GENETICS

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IS HERE

genetics made simple

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TOPIC COVARAGE

- Introduction
- Concepts in inheritance:
- Monohybrid crosses:
- Sex determination
- Sex-linked Inheritance
- Blood grouping
- Dihybrid crosses
- Genetic lineages/pedigree
- Mutations
- Genetic Engineering
- Paternity testing
- Genetic Links

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INTRODUCTION

- Father of Genetics. Gregor Mendel, through his work on pea plants, discovered the fundamental laws of inheritance.
- He deduced that genes come in pairs and are inherited as distinct units, one from each parent.

CONCEPTS IN INHERITANCE:

- 1.CHROMATIN:
- is a substance within a chromosome consisting of DNA and protein

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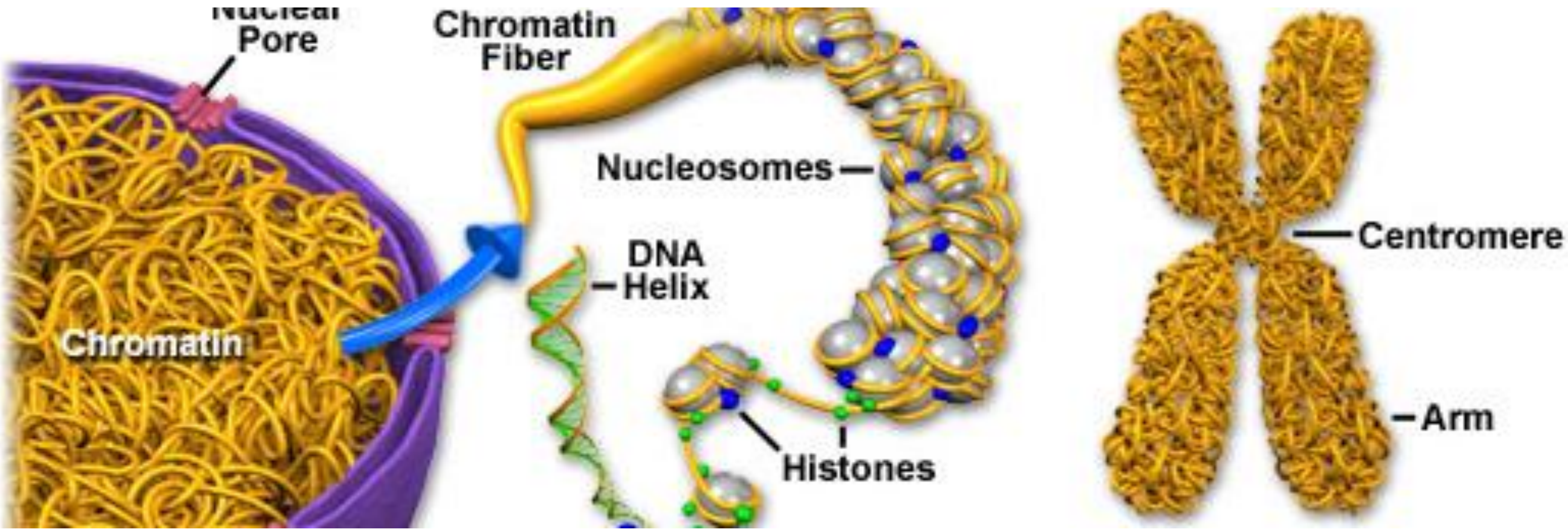
- 2.CHROMOSOMES
- are **threadlike** structures of **nucleic acids** and found in the **nucleus carrying genetic** information.



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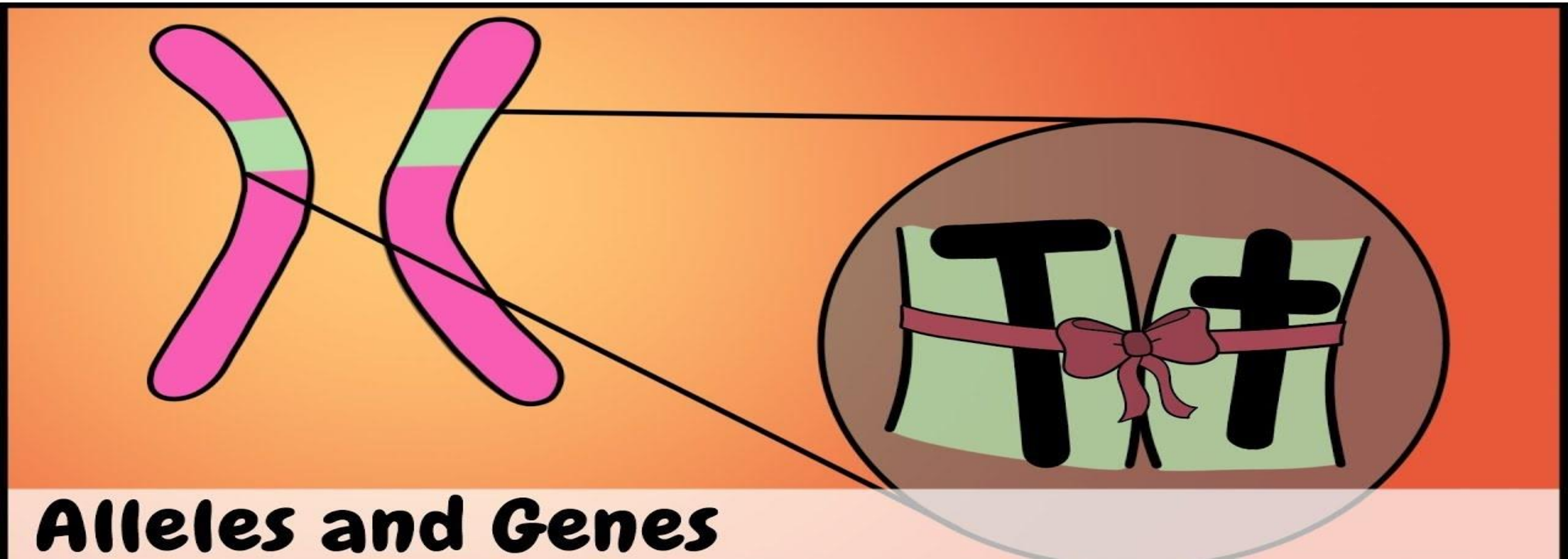
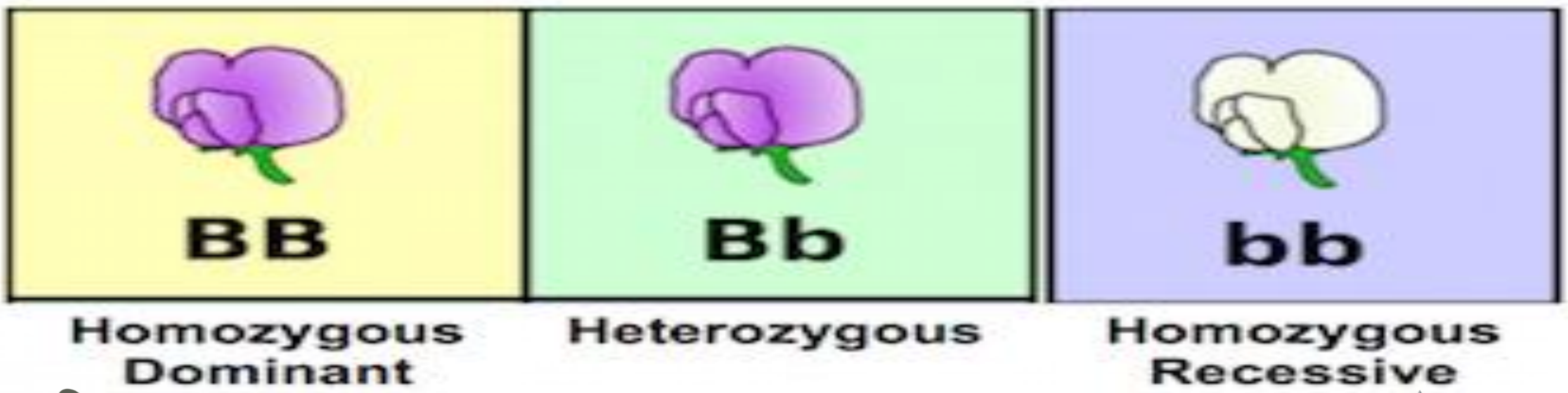
- 3.GENES:
- A small portion of DNA coding for a particular Characteristic/ protein
- 4.ALLELES:
- each of two or more alternative forms of a gene found at the same (locus) place on a chromosome.

