Inheritance

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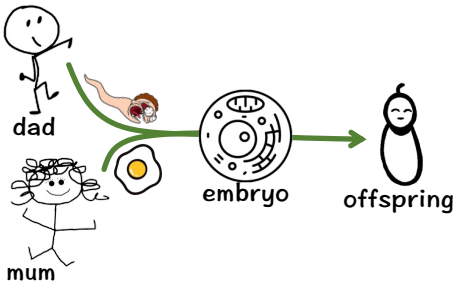
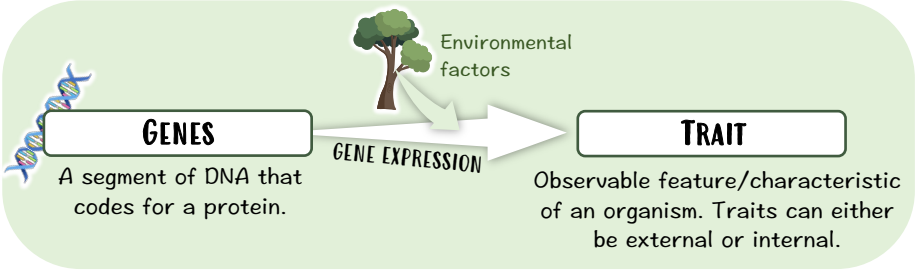
Part 1 The Basics of Inheritance

1. What is inheritance?

Inheritance is the transmission of genetic information from parents to offspring through DNA.

The GENETIC INFORMATION inherited from both parents can be expressed (learned in D2.2) to be displayed as a trait. Traits are influenced not only by genetics but also environmental factors:





Two haploid cells (n), the egg from the mother and sperm from the father, come together in a processes called FERTILIZATION. The resulting cell is diploid (2n), a zygote, which will divide to eventually form the full baby.

Sperm cells are formed through spermatogenesis, and egg cells are formed through oogenesis (see D3.1- HL)

Traits may be influenced exclusively by genetics, exclusively by the environment or by both. Here are some examples:

Exclusively determined by GENETICS

Features:
- ABO blood type

Disorders:
- Huntington's disease
- Hemophilia
- Color blindness

Exclusively determined by ENVIRONMENT

Learned behaviors
(math, learning new song, etc...)

Acquired physical traits:
- Scar
- Tattoos

Determined by both GENETICS & ENVIRONMENT

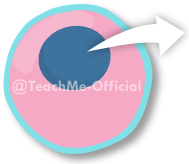
(1) Height
- Max height is genetic. Need nutrition in order to reach your potential.
- May have genetic component, but can also be triggered by cancer causing substances from the environment.

(3) Muscle building
- Need to train, but your muscle building potential depends on your genetics.

Inheritance

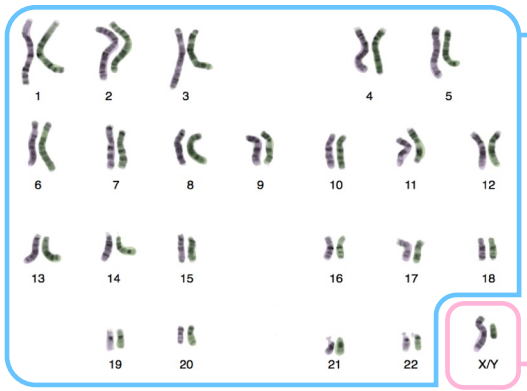
2. Genetic Information

But how is this GENETIC INFORMATION stored in human beings?



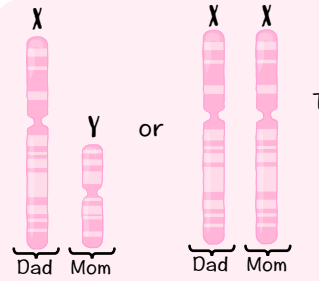
Each cell of your body contains 46 CHROMOSOMES (44 autosomes and 2 sex chromosomes)— with the exception of your gametes (egg or sperm cell) which contain half: 23 CHROMOSOMES.

A KARYOGRAM is an image/diagram that shows the chromosomes of an organism arranged in homologous pairs and in order of decreasing size.



AUTOSOMES
From 1 to 22

They contain a copy from the dad and a copy from the mum.

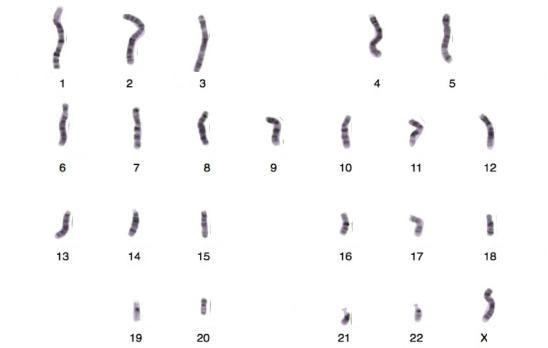


They contain either two X's or an X and a Y.

SEX CHROMOSOMES
23 (either XY or XX)

Diploid: *A cell with two sets of chromosomes.*
Where: *Somatic cells (other than sex cells.)*

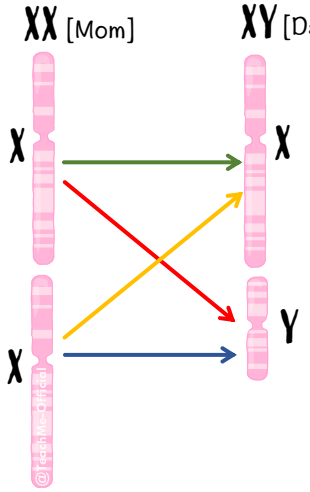
Karyogram of an egg or sperm, with an X chromosome:



Haploid: *A cell with one set of chromosomes.*
Where: *Sperm or egg cells (gametes)*

How is sex/gender determined?

*"Possibility of having a **girl** or **boy** is 50%"*



XX
XX
Female = 50%

XY
XY
Male = 50%

BIG BRAIN TIP!
For males it is conventional to place the X before the Y

Your gender is determined by the sex chromosomes you inherit from each parent. You could inherit either an X or Y from your dad, and an X from your mother. The combination you inherit will determine your gender.

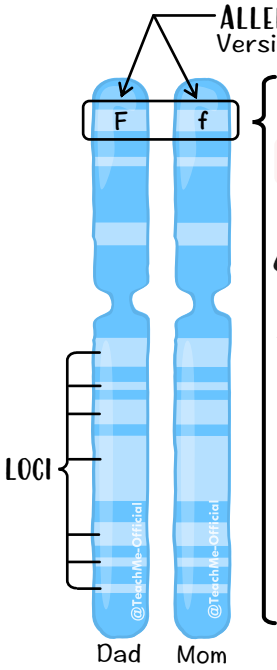
Inheritance

3. Genes and Alleles

Chromosomes each contain various genes. The position of each gene on a chromosome is called a locus. Every gene's locus is fixed. The same gene is located on the same place on the same chromosome on different individuals.

Gene A section of DNA that codes for a protein.

Locus (pl. Loci) Specific position of a gene on a chromosome. A gene locus is fixed.



Alleles can either be **Dominant** or **Recessive**:

Dominant (shown as capital letter)

Simply said, the dominant allele is the stronger allele. It masks the recessive (weaker) allele.

The dominant allele is one that has the same effect on the phenotype whether it is in **homozygous** (both parents gave it) or **heterozygous** state (one parent gave it).

Recessive (shown as lower-case letter)

Simply said, the recessive allele is the weaker allele. It is masked in the presence of a dominant (stronger) allele.

The recessive allele, only has an effect on the phenotype when expressed in **homozygous** state (both parents gave it.)

Big Brain Tip!

