# CB3 – Genetics (paper 1)

CB3a Meiosis

What is a zygote?
Humans start life as a single fertilised egg cell (a zygote). This is formed when 2 gametes fused during fertilisation. The zygote forms a ball of cells using a form of cell division called mitosis.

What is DNA?
The instructions controlling each individual cell are found as a code in a molecule called DNA. The DNA of an organism is its

D meiosis (simplified to show just two different types of chromosome)
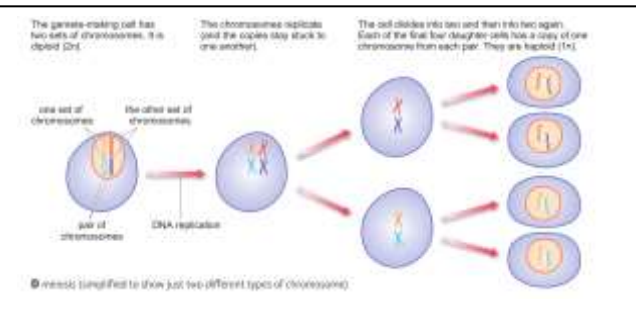
What is the human genome?
The human genome is found on 46 very long molecules of DNA and each molecule is inside a chromosome. Along the length of a DNA molecule are sections that contain a code for making a protein. The DNA sections are genes.

What is a protein?
Proteins are polymers made by linking different amino acids together in a chain.

Why do gametes have to be haploid?
If you diploid cells joined in fertilisation, the zygote would have 4 sets of chromosomes, so gamete need to have just 1 set of chromosomes. They have to be haploid (the shorthand for a haploid cell is 1n).

How many chromosomes does a human have?
There are 23 different chromosomes in humans and most nuclei contain 2 of each type. So a human body contains 2 sets of 23 chromosomes making 46 in total. A cell like this is diploid (written as 2n in shorthand).

CB3bi DNA

Describe a molecule of DNA
A molecule of DNA contains 2 strands, each of which forms a shape called a helix. The strands

How many bases are there in DNA?
There are 4 bases in DNA: adenine, thymine,

What are bases attached to?
Each base is attached to a sugar and each sugar is attached to a phosphate group. The sugars and phosphate groups form the backbone of the DNA strands.

Why do parts of DNA bases have a hydrogen bond?
Parts of DNA bases have very slight electrical charges. A negatively charged part of one base attracts a positively charged part of another

What is the DNA code?
The order of bases in a gene contains the coded instructions for a protein. We all have slight difference in our

DNA double helix
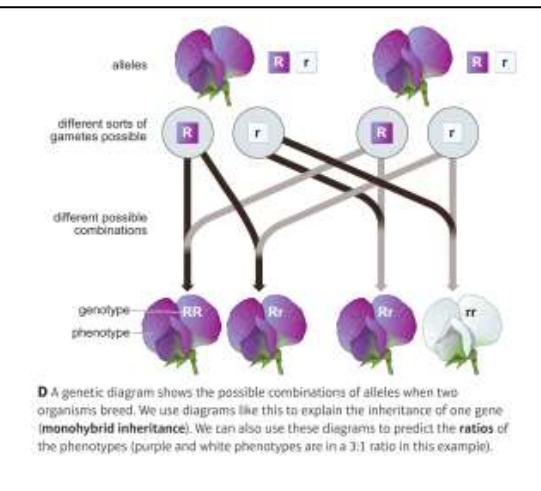
B the structure of DNA
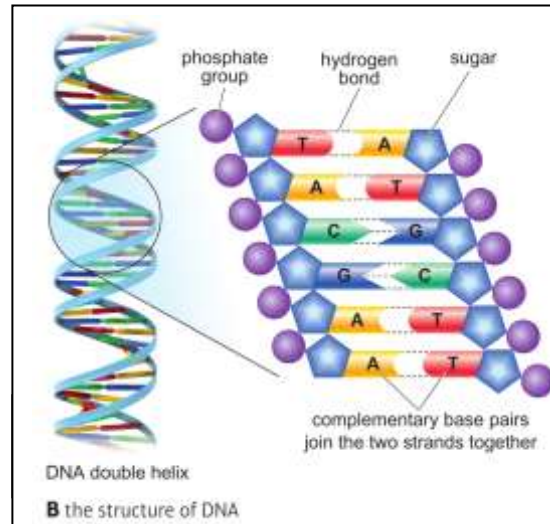
CB3e Gene Mutation

Why do we use Punnett squares?
Punnett squares are used to work out the probability of different phenotypes caused by alleles.

How do mutations happen?
Mutations happen when there is a mistake in copying DNA during cell division. For example one base in a DNA sequence might be replace by another. This can happen naturally of if there is damage to the DNA caused by radiation or other harmful substances.

