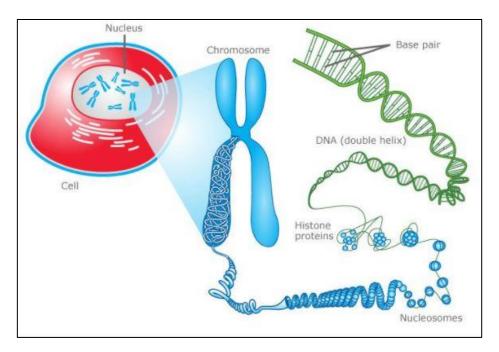
# Module 3 Genetics & Information Transfer

## • Understanding Genetics: A Bird's Eye View

Genetics is the study of how traits are inherited, and the role genes play in determining our characteristics, from physical attributes to disease susceptibility. Through his groundbreaking pea plant experiments in the 19th century, Gregor Mendel laid the foundation for modern genetics by introducing concepts like dominant and recessive traits.

Genes, segments of DNA, are the carriers of genetic information and hold the instructions for building and maintaining an organism. The field of genetics has far-reaching implications, influencing advancements in medicine, agriculture, biotechnology, and even our understanding of human history and evolution.



DNA, the molecule of life, serves as the intricate code that dictates everything about an organism, from its appearance to its functioning. Genetic engineering, made possible through our understanding of genetics, has revolutionized industries by allowing us to modify genes for various purposes, from creating disease-resistant crops to developing cutting-edge medical treatments.

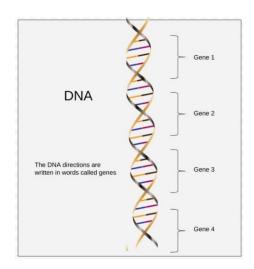
Genetics is not static but a constantly evolving field, continually uncovering new insights into heredity, genetic variation, and the complex web of genes and traits that make up life as we know it.

## What is genetics?

Genetics is the scientific study of heredity, exploring how genetic information is passed from generation to generation. It delves into the mechanisms that determine an organism's traits

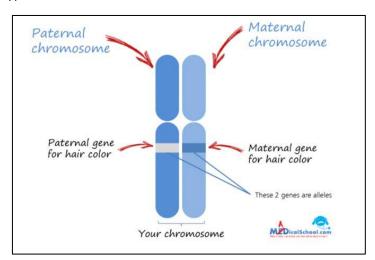
and characteristics, be it the color of a flower's petals or the predisposition to certain diseases in humans. At the core of genetics lies the understanding of genes, segments of DNA that carry this vital information.

Through a complex yet fascinating process, genes are inherited from parents, shaping the unique features of offspring. This intergenerational transfer of genetic material is central to the perpetuation of life on Earth and plays a pivotal role in the diversity and continuity of species.



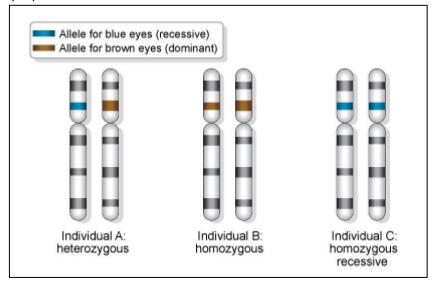
#### #Terminology

- **Genes:** Genes are the units of heredity that determine specific traits in living organisms. For example, the gene for eye color in humans can have variations like blue, brown, or green.
- **DNA (Deoxyribonucleic Acid):** DNA is the molecule that carries genetic information in all living cells. An example is the DNA double helix structure discovered by James Watson and Francis Crick.
- Allele: Alleles are different forms of a gene that can produce variations in a trait. In humans, the gene for blood type has alleles A, B, and O.

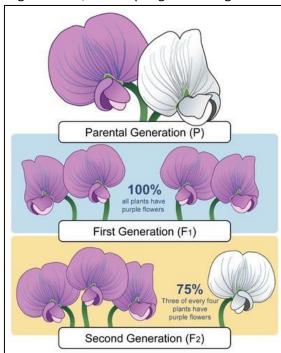


- Homozygous Dominant: Homozygous dominant refers to an individual with two identical dominant alleles for a specific trait. For instance, an individual with two dominant alleles for freckles (FF) will express freckles.
- Homozygous Recessive: Homozygous recessive individuals have two identical recessive alleles for
  a trait. In Mendel's pea plant experiments, a homozygous recessive plant for flower color would
  be "pp" for purple flowers.

• **Heterozygous:** Heterozygous individuals have one dominant and one recessive allele for a particular trait. For example, a heterozygous plant for flower color in Mendel's experiments would have "Pp" for purple flowers.



- Monohybrid Cross: A monohybrid cross involves the study of one trait in a genetic cross. For
  example, when Mendel crossed two pea plants differing in flower color (purple and white), it was
  a monohybrid cross.
- **Dihybrid Cross:** A dihybrid cross studies two different traits simultaneously. Mendel's experiments with pea plants, considering both flower color and seed texture, represent a dihybrid cross.
- P1, F1, and F2 Generation: "P1" stands for the parental generation, the starting generation of a genetic cross. "F1" represents the first filial generation, the offspring of the P1 generation. "F2" represents the second filial generation, the offspring of the F1 generation.



