



Pattern of inheritance II

Sep 6, 2022

Objectives

By the end of this session, the students should be able to:

1. Identify and recognize various mode of inheritance from family tree
2. Analyse the family pedigree to determine mode of inheritance
3. Rationalize the mode of inheritance in each illustrated pedigree



Pedigree Analysis

When you analyse a pedigree ask yourself the following:

1. Are both males and females affected? only males? only females? (autosomal vs sex-linked)

If both → AR or AD or mitochondrial

If males → X-linked recessive or Y-linked

If more females → X-linked dominant

2. Does disorder skip generation? Or seen in all generations? (dominant vs recessive)

3. Is transmission through mother or father? Is there male to male transmission? (to differentiate between Y-linked vs mitochondrial vs x-linked)

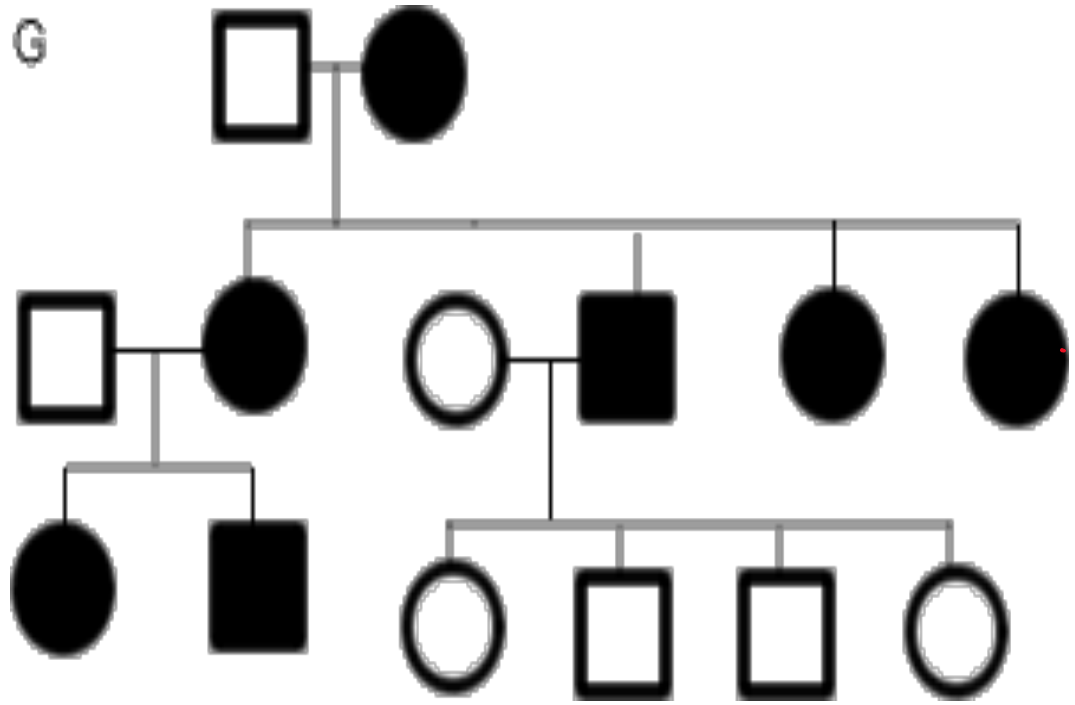
Steps to interpret a family pedigree

A-Determine if the pedigree chart shows an **autosomal** or **sex-linked** disease:

- If most of the males in the pedigree are affected, then the disorder is **sex-linked**
- If it is a **50/50** ratio between men and women the disorder is **autosomal**.

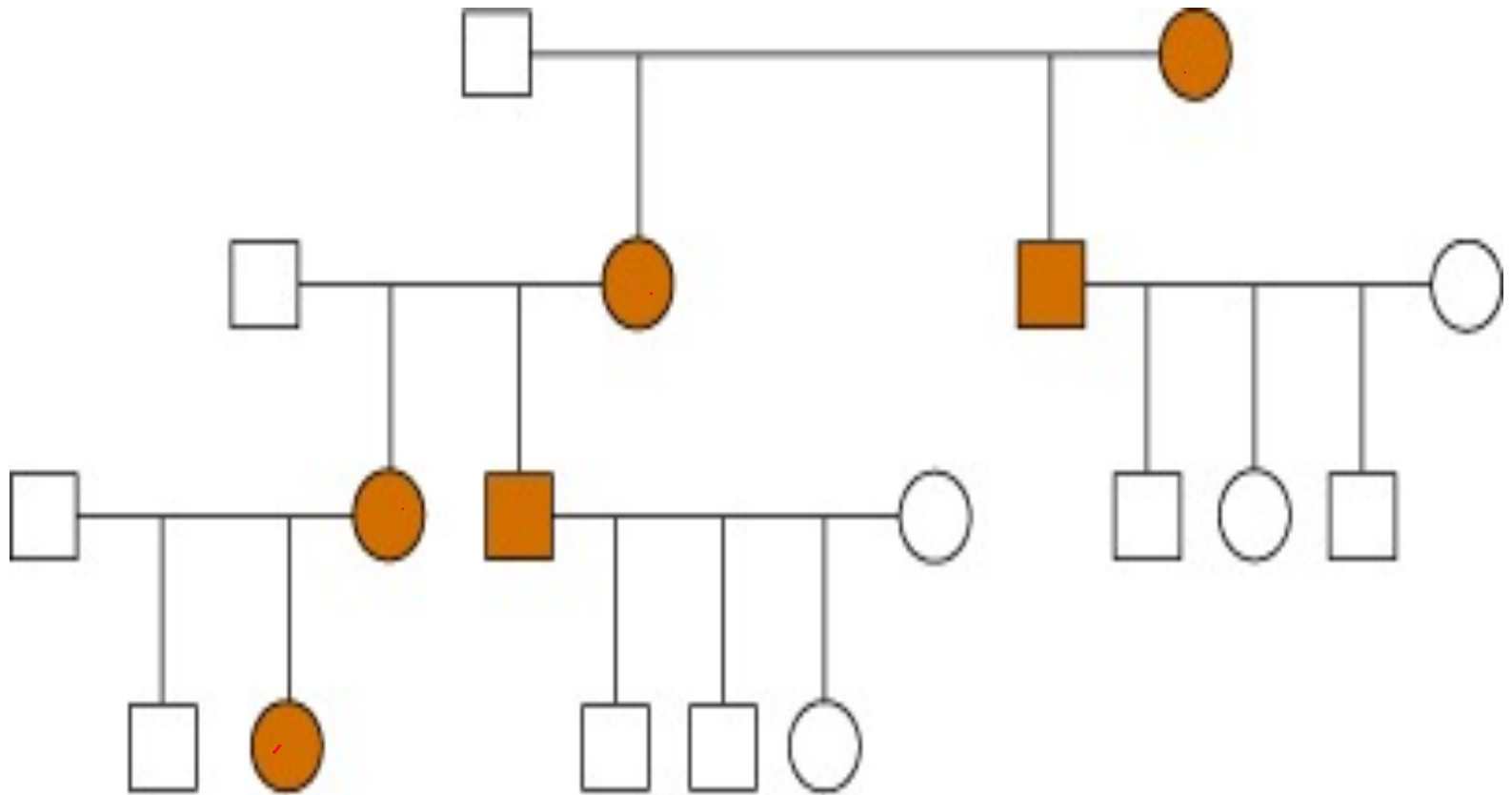
B-Determine whether the disorder is **dominant** or **recessive**:

- If the disorder is **dominant**, **one** of the **parents must** have the **disorder**.
- If the disorder is **recessive**, neither **parent** is affected because they are **heterozygous**.



