell membrane).
 - 2. Cutting-restriction enzyme is added to cut DNA at specific lengths. (everyone's is diff)
 - 3. Separation-gel electrophoresis separates fragments of DNA based on their lengths.
 - 4. <u>Pattern analysis</u>-the gel is *stained* and viewed under *ultraviolet light* and the individuals DNA pattern (AKA DNA profile) is seen.
- > **Genetic screening** is a test of a person's DNA to see if an altered/mutated gene is present.

(E.g. cystic fibrosis= build up of mucus in the lungs)

<u>RNA</u> = ribonucleic acid (mostly found free in the cytoplasm in the form of **ribosome's**). RNA is single-stranded, contains the bases C, G, A and U (uracil) and the sugar ribose.

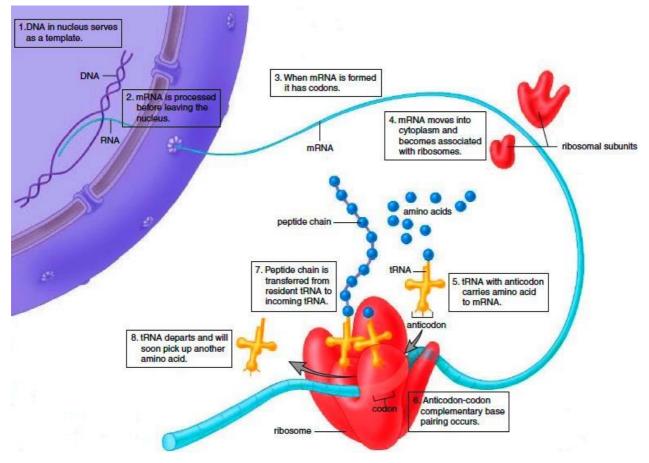
Protein Synthesis

Protein synthesis is the making of a protein using amino acids and the code in messenger RNA.

 Occurs in the cytosol on the surface of *ribosomes* and involves three distinct stages: *transcription, translation* and *protein folding.*

Watch this video from 0:27-2:40 http://www.youtube.com/watch?v=5iS4CRPPDus

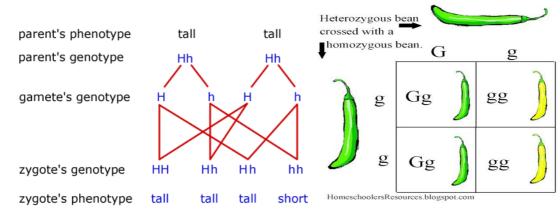
- <u>Transcription</u> is the making of mRNA using a DNA template.
 It occurs in the *nucleus*. Enzymes unwind and break the 'H' bonds between the strands of DNA, exposing *nucleotide bases only at the site of the gene to be transcribed*. The anabolic enzyme, *RNA polymerase synthesises messenger RNA (mRNA*) using one of the strands of DNA.
- <u>Translation</u> is the making of a protein using the code n mRNA.
 It occurs on ribosomes in the cytoplasm. Ribosomes are made from *ribosomal RNA (rRNA).* mRNA interacts with rRNA and slides through its subunits.
- All proteins have their own characteristic shape enabling them to carry out their specific function.
 The long chain of *amino acids* that has been produced *folds* over on itself (it may also attach with other proteins to give the final structure). It is then ready to carry out its function.



- A <u>triplet</u> or <u>codon</u> is a sequence of three bases present on mRNA that codes for one amino acid.
 Amino acids are transported to the ribosome by *transfer RNA (tRNA)*.
- An <u>anti codon</u> is a sequence of three bases present on tRNA that will be complimentary to a codon on mRNA.

Inheritance and Genetic crosses

- ✓ **<u>Genetics</u>** is the study of inheritance.
- ✓ **Inheritance** is the passing on of traits from one generation to the next.
- ✓ **Traits** are physical and chemical characteristics that a living organism possesses.
- ✓ **Gametes** are haploid sex cells.
- ✓ **Fertilisation** is the fusion of 2 haploid gametes to produce a zygote.
- ✓ **<u>Alleles</u>** are alternate forms of genes.
- ✓ **Locus** is the position of an allele or gene on a chromosome.
- ✓ Homozygous: 2 alleles are the same. (often referred to as pure breeding)
- ✓ Heterozygous: 2 alleles are different.
- ✓ **Dominance** occurs where one allele (the dominant one) masks the effect of another allele.
- <u>Recessive</u> occurs where an allele's effect is only expressed when in the homozygous condition.
 We use <u>CAPITAL LETTERS to refer to dominant alleles</u> and <u>lower case letters to refer to recessive</u> <u>alleles</u>. (E.g. A person with brown eyes could have two brown alleles 'BB' (AKA homozygous dominant) or one brown and one blue allele 'Bb', a person with blue eyes can only have 'bb' (AKA homozygous recessive) alleles as the blue allele is recessive).
- ✓ **Genotype** is the genetic make-up of an individual. (BB, Bb, bb)
- ✓ **Phenotype** is the physical make-up of an individual. (brown or blue eyes)
- ✓ A **monohybrid cross** is a genetic mating between two organisms where one gene is studied.
- The *first filial generation* refers to the first generation offspring of any two parents from any living organisms.
- ✓ A genetic cross is a diagram or table showing how characteristics are inherited.



