TOPIC: PATTERNS OF INHERITANCE

Key Knowledge:

- Pedigree charts and patterns of inheritance, including autosomal and sex-linked inheritance
- Predicted genetic outcomes for a monohybrid cross and a monohybrid test cross
- Predicted genetic outcomes for two genes that are either linked or assort independently

PEDIGREE CHARTS

A pedigree is a chart of the genetic history of a family over several generations. Males are represented as squares (☐), while females are represented as circles (○). **Shaded symbols** denote a condition of interest. A horizontal line between man and woman represents mating, and all offspring are shown as offshoots. Generations are listed using roman numerals (I, II, III, IV, etc.) and individuals are numbered from eldest to youngest (for example: the youngest of three grandchildren would be labelled as III-3).

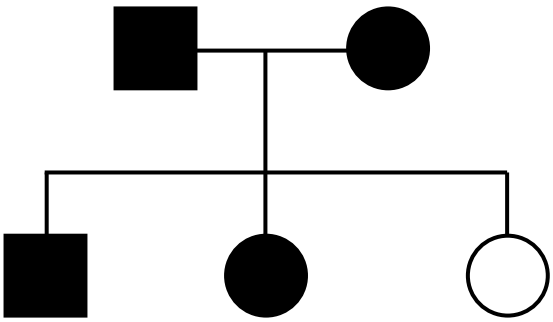
Certain patterns in a pedigree chart can be used to identify the mode of inheritance for a given condition:

- **Autosomal dominant:** If both parents are *affected* and any offspring are not, the condition must be dominant (unaffected offspring are homozygous recessive and both parents must be heterozygous).
- **Autosomal recessive:** If both parents are *unaffected* and any offspring are not, the condition must be recessive (affected offspring are homozygous recessive and both parents must be heterozygous).

It is <u>not</u> possible to conclusively determine sex linkage from pedigree charts, as autosomal traits could potentially generate the same results. However certain trends can be used to suggest sex linkage:

- **X-linked dominant:** Trait tends to be more common in *females*. If a male is affected, all daughters <u>must</u> be affected. An unaffected mother <u>cannot</u> have an affected son (or an affected father).
- **X-linked recessive:** Trait tends to be more common in *males*. If a female is affected, all sons <u>must</u> be affected. An unaffected mother can have affected sons if she is a carrier (heterozygous).
- **Y-linked conditions:** If a male is affected, all sons <u>must</u> be affected. No female can ever be affected. Because males only have one Y chromosome, there is no distinction between dominant and recessive.

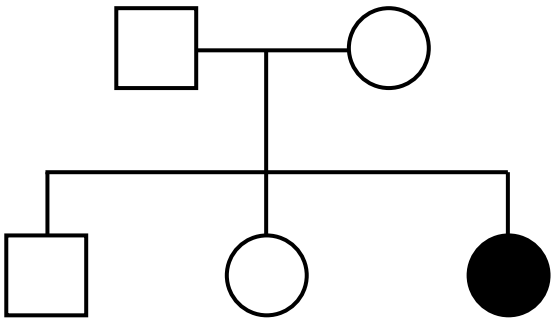
AUTOSOMAL DOMINANT



Both parents are affected and a child is not

Parents must be heterozygous

AUTOSOMAL RECESSIVE



Neither parent is affected but a child is

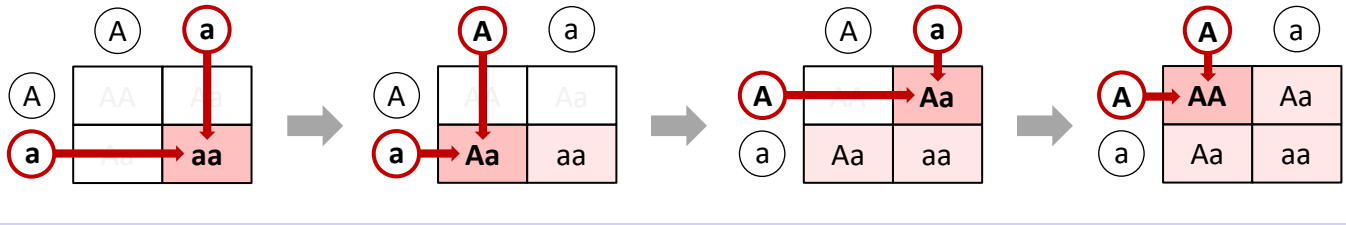
Parents must be heterozygous

MONOHYBRID CROSSES

A monohybrid cross determines the allele combinations for potential offspring with reference to a **single gene** (i.e. monogenic). Monohybrid crosses can be performed according to the following steps:

- Designate potential alleles (dominant = capital letter, recessive = lowercase, codominant = superscript)
- Write down the genotype and phenotype of the prospective parents (this is the P generation)
- Draw a grid with maternal gametes on the top and paternal gametes on the left (this is a **Punnett grid**)
- Complete the Punnett grid to determine the genotypes and phenotypes of offspring (F_1 generation)

