

### Key words for topic

**Genotype** Genetics makeup of an organisms - the alleles present.

**Phenotype** The expression of genes (also affected by the environment).

**Homozygous** A pair of homologous chromosomes carrying same allele for a gene (e.g. AA, aa).

**Heterozygous** A pair of homologous chromosomes carrying different alleles for a gene (e.g. Aa).

**Recessive allele** Allele only expressed in the absence of dominant alleles

**Dominant allele** Allele always expressed in phenotype.

**Codominant** Multiple alleles are equally dominant and expressed in the phenotype.

**Sex-linkage** A gene with a locus in the X chromosome.

**Autosomal linkage** Genes in the same chromosome (not sex chromosome). An autosomal chromosome is any chromosome other than sex chromosomes.

**Epistasis** When one gene modifies the expression of a different gene.

**Monohybrid** Genetics inheritance cross of trait determined by one gene.

**Dihybrid** Genetics inheritance cross of trait determined by two gene.

**Gene pool** All alleles of all genes within population.

### Key words for topic (cont)

**Allele frequency** Proportion of an allele in a gene pool.

Some of these terms will be further explained in this cheat sheet.

### Notation systems for topic

**Type of inheritance** **Coding**

**Monohybrid** Single letter capital / lower case e.g. A, a

**Codominance** Gene<sup>allele</sup> e.g. C<sup>W</sup>C<sup>R</sup>, I<sup>A</sup>I<sup>B</sup>

**Multiple alleles for one gene (more than two)** Gene<sup>allele</sup> e.g. I<sup>A</sup>I<sup>O</sup>

**Sex linkage** Chromosome<sup>allele</sup> e.g. X<sup>R</sup>X<sup>r</sup>, X<sup>r</sup>Y

**Autosomal linkage and epistasis** Single letters e.g. Aa Bb

### Monohybrid crosses

**Worked example:** Cystic fibrosis is caused by a recessive allele, what is the probability that two carrier parents will have a child with cystic fibrosis?

Parent genotypes ("both carriers"): Ff x Ff

Each gamete will carry either the F or f allele and can fuse with gametes containing either F or f alleles too because parents are heterozygous.

	F	f
F	FF (no CF)	Ff (no CF)
f	Ff (no CF)	ff (CF)

Therefore probability of child with cystic fibrosis = 1/4 = 25%.

### Codominant inheritance

**Worked example:** A flower can be three colours: white, red or pink. The alleles for white and red are codominant to form pink flowers. If two pink flowers reproduce, what is the probability of forming a red offspring flower?

Pink flowers must have the alleles both for red and white as they are codominant to produce pink. They are therefore heterozygous.

Parent genotype: C<sup>R</sup>C<sup>W</sup> x C<sup>R</sup>C<sup>W</sup>

	C <sup>R</sup>	C <sup>W</sup>
C <sup>R</sup>	C <sup>R</sup> C <sup>R</sup> (red)	C <sup>R</sup> C <sup>W</sup> (pink)
C <sup>W</sup>	C <sup>R</sup> C <sup>W</sup> (pink)	C <sup>W</sup> C <sup>W</sup> (white)

Therefore probability of forming a red offspring is 1/4 = 25%.

### Multiple alleles inheritance

**Key example: Blood groups**

There are three alleles for blood group: A, B and O, represented as I<sup>A</sup>, I<sup>B</sup> and I<sup>O</sup>.

Both I<sup>A</sup> and I<sup>B</sup> are codominant to form phenotype group AB, and I<sup>O</sup> is recessive.

**Worked example:** parents with blood group AB and O reproduce. What so the probability that they will produce an offspring with blood group A?

Parent genotype: I<sup>A</sup>I<sup>B</sup> x I<sup>O</sup>I<sup>O</sup>

	I <sup>A</sup>	I <sup>B</sup>
I <sup>O</sup>	I <sup>A</sup> I <sup>O</sup> (group A)	I <sup>B</sup> I <sup>O</sup> (group B)
I <sup>O</sup>	I <sup>A</sup> I <sup>O</sup> (group A)	I <sup>B</sup> I <sup>O</sup> (group B)

Therefore probability is 2/4 = 50%.

### Sex-linkage

Sex linked alleles only occur in X chromosomes as Y chromosomes contain less genetic information (too small to carry more). This makes males more likely to carry recessive sex-linked disorders as their homologous Y chromosome cannot contain the dominant allele.