Never use letters like:
O, S, X, C, V, K, U, W, X or Z
[Hard to distinguish capital and lower case]

Allele Combinations

Since you inherit one allele from each parent, you will possess **two** alleles per gene.
The combination of those two are either called **homozygous** or **heterozygous**:

Homozygous e.g. FF and ff

Both alleles are the same allele (dominant or recessive)

Heterozygous e.g. Ff

Both alleles are different (dominant and recessive)

Big Brain Tip!

It is conventional to place a capital letter first: Bb instead of bB (but it is not incorrect)

"Some traits are controlled by a **single** gene. Others are controlled by **multiple** genes."

Genotype

The allele combinations (genetic makeup) that you possess.

→ FF
→ Ff
→ ff

Gene expression

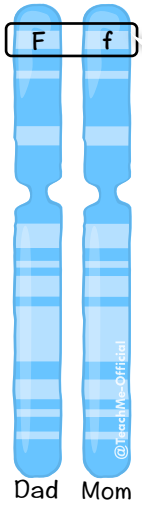
Environmental factors

Phenotype

Observable feature/characteristic of an organism. Traits can either be external or internal.

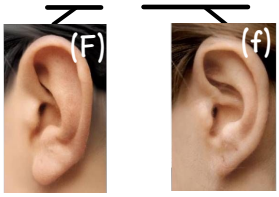
Example

The gene at this loci on chromosome 3 determines ear lobe attachment in humans.



Ear lobe attachment gene

Two different types of ear attachment exist: free or attached ear lobes



Two alleles exist:

(F) – the dominant allele = free ear lobes

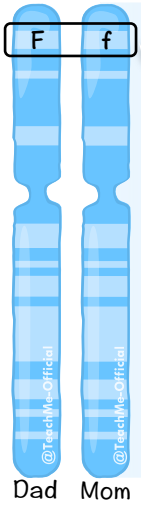
(f) – the recessive allele = attached ear lobes

Inheritance

Part 2 Inheritance Patterns

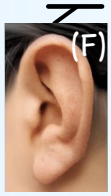
Autosomal dominant

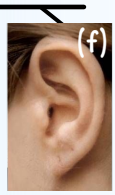
A trait that presents if one or more alleles for it is present.



Example case: Ear lobe attachment gene

Two different phenotypes of ear attachment exist: free or attached ear lobes





Two alleles exist:

(F) – the dominant allele = free ear lobes

(f) – the recessive allele = attached ear lobes

Autosomal dominant diseases:

- Huntington's
- Marfan syndrome
- Retinoblastoma
- Achondroplasia

Since each person inherits two alleles for each gene, there are three possible genotypes:

Genotypes	Phenotypes
FF	Free ear lobes
Ff	Free ear lobes
ff	Attached ear lobes

CARRIER

Since F is dominant over f (is masked), the phenotype is the same whether FF or Ff

Carrier

An individual who has a recessive allele of a gene that leads to a trait/disease if homozygous.

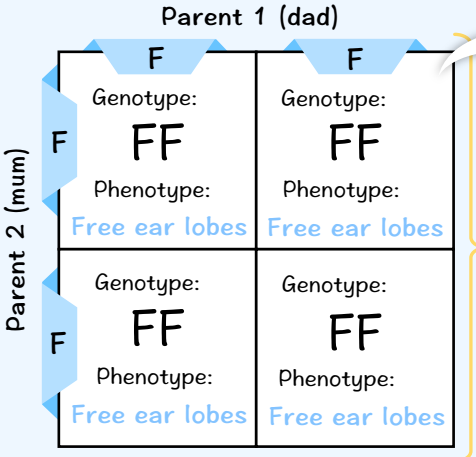
In this example, individuals with genotype Ff are carriers, as they have a recessive allele f. They don't have the phenotype of the recessive allele however, since both are required.

Example 1

What genotype and phenotype outcomes are possible in the offspring if the parents are both HOMOZYGOUS DOMINANT?

1. To answer such questions, we need to first determine the genotype of both parents for the trait:
   Since both are homozygous dominant: F – F Dad: FF F – F Mum: FF

2. We can then construct a PUNNETT GRID (as seen below) in which we cross (breed/mate) the alleles of both parents to reveal all possible allele combinations possible by these two parents.



PUNNETT GRID / SQUARE

OFFSPRING

Genotype:
100% Homozygous dominant

Phenotype:
100% Free ear lobes

BIG BRAIN TIP!

Remember during reproduction, the gametes made only contain half the DNA of each parent: so only ONE allele from each parent can be inherited.

This is an example of a Monohybrid cross (a cross involving a single gene):

MONOHYBRID CROSS
One
Mix (gene)
Mating

Inheritance

Example 2

What genotype and phenotype outcomes are possible in the offspring if the parents are both HETEROZYGOUS?

1. Start by determining the parent's genotypes:

Parent 1 (dad)
Genotype: Ff
Heterozygous
Phenotype: Free lobes

Parent 2 (Mum)
Genotype: Ff
Heterozygous
Phenotype: Free lobes

2. Construct the Punnett grid:

Parent 2 (mum) \ Parent 1 (dad)	F	f
F	Genotype: FF Phenotype: Free ear lobes	Genotype: Ff Phenotype: Free ear lobes
f	Genotype: Ff Phenotype: Free ear lobes	Genotype: ff Phenotype: Attached ear lobes

OFFSPRING

Genotype:
- 25% Homozygous dominant
- 50% Heterozygous
- 25% Homozygous recessive

Phenotype:
- 75% Free ear lobes
- 25% Attached ear lobes

IMPORTANT TO NOTICE!

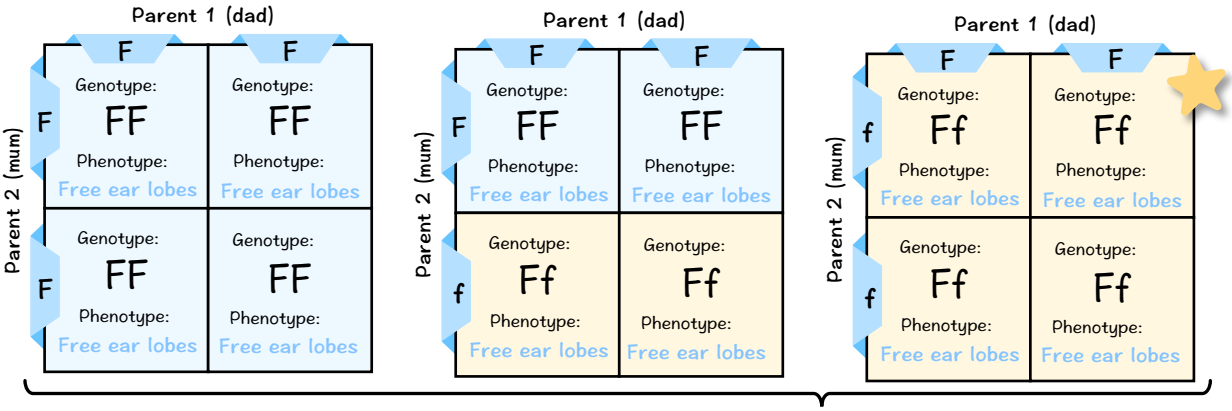
This is a concept of CHANCE: it does not necessarily mean if you have 4 kids, and both you and your partner are heterozygous for the trait that one will have attached earlobes. Instead, each kid has in ¼ CHANCE of having have attached earlobes.

BIG BRAIN TIP!

