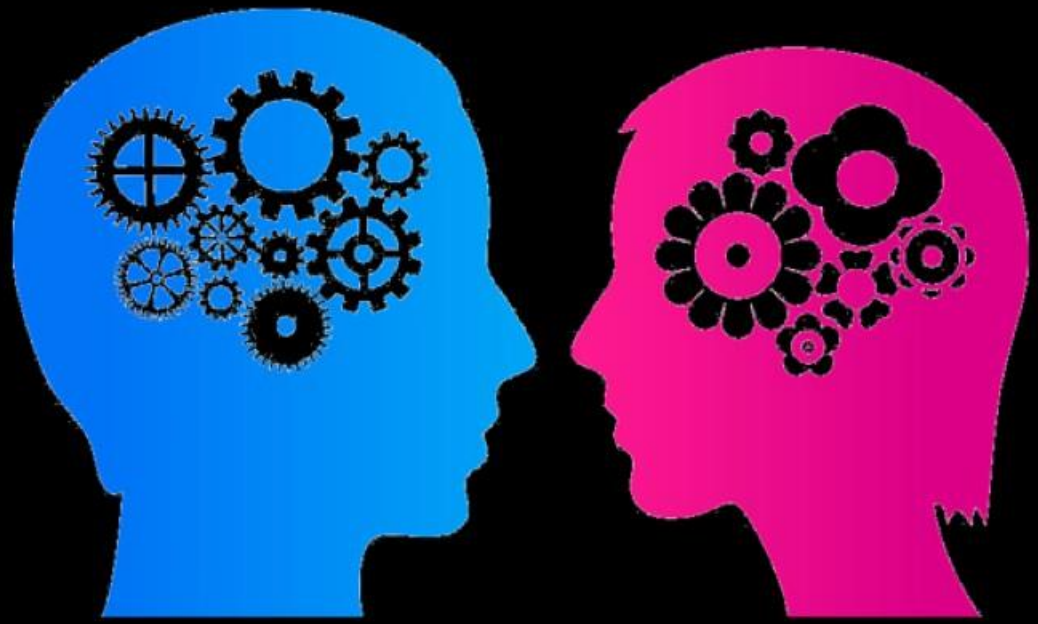
Part 6

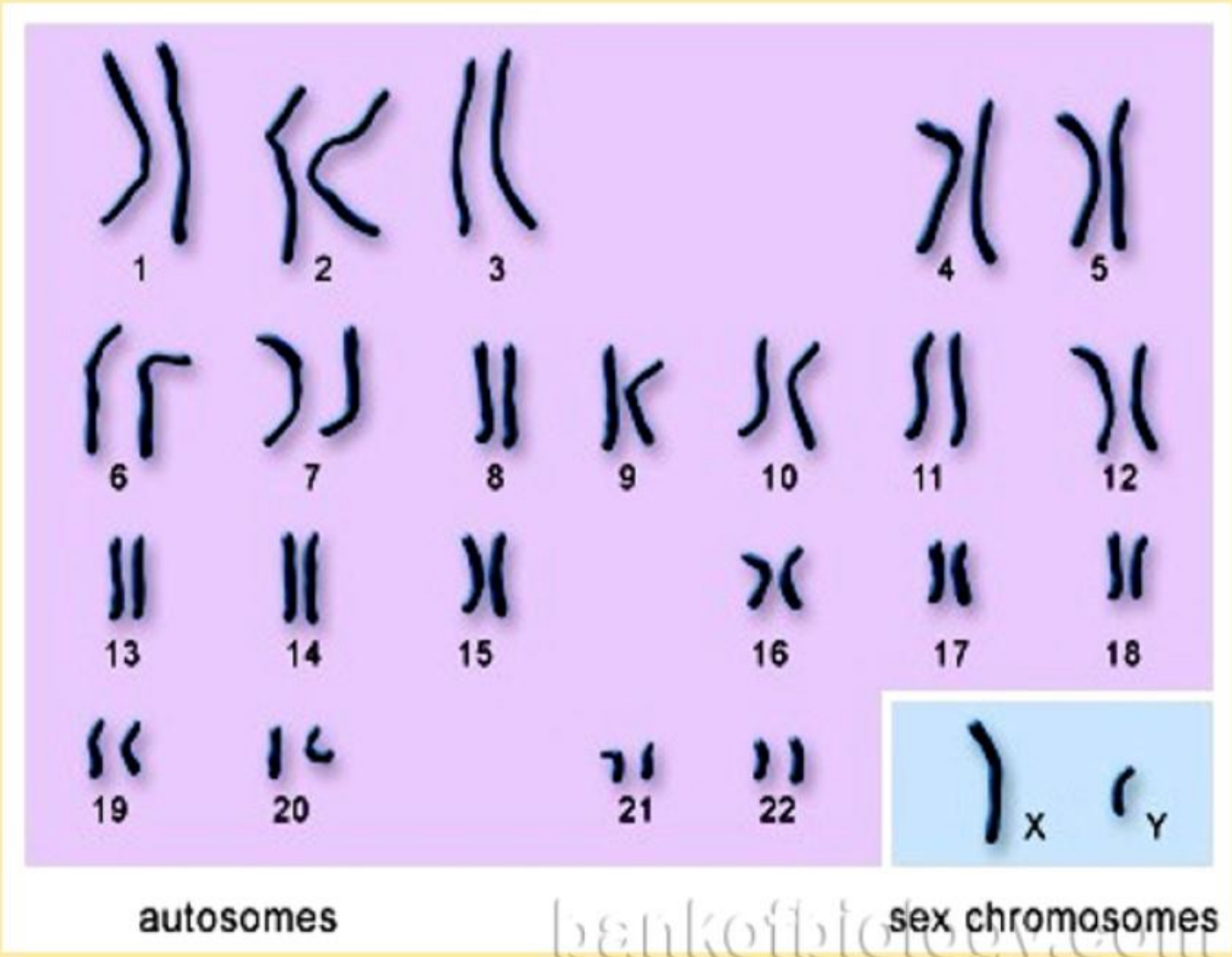


SEX DETERMINATION

SEX DETERMINATION

Autosomes and Sex chromosomes (allosomes)

- The chromosomes that are involved in sex determination are called sex chromosomes (allosomes).
- They include X & Y chromosomes.
- Autosomes are chromosomes other than sex chromosomes.
- Number of autosomes is same in males and females.



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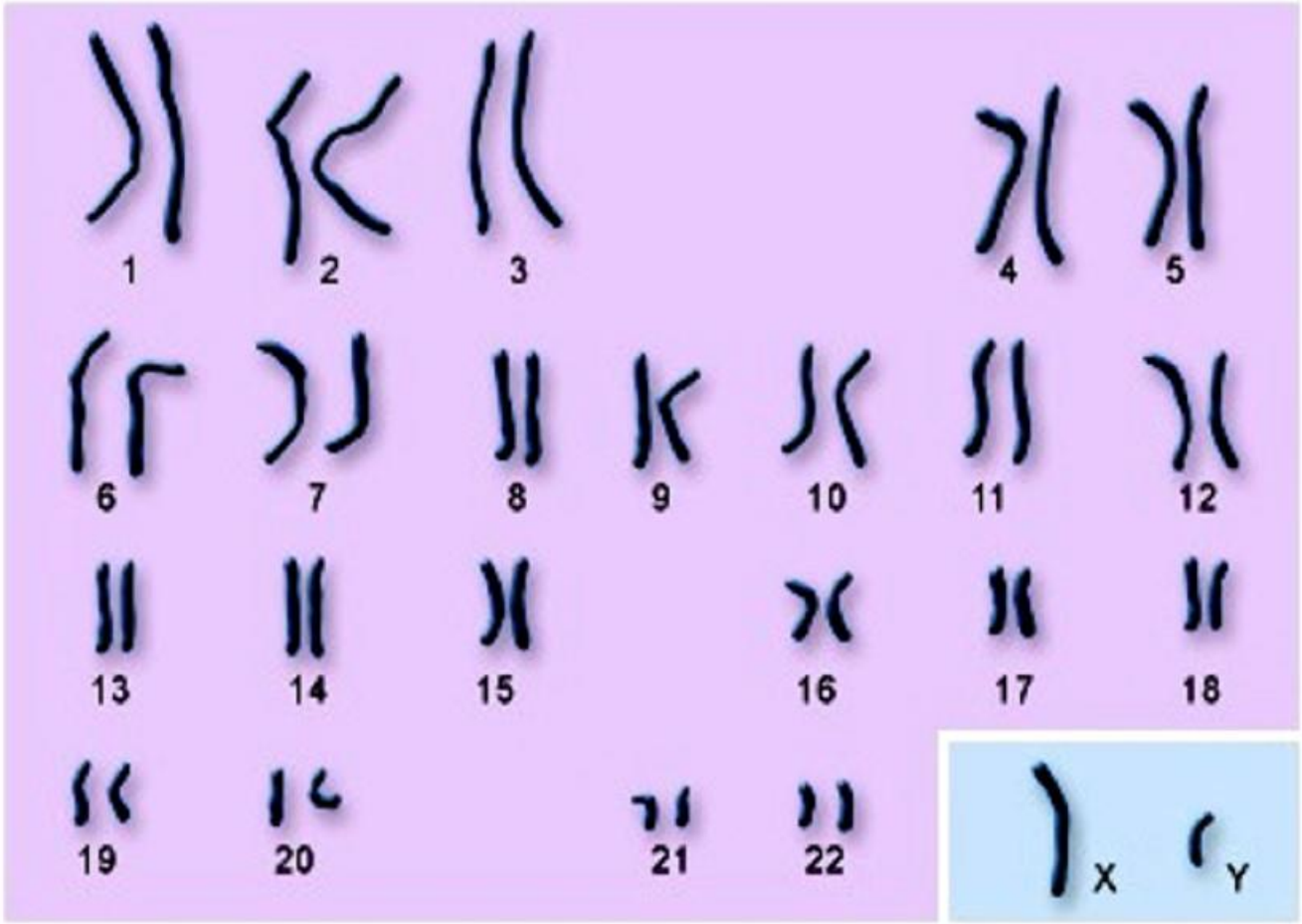
SEX DETERMINATION