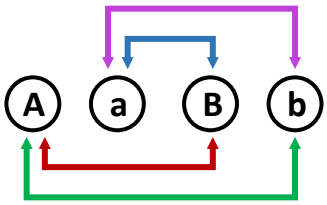
Parental Generation: Aa × Aa



F_1 Generation: 1 AA : 2 Aa : 1 aa (3 dominant : 1 recessive)

DIHYBRID CROSSES

A dihybrid cross determines the allele combinations for potential offspring with reference to **two genes**. Dihybrid crosses can also be performed using a Punnett grid; however, because two genes are involved, there may be up to four different gamete combinations. The easiest way to work out these combinations is by using the **FOIL** method (**F**irst / **O**utside / **I**nside / **L**ast).



When performing a dihybrid cross, always pair the alleles from the same gene together (with the dominant allele written first). Only include the unique gamete combinations for each parent when constructing the Punnett grid, as any repeats will not change the final genotype and phenotype ratios. This will mean that a 4 × 4 grid is only ever needed when crossing two individuals who are heterozygous for both characteristics.

Parental Generation: Aa Bb × Aa Bb

A = Long arms *a = short arms* *B = Black hair* *b = red hair*

	A B	**A b**	**a B**	**a b**
A B	A A B B	A A B b	A a B B	A a B b
A b	A A B b	A A b b	A a B b	A a b b
a B	A a B B	A a B b	a a B B	a a B b
a b	A a B b	A a b b	a a B b	a a b b

F_1 Generation: 9 Long / Black (◯) : 3 Long / Red (◯) : 3 Short / Black (◯) : 1 Short / Red (◯)

TEST CROSSES

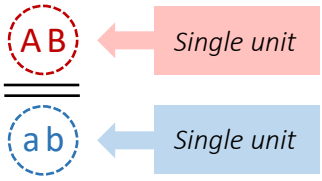
A test cross involves mating an organism with an unknown dominant genotype with an organism that is homozygous recessive. This is because recessive alleles will always be masked by dominant alleles and so the phenotype of any offspring must therefore reflect the genotype of the unknown parent:

- If the parent is homozygous dominant, all offspring must inherit the dominant phenotype
- If the offspring is heterozygous, a proportion of offspring will inherit the recessive phenotype

Test crosses can also be used to determine if two genes are linked or unlinked based on the phenotypic frequencies identified in the offspring (linked and unlinked genes will produce different phenotype ratios).

GENE LINKAGE

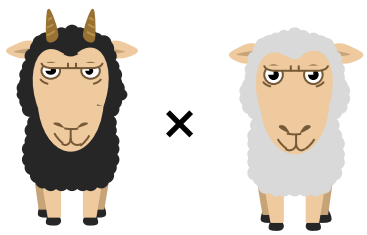
A linkage group is a group of genes whose loci are on a **single chromosome** and hence will not typically be inherited independently of one another. The inheritance pattern will be similar to a monohybrid cross, as the two genes are inherited as a single unit. Linked genes are represented as vertical pairs.



LINKED VERSUS UNLINKED

Because linked genes are on the same chromosome, they do not follow the expected ratios of a dihybrid cross. Offspring should only inherit to phenotypic combinations seen in the parents for the two traits. However, linked genes can be separated via crossing over (in Prophase I of meiosis) to form **recombinant phenotypes** (i.e. the combinations **not** seen in the parents). Because recombination is an uncommon event (crossing over does not happen every time), the recombinant phenotypes will be inherited at a significantly lower frequency than would be expected to occur if the genes were unlinked. Hence, by performing a test cross, the relationship between two genes (linked versus unlinked) can be determined.

TEST CROSS



Heterozygote × Recessive

ALLELES:

B = *black hair* ***H*** = *horns*

b = *white hair* ***h*** = *hornless*

IF TRAITS ARE UNLINKED

	BH	Bh	bH	bh
bh	BbHh	Bbhh	bbHh	bbhh

IF TRAITS ARE LINKED

	Linked		*Recombinants*	
	BH	bh	Bh	bH
bh	BH / bh	bh / bh	Bh / bh	bH / bh

RATIOS

UNLINKED:
1 : 1 : 1 : 1

LINKED:
1 : 1 : 0.1 : 0.1

GENETIC SCREENING

Genetic screening involves testing individuals for either a genetic disease or a predisposition. Genetic markers can be detected by specific **gene probes**, which are single-stranded sequences that are labelled with a radioactive or fluorescent tag. Genetic screening can be useful for family planning (by determining the genetic risks to offspring), but there are issues associated with the confidentiality of the data collected (the genetic information collected for an individual could provide data about all related family members).