On the exam, if asked to draw a punnett grid, this is sufficient:

	F	f
F	FF	Ff
f	Ff	ff

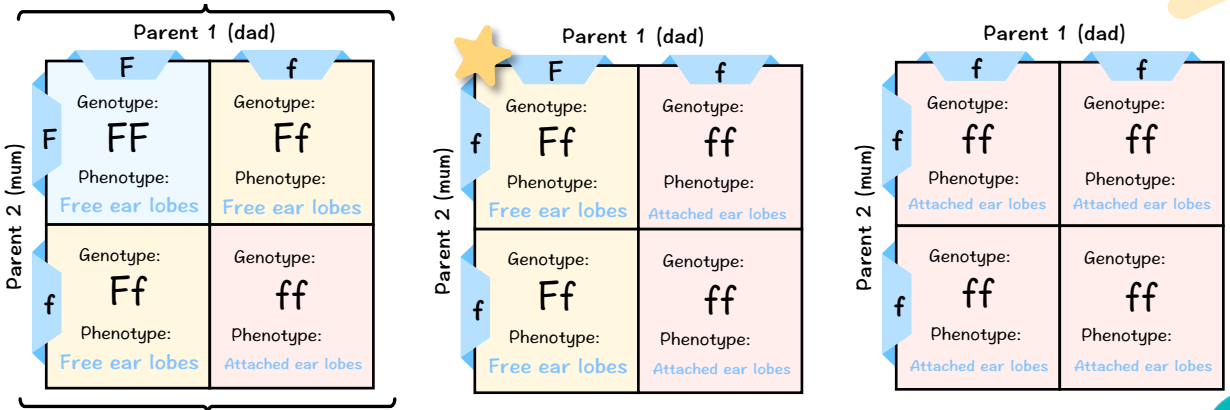
When considering an AUTOSOMAL DOMINANT trait, there are various **KEY TAKAWAYS** from the crosses we can make:



If one of the parents is homozygous dominant, then all the children will display the phenotype, regardless of their genotype.

Despite BOTH parents having the phenotype, they could produce offspring do not have the phenotype.

These two examples are TEST CROSSES.
See more on next page.



Phenotype ratio 3:1 if parents are both heterozygous.

*the colors (blue, yellow and red) are only to help you identify the genotypes at a glance!

FF (Free ear lobes) Ff (Free ear lobes) Ff (Attached ear lobes)

Inheritance

There is one more special type of cross: the **TEST CROSS**.

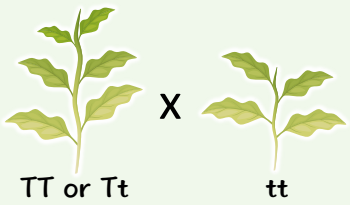
TEST CROSS *A genetic cross used to determine the genotype of an organism displaying a dominant phenotype.*

Example

The height of a pea plant is an autosomal dominant trait: T is the tall allele, and t is the short allele.

You find a tall pea plant, but you do not know what its genotype is: it could either be TT or Tt.
To figure it out, you perform a test cross:

1. You cross this individual with a homozygous recessive (tt) individual.
2. If all the children have dominant phenotype (are tall)
   → then the unknown plants genotype must be homozygous dominant (TT).
3. If half the children have dominant phenotype (half tall, half short)
   → then the unknown plants genotype must be heterozygous (Tt).



EITHER

Parent 1 (unknown) / Parent 2 (short)	T	t
t	Genotype: Tt Phenotype: Tall	Genotype: tt Phenotype: Short
t	Genotype: Tt Phenotype: Tall	Genotype: tt Phenotype: Short

OR

Parent 1 (unknown) / Parent 2 (short)	T	T
t	Genotype: Tt Phenotype: Tall	Genotype: Tt Phenotype: Tall
t	Genotype: Tt Phenotype: Tall	Genotype: Tt Phenotype: Tall

Try for yourself!

Question: A test cross was carried out, and the offspring showed a ratio of 1:1 for the dominant : recessive phenotypes. What are the genotypes of the parents?

Working steps:

STEP 1 *We know this is a test cross, therefor one of the parents must be homozygous recessive. The other must have at least one dominant allele. Start by setting up a Punnett grid to show this information:*

Parent 1 / Parent 2	A	?
a	Genotype: Aa	Genotype: ?a
a	Genotype: Aa	Genotype: ?a

STEP 2 *We also know that half the offspring presented a dominant phenotype, while the other half presented the recessive phenotype. Fill this in the Punnett grid.*

Parent 1 / Parent 2	A	?
a	Genotype: Aa Phenotype: Dominant	Genotype: ?a Phenotype: Recessive
a	Genotype: Aa Phenotype: Dominant	Genotype: ?a Phenotype: Recessive

STEP 3 *Finish filling in the Punnett grid with the allele which would give the correct phenotypes to the offspring. In this case, it must be a recessive allele.*

Parent 1 / Parent 2	A	a
a	Genotype: Aa Phenotype: Dominant	Genotype: aa Phenotype: Recessive
a	Genotype: Aa Phenotype: Dominant	Genotype: aa Phenotype: Recessive

Final answer:

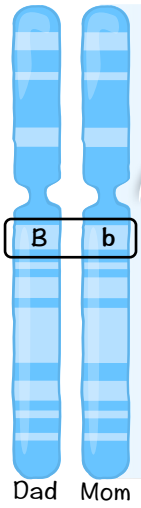
Therefore, the parents' alleles must be:
Homozygous recessive (aa) and heterozygous (Aa).

*NOTE: you can use any letter to represent the alleles when not stated in the question.

Inheritance

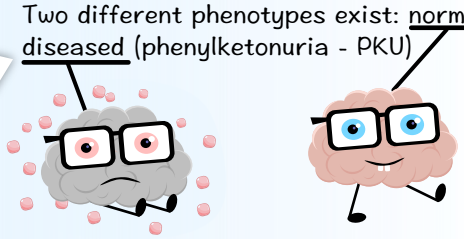
Autosomal Recessive

A trait that only presents when BOTH alleles for it is present.



Example case: Phenylketonuria (PKU)

Two different phenotypes exist: normal or diseased (phenylketonuria - PKU)



Two alleles exist:

(B) – the dominant allele = normal

(b) – the recessive allele = abnormal (PKU)

Since each person inherits two alleles for each gene, there are three possible genotypes:

Genotypes	Phenotypes
BB	Normal
Bb	Normal
bb	Phenylketonuria

CARRIER

Since B is dominant over b (is masked), the phenotype is the same whether BB or Bb

CARRIER

An individual who has a recessive allele of a gene that leads to a trait/disease if homozygous.

In this example, individuals with genotype Bb are carriers, as they have the recessive allele b.

Autosomal recessive diseases:

- Albinism
- Cystic fibrosis
- Sickle cells disease
- Tay Sachs disease
- Thalassemia
- Phenylketonuria (PKU)

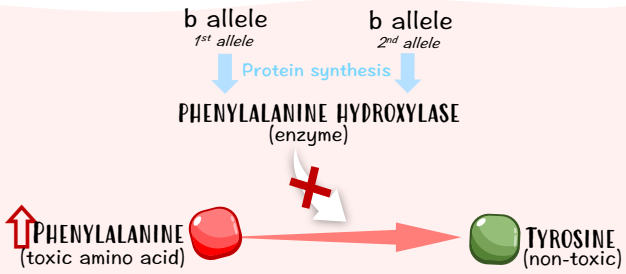
MECHANISM OF DISEASE

Phenylketonuria (PKU)

You have an amino acid in your body called phenylalanine. This amino acid is toxic however. Therefore an enzyme (PHENYLALANINE HYDROXYLASE) converts it to tyrosine (which is non-toxic).

PAH GENE codes for phenylalanine hydroxylase. There are two alleles of this PAH gene: B or b. When transcribed and translated, this gene produces the protein:

*The B allele gives rise to a **normal** protein (enzyme).*
*The b allele giver rise to **defective** protein (enzyme).*



- **Genotype BB** will produce only normal enzymes.
- **Genotype Bb** will produce some normal and some defective proteins: but the number of normal enzymes is enough to prevent PKU.
- **Genotype bb** will only produce defective enzymes. You are therefore **phenylalanine hydroxylase deficient** and have the disease (PKU).

