INTRODUCTION TO GENETICS

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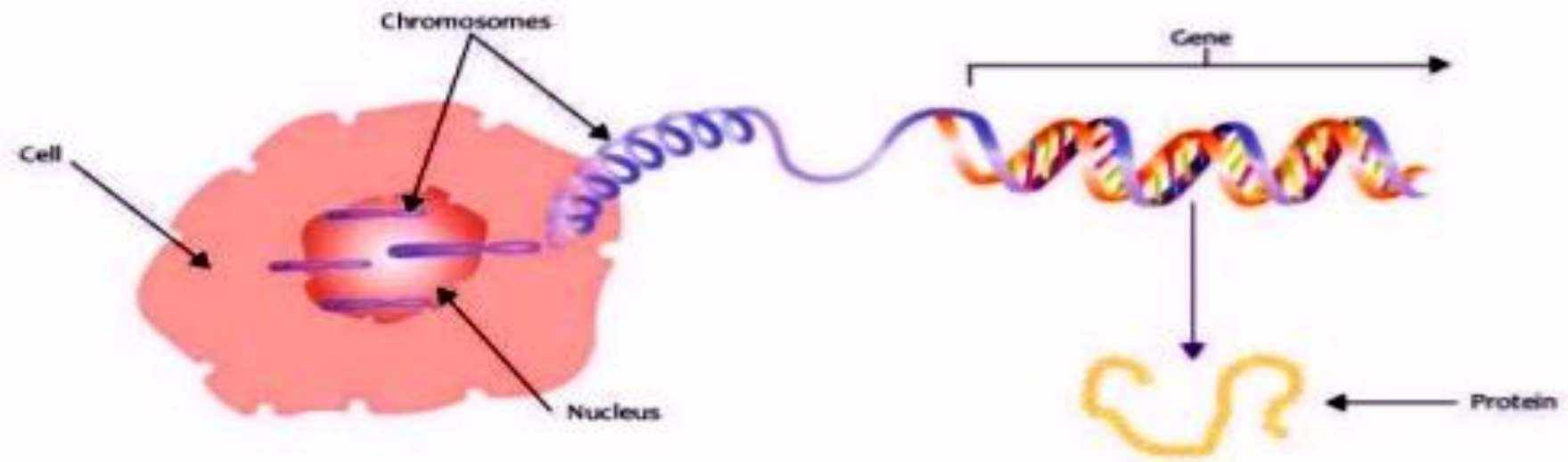
LEARNING OBJECTIVES

- Understand the role and structure of DNA, genes and chromosomes.
- Understand that proteins are encoded by genes.
- Alteration in genetic material can cause disease.
- To know some terms like alleles, mutations, genotype, phenotype, codon, Mendelian disorder.

GENETICS

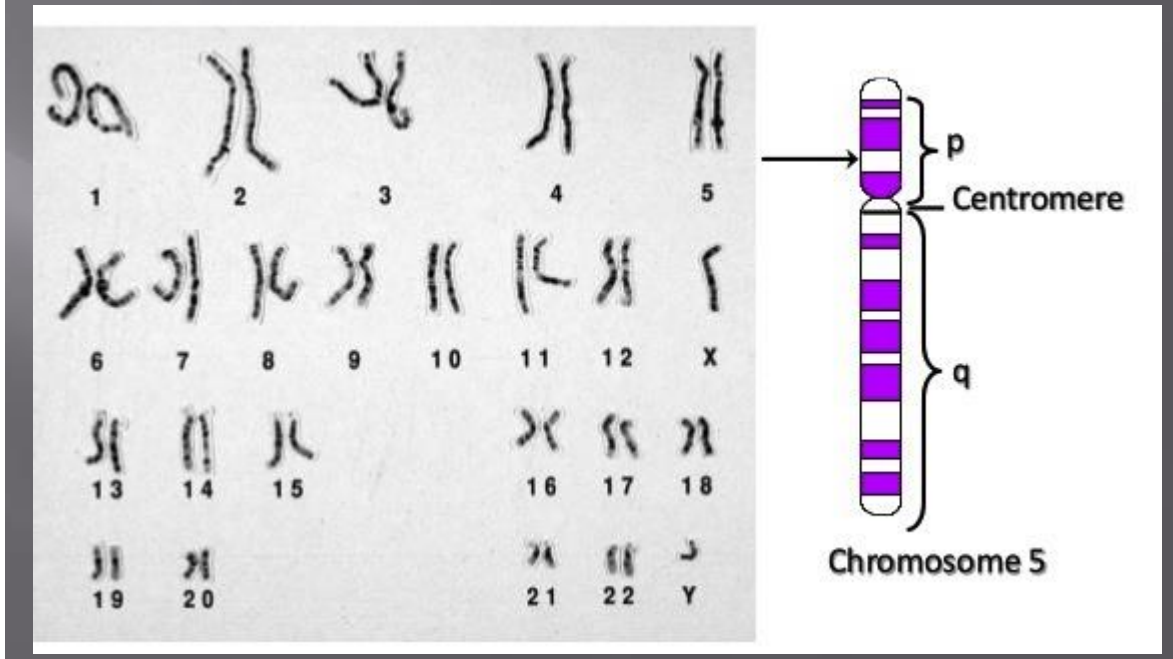
- It is the study of genes, genetic variation, and heredity in organisms.
- Gregor Mendel, a scientist working in the 19th century, was the first to study genetics scientifically.
- Genetics derives from the ancient Greek meaning "genitive"/"generative", meaning "origin