• **Genotypic Ratio:** The genotypic ratio shows the ratio of different genetic combinations in offspring. In a monohybrid cross, the genotypic ratio for a heterozygous dominant parent crossed

with a homozygous recessive parent is 1:1, meaning there's a 1/4 chance of homozygous dominant (HH), a 1/2 chance of heterozygous (Hh), and a 1/4 chance of homozygous recessive (hh) offspring.

• **Phenotypic:** Phenotypic traits are the observable physical characteristics of an organism, resulting from the interaction of genes with the environment.

For example, in a dihybrid cross between pea plants with yellow, round seeds (YYRR) and green, wrinkled seeds (yyrr), the phenotypic ratio of the F2 generation might be 9:3:3:1 for yellow-round, yellow-wrinkled, green-round, and green-wrinkled seeds, respectively.

#### - Punnett Square: Unraveling Genetic Inheritance

**Origin:** The Punnett Square technique, named after British geneticist Reginald Punnett, was developed in the early 20<sup>th</sup> century. Reginald Punnett, along with his colleague William Bateson, revolutionized the study of genetics with this graphical tool.

**Significance:** The Punnett Square is a powerful tool in genetics, designed to predict the possible genotypes and phenotypes of offspring when two individuals with known genotypes are crossed. It simplifies complex genetic calculations, making it accessible to students and researchers alike.

# **Applications:**

- 1. <u>Understanding Inheritance:</u> The Punnett Square is a fundamental tool for comprehending the principles of inheritance. It helps visualize the likelihood of traits being passed from parents to offspring.
- 2. <u>Plant and Animal Breeding:</u> Breeders employ Punnett Squares to predict the outcomes of controlled crosses, enhancing the breeding of plants and animals with desired traits, such as crop yield or disease resistance.
- 3. <u>Medical Genetics:</u> In the field of medical genetics, Punnett Squares aid in assessing the risk of genetic disorders within families and understanding how genetic diseases can be inherited.

**Example:** Consider a simple Punnett Square scenario involving flower color in pea plants. Let's say we have two pea plants, one with purple flowers (PP) and another with white flowers (pp). We want to predict the flower color of their offspring.

## 1. Parental Genotypes:

Parent 1: Purple flowers (PP)

Parent 2: White flowers (pp)

### 2. Create the Punnett Square:

X	Р	р
Р	PP	Рр
р	Рр	рр

## 3. Analyze the Offspring Genotypes:

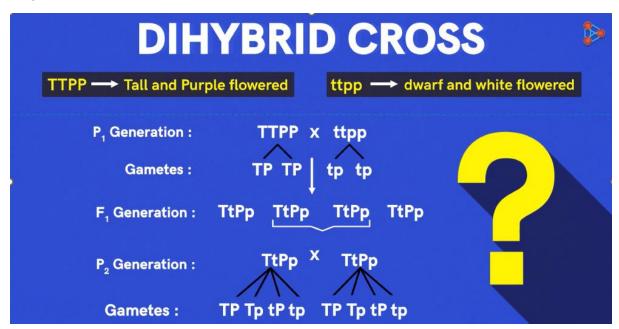
In this case, the Punnett Square shows that all offspring will have the genotype Pp, meaning they will have purple flowers. This is because the dominant allele (P) for purple flower color masks the expression of the recessive allele (p) for white flowers.

# 4. Genotypic and Phenotypic Ratios:

**Genotypic Ratio:** 1 (Homozygous Dominant): 2 (Heterogeneous): 1 (Homozygous Recessive) **Phenotypic Ratio:** 3 (Purple flower-producing plant): 1 (White flower-producing plant)

Exercise: Determine the F2 generation resulting from the following dihybrid cross.

# **Dihybrid cross:**



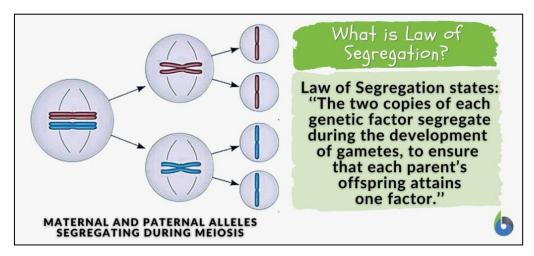
# 1. Law of Segregation: Understanding Allele Separation

The Law of Segregation, proposed by Gregor Mendel, is a fundamental principle of genetics that describes the separation of alleles during the formation of gametes (sperm and egg cells). Alleles are alternative versions of a gene that can code for different traits. This law explains how genetic diversity is maintained in populations and how traits are passed from one generation to the next.

# **Key Points:**

- 1. **Allele Separation:** Each individual carries two alleles for each gene, one inherited from each parent. During the formation of gametes, these alleles segregate or separate, ensuring that each gamete carries only one allele for a specific trait.
- 2. **Random Segregation:** The segregation of alleles is a random process. It means that the allele a gamete receives from one parent is independent of the allele received from the other parent. This random assortment of alleles contributes to genetic diversity.

3. **Genotype and Phenotype:** The genotype represents an individual's genetic makeup, which consists of the combination of alleles they carry. The phenotype is the observable physical expression of those alleles.



