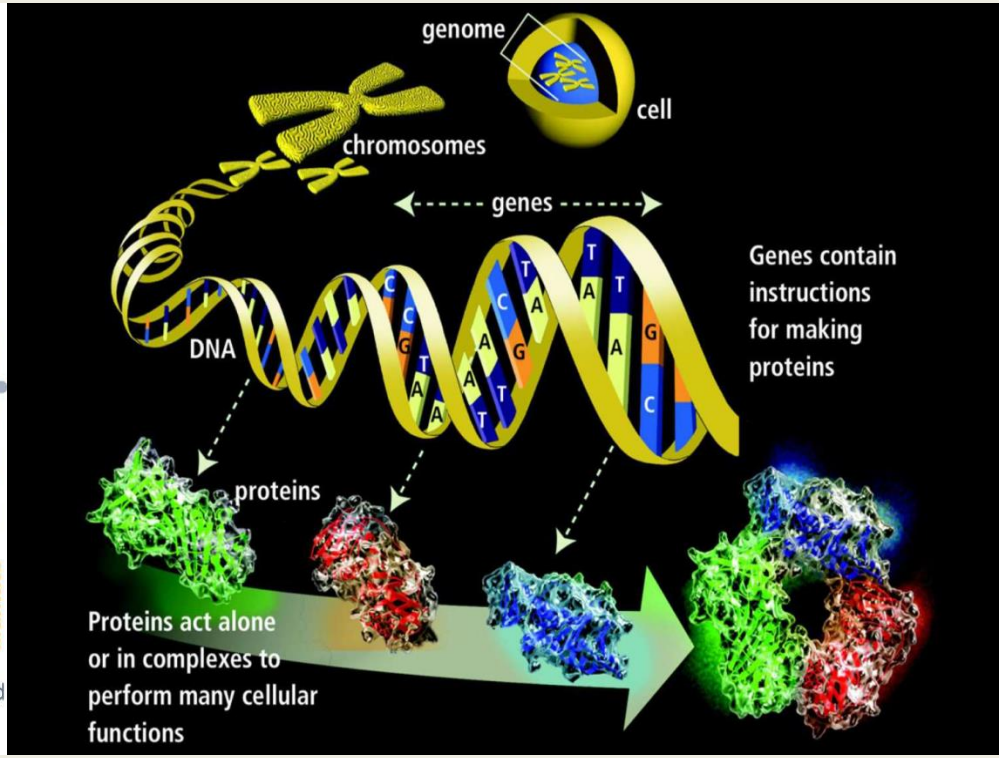
Biology Lesson Note For 11th Graders





Unit 4 - Genetics

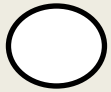
4.8 Human pedigree analysis and its importance
4.9 Genetic disorders
4.10 Genetic testing and counseling
4.11 Gene therapy
4.12 Breeding
4.13 Bioinformatics introduction

Part - III

By: Getaneh T.

4.8 Human pedigree analysis and its importance

- Genetic researchers analyze pedigrees, or family trees, to understand how human traits and diseases are inherited.
- A pedigree can show whether a trait is an autosomal dominant, autosomal recessive or X-linked trait.
- Pedigrees show relationships and identify individuals with a given trait from the history of a family.
- **In a pedigree or family tree;**

1. The square  represents the male, the circle  represents the female, and the line ——— between them represents the marriage.
2. The red are affected individuals and the blue are unaffected individuals
3. The top row of a pedigree is the original pair.
4. Two individuals who are connected by the horizontal line are breeding pairs.
5. The children of the pair are connected to them by vertical lines.
6. The next row of the pedigree shows the pair's children, as well as the partners of the children.
7. The third row of the pedigree shows the next generation, the grandchildren of the pair at the top of the pedigree).

