Genetics Notes

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Genetics is one of those topics that people sometimes are a bit put off by. But I have found that if you can master the main definitions then the rest of the topic is a matter of applying those definitions. There is a little bit of problem solving but nothing too difficult at all, let's get straight into the definitions 😊

Chromosome: a thread like structure found in the nucleus of a cell. Each chromosome contains one enormously long DNA molecule which is packed with proteins. Humans have 46 chromosomes.

Gene: a short section of DNA that codes for the production of a protein.

Allele: a single version of a gene. For example, the gene for blue eyes is "bb", each of these letters represents an allele. Two alleles make a gene.

Dominant: a dominant allele is always expressed if it is present. Dominant alleles are represented by a capital letter.

Recessive: a recessive allele is only expressed if both alleles are recessive. Recessive alleles are represented by a lowercase letter.

Homozygous: when both alleles for a gene are the same.
There are two types of homozygous, you can have **homozygous dominant**, where both alleles are dominant for example "BB". You can also have **homozygous recessive**, where both alleles are recessive for example "bb".

Heterozygous: when both the alleles for a gene are different. We **must** describe that one allele in a heterozygous gene is dominant and one is recessive. For example, "Bb or Hh"

Genotype: the gene for a certain characteristic found in an organism, for example the genotype for blue eyes is "bb" or the genotype for brown eyes could be "Bb or BB".

Phenotype: the physical appearance of the organism. So, what the organism looks like.

Gamete: a sex cell. In humans these are the ova or eggs in females and sperm in males. Each has 23 chromosomes in the nucleus of the cell.

Zygote: a diploid cell formed by the fusion of two haploid gametes during fertilisation.

Please learn all of the above off by heart, it will make the rest of the topic much easier to understand and boost your ability to a correctly answer questions 😊. It is a lot of definitions… but break it up… learn 2 or 3 a day.

So, where does our modern understanding of genetics come from? Interestingly, an Austrian monk.

Gregor Mendel

Who was Gregor Mendel?
Gregor Mendel (1822-1884) was an Austrian monk, he is the father of modern genetics. He studied the inheritance of different characteristics in pea plants.

What sort of work did Mendel do?
Mendel bred pea plants to see what characteristics they would inherit from their parents. For example, he bred tall pea plants with short pea plants, all the offspring produced were tall. As far as people knew at the time this went against the prediction, it was thought that the heights would blend to produce medium height plants. If he bred two of the offspring together, most of the offspring were tall, but some were small. We know this makes sense as the allele for a tall height in pea plants is dominant and the allele for a short height is recessive.

Why was Mendel's work difficult?
Mendel's work was not easy as almost nothing was known about DNA, genes or alleles so he called the traits like colour and height "units".

Why was Mendel's work not accepted during his lifetime?
Mendel's work was not accepted by most scientists during his own life for three main reasons:
1. Mendel found it difficult to communicate his results with other scientists.
2. Mendel published in a scientific journal that was not very well known so not many people read it.
3. Mendel of course could not explain the science behind why characteristics were inherited.

Mono Hybrid Inheritance

Quite a lot of exam questions about genetics have within them a question or problem involving monohybrid inheritance. **Mono** of course **means one** or single and **hybrid**, in this case, relating to a particular **trait**.

Monohybrid inheritance: is the inheritance of characteristics controlled by a single gene. For example, eye colour.

Monohybrid inheritance is usually done using a Punnett square or a genetic diagram. They both get the same result so whichever one you prefer really. I find the Punnett squares the most straight forward to use 😊

First, let's look at how you do a Punnett Square.

- **State the genotypes (gene) of the parents**. Usually, the question will tell you what letters it wants you to use for a gene. If it doesn't you can use any letter. But pick one where the capital is quite different from the lower case version.
- **Give the alleles for each parent.**
- **Treat the Punnett square like a multiplication grid and complete the possible outcomes.**

Below I have shown both the **Punnett square** for the inheritance of height in pea plants.
Let's cross a homozygous dominant individual (TT) with a heterozygous (Tt) individual using a **Punnett square**:

Parent 1 – homozygous dominant

Parent 2 – Heterozygous

	T	T
T	TT	TT
t	Tt	Tt

Offspring

This cross produces **all tall plants**, as the allele for height is dominant. The offspring produced from the first cross are called the **F1 generation**.

F1 generation:

Phenotype: Tall

Genotype: TT & Tt

Now let's cross the two heterozygous individuals from the first cross.

