Mendelian Modes of Inheritance

Introduction: Mendel's Legacy in Human Genetics

The science of inheritance began with the humble experiments of **Gregor Johann Mendel**, an Austrian monk who studied how traits passed from one generation to the next in pea plants.

His work, published in 1866, laid the foundation for the field of genetics. Though unrecognized during his lifetime, Mendel's principles of **segregation** and **independent assortment** later became the core of classical or Mendelian genetics.

Mendel's laws are not just about plants—they explain how humans inherit traits like eye color, blood groups, and even diseases. These **Mendelian modes of inheritance** are still essential in understanding genetic disorders, population genetics, and human diversity in anthropology.

I. Autosomal Inheritance

Autosomal inheritance refers to traits controlled by genes located on **autosomes**—the non-sex chromosomes (humans have 22 pairs of autosomes and one pair of sex chromosomes). These traits are inherited regardless of whether the individual is male or female.

1. Autosomal Dominant Inheritance

In **autosomal dominant inheritance**, a person only needs **one copy of the dominant gene** from either parent to express the trait or disorder.

This means that even if the other gene copy is normal, the dominant gene will "overpower" it and show its effect. Because only one copy is needed, the trait is often seen in **every generation**, and it does **not skip generations**.

This type of inheritance occurs in genes located on **autosomes**—the 22 pairs of non-sex chromosomes that are the same in both males and females. As a result, the trait appears equally in **males and females**, and either parent can pass it to their children.

Examples:

1. **Achondroplasia**
   This is a well-known form of **dwarfism** caused by a mutation in the **FGFR3 gene**. People with achondroplasia typically have **short limbs, a large head, and normal torso size**.

- If a child inherits one mutated gene, they will have the condition.
- If they inherit two copies (from both parents), the condition is often fatal before or shortly after birth.
- In many cases, the mutation happens spontaneously (not inherited), but it can also be passed down.

2. **Huntington's Disease**
   A serious **neurodegenerative disorder** that typically starts in **middle age**. It is caused by a defect in the **HTT gene**, leading to uncontrolled movements (chorea), memory loss, and personality changes.
   - Affected individuals usually inherit the gene from an affected parent.
   - Because symptoms often begin **after reproductive age**, people may unknowingly pass the gene to their children before the disease shows up in themselves.

Key Features:

- **Equal occurrence** in both males and females
- Usually, **one affected parent** can pass the gene
- If **one parent is affected** (heterozygous), there is a **50% chance** that each child will inherit the trait
- The trait usually appears in **every generation** of an affected family
- If a child does **not inherit the gene**, they cannot pass it on to their children

2. Autosomal Recessive Inheritance

In **autosomal recessive inheritance**, a person must inherit **two copies of the recessive gene**—one from each parent—for the trait or disorder to appear.

If the person has only **one copy of the recessive gene**, they are called a **carrier**. Carriers do not show symptoms themselves but can pass the gene to their children.

Because both copies of the gene need to be faulty, the condition often appears when **both parents are carriers**, even if they look healthy. If two carriers have a child:

- There is a **25% chance** the child will have the disorder
- A **50% chance** the child will be a carrier
- A **25% chance** the child will inherit two normal genes

This type of inheritance occurs on **autosomes**, which means it affects **males and females equally**.

Examples:

1. **Cystic Fibrosis**
   A serious genetic disorder that affects the **lungs and digestive system**. It is

caused by mutations in the **CFTR gene**, leading to the buildup of **thick mucus** in the lungs and other organs.

- People with cystic fibrosis often experience **chronic lung infections** and **difficulty digesting food**.
- Carriers usually don't have symptoms, so the condition can go unnoticed in families for generations.

2. **Sickle Cell Anemia**
   A **blood disorder** caused by a mutation in the gene that codes for **hemoglobin**. This results in red blood cells becoming **sickle-shaped**, making it hard for them to carry oxygen and move through blood vessels smoothly.
   - People with two copies of the sickle cell gene (homozygous) have **sickle cell anemia**, which causes pain, fatigue, and organ damage.
   - People with one copy (heterozygous) have **sickle cell trait**, which usually causes no symptoms but provides some **resistance to malaria**—an advantage in malaria-endemic regions.