Autosomes and Sex chromosomes (allosomes)

- **Henking** (1891) studied spermatogenesis in some insects and observed that 50% of sperm received a nuclear structure after spermatogenesis, and other 50 % sperm did not receive it.
- Henking called this structure as the **X body** (now it is called as **X-chromosome**).



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SEX DETERMINATION

Mechanism of sex determination

- **XX – XO Mechanism**
- **XX – XY Mechanism**
- **ZZ – ZW Mechanism**

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SEX DETERMINATION

Mechanism of Sex Determination

1. XX-XO Mechanism

- Here, male is heterogametic, i.e. XO (Gametes with X and gametes without X) and female is homogametic, i.e. XX (all gametes are with X-chromosomes).
- E.g. Many insects such as grasshopper.

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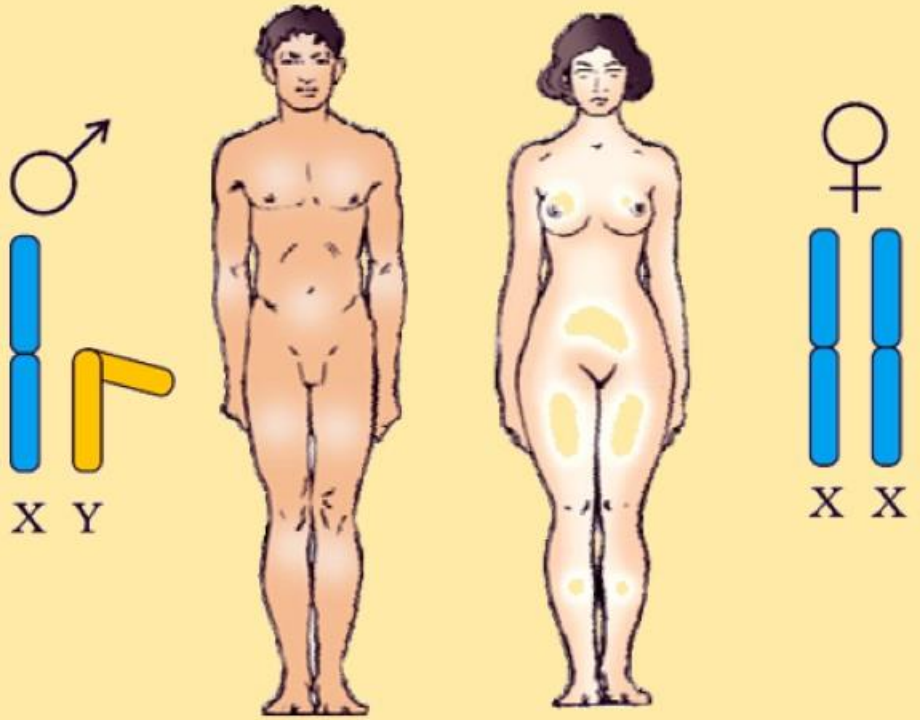
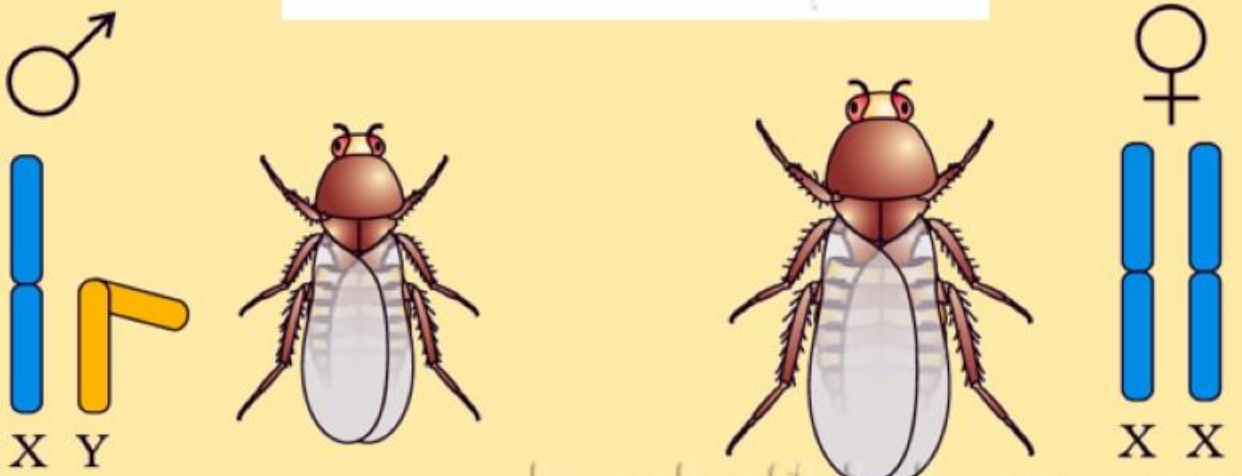
SEX DETERMINATION

Mechanism of Sex Determination

2. XX-XY Mechanism

- **Male is heterogametic (X & Y) and female is homogametic (X only).**
- **E.g. Human & *Drosophila*.**





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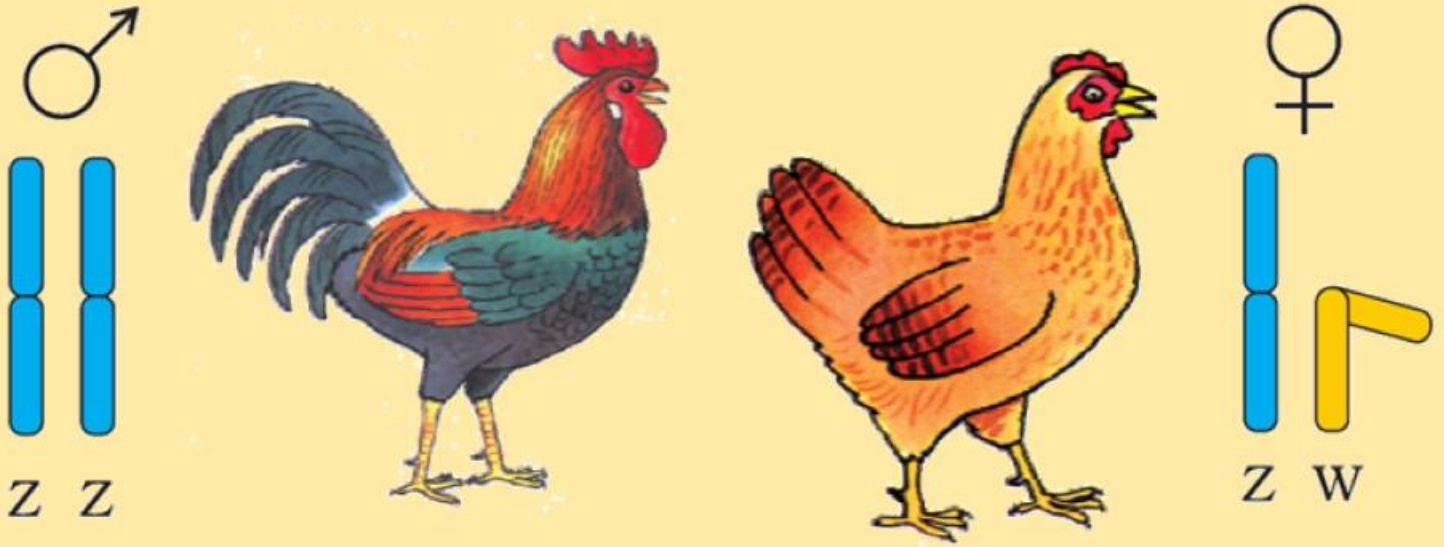
SEX DETERMINATION

Mechanism of Sex Determination

3. ZZ-ZW Mechanism

- Male is homogametic (ZZ) and female is heterogametic (Z & W).
- E.g. Birds.