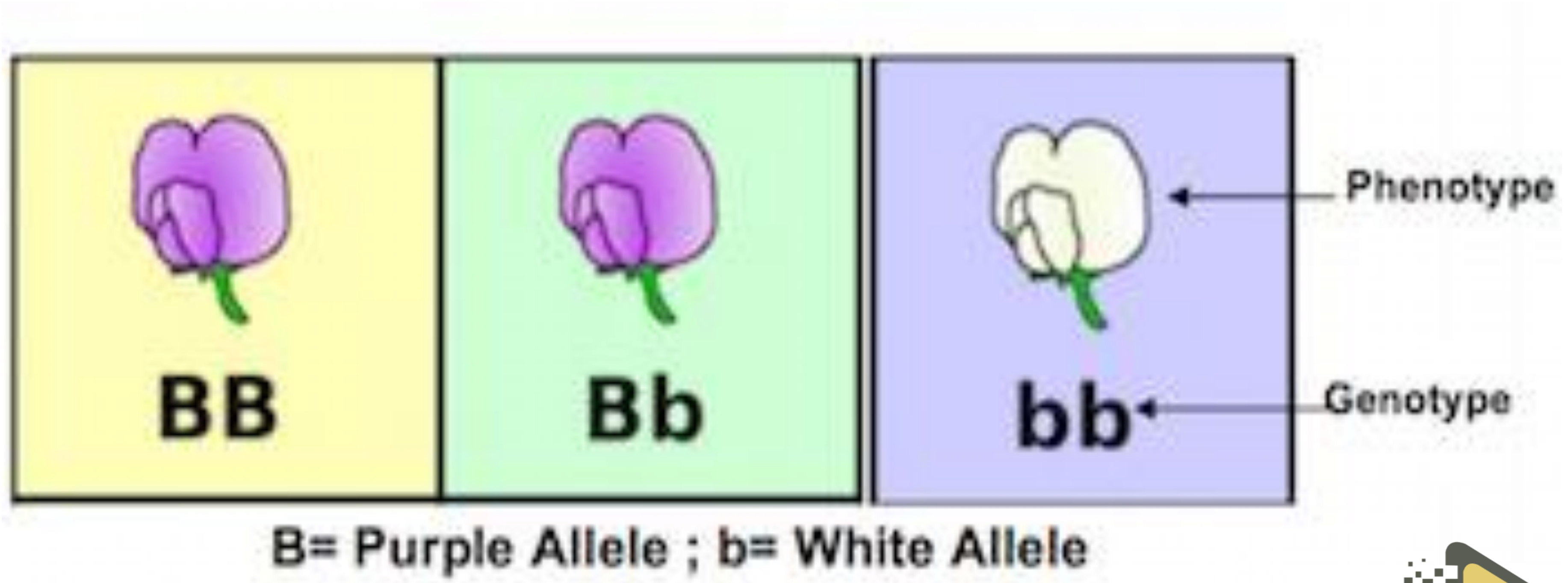


- **5.DOMINANT ALLELES**
- An allele that is expressed in the phenotype when found in the heterozygous (Tt) and
- homozygous (TT) condition.
- **6.RECESSIVE ALLELES**
- An allele that is masked in the phenotype when found in the heterozygous (Tt) condition.
- It is only expressed in the homozygous (tt) condition.



B= Purple Allele ; b= White Allele

- PHENOTYPE
- The physical appearance of an organism determined by the genotype, e.g. tall, short.
- GENOTYPE
- Genetic composition (make- Alleles up) of an organism.



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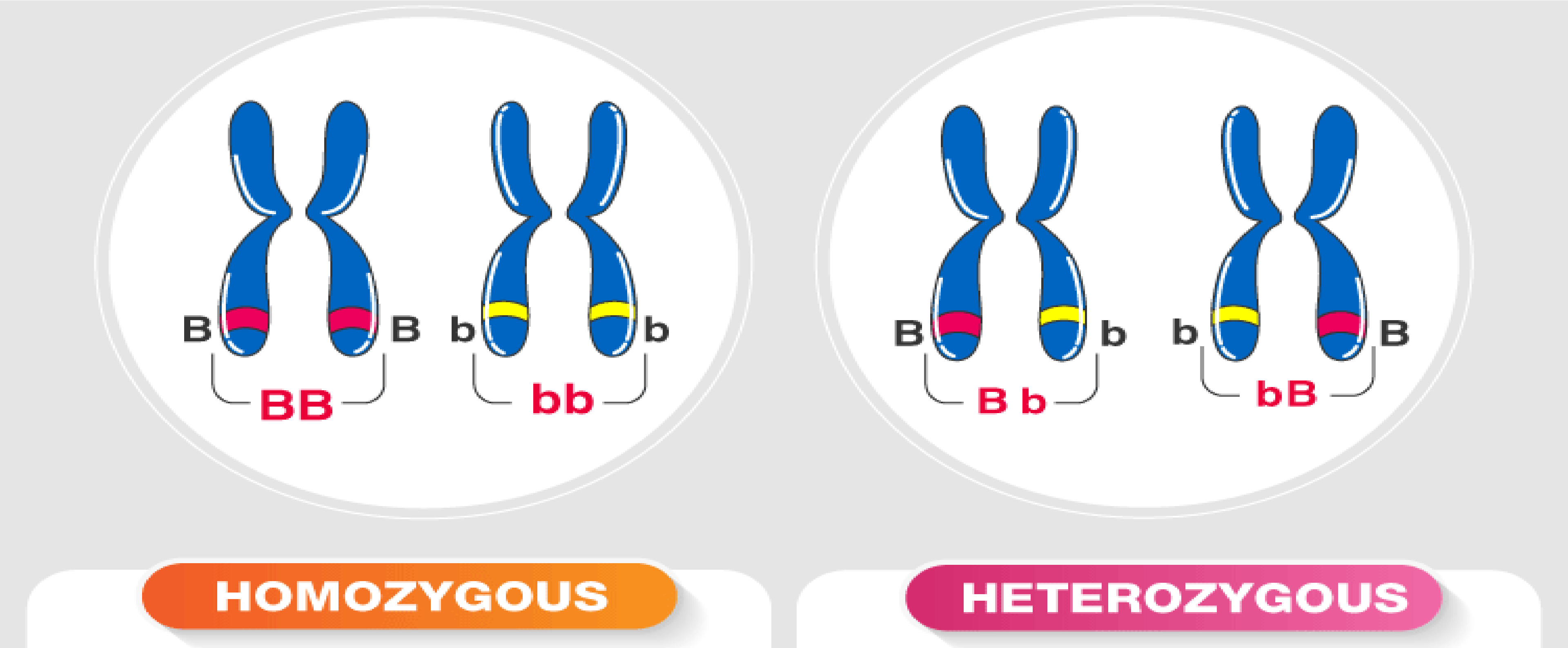
- Homozygous
- Two identical alleles for a particular characteristic, BB or bb.
- Heterozygous
- Two different alleles for a particular characteristic, Bb.



MONOHYBRID CROSSES:

- Is a cross which involve only one characteristic or trait is being shown in the genetic cross.
- **Mendel's Law of Segregation**
- states that a **diploid organism** passes a **randomly** selected allele for a trait to its offspring, such that the offspring receives one allele from each parent

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FORMAT FOR REPRESENTING A GENETICS CROSS

P_1 Phenotype -------- x --------

Genotype -------- x --------

Meiosis

Gametes ---- , --- -----, -------

Fertilisation

F_1

Gametes	-----	----

Genotype:
Ratio:
Phenotype:
Ratio:

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How do we mark a genetic cross

P_1 Phenotype × ✓

Genotype × ✓

Meiosis

✓ ✓ **Gametes** × ✓

Fertilisation

F_1 Genotype ✓

Phenotype ✓

OR

Gametes		

1 mark for correct gametes

1 mark for correct genotypes

[6]

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TYPES OF DOMINANCE

- 1.COMPLETE DOMINANCE
- – genetic cross where the dominant allele masks (blocks) the expression of a recessive allele in the heterozygous condition.
- INCOMPLETE DOMINANCE
- genetic cross where none of the two alleles of a gene are dominant over one another, resulting in an intermediate phenotype in the heterozygous condition
- CO-DOMINANCE
- A genetic cross in which both alleles are expressed equally in the phenotype.

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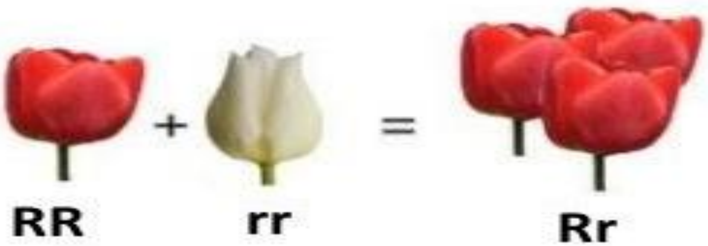
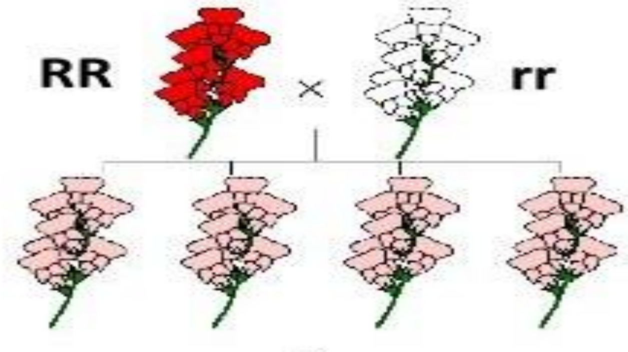
Complete dominance