STRUCTURE OF DNA, GENES,CHROMOSOMES



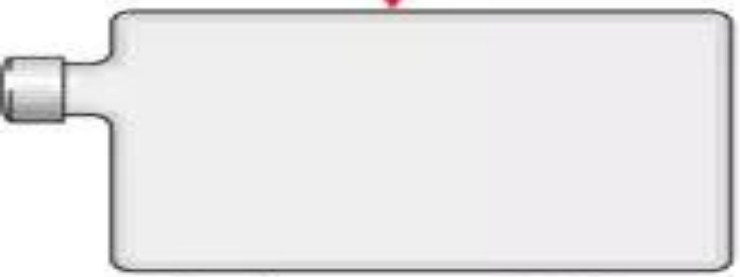
CHROMOSOMES

- Chromosomes are made of DNA.
- Each contains genes in a linear order.
- Human body cells contain 46 chromosomes in 23 pairs – one of each pair inherited from each parent
- Chromosome pairs 1 – 22 are called autosomes.
- The 23rd pair are called sex chromosomes:
  XX is female, XY is male.



5 mL venous blood

Add phytohemagglutinin
and culture medium



Culture at 37°C
for 3 days

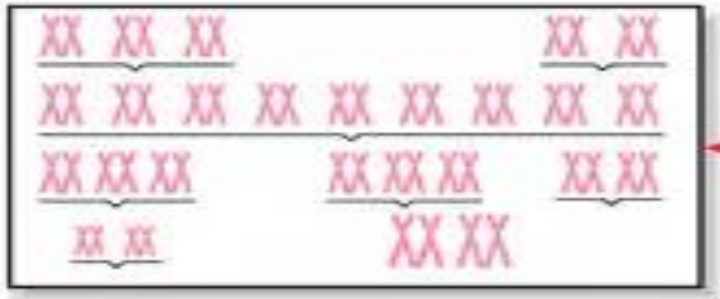
Add colchicine and
hypotonic saline

Cells fixed

Spread cells onto
slide by dropping

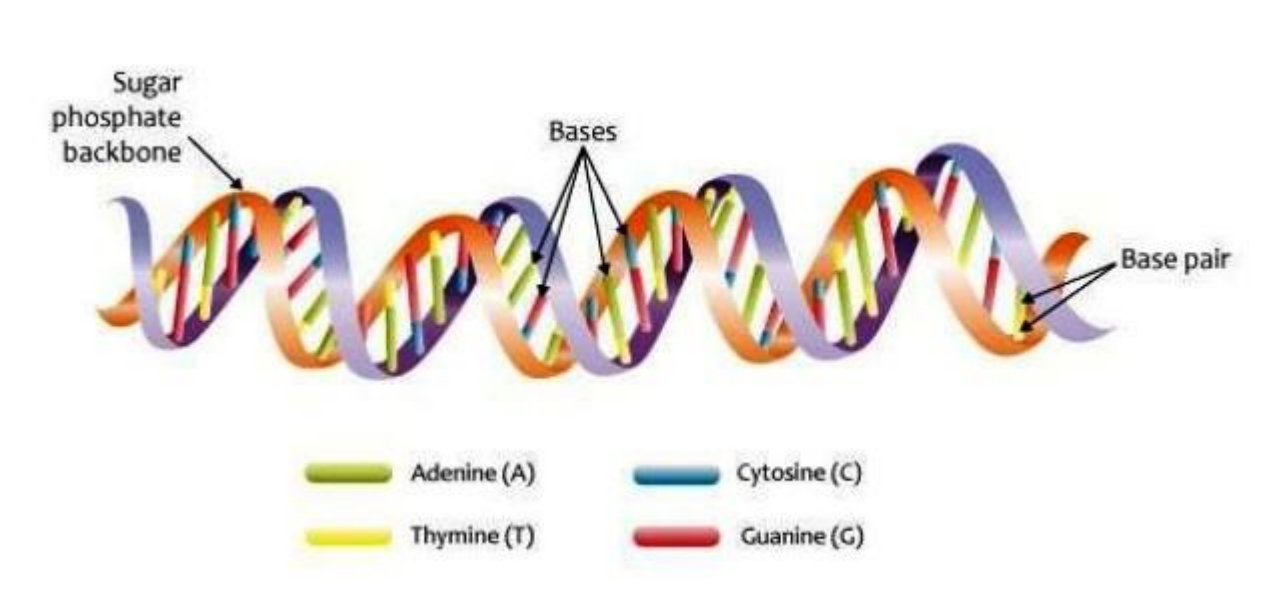
Digest with trypsin
and stain
with Giemsa