XX-XO and XX-XY mechanisms show male heterogamety.
ZZ-ZW mechanism shows female heterogamety.

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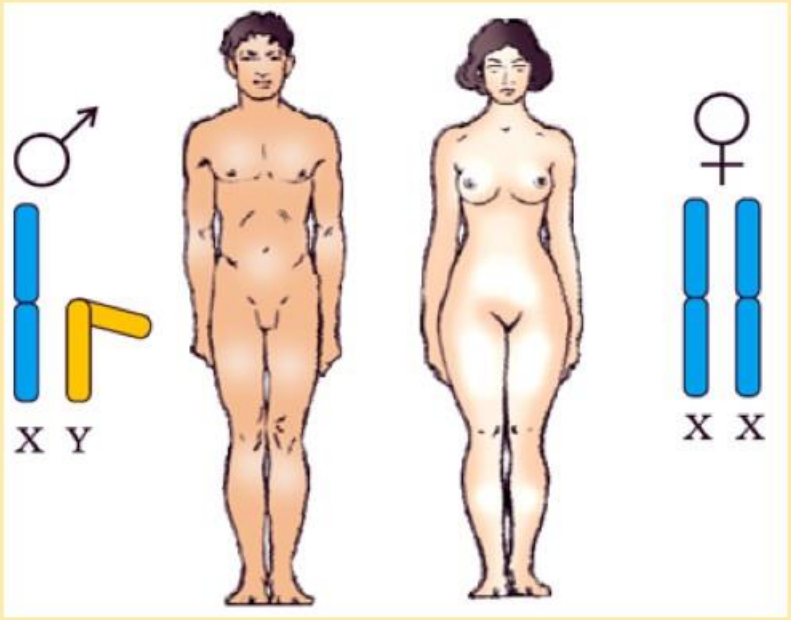
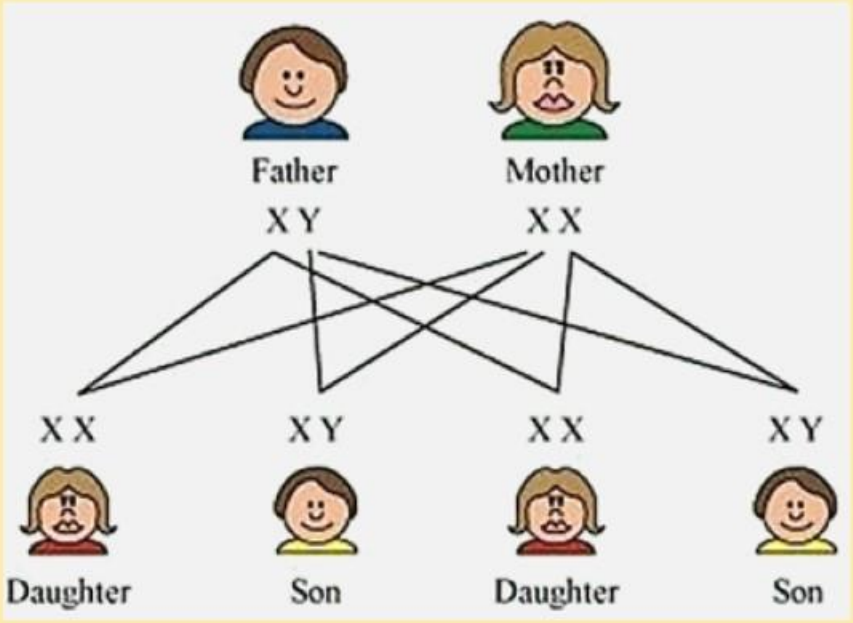
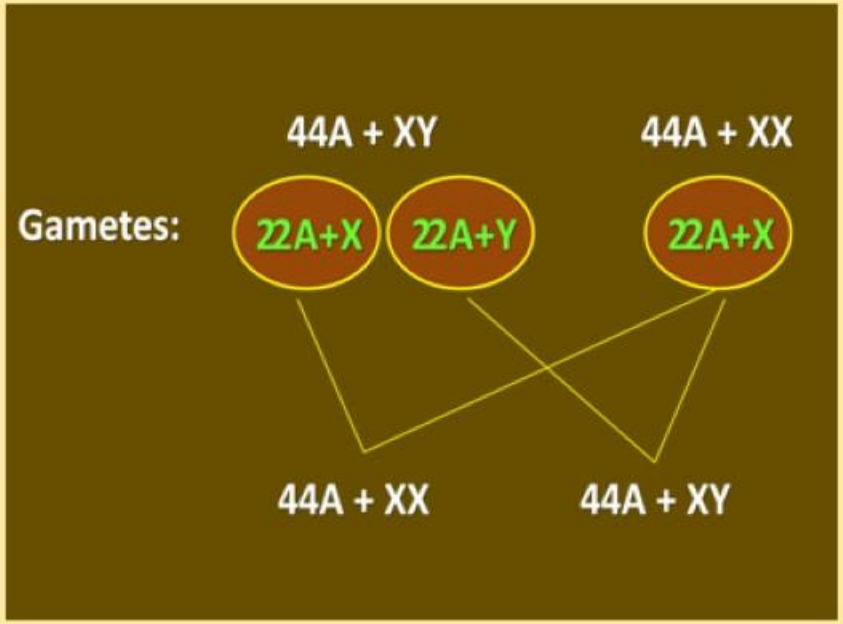
SEX DETERMINATION

Sex Determination in Humans (XX-XY type)







- Human has 23 pairs of chromosomes (22 pairs of autosomes + 1 pair of sex chromosomes).
- A pair of X-chromosomes (XX) is present in female.
- X & Y chromosomes (XY) are present in male.