C-Transmission:

- If the disorder is transmitted through **mother i.e NO male to male transmission** → mitochondrial or X-linked dominant or X-linked recessive
- If the disorder is transmitted through **father i.e male to male transmission** → Y-linked



Pedigree showing transmission and expression of a **mitochondrial** trait. Note that **transmission occurs only through females** .

Rules of inheritance

Autosomal Recessive

- Appears in both sexes with equal frequency
- Trait tend to skip generations

Affected offspring are usually born to unaffected parents

- When both parents are heterozygous, approx. $1/4$ of the progeny will be affected
- Appears more frequently among the children of consanguine marriages

Rules of inheritance

Autosomal Dominant

- Appears in **both sexes** with **equal frequency**
- Both sexes transmit the trait to their offspring
- Does **not skip generations**
- Affected offspring must have an affected parent **unless** they **posses a new mutation**
- When **one parent is affected** (het.) and the **other** parent is **unaffected**, approx. **1/2** of the offspring will be **affected**
- Unaffected parents do not transmit the trait

Rules of inheritance

X-Linked Dominant

- Both males and females are affected; often more females than males are affected
- Does not skip generations .
 - Affected sons must have an affected mother ;
 - affected daughters must have either an affected mother or an affected father
- Affected fathers will pass the trait on to all their daughters
- Affected mothers if heterozygous will pass the trait on to $1/2$ of their sons and $1/2$ of their daughters

Rules of inheritance

X-Linked Recessive

- More males than females are affected
- Affected sons are usually born to unaffected mothers, thus the trait skips generations
- •Approximately $1/2$ of carrier mothers' sons are affected
- •It is never passed from father to son
- •All daughters of affected fathers are carriers

Rules of inheritance

Y-Linked Dominant

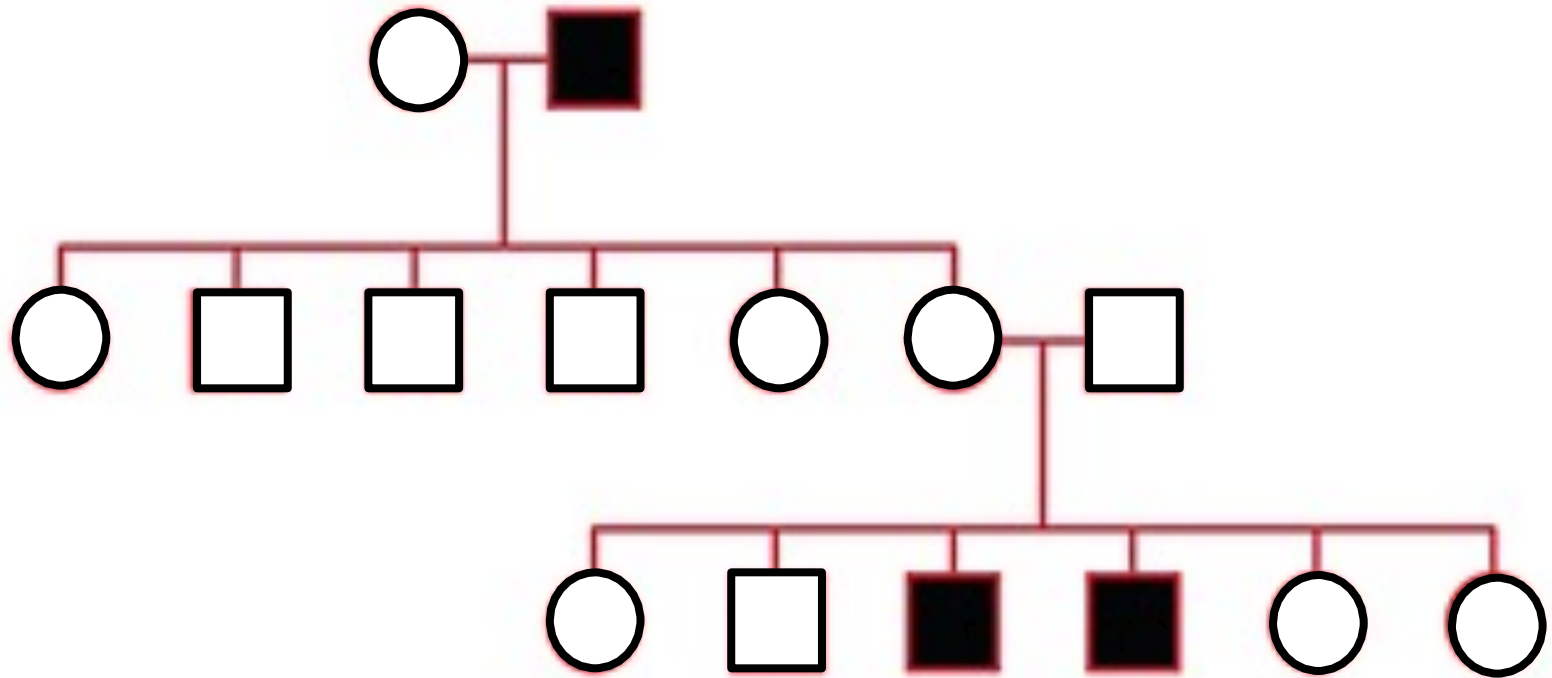
- Only males are affected
- It is passed on from father to all sons
- It does not skip generations