Analyze "metaphase spread



Karyotype

DNA…. Double Helix



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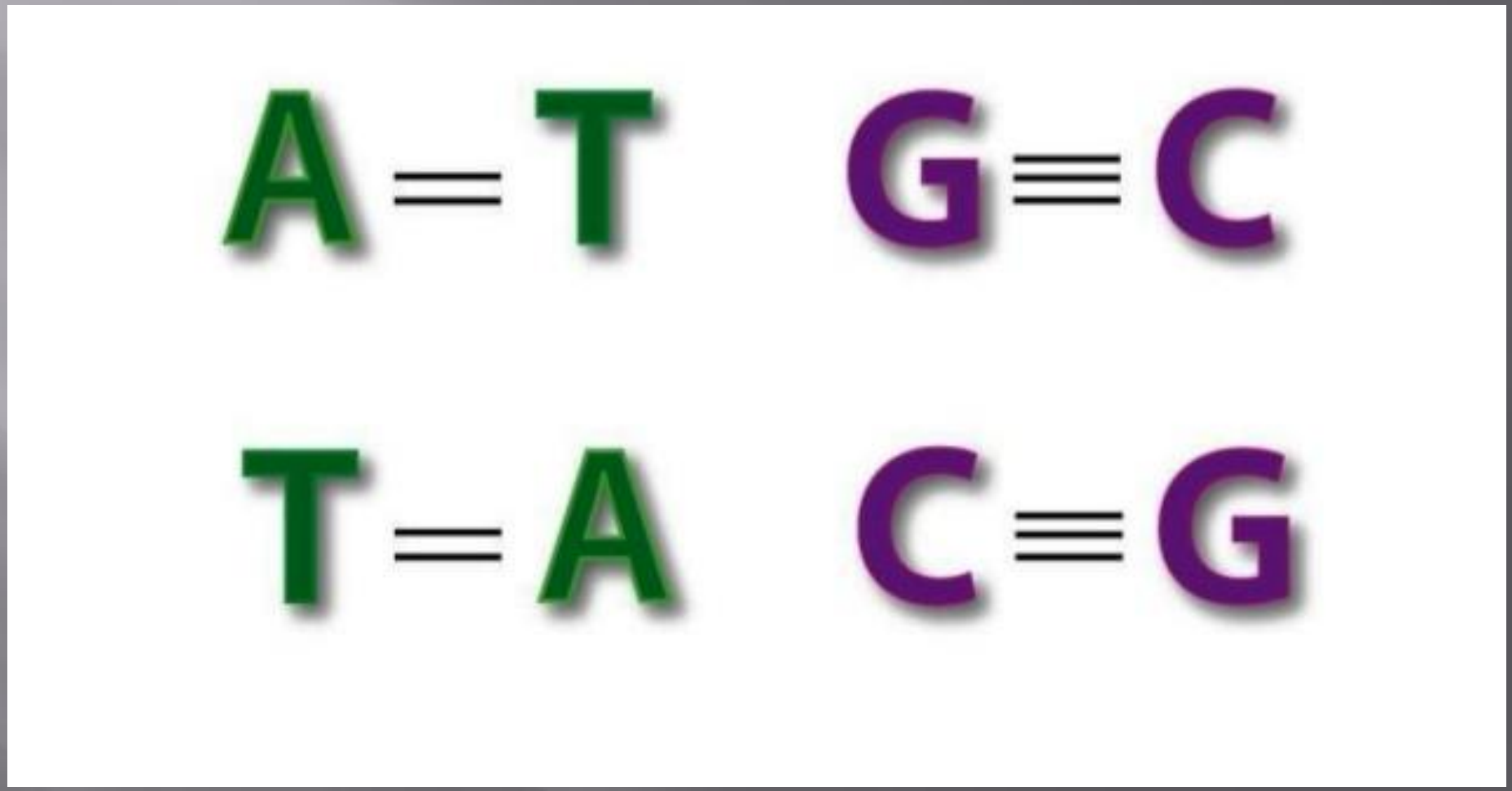
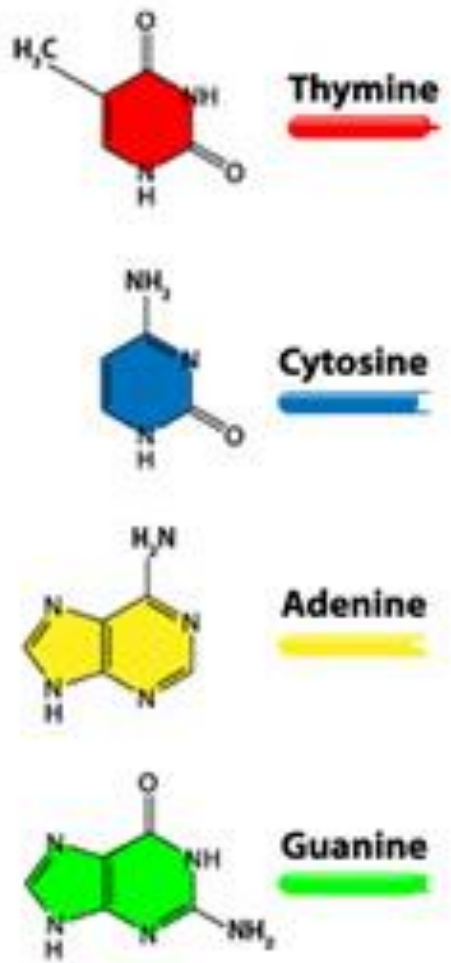
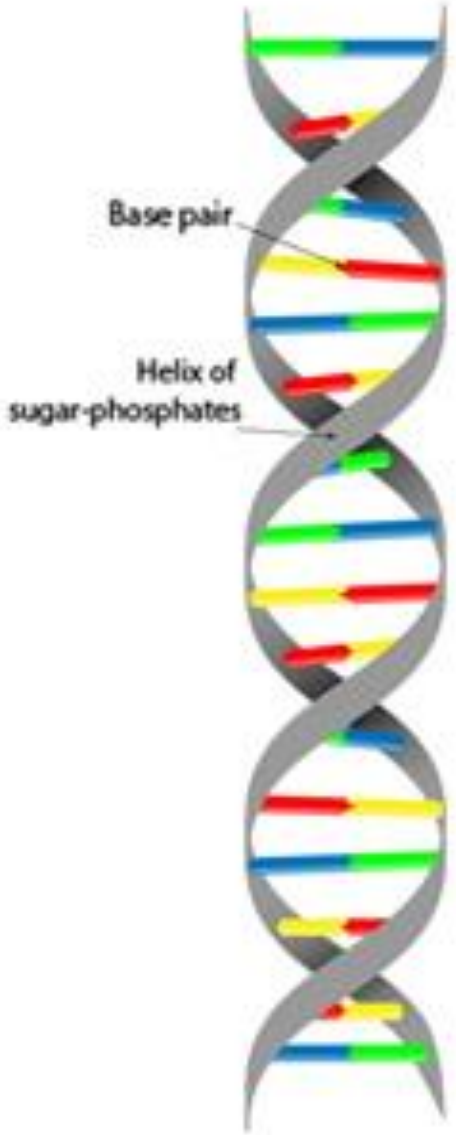