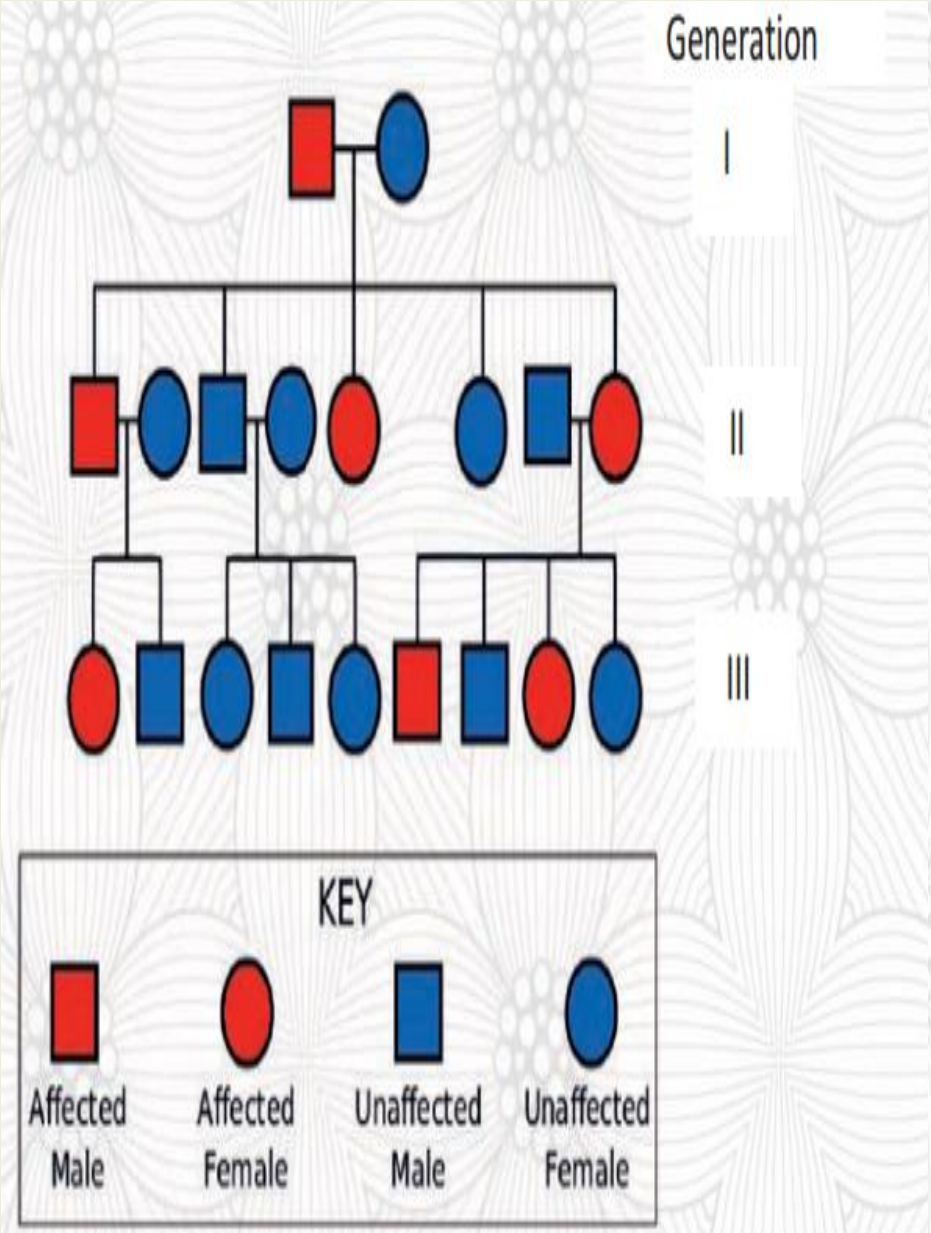


Figure 4.34 Pedigree of an autosomal dominant trait

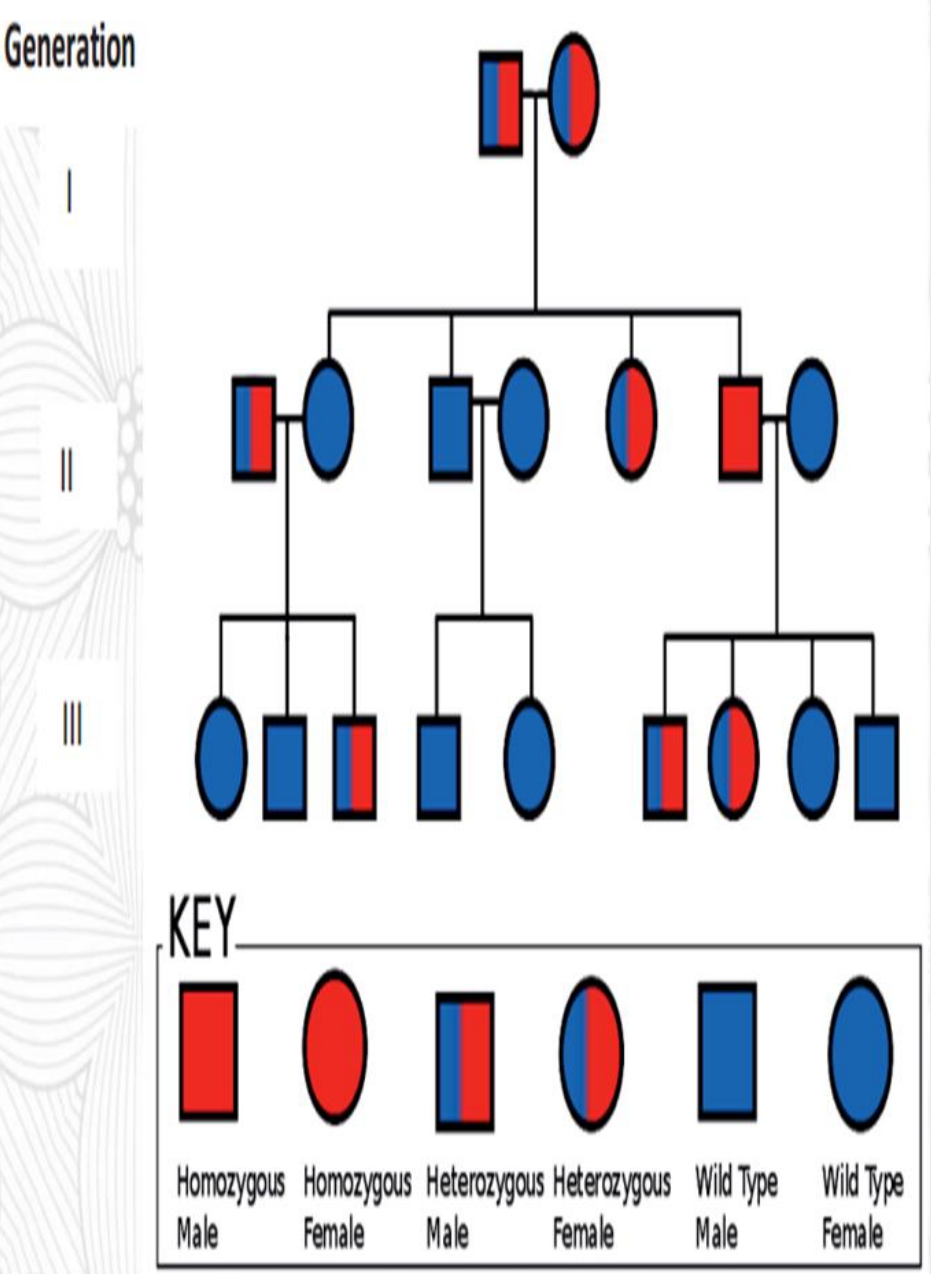


Figure 4.35 Pedigree of an autosomal recessive trait

Pedigree analysis

- Pedigree analysis can provide information about trait inheritance by examining the presence and absence of a trait throughout the history of a family.
- Pedigrees can give important clues about the risk of disease inheritance and propagation.

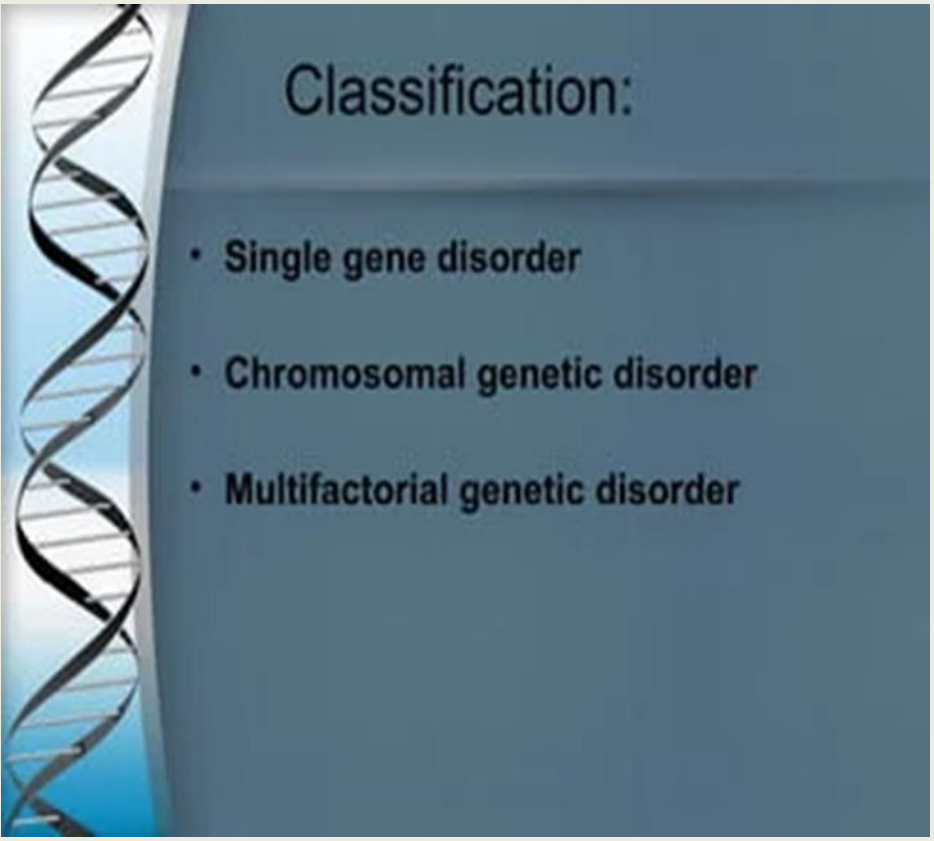
- **In general analyzing pedigrees can reveal the following:**
  1. Whether a trait is dominant or recessive
  2. The type of chromosome, autosomal or sex, a trait is linked to
  3. The genotypes of family members, and
  4. The probabilities of phenotypes in the future generations.

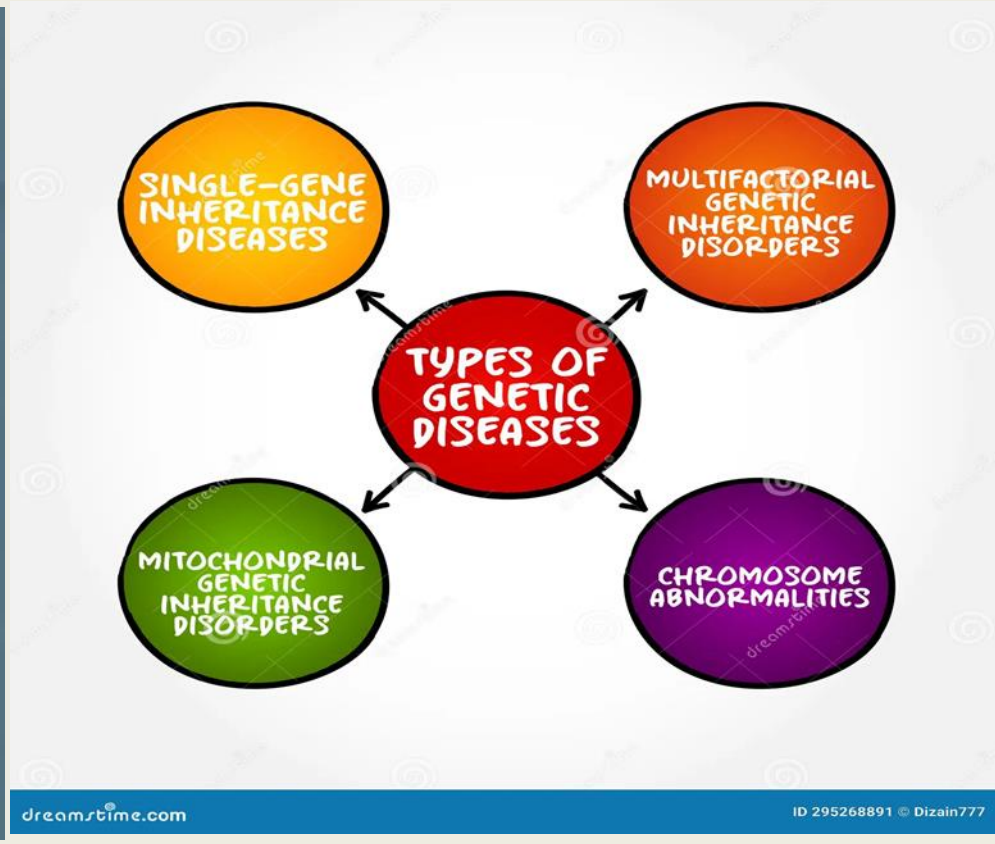
Activity

1. Find a pedigree (or draw a pedigree) that shows the transfer of **freckles**, **hemophilia** and **color blindness** in a family.