Mitochondrial

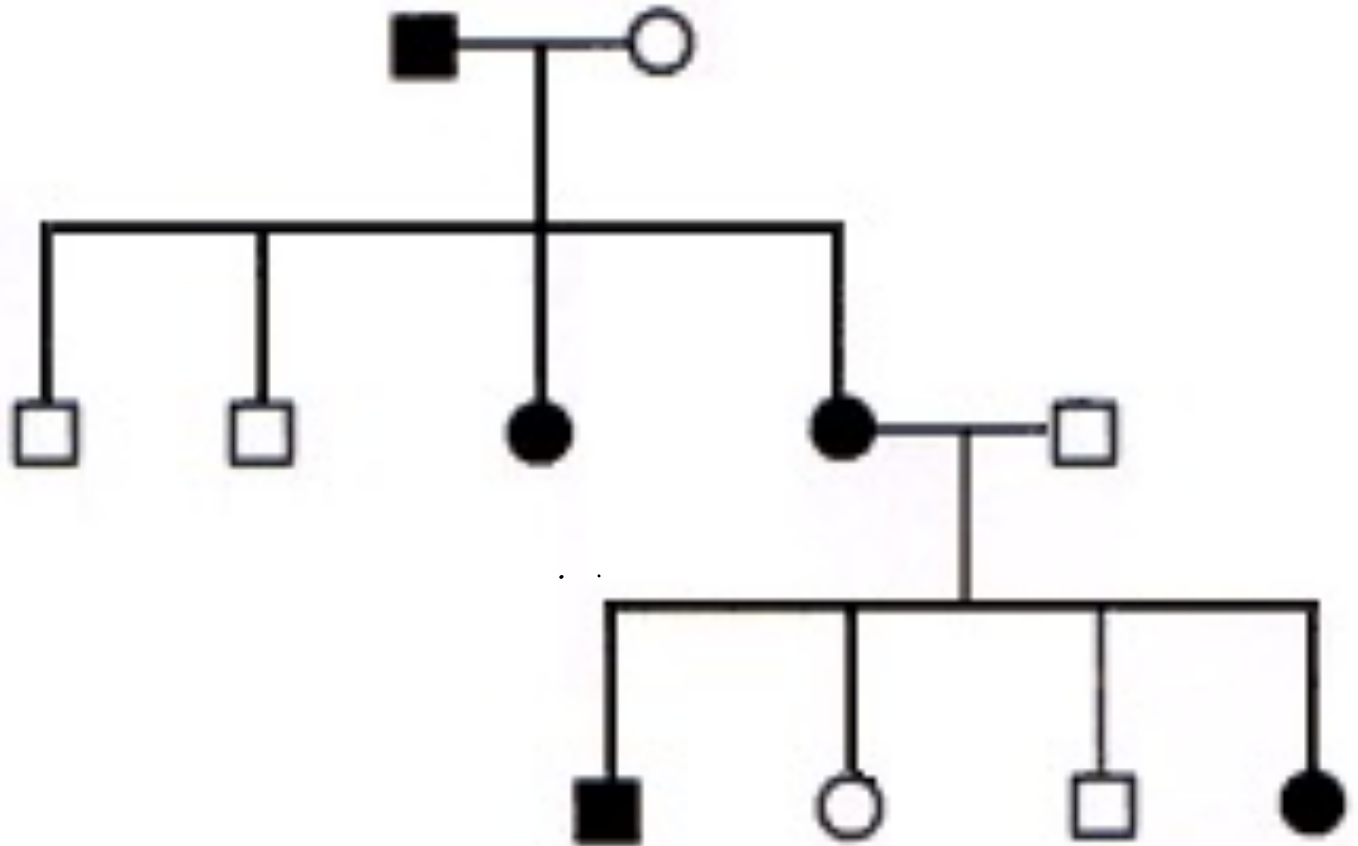
- Trait is inherited from mother only
- All children of a mother are at risk to be affected or carriers