Key Features:

- Often **skips generations** because carriers do not show symptoms
- Affects **males and females equally**
- **Unaffected parents** can have an affected child if both are carriers
- The condition is more likely in **consanguineous marriages** (between relatives), where both parents might carry the same gene

3. Autosomal Codominance

Codominance is a special type of inheritance where **both alleles in a gene pair are equally and fully expressed** in the individual. Unlike dominant-recessive inheritance—where one allele masks the other—in codominance, **both traits appear side by side** in the phenotype.

This means that if a person inherits one allele from each parent, instead of blending the traits or one being hidden, **both will be visible**.

Classic Example: ABO Blood Group System

The **ABO blood group** system is the best-known example of codominance in humans. This system is controlled by a single gene with **three alleles**:

- **IA** (codes for A antigen)
- **IB** (codes for B antigen)
- **i** (codes for no antigen)
- If a person inherits **IA and IB**, they will have **AB blood group**.

- In this case, both A and B antigens are present **together** on the surface of red blood cells.
- Neither A nor B is dominant over the other—they are **codominant**.

Key Features:

- Both alleles are **expressed equally and visibly**
- The resulting phenotype is a **combination** of both traits, not a blend
- No masking or suppression of one allele by the other

II. Sex-linked Inheritance

Sex-linked traits are carried on the **sex chromosomes**, particularly the **X chromosome**. Since males have one X and one Y chromosome (XY), and females have two X's (XX), X-linked traits often behave differently in males and females.

4. X-linked Recessive Inheritance

X-linked recessive inheritance refers to genetic traits or disorders caused by **mutations on the X chromosome**, one of the two sex chromosomes.

Since males have only **one X chromosome** (and one Y), a single copy of a faulty gene on the X is enough to cause the disorder.

In contrast, **females have two X chromosomes**, so a faulty gene on one X can be masked by a healthy gene on the other. As a result, **males are more likely to be affected**, while **females usually act as carriers**.

Why Males Are More Affected

- Males: **XY** → If the **X** has the mutation, there's **no second X** to compensate
- Females: **XX** → If one X has the mutation and the other is normal, the normal one usually **prevents symptoms**, but the female can **pass the gene to children**

This pattern makes **X-linked recessive disorders** more visible in **sons**, while **daughters** may be unaffected carriers.

Examples:

1. **Haemophilia A**
   A genetic disorder where blood **does not clot properly** due to a lack of **clotting factor VIII**.
   - Small injuries can lead to **prolonged bleeding**, joint damage, or even internal bleeding.
   - Common in males; carrier females may not show symptoms.
   - Famous historical example: **descendants of Queen Victoria**, who was a carrier, passed haemophilia into the royal families of **Russia, Spain,**

and Germany. This case is often studied in medical genetics and anthropology.

2. **Red-Green Colour Blindness**
   A condition where people have trouble **distinguishing between red and green hues**.
   - Affects about **8% of males** but only **0.5% of females** globally.
   - This condition is **non-lethal** and often goes unnoticed, but it serves as a useful model for studying **X-linked inheritance** in population studies.

Key Features of X-linked Recessive Inheritance:

- **No male-to-male transmission**: A father **cannot pass his X chromosome to his sons**, only to daughters.
- **Affected males pass the faulty gene to all daughters**, who become **carriers**.
- **Carrier females** have a **50% chance of passing the gene** to their **sons (affected)** and a **50% chance to daughters (carriers)**.

5. Sex-Influenced Inheritance

Sex-influenced inheritance refers to a pattern where a gene is **not located on a sex chromosome**, but its **expression is influenced by the biological sex** of the individual—usually due to **hormonal differences** like testosterone or estrogen.

In this case, the **same gene** behaves **differently in males and females**. A gene may appear **dominant in one sex** and **recessive in the other**, depending on the hormone levels present in the body.

This type of inheritance typically involves genes on the **autosomes** (non-sex chromosomes), but the **phenotype depends on the individual's sex**.

Example: Male-Pattern Baldness

One of the best-known examples of sex-influenced inheritance is **male-pattern baldness**, which is **genetically inherited but hormonally driven**.