**Example:** Consider a monohybrid cross between two pea plants. One parent plant has two dominant alleles for tall height (TT), and the other has two recessive alleles for short height (tt).

## Parental Genotypes:

- Parent 1: TT (homozygous dominant, tall)
- Parent 2: tt (homozygous recessive, short)

#### Gamete Formation:

• Parent 1 produces gametes with the allele 'T,' and Parent 2 produces gametes with the allele 't' because of allele segregation.

# • Offspring Genotypes:

 When these gametes combine during fertilization, the resulting offspring inherit one allele from each parent. The possible genotypes of the offspring are Tt (heterozygous, tall).

#### Phenotypic Ratio:

• The phenotypic ratio of the offspring is 100% tall, even though they carry one dominant and one recessive allele. This is because the dominant allele 'T' masks the expression of the recessive allele 't.'

The Law of Segregation is crucial in understanding how traits are passed from generation to generation and how genetic diversity is maintained within populations. It laid the foundation for our understanding of genetics and heredity.

#### 2. Law of Independent Assortment: Embracing Genetic Diversity

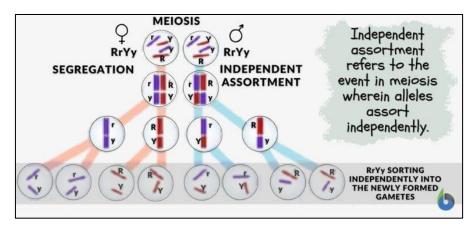
The Law of Independent Assortment, another pivotal concept proposed by Gregor Mendel, is a fundamental principle in genetics that underscores the independent inheritance of genes for different

traits. This law highlights how the assortment of alleles for one trait does not influence the assortment of alleles for another trait during gamete formation, thus contributing to genetic diversity.

## **Key Points:**

- 1. **Assortment of Alleles:** According to this law, genes located on different chromosomes assort independently during gamete formation. This means that the inheritance of one trait is not dependent on the inheritance of another trait.
- 2. **Random Distribution:** The random distribution of alleles from parents to offspring results in an assortment of gene combinations in offspring. This randomness leads to the creation of unique genetic profiles in each individual.
- 3. **Dihybrid Crosses:** The Law of Independent Assortment is particularly evident in dihybrid crosses, where two different traits are considered simultaneously. The genes for these traits segregate independently, leading to various combinations of alleles in the offspring.

**Example:** Let's consider a dihybrid cross involving pea plants. One parent plant has the genotype YyRr, where 'Y' represents yellow seed color (dominant) and 'R' represents round seed texture (dominant). The other parent plant also has the genotype YyRr.



## Parental Genotypes:

- Parent 1: YyRr (heterozygous for both traits)
- Parent 2: YyRr (heterozygous for both traits)

## Gamete Formation:

• During gamete formation, the alleles segregate independently. The gametes from both parents can carry any combination of 'Y' or 'y' alleles for seed color and 'R' or 'r' alleles for seed texture.

# • Offspring Genotypes:

 When these gametes combine during fertilization, the resulting offspring inherit a combination of alleles independently for both traits. The possible genotypes of the offspring include YYRR, YyRR, YyRr, Yyrr, and so on.

#### Phenotypic Variety:

• This independent assortment of alleles results in a diverse phenotypic range among the offspring, contributing to genetic diversity within the population. Each offspring can have a unique combination of seed color and texture.

The Law of Independent Assortment is essential in understanding how genes for different traits are inherited independently, leading to the vast genetic diversity observed in populations. This concept has profound implications in genetics, evolution, and the study of inheritance patterns.

## 3. Laws of Dominance: Understanding Inheritance Patterns

The Laws of Dominance, encompassing both complete dominance and its variations, incomplete dominance and codominance, elucidate how specific alleles interact to determine phenotypic traits in offspring. These laws provide insights into the expression of dominant and recessive alleles, leading to a spectrum of phenotypes and enhancing our comprehension of genetic inheritance.

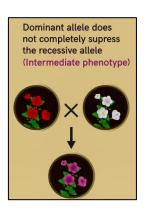
#### **Key Points:**

## 1. Complete Dominance:

- In the context of complete dominance, one allele (the dominant allele) fully masks the expression of the other allele (the recessive allele) in a heterozygous individual. The dominant allele's phenotype is observed.
- Example: In Mendel's pea plant experiments, the allele for purple flower color (P) is dominant over the allele for white flower color (p). Heterozygous plants (Pp) exhibit the dominant purple phenotype.

# 2. Incomplete Dominance (Blending Inheritance):

- In incomplete dominance, neither allele is completely dominant over the other. Instead, they blend, resulting in an intermediate or blended phenotype in heterozygous individuals.
- Example: In snapdragons, a red-flowered plant (RR) crossed with a white-flowered plant (WW) produces heterozygous offspring (RW) with pink flowers, illustrating incomplete dominance.



#### 3. Codominance:

- Codominance occurs when both alleles in a heterozygous individual are fully expressed, without one being dominant over the other. This leads to a phenotype that displays traits from both alleles simultaneously.
- Example: In the ABO blood group system, individuals with alleles for blood types A (IA) and B (IB) express both A and B antigens on their red blood cells, resulting in type AB blood. Neither allele is dominant, and both are visibly expressed.