What affect will a mutation have?
Sometimes a mutation produces an allele that causes a big change in the protein that is produced. This will affect how the body works. Some mutations may only have a small effect on the protein that is produced, while some will not change the protein at all.

What is the human genome project?
The human genome project mapped the bases pairs in the human genome. Mapping a person's genome can indicate their risk of developing diseases that are caused by different alleles of genes. It can also help identify which medicines might be best to treat a persons' illness.

CB3d Inheritance

D A genetic diagram shows the possible combinations of alleles when two organisms breed. We use diagrams like this to explain the inheritance of one gene (monohybrid inheritance). We can also use these diagrams to predict the ratios of the phenotypes (purple and white phenotypes are in a 3:1 ratio in this example).

What are your sex chromosomes?
Two of your chromosomes determine what sex you are. They are your sex chromosomes and there are two types: X and Y. Females have two X sex chromosomes and males have one X and one Y.

What is the difference between an egg cell and a sperm cell?
A woman's gametes all contain an X sex chromosome. The male sperm cells may contain either an X or a Y (diagram A).

What is a Punnett square?
Punnett squares (diagram B) demonstrate inheritance. The boxes show the possible genotypes. Two boxes contain XX and two contain XY. The probability of being male is 50%.

Why do we use Punnett squares?
Punnett squares are used to work out the probability of different phenotypes caused by alleles.

The boxes show the possible combinations in the offspring.

B Punnett square for human sex determination
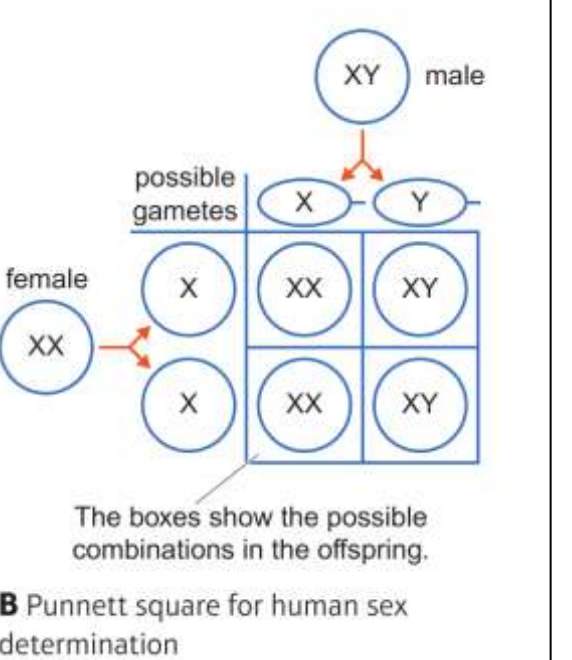
CB3c Alleles

What is an allele?
Genes for the same characteristic (e.g. eye colour) can contain slightly different instructions that create variations. Different forms of the same gene are called alleles.

What produces genetic variations?
There are 2 copies of every chromosome in a body cell nucleus, a body cell contains 2 copies of every gene. Each copy of a gene may be a different allele. The different combination of alleles in each person give us slightly different characteristics.
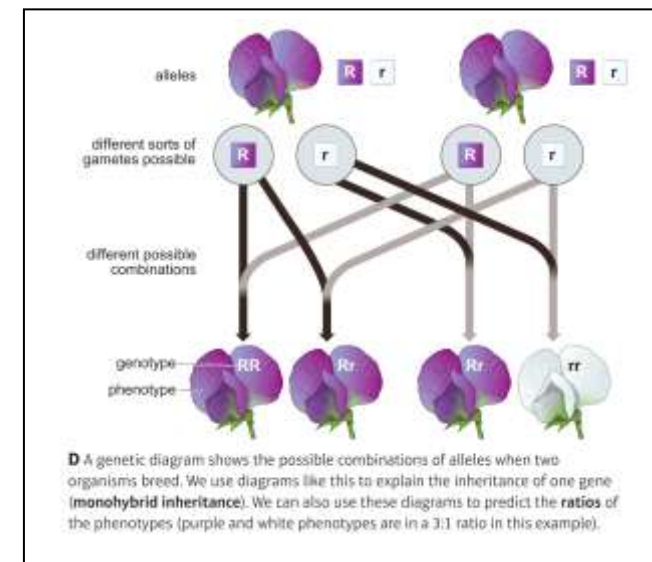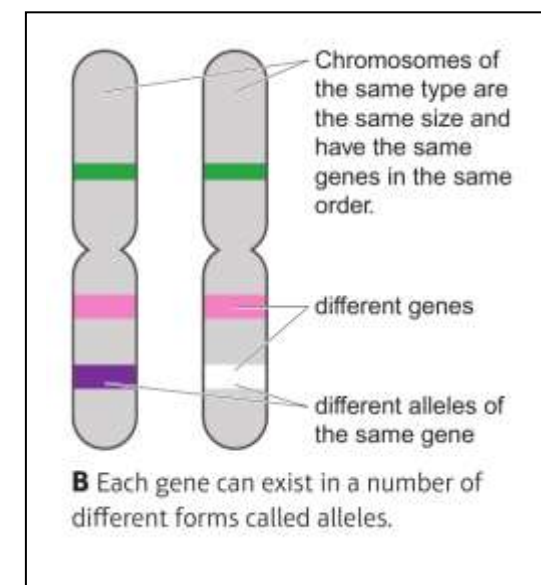
Explain the term homozygous
If both alleles for one genes are the same an organism is homozygous for that gene.