4.9 Genetic disorders

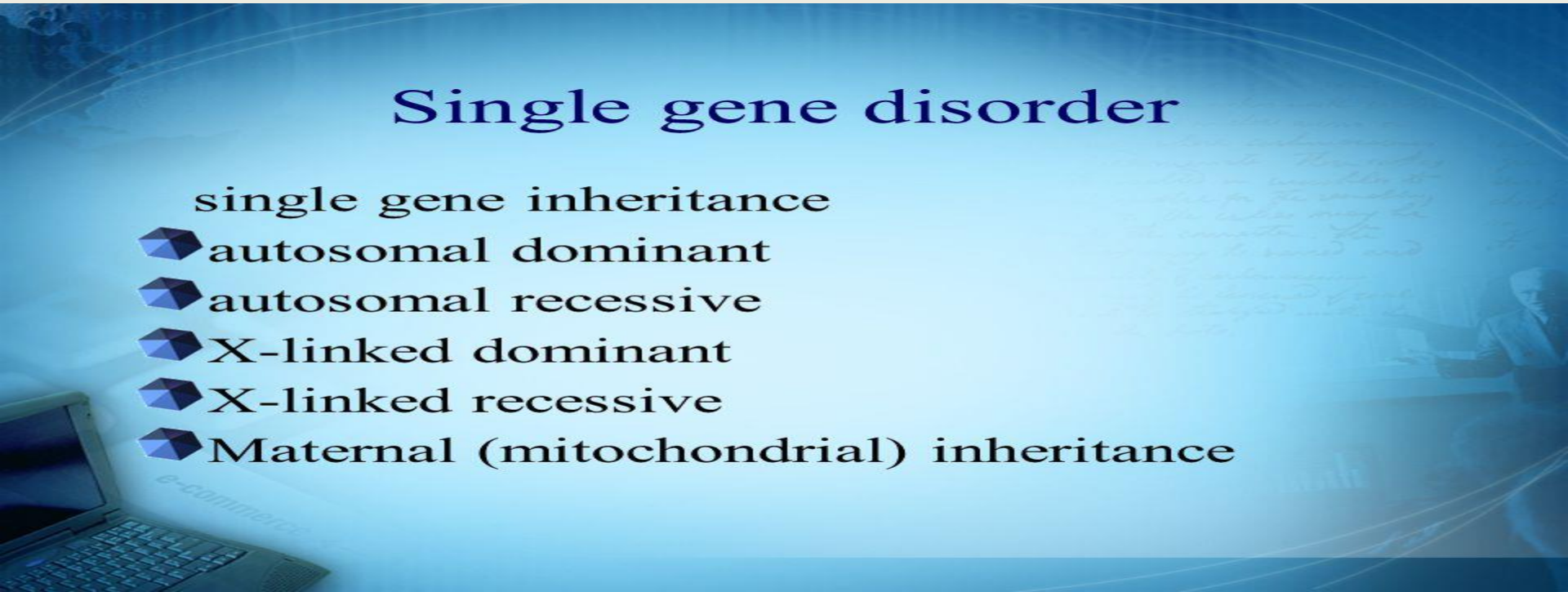
- **A genetic disorder** is a disease that is caused by a mutation in an individual's DNA.
- **Mutation** is a change in the DNA sequence of an organism due to either errors that occur during DNA replication or environmental factors**.**
- Parents pass genes on to their children, and some of these genes may contain genetic disorders.
- Some genetic disorders are carried by a dominant allele, whereas others are carried by a recessive allele.





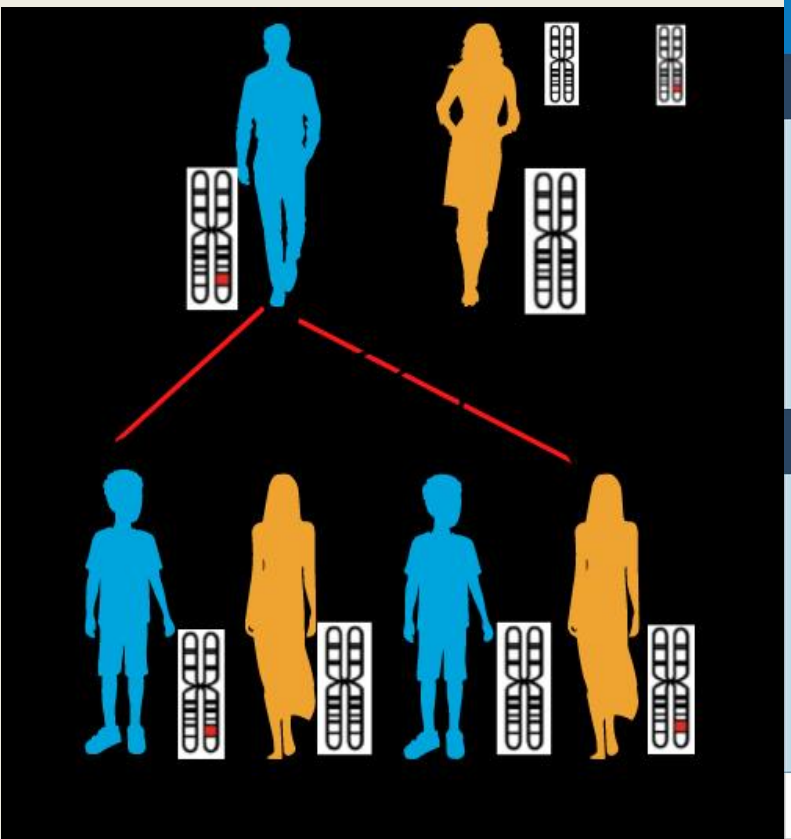
4.9.1 Single-gene disorders

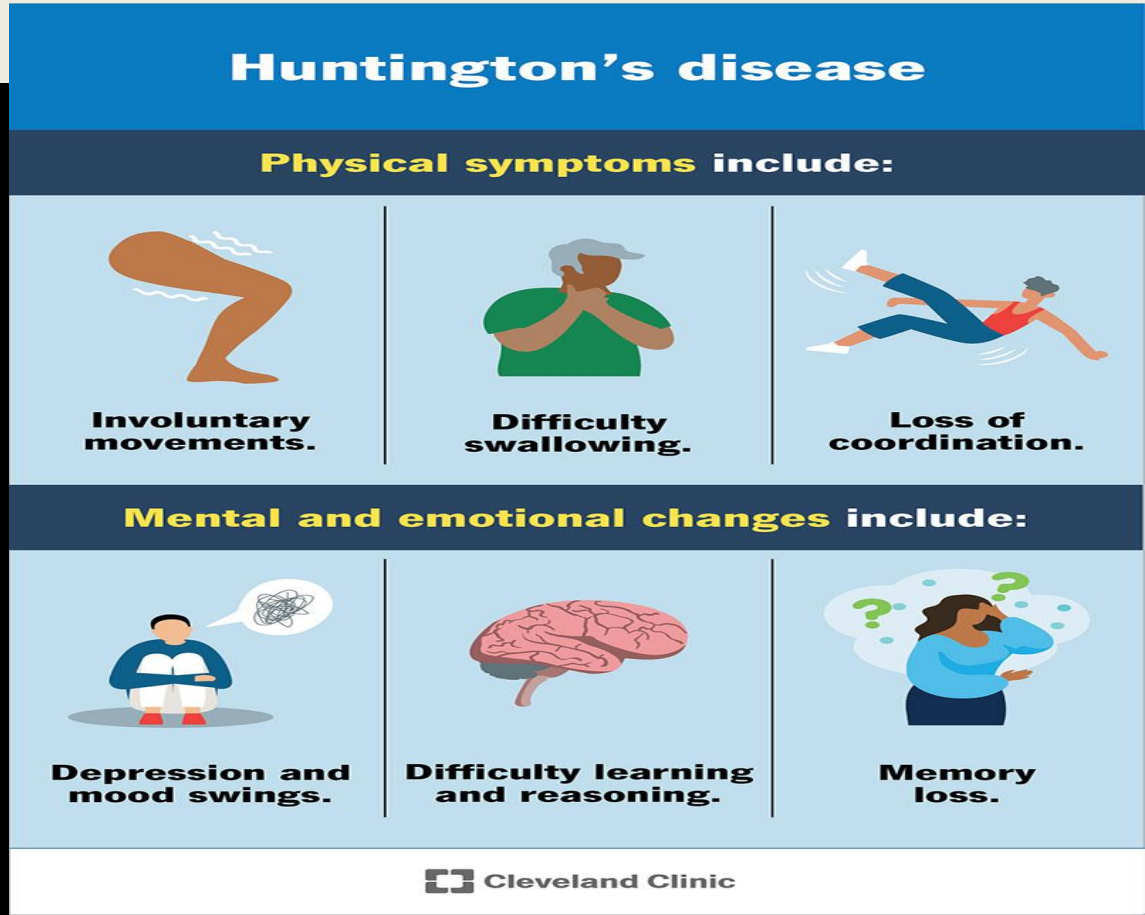
- Also known as **monogenic disorders** or **Mendelian disorders**.
- Single gene disorders are caused by defects in **one particular gene** due to changes or mutations that occur in the DNA.
- The pattern of inheritance depends on whether they are controlled by genes on autosomes or by genes on sex chromosomes.
- The three major patterns of Mendelian inheritance for genetic disorders and diseases are **autosomal dominant**, **autosomal recessive**, and **X-linked**.