## Significance:

 These laws of dominance demonstrate the complex interplay of alleles in determining phenotypic traits.

 They highlight the versatility of genetic inheritance, with some traits following traditional Mendelian patterns of complete dominance, while others exhibit unique variations such as incomplete dominance and codominance.

• Understanding these laws enhances our ability to predict and explain a wide range of genetic traits and their expressions in populations.

#### **Epistasis: The Intricate Gene Interaction**

Epistasis is a genetic phenomenon where the expression of one gene (the epistatic gene) masks or modifies the expression of another gene (the hypostatic gene) at a different locus. In simpler terms, epistasis occurs when one gene controls or influences the expression of another gene, leading to unexpected genetic outcomes.

# **Key Points:**

## 1. Epistatic Gene:

- The epistatic gene is the gene that exerts control over the expression of another gene (the hypostatic gene). It can either enhance or suppress the expression of the hypostatic gene.
- Example: In Labrador Retrievers, the gene responsible for coat color (the B gene) is epistatic to the gene controlling pigment deposition (the E gene). The B gene determines whether the dog will have black (BB or Bb) or brown (bb) fur, while the E gene affects the distribution of pigment in the fur.

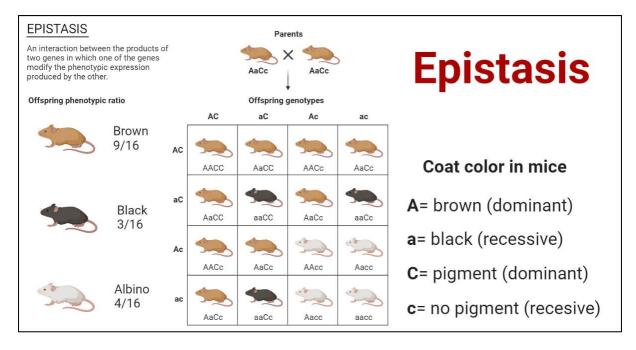
#### 2. Hypostatic Gene:

- The hypostatic gene is the gene whose expression is influenced or modified by the presence or absence of the epistatic gene.
- Example: In the case of Labrador Retrievers, the E gene controls whether pigment is deposited evenly or not in the fur. The presence or absence of the B gene (epistatic gene) determines whether the fur color will be black or brown, but the E gene influences how that color is distributed.

#### Significance:

- Epistasis adds complexity to genetic interactions by revealing that the effect of one gene is contingent on the presence or absence of another gene.
- Understanding epistasis is essential in studying traits that result from multiple genes, as it
  allows us to unravel the intricate relationships between genes and their collective impact on
  phenotypes.

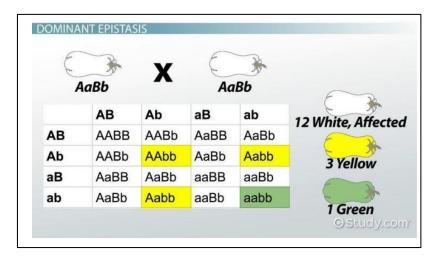
**Example:** In mice, the presence of a particular allele (Cc) for coat color is epistatic to another gene (Aa) that controls the presence of pigment in the fur. If the mouse has at least one "C" allele, it will have a colored coat, regardless of the "A" allele's presence or absence. However, if the mouse lacks the "C" allele and has "cc," its coat color will depend on the "A" allele's presence: "Aa" will produce a colored coat, while "aa" will result in an albino coat. Here, the epistatic gene "C" controls the expression of the hypostatic gene "A" and determines the presence or absence of pigment in the fur.



**Recessive Epistasis:** The example provided is a demonstration of recessive epistasis. In recessive epistasis, the presence of a recessive allele at the epistatic gene locus (in this case, the "cc" genotype) masks or suppresses the expression of alleles at the hypostatic gene locus (the "Aa" and "aa" genotypes) and affects the phenotype.

In the example, when the mouse has the "cc" genotype (recessive epistatic gene), it doesn't matter whether it carries the "Aa" or "aa" genotype at the hypostatic gene locus; the presence of "cc" results in a colorless (albino) coat. This illustrates the classic pattern of recessive epistasis, where the recessive allele at the epistatic locus determines the phenotype regardless of the alleles at the hypostatic locus.

**Dominant Epistasis:** In summer squash, the color of the fruit is determined by the interplay of two genes, gene A and gene B. Gene A is responsible for producing yellow color in the squash fruit. Gene B, on the other hand, codes for a dominant trait that results in white fruit color.

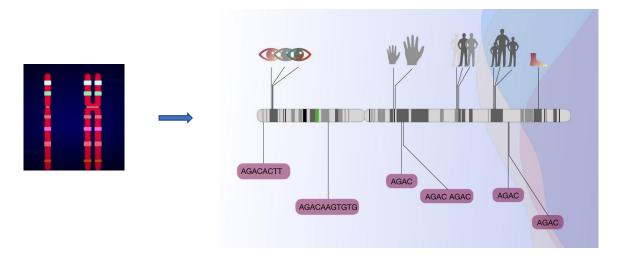


The interesting aspect of this genetic interaction is that gene B exhibits dominant epistasis over gene A. Dominant epistasis means that the presence of the dominant allele of one gene (B) suppresses the expression of the second gene (A), preventing it from producing its characteristic yellow color.