The dominant allele completely masks the recessive one

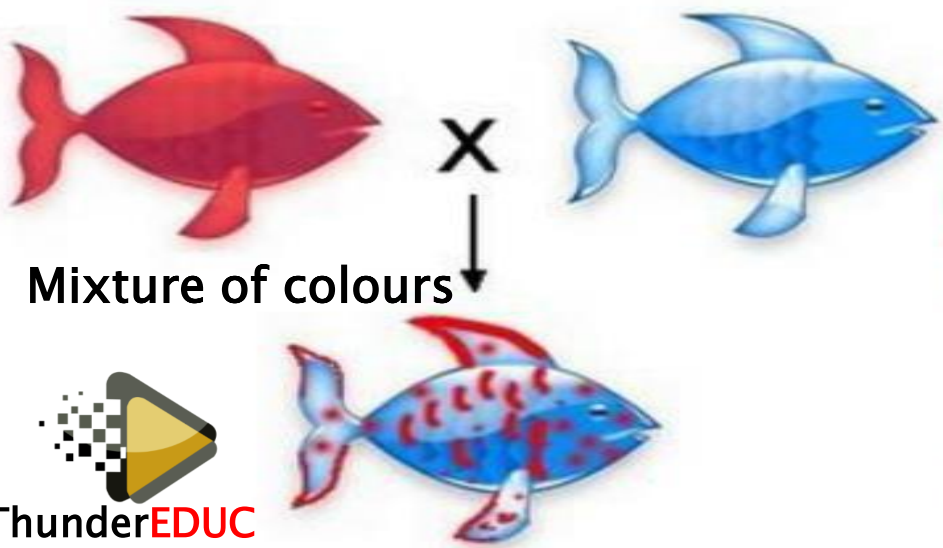


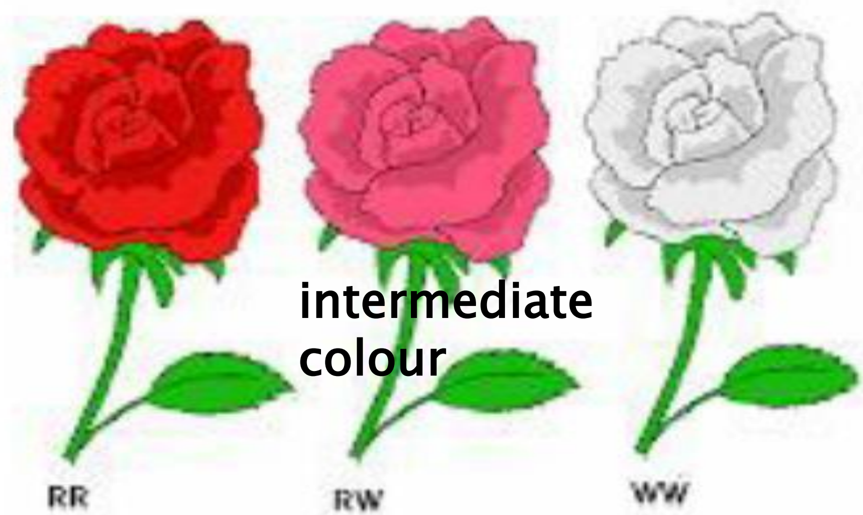
Incomplete dominance

Neither allele is dominant



Codominant vs Incomplete Dominant- What's the difference?





Genetics problems involving each of the three types of dominance

1.COMPLETE DOMINANCE

P_1 phenotyp **Tall x Short**
genotype **TT x tt**

Meiosis

Gametes T , T x t , t

Fertilisation

F_1 Tt Tt Tt Tt

gametes	T	T
t	Tt	Tt
t	Tt	Tt

Genotype: all **Tt**
Phenotype: all **Tall**

(Individuals of F_1 all display the dominant characteristic)

Allele for Tallness (T)is dominant over Allele for shortness (t). A man who is homozygous tall marries a women who sis homozygous short and have children ; using a a genetic cross to determine the possible genotypes and phenotypes of the children.

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2.INCOMPLETE DOMINANCE

P_1 phenotype **Red** x **White**
genotype **RR** x **WW**

Meiosis

Gametes **R , R** x **W , W**

Fertilisation

gametes	R	R
W	RW	RW
W	RW	RW

F_1 **RW RW RW RW**

Genotype: all **RW**
Phenotype: all **Pink**

(Offspring have intermediate forms of traits of parents)

Allele for Red (R) dominant and Allele for white (W) . When homozygous red crosses with homozygous white; using a a genetic cross to determine the possible genotypes and phenotypes of the flowers produced .

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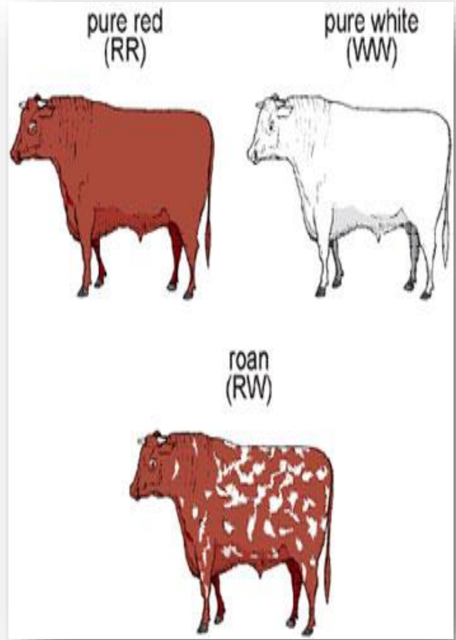
3. CO-DOMINANCE



Cows with white fur (W) were crossed with bulls with red fur (R): as shown in the diagram , use a genetic cross to show how Roan was got.

P_1 phenotype **Red x White**
genotype **RR x WW**

Meiosis

Gametes R , R x W , W

Fertilisation

	R	R
W	RW	RW
W	RW	RW

F_1 **RW RW RW RW**

Genotype: **RW**
Phenotype: **Roan (both red and white)**

(Both alleles are equally dominant and are expressed in the phenotype)

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Proportion and ratio of genotypes and phenotypes

- genotypic ratio describes the number of times a **genotype** would appear in the offspring after a test cross. For example, a test cross between two organisms with same **genotype**, Rr, for a heterozygous dominant trait will result in offspring with **genotypes**: RR, Rr, Rr, and rr. Hence 1: 2:1
- **Phenotypic ratio** pertains to the relative number of offspring manifesting a particular trait or combination of traits. Eg 3 red, 1 white hence 3:1

NOTE: let students practice different examples with crosses

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Representation of a genetic cross to show the inheritance of sex

P_1 phenotype **man** **x** **woman**
genotype **XY** **x** **XX**

Meiosis

Gametes X , Y x X , X

Fertilisation

gametes	X	X
X	XX	XY
Y	XY	XY

F_1 XX XX XY XY

Genotype: 2 **XX , 2 XY**
Phenotype: 2FEMALE , 2MALE

A man marries a woman, and have children. using a a genetic cross to determine the possible genotypes and phenotypes of the children produced .

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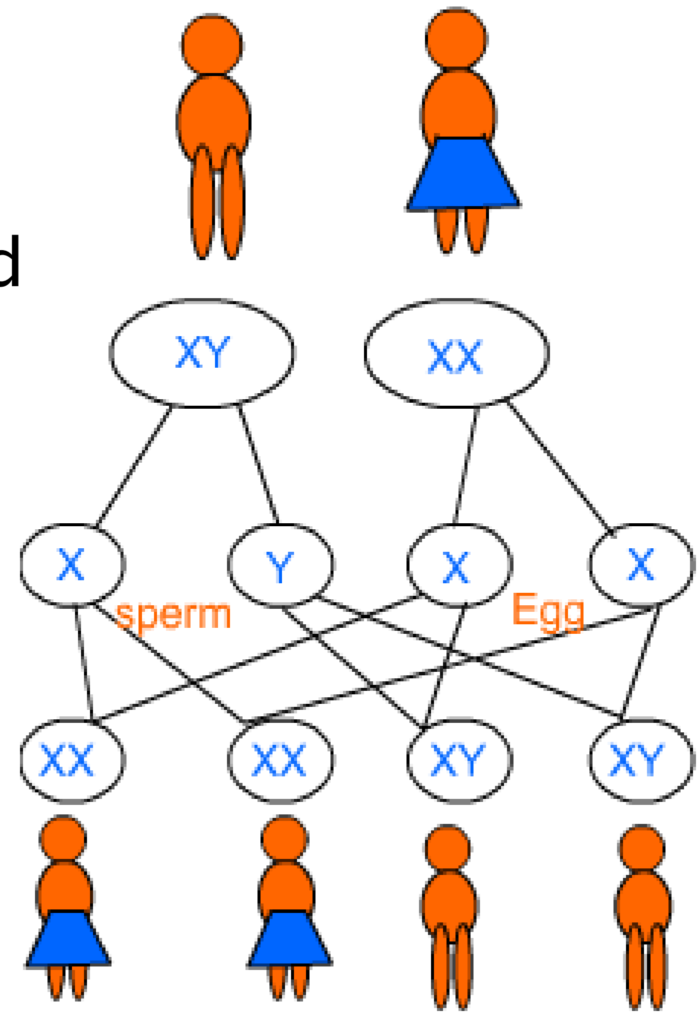
SEX DETERMINATION

- 22 pairs of chromosomes in humans are autosomes and 1 pair of chromosomes are sex chromosomes/gonosomes
- Males have XY chromosomes and females have XX chromosomes
- If the sperm carrying Y chromosome fertilizes the ovum with X, the individual is going to have XY and is a male
- If the sperm carying X chrosome fertilizes the ovum with X, the individual is going to have XX and is a female. There is 50% chance of having a boy or girl



SEX-LINKED INHERITANCE

- Sex linked genes/alleles are genes that are carried on **sex** chromosomes
- Sex-linked characteristics are characteristics (traits) that are carried on the sex chromosomes.