Examples

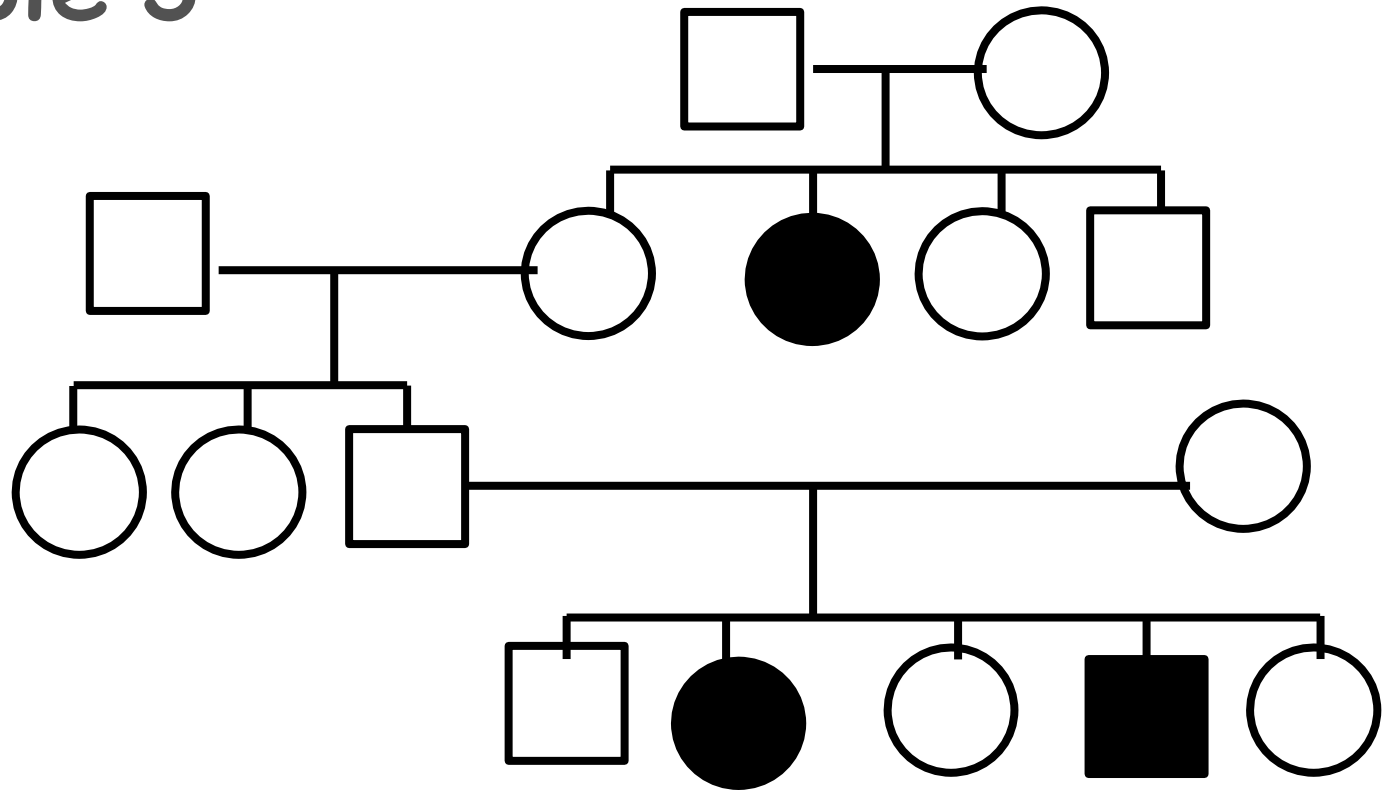
Example 1



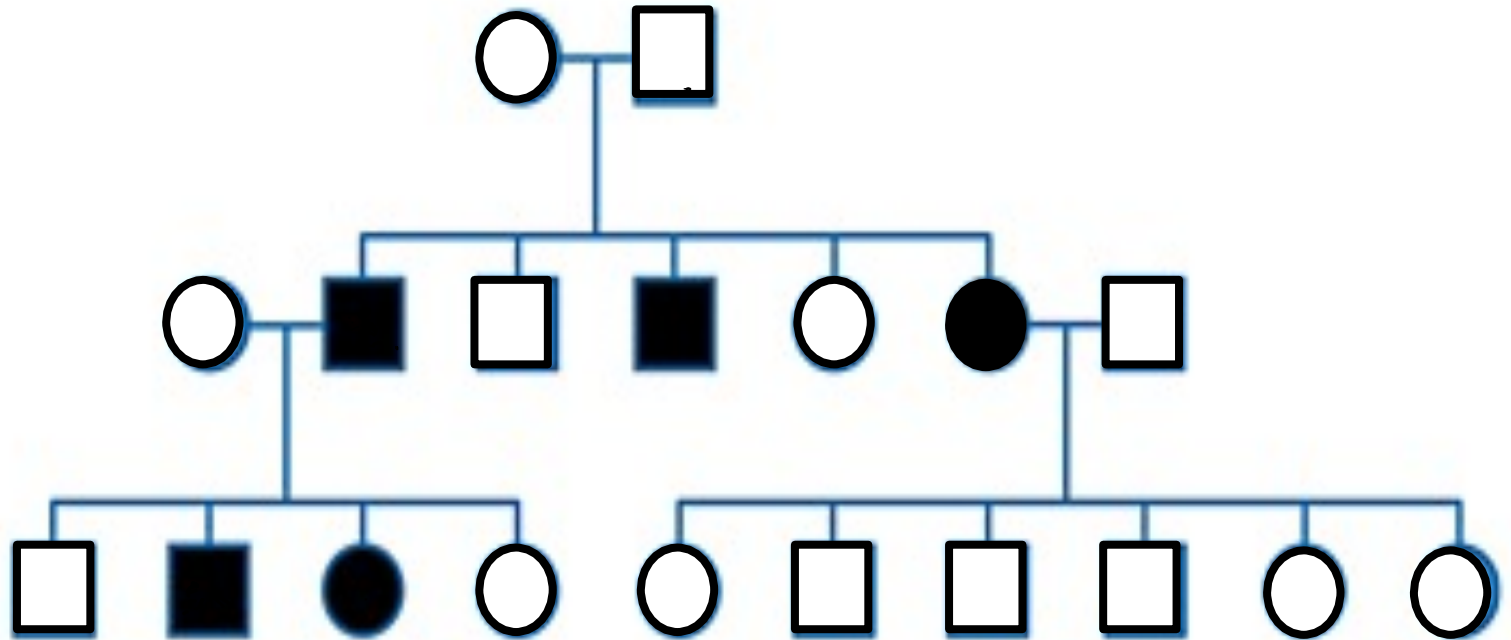
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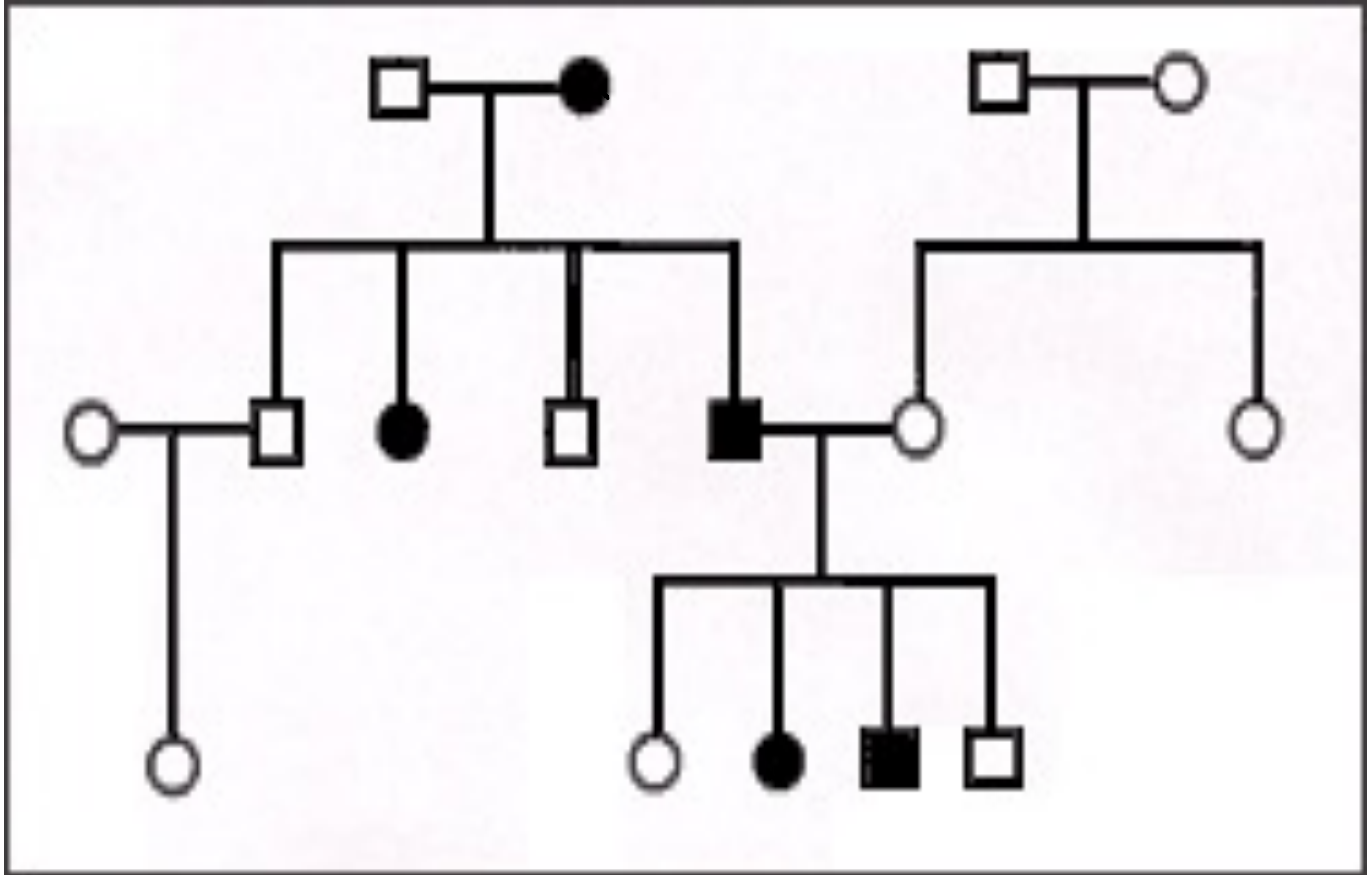
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