Consequently, in the presence of at least one dominant B allele, the squash fruit will appear white, regardless of whether the individual also carries alleles of gene A.

Yellow fruit color only manifests when both alleles at the B locus are recessive (bb), allowing the expression of gene A to prevail. However, in the absence of both dominant B alleles (bb) and recessive A alleles (aa), a completely different phenotype emerges: green squash fruit color. This demonstrates how the interaction between these two genes determines the range of colors observed in summer squash, with the dominance of B effectively masking the expression of A, and the absence of both, resulting in the green fruit color phenotype.

**Gene Mapping:** Gene mapping involves locating genes on chromosomes to understand genetic traits and diseases. Researchers start by collecting samples from individuals with and without the trait or disease of interest. They then isolate DNA from these samples, examining it for unique patterns called polymorphisms or markers. These markers can be any distinguishable sequence feature in the DNA. Genes are represented as traits that differ between parents and can serve as genetic markers. Gene mapping is essential for genetics, genomics, and disease research and crop improvement applications.



## **Single Gene Disorders:**

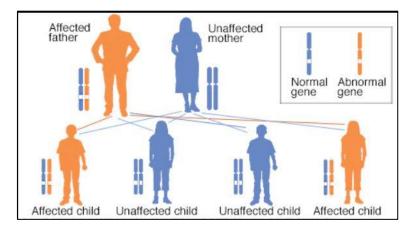
- Single-gene disorders, also known as Mendelian or monogenic disorders, are genetic disorders
  caused by mutations in a single gene. These mutations can be inherited from one or both
  parents.
- 2. **Genetic Basis**: These disorders are typically caused by mutations in one specific gene that disrupts the normal functioning of that gene's protein product.

#### 3. Inheritance Patterns:

**Autosomal Dominant**: Inheritance of a single mutated copy of the gene from either parent is sufficient to cause the disorder (e.g., Huntington's disease).

Dominant diseases are single gene disorders that occur in the heterozygous state –
 when an individual has one mutant copy of the relevant gene and one healthy copy.

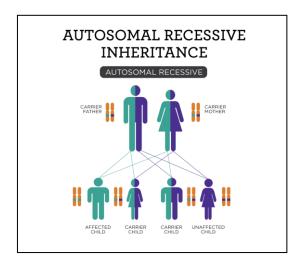
• The effects of the mutant version of the gene (allele) override the effects of the healthy version of the gene. So, the mutant allele causes disease symptoms even though a healthy allele is present.



- Dominant disorders tend to crop up in every generation of an affected family because everyone carrying a dominant mutant allele shows the symptoms of the disease. Dominant disorders spread vertically down family trees, from parent to child.
- In rare cases, when an individual has two copies of the mutant gene (also known as being homozygous), the disorder symptoms are generally more severe.
- An example of a dominant single-gene disorder is Huntington's disease, a nervous system disease.

**Autosomal Recessive**: Both copies of the gene must be mutated to manifest the disorder (e.g., cystic fibrosis).

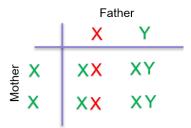
- Recessive diseases are single-gene disorders that only occur in the homozygous state when an individual carries two mutant versions (alleles) of the relevant gene.
- The effects of the healthy allele can compensate for the effects of the mutant allele. The mutant allele does not cause disease symptoms when a healthy allele is also present.
- However, if a parent inherits two mutant alleles, there are no healthy alleles, so the mutant allele can exert its effect.



 Recessive diseases are more difficult to trace through family trees because carriers of a mutant allele do not show symptoms of the disease.

- It, therefore, appears that the disease has skipped a generation when it is seen in groups of children within a family.
- The risk of an individual having a recessive disorder increases when two people who are closely related have a child together (consanguinity).
- This is because there is a much greater chance that the same mutant allele will be present in related parents.

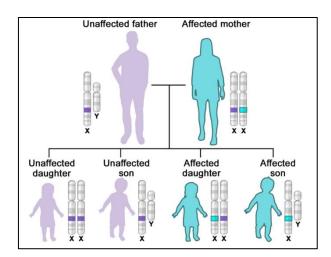
X-linked: The mutated gene is located on the X chromosome, and the disorder predominantly affects males (e.g., haemophilia).



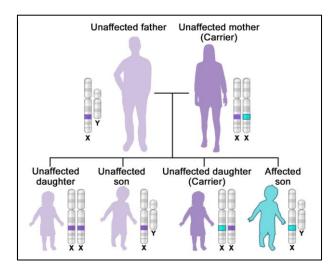
Father can pass x-linked disorders only to daughter, and never to son.

- X-linked disorders are single-gene disorders that result from the presence of a mutated gene on the X chromosome.
- Because females (XX) have two copies of the X chromosome, but males (XY) only have one copy, X-linked disorders are more common in males.
- If a male's single copy on the X chromosome is mutated, he has no healthy copy to restore healthy function.
- The inheritance patterns of X-linked diseases are simplified by the fact that males always pass their X chromosome to their daughters but never to their sons.
- Like other single-gene disorders, X-linked disorders can be either recessive or dominant.