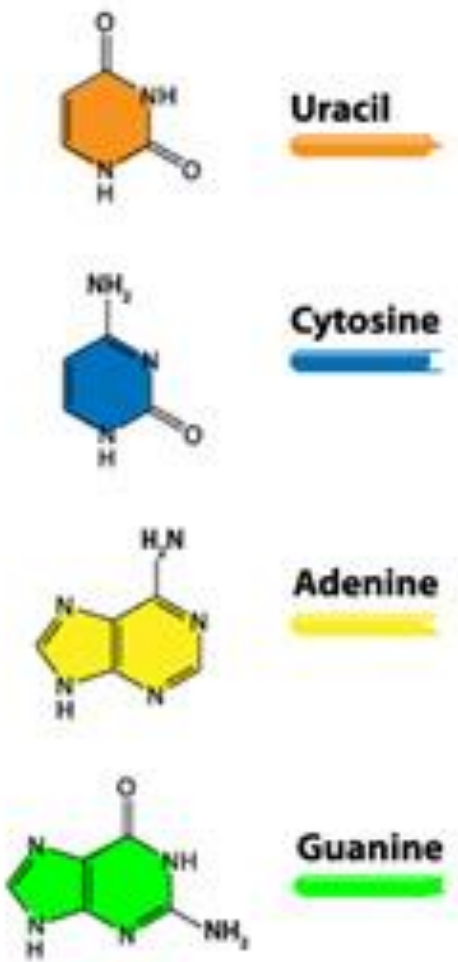


Nucelobases of DNA



DNA
Deoxyribonucleic Acid

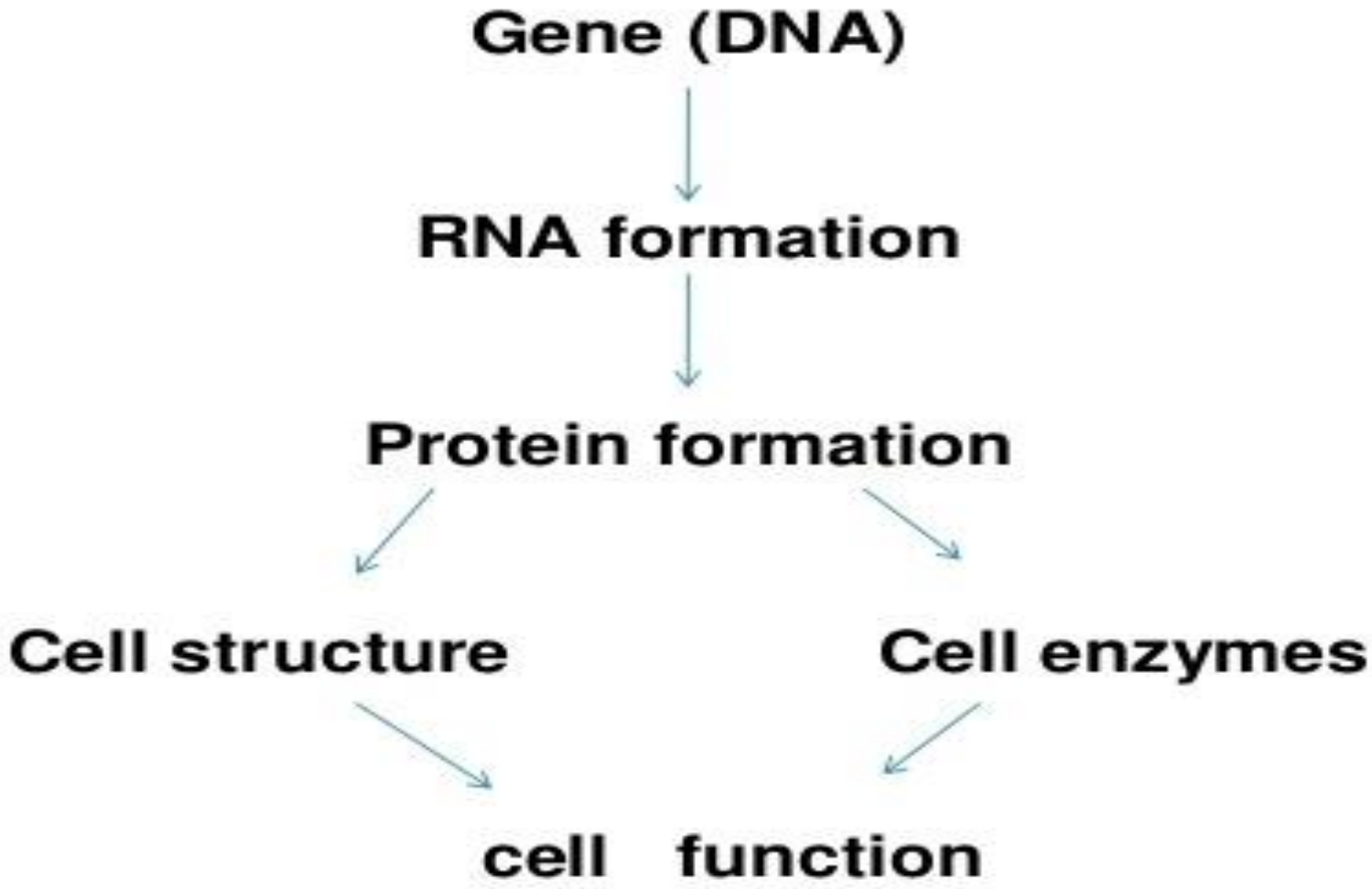
RNA
Ribonucleic Acid



Nucelobases of RNA

GENES

- **Biological unit of hereditary.**
- **Gene…. Hold the information** to build and maintain their cells and pass genetic traits to off springs.
- In cells, gene is a portion of DNA.

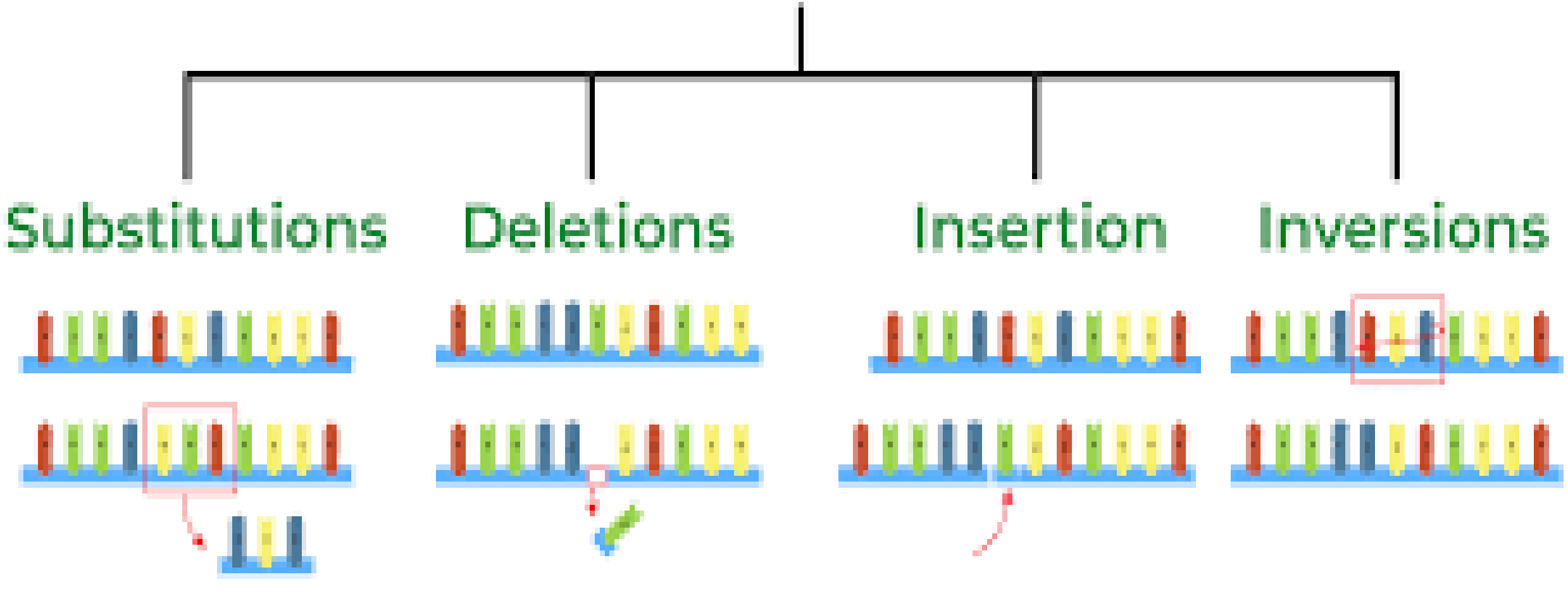

Gene (DNA)
RNA formation
Protein formation
Cell structure
Cell enzymes
cell
function

MUTATIONS

- **MUTATIONS** are the permanent changes in the DNA.
- Is the source of all genetic variation.
- **MUTATIONS**….affecting **germ cells** are transmitted to progeny…… **INHERITED DISEAESES.**
- Occurring in **somatic cells** …… are important in the genesis of cancers and congenital malformations.

Types of Mutations

(At the DNA level)



MUTATIONS

- During the DNA replication , errors occur occasionally … MUTATIONS.
- An alteration in the nucleotide sequence of the genome of an organism.
- Mutations result from errors during DNA replication, mitosis, or meiosis or other types of damage to DNA.

HEREDITARY

- The sum of all biological processes by which particular characteristics are transmitted from parents to their offspring.
- Both aspects of heredity can be explained by genes, the functional units of heritable material that are found within all living cells.