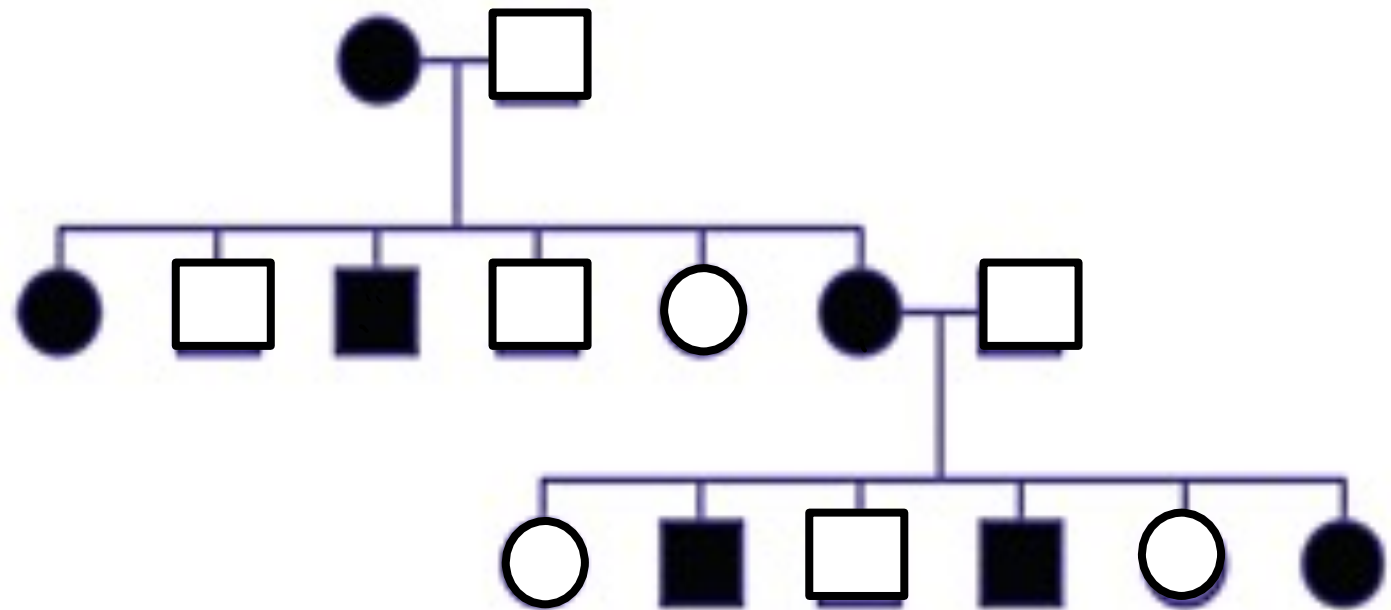
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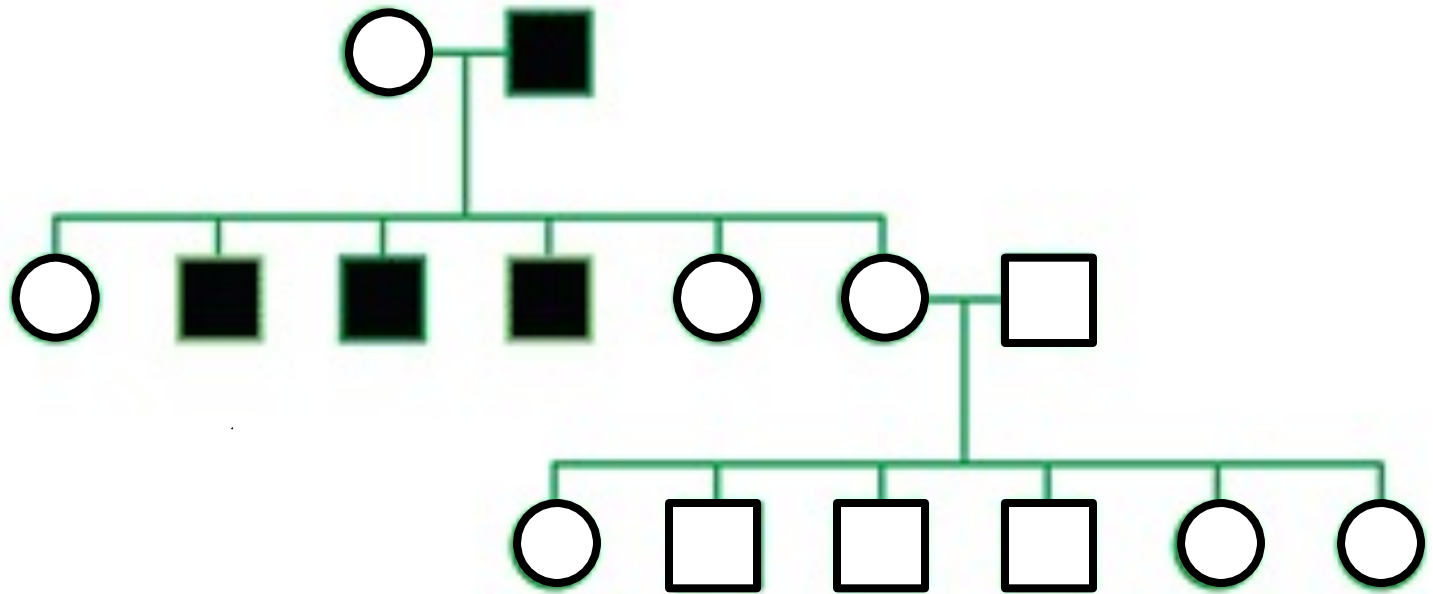
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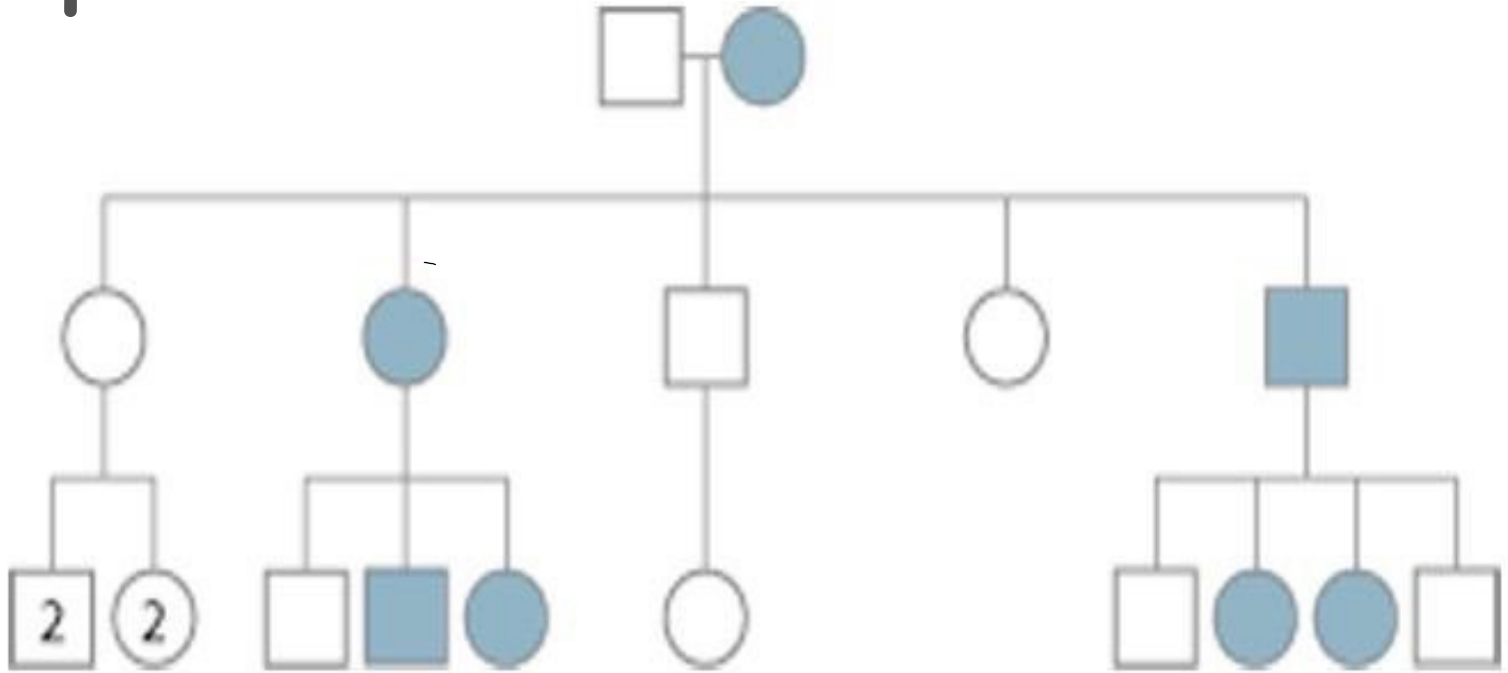
Example 6