## - X-linked dominant diseases:



#### - X-linked recessive diseases:



## 4. Examples of Single Gene Disorders:

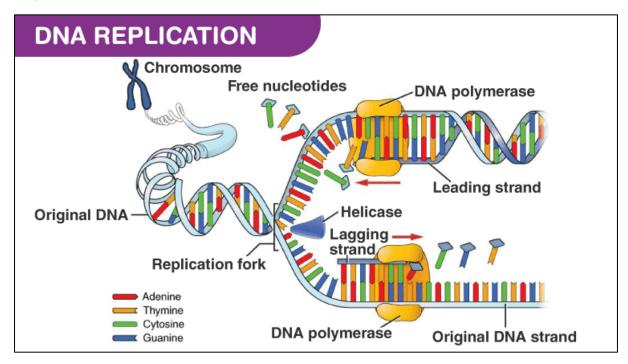
- **Cystic Fibrosis**: Caused by mutations in the CFTR gene, resulting in thick mucus production that affects the lungs, digestive system, and other organs.
- **Huntington's Disease**: Arises from mutations in the HTT gene, leading to progressive neurological degeneration.
- **Sickle Cell Anaemia**: Caused by mutations in the HBB gene, resulting in misshaped red blood cells and various health complications.
- **Duchenne Muscular Dystrophy**: Results from mutations in the DMD gene, leading to muscle weakness and degeneration.
- 5. **Diagnosis**: Genetic testing, family history analysis, and clinical symptoms are used to diagnose single gene disorders.
- 6. **Treatment**: Treatment options vary depending on the disorder but may include medication management, gene therapy, or supportive care to alleviate symptoms.
- 7. **Prevention**: In some cases, carrier screening and genetic counseling can help individuals understand their risk of passing on a genetic disorder to their offspring.
- 8. **Frequency**: Single gene disorders are relatively rare individually but collectively account for a significant portion of genetic disorders.
- 9. **Research and Advancements**: Advances in genetic research, including gene editing technologies like CRISPR-Cas9, offer promising avenues for potential treatments and cures for some single gene disorders.

In summary, single-gene disorders result from mutations in a single gene and follow specific inheritance patterns. Understanding the genetic basis of these disorders is crucial for diagnosis, treatment, and genetic counselling to manage and prevent them in affected individuals and their families.

### **DNA Replication:**

DNA replication, or semi-conservative replication, is the process by which DNA is essentially doubled. It is an important process that takes place within the dividing cell.

DNA replication is a complex process that ensures the accurate duplication of genetic material. It involves the coordination of various enzymes to unwind the DNA double helix, synthesize new strands, and proofread for errors. Here is a step-by-step explanation of DNA replication, along with the six enzymes involved:



Step 1: Helicase

Function: Helicase is the initial enzyme in DNA replication. Its primary function is to unwind the DNA double helix. It accomplishes this by breaking the hydrogen bonds between the complementary base pairs, such as A-T and G-C. By doing so, helicase separates the two DNA strands, creating what is called a "replication fork." This single-stranded DNA region is where the actual replication process will take place.

# Step 2: Primase

Function: Primase follows helicase. Its role is to synthesize RNA primers. Primase adds short RNA sequences, known as primers, to the DNA template strands. These primers are essential because DNA Polymerase, the enzyme responsible for adding new DNA nucleotides during replication, can only extend an existing strand. On the leading strand, primase synthesizes a single RNA primer at the 5' end, providing a starting point for DNA Polymerase.

# Step 3: DNA Polymerase III

Function: DNA Polymerase III is the primary enzyme responsible for DNA synthesis during replication. It adds nucleotides to the growing DNA strand. On the leading strand, DNA Polymerase III synthesizes

the new DNA strand in a continuous manner by extending from the 5' to 3' direction, using the parental DNA strand as a template. This strand doesn't encounter the same challenges as the lagging strand, which requires a more intricate process.

#### Step 4: Exonuclease

Function: Exonucleases come into play after DNA Polymerase III. Their function is to remove RNA primers from the DNA template. Once the RNA primers are synthesized by primase, exonucleases degrade them, creating gaps in the DNA strands. On the lagging strand, multiple RNA primers are synthesized at different points along the template to provide starting points for DNA Polymerase.

## Step 5: DNA Polymerase I

Function: DNA Polymerase I is an enzyme responsible for gap-filling. After the removal of RNA primers by exonucleases, DNA Polymerase I replaces the RNA nucleotides with DNA nucleotides. On the lagging strand, DNA Polymerase I works in a discontinuous manner, filling in the gaps created by the removal of RNA primer.

#### Step 6: Ligase

Function: Ligase is the final enzyme involved in DNA replication. Its function is to seal the nicks or gaps in the DNA backbone. It does so by catalyzing the formation of phosphodiester bonds between adjacent nucleotides. On the lagging strand, ligase joins the Okazaki fragments together, ensuring the integrity and continuity of the newly synthesized DNA strand.

In conclusion, DNA replication involves two strands: the leading strand, which is synthesized continuously, and the lagging strand, which is synthesized discontinuously in Okazaki fragments. Different enzymes work together to ensure the accurate replication of both strands, preserving genetic information during cell division and ensuring the inheritance of genetic traits.