### Sex-linkage (cont)

**Worked example:** Colour blindness is caused by a recessive sex-linked allele only found in the X chromosome. If a colour blind male reproduces with a heterozygous female, what is the probability that they would have a colour blind child?

**Parent genotype:**  $X^B X^b \times X^b Y$

	$X^B$	$X^b$
$X^b$	$X^B X^b$	$X^b X^b$
Y	$X^B Y$	$X^b Y$

The probability of a colour blind child is therefore  $2/4 = 50\%$ .

Be sure to read the question carefully.

Some exam questions will choose animals where the female has XY chromosomes and the male has XX chromosomes which can throw you off.

### Epistasis

One gene affects the expression of another, therefore multiple genes will be at play here.

**(Common) worked example:** Labradors have two genes which will affect their fur colour. The first gene controls which colour is expressed. If allele B is expressed, the dog will be black. If allele b is expressed, the dog will be brown. The second gene codes for pigment production. If allele E is expressed, pigment will be produced. If allele e is expressed, no pigment will be produced and the dog will be yellow. Parents heterozygous for both genes reproduce.

**Parent genotype:**  $Bb Ee \times Bb Ee$

**Parent possible gametes:** BE, Be, bE, be x BE, Be, bE, be

	BE	Be	bE	be
BE	BBEE	BBEe	BbEE	BbEe
Be	BBEe	BBee	BbEe	Bbee
bE	BbEe	BbEe	bbEE	bbEe
be	BbEe	Bbee	bbEe	bbee

### Epistasis (cont)

**Offspring phenotypes:**

Black : Brown : Yellow

9:3:4

### Dihybrid crosses

Two genes considered at the same time.

**Key example: Mendel's peas**

Peas can be two different colours, yellow or green. They can also be either round or wrinkled. Roundness is a dominant allele (R) and yellow colour is also dominant (Y).

Show the proportions of phenotypes for the offsprings of two heterozygous peas.

**Parent genotype:**  $RrYy \times RrYy$

**Gametes:** RY, Ry, rY, ry x RY, Ry, rY, ry

	RY	Ry	rY	ry
RY	RRYY	RRYy	RrYY	RrYy
Ry	RRYy	RRyy	RrYy	Rryy
rY	RrYy	RrYy	rrYY	rrYy
ry	RrYy	Rryy	rrYy	rryy

**Offspring phenotypes:**

Round and yellow : Wrinkled and yellow : Round and green : Wrinkled and green

9:3:3:1

This ratio will always be present in dihybrid heterozygous crosses.

This proportion is only true IF:

- There is no autosomal linkage.
- There is no crossing over during meiosis.
- There are no mutations.
- There is no sexual selection (e.g. black rabbits only mate with other black rabbits).
- There is no epistasis.

### Autosomal linkage and crossing over

When genes are linked, it means they occur on the same chromosome.

For example, R and Y are on one chromosome and r and y on the other.

### Autosomal linkage and crossing over (cont)

This means it is not possible to get all the gametes predicted in the previous box.

R and y cannot form a gamete because R and Y are always inherited together.

In the example we used, the only possible gametes for heterozygous parents if the genes are linked are:

RY, ry x RY, ry

	RY	ry
RY	RRYY	RrYy
ry	RrYy	rryy

The ratio therefore changes from 9:3:3:1 to 3:1 (in this example.)

### Crossing over (meiosis)

It is, however, possible for observations to show more than just two phenotypes, or unexpected proportions of these phenotypes.

This is because of crossing over in meiosis.

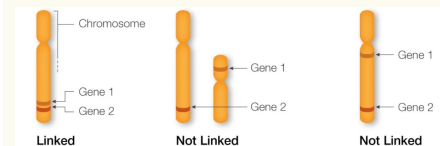
Homologous chromosomes can overlap and swap parts of non-sister chromatids.

This can form new gametes.

In our example, part of the chromosome containing the Y and another chromatid containing the y allele could swap, creating the gametes Ry and rY. These are called **recombinant genes**.

Genes are less likely to split if they are close together, because there is less space for a chiasmata to form between them so they are less likely to be separated and will be inherited together (linked).

### Autosomal linkage



Note the genes occurring on the same chromosome but far apart as marked as not linked because they are likely to be separated by a chiasmata during meiosis.



