الوراثة الطبية 1 / الكورس الاول

Family Pedigree: Symbols and Interpretation

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الجانب النظري : الوراثة الطبية

Theoretical : Medical Genetics

محاضرة رقم 8

Lecture No. 8

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Learning Objectives

By the end of this lecture, you will be able to:

- Define the concept of pedigree in medical genetics.
- Recognize the standard symbols used in drawing a family pedigree.
- Understand how to trace genetic traits or diseases through generations.
- Analyze patterns of inheritance (dominant, recessive, sex-linked, multifactorial) using pedigree charts.
- Apply these concepts to real-world examples of genetic diseases.

Introduction

- The **family pedigree** is one of the most essential tools in **clinical genetics**. It allows clinicians and researchers to trace the transmission of **genetic traits or inherited diseases** across generations.

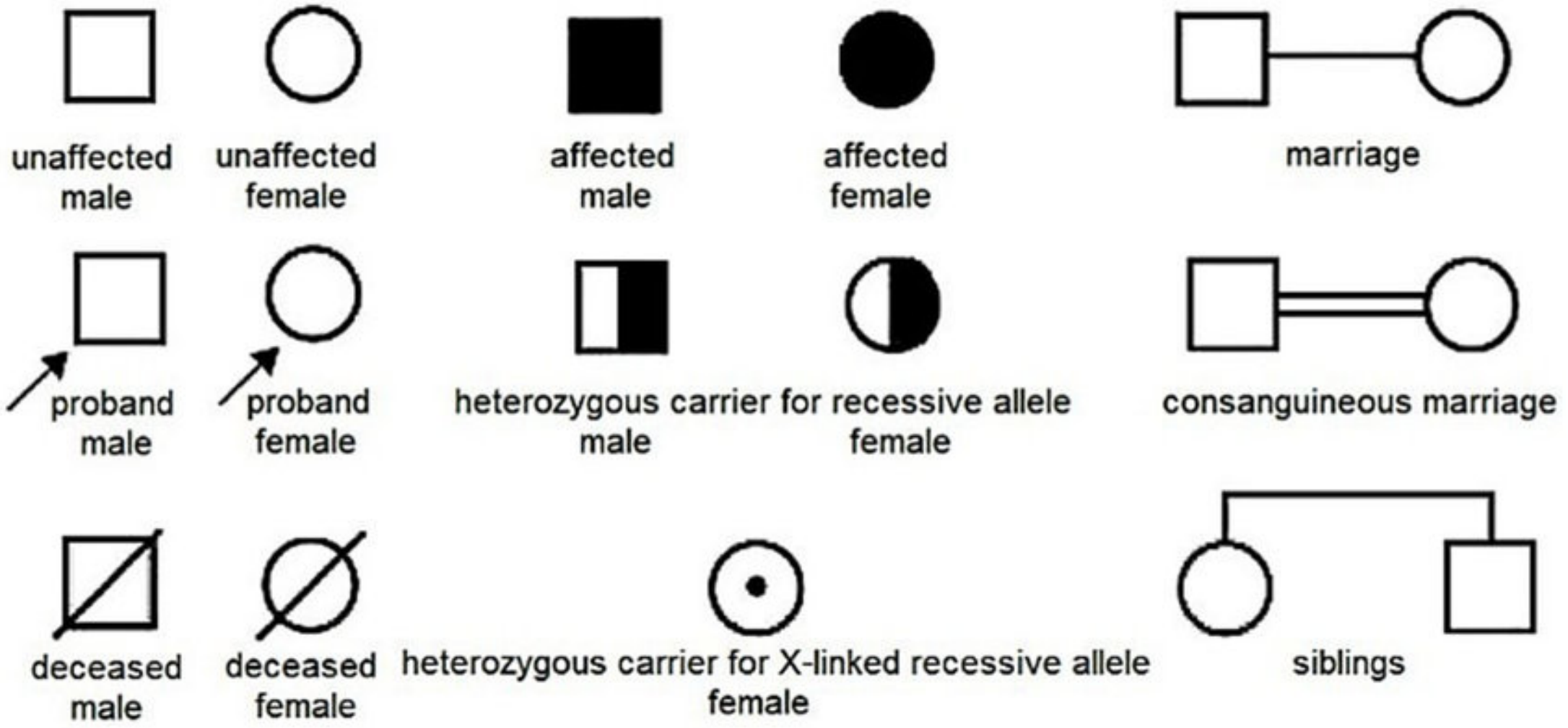
  A pedigree uses **standard symbols** universally accepted to represent individuals, gender, relationships, and disease or carrier status.

