HUMAN GENETICS

GLOSSARY

Chromosome – cellular structure composed of DNA which contains inherited information
Gamete – sex cell (sperm & eggs)
Dominant – a gene that mask the other gene in a pair
Recessive – a gene that is hidden by the dominant gene, both genes must be present for this trait to appear in the phenotype
Heterozygous – where the two genes control a specific trait are different
Homozygous – where the two genes which control a specific trait are the same
Genotype – the pair of genes
Phenotype – how the genes are expressed (appearance)
Allele – one member of pair of genes
Diploid cells – cells with two chromosomes from each homologous pair
Haploid cells – cells with one chromosomes from each homologous pair found in the sex cells in humans
Gene 0 – a unit of heredity
Locus – a specific location on a chromosome where a gene can be found

GREGOR MENDEL

The discoveries of Mendel formed the basis of modern genetics. The time he devoted to plant breeding research enabled him to discover basic principles of heredity. For his experiments he used pea plants because they displayed several traits in one of two contrasting forms. For example, the seeds of a plant were either wrinkled or smooth. The stem length was either tall or short. He crossed plants with contrasting traits enabling him to ultimately explain these events with his basic laws of heredity.

> *Principle of Dominance* – states that genes which prevent the expression of a recessive trait are dominant over those recessive genes
>
> *Principle of Segregation* – states that during meiosis only one gene from each gene pair will be passed on to future offspring
>
> *Principle of Independent Assortment* – states that during meiosis all gene pair sorted independently thus resulting in a variety of gene combinations
>
> *Principle of Unit Characteristics* – states that all traits are controlled by two genes, one from the egg and one from the sperm

Probability & the Punnett Square

The principle of heredity helps geneticists to predict the *likely* results of genetics crosses. For example, Mendel's experiments revealed that when 2 plants, heterozygous for yellow seeds are crossed, approximately 1 of every 4 seeds will be green. Using the Punnett Square, we can predict

this and understand what has happened. Because the chromosomes pairs separate during the meiosis, there will be two possible gametes from each parent. Since we are dealing with only one trait we call this a **monohybrid cross**.
To set up a Punnett Square we need to start with the following information:

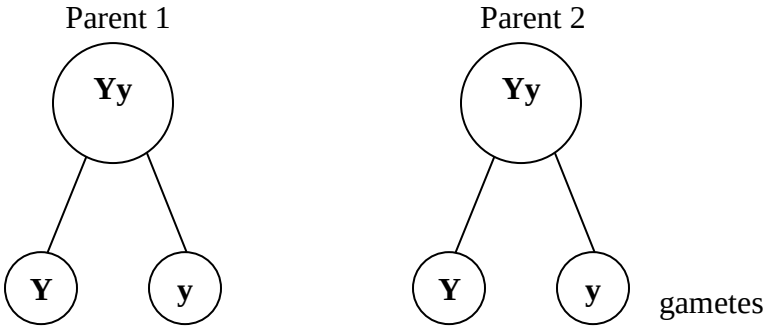
Y – Yellow (dominant)
y – green (recessive)

We then can find the genotype of each parent:

In this case our cross will be **Yy ✕ Yy**

From this we determine the possible gametes from each parent:

In our example **Yy ✕ Yy,** each parent can contribute one dominant gene, here denoted by the **Y** and one recessive gene, here detonated by the **y .**



When then set up your Punnett square in the following manner:

	Y	y
Y	YY	Yy
y	Yy	yy

From this Punnett square we can extract the following information:

Genotypic Ratio: 1 YY: 2Yy : 1yy
or 25% YY : 50% Yy : 25% yy
or ¼ YY: ½ Yy : ¼ yy

Phenotypic Ratio: 3 Yellow : 1 Green
or 75% Yellow : 25% Green
or ¾ Yellow : ¼ Green

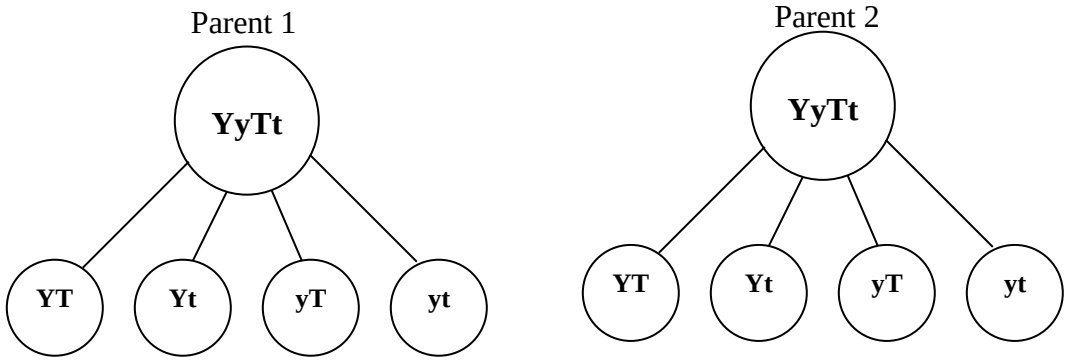
A **dihybrid cross** involves 2 pairs of alleles which code for 2 separates traits.
We first determine the traits we are dealing with :

Y – Yellow (dominant)
y – Green (recessive)
T – Tall (dominant)
t – Short (recessive)

Our cross involves two plants, both of which are heterozygous for both traits. We can then determine the genotype of both plants:

YyTt ✕YyTt

We then must determine the gametes



	YT	Yt	yT	yt
YT	YYTT	YYTt	YyTT	YyTt
Yt	YYTt	YYtt	YyTt	Yytt
yT	YyTT	YyTt	yyTT	yyTt
yt	YyTt	Yytt	yyTt	yytt

From this we can determine the following :

Phenotypic Ratio: 9 Tall &Yellow: 3 Tall & Green : 3 Short & Yellow : 1 Short & Green

Or $^{9}/_{16}$ Tall & Yellow: $^{3}/_{16}$ Tall & Green : $^{3}/_{16}$ Short & Yellow : $^{1}/_{16}$ Short & Green

Genotypic Ratio: 1YYTT: 2 YYTt : 1 YYtt : 2YyTT : 4YyTt : 2Yytt : 1yyTT: 2yyTt : 1yytt

Or $^{1}/_{16}$YYTT: $^{2}/_{16}$ YYTt : $^{1}/_{16}$ YYtt : $^{2}/_{16}$YyTT : $^{4}/_{16}$YyTt : $^{2}/_{16}$Yytt : $^{1}/_{16}$ yyTT:

$^{2}/_{16}$ yyTt : $^{1}/_{16}$ yytt