HEREDITARY

- **Inheritance** or **biological inheritance**, is the passing on of traits from parents to their offspring, either through asexual reproduction or sexual reproduction.
- The offspring cells or organisms acquire the genetic information of their parents.
- The study of heredity in biology is **GENETICS.**

CONGENITAL

- **Congenital** conditions are those present from birth. Birth defects are described as being **congenital**.
- They can be caused by a **genetic** mutation, an unfavorable environment in the uterus, or a combination of both factors.
- A **congenital** condition is not linked to **genetics**

CONGENITAL

- **Congenital** disorders are present from birth, and **hereditary** disorders are transmitted from parents to their children through the genes.
- Not all congenital diseases are genetic (congenital syphilis) and not all genetic diseases are congenital (Huntington disease)

GENOTYPE.

- Inherited traits are controlled by genes and the complete set of genes within an organism's genome is called its GENOTYPE.
- Genotype is an organism's full hereditary information.
- Genotype is inherited from an organism's parents, the phenotype is not.
- A phenotype influenced the Genotype, genotype does not equal phenotype

PHENOTYPE

- Phenotype is an organism's actual observed properties, such as morphology, development, or behavior.
- The complete set of observable traits of the structure and behavior of an organism is called its phenotype.
- These traits arise from the interaction of its genotype with the environment.
- Many aspects of an organism's phenotype are not inherited.

PHENOTYPE

- Examples of phenotypes include height, wing length, and hair color.
- Phenotypes also include observable characteristics that can be measured in the laboratory, such as levels of hormones or blood cells.

