

Basic Patterns of Human Inheritance: Pedigrees



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المزيد من مادة
علوم:

إعداد: Sailux

التواصل الاجتماعي بحسب الصف الحادي عشر العام



صفحة المناهج
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المزيد من الملفات بحسب الصف الحادي عشر العام والمادة علوم في الفصل الثالث

Applied Genetics

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Pedigrees

Review **Table 1**, and recall the following:

- A **recessive** trait is expressed when the individual is **homozygous** recessive for that trait.
- Those with at least one dominant allele will not express the recessive trait.
- An individual who is **heterozygous** for a recessive disorder is called a **carrier**.

This information is applied to help study:

- Patterns of human inheritance, including:
 - Inheritance of dominant disorders
 - Inheritance of recessive disorders

Term	Example	Definition
Homozygous	True-breeding yellow-seed pea plants would be YY, and green-seed pea plants would be yy. 	An organism with two of the same alleles for a particular trait is said to be homozygous for that trait.
Heterozygous	A plant that is Yy would be a yellow-seed pea. 	An organism with two different alleles for a particular trait is said to be heterozygous for that trait. When alleles are present in the heterozygous state, the dominant trait will be observed.

Table 1

Homozygous:

يعني إن الكائن الحي عنده نسختين متطابقتين من الجين المسؤول عن صفة معينة (نفس اللون) YY أو yy

Heterozygous:

يعني إن الكائن الحي عنده نسختين مختلفتين من الجين، واحدة سائدة وواحدة متنحية والصفة السائدة هي التي بتظهر في الشكل الخارجي
هي الأقوى Y النبات بيطلع لونه أصفر لأن Yy



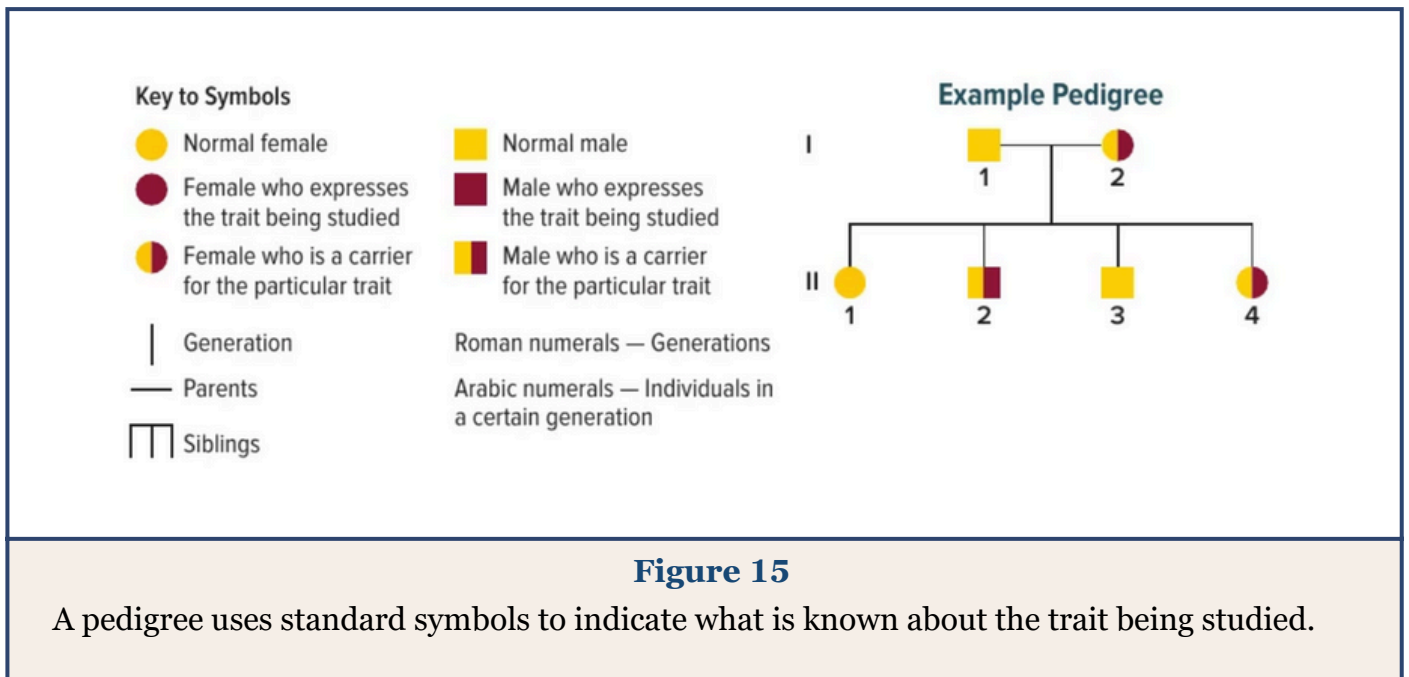
In organisms such as peas and fruit flies, scientists can perform crosses to study **genetic relationships**.

