SAWA UNIVERSITY

الوراثة الطبية 1 / الكورس الاول

Dominant Inheritance and Recessive inheritance

جامعـــــــة ســـــــــاوة الاهلية — Sawa University

كليـــــة التقنيـــات الصحيـة والطبيـة — College of health and medical techniques

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المرحلـــــــــــــة الثالثة — Third Stage

الجانب النظري : الوراثة الطبية

Theoretical : Medical Genetics

محاضرة رقم 11 و 12

Lecture No. 11 +12

تدريسي المادة : م. د عباس علي

Learning Objectives

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

Differentiate between dominant and recessive alleles and describe how each contributes to trait expression.

Dominant Inheritance

- Dominant inheritance occurs when a single dominant allele is sufficient to express a trait.

1. Genotypes that show the dominant trait:

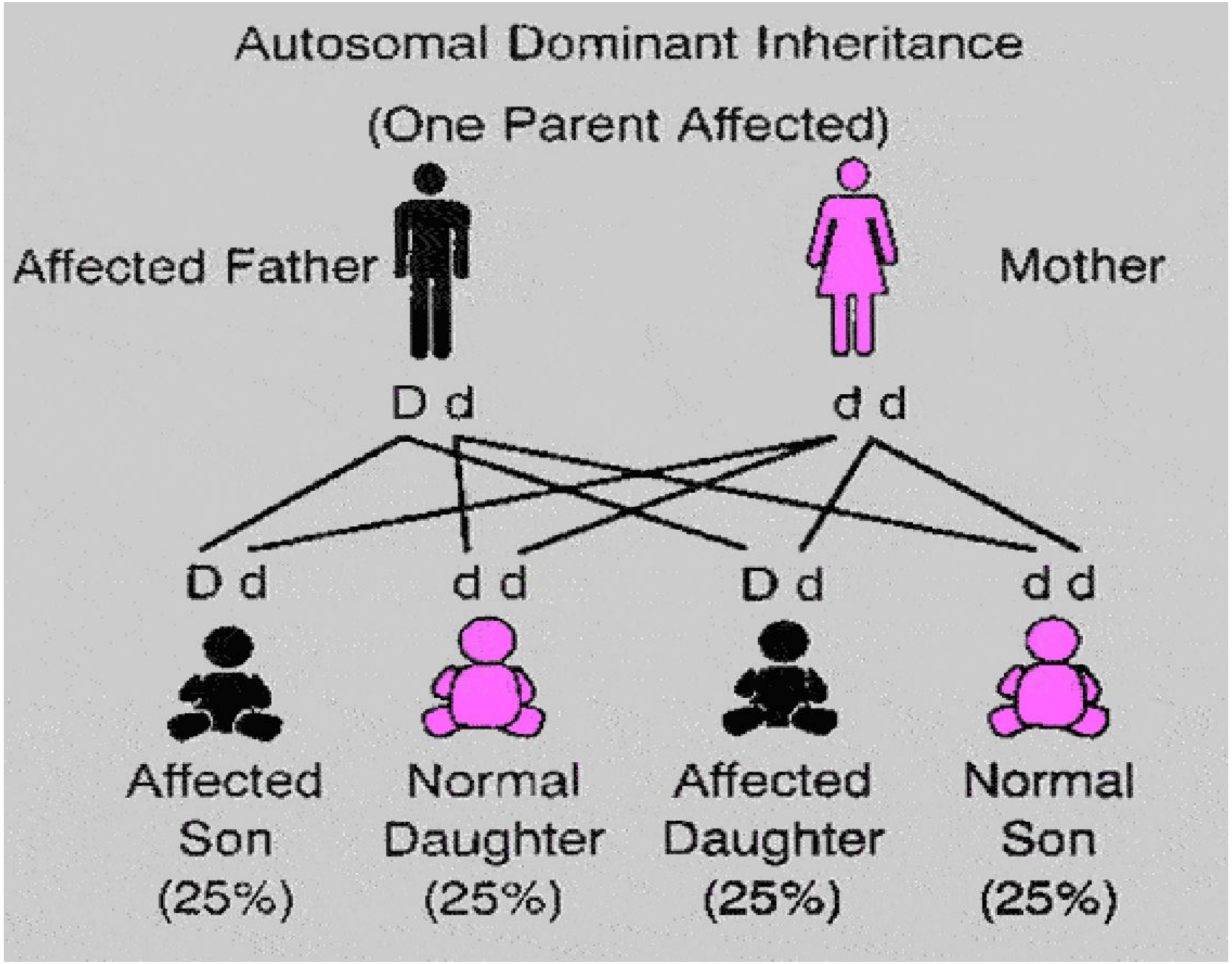
- **AA** → dominant
- **Aa** → dominant

2.Genotypes that show the recessive trait:

- **aa** → recessive (dominant trait absent)

3. Characteristics of Dominant Inheritance

- The trait typically appears in every generation.
- Only one affected parent is needed to pass on the trait.
- Two recessive individuals cannot produce a dominant offspring.
- Affected individuals have at least one dominant allele.


Autosomal Dominant Inheritance
(One Parent Affected)
Affected Father
Mother
D d
d d
D d
d d
D d
d d
Affected Son (25%)
Normal Daughter (25%)
Affected Daughter (25%)
Normal Son (25%)

N*N------NN

N*N N*N NN NN

25% affected 25% affected normal normal

50% 50%

Dominant Inheritance

4- With every autosomal dominant disease, some patients do not have affected parents, so there is new mutation involving either the egg or the sperm from which they derived. Their siblings are neither affected nor at high risk of the disease.

5- Clinical features can be modified by reduced penterance &variable expressivity.

reduced penterance: Some patients inherit the mutant gene but are phenotypically normal.

variable expressivity: If the trait is seen in all individuals carrying the mutant gene but is expressed differently among individuals…

6- In many conditions, signs &symptoms do not appear until adulthood.

7- 50% reduction in normal gene products is associated with clinical symptoms.

Examples of autosomal dominant disorders:

Marfan syndrome

hypercholesterolemia

Polycystic kidney disease

Hereditary spherocytosis

Familial polyposis coli

Autosomal recessive disorders:

Occur when both of the alleles at a given locus are mutants, so characterized by the following features:

1- The trait does not usually affect the parents, but siblings may show the disease.

2- Siblings have 25% risk for each birth (one chance in four).

3- If the mutant gene occurs with a low frequency in the population, there is a strong likelihood that the proband is the product of a consanguineous marriage.

4- The expression of the defect tends to be more uniform than autosomal dominant.

5- Complete penterance is common.

6- Onset of signs &symptoms early in life.

7- New mutation occur but rarely detected clinically because the affected person is a symptomatic heterozygote, unless this heterozygous marry other heterozygous &produce affected offspring.

N*N N*N

N*N*	N*N	N*N	NN
affected	carrier	carrier	normal
25%	25%	25%	25%

Examples of autosomal recessive disorders:

Sickle cell anemia

Thalassemias

Cystic fibrosis

Phenylketonuria

Wilson disease

Glycogen storage diseases

Albinism

Any questions?