What is the difference between a genotype and a phenotype?
The alleles in an organism is its genotype. What the organism looks like is its phenotype.

What is a dominant characteristic?
Characteristics from a dominant allele will always be expressed. A dominant allele is shown by a capital letter, the recessive allele has the lower case version of the same letter. The letter for the dominant allele is always written before the recessive one.

What is a recessive characteristic?
A recessive characteristic is only seen if both alleles are recessive (diagram D).

Explain the term heterozygous
If the alleles are different an organism is heterozygous.

Chromosomes of the same type are the same size and have the same genes in the same order.

different genes

different alleles of the same gene

B Each gene can exist in a number of different forms called alleles.

D A genetic diagram shows the possible combinations of alleles when two organisms breed. We use diagrams like this to explain the inheritance of one gene (monohybrid inheritance). We can also use these diagrams to predict the ratios of the phenotypes (purple and white phenotypes are in a 3:1 ratio in this example).

CB3f Variation

What is genetic variation?
Genetic variation is caused by the different alleles inherited during sexual reproduction.

What is environmental variation?
Many characteristics also show environmental variation because they are affected by their surroundings. For example how well a plant grows is affected by how much light, water and nutrients it gets.

In what two ways can variation be grouped?
A discontinuous variation is where the data can only take a limited set of values e.g. the number of leaves on a plant. Continuous variation is where the data can be any value in a range e.g. the length of a leaf on a tree. Continuous data for variation often forms a bell-shaped curve known as a normal distribution.

Key Vocabulary Definitions

Acquired characteristics

Alleles – most genes come in different versions called alleles.

Bases – a substance that helps make up DNA. There are four bases in DNA, shown by the letters A, C, G and T.

Chromosome – a thread-like structure found in the nuclei of cells. Each chromosome contains one long DNA molecule packed with proteins.

Complementary base pairs - two DNA bases that fit into each and link by hydrogen bonds

Daughter cell – a new cell produced from the division of a parent cell.

Diploid – a cell or nucleus that has two sets of chromosomes.

Dominant – describes an allele that will always affect a phenotype as opposed to a recessive allele, whose effect will not be seen if a dominant allele is present.

Double helix – the shape of a DNA molecule consisting of two helices.

Family pedigree chart – a chart showing the phenotypes and sexes of several generations of the same family to track how characteristics have been inherited.

Gametes – a haploid cell produced the meiosis used for sexual reproduction.

Genes – A section of the long strand of DNA found in a chromosome which often contains instructions for a specific protein.

Genetic diagram – a diagram showing how the alleles in two parents may form different combinations in offspring when the parents reproduce.

Genome – all of the DNA in an organism. Each body cell contains a copy of the genome.

Genotype – the alleles for a certain characteristic that are found in an organism.

Haploid – a cell or nucleus that has one set of chromosomes. Gametes are haploid.

Hydrogen bond – a weak force of attraction caused by differences in the electrical charge on different parts of different molecules.

Meiosis – a form of cell division in which one parent cell produces four haploid daughter cells.

Mutation – a change to a gene caused by a mistake in copying the DNA base pairs during cell division, or by the effects of radiation or certain chemicals.

Normal distribution – when many individuals have a middle for a feature with fewer individuals having greater or lesser values. This sort of data forms a bell shape on charts and graphs.

Phenotype – the characteristics produced by a certain set of alleles.

Polymers – a long-chained molecule made by joining many smaller molecules (monomers) together.

Punnett square – a diagram used to predict the characteristics of offspring produced by two organisms with known combinations of allele.

Recessive – describes an allele that will only affect the phenotype if the other allele is also recessive. It has no effect if the other allele is dominant.

Replicate – when DNA replicates, it makes a copy of itself.

Sex chromosomes – a chromosome that determines the sex of an organism.

Variation – difference in the characteristics of organisms.

Zygote – a fertilised egg cell.

Calculations