In the case of humans, a scientist studies a family history using a pedigree:

- A diagram that traces the inheritance of a particular trait through several generations.

A pedigree, such as the one shown in Figure 15, uses symbols to illustrate inheritance of the trait:

- Males are represented by squares.
- Females are represented by circles.
- One who expresses the trait is represented by a dark (filled) square or circle, depending on their gender.
- One who does not express the trait is represented by an **unfilled** square or circle.



- A **horizontal line** between two symbols shows that these individuals are the parents of the offspring listed below them.
- **Offspring:**
 - Are listed in descending birth order from left to right.
 - Are connected to each other and to their parents.
- **A pedigree uses a numbering system:**
 - a. Roman numerals represent **generations**.
 - b. Arabic numbers represent the **birth order within a generation**.
- **Example:**
 - In **Figure 15**, individual II1 is a female who is the firstborn in generation II.



EXTRA

Generation	Individual	Sex	Genotype (assumed)	Phenotype
I	1	♂ Male (square)	AA (Normal, not carrier)	Healthy
I	2	♀ Female (circle, half-shaded)	Aa (Carrier)	Healthy (carrier)
II	1	♀ Female (circle, yellow only)	AA (Normal)	Healthy
II	2	♂ Male (square, half-shaded)	Aa (Carrier)	Healthy (carrier)
II	3	♂ Male (square, yellow only)	AA (Normal)	Healthy
II	4	♀ Female (circle, half-shaded)	Aa (Carrier)	Healthy (carrier)

Notes:

- **Shaded red = affected (aa).** None are fully shaded → no affected individuals.
- **Half shaded = carrier (Aa).**
- **Unshaded = normal (AA).**
- **Disorder is autosomal recessive:**
 - Affected: aa
 - Carrier: Aa
 - Normal: AA

Analyzing Pedigrees

- **Purpose of Pedigrees:**
 - Used to study inheritance of both recessive and dominant traits within a family.
 - Constructed using all **available information** about the presence¹ or absence² of a particular trait in family members.
 - Analyzed to reveal patterns of inheritance.
- **Applications of Pedigree Analysis:**
 - Helps determine whether a trait is **X-linked or autosomal**.
 - Patterns can reveal the mode of inheritance (e.g., of a disease, disorder, or trait).
 - Can infer genotypes of specific individuals.
 - Used to predict the possibility of disorders in future offspring.



Recessive Genetic Disorders

- A pedigree illustrating Tay-Sachs disease is shown in **Figure 16**.

Tay-Sachs disease:

- Is a recessive genetic disorder.
- Caused by the **lack of an enzyme involved in lipid metabolism**.
- The missing enzyme causes lipids to build up in the central nervous system, which can lead to death.

Pedigree analysis (**Figure 16**):

- Two unaffected parents (I1 and I2) have an affected child (II3).
- **This indicates:**
 - Each parent has one recessive allele.
 - Both are heterozygous → carriers of the trait.
- The half-filled square and circle show that both parents are carriers.

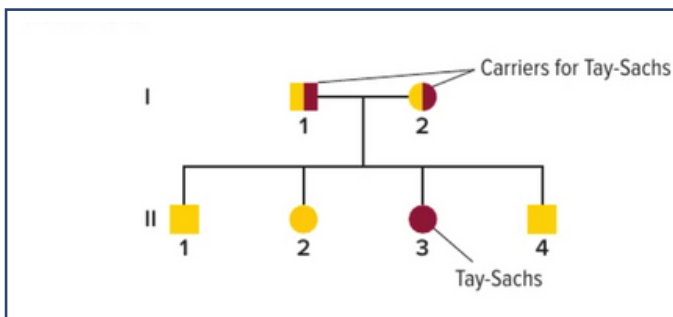


Figure 16

This pedigree illustrates the inheritance of the recessive disorder Tay-Sachs disease.

Note:

- Two unaffected parents (I1 and I2)
- Can have an affected child (II3)

Dominant Genetic Disorders

The pedigree in **Figure 17** shows the inheritance of the dominant genetic disorder polydactyly (extra fingers or toes -Pah lee or tuh lee).

Dominant inheritance logic:

- The trait is expressed when **at least one** dominant allele is present (Pp or PP).

If one parent is unaffected (pp) and the other parent is affected (P_), the child's possible genotypes are:

- Pp (heterozygous, affected with polydactyly)
- pp (homozygous recessive, unaffected)

- Each unaffected person is homozygous recessive (pp).