- **Pedigree analysis** is widely used in:
  - Diagnosing **hereditary diseases**.
  - Determining the **pattern of inheritance** (dominant, recessive, sex-linked).
  - Estimating the **risk of recurrence** in future generations.
  - Performing **genetic research** and family studies.

I. Definition of Pedigree

- A **pedigree** is a graphical chart that shows the **genetic and familial relationships** among individuals, usually drawn as a **family tree** beginning with the **oldest generation (grandparents)** and extending to the **current generation**.
- Each generation is labeled with a **Roman numeral (I, II, III, ...)**
- Each individual within a generation is given an **Arabic number (1, 2, 3, ...)**.
- **For example:**

  The third individual in the second generation is written as **II-3**.



Standard Pedigree Symbols

III. How to Read a Pedigree

When analyzing a pedigree:

- **Start from the upper generation** (the grandparents).
- **Identify the affected individuals** using shaded symbols.
- **Trace the disease transmission** through generations.
- Observe whether the disease appears in **both sexes or one sex only**.
- Determine whether the disease **skips generations** (recessive) or **appears in every generation** (dominant).

IV. Steps in Drawing a Pedigree Chart

- **Start with the oldest generation** at the top (grandparents).
- **Connect couples with a horizontal line.**
- **Draw vertical lines** leading to their children.
- **Use appropriate symbols** to represent sex and disease status.
- **Assign numbers** to indicate the order of individuals.
- **Add a key (legend)** explaining all symbols used.

V. Patterns of Inheritance Identified by Pedigree Analysis

1. Autosomal Dominant

- Appears in every generation.
- Affects both males and females equally.
- Each affected individual usually has an affected parent.
- **Examples:** *Huntington's disease*, *Familial hypercholesterolemia.*

2. Autosomal Recessive

- May skip generations.
- Affects both males and females equally.
- Often occurs in **consanguineous marriages**.
- **Examples:** *Cystic fibrosis*, *Sickle cell anemia.*

3. X-linked Dominant

- Affects both sexes but females are often more affected.
- An affected father transmits the condition to **all daughters** but **no sons**.
- **Examples:** *Rett syndrome.*

4. X-linked Recessive

- More commonly observed in **males**..
- Females are usually **carriers** without symptoms.
- An affected father **does not transmit** the disease to his sons.
- **Examples:** *Hemophilia A.*

5. Mitochondrial Inheritance

- Transmitted **only from the mother**.
- An affected mother can pass the condition to **all her offspring**, with variable expression.
- **Example:** *Leber's hereditary optic neuropathy (LHON).*

VI. Example (Case Analysis)

- **Case 1:**

  A hereditary disease appears in **every generation**, affects **both sexes equally**, and **each affected person has an affected parent.**

✔ **Interpretation:** The pattern is **Autosomal Dominant Inheritance.**

- **Case 2:**

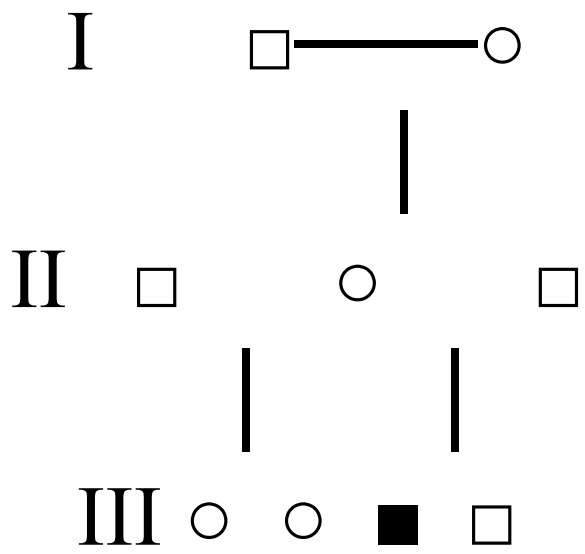
  A disease appears **only in males**, while mothers are healthy but have affected sons.