A. Autosomal dominant

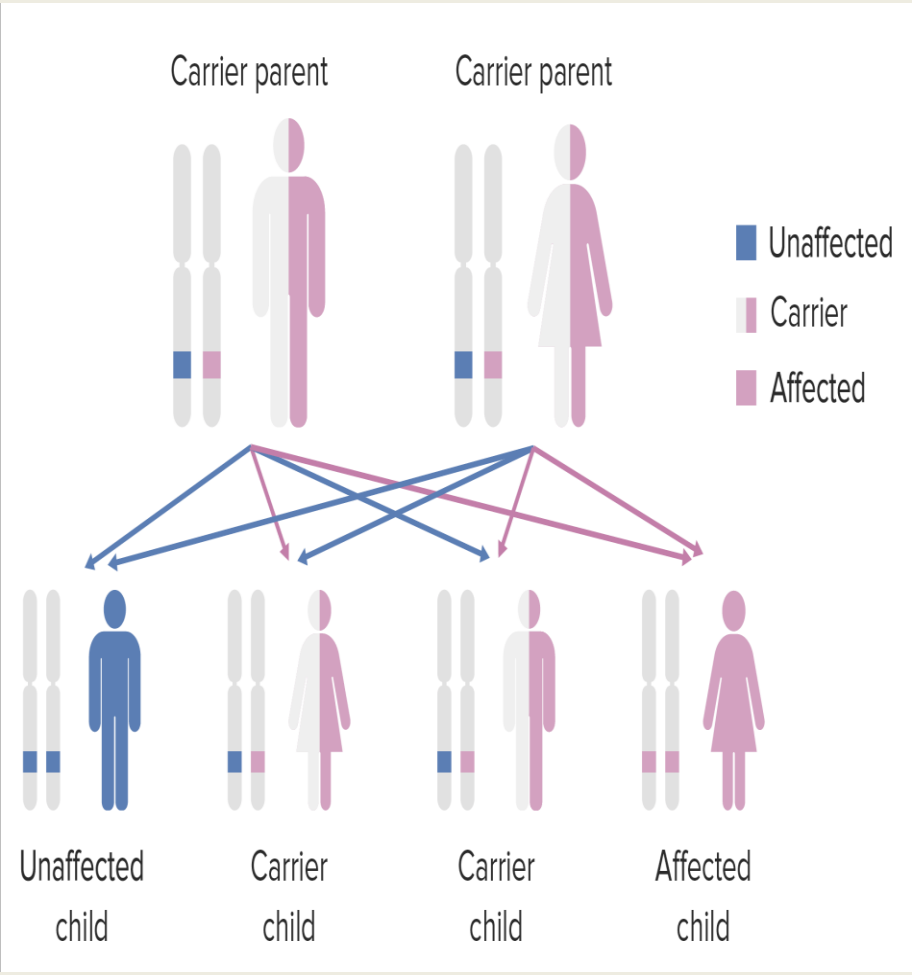
- Autosomal-dominant disorders are expressed in the **heterozygous condition**.
- Autosomal-dominant disorders are controlled by genes on **one** of human autosomes that do not differ between males and females.
- Therefore, autosomal dominant disorders are inherited in the same way regardless of the sex of the parent or offspring.
- **Huntington's disease** is a well-known example of an autosomal dominant single gene disorder**.**