Example in Figure 17:

- Individual I2:
 - Has polydactyly → shown by a dark circle.
 - Therefore, she is either:
 - **Homozygous dominant**, or
 - **Heterozygous**
 - Based on her children (II3 and II4) being unaffected:
 - She must be heterozygous (one dominant + one recessive gene)



Inference about II2:

- II6 and II7 are unaffected parents
- Their child (III2) is also unaffected

Question: What can be inferred about II2, based on:

- The phenotype of her parents, and
- Her offspring?

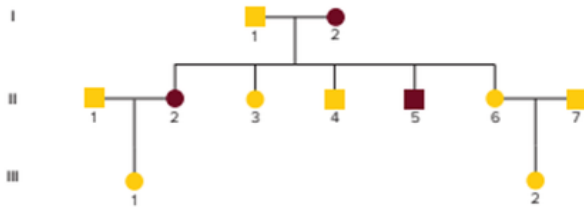


Figure 17

1. This pedigree illustrates the inheritance of a dominant disorder.
2. Note that affected parents can pass on their genes (II2, II5).
3. But unaffected parents cannot have an affected child (III2).

N.B

Person	Sex	Affected?	Genotype
I-1	♂	Unaffected	pp
I-2	♀	Affected	Pp
II-2	♀	Affected	Pp
II-3	♀	Unaffected	pp
II-4	♂	Unaffected	pp
II-5	♂	Affected	Pp
II-6	♀	Unaffected	pp
II-7	♂	Unaffected	pp
III-1	♀ (child of II-2)	Unaffected	pp
III-2	♀ (child of II-6)	Unaffected	pp



Inferring Genotypes

1. General Purpose of Pedigrees:

- Pedigrees are used to infer genotypes from the **observation of phenotypes**.
- By knowing physical traits, genealogists can determine what genes an individual is most likely to have.
- Phenotypes of entire families are analyzed to determine family genotypes.

2. Use in Genetic Counseling:

- Pedigrees help genetic counselors determine whether inheritance patterns are **dominant or recessive**.
- Once the pattern is determined using available data, the genotypes of individuals can be identified through pedigree analysis.

3. How Analysis Is Performed:

- One particular trait is selected to be studied.
- A decision is made whether the trait is dominant or recessive.
- Dominant traits are easier to recognize because they are exhibited in the phenotype of individuals.

4. Understanding Recessive Traits:

- A recessive trait is not expressed unless the person is **homozygous recessive**.
- A recessive allele must be passed on by each parent.

When recessive traits are expressed:

- The ancestry of the person expressing the trait is traced across generations.
- This helps identify which parents and grandparents were carriers of the recessive allele.

Predicting Disorders

1. If good records have been kept within families, the possibility of disorders occurring in future offspring can be predicted.
2. However, more accuracy can be expected if several individuals within the family can be evaluated.
3. Having information about several generations of family members can provide valuable information.
4. The study of human genetics can be difficult because scientists are limited by:
 - Time
 - Ethics
 - Availability of information

Example:

- It takes decades for each generation to mature and have offspring when the study involves humans.
- Therefore, good record keeping, where it exists, helps scientists:
 - Use pedigree analysis
 - Study inheritance patterns
 - Determine phenotypes
 - Ascertain genotypes within a family



Types of Recessive Genetic Disorders

Disorder	Occurrence in the U.S.	Cause	Effect	Cure/Treatment
Albinism	1 in 17,000	Genes do not produce normal amounts of the pigment melanin.	- No color in the skin, eyes and hair - Skin susceptible to UV damage - Vision problems	- No cure - Protect skin from the Sun and other environmental factors - Visual rehabilitation
Cystic fibrosis	1 in 3500	The gene that codes for a membrane protein is defective.	- Excessive mucus production - Digestive and respiratory failure	- No cure - Daily cleaning of mucus from the lungs - Mucus-thinning drugs - Pancreatic enzyme supplements
Galactose mia	1 in 50,000 to 70,000	Absence of the gene that codes for the enzyme that breaks down galactose	- Mental disabilities - Enlarged liver - Kidney failure	- No cure - Restriction of lactose/galactose in the diet
Tay-Sachs disease	1 in 2500 (affects people of Jewish descent)	Absence of a necessary enzyme that breaks down fatty substances	- Buildup of fatty deposits in the brain - Mental disabilities	- No cure or treatment - Death by age 5

Table 2: Recessive Genetic Disorders in Humans

Albinism

- In humans, albinism is caused by altered genes, resulting in the absence of the **skin pigment melanin** in hair and eyes.
- Albinism is also found in other animals.

A person with albinism typically has:

- White hair
- Very pale skin
- Pink pupils

- The absence of pigment in the eyes can cause **vision problems**.
- Although everyone should protect their skin from the Sun's ultraviolet (UV) radiation, individuals with albinism need to be especially careful.