Consequences:

Impair brain development

Treatment:

Omit foods high in phenylalanine. examples; eggs, chicken, nuts. Need dietary supplements.

Example 1

Parent 1 (dad)

Parent 2 (mum)	B	b
B	Genotype: BB Phenotype: Normal	Genotype: Bb Phenotype: Normal
b	Genotype: Bb Phenotype: Normal	Genotype: bb Phenotype: PKU

Notice that neither parent has the disease. But they are both carriers.

OFFSPRING

Genotype:		Phenotype:	
25%	Homozygous dominant	75%	Normal
50%	Heterozygous	25%	PKU
25%	Homozygous recessive		

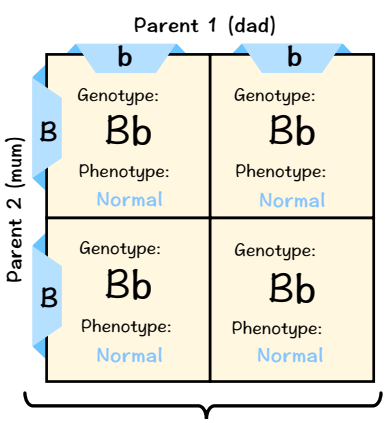
COMMON CONFUSION:

For example, if we consider the height of the pea plants from earlier, we can say that the trait (phenotype) of TALL plants is inherited in an AUTOSOMAL DOMINANT fashion. Whereas the trait of SHORT plants is inherited in an AUTOSOMAL RECESSIVE fashion. It all depends on WHICH trait you are considering!

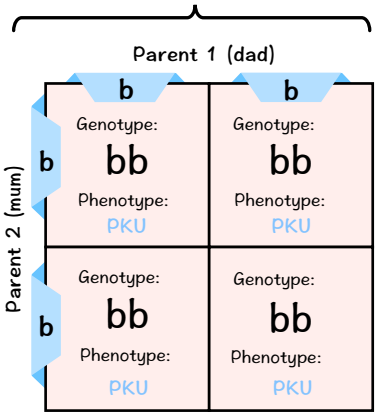
Inheritance

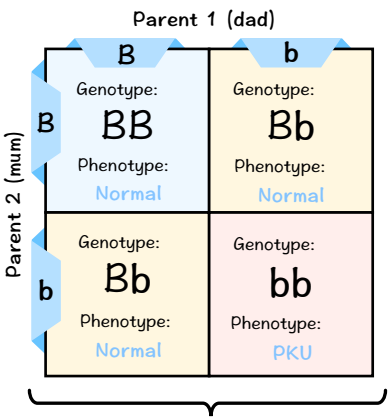
When considering an **AUTOSOMAL RECESSIVE** trait, there are various **KEY TAKAWAYS** from the crosses we can make:

If both parents are affected, then the child will also be affected (not true for autosomal dominant trait).



If one parent is affected (homozygous), it does not mean the child will be affected (not true for autosomal dominant trait).





If a child is affected, but both parents are not affected, then both parents must be carriers.

*the colors (blue, yellow and red) are only to help you identify the genotypes at a glance!

BB (normal) | Bb (normal - carrier) | bb (PKU)

Inheritance

XY

Sex-linked

A trait whose gene is located on one of the sex chromosomes (X or Y).

Sex-linked traits may either be on the X or Y chromosome. X-linked genes may be inherited in a dominant or recessive manner.

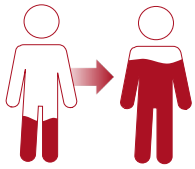
- Sex-Linked
  - X-Linked
    - X linked dominant
    - X linked recessive
  - Y-Linked

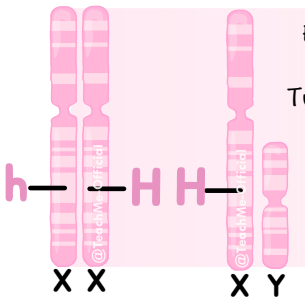
NOTE! For IB, we only focus on X-linked recessive disorders.

Mechanism of Disease
Haemophilia

This is a disorder in which blood does not clot properly. Causing the individual to be at a high risk of bleeding to death from a "minor injury". Bleeding can be either internal (e.g. inside your joints) or external (e.g. knife wound).

Medical treatments give people affected by haemophilia a better quality of life.





Example case: Hemophilia (X-linked recessive)

Two alleles exist:

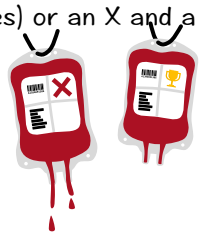
(X^H) – the dominant allele = normal blood clotting

(X^h) – the recessive allele = hemophilia

No allele is present on the Y chromosome

Since each person inherits either two X's (females) or an X and a Y (males), five combinations are possible:

In Females
- X^HX^H Normal blood clotting
- X^HX^h Normal blood clotting, carrier
  Only females can be carriers (not males)
- X^hX^h Hemophiliac



In Males
- X^HY Normal blood clotting
- X^hY Hemophiliac

NOTICE! Notice how the annotation for sex-linked disorders is different compared to autosomal disorders. A prefix (X or Y) is used to indicate that the gene occurs on a sex chromosome. And a superscript is used to show the allele as dominant or recessive.

X-linked recessive diseases:
- Hemophilia
- Color blindness (in humans)
- Duchenne muscular dystrophy (DMD)

When considering an **X-LINKED RECESSIVE** trait, there are various **KEY TAKAWAYS** from the crosses we can make:

For a girl to get a sex-linked (recessive) trait the father must have the disease, and the mother must be at least a carrier.

Parent 2 (mum) \ Parent 1 (dad)	X^h	Y
X^H	Genotype: X^HX^h Phenotype: Carrier	Genotype: X^HY Phenotype: Normal
X^h	Genotype: X^hX^h Phenotype: Color Blind	Genotype: X^hY Phenotype: Color blind

Boys can only get sex linked traits from their mom. Since they always get one Y chromosome from dad, the X chromosome must be from mom.

If the mom has the disease (recessive), the boys will always get the disease.

Parent 2 (mum) \ Parent 1 (dad)	X^H	Y
X^h	Genotype: X^HX^h Phenotype: Carrier	Genotype: X^hY Phenotype: Color Blind
X^h	Genotype: X^HX^h Phenotype: Carrier	Genotype: X^hY Phenotype: Color Blind

Males are affected more than females.

Parent 2 (mum) \ Parent 1 (dad)	X^h	Y
X^H	Genotype: X^HX^h Phenotype: Carrier	Genotype: X^HY Phenotype: Normal
X^H	Genotype: X^HX^h Phenotype: Carrier	Genotype: X^HY Phenotype: Normal

Only females can be carriers, not males.

X^HY or X^HX^H (normal) | X^hY or X^hX^h (Color Blind) | X^HX^h (normal - carrier)

*the colors (blue, yellow and red) are only to help you identify the genotypes at a glance!

Inheritance

Codominance

Two alleles for a particular trait are both expressed equally in the phenotype of an organism. Neither allele is dominant or recessive: both contribute to the organism's appearance, neither is masked.

Example Human Blood Groups

Blood Type

Three possible alleles exist:

- (I^A) – blood group A
- (I^B) – blood group B

} Codominance

- (i) – blood group O

NOTICE!

Prefix "I or i" refers to blood type.
I refers to the dominant alleles (A & B).
i refers to the recessive allele (O).
A and B are codominant alleles.