✔ **Interpretation:** The pattern is **X-linked Recessive Inheritance.**

VII. Importance of Pedigree in Medical Practice

- Aids in **diagnosing genetic disorders.**
- Estimates **risk of inheritance or recurrence.**
- Provides essential information for **genetic counseling**.
- Helps in selecting **appropriate genetic testing.**
- Useful in **population and family genetics research.**

Example of a Simple Family Pedigree



Understanding Genotypes and Phenotypes

- In genetics, every trait is controlled by a pair of alleles, which are inherited from the two parents.
- One allele may be dominant (symbolized by a **capital letter**, A) and the other recessive (**small letter**, a).

Example/ eyes color

Genotype	Phenotype
BB	brown eyes
Bb	brown eyes
bb	blue/green eyes

- **Dominant allele (A)** masks the effect of the other allele.

Consequently, a person whose genotype is either homozygous dominant (AA) or heterozygous (Aa) will show the dominant phenotype.

- **Recessive allele (a)** is only expressed when both copies are present (aa).

by contrast, can influence the phenotype only when both copies are recessive, i.e. in the homozygous recessive state (aa).

Example

❖ **Trait: Human hair color**

- Allele B (brown) = dominant
- Allele b (blond) = recessive

1. What hair color is expected for genotype BB?
2. What hair color is expected for genotype Bb?
3. What hair color is expected for genotype bb?
4. How many copies of allele b are needed for a person to show blond hair?
5. Write the only genotype that gives blond hair.
6. List all genotypes that give brown hair.
7. Can two brown-haired parents have a blond child? Explain with genotypes.
8. Bb male couples with Bb female. Give the expected genotype and phenotype of each child.

Example of a Simple Family Pedigree

Q/ A pair of heterozygous parents (Aa × Aa) are expecting four children.
Albinism (autosomal-recessive) is only seen in the homozygous-recessive genotype.

1. Which genotype is **affected**?
2. Write the full genotype **and** corresponding phenotype (affected / unaffected-carrier / unaffected-non-carrier) for each of the four possible offspring.

- Answer:
- The **affected** genotype is: **aa**
- The four possible offspring:

Genotype	Phenotype
AA	**unaffected (non-carrier)**
Aa	**unaffected (carrier)**
Aa	**unaffected (carrier)**
aa	**affected**

Punnett Square:

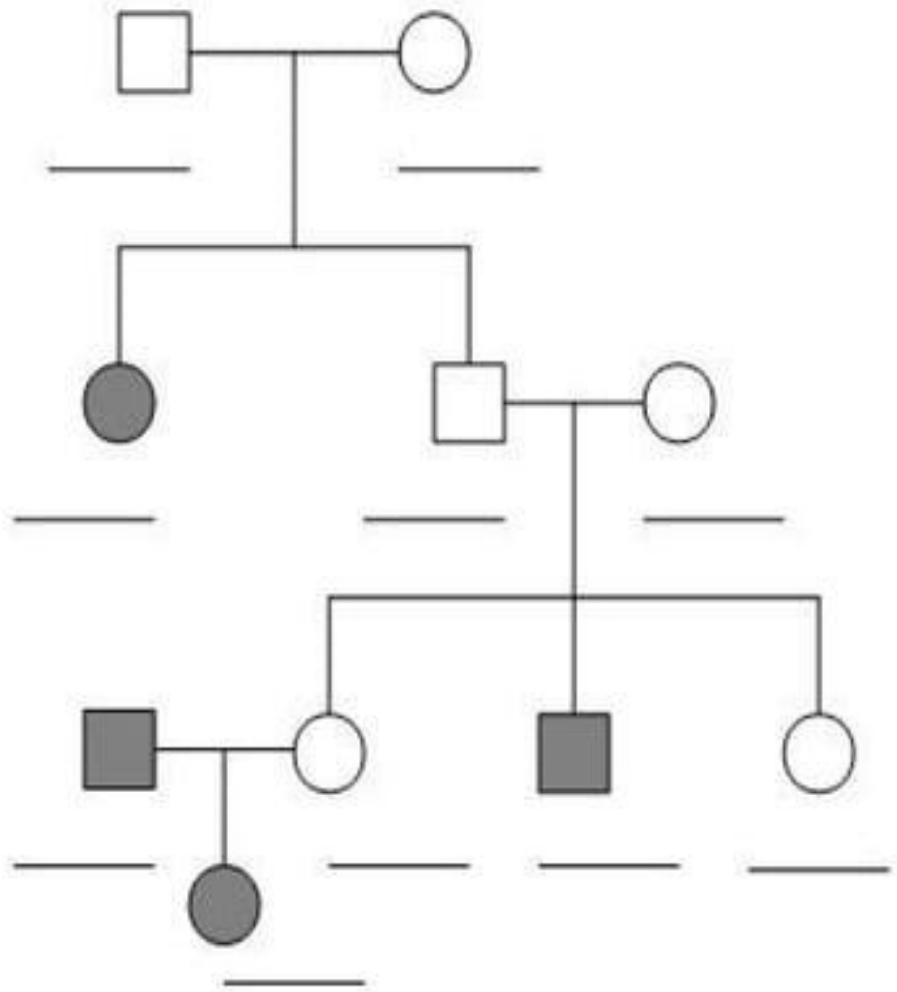
	A	**a**
A	AA	Aa
a	Aa	aa

Example of a Simple Family Pedigree

Question: The pedigree below shows the inheritance of albinism, an autosomal recessive disorder.

Using A = dominant allele and
a = recessive allele.

1. How many children does this family have?
2. What are the sexes of the children?
3. Fill in the genotypes (AA, Aa, or aa) for every individual shown in the pedigree.



Example of a Simple Family Pedigree

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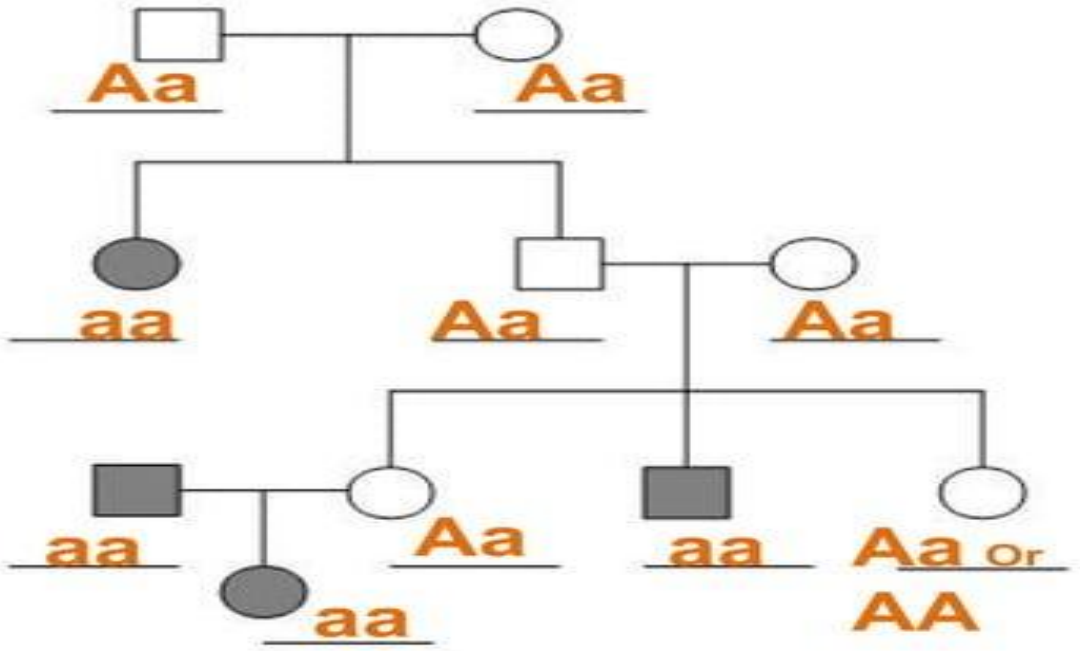
Answers

3. How many children does this family have? **2**
4. What are the sexes of the children? **Female, Male**
5. Fill out the blanks of the pedigree below (AA, Aa, aa)



Any questions?