SEX-LINKED DISORDERS

- haemophilia, in ability for blood to clot
- colour blindness,
- night blindness

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HAEMOPHILIA

Haemophilia is caused by a recessive allele on X chromosome, h, a man who is haemophilic marries a lady who is normal but a carrier and produced children. Using genetic cross show the phenotypes and genotypes of children.

P_1 phenotype **Male haemophiliac** **x** **Female normal**

genotype **X^hY** **x** **X^HX^h**

Meiosis

Gametes **X^h** **Y** **X^H** **X^h**

Fertilisation

Gametes	X^h	Y
X^H	X^HX^h	X^HY
X^h	X^hX^h	X^hY

F_1 Genotype: **X^HX^h,** **X^hX^h,** **X^HY,** **X^hY**

Phenotype: Normal Haemophilic Normal
Haemophilic

female female male male

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PRACTICE QUESTIONs (teacher must guide)

- 1. Colour blindness is another example of genetic disorder caused by a recessive allele **c** on the **X** chromosome. A man who is colour blind, marries homozygous nornal women. Do a genetic cross to show the possible genotypes of their children.

  Use B for normal and b for colour blindness. (6)
- 2. Why are males affected by colour blindness than female? (3)

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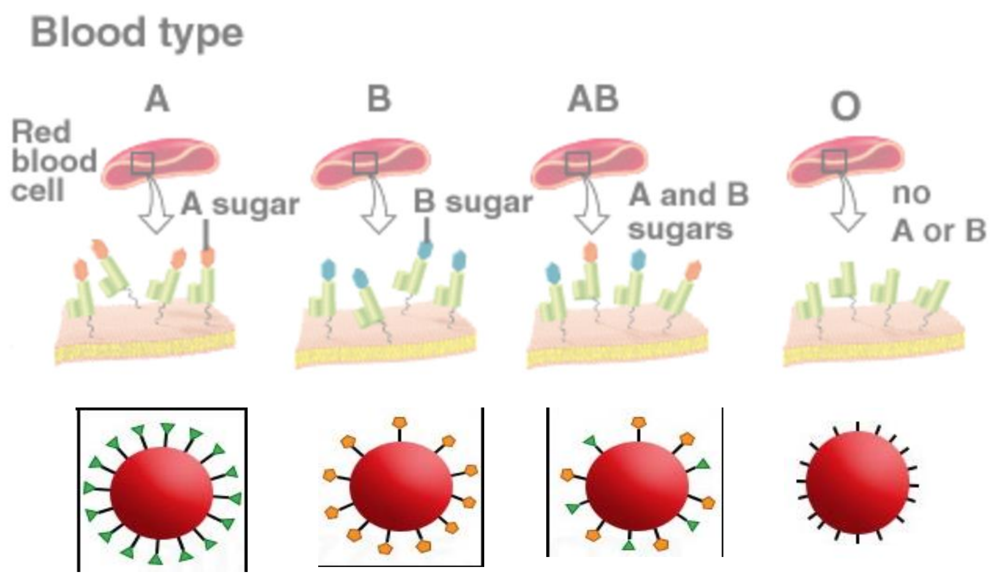
BLOOD GROUPING

- Different blood groups are a result of **multiple alleles eg blood group**
- Three or more alternative forms of a gene (**alleles**) that can occupy the same locus.
- However, only two of the **alleles** can be **present** in a single **organism.**

Phenotype	Genotype
Type A	I^AI^A, I^Ai
Type B	I^BI^B, I^Bi
Type AB	I^AI^B
Type O	ii



- The alleles I^A, I^B and **i** in different combinations, result in 4 blood groups
- **three different alleles I^A, I^B and i**
- **Four blood types A, B , AB, O**

Blood group (Phenotype)	Alleles (Genotype)
A	I^AI^A or I^Ai
B	I^BI^B or I^Bi
AB	I^AI^B
O	ii

GENETICS PROBLEMS FOR BLOOD GROUP

P_1 Phenotype AB x O

Genotype I^AI^B x ii

Meiosis

Gametes I^B I^B i i

Fertilisation

Gametes	I^A	I^B
i	I^A i	I^B i
i	I^A i	I^B i

F_1 Genotype: I^Ai **and** I^Bi
Ratio: 1 : 1
Phenotype: **Blood group A** **and** **Blood group B**
Ratio: 1 : 1

susan with blood group AB marries a John with blood group O. What will be the possible genotypes of their offspring

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PRACTICE QUESTIONs (teacher must guide)

1. Kabelo of blood group A, marries karabo of blood group B. And produced children of blood group O, Use a genetic cross to show the phenotypic ratio of their offspring
2. How many alleles controlling blood groups
3. How many blood group types do humans have

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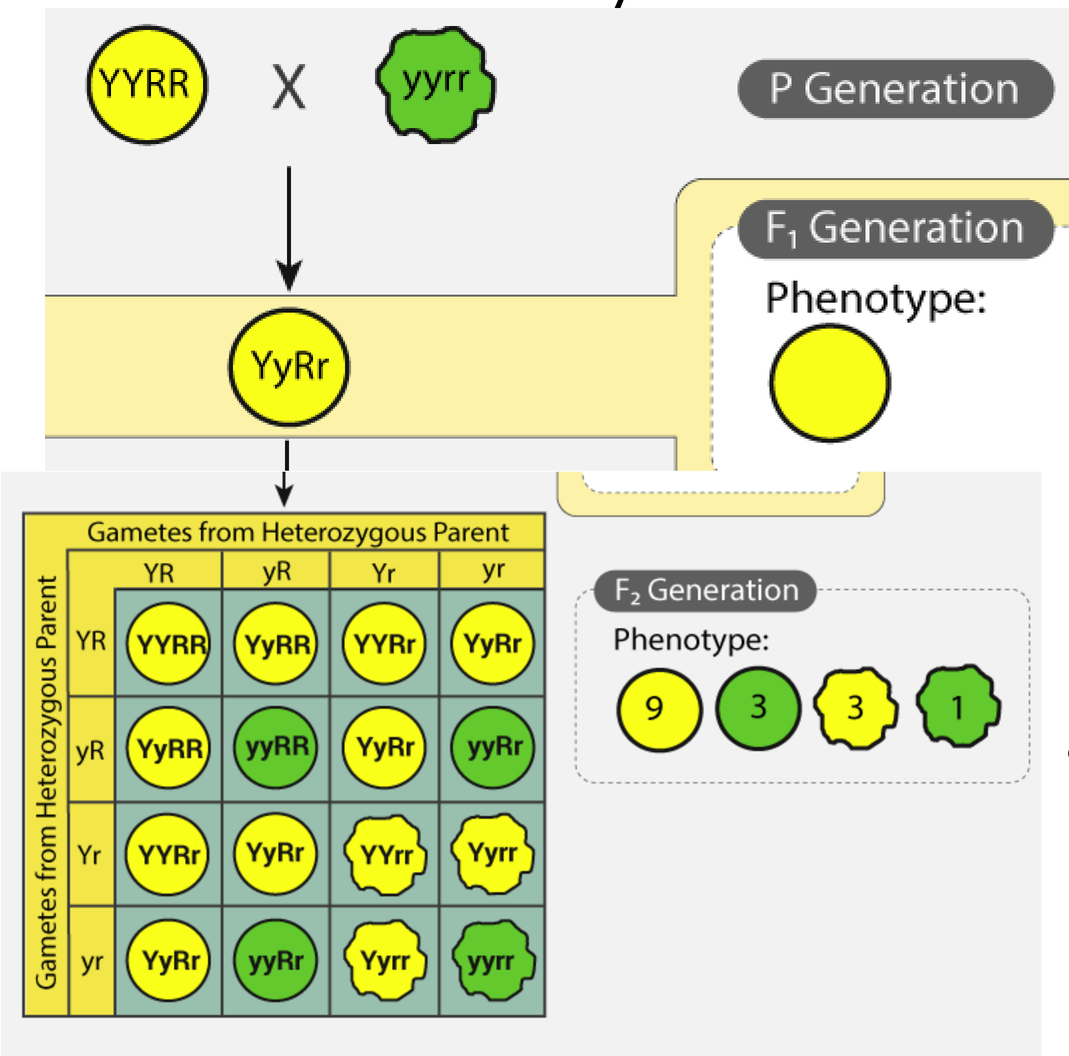
DIHYBRID CROSSES

- A dihybrid cross involves the inheritance of two characteristics.
- **Mendel** explained the results obtained from dihybrid crosses according to his **Principle of Independent**
- It describes how different genes/alleles **independently** separate from one another when reproductive cells develop (meiosis).