Example 7



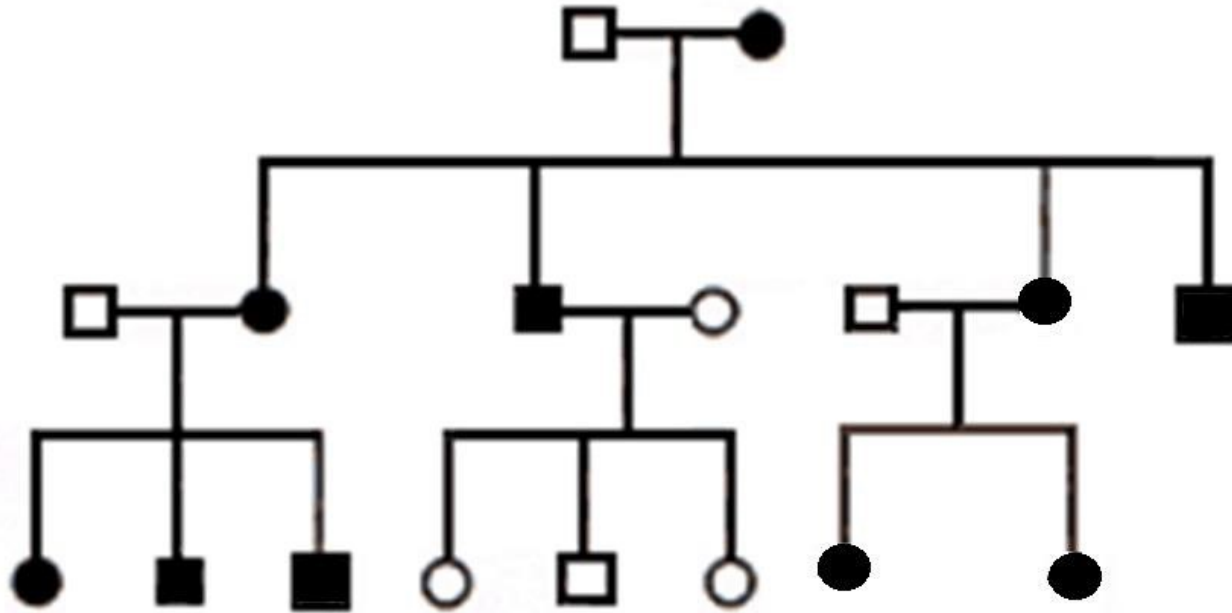
Example 8



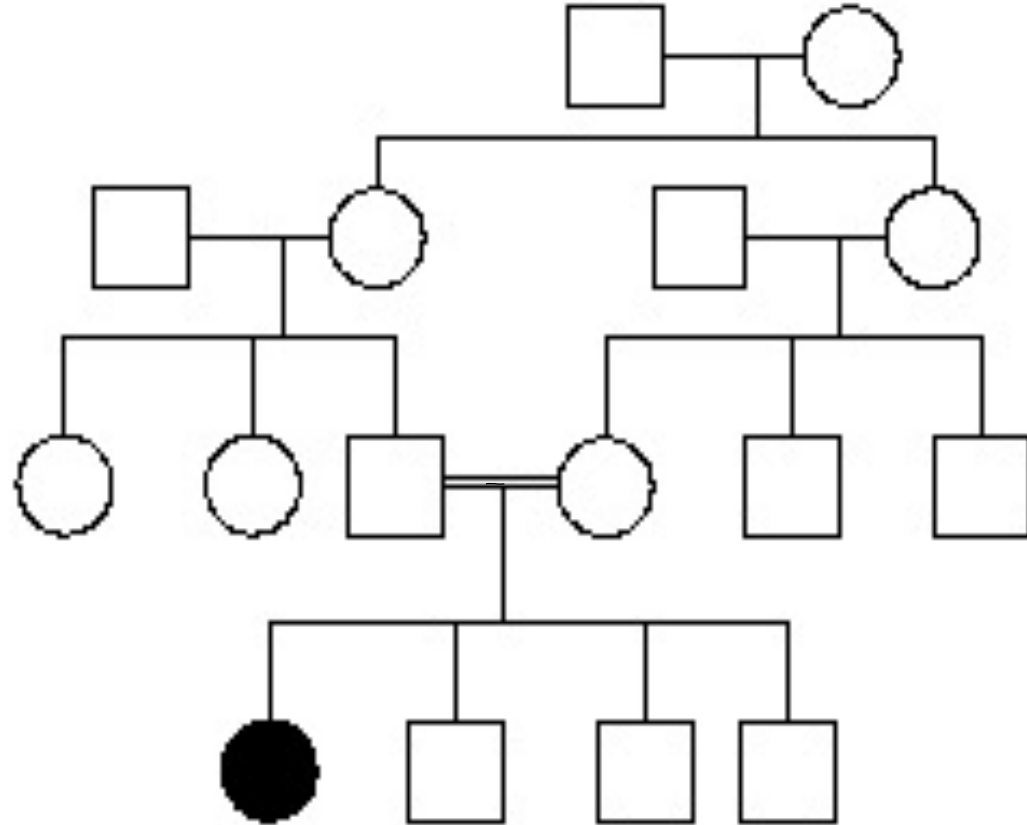
Example 9



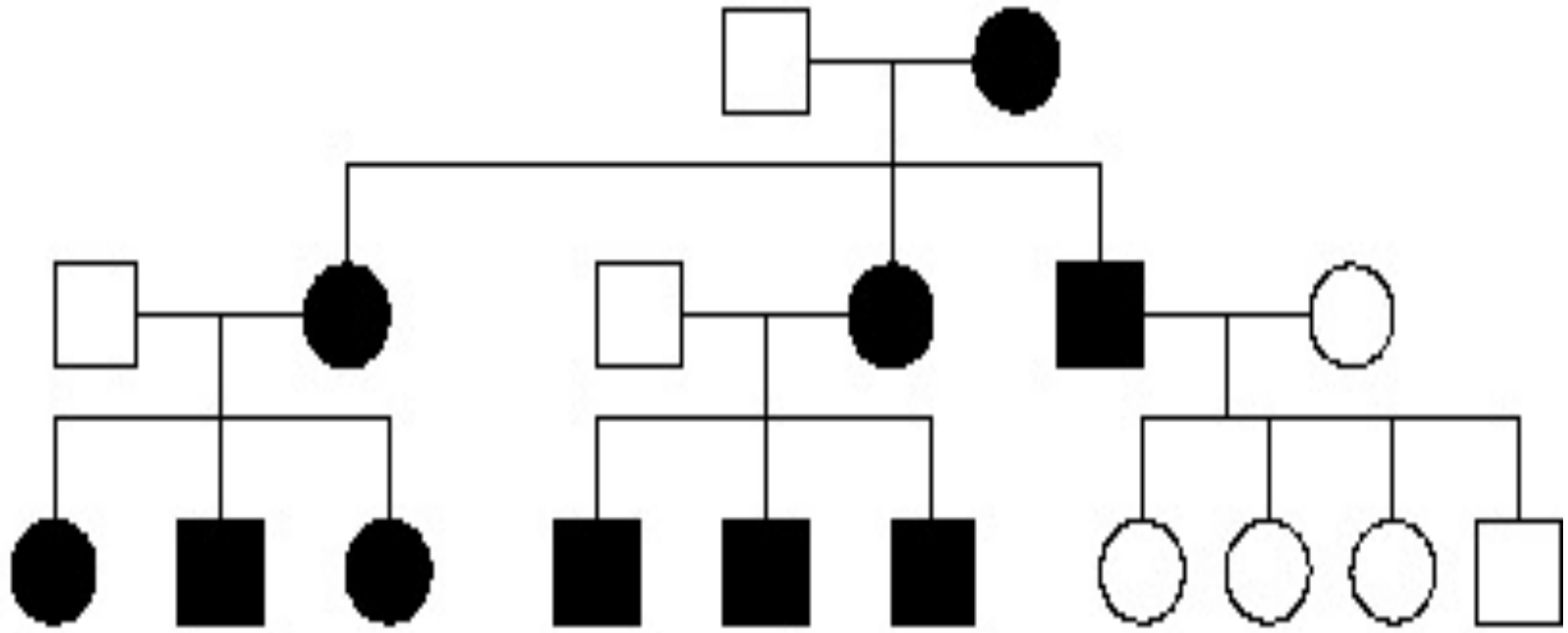
Example 10



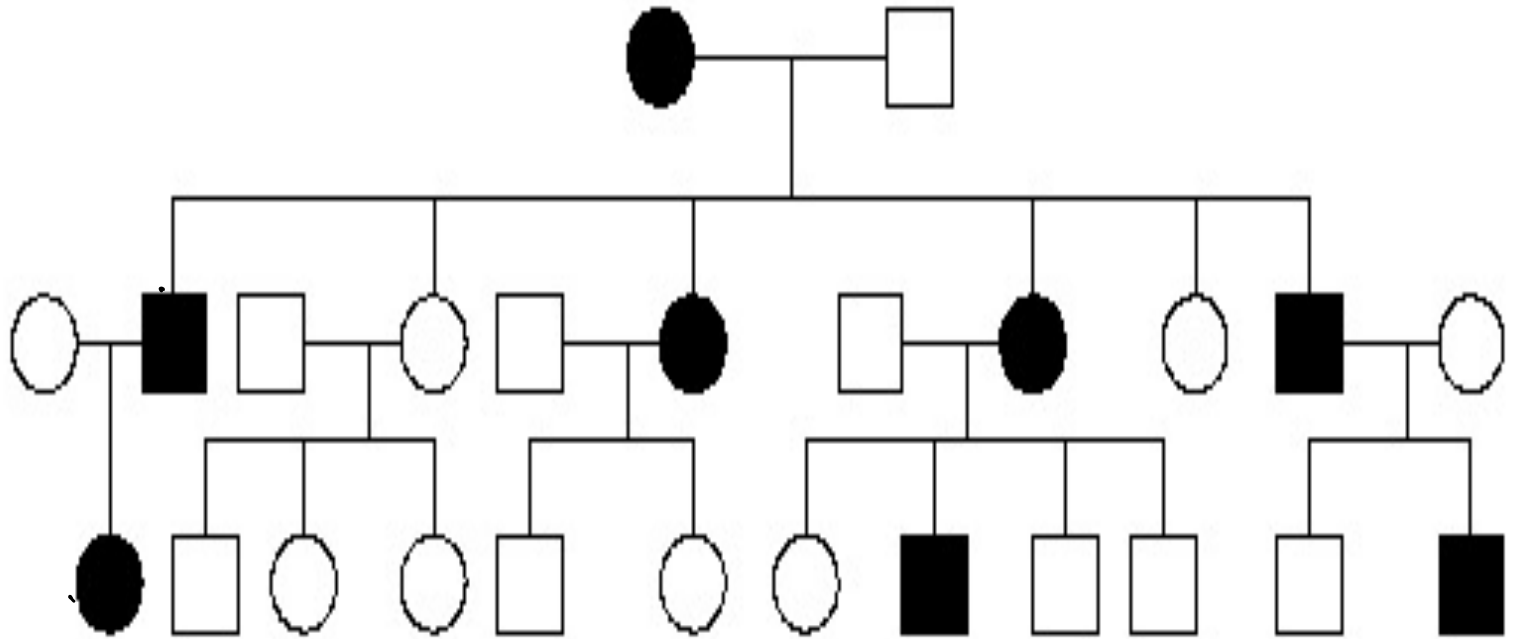
Example 11



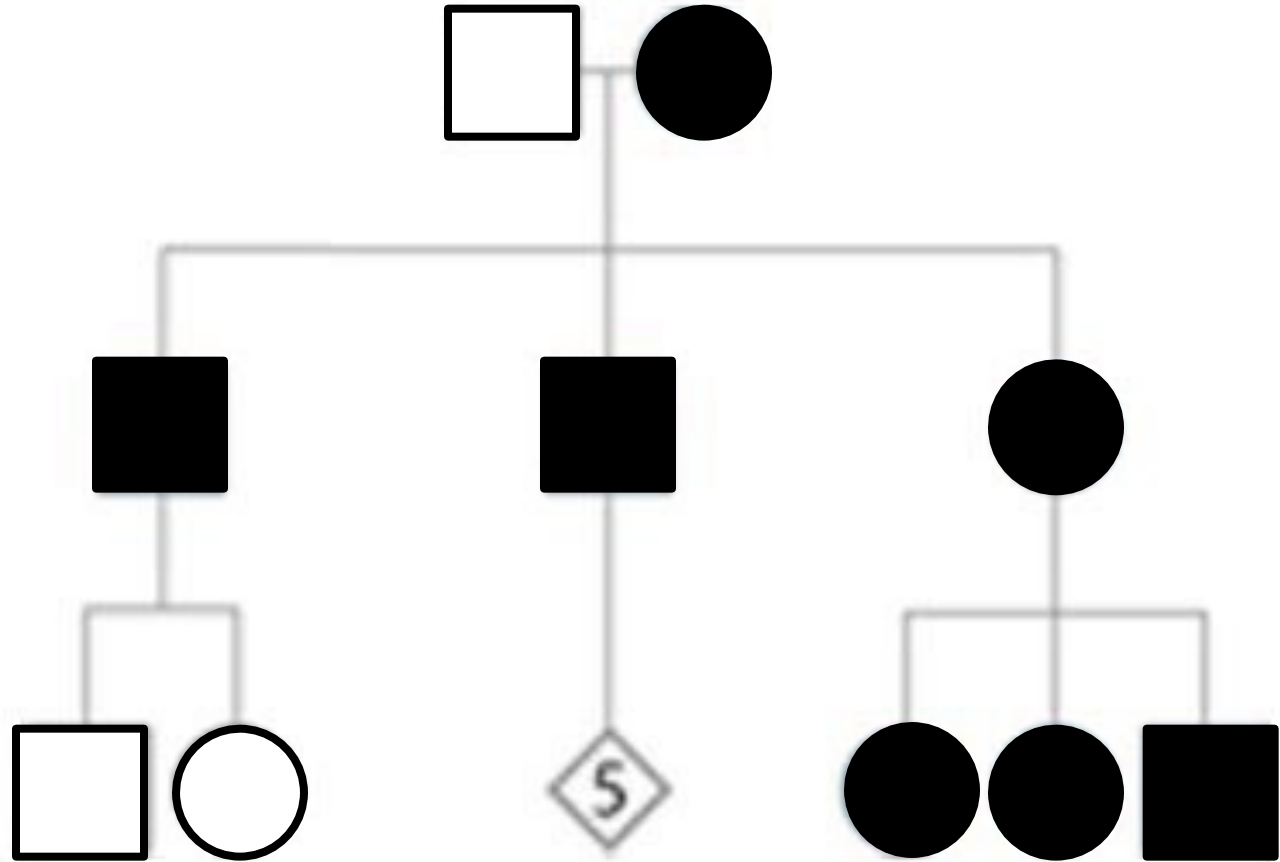
Example 12



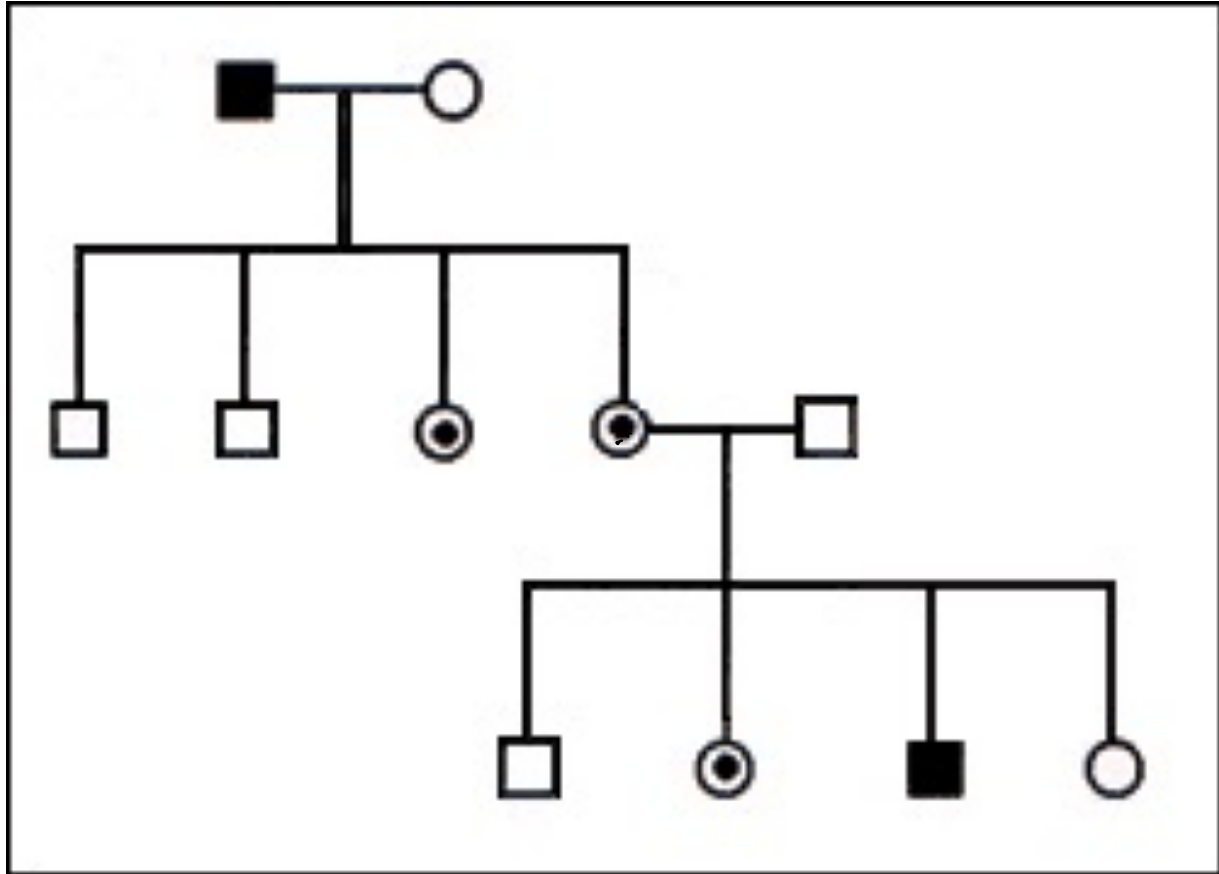