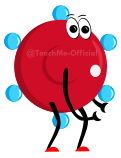
Each allele codes for a different antigen (protein) that will be placed on your red blood cells. I^A codes for A antigens, I^B codes for B antigens, and i codes for no antigens. An antigen is like a label that identifies a cell as either your own body cell or a foreign cell (not your own body cell).

If you are blood group A with genotype I^AI^A then your body will make anti-B antibodies (little weapons) that attack/eliminate red blood cells with B antigens as they are not part of your own body cells. This is why you cannot receive blood from someone with a different blood type to you.

Parent 1 (dad, Blood Group A)

Parent 2 (mum, Blood Group B)	I^A	i
I^B	Genotype: I^AI^B Phenotype: Blood Group AB	Genotype: I^Bi Phenotype: Blood Group B
i	Genotype: I^Ai Phenotype: Blood Group A	Genotype: ii Phenotype: Blood Group O

Four different phenotypes can be created from the alleles (you learned blood groups in C3.2):

	Blood Group A	Blood Group B	Blood Group AB	Blood Group O
Phenotype				
Antigens	A	B	A,B	NONE
Antibody	Anti-B Antibody	Anti-A Antibody	Absence of Anti-A or Anti-B antibody	Anti-A antibodies Anti-B antibodies
Genotype	I^AI^A I^Ai	I^BI^B I^Bi	I^AI^B	ii

BIG BRAIN TIP!

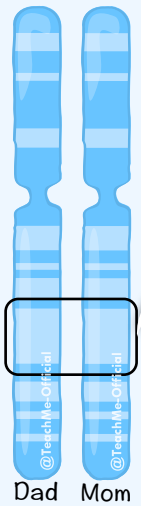
In codominance both alleles are shown in the phenotype. In **incomplete** dominance (see the next page), both alleles are shown **incompletely**. Instead, the phenotype is an intermediate/blend between the two alleles.

Inheritance

Incomplete Dominance

Neither allele is completely dominant over the other, resulting in a blended (intermediate) phenotype in a heterozygous individual. Neither allele is dominant or recessive: both contribute to the organism's appearance as a blend.

Example 1 Hair shape in humans



Two alleles exist:

- (H^S) – straight hair (S)
- (H^C) – curly hair (C)

Hair shape gene

Since each person inherits two alleles for each gene, there are three possible genotypes:

Genotypes	Phenotypes
H^SH^S	Straight hair
H^SH^C	Wavy hair
H^CH^C	Curly hair

Intermediate
A mix of both curly and straight

Parent 1 (dad, staight hair)

Parent 2 (mum, curly hair)	H^S	H^S
H^C	Genotype: H^SH^C Phenotype: Wavy hair	Genotype: H^SH^C Phenotype: Wavy hair
H^C	Genotype: H^SH^C Phenotype: Wavy hair	Genotype: H^SH^C Phenotype: Wavy hair

Notice that neither parent has wavy hair but all their offspring do.

NOTE!

The prefix (here "H") can be any letter you want. Here we picked H for hair. Try not to use X however, as it is easy to confuse with X-linked genes. The superscript (S or C) refers to the specific hair type. We don't use upper-case and lower-case for a single letter as neither allele is dominant or recessive. They are both going to contribute equally to the phenotype as a blend. Hence, we use two different letters

Example 2 The 4 o'clock flower *Mirabilis jalapa* - marvel of Peru

Two alleles exist:

- (C^R) – red flowers (R)
- (C^W) – white flowers (W)

Flower pigmentation

Since each person inherits two alleles for each gene, there are three possible genotypes:

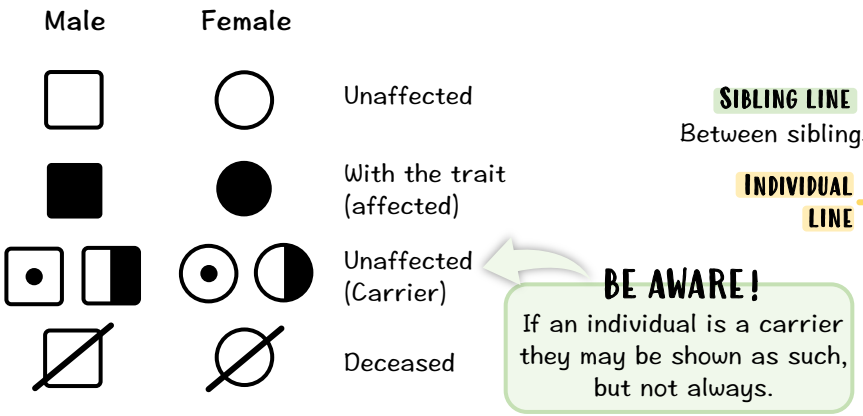
Genotypes	Phenotypes
C^RC^R	Red flowers
C^RC^W	Pink flowers
C^WC^W	White flowers

Intermediate
A mix of both red and white pigmentation

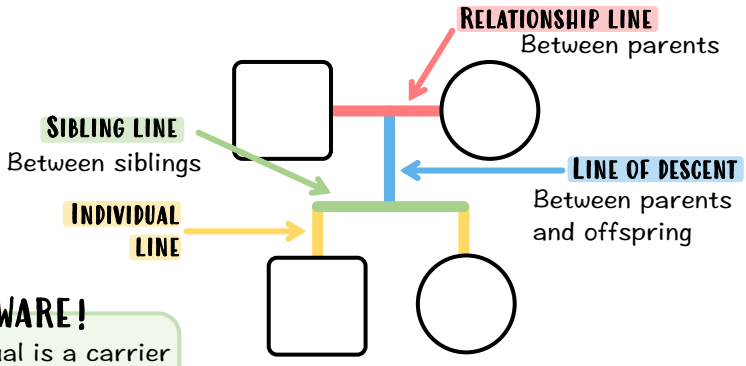
Inheritance

Part 3 Pedigree Charts

Pedigree charts are diagrams that are constructed to show biological relationships. In genetics, they are used to show how a trait can pass from one generation to the next.



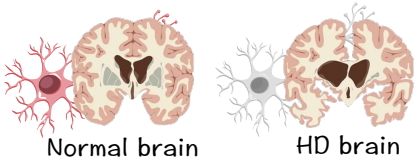
BE AWARE!
If an individual is a carrier they may be shown as such, but not always.



You need to be able to recognize the type of inheritance for a given trait based on the patterns seen on the pedigree chart (specifically autosomal dominant, autosomal recessive and x-linked recessive). Each type of inheritance presents within families with distinct patterns:

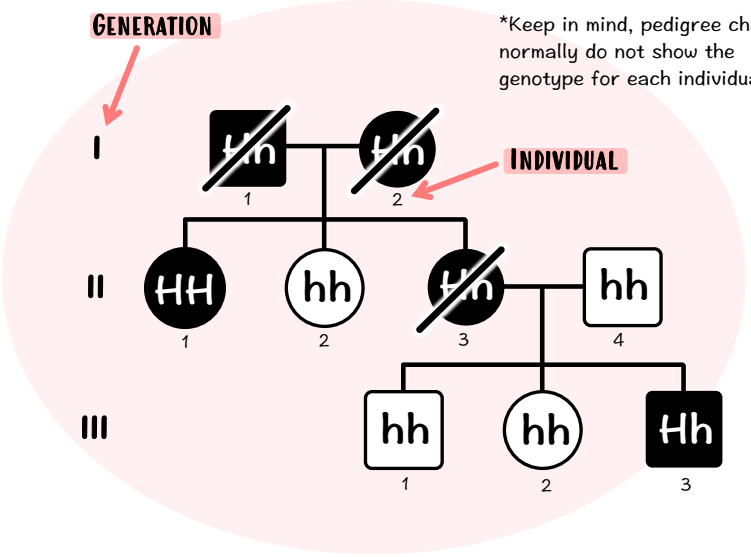
1. Autosomal Dominant

e.g. Huntington's disease



Two alleles exist:
- (H) – Huntington's allele (dominant)
- (h) – Normal allele (recessive)

Genotypes	Phenotypes
HH	Huntington's disease
Hh	Huntington's disease
hh	Normal trait



*Keep in mind, pedigree charts normally do not show the genotype for each individual.