Example

- Mendel crossed garden peas, he crossed round and yellow with black and wrinkled. But all offspring in F1 were round and yellow.



- It means that yellow was dominant over black
- Round was dominant over wrinkled

He interbreed the offspring, and he obtained a ratio of 9:3:3:1 As shown

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EXAMPLES OF DIHYBRID GENETIC CROSS

In sheep black fur (B), long fur (L) are dominant over brown (b), short fur (l). If a male sheep crossed with a female sheep both homozygous and produces F1 generation, where the offspring were interbreed, use a genetic cross to show the genotype and phenotypes of the F2 generation with their phenotypic ratios.

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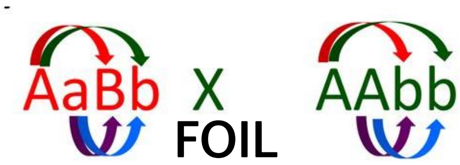

P_2 Phenotype:

black and long fur x black and long fur

Genotype: BbLl x BbLl

Meiosis

Gametes: BL, Bl, bL, bl x BL, Bl, bL, bl

Fertilisation

F_2 genotype:

	BL	**Bl**	**bL**	**bl**
BL	BBLL	BBLl	BbLL	BbLl
Bl	BBLl	BBll	BbLl	Bbll
bL	BbLL	BbLl	bbLL	bbLl
bl	BbLl	Bbll	bbLl	bbll

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Phenotype ratio

Phenotype	9 black long
	3 black short fur
	3 brown long fur
	1 brown short fur

Black & long : black & short : brown & long : brown short

9 : 3 : 3 : 1

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GENETIC LINEAGES/PEDIGREE

- A genetic lineage/pedigree traces the the inheritance of characteristics over many generations

- **NOTE**

- Determine whether the **trait/ character** is dominant or recessive.
- Determine if the chart shows an autosomal or sex-linked trait.
- Sex linked character is always stated in the question

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INTERPRETATION OF PEDIGREE DIAGRAMS

- 1 non sex linked cross (autosomal trait)
- Eg albinism
- Albinism is an inherited condition caused by a recessive gene mutation.
- This mutation results in the absence of the protein melanin in the skin.

- **QUESTION**
- The pedigree diagram shows inheritance of eye colour in humans over three generations of a family. Brown eye colour (A) is dominant over blue eye colour (a). Study the diagram and then answer the questions that follow.

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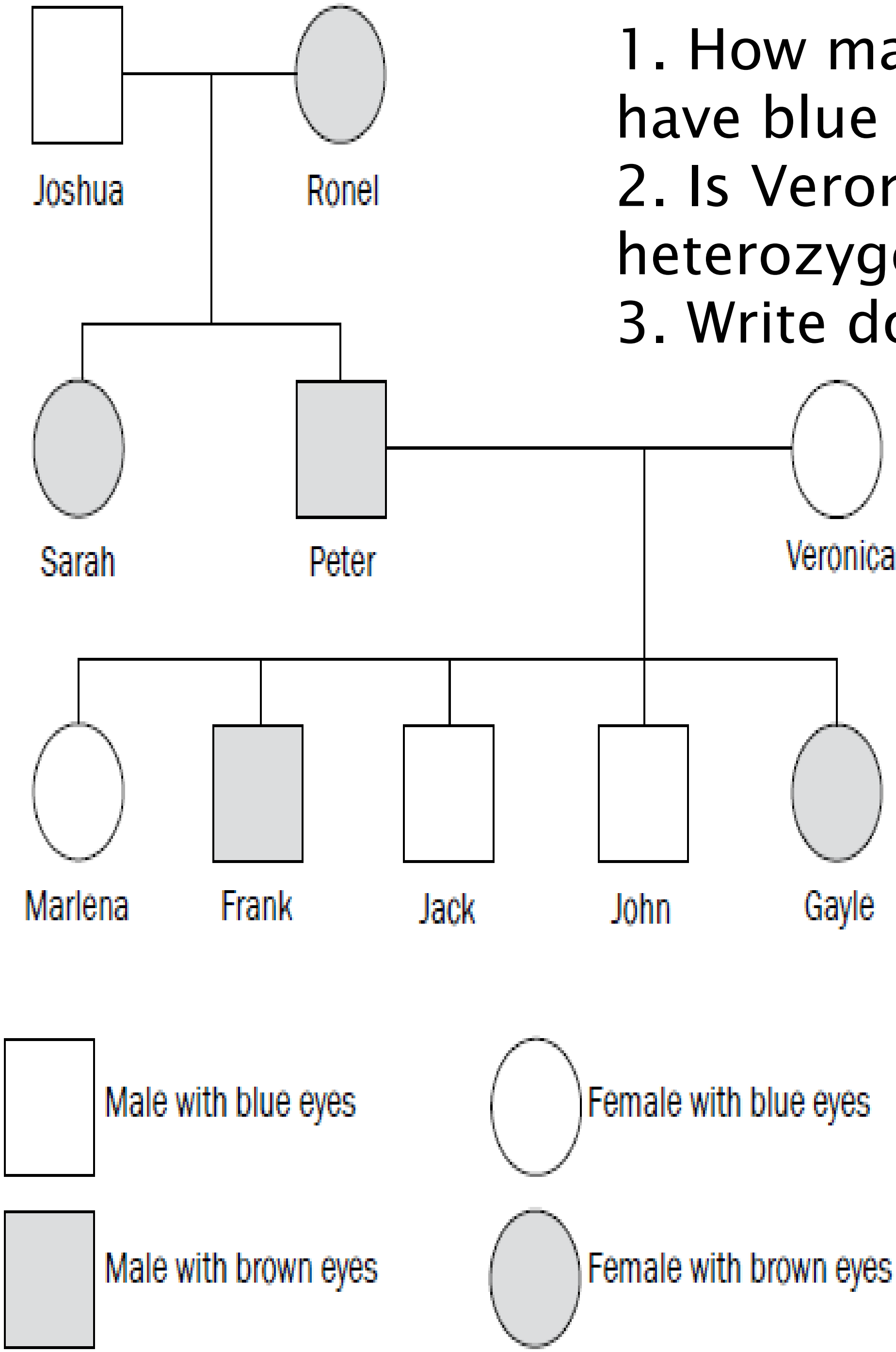

1. How many members of the family have blue eyes? (1)
2. Is Veronica homozygous or heterozygous for eye colour? (1)
3. Write down the genotype of:
    - a) Joshua (2)
    - b) Ronel (2)
    - c) Frank

4 Sarah married Jack and produced a kid with blue eyes. Show how possible could this be using genetic cross

1. 5 (1)
2. Homozygous (1)
3. a) aa (2)

b) AA/Aa (2)

c) Aa(2)

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NOTE : THIS IS CAUSED BY A **DOMINANT** ALLELE

- P2 phenotype: Sarah X Jack
- Genotype: Aa X Aa
- Meisosis
- Gametes A a A a
- Fertilization

Gametes	A	a
A	AA	Aa
a	Aa	aa

- F2 offspring Phenotypes: 3 brown eyes ,
  1 blue eyes

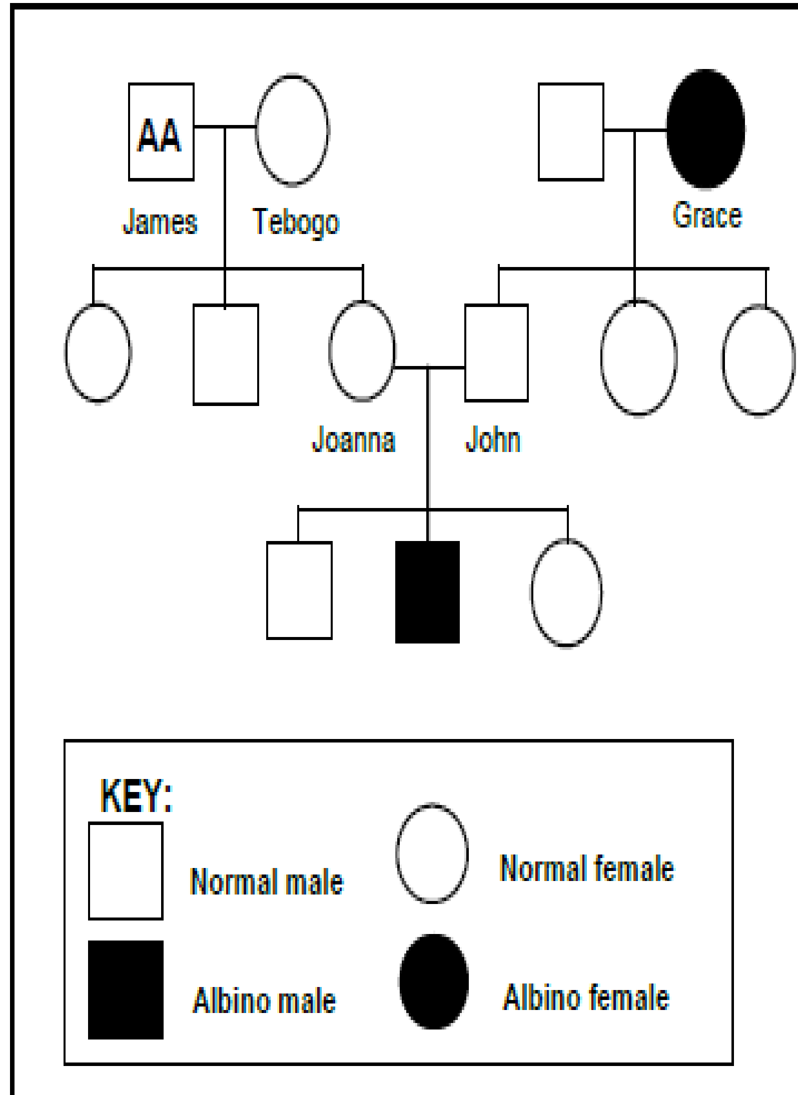
PRACTICE QUESTIONs (teacher must guide)

NOTE :

- 1. THIS IS CAUSED BY A **RECESSIVE** ALLELE
- 2. WE MIGHT GIVE YOU A **KEY** OR **NOT**

- **QN**: The pedigree diagram below shows the inheritance of albinism in a family. The genotype of James is shown in the diagram.

