WEEK ONE: BIOLOGICAL DEVELOPMENT

Genes and Genetics

- Parents pass on distinguishable characteristics (eg. eye colour, hair colour). These are genes, the blueprint for our bodies
- Health conditions and behavioural characteristics can also be genetic
- Genes are stored in body cells in the nucleus and are part of chromosomes (long strands of DNA) therefore genes are made up of DNA
- Letter sequence of each gene contains information of building specific molecules
- Genes are copied to a similar substance called RNA. The working parts of the cell read the RNA to create the protein or hormone according the instructions in the DNA
- One characteristic (eg. eye colour) may be influenced by any genes. Each gene codes the instruction for one single protein
- Sometimes genes can contain a *variation* (can be spontaneous or inherited) that disrupts the gene's coded message – this makes the gene not work properly and cause mutations which can directly or indirectly lead to a wide range of conditions

Chromosomes and Sperm and Egg Cells:

- Human cells have 46 paired chromosomes with about 23,000 genes
- Chromosomes numbered from 1 to 22 according to size (1 being the biggest)- numbered chromosomes are called *autosomes*
- In addition to 44 autosomes there are 2 sex chromosomes (XX for women and XY for men)
- The 23,000 genes come in pairs. One gene in each pair is from mother and other from father
- A sperm and egg contain one copy of every gene needed to make up a person (one set of 23 each)
- There are many genes located on the X chromosome but only a few on the Y chromosome

How we Inherit Characteristics:

- We can inherit characteristics in many different ways (eg. blood type can be A, B, AB or O)
- Each variation of a gene is called an ***allele***

Dominant and Recessive Genes:

- Actions of some genes contained in each set of chromosomes appear to be more 'dominant' over others (eg. brown eye colour is usually more dominant than blue eye colour)

Dominant and Recessive Blood-Group Inheritance:

- The A allele is dominant over the O allele so someone with one of each has the blood group AO (a person needs two O alleles to have the blood group O)
- If someone has an A and O allele (AO), their blood group will be A

Co-Dominant Genes:

- Sometimes each allele in a gene pair carries equal weight and will show up as a combined physical characteristic (eg. with blood groups, the A allele is as strong as the B allele – person will be AB)

Genotype and Phenotype:

- ***Phenotype*** is the observable trait (eg. blood group A)
- ***Genotype*** is the genes that produce the observable trait (eg. AO)

Chemical Communication:

- Cells only need some specific genes to be turned on to perform particular functions. Unnecessary genes are switched off
- Genes communicate with the cell in chemical code (genetic code)
- Cells carry out the gene instructions to the letter
- Cells reproduce by copying its genetic information then splitting it in half, forming two individual cells. Genetic mutations can sometimes be made
- Genetic mutations are permanent – some causes of mutation include exposure to:
    - Radiation
    - Chemicals
    - Cigarette smoke

Variations in the Genes in the Cells:

- Sperm and egg cells known as ***germ cells***
- All other body cells are called ***somatic cells***
- If variation in the gene occurs spontaneously in the somatic cells they may develop a condition related to gene change however won't pass this onto their children (eg. skin cancer from damaged skin cells from UV radiation)
- If variation occurs in germ cells, person's children each have 50% chance of inheriting the mutated gene.
- People can still be genetic carriers of faulty genes even if they themselves do not have the condition
- A correct copy of a gene overrides the faulty copy

Genetic Conditions:

- To date 1,700 conditions have been found cause directly or indirectly by changes in genes
- Approx. half of all miscarriages are caused by changes in the total number of genes in the developing baby
- Approx. half the Australian population will be affected at some point in their life by an illness that is at least partly genetic in origin
- Three ways in which genetic conditions can happen:
    1. The variation in gene makes it faulty (a maturation) happens spontaneously in the formation of the egg or sperm or at contraception
    2. The faulty gene is passed from parent to child and may directly cause a problem that affects the child at birth or later in life
    3. The faulty gene is passed from parent to child and may cause a genetic susceptibility. Environmental factors such as diet and exposure to chemicals, combine with this susceptibility to trigger the onset of the disorder

Genetic Predisposition (Inherited Susceptibility):

- In many cases being born with a faulty gene just means you are at increased risk of developing a certain condition
- Many conditions involving genetic susceptibility (eg. types of cancer) need to be triggered by environmental factors such as diet and lifestyle (eg. prolonged exposure to the sun is linked to melanoma)

Genes and Genetics – Related Parents:

- Relayed parents are more likely than unrelated parents to have children with health problems or genetic disorders because the two parents share one or more common ancestors and so carry some of the same genetic material.
- Increases risk of both parents carrying the same mutated gene

Autosomal Recessive Genetic Disorders:

- If two parents have the same altered gene they may pass their copy onto a child so the child receives both altered copies – ***autosomal recessive inheritance*** (the parents are carriers of the genetic condition but are unaffected themselves)
- Autosomal recessive genetic disorders include:
    - Cystic Fibrosis
    - Phenylketonuria (PKU)

Degrees of Relationship:

- ***First degree relative*** – shares half their genetic information (eg. siblings, non-identical twin, parents, children)
- ***Second degree relative*** – shares one quarter of their genetic information (eg. half-siblings, uncles, aunts, nephews, nieces, grandparents)
- ***Third degree relatives*** – share one eighth of their genetic information (eg. first cousins, half-uncles, half-aunts, half-nephews, half-nieces