Parent 1 – heterozygous

Parent 2 Heterozygous

	T	t
T	TT	Tt
t	Tt	tt

Offspring

F2 Generation:

Phenotype: 3 x tall and 1 x small

Genotype: TT, Tt & tt

We could be asked for probabilities in these kinds of questions, this is usually given as a fraction or as a ratio.
A question could ask:

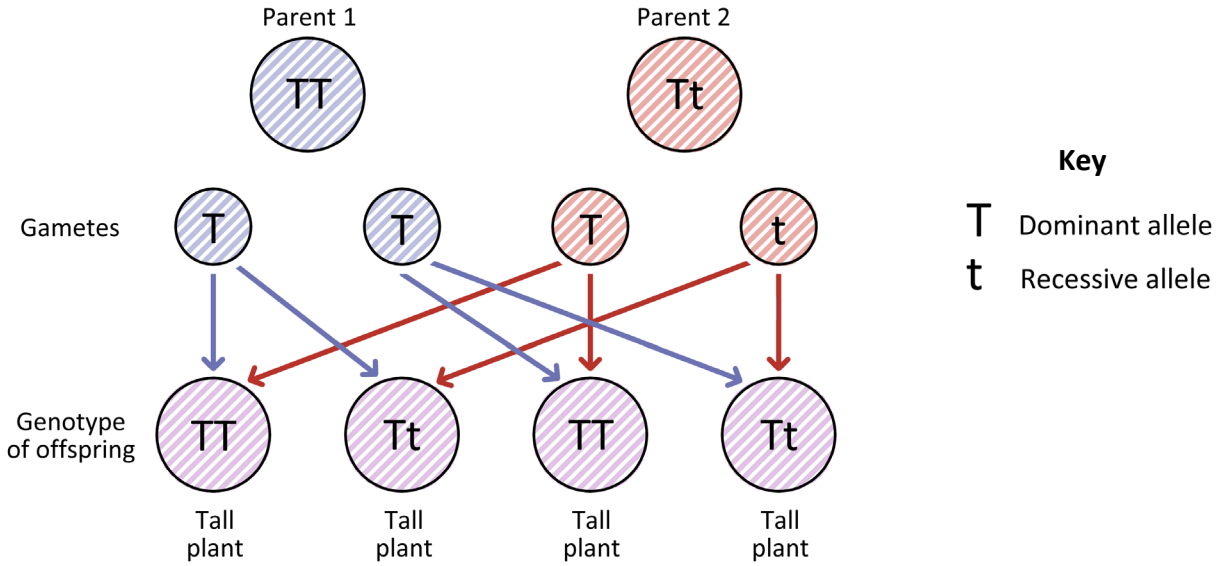
"What is the probability of a short plant being produced from two heterozygous parents?"

Any answer that communicates one quarter, or 25% or 0.25 is perfectly correct.

Genetic Diagrams

Let's cross a homozygous dominant individual with a heterozygous individual using a **genetic diagram**.

In this type of diagram, you must break the two parents' genotype down into their four individual alleles and cross the alleles to determine their outcomes.



This cross produces **all tall plants**, as the allele for height is dominant. The offspring produced from the first cross are called the **F1 generation**.

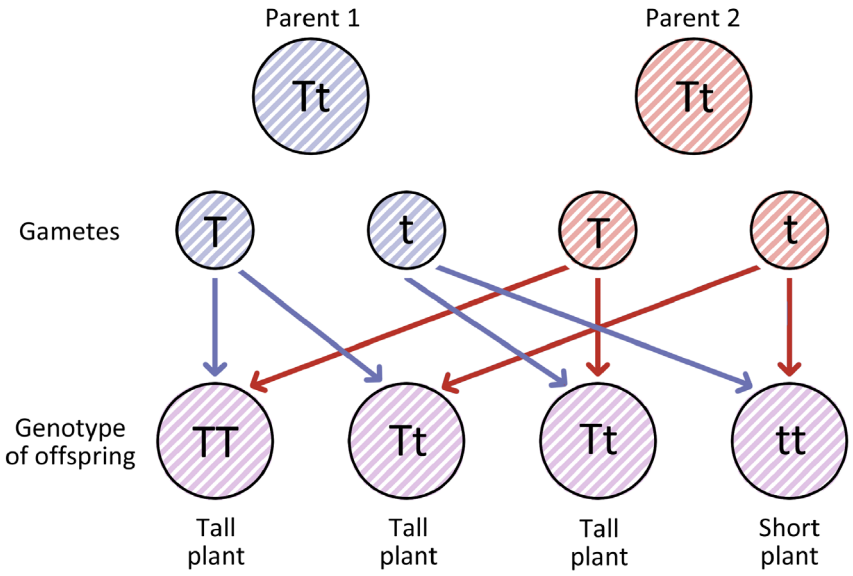
F1 generation:

Phenotype: Tall

Genotype: TT & Tt

Exam note: as you can see the Punnett square and genetic diagram for the cross between a homozygous dominant individual and a heterozygous individual produce the same answer.