How to recognize Autosomal Dominant

- If an individual as the disease, one of the parents must have the disease.
- If both parents have the condition, the kids won't always have the condition.
  I1 and I2 are both heterozygous, but they have one unaffected kid (II2).
- It can't skip generations – all generations show the trait.

Notice the high rate of death in this pedigree, this is associated with the high mortality of Huntington's disease.

Another way of talking about the different generations:
P – parent generation (I)
F1 – first generation of offspring (II)
F2 – second generation of offspring (III)
...
"F" = filial (refers to offspring generations)

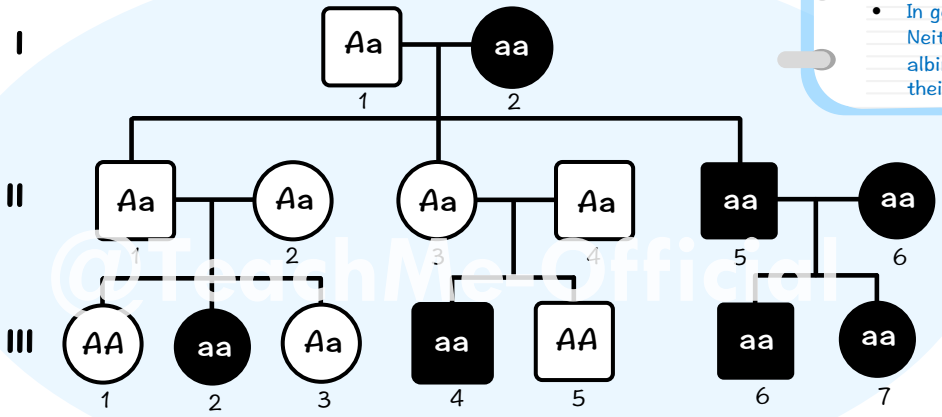
Inheritance

2. Autosomal Recessive

e.g. Albinism

Two alleles exist:
- (A) – Normal allele (dominant)
- (a) – Albinism allele (recessive)

Genotypes	Phenotypes
AA	Unaffected
Aa	Unaffected
aa	Albinism



How to recognize Autosomal Recessive

- If both parents have the trait, then the kids will all have it.
  - II5 and II6 both have it: all their kids get it (III6 and III7)
- Kids can have it even if both parents don't have it. Since parents can be heterozygous.
  - II1 and II2 both are heterozygous, they have one kid affected (III2).

Generations can be skipped.

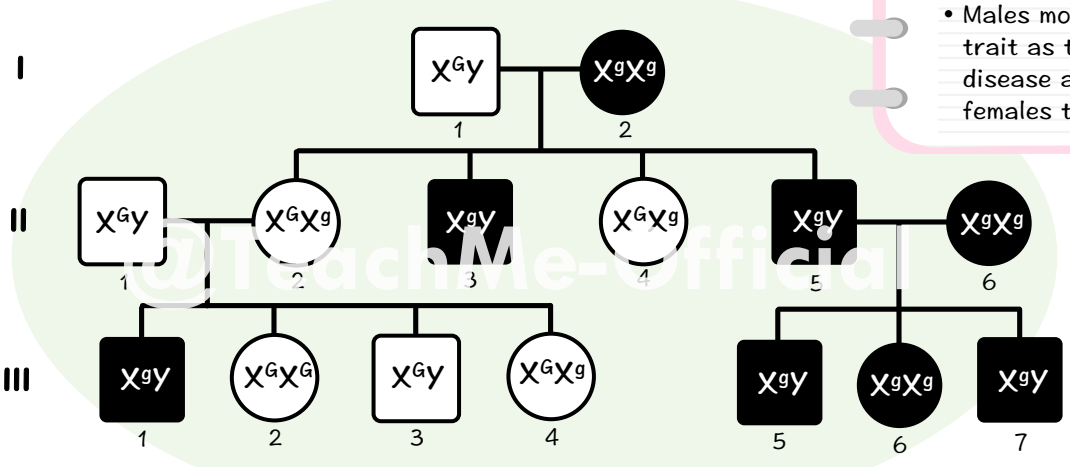
- In generation 1, individual I2 has albinism. Neither individual II1 nor his wife II2 has albinism (generation skipped), but one of their kids have albinism (III2).

3. X-linked Recessive

e.g. Red Green Colorblindness

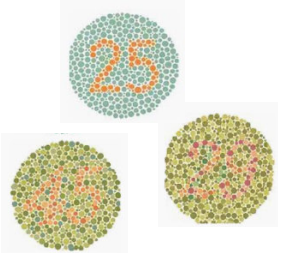
Two alleles exist:
- (G) – Normal allele (dominant)
- (g) – Colorblindness allele (recessive)

Genotypes	Phenotypes
X^GX^G	Unaffected female
X^GX^g	Unaffected female
X^gX^g	Color Blind female
X^GY	Unaffected male
X^gY	Color Blind male



How to recognize X-linked Recessive

- For a girl to get the trait, the dad must have the trait.
  - III6 is the only girl with the trait. her dad also has the trait.
- If the mom has the trait, all sons will get the trait.
  - II6's sons all have the trait.
- Males more commonly have the trait as they only need one disease allele, compared to females that need both.



Inheritance

The following two patterns are more for your own interest, you would not need to identify these kinds of inheritance patterns on a pedigree.

4. Incomplete Dominance

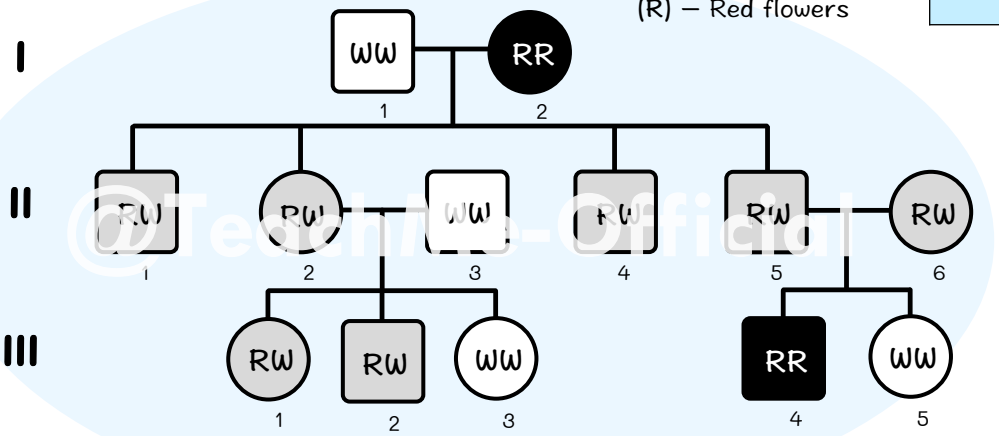
e.g. 4 o'clock flower petal color

Two alleles exist:

(W) – White flowers

(R) – Red flowers

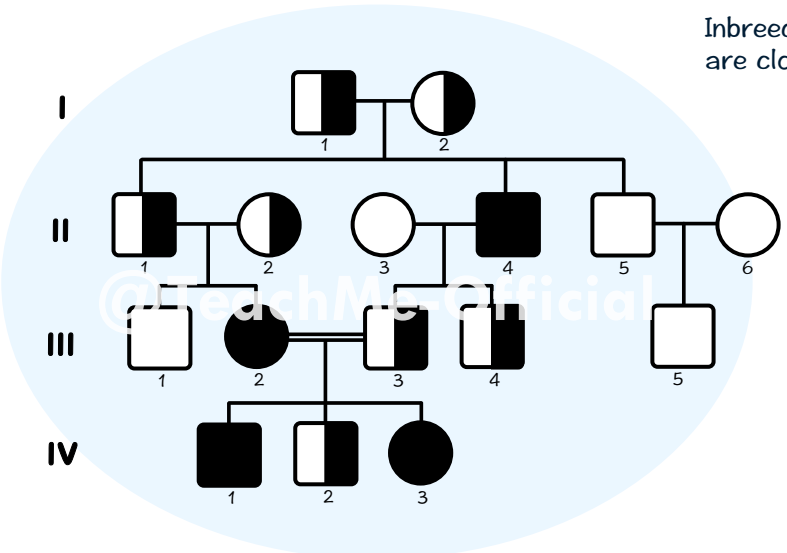
Genotypes	Phenotypes
RR	Red flower
RW	Pink flower
WW	White flower



5. Inbreeding (BONUS)

For your own interest; notice how inbreeding causes an increased frequency of recessive disorders:

Inbreeding is the mating or breeding of individuals that are closely related genetically (cousins, siblings, etc.)



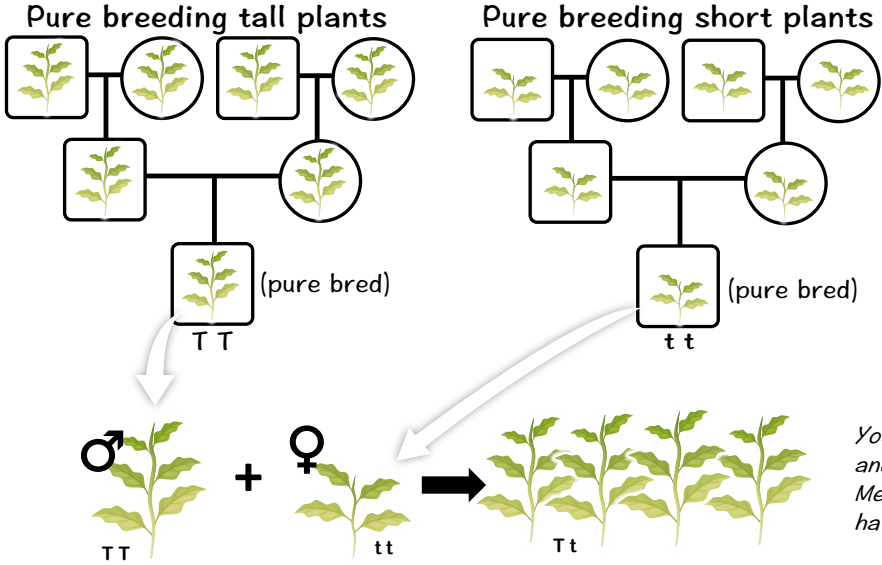
Double lines indicate inbreeding

Inheritance

Part 4 Mendel, Genetics & Data

1. Gregor Mendel Experiments

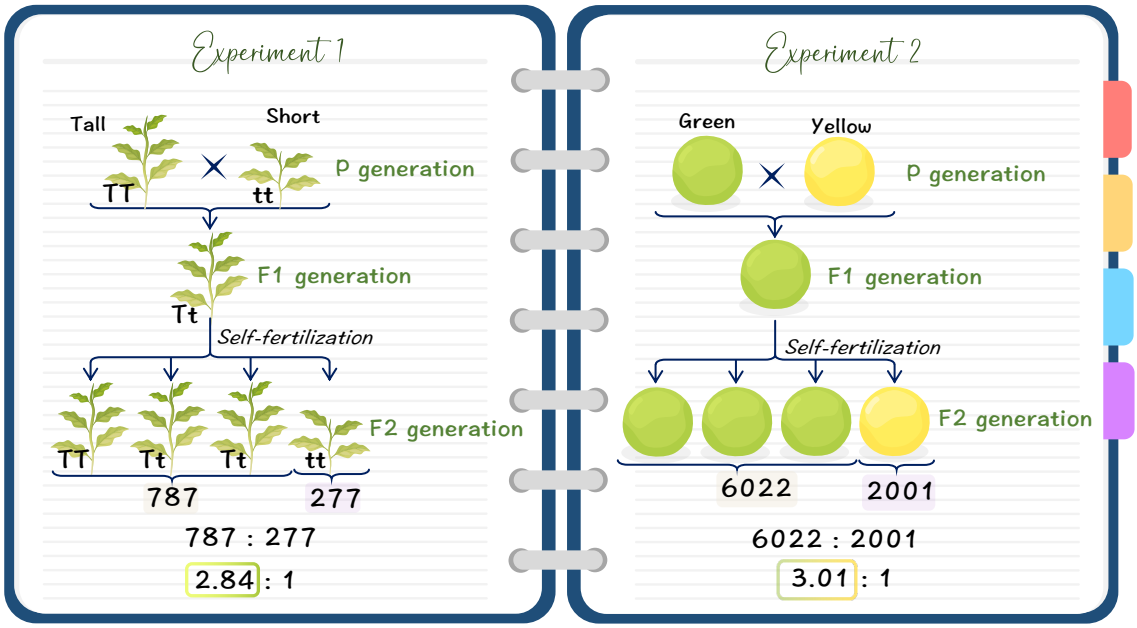
Gregor Mendel, a farmer, cross pollinated **purebred** pea plants in a series of **experiments**.
Transferred pollen (male sperm cells) to the ovules (female egg cells.)



Pure breeding
A cross that always produces offspring of the same phenotype (are homozygous for the trait). Simply put, when an individual has a trait that all its ancestors have it is a pure-bred organism.

You can notice that although the one parent is tall and the other is short, all the offspring are tall. Mendel would then refer to "factors": the tall plant having a stronger factor over the short plant.

"He would then self pollinate the offsprings (F1) and notice interesting pattens (see below) of inheritance in F2"



At the time, the role of DNA and alleles were not discovered yet. Today we know that the tall allele is dominant over the recessive short plant allele. Thus, heterozygous offspring (F1 generation) would have a dominant phenotype. Because of our understanding of inheritance today, we are also able to explain the 3:1 ratio of the F2 generation.

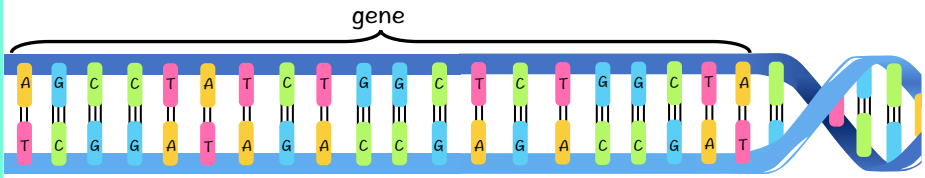
Mendel experimented with various other traits, such as **White** ✕ **Purple** and **Smooth** ✕ **Wrinkled**. Noticing everytime that the ratio between the phenotypes approched 3:1 in the F2 generation.

Due to to his contributions to modern genetics, Mendel is considered the father of genetics and thus the patterns of inheritance mentioned earlier can be called Mendelian patterns of inheritance.