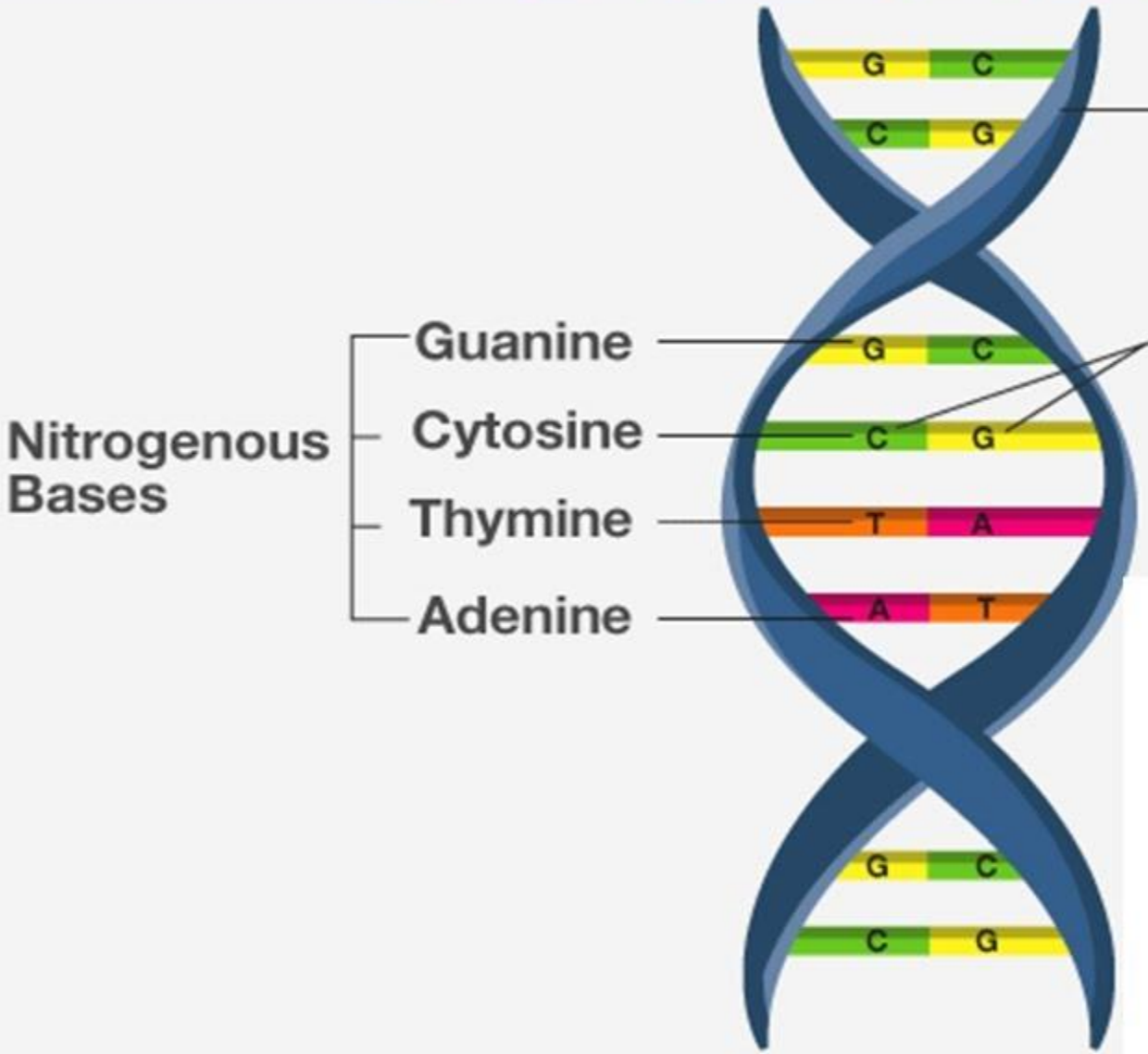
GENOTYPE VS PHENOTYPE

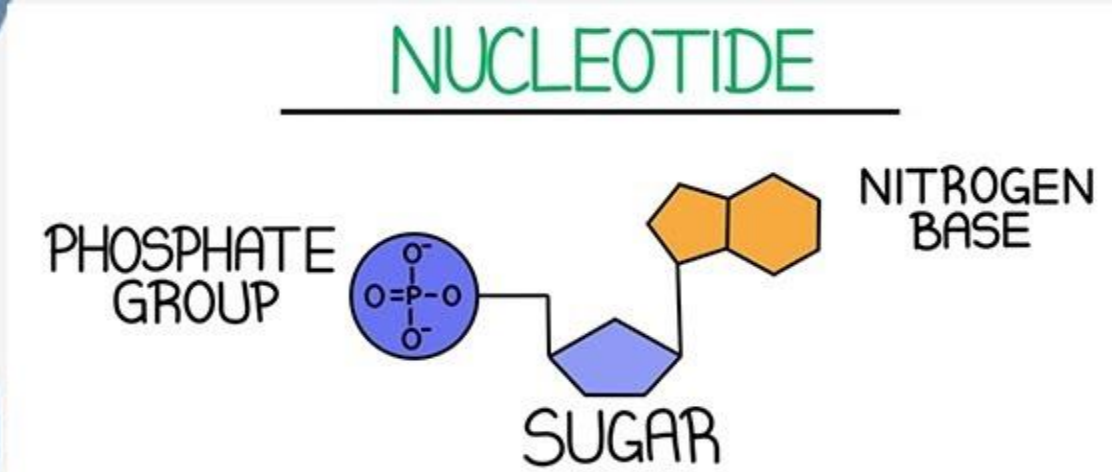
- A genotype refers to the genetic characteristics of an organism.
- A phenotype refers to the physical characteristics. For example, having blue eyes (an autosomal recessive trait) is a phenotype; lacking the gene for brown eyes is a genotype.

CODON

- A **codon** is a **trinucleotide sequence of DNA or RNA** that corresponds to a specific amino acid.
- The genetic code describes the relationship between the sequence of DNA bases (A, C, G, and T) in a gene and the corresponding protein sequence that it encodes.
- The cell reads the sequence of the gene in groups of three bases.

DNA Structure





MENDELIAN DISORDER ?

- Inheritance is in accordance to the Mendel law's of inheritance.
- Occur as a result of mutation.
- **Single gene disorder.**
- **Types:**

. Autosomal Dominant

. Autosomal Recessive

. X-linked Dominant

. X-linked Recessive

. Mitochondrial

MENDELS' LAWS OF INHERITANCE

- **1st Law : Law of dominance**

For a group of genes , the one which expresses is called Dominant while the other is recessive.

Example.

In pea plant , for the height, T (tall) is dominant gene and t (short) is recessive. Thus in condition such as Tt, T will express itself and the pea plant will be tall.

MENDELS' LAWS OF INHERITANCE

- **2nd law of segregation**:

The two members of gene pair (alleles) segregate from each other in the formation of gametes.

Half the gametes carry one allele, the other half carry the other allele

MENDELS' LAWS OF INHERITANCE

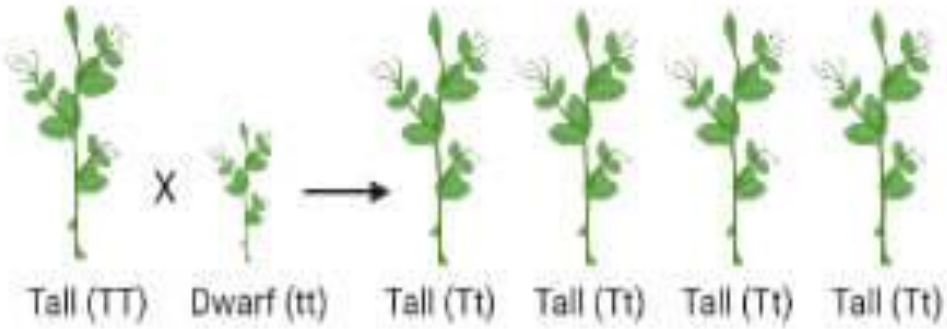
- **3rd law of independent assortment:**

  Genes for **different traits** assort independently of one another in the formation of gametes.

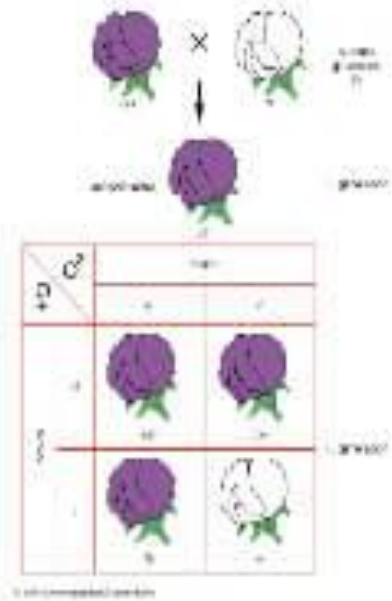
  Example:

	RRYY	**rryy**
Parental gametes	RY	ry
1st generation	RrYy (round yellow seeds)	
Self pollination	9 yellow round seeds, 3 yellow wrinkled seeds, 3 green round and 1 green wrinkled seeds	