Now let's cross the two heterozygous individuals from the first cross.



This cross produces **3 tall and 1 small plants**, as the allele for height is dominant. The offspring produced from the second cross are called the **F2 generation**.

F2 generation:

Phenotype: 3 x tall and 1 x short plant

Genotype(s): TT, Tt & tt

Pedigree Analysis

Questions involving pedigree analysis appear quite regularly at GCSE. These are family trees which can show how traits or genetic diseases are inherited in a family. These questions always have a **key** which shows the **gender of the individuals** and if they are **affected** by a trait, if they are **carriers** of the trait or if they do **not carry** the trait at all. Please see the example of a family tree below.

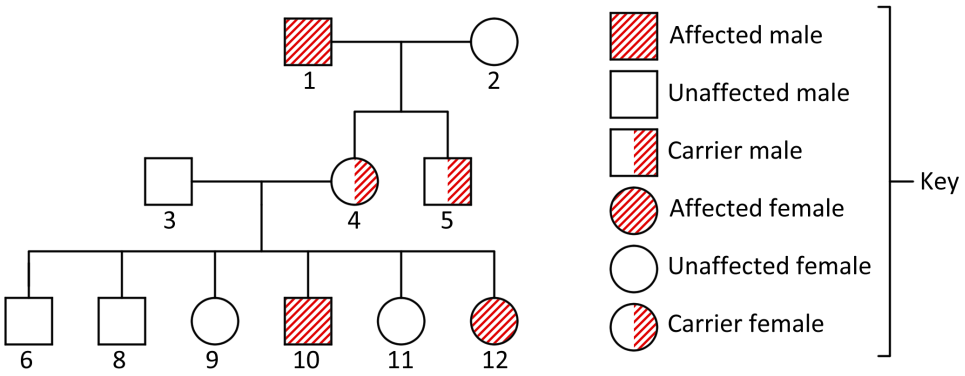
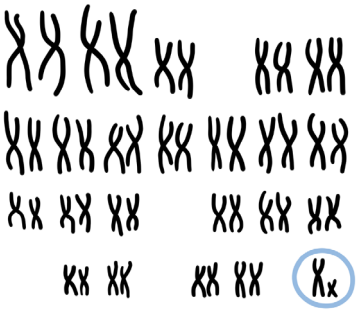


Diagram note: Females are traditionally represented by **circles** and **males** are traditionally represented by **squares**. Affected individuals are often represented by greyed shapes. Individuals that are carriers are often represented by shapes that are half grey and half white.

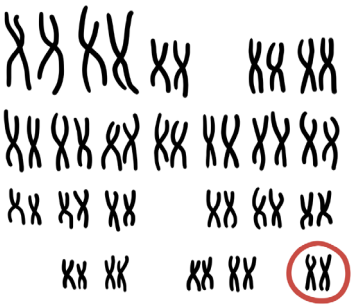
Sex chromosomes and Sex Determination

Humans have 23 pairs of chromosomes (46 individual chromosomes) the **sex chromosomes are your 23rd pair (the last two chromosomes)**. These chromosomes determine your gender, all ovum cells carry an X chromosome, it's the incoming sperm that carries either another X or a Y chromosome.