- 7.1 How many grandsons do James and Tebogo have?
- 7.2 What is:
- (a) Grace's phenotype
- (b) John's genotype
- 7.3 John and Joanna wish to have another child. What Is the percentage chance that the child will:
- (a) Be a girl
- (b) Have albinism



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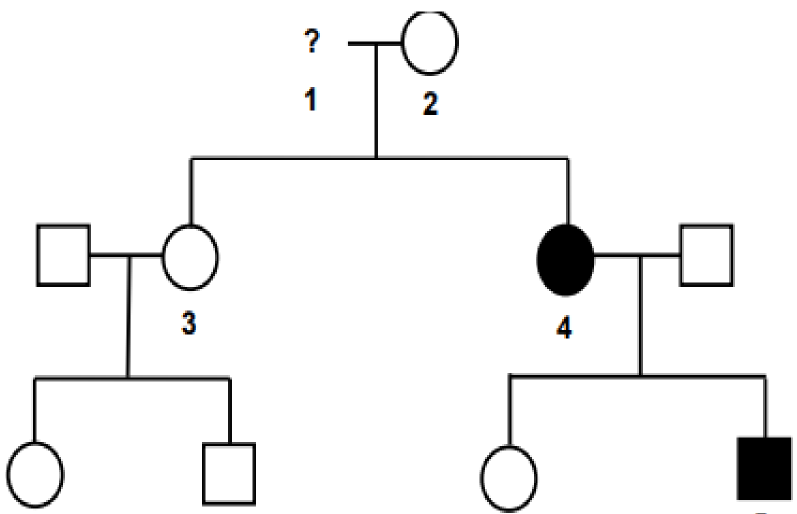
CROSSES WITH ASEX-LINKED TRAIT

- **QN** Severe combined immune deficiency syndrome (SCID) is a disorder affecting the immune system. It is caused by a sex-linked
- The diagram below shows the inheritance of the disorder in a family. It is not known if individual 1 has the disorder or not. Recessive allele (X^d).



Give the:

(a) Phenotype of individual **2**

(b) Phenotype of individual **1**

(c) Genotype of individual **3**

- A- Normal female
- B-male with SCID
- C-X^DX^d/X^DX^D

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QUESTION 2

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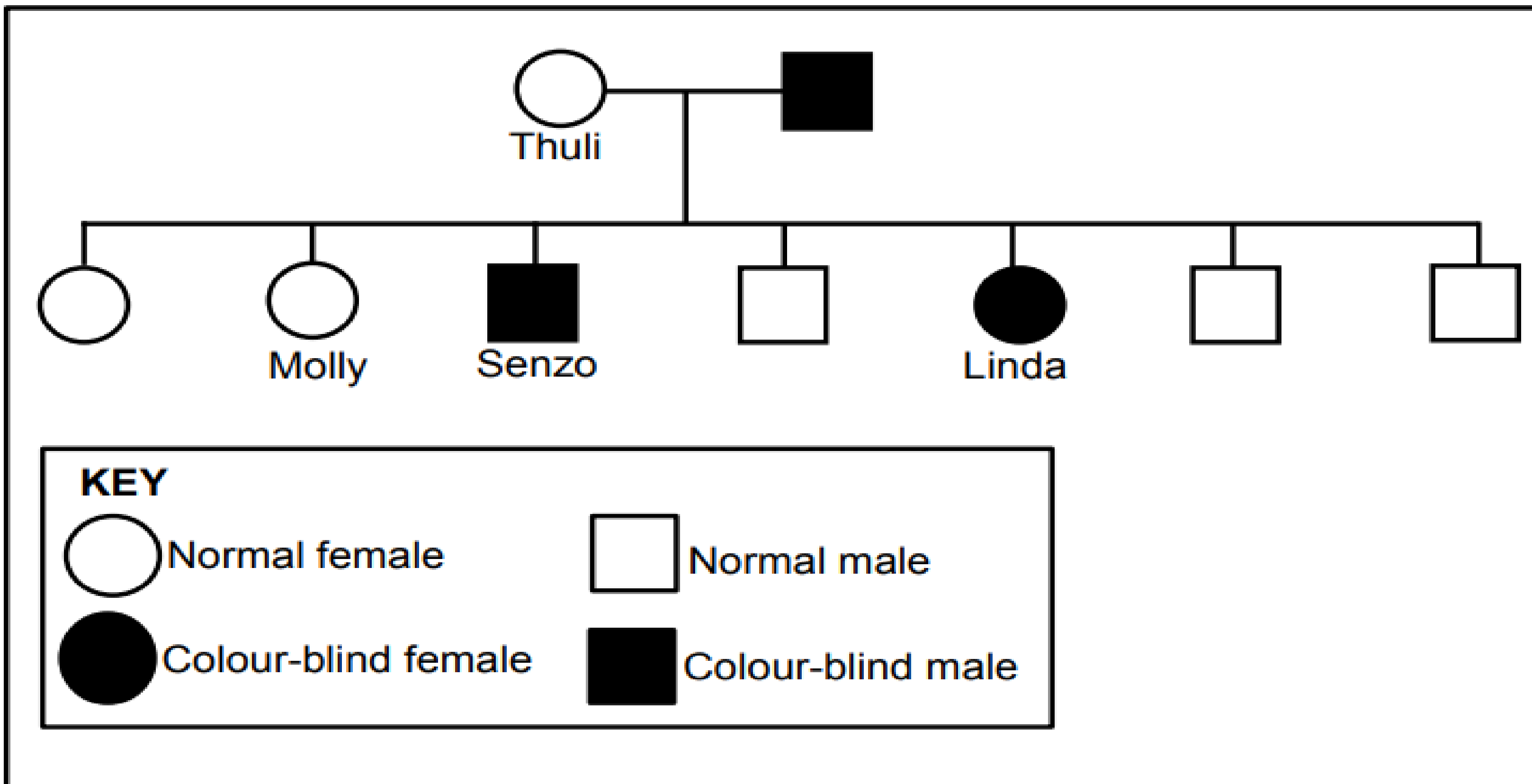
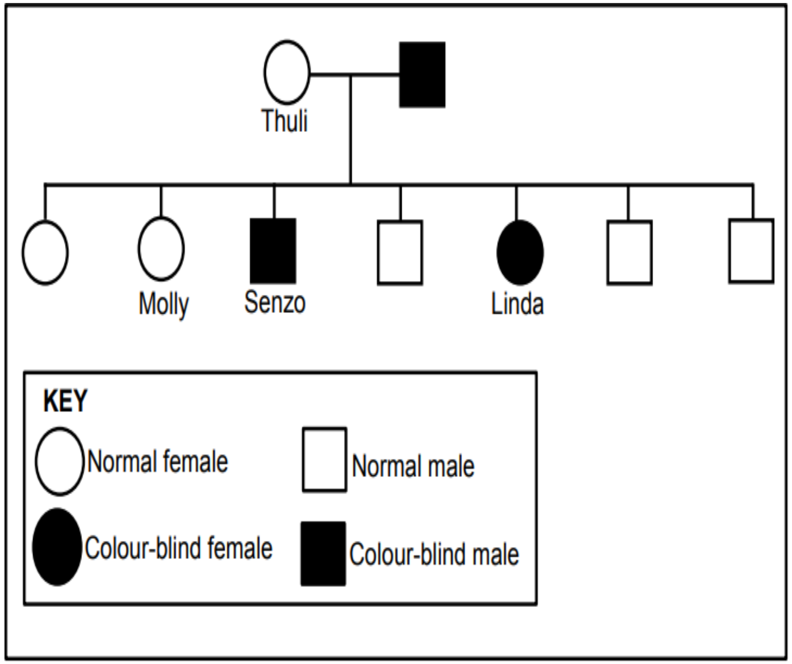
- Colour-blindness (Daltonism) is a sex-linked disorder caused by a recessive allele (X^d).
- The diagram below shows the inheritance of this disorder in a family.



- 2.3.1 Give the:
- (a) Phenotype of Senzo
- (b) Genotype of Thuli
- 2.3.2 Describe how Linda inherited colour-blindness
- 2.3.3 Explain why there are generally more males than females with colourblindness in a population.