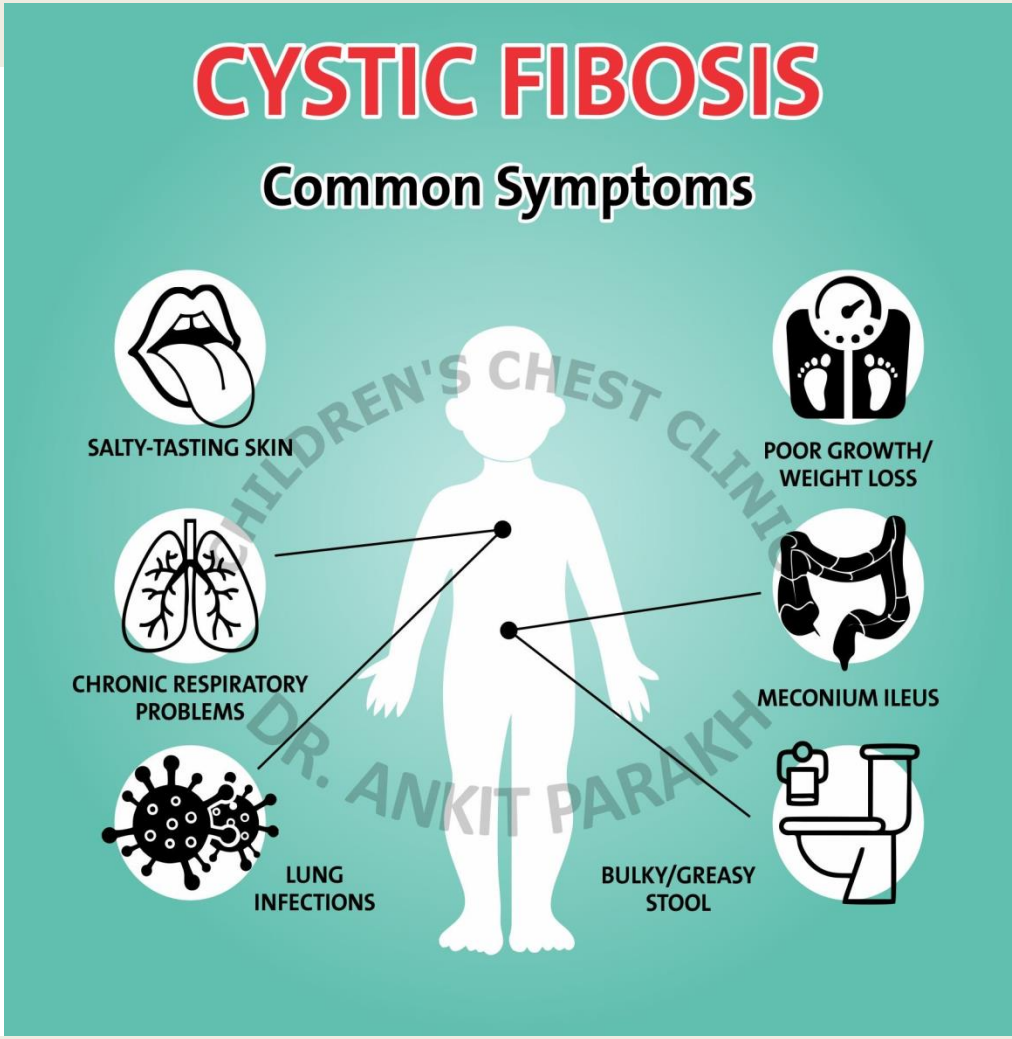




B. Autosomal recessive

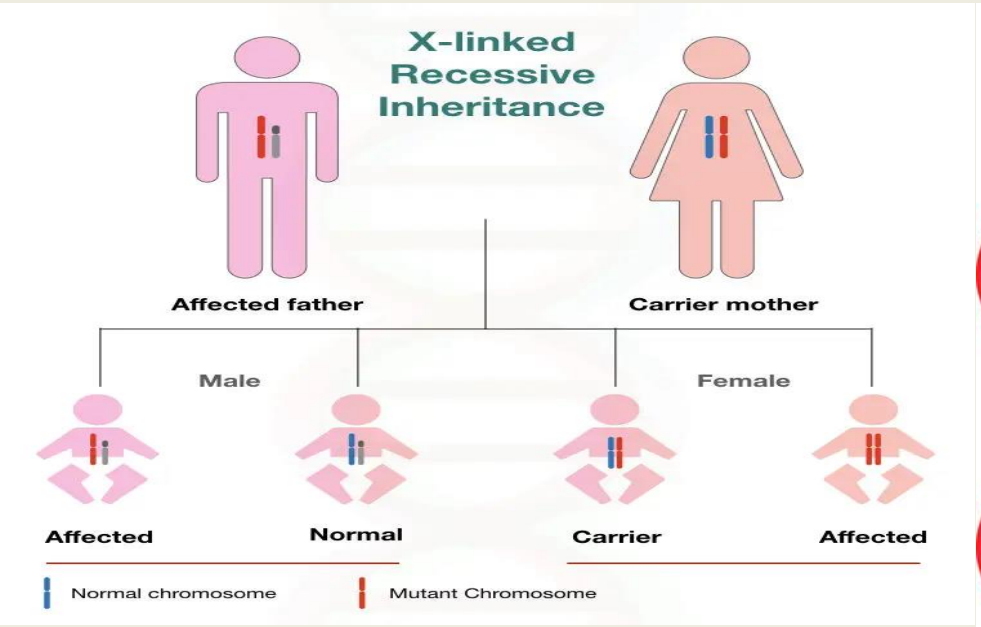
- Autosomal-recessive disorders are expressed in **homozygous conditions**.
- An autosomal recessive disorder will most commonly occur when **both parents** carry the trait and the offspring receives the defected gene from each parent.
- **Cystic fibrosis, Albinism, Sickle-cell anemia** and **Tay-Sachs disease** are examples of autosomal recessive disorders.

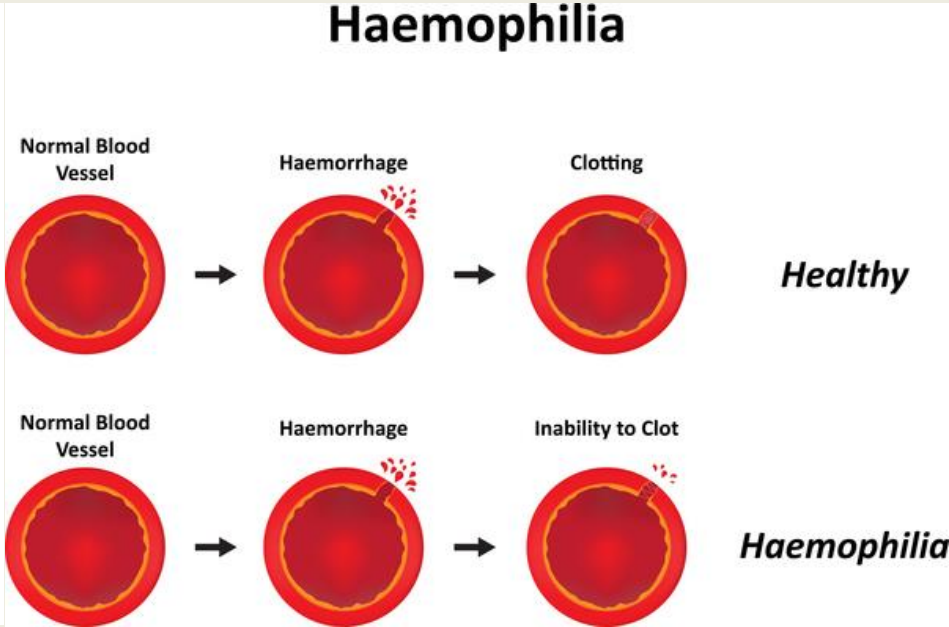




C. X-linked disorders

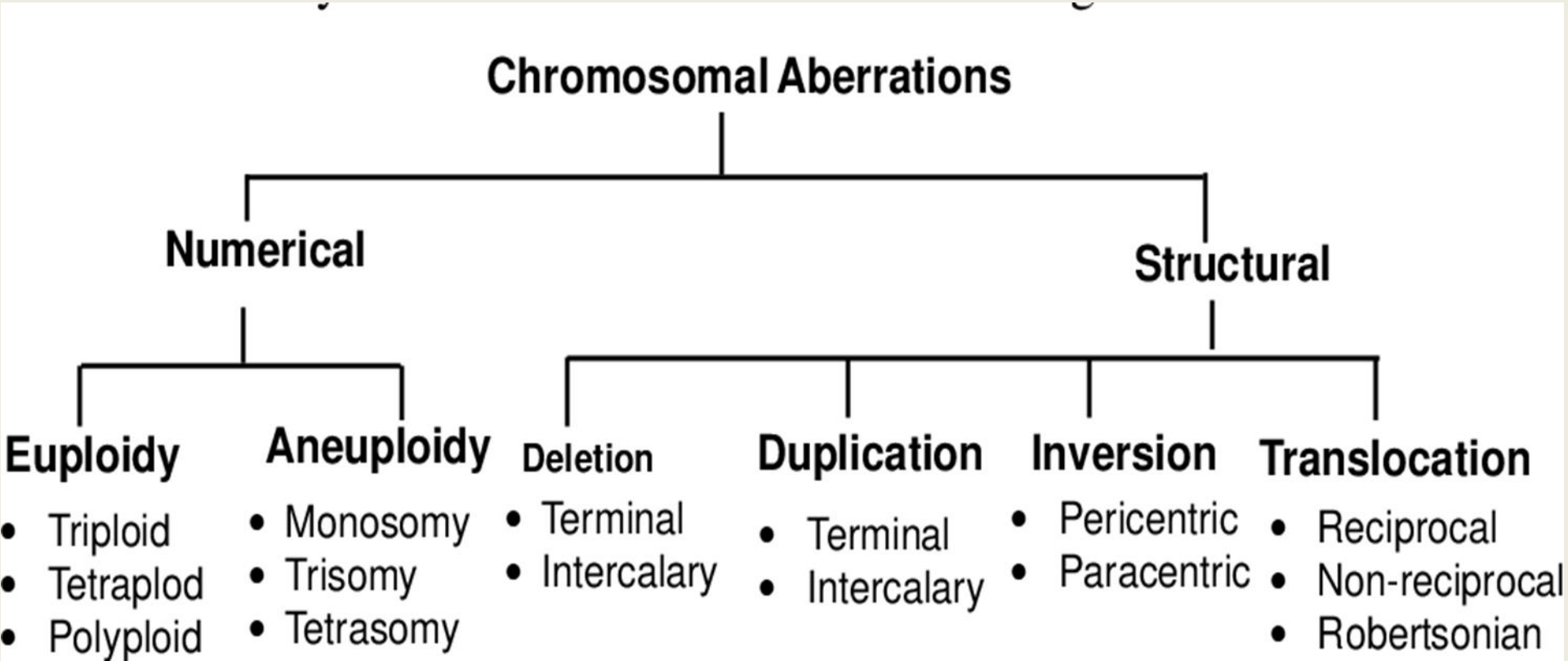
- X-linked disorders are caused by variants in genes on the X chromosome.
- X-linked disorders can be dominant or recessive.
- Females have two X chromosomes (XX) and need to inherit two copies of the alleles to be affected by recessive X-linked disorder (from both parents).
- Males have only one X chromosome (XY) and need to inherit only one copy of the allele to be affected by recessive X-linked disorder (from their mother).
- **Hemophilia** and **red-green color blindness** are examples of X-linked recessive disorders.
- Both males and females need to inherit only one copy of the allele to be affected by X-linked dominant disorder such as fragile X syndrome.





4.9.2 Chromosome disorders

- Chromosome disorders are disorders resulting from changes in the number or structure of chromosomes.
- Because chromosomes are the carriers of the genetic material, abnormalities in the chromosome number or structure can result in disease.
- Chromosomal abnormalities typically occur due to errors during cell division.
- **Down syndrome** or **trisomy 21**(2n+1), is an example of the most commonly known genetic disorder that occurs when a person has three copies of chromosome 21.



