



**ثانوية أجيال**  
AJYAL HIGH SCHOOL

**Subject: Biology**

**Grade: 12**

**Section: LS**

**Teacher: Abdallah Nassour**

**Unit: Reproduction and Genetics**

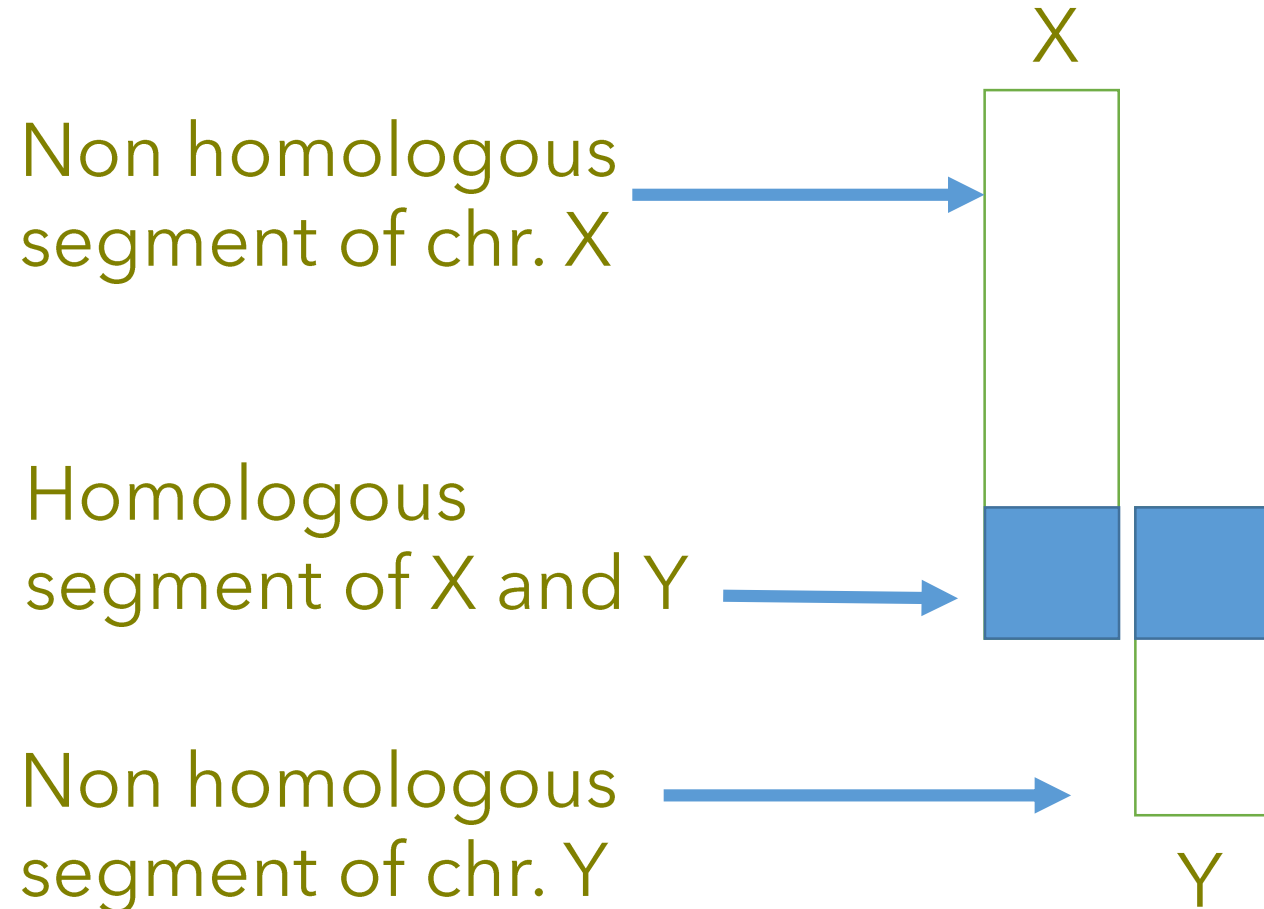
**Chapter 5: Human genetics**

**Document 3: Sex -linked diseases**

Some hereditary diseases are more frequent in males than in females.  
Where are the genes responsible for such diseases located? How are they inherited?