XX = Female **XY** = Male

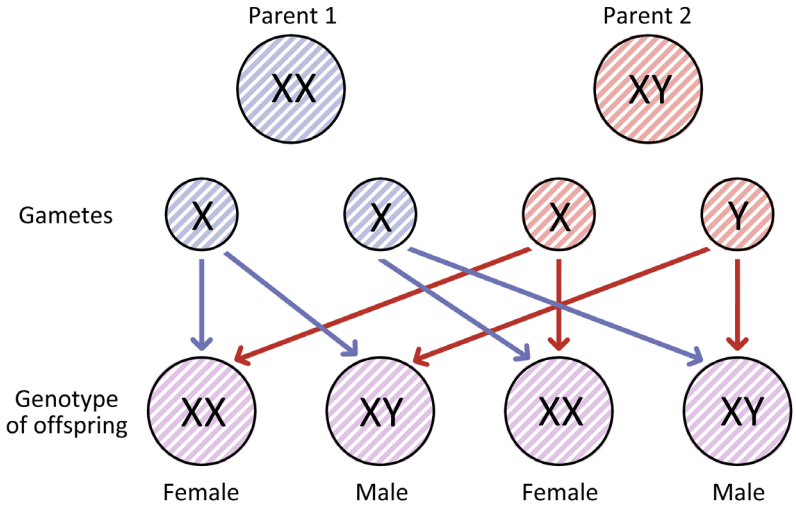


The chromosomes of a human male.



The chromosomes of a human female.

Below is the genetic diagram showing the formation of a person's **sex chromosomes**. You could be asked to do a Punnett square of the cross, the results are exactly the same of course.



Sources of Variation

Genetic variation – different characteristics caused by mutations and sexual reproduction.

A mutation: a change in a gene or chromosome. It is not a very common occurrence; it is a random change in the genetic material and it can be inherited.

Mutagen: a substance that causes a mutation.

Mutations occur naturally, in fact they are how organisms evolve and change over time. Mutations can have positive as well as negative effects.

Mutations can be caused by:

- **A chemical mutagen**, such as tar in cigarette smoke.
- **Ionising radiation** – such as UV, X rays and gamma rays.

The higher the dose of either mutagen the higher the chance of a mutation occurring.

Recall that: a mutation is a change in gene or chromosome.

So, the sequence of DNA bases A, T, C & G is altered. If the protein produced is an enzyme, a mutation may change the shape of the active site, causing the protein to not function or function better, or have no effect at all.

Most mutations have no effect on the phenotype.

Sexual reproduction – the random nature of sexual reproduction, in that any one of hundreds of millions of sperm can fertilise any one of thousands of eggs.

Environmental variation – all organisms are affected by the environment in which they live.
A good example of environmental variation is a scar on your body. Any scars may have come about due to an accident or disease; these cannot be inherited from a parent.

Also, if it's quite sunny where you live you might have tan. This could fade away in the winter.

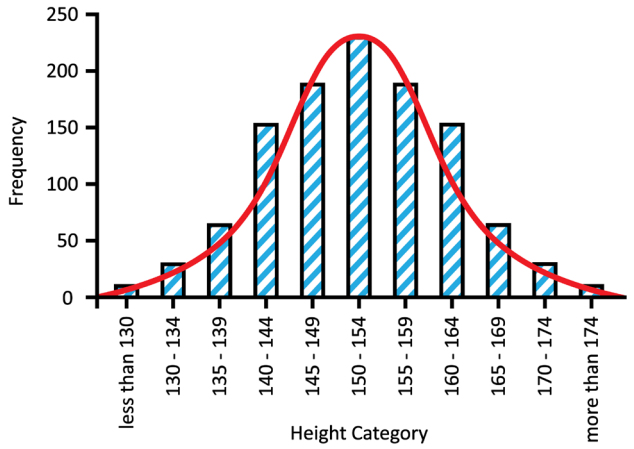
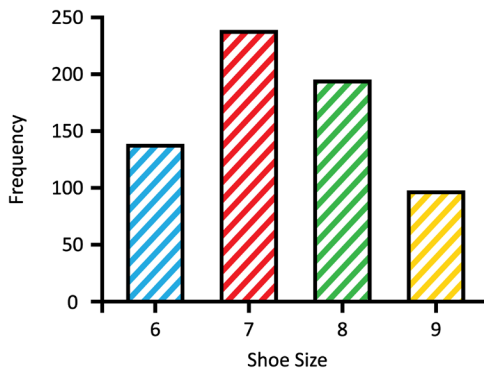
Note: many characteristics are controlled by both a combination of both genetic and environmental causes. This often leads to arguments of "nature versus nurture" but that's more of a political thing.

There is extensive genetic variation between individuals in a population of a species. This is due to mutations. Variation can be grouped into two categories.

- **Continuous variation** – this is where the data can be any value. A good example of continuous data is height, everyone has a slightly different height.
- **Discontinuous variation** – this is where the data must fall into categories. A good example is shoe size, for example you could have a size 3 or 4 or 10 shoes etc.

Be aware how each type of variation looks as a graph, this has come up as a multiple choice question many times.

Continuous Data



Discontinuous Data