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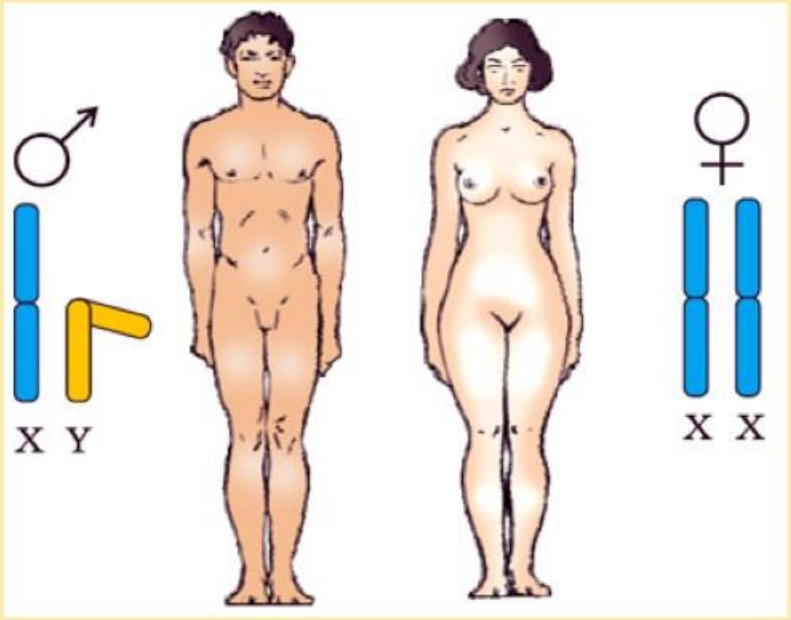
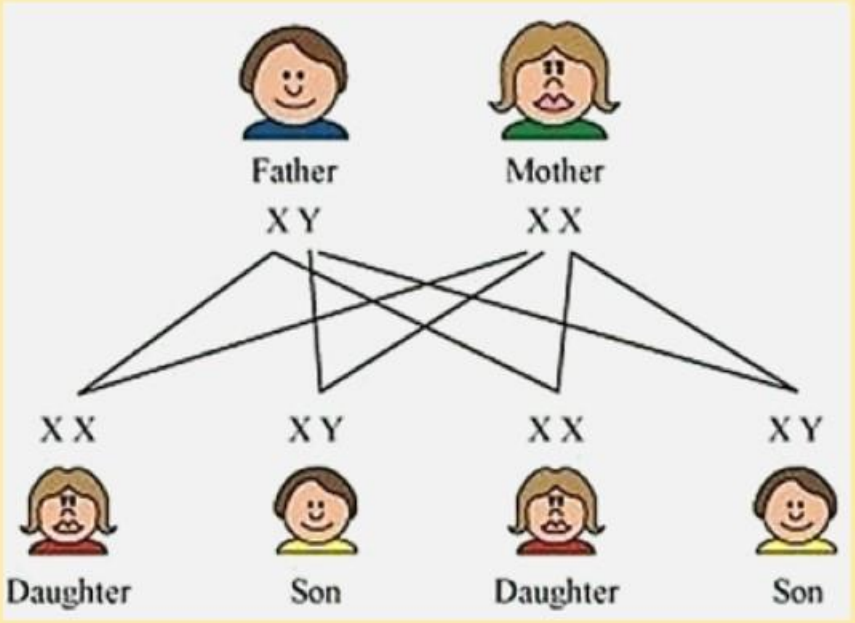
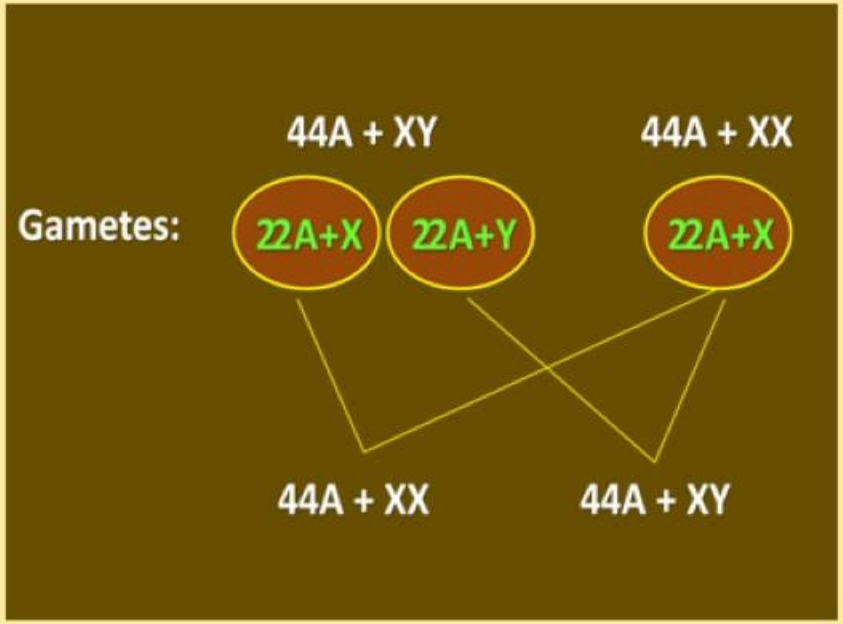
SEX DETERMINATION

Sex Determination in Humans (XX-XY type)







- During spermatogenesis, males produce 2 types of gametes- 50% with X chromosome and 50% with Y chromosome.
- Females produce only ovum with an X-chromosome.

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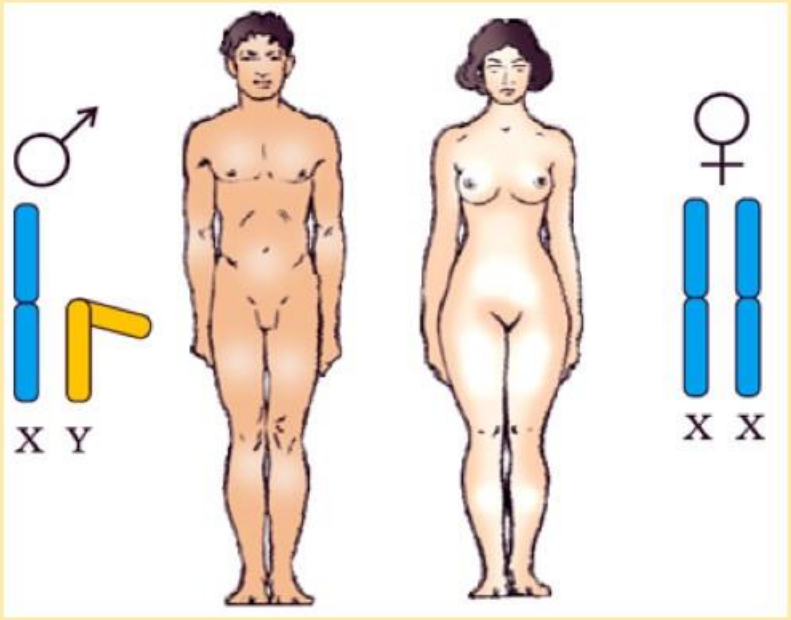
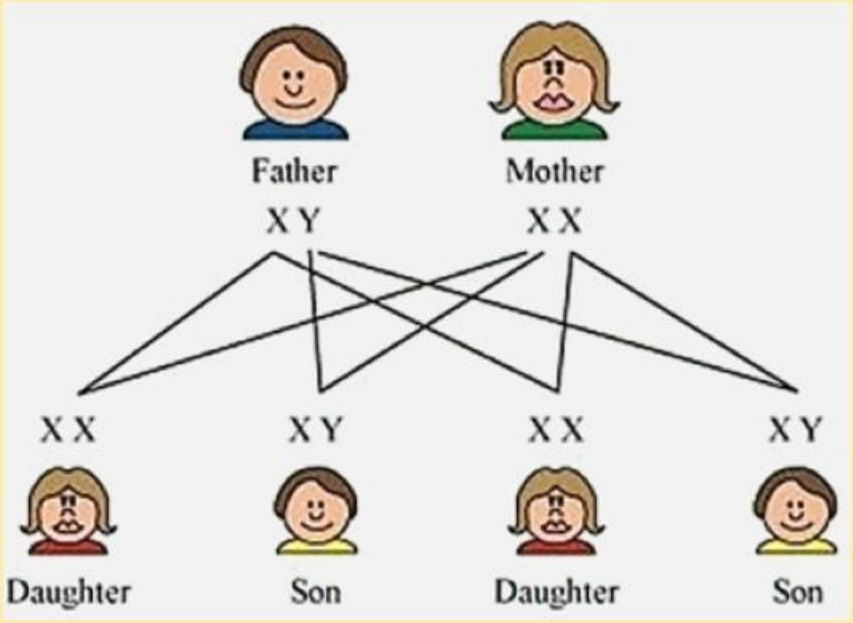
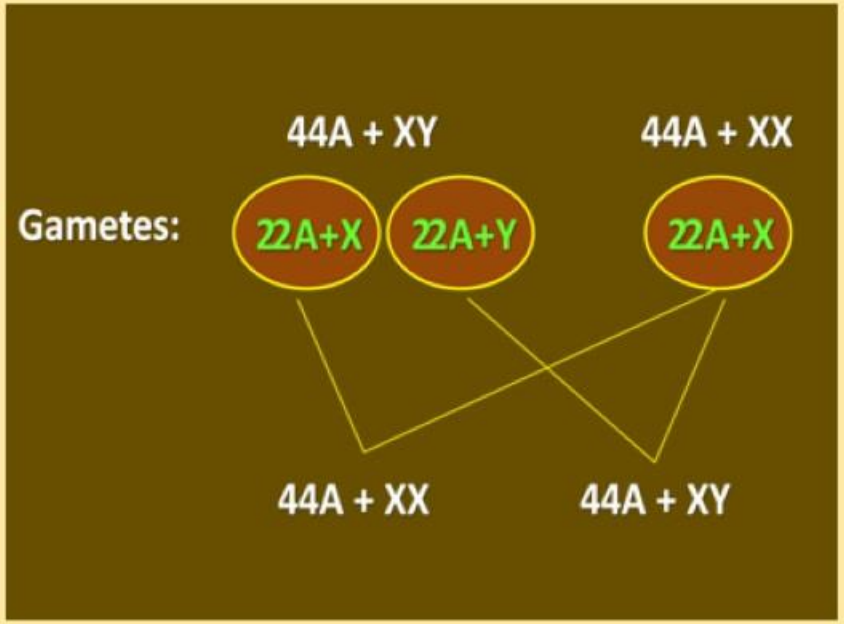
SEX DETERMINATION

Sex Determination in Humans (XX-XY type)







- There is an equal probability of fertilization of ovum with the sperm carrying either X or Y chromosome.