Galactosemia

(Pronounced: guh lak tuh SEE mee uh)

Definition: A disorder characterized by the inability of the body to digest galactose.

Process:

During digestion, lactose (from milk) breaks down into:

- Galactose
- Glucose (used by the body for energy)

(Galactose is converted into glucose by the enzyme GALT)

Problem:

- Individuals who lack or have defective GALT cannot digest galactose.

Management:

- These individuals should avoid milk products.



Cystic Fibrosis

Definition & Prevalence

- A common recessive genetic disorder, especially among Caucasians.

Affects:

- Mucus-producing glands
- Digestive enzymes
- Sweat glands

Cause & Pathophysiology

Chloride ions are not properly transported out of cells.

This leads to:

- High intracellular (in cells) chloride
- Poor water diffusion from cells (doesn't exist)
- Thick mucus secretion

Effects on the Body

Thick mucus:

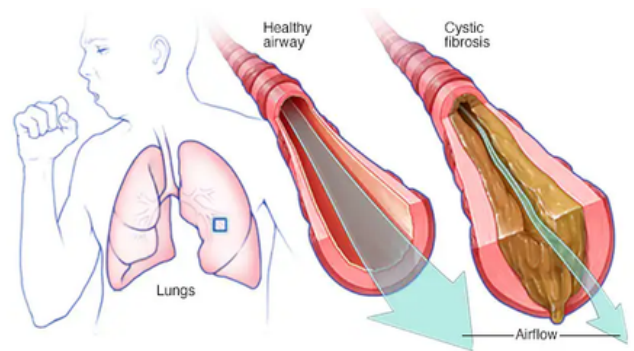
- Clogs ducts in the pancreas
- Interrupts digestion
- Blocks tiny airways in the lungs
- Increases the risk of lung infections due to mucus buildup.

Treatment Options

- Physical therapy
- Medication
- Special diets
- Replacement digestive enzymes

Other Notes

- Genetic tests are available to determine if a person is a carrier.





Tay-Sachs Disease

(Pronounced: TAY saks)

General Info

- Tay-Sachs is a recessive genetic disorder in humans.
- The gene is located on chromosome 15.
- Often identified by a **cherry-red spot** on the back of the eye.
- Most common in Jews of eastern European descent.

Cause

- Caused by absence of enzymes that break down fatty acids called gangliosides.

Normal Process

- **Gangliosides are:**
 - Made naturally in the brain.
 - Later broken down as the brain develops.

In Tay-Sachs

- Gangliosides are **not broken down**.
- They accumulate in the brain.
- **This causes:**
 - Inflation and damage to brain nerve cells.
 - Mental deterioration.



Types of Dominant Genetic Disorders

- Some human disorders — such as the rare disorder Huntington's disease — are caused by dominant alleles.
- This means that:
 - Individuals who do not have the disorder are homozygous recessive for the trait.
- Review **Table 3** as you read about several dominant genetic disorders that occur in humans.



Disorder	Occurrence in the U.S.	Cause	Effect	Cure/Treatment
Huntington's disease	1 in 10,000	A gene affecting neurological function is defective.	• Decline of mental and neurological functions • Ability to move deteriorates	No cure or treatment
Achondroplasia	1 in 25,000	A gene that affects bone growth is abnormal.	• Short arms and legs • Large head	No cure or treatment

Table 3
Dominant Genetic Disorders in Humans

Huntington's disease

Type

Dominant genetic disorder affecting the nervous system

Prevalence

Affects 1 in 10,000 people in the U.S.

Symptoms

Appear between ages 30 and 50

Include:

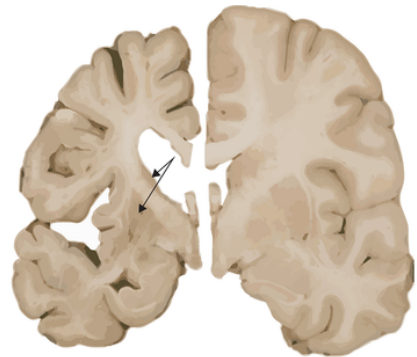
- Gradual loss of brain function
- Uncontrollable movements
- Emotional disturbances

Diagnosis

Genetic tests are available to detect the dominant allele

Treatment

No preventive treatment or cure exists



Huntington Disease Brain

Normal Brain



Achondroplasia

Pronounced: a kahn droh PLAY zhee uh

Key Features

- **Affected individuals have:**
 - Small body size
 - Short limbs (comparatively)
- **It is the most common form of dwarfism.**

Adult Characteristics

- Adult height $\approx$ about 4 feet
- Normal life expectancy

Inheritance

- 75% of individuals with achondroplasia are born to parents of average size.

Cause

- When parents are average-sized, the condition is likely due to a new mutation or genetic change.