# **DNA Replication:**

### Stages of DNA replication

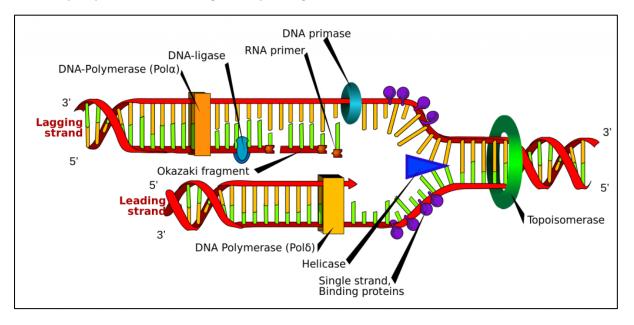
DNA replication can be thought of in three stages: Initiation, Elongation, Termination

#### Initiation

DNA synthesis is initiated at particular points within the DNA strand known as 'origins', which are specific coding regions. These origins are targeted by initiator proteins, which go on to recruit more proteins that help aid the replication process, forming a replication complex around the DNA origin. There are multiple origin sites, and when replication of DNA begins, these sites are referred to as **replication forks**.

Within the replication complex is the enzyme **DNA Helicase**, which unwinds the double helix and exposes each of the two strands, so that they can be used as a template for replication. It does this by hydrolyzing the ATP used to form the bonds between the nucleobases, therefore breaking the bond holding the two strands together.

DNA Primase is another enzyme that is important in DNA replication. It synthesizes a small **RNA primer**, which acts as a 'kick-starter' for **DNA Polymerase**. DNA Polymerase is the enzyme that is ultimately responsible for creating and expanding the new strands of DNA.



## **Elongation**

Once the DNA Polymerase has attached to the original, unzipped two strands of DNA (i.e. the **template** strands), it is able to start synthesizing the new DNA to match the templates. It is essential to note that DNA polymerase can only extend the primer by adding free nucleotides to the **3'** end.

One of the templates is read in a 3' to 5' direction, meaning the new strand will be formed in a 5' to 3' direction. This newly formed strand is referred to as the **Leading Strand**. Along this strand, DNA Primase only needs to synthesize an **RNA primer** once, at the beginning, to initiate DNA Polymerase. This is because DNA Polymerase can extend the new DNA strand by reading the template 3' to 5', synthesizing in a 5' to 3' direction as noted above.

However, the other template strand (the **lagging strand**) is antiparallel and is therefore read in a **5' to 3'** direction. Continuous DNA synthesis, as in the **leading strand**, would need to be in the 3' to 5' direction, which is impossible as we cannot add bases to the 5' end. Instead, as the helix unwinds, RNA primers are added to the newly exposed bases on the **lagging strand** and DNA synthesis occurs **in fragments**, but still in the 5' to 3' direction as before. These fragments are known as **Okazaki fragments**.

#### **Termination**

Expanding the new DNA strands continues until there is no more DNA template left to replicate (i.e. at the end of the chromosome), or two replication forks meet and subsequently **terminate.** The meeting of two replication forks is not regulated and happens randomly along the course of the chromosome.

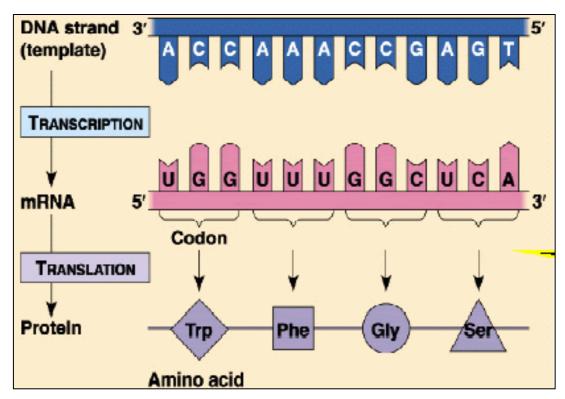
Once DNA synthesis has finished, it is important that the newly synthesized strands are bound and stabilized. With regards to the lagging strand, two enzymes are needed to achieve this: **RNAase H** removes the RNA primer that is at the beginning of each Okazaki fragment, and **DNA Ligase** joins fragments together to create one complete strand.

#### Video content:

https://www.youtube.com/watch?v=ISvF5-rBRGQ&ab channel=FuseSchool-GlobalEducation

## **Central Dogma of Life:**

Central Dogma - An Inheritance Mechanism

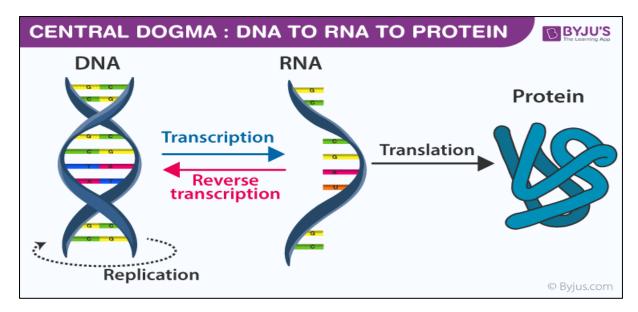


In molecular biology, central dogma illustrates the flow of genetic information from DNA to RNA to protein. It is defined as a process in which the information in DNA is converted into a functional product.