➢2.3.4 Molly married a 'normal male'. Use a genetic cross to show the possible genotypes and phenotypes of their children.

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2.3.1 (a) Colour blind male✓/male with Daltonism (1)

(b) X^DX^d✓ (1)

2.3.2 - Linda inherited one recessive allele/X^d from her father✓
- and one recessive allele/X^d from her mother✓ (2)

2.3.3 - Males only have one X-chromosome✓
- If this chromosome carries the recessive allele✓/X^d
- the male will be colour blind✓
- Females have 2 X-chromosomes✓
- They need to have two recessive alleles✓/X^d X^d to be affected
- A dominant allele on the other X-chromosome will mask the effect✓

Any 4 (4)

P₁	Phenotype	Normal female	x	Normal male ✓
	Genotype	X^DX^d	x	X^DY ✓
Meiosis				
	G/gametes	X^D, X^d	x	X^D,Y✓
Fertilisation				
F₁	Genotype	X^DX^D X^DY X^DX^d X^dY✓ *		
	Phenotype	Normal females, Normal male, Colour blind male } ✓ *		

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OR

P_1	Phenotype	Normal female	X	Normal male ✓
	Genotype	X^DX^d	X	X^DY ✓

Meiosis

Fertilisation

Gametes	X^D	Y
X^D	X^DX^D	X^DY
X^d	X^DX^d	X^dY

1 mark for correct gametes✓

1 mark for correct genotypes✓ *

F_1 Phenotype: Normal females, Normal male, Colour blind male ✓ *

P_1 and F_1✓

Meiosis and fertilisation✓

***Compulsory** 2 + Any 4

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MUTATIONS

- Is the changing of the structure of a gene
- **EFFECTS OF MUTATIONS:**
- harmful mutations:may cause genetic disorders or cancer. Eg cystic
- harmless mutations: many codons specify the same amino acid, eg the codons CGU, CGC, CGA, and CGG all code for the amino acid Arg hence harmless
- useful mutations:have a positive effect on the organism in which they occur.eg HIV resistance

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CHROMOSOMAL MUTATION

- **3. Down syndrome** – due to an extra copy of chromosome 21 as a result of non disjunction during meiosis

GENETIC ENGINEERING

is the process whereby the genes on the DNA are changed, transferred or manipulated to produce a different organism.

Genetic engineering uses biotechnology to satisfy human needs e.g Stem cell research, Genetically modified organisms, Cloning.

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- **Mutations** causes the organism's DNA to change that create diversity within a population by introducing new alleles. This cause variation among organisms
- GENE MUTATION
- is a permanent alteration in the DNA sequence that makes up a **gene** eg
- 1.Haemophilia – due to absence of blood clotting factors
- 2. Colour blindness – due to absence of the proteins that comprise either the red or green cones/photoreceptors in the eye

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FOUR DISADVANTAGES OF GENETIC ENGINEERING/ BIOTECHNOLOGY

- • Expensive/research money could be used for other needs
- • Interfering with nature/immoral
- • Potential health impacts
- • Unsure of long-term effects

FOUR ADVANTAGES OF GENETIC ENGINEERING/ BIOTECHNOLOGY

- • Production of medication/resources cheaply
- • Control pests with specific genes inserted into a crop
- • Using specific genes to increase crop yields/food security
- • Selecting genes to increase shelf-life of plant products

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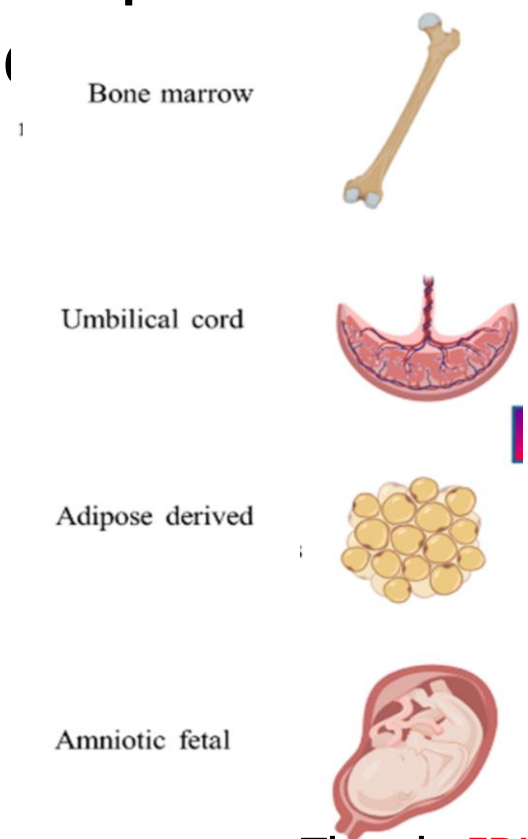
BIOTECHNOLOGY: living organisms are used to make useful chemicals and products or to perform an industrial task

STEM CELLS

- Cells with unique ability to develop into specialised **cell** types in the bo
- SOURCES OF STEM CELLS
- **Bone marrow.**
- **Umbilical cord blood.**
- **Adipose tissue.**
- **Amniotic fluid.**



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USES OF STEM CELLS

- grow new **cells** in a laboratory to replace damaged organs or tissues.
- correct parts of organs that don't work properly.
- research causes of genetic defects in **cells**.
- research how diseases occur or why certain **cells** develop into cancer **cells**

CLONING

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- It allows farmers and ranchers to accelerate the reproduction of their most productive livestock in order to better produce safe and healthy food.
- **Cloning** reproduces the healthiest animals, thus minimizing the use of antibiotics, growth hormones and other chemicals.

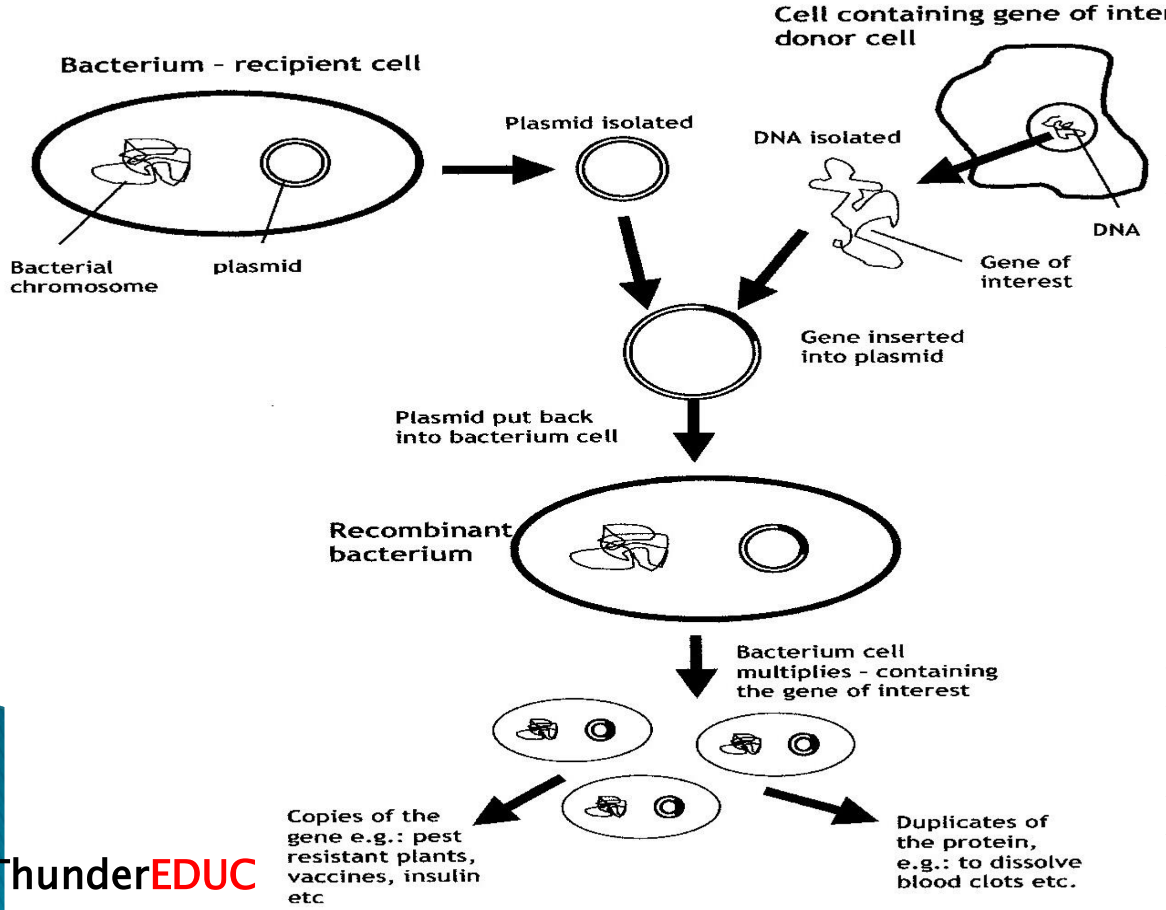
➢CLONING STEPS/ PROCESS

➢Isolation of target DNA fragments (often referred to as inserts)