- **The most common types of chromosomal disorders occur due to the following;**
- **Aneuploidy**: loss or addition of a chromosome (2n-1, 2n-2, 2n+1, 2n+2)
- **Deletion:** a part of a chromosome is missing or removed.
- **Inversion**: occurs when a segment breaks off and reattaches within the same chromosome, but in reverse (opposite) orientation.
- **Translocation**: occurs when a segment of a chromosome breaks off and then attaches to a completely different chromosome.
-



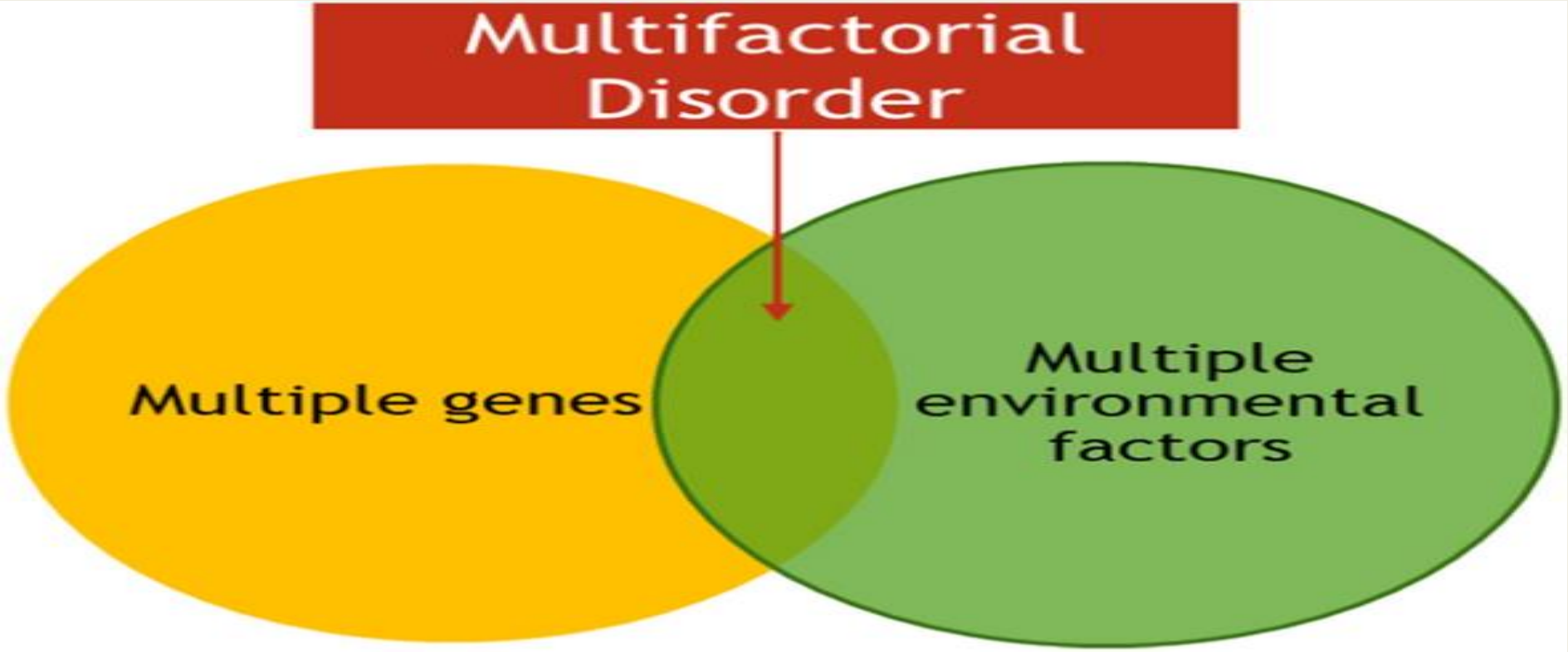
Figure 4.36 Types of changes in chromosome structure

Disorder	Cause	Chromosomes
Down Syndrome	Extra copy of chromosome 21	
Patau Syndrome	Extra copy of chromosome 13	
Edward's Syndrome	Extra copy of chromosome 18	
Triple X Syndrome	Extra X chromosome in females	
Turner Syndrome	One X chromosome missing in females	
Klinefelter Syndrome	Extra X chromosome in males	
XYY Syndrome	Extra Y chromosome in males	
Cri-du-Chat Syndrome	Deletion on chromosome 5	
Fragile X Syndrome	Duplications on X chromosome	
Acute Myelogenous Leukemia	Translocation in which part of chromosome 22 moved to 9	

4.9.3 Multifactorial disorders

- **Multifactorial disorders** are complex disorders caused by changes in the combination of multiple **genes**, complex interaction with **environmental** and **lifestyle factors** such as **smoking**, **drinking alcohol**, **eating an unhealthful diet**, **not getting enough sleep** and living in an area that has high levels of **air pollution**.

- **Examples** of multifactorial inheritance include **diabetes**, **cancer**, **Alzheimer's disease,** heart disease, epilepsy, etc…



4.10 Genetic testing and counseling

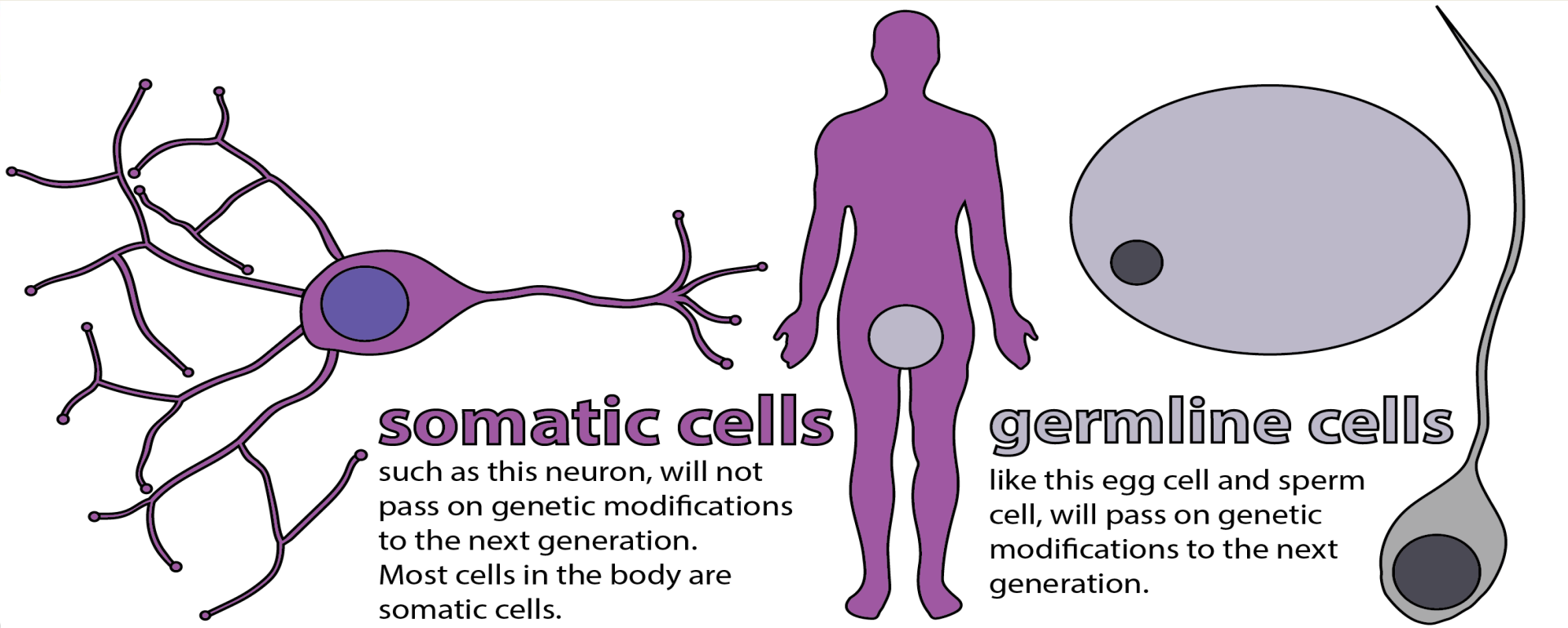
- **Genetic testing** involves examining a person's blood or other tissues to determine whether he or she has a change in his or her genetic material.
- **Genetic testing**
  - examines the genetic material and tells an individual about the likelihood and risk of passing genetic disorders on to children.
  - identifies the likelihood of parents who pass a genetic disease or disorder to their children.
  - examines if there are any change in our DNA that can inform us the well-being of our health and our family.