The sperm determines whether the offspring male or female.

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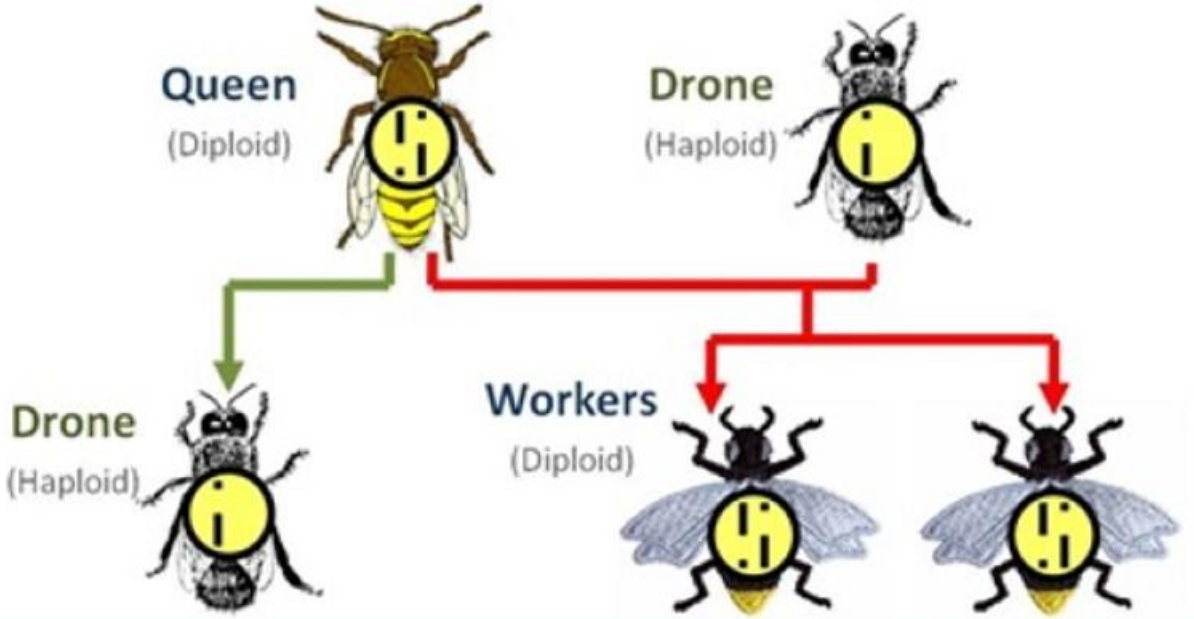
SEX DETERMINATION

Sex Determination in Honeybee



Parents:	Female (32)	Male (16)
	↓*Meiosis*	↓*Mitosis*
Gametes:	16 16	16
F_1:	Male (16)	Female (32)

- It is based on the number of sets of chromosomes an individual receives.
- Fertilised egg develops as a female (queen or worker).
- An unfertilised egg develops as a male (drone). It is called parthenogenesis.

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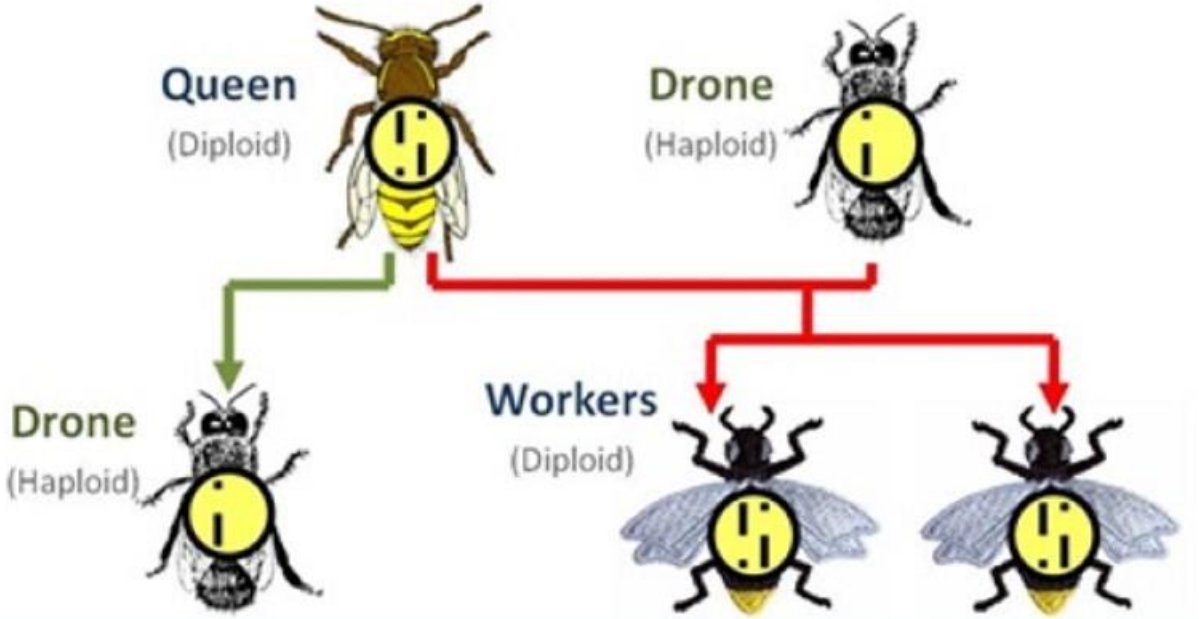
SEX DETERMINATION

Sex Determination in Honeybee



Parents:	Female (32)	Male (16)
	↓*Meiosis*	↓*Mitosis*
Gametes:	16 16	16
	↓	↓
F_1:	Male (16)	Female (32)

- **Therefore, the females are diploid (32 chromosomes) and males are haploid (16 chromosomes). This is called as haplodiploid sex determination system.**
- **In this system, the males produce sperms by mitosis. They do not have father and thus cannot have sons, but have a grandfather and can have grandsons.**

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Part 7
MUTATION

MUTATION

- It is a sudden heritable change occurring in DNA sequences that results changes in the genotype and the phenotype of an organism.
- Recombination and mutation leads to variation in DNA.

Types of mutation

- Point mutation
- Frame shift mutation

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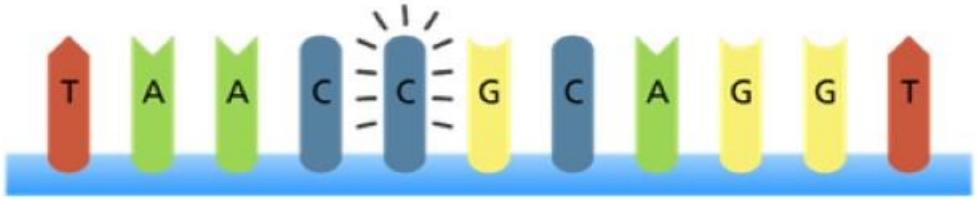
MUTATION

POINT MUTATION

- It is the mutation due to change in a single base pair of DNA.
- E.g. sickle cell anaemia.