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# I- Sex chromosomes

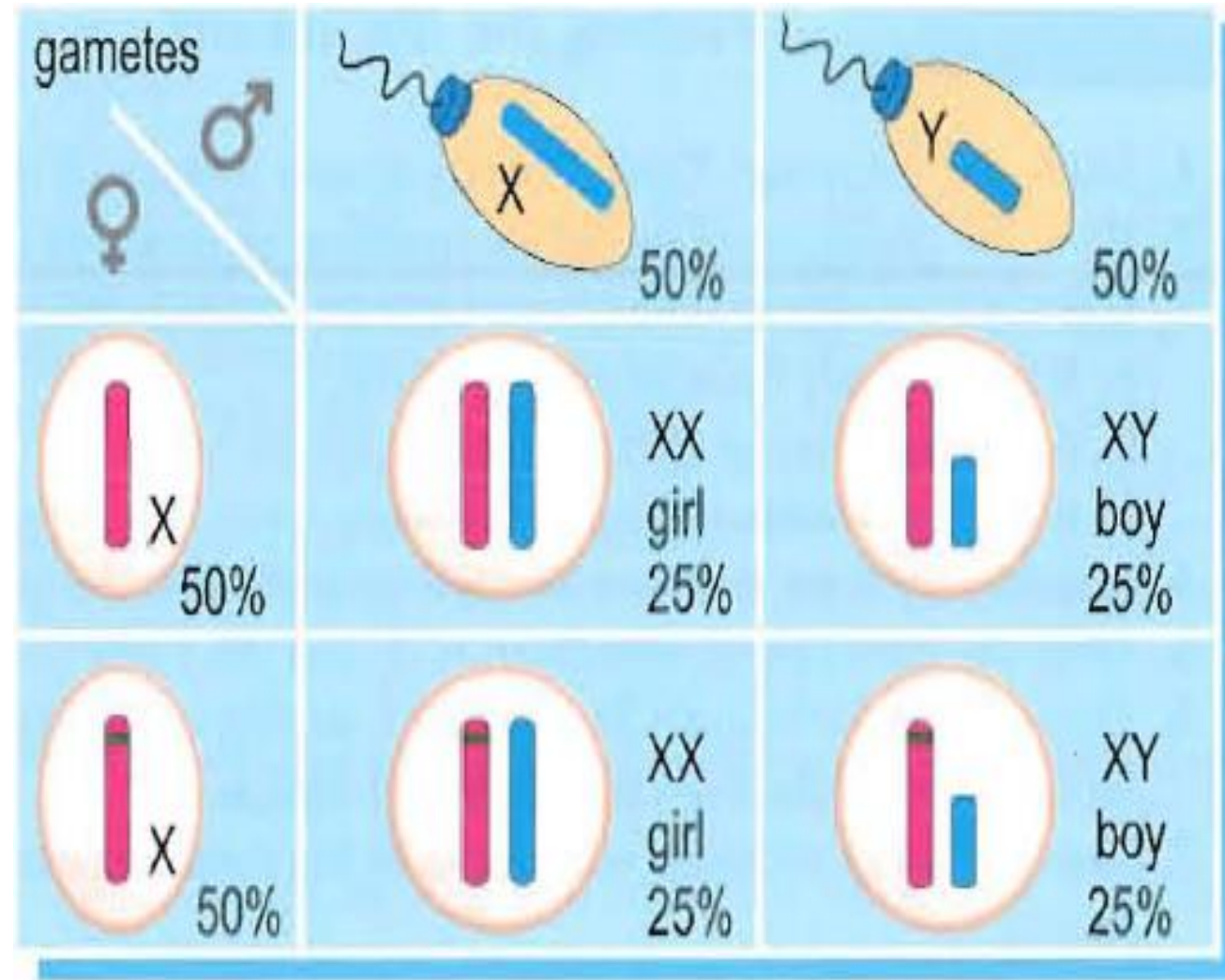


## 2 – The chromosomal basis for sex determination

During fertilization, it is the sperm cell that determines the sex of the future child

Since gametes meet randomly, a sperm cell carrying an X chromosome has the same probability as a sperm cell carrying a Y chromosome to fertilize an oocyte.

In fact, this equal (or about equal) probability of both sexes is observed in the population.