- **Genetic counseling** is the process of checking a family with regard to the medical history and medical records, ordering genetic tests, evaluating the results of these tests and recording, and helping parents understand and reach decisions about what to do next.
- **Genetic counselors** can help people in identifying and interpreting the risks of an inherited disorder, explaining inheritance patterns and suggesting genetic testing.

4.11 Gene therapy

- **Gene therapy** is a medical approach that involves manipulating genes within an individual's cells to treat or prevent disease.
- **There are two different types of gene therapy depending on the types of cells are treated. These are:**

1. **Somatic gene therapy** refers to transferring a section of DNA to somatic cells and the effects will not be passed onto the patient's children.
2. **Germ line gene therapy** refers to transferring a section of DNA to sex cells and the effects will be passed onto the patient's children and subsequent generations.



- **Gene therapy can be done through different mechanisms, such as by;**

1. replacing a disease-causing gene with a healthy copy of the gene
2. inactivating a disease causing gene that is not functioning properly and
3. introducing a new or modified gene into the body to help treat the disease.

- **There are different types of gene therapy products that can be used to introduce curative genes into the patient and edit the infected genes.**

a. **Plasmid DNA -** uses bacterial non-chromosomal DNA molecules to carry curative genes into human cells.
b. **Viral vectors** uses modified viruses as vectors (vehicles) to carry curative genes into human cells.
c. **Bacterial vectors** use modified bacteria as vectors (vehicles) to carry curative genes into human tissues.
d. **Human gene editing technology** is used to disrupt harmful genes or to repair mutated genes. E.g. **CRISPR**
e. **Patient-derived cellular gene therapy products** use cells removed from the patient genetically modified and then returned them to the patient.

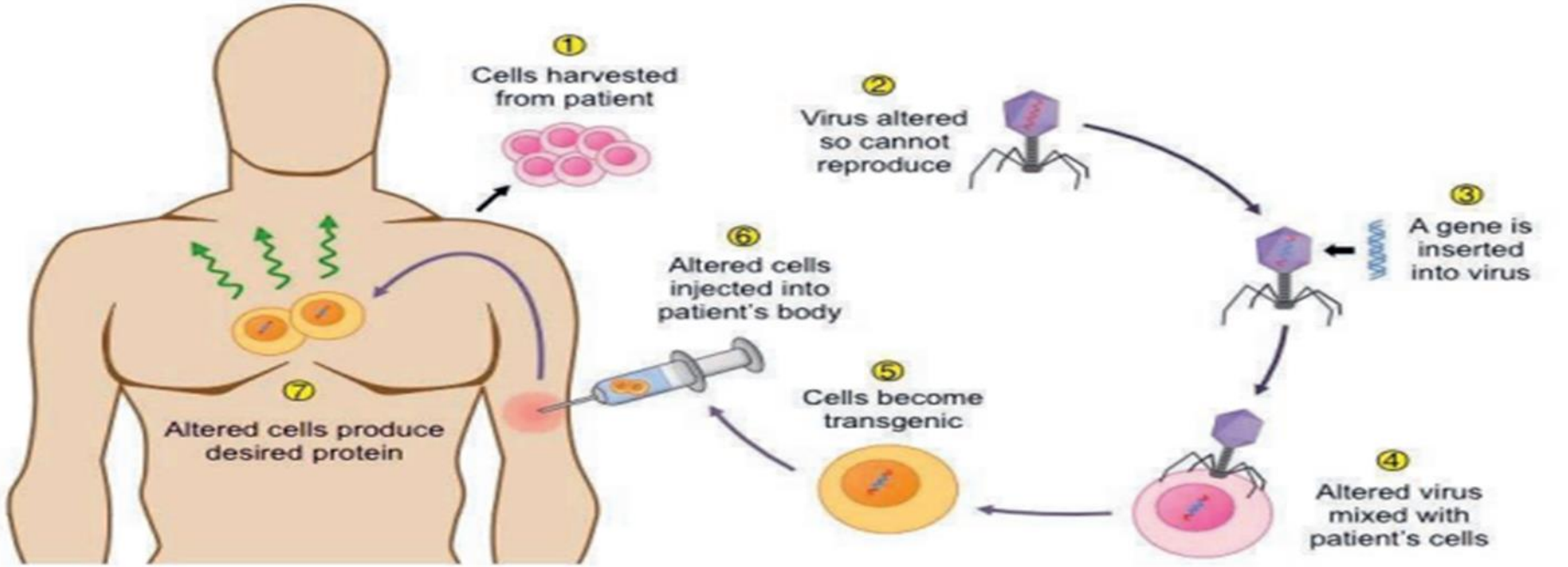


Figure 4.37 gene therapy procedures

- Gene therapy is a **promising treatment option** for a number of diseases such as cystic fibrosis, muscular dystrophy, and sickle cell anemia, as well as certain types of cancer and other acquired diseases.

- There are **challenges of gene therapy** - related to delivering the gene to the right place and switching it on, avoiding the immune response, making sure the new gene does not disrupt the function of other genes and the cost of gene therapy.

4.12 Breeding

- Breeding is the application of genetic principles in animal husbandry, agriculture, and horticulture to improve desirable qualities.
- Breeding can occur through **selective breeding/artificial selection** or **natural selection.**

Selective breeding/Artificial selection

- **Selective breeding** is the selection of individual animals or plants that show the most desirable characteristics for the next generation in the breeding program.
- It involves choosing parents with particular characteristics to breed with each other to produce offspring with more desirable characteristics.
- It utilizes the natural variations in traits that exist among members of any population.

Plant breeding

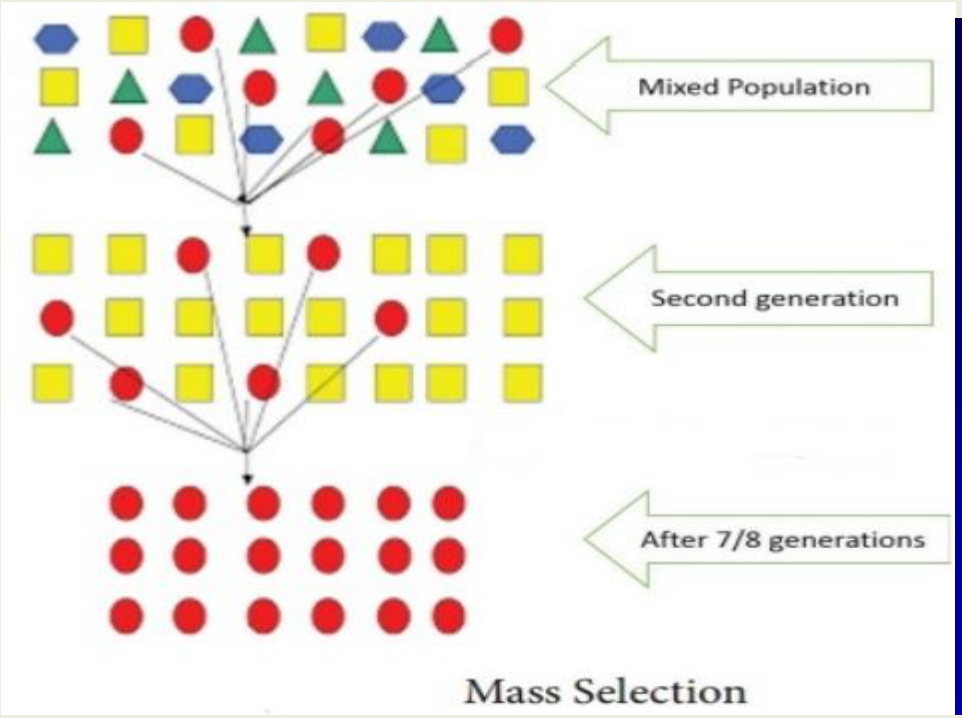
- **Plant breeding** is the science of changing the traits of plants in order to produce desired characteristics.