Example 13



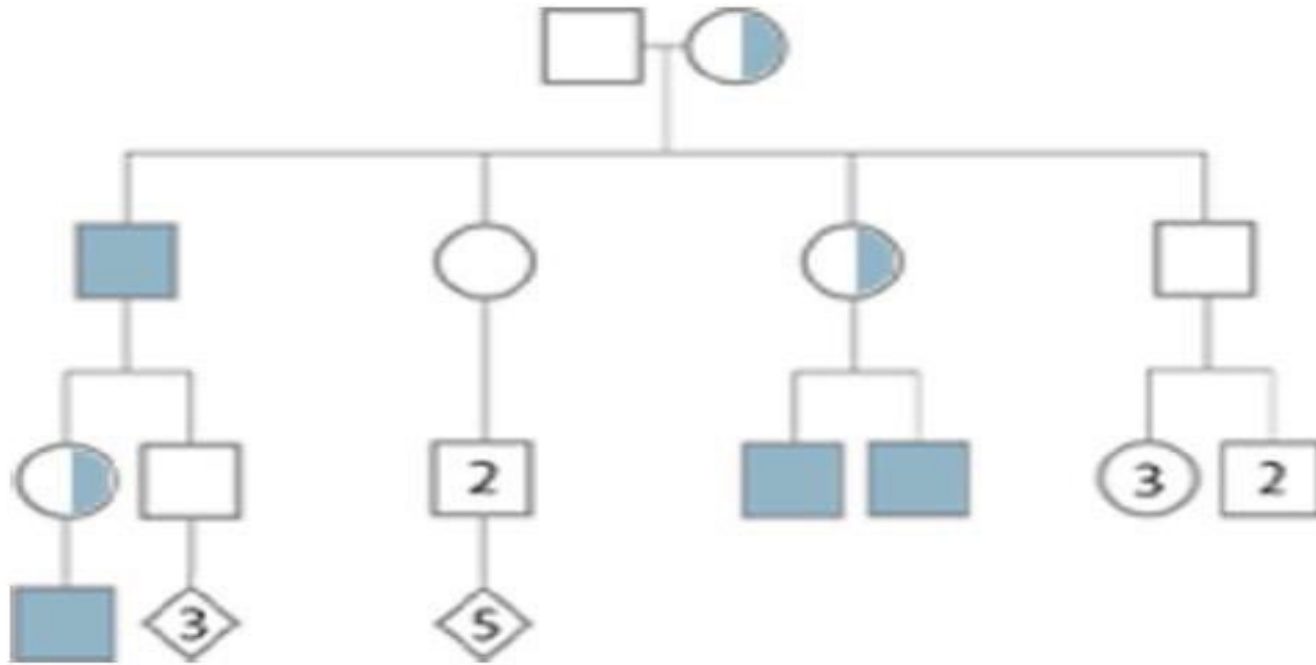
Example 14



Example 15



Example 16



Answers:

1	X-Linked Recessive
2	X-Linked Dominant
3	Autosomal Recessive
4	Autosomal Recessive
5	Autosomal Dominant
6	Autosomal Dominant
7	Y-Linked
8	X-Linked Dominant
9	Autosomal Dominant
10	Mitochondrial
11	Autosomal Recessive
12	Mitochondrial
13	Autosomal Dominant
14	Mitochondrial
15	X-Linked Recessive
16	X-Linked Recessive

References

- Standardized human pedigree nomenclature: update and assessment of the recommendations of the National Society of Genetic Counselors. Robin Bennett et al. J Genet Couns. 2008 Oct;17(5):424-33.



THANK YOU