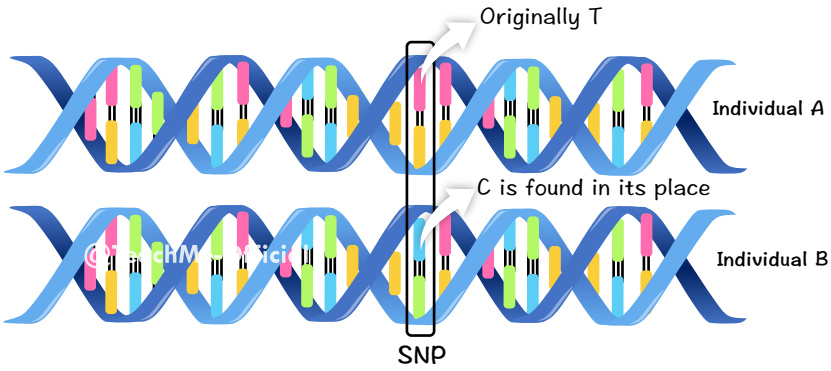
Inheritance

2. Single-Nucleotide Polymorphisms (SNPs)

What do different alleles exist? Often times, different alleles are made by SNPs.



These occur when a nucleotide of a gene, such as T, is not found where it is expected, and instead an individual will have another one, such as C, at that position.



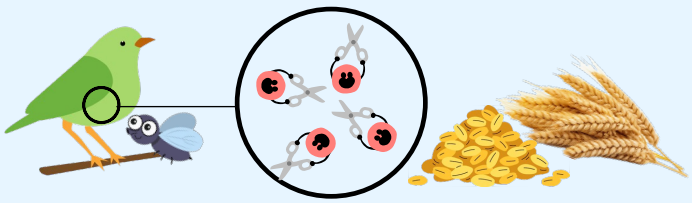
If the difference occurs in a coding region, it may cause an amino acid change, and hence protein functional change. Such a variation in a nucleotide indicates a **different allele**.

3. Phenotypic Plasticity

What: An organism's ability to express its phenotype differently depending on the environment. It does so by varying its patterns of gene expression. A phenotype change without a genotype change.

Advantage: Effective way of adapting. Allow organisms to adapt to subtle changes in environment without need of genetic changes.

Example #1



Environment has ample insects:

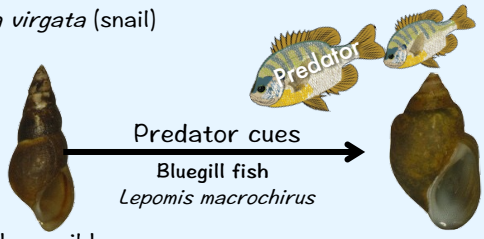
No need to make much maltase to break down grain. Enough nutrition from insects.

Environment has few insects:

Need to increase expression of maltase genes, since not enough nutrition from insects alone.

Example #2

e.g. physa virgata (snail)

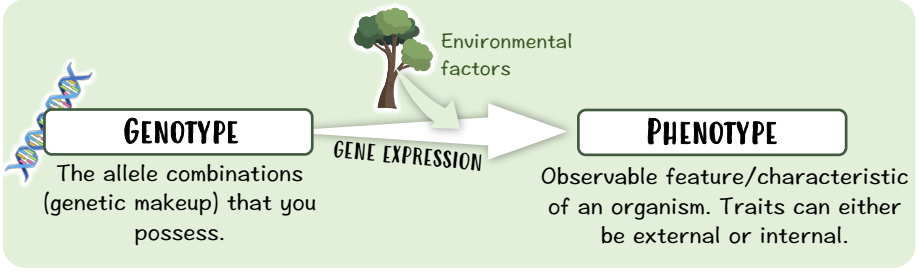


Normally, the snail has an elongated shell

Round/shorter shell

Depending on predator cues, the snail will be expressing its genes slightly differently:

More difficult for its predator to crush. There is NO genotype change (not a mutation): the snail still has the same alleles.



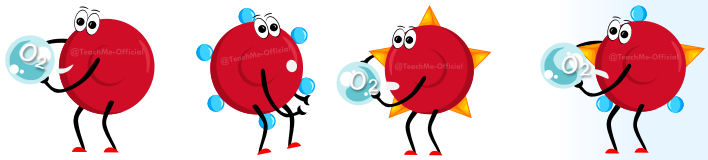
Inheritance

4. Data Analysis

Discontinuous Variation
(discrete)

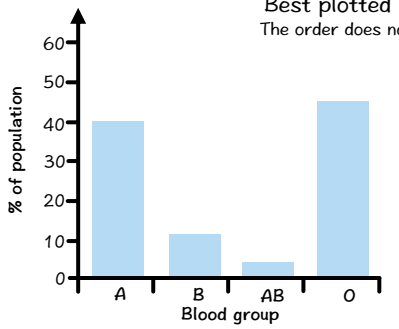
What: When variation can be placed in distinct categories.

Example: Blood Type.



Key Features:

Best plotted as a bar chart.
The order does not matter (no trend)



Can determine a mode, but not a mean.

Usually occurs when a trait is determined by a single gene.

Continuous Variation

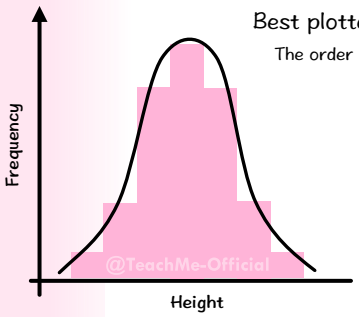
What: When variation has a wide range of possibilities.
No distinct categories.

Example: Height, eye color, skin color

"With multiple genes, the number of possibilities for a single trait increases."

Key Features:

Best plotted as a curve.
The order matters (trend)



Can determine a mean, to show central tendency.

Usually occurs when one trait is determined by two or more genes (polygenetic inheritance).

"Some traits are controlled by a single gene, while others are controlled by multiple genes."

Box-and-Whisker Plots

Key words: Minimum, Maximum, Median, Lower quartile, Upper quartile, IQR, outlier.

Learning with an example (continuous trait) - Height in different countries:

South Africa

139, 140, 155, **160**, 160, 175, **178**, 185, 186, **190**, 195, 200, **205**

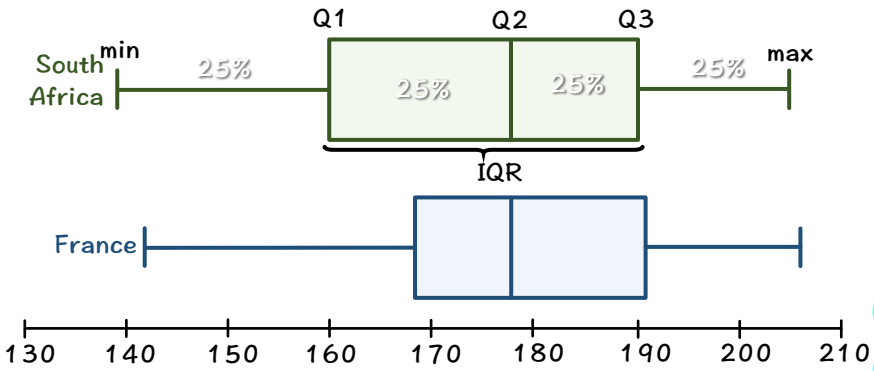
France

142, 149, 157, **168**, 169, 174, **178**, 182, 186, **191**, 197, 200, **206**

Big Brain Tip!
Median ≠ Mean (average)

		Height in South Africa	Height in France
Minimum	Min	139	142
Maximum	Max	205	206
Median	Med (Q2)	178	178
Quartile 1	Q1	160	168
Quartile 3	Q3	190	191
Interquartile Range	IQR (Q3-Q1)	30	23
Outlier	$Q1 - 1.5\times IQR$ $Q3 + 1.5\times IQR$	115* 235	133.5 225.5

*example calculation: 160−1.5×30=115



Key Takeaways!

- Shows visually the SPREAD of the data, NOT an average.
  - IQR shows where the middle 50% of data is.
- Identifies the median (middle number) and outliers.
- Useful to compare data sets (e.g. heights between countries).

You cover more on box-and-whisker plots in your math class!

notes