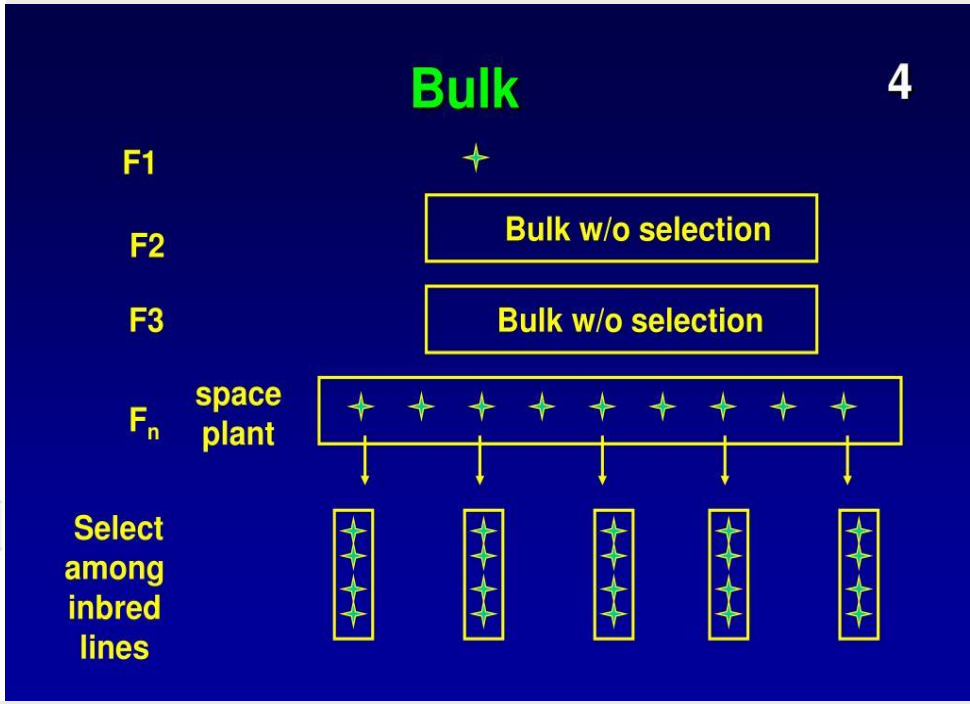
The goals of plant breeding includes;

1. Increase yield or productivity of crop plants
2. Producing disease and pest resistance plants
3. Producing environmental stress (drought) tolerance plants
4. Enhancing the quality (nutritional value, taste, texture & appearance) of plants
5. Preserving of genetic diversity within plant species
6. Developing crops that require fewer chemical inputs such as fertilizers and pesticides.

- **Selection methods (techniques) in plant breeding include;**

1. **Mass selection method:** - a large number of plants of similar phenotype are selected and their seeds are mixed together to constitute the new variety.
   - is the simplest form of plant breeding that was practiced in agriculture by early humans (inexperienced farmers).
2. **Pure line selection:** - a large number of plants are selected from self-pollinated crops and are harvested individually by which individual plant progenies from them are evaluated and best progeny is released as pure line variety.
3. **Bulk method**: - the F2 and the subsequent generations are harvested in mass or as bulk to raise the next generation and finally, individual plants are selected & evaluated.





Animal breeding

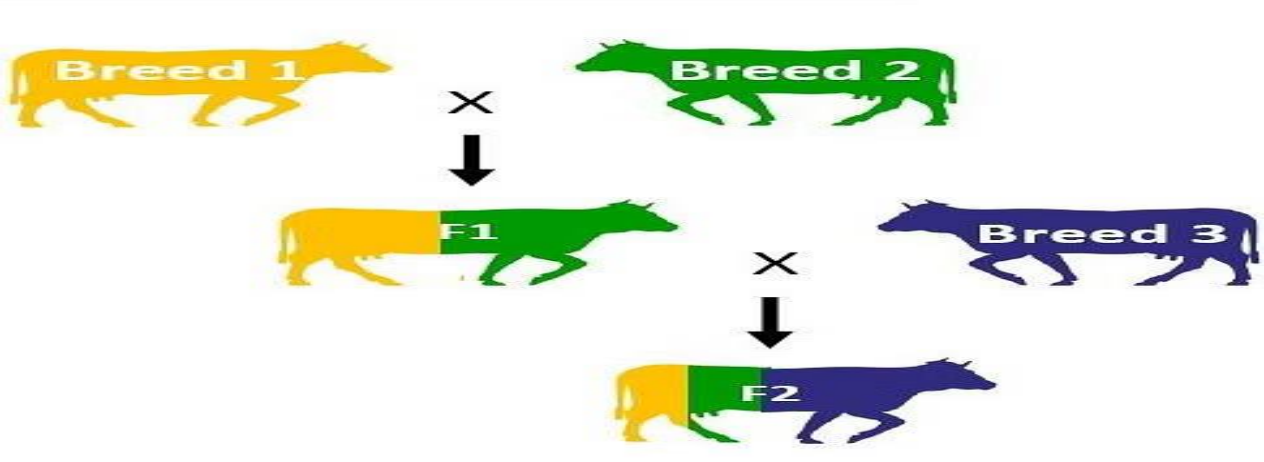
- **Animal breeding** consists of choosing the desired quality characteristics, selecting the breeding stock, and determining the breeding system.
- Humans have selectively bred plants and animals for thousands of years.
- Selective breeding involves choosing parents with particular characteristics to breed with each other to produce offspring with more desirable characteristics.
- **Types of selective breeding/artificial selection;**

1. **Inbreeding** – is the process of producing offspring through mating **genetically similar** organisms.

- After many generations of inbreeding, the offspring will be almost genetically identical and will produce identical offspring.
- Pure breeds are animals with homogeneous appearance, behavior and other characteristics.
- Inbreeding increase **homozygosity.**
- Purebred breeding aims to establish and maintain stable traits that the animal can pass on to the next generation that could help to develop superior qualities.
- The negative consequence of inbreeding is that it makes the expression of undesired recessive traits in the family more likely.

2. **Crossbreeding** - is the process of producing offspring through mating two purebred individuals that come from **different** breeds, varieties, or even species.

- Crossbreeding involves breeding two unrelated individuals. For instance, crossbreeding of the local breeds with exotic sires.
- Crossbreeding increase **hetrozygosity** (hybrid vigor).
- Crossbreeding is incompatible with the conservation of indigenous breeds.



Natural selection

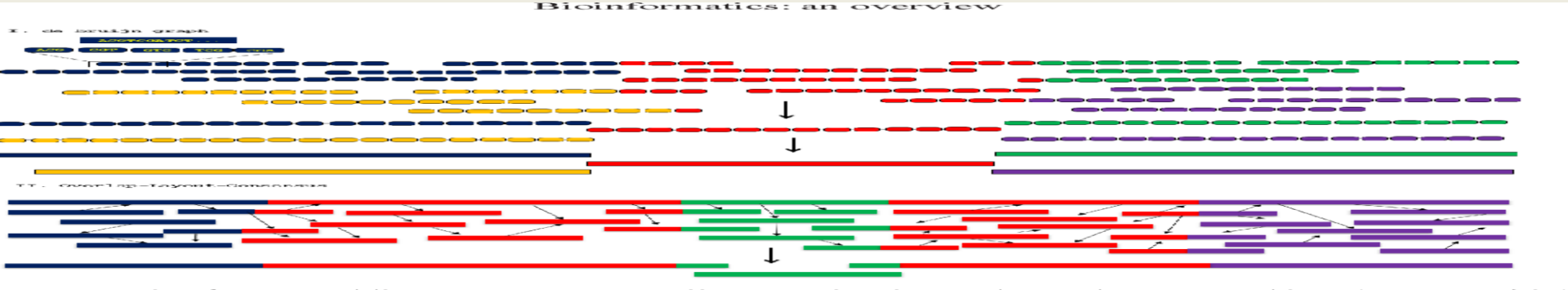
- Natural selection is the selection of certain traits without any human intervention.
- Plants and animals that are better adapted to their environment have a higher chance of survival and producing more offspring than less adapted plants and animals.

4.13 Bioinformatics introduction

- **Bioinformatics** is an interdisciplinary field that combines biology, computer science, and information technology to analyze, interpret, and manage biological data.
- particularly data related to DNA sequences, protein structures, and other bio-molecular information.
- **The field of bioinformatics incorporates three main areas:**

  1. Genomics 2. proteomics, and 3. systems biology.

- **Genomics** includes **DNA** sequence data, whereas **proteomics** deals with the function, shapes, interactions, and abundance of **proteins**.
- **Systems biology** examines the extensive role of **protein** and **DNA interactions** on the function of cells, tissues, and organs.
  - It can describe the pathway of enzymes and their various metabolites by using computer data models.
  - It can illustrate brain function by using computer images.
  - It is the most recent and complex branch in the field of bioinformatics.





“Believe you can and you are halfway there!!”