Original sequence

Point mutation

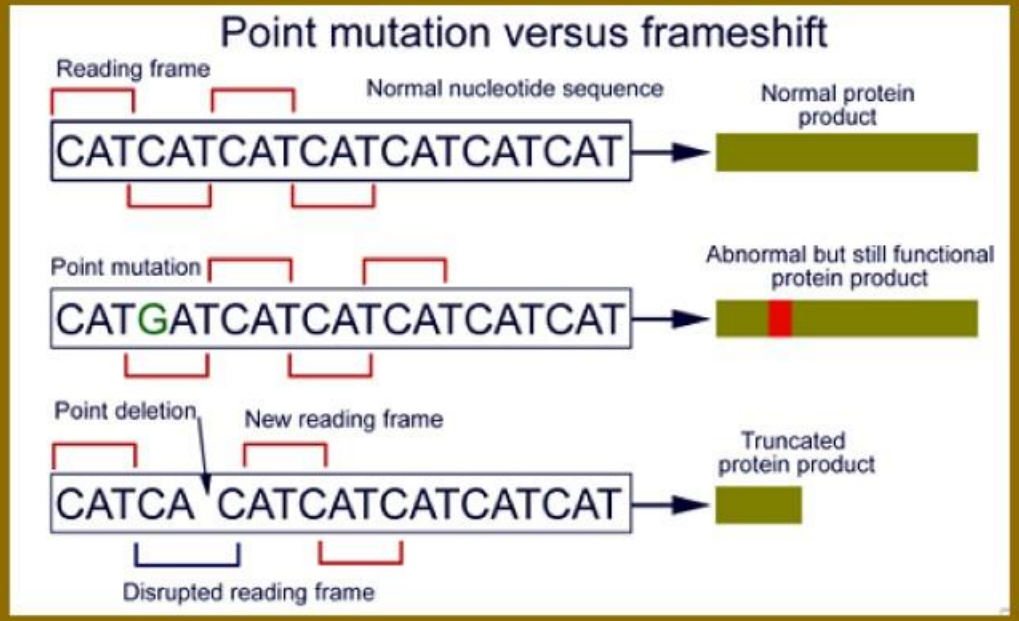


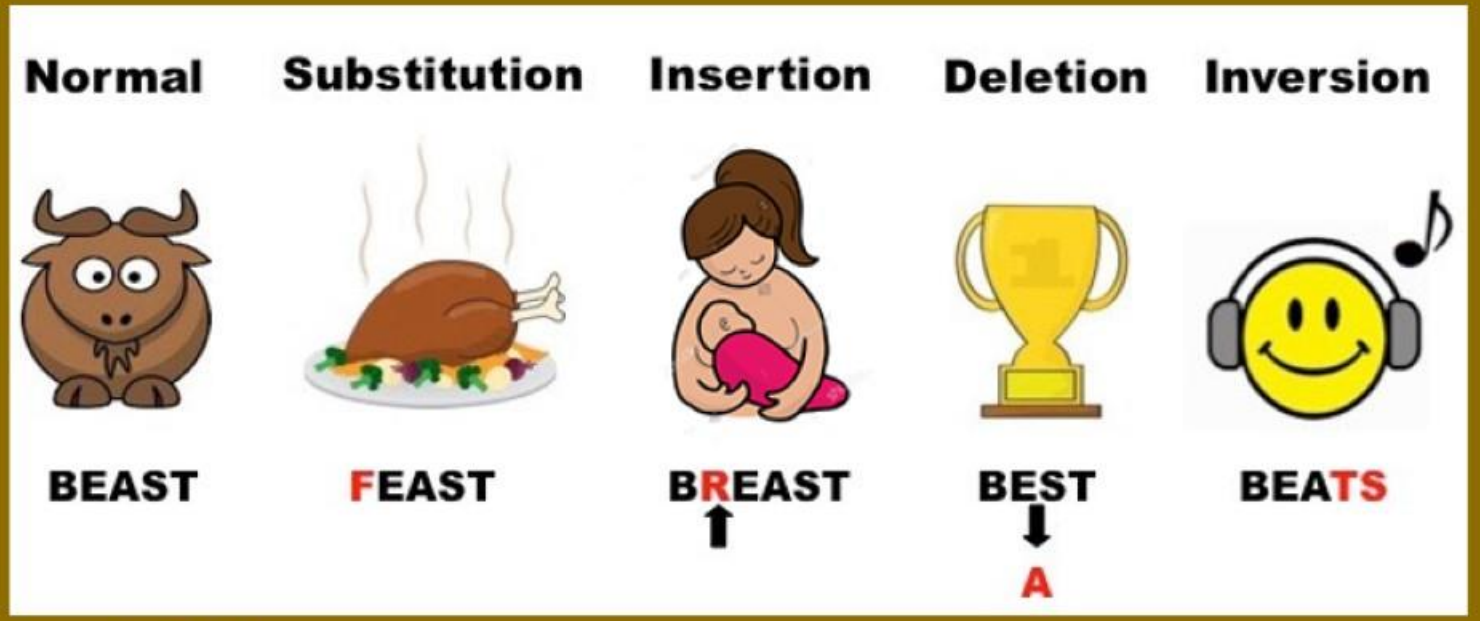
	No mutation	Point mutations			
		Silent	Nonsense	Missense	
				conservative	non-conservative
DNA level	TTC	TTT	ATC	TCC	TGC
mRNA level	AAG	AAA	UAG	AGG	ACG
protein level	Lys	Lys	STOP	Arg	Thr

MUTATION

FRAME SHIFT MUTATION

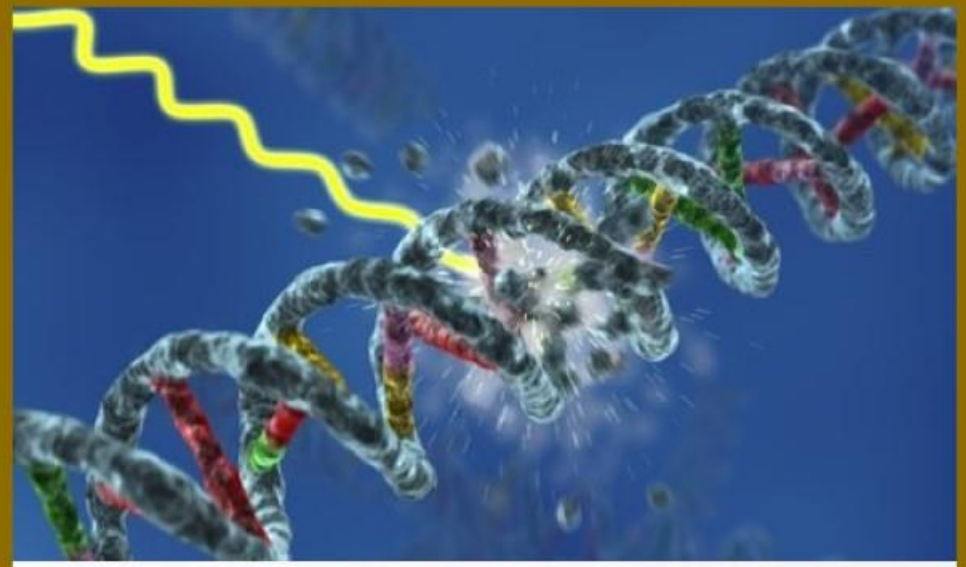
- It is the deletion or insertion of base pairs resulting in the shifting of DNA sequences.
- Loss (deletion) or gain (insertion/ duplication) of DNA segment cause Chromosomal abnormalities (aberrations).
- Chromosomal aberrations are seen in cancer cells.

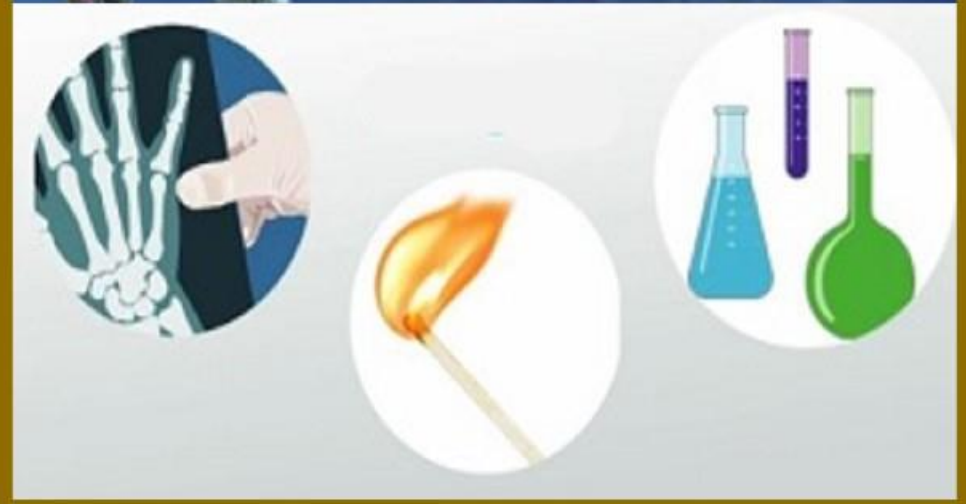




MUTATION

MUTAGENS





Mutagens are the agents that induce mutation.