- In **males**, the gene behaves as **dominant**. So, even **one copy** of the baldness gene is enough to cause **significant hair loss**.
- In **females**, the same gene behaves as **recessive**. So, a woman usually needs **two copies** of the gene (from both parents) to show **visible thinning or hair loss**.
- However, due to lower levels of **testosterone**, women tend to show **milder symptoms**, even if they inherit both copies.

This example shows how **sex hormones** like **testosterone** can influence how strongly a gene is expressed, even when the gene itself is not sex-linked.

Key Features:

- The **gene is autosomal**, not located on the X or Y chromosome
- The **expression depends on the individual's sex** and their hormone levels
- The trait may **appear dominant in one sex** but **recessive in the other**
- Affected individuals may show **different levels of severity** based on sex

6. Sex-Limited Inheritance

Sex-limited inheritance refers to traits that are **expressed only in one biological sex**, even though the **genes for the trait exist in both males and females**. The reason these traits show up only in one sex is because their **expression depends on sex-specific hormones**, such as **testosterone or estrogen**.

These traits are usually controlled by **autosomal genes** (not on the sex chromosomes), but they are **"switched on" only in one sex** due to the hormonal environment. The other sex may carry the gene silently without ever expressing the trait.

Examples:

1. **Milk Production in Females**
    - Both males and females carry the **genes required for lactation** (milk production), but the trait is **only expressed in females** after childbirth.
    - This is because **high levels of estrogen and prolactin**, which rise during pregnancy and after delivery, are required to activate those genes.
    - In males, although the same genes are present, the **lack of these hormones** prevents expression.
2. **Beard Growth in Males**
    - The genes responsible for **facial hair growth** exist in both sexes.
    - However, only **males grow thick beards**, typically after puberty, because of the **presence of high levels of testosterone**.
    - In females, the **same genes remain inactive** due to low testosterone levels.

These examples show that it's not just the gene that determines the trait—it's also the **hormonal "environment"** of the body.

Key Features:

- Traits are **limited to one sex**, despite the gene being present in **both sexes**
- The gene is usually **autosomal**, but its expression is **dependent on sex-specific hormones**
- Often linked with **reproductive roles** or **secondary sexual characteristics**

PYQ Insights

1. UGC NET frequently tests basic definitions and core concepts of inheritance patterns.
Example: Which of the following shows autosomal dominant inheritance?

2. Match-the-following questions are commonly used to assess recognition of trait-inheritance type links.
Example: Match traits like haemophilia, AB blood group, milk production, and sickle cell anemia with their correct modes of inheritance.

3. Questions involving pedigree analysis test understanding of how traits pass across generations.
Example: A trait skips generations and affects both sexes equally — what is the likely inheritance pattern?

4. X-linked inheritance is often examined through scenario-based questions involving carrier mothers and affected sons.
Example: A carrier mother and a normal father have children — what's the chance their son is affected by an X-linked disorder?

5. Assertion-Reason questions are used to check conceptual depth, especially for sex-influenced traits.
Example: Assertion: Male-pattern baldness is sex-influenced. Reason: It is inherited through the Y chromosome.

6. Codominance is usually tested using the ABO blood group system and allele interactions.
Example: A person with IAIB genotype belongs to which blood group?

7. Anthropological questions focus on inheritance patterns in specific populations or tribal groups.
Example: Which Indian tribal communities show high frequency of the sickle cell trait, and what is its mode of inheritance?

8. Questions often test the difference between sex-influenced and sex-limited traits.
Example: Which of the following traits is sex-limited: beard growth, baldness, or color blindness?

Conclusion

Mendelian inheritance explains how traits are passed from parents to offspring based on Gregor Mendel's principles of segregation and independent assortment.

Autosomal dominant traits require only one copy of a gene to be expressed (e.g., Huntington's disease), while **autosomal recessive traits** need two copies (e.g., sickle cell anemia).

In **codominance**, both alleles are fully expressed, as seen in the AB blood group. **X-linked recessive traits**, like haemophilia, mainly affect males because they have only one X chromosome.

Sex-influenced traits, such as male-pattern baldness, are affected by hormones and show different patterns in males and females. In contrast, **sex-limited traits** like milk production appear only in one sex despite being present in both. These inheritance patterns are crucial in anthropology for studying human diversity, population genetics, and gene-environment interactions.

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