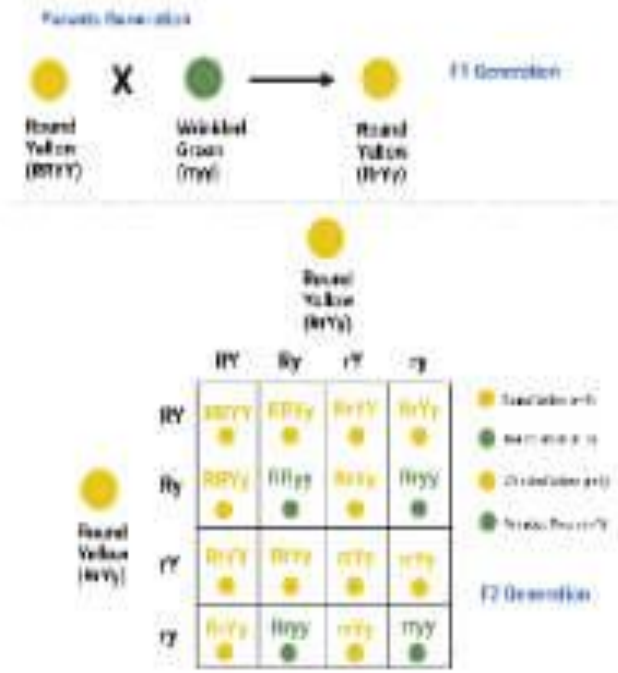
Law of Dominance



Law of Segregation



Law of Independent Assortment

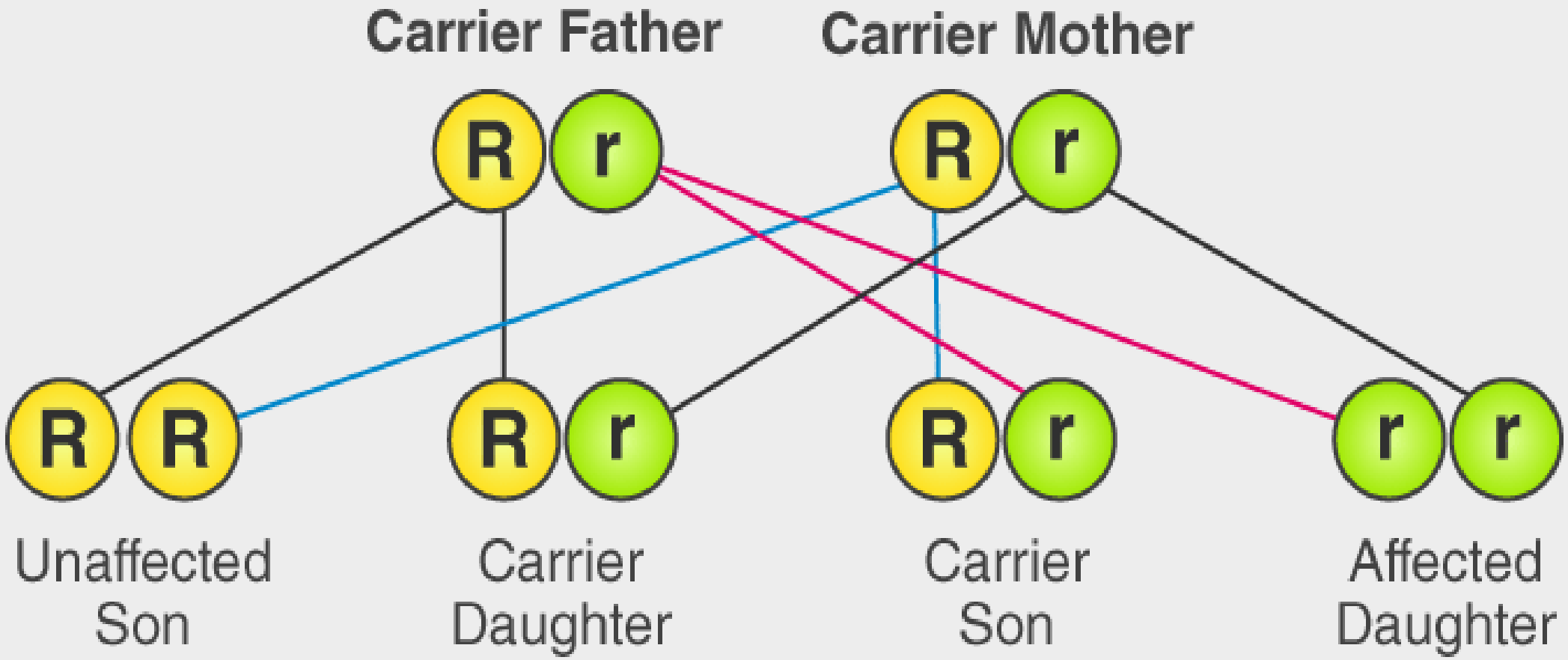


MENDELIAN DISORDER

- **Genetic disease** caused by **single gene mutation** in the structure of the DNA, causing large effects.
- Such a condition can be seen **since birth**.
- These genetic disorders are quite rare and may affect one person in every thousand or a million.
- The defect can be predicted through **pedigree analysis**.
- The disorder is transmitted to the **progeny**

MENDELIAN DISORDERS IN HUMANS

(a) Inheritance pattern of thalassemia




Thank you


Frozen bubbles in canada
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Pink lake austrailia


russia