Types of mutagens

Physical mutagens

UV radiation,
α, β, γ rays,
X-ray etc.

Chemical mutagens

Mustard gas, phenol, formalin, acetic acid, formic acid, NH_3 etc.

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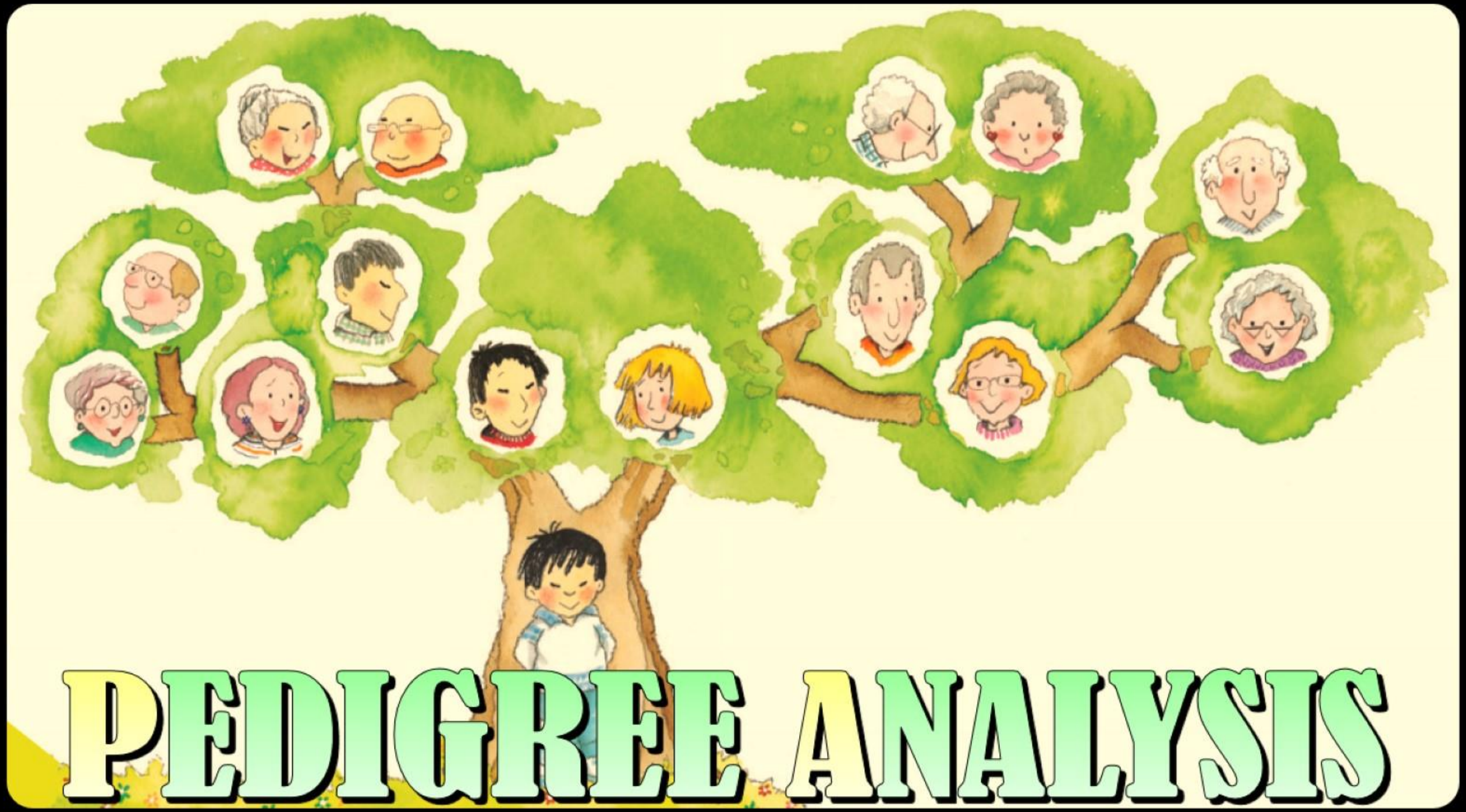
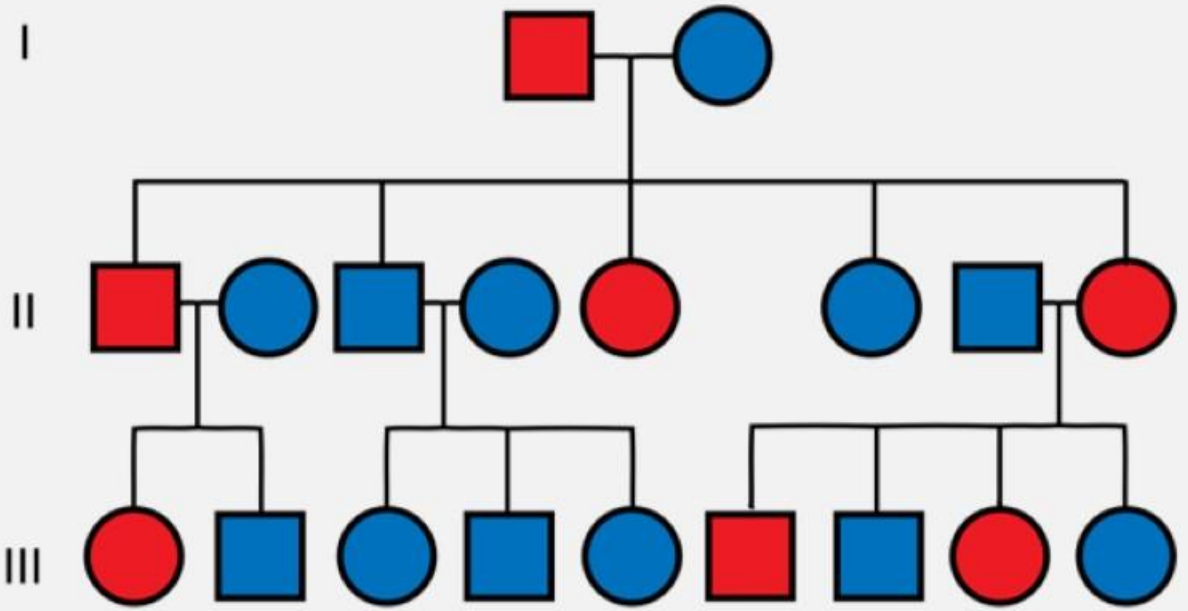

PEDIGREE ANALYSIS

PEDIGREE ANALYSIS



- In human, control crosses are not possible. So the study of family history about inheritance is used.
- Such an analysis of genetic traits in several generations of a family is called **pedigree analysis.**

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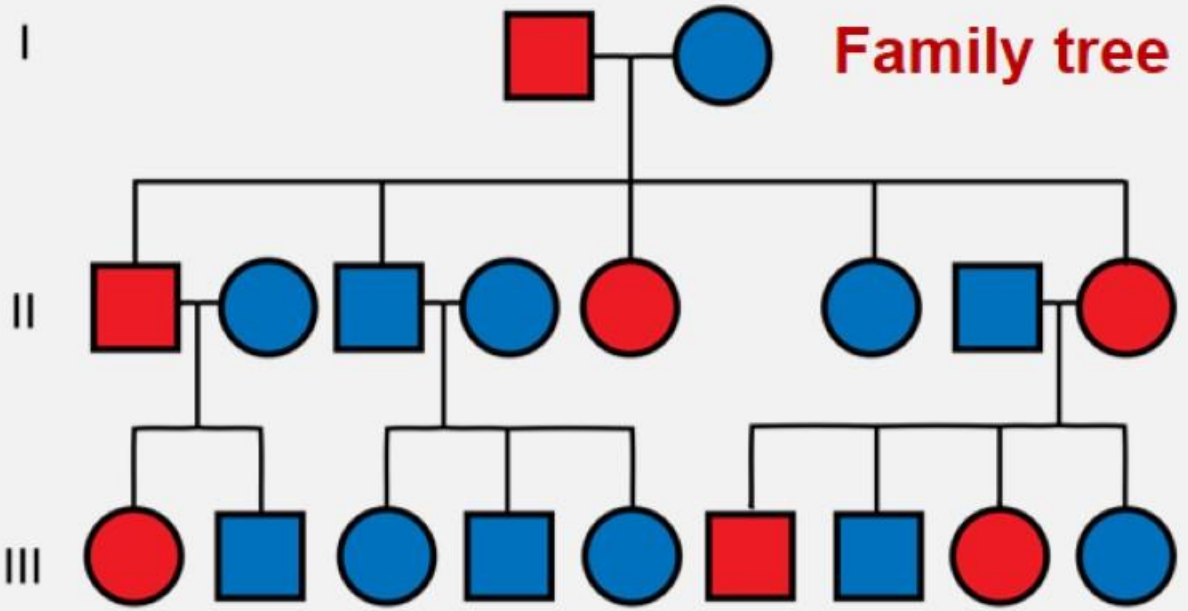
PEDIGREE ANALYSIS



- The representation or chart showing family history is called **family tree (pedigree).**
- In human genetics, pedigree study is utilized to trace the inheritance of a specific trait, abnormality or disease.