It is suggested that the information present in DNA is essential to make up all proteins and RNA acts as a messenger that carries information through the ribosomes.

Central Dogma Definition

"Central dogma is the process in which the genetic information flows from DNA to RNA, to make a functional product protein."

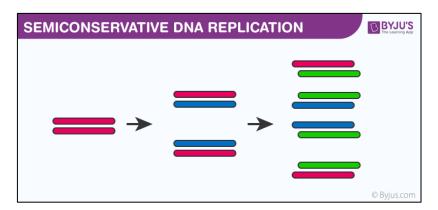


## What is Central Dogma?

The central dogma illustrates the flow of genetic information in cells, the DNA replication, and coding for the RNA through the transcription process and further RNA codes for the proteins by translation.

The concept of a sequence of interactions can be understood through the framework. The most common include biopolymers. The major category of biopolymers include Proteins, RNA, and DNA that are further divided into general transfers, unknown transfers, and special transfers.

Special transfers occur in an exceptional case in the laboratory. General transfer occurs in almost all cells. It describes the regular flow of information through transcription and translation. Unknown transfers are said never to occur.



The new DNA strands are formed, with one strand of the parent DNA and the other is newly synthesized, this process is called semiconservative DNA replication.

# Central Dogma - Steps

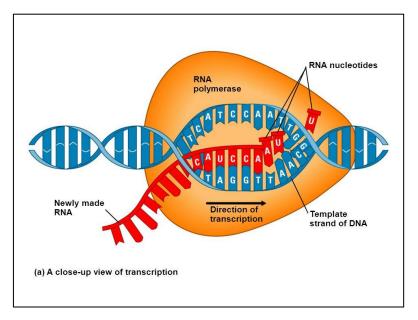
The central dogma takes place in two different steps:

## Transcription

Transcription is the process by which the information is transferred from one strand of the DNA to RNA by the enzyme RNA Polymerase. The DNA strand which undergoes this process consists of three parts namely promoter, structural gene, and a terminator.

The DNA strand that synthesizes the RNA is called the template strand and the other strand is called the coding strand. The DNA-dependent RNA polymerase binds to the promoter and catalyzes the polymerization in the 3' to 5' direction.

As it approaches the terminator sequence, it terminates and releases the newly synthesized RNA strand. The newly released RNA strand further undergoes post-transcriptional modifications.

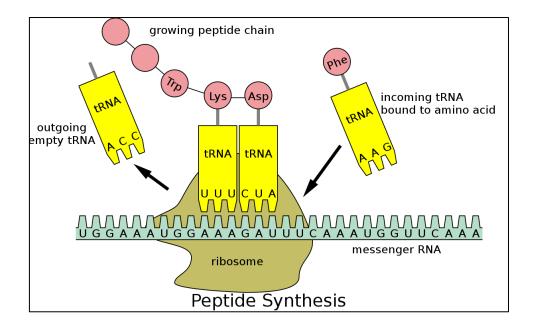


## **Translation**

Translation is the process by which the RNA codes for specific proteins. It is an active process which requires energy. This energy is provided by the charged tRNA molecules.

Ribosomes initiate the translation process. The ribosomes consist of a larger subunit and a smaller subunit. The larger subunit, in turn, consists of two tRNA molecules placed close enough so that peptide bond can be formed at the expense of enough energy.

The mRNA enters the smaller subunit which is then held by the tRNA molecules of the complementary codon present in the larger subunit. Thus, two codons are held by two tRNA molecules placed close to each other and a peptide bond is formed between them. As this process repeats, long polypeptide chains of amino acids are synthesized.



## **Assignment:**

1. What is the role of DNA (Deoxyribonucleic Acid) in genetics and how does it contribute to the inheritance of traits?

- 2. Define and differentiate between homozygous dominant, homozygous recessive, and heterozygous genotypes. Provide examples for each.
- 3. Explain the significance of monohybrid and dihybrid crosses in understanding the inheritance patterns of single and multiple traits, respectively.
- 4. Describe the P1, F1, and F2 generations in genetic crosses, and discuss the genotypic and phenotypic ratios observed in these generations.
- 5. Provide examples illustrating the application of the Laws of Dominance, Segregation, and Independent Assortment in explaining patterns of inheritance in specific genetic crosses.
- 6. Differentiate between recessive epistasis and dominant epistasis and provide examples illustrating each type of epistatic interaction.
- 7. Provide examples of single gene disorders, both autosomal and X-linked, and briefly describe the associated symptoms or characteristics of each disorder.
- 8. Describe the process of DNA replication, including the key steps involved and the enzymes responsible. Additionally, provide a diagram illustrating the process of DNA replication and explain the significance of replication fidelity in maintaining genetic stability.
- 9. Explain the process of transcription in molecular biology, including the key steps involved and the enzymes responsible. Additionally, provide a diagram illustrating the process of transcription and discuss the importance of transcription in gene expression and protein synthesis.
- 10. Describe the process of translation in molecular biology, including the key steps involved and the components required. Additionally, provide a diagram illustrating the process of translation and explain the role of ribosomes, transfer RNA (tRNA), and messenger RNA (mRNA) in protein synthesis.