➢inserts into an appropriate cloning vector, creating recombinant molecules (e.g., plasmids)

- **Transformation** of <u>recombinant</u> plasmids **into bacteria** or other suitable host for propagation
- **selection of hosts** containing the intended recombinant plasmid

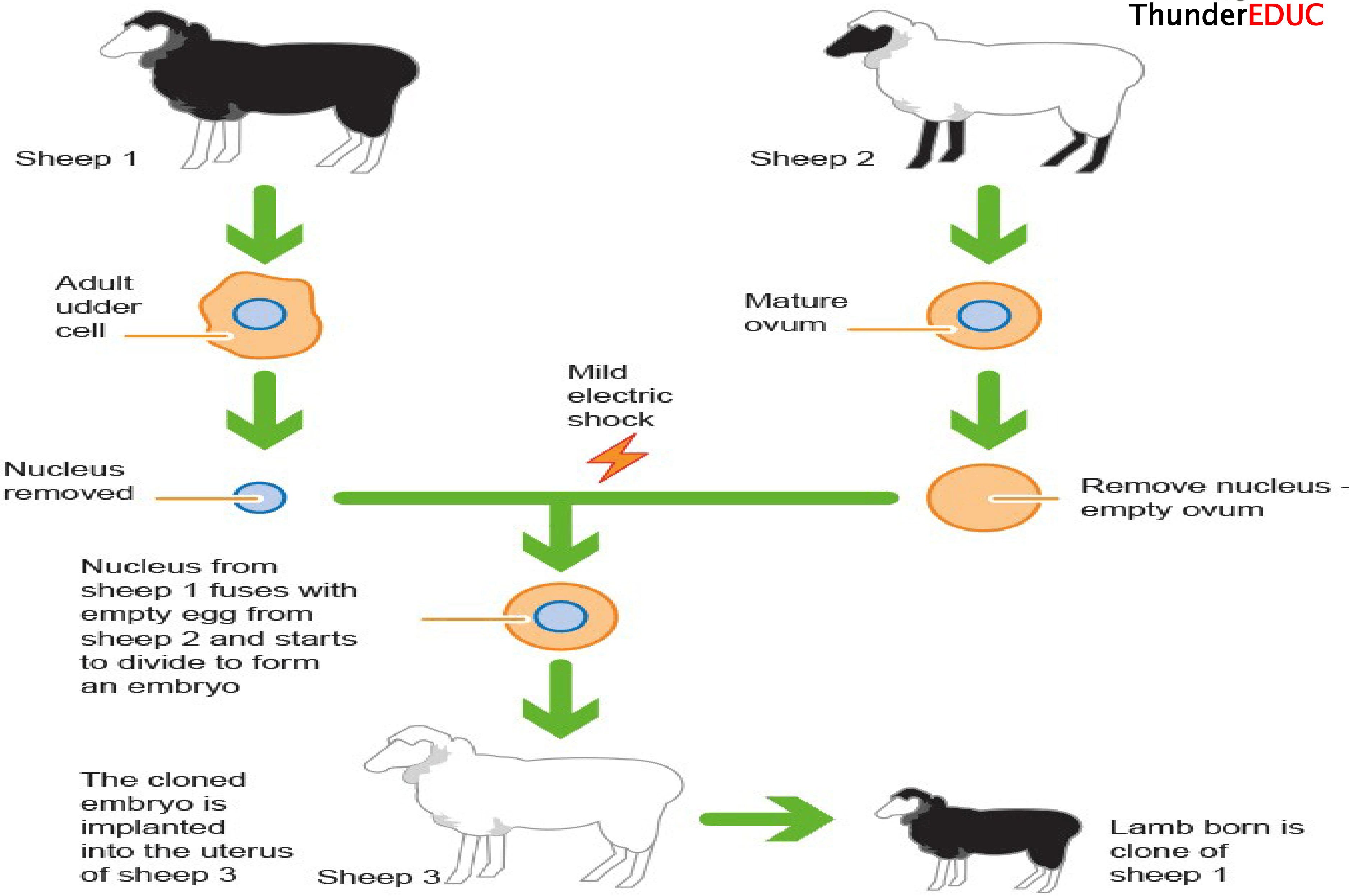
PRODUCTION OF INSULIN USING BIOTECHNOLOGY/CLONNING



STEPS

- A plasmid/ circular DNA is removed from the bacterial cell·
- – It is cut· using enzymes
- – The insulin gene is removed from a human cell· and
- – inserted into the plasmid·
- to form the recombinant DNA

CLONING OF A DOLLY SHEEP



2. DNA PROFILING

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- – A child received DNA from both parents.
- – The DNA profiles of the mother, child and the possible father are determined.
- – A comparison of the DNA bands of the mother and the child is made.
- – The remaining DNA bands are compared to the possible father's DNA bands.
- – If all the remaining DNA bands in the child's profile match the possible father's DNA bands.
- – then the possible father is the biological father.
- – If all the remaining DNA bands in the child's profile does not match the possible father's DNA bands.
- – then the possible father is not the biological father.

PATERNITY TESTING

is test, to determine whether a man is the father of a particular child.

- 1.BLOOD GROUPING
- – The blood group of a child is determined by the alleles received from both parents.
- – The blood group of the mother, the child and the possible father is determined.
- – If the blood group of the mother and possible father cannot lead to the blood group of the child.
- – the man is not the father.
- – If the blood group of the mother and the possible father can lead to the blood group of the child.
- – the man might be the father.
- – This is not conclusive.
- – because many men have the same blood group.

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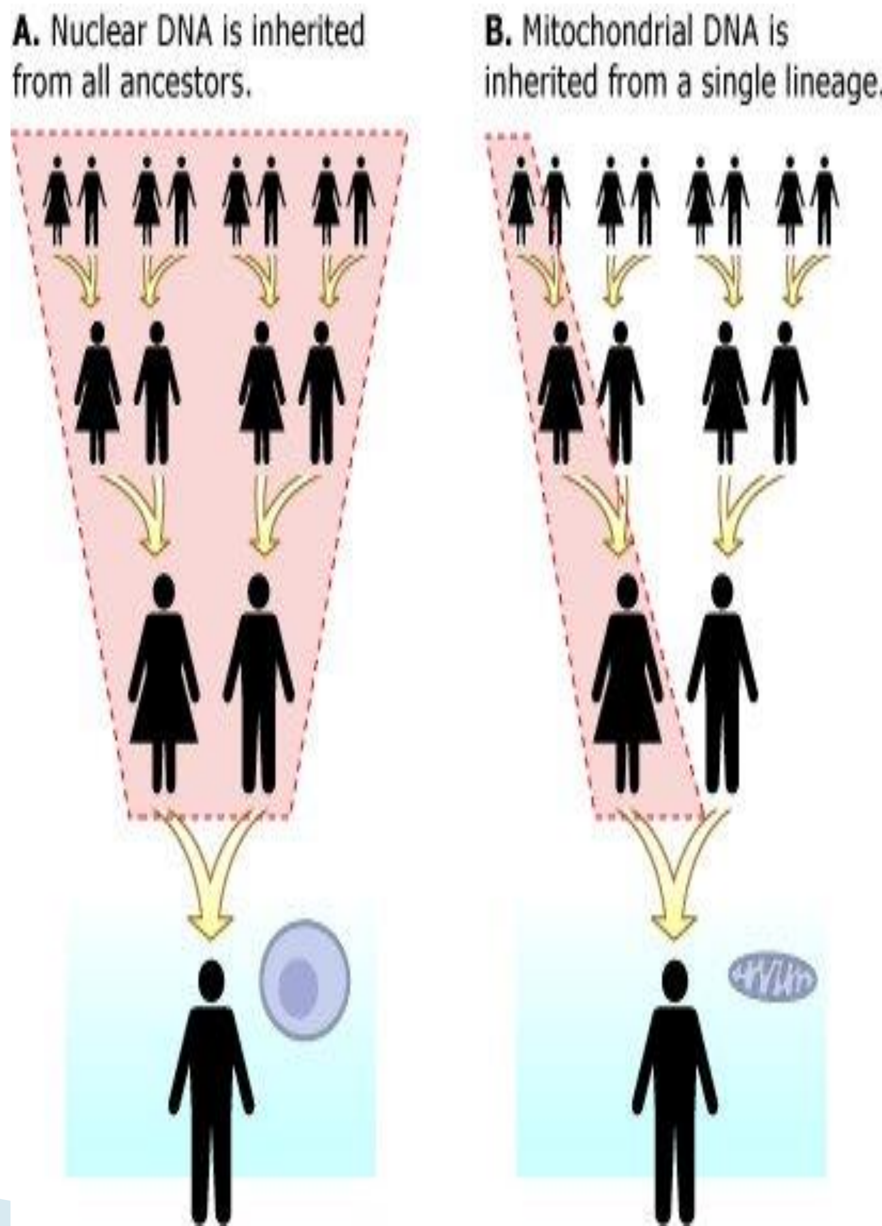
MITOCHONDRIAL DNA

- Human mitochondrial DNA is much smaller than human nuclear DNA. For both of these reasons, the sequence of mitochondrial DNA stays the same over generations, and thus is a useful tool for looking at maternal ancestry.



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