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PEDIGREE ANALYSIS

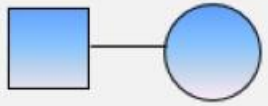
SYMBOLS USED IN PEDIGREE ANALYSIS

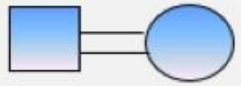
Male 

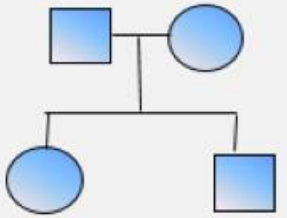
Female 

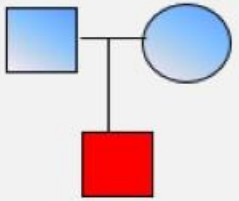
Sex unspecified 

Affected individuals 

Mating 

Mating b/w relatives (consanguineous mating) 

Parents above & children below 

Parents with affected male child 

Five unaffected offspring 

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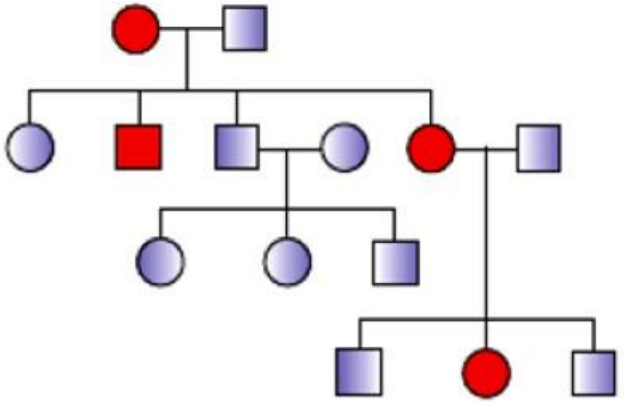
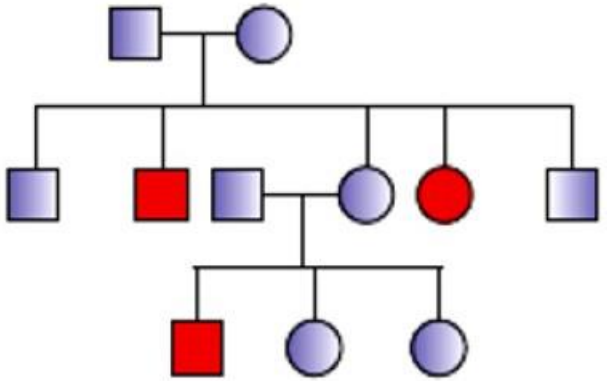
PEDIGREE ANALYSIS

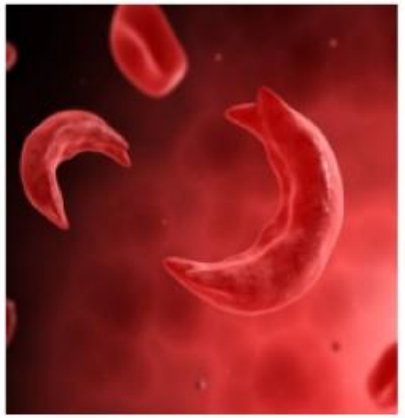
Pedigree analysis helps to understand whether a trait is dominant or recessive.



Pedigree analysis of Autosomal dominant trait (E.g. Myotonic dystrophy)



Pedigree analysis of Autosomal recessive trait (E.g. Sickle-cell anaemia)



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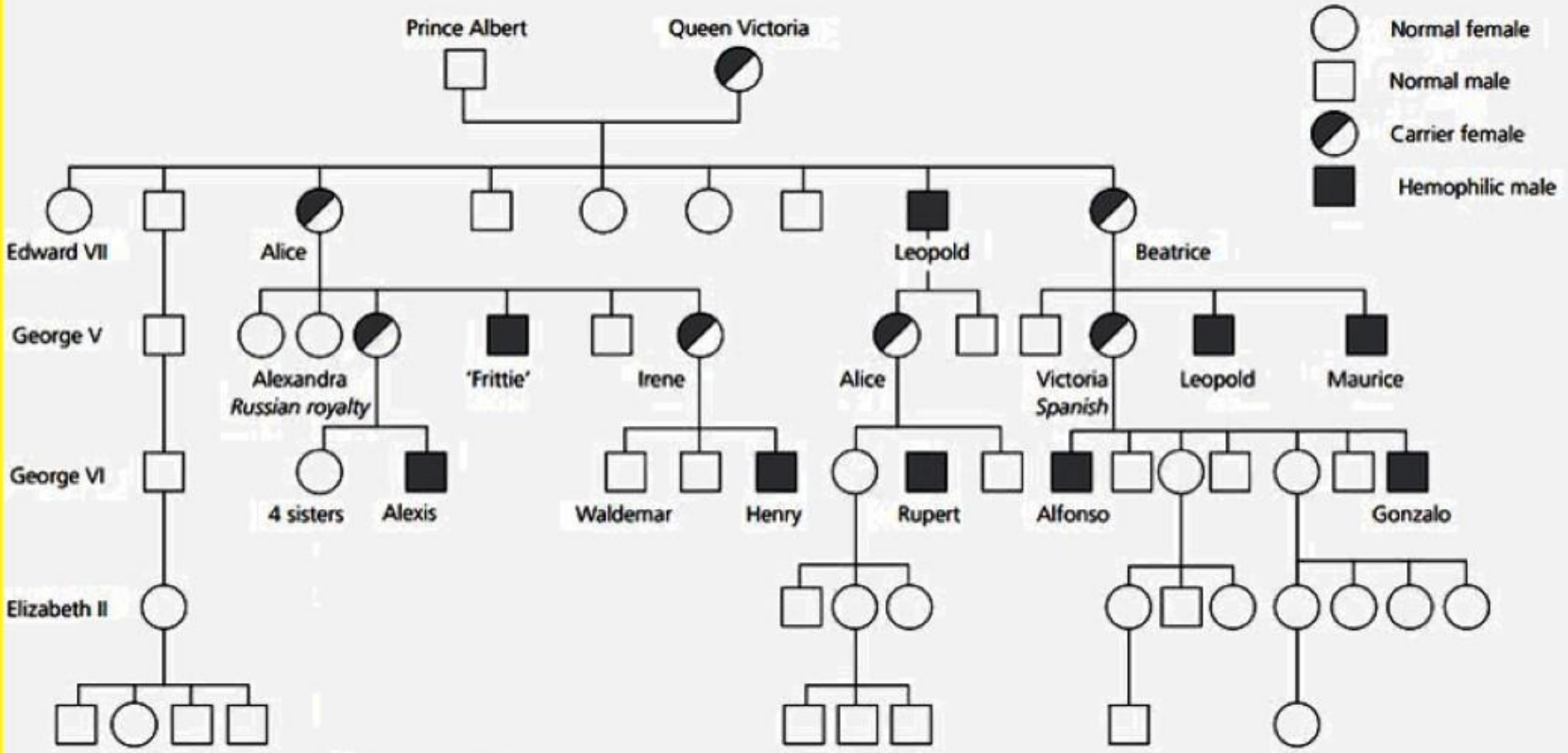
PEDIGREE ANALYSIS

Queen Victoria was a carrier of hemophilia. So her family pedigree shows many haemophilic descendants.



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