- ✓ Incomplete dominance occurs when a cross between organisms of two different phenotypes produces offspring with a third phenotype that is a mixture of the parental phenotypes.
- ✓ Mendel's first law of segregation:

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- Each cell contains two *factors* (now called alleles) for each trait.
- These factors *separate at gamete formation*, so that each gamete contains only one factor from each pair of factors. (now known as meiosis)
- At fertilisation, the new organism will have two factors for each trait, one from each parent.

✓ Mendel's second law of independent assortment:

Members from one pair of factors separate independently of another pair of factors during gamete formation.

- ✓ A <u>dihybrid cross</u> is a genetic mating between two organisms where two separate genes are studied.
- ✓ Linked genes are genes present on the same chromosome.
- ✓ **Sex linkage** is where a gene is located on a sex chromosome.

The genes for colour vision and blood clotting are present on the 'x' chromosome but not the 'y', hence higher instances of colour blindness and haemophilia are found in males as they only have one 'x' chromosome.

- ✓ A **pedigree study** is a diagram of individuals of a family used to determine patterns of inheritance.
- Non-nuclear inheritance is the passing on of features from one generation to the next without the use of the nucleus.

Mitochondria and chloroplasts are inherited independently of the nucleus. They've their own DNA. Mitochondria are inherited maternally in animals as the cytoplasm of the egg carries the mitochondria. (The sperm cell does have mitochondria, but only the nucleus from the sperm enters the egg)

Genetic Engineering

- **Genetic engineering** is the artificial manipulation and alteration of genes is a process whereby genes are transferred from one organism to another.
- **1.** <u>Isolation</u> (removing DNA from a cell).
- <u>Cutting</u> (removing a gene from a piece of DNA using a *restriction enzyme*) and <u>ligation</u> (joining the gene to a *vector* (a piece of DNA, such as a bacterial plasmid that will carry the gene of interest into a host cell) using *DNA Ligase*).
- 3. <u>Transformation</u> (the uptake of *recombinant DNA*(DNA from 2 diff species)into a bacterial cell)

- **4.** <u>Selection</u> (killing any cells that didn't take up the recombinant DNA) and <u>cloning</u> (producing identical copies of a cell)
- **5.** <u>**Expression**</u> (the stimulation of a cell to produce the product of a particular gene) Applications:
- **<u>Plants:</u>** herbicide-and insect-resistant corn.
- **<u>Animals:</u>** mosquitoes resistant to malaria.
- Microorganisms: insulin produced by bacteria.

A **cloning vector** is a piece of DNA that can accept the **target gene** and replicate e.g. plasmid in bacteria.

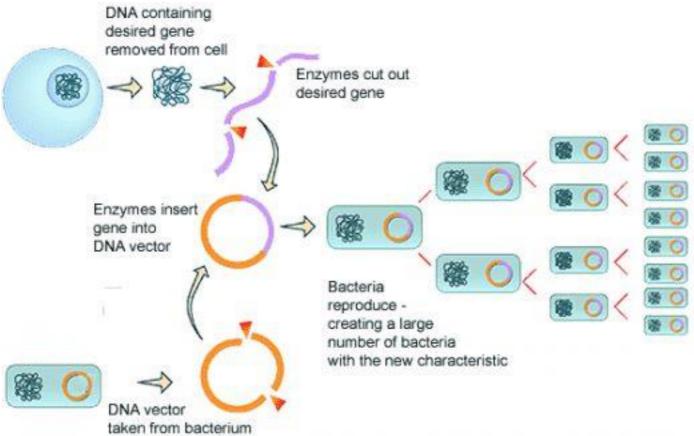
Restriction enzymes cut DNA at specific sites. Genetic 'scissors' that recognises a specific sequence of bases.

DNA ligase is an enzyme that is used to get the foreign DNA to join with the DNA of the cloning vector. Genetic 'glue'.

The altered DNA is called **recombinant** DNA because it recombines after the small section of DNA is inserted into it.

Transgenic organisms are organisms that have been altered using genetic engineering.

GMOs are genetically modified organisms – living things whose DNA has been altered artificially.'Pharming' is the production of foreign proteins by GMOs.



Applications of genetic engineering: (know 1 plant, 1 animal, 1 m/o)

Plant

Weedkiller-resistant crops

Inserting a bacterial gene for herbicide resistance in to **crop plants**, so that when the herbicide is sprayed it will kill weeds but it will not kill the plant.

Animals

Sheep produce a protein to treat emphysema (collapse of the alveoli because they cannot produce a protective protein in the lungs). A human gene for this protein (AAT) has been inserted inot sheep DNA and they can the produce the protein in their milk.

Microorganisms

Bacteria make insulin

Inserting the gene for human insulin into a **bacterium** which then produces human insulin for use by diabetics. This overcomes the danger of people producing antibodies to the pig insulin.

Ethical issues of genetic engineering

Release of GMOs into the environment.

Use of GMOs as a food source.

Concern that animals will suffer as a result of being genetically modified.

The fear that humans, especially zygotes, may be genetically modified – 'designer babies'.