### Chi-squared

Statistical test used to calculate whether what we expect is different from what we actually observe.

$$(O-E)^2/E$$

Degrees of freedom = number of categories - 1

**Worked example:** Corn can be yellow (Y) or purple (y). Heterozygous cross - Mendelian genetics expected:

	Y	y
Y	YY	Yy
y	Yy	yy

yellow : purple = 3:1

A student observed 21 yellow and 13 purple kernels. Is this significantly different from expectation?

*Null hypothesis: no significant difference between expected and observed colour of corn.*

1. Make expected a proportion	21+13 = 34 3+1 = 4 34/4 = 8.5 3x8.5 = 25.5 1x8.5 = 8.5
2. Observed - Expected	21-25.5 = -4.5 13-8.5 = 4.5
3. Square answers	-4.5 <sup>2</sup> = 20.25 4.5 <sup>2</sup> = 20.25
4. Divide by expected value	20.25/25.5 = 0.794 20.25/8.5 = 2.382
5. Add up values	Chi squared = 3.176
6. Calculate degrees of freedom and compare calculated value to critical value table	2-1 = 1 critical value = 3.841

### Chi-squared (cont)

*Because our calculated value is lower than the critical value, there is more than 5% probability that any differences is due to chance. This means results are not significantly different and we accept the null hypothesis.*

*So the colour observed matched the Mendelian genetics we originally calculated.*

If the observed data does not match the expected data, this means that genes are linked and therefore not subject to independent assortment.

### The Hardy-Weinberg principle

Mathematical model used to determine the frequency of alleles.

$$p^2 + 2pq + q^2 = 1$$

$p^2$  = frequency of homozygous dominant genotype (AA)

$2pq$  = frequency of heterozygous genotype (Aa)

$q^2$  = frequency of homozygous recessive genotype (aa)

$$p + q = 1$$

$p$  = frequency of dominant allele

$q$  = frequency of

**Worked example:** Haemochromatosis is caused by a recessive allele. In one country, every 1 in 400 people have haemochromatosis. What percentage of the population is a carrier for the haemochromatosis recessive gene?

*The data given to us is the proportion of homozygous recessive individuals, so  $q^2$ . Therefore,  $q^2 = 1/400 = 0.0025$ .*

1. Use the information given to work out other values.	$\sqrt{q^2} = q$
<i>If we know <math>q^2</math>, then we can find <math>q</math> then <math>p</math>.</i>	$\sqrt{0.0025} = q = 0.05$
	$p+q = 1$ , therefore $p + 0.05 = 1$
	$1-0.05 = p = 0.95$

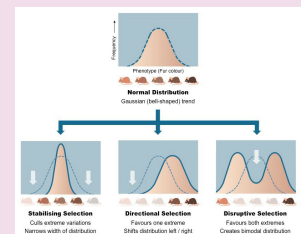
### The Hardy-Weinberg principle (cont)

2. Substitute for answer	Carriers = heterozygous, so $2pq$ $2 \times 0.95 \times 0.05 = 0.095$ $0.095 \times 100 = 9.5\%$
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The Hardy-Weinberg principle does make assumptions which could make it inappropriate to use for some contexts. It relies on:

- A large population.
- Random mating.
- No natural selection.
- No migration / gene flow.
- No mutations.

### Types of selection



### Speciation

Creation of new species. A group gets reproductively isolated from the population (cannot breed together) and develop differences in gene pools.

**Allopatric speciation** Become geographically isolated (e.g. new mountain range). Evolve separately through natural selection, accumulate mutations.

**Sympatric speciation** Occupy same area but behaviours change so don't breed together.

### Genetic drift

Change in allele frequency in population. Impacts are greater in smaller populations.

**Factors that can lead to genetic drift are:**



### Genetic drift (cont)

*Genetic bottle-necks* Event (e.g. natural disaster, overfishing...) kills off most of a population, leaving a few survivors behind (small gene pool).

*Founder effect* A few individuals (small gene pool) first colonise an area, isolated from original population. Can even make rare homozygotic recessive phenotypes more frequent.

### Natural v. Artificial selection

*Natural selection* Evolution. Variety in phenotypes due to genetic and environmental factors.

*Artificial selection* Humans select desirable features and breed those individuals together.

**Ethical concern:** due to selected features, some animals (e.g. pugs) will have medical issues.

### Genetic banks

Gene banks store DNA from plants or animals.

Selective breeding often involves inbreeding, so gene banks can be used to reduce this and increase genetic diversity of a species.



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