### 3 – Duchenne muscular dystrophy, a recessive disease

Duchenne muscular dystrophy (DMD) is characterized by the weakening of voluntary muscles. It appears at the age of two; the child waddles along and falls frequently. At the age of twelve, the child can no longer walk and needs a wheel chair. This disease occurs with a frequency of 1 in 5000

The protein whose deficiency causes the disease is called dystrophin. Studying the pedigree of a family in which some of its members are affected by this disease (myopathy) allows us to establish the pattern of inheritance of this disease (*Doc b*). Only affected girls are not viable

1- Specify if the allele responsible for the myopathy is recessive or dominant. Justify the answer

The boy II-3 has received at least one allele responsible for the disease from one parent, who therefore must be a carrier of this mutant allele although phenotypically normal. We can conclude that the allele responsible for the myopathy is recessive to the normal allele.

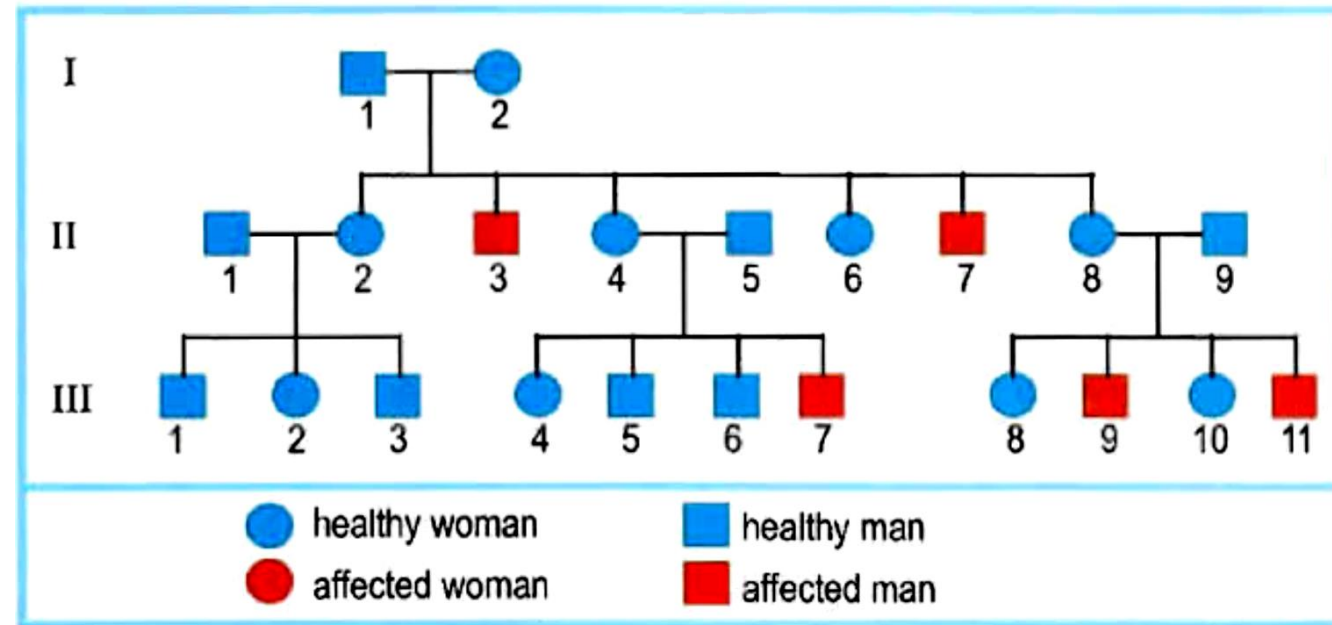
2- Determine the chromosomal location of the gene responsible for the DMD disease

The pedigree analysis shows that **only boys are affected**, which is consistent with a sex-linked type of inheritance. The myopathy gene can be carried by the non-homologous (proper part) of either chromosome Y or chromosome X. If it is carried by the proper part of the Y chromosome, the Y chromosome carrying the gene is surely transmitted from father to son, who will both be either affected or not affected; but this is not the case, because the normal father I-1 has two II-3 and II-7 affected children. The gene is thus carried by the non-homologous part of chromosome X

3- Give the probability for a couple (II-4 and II-5) to have an affected child

The probability between boys is: 50% affected with Duchenne's myopathy.

Or the probability between the children is: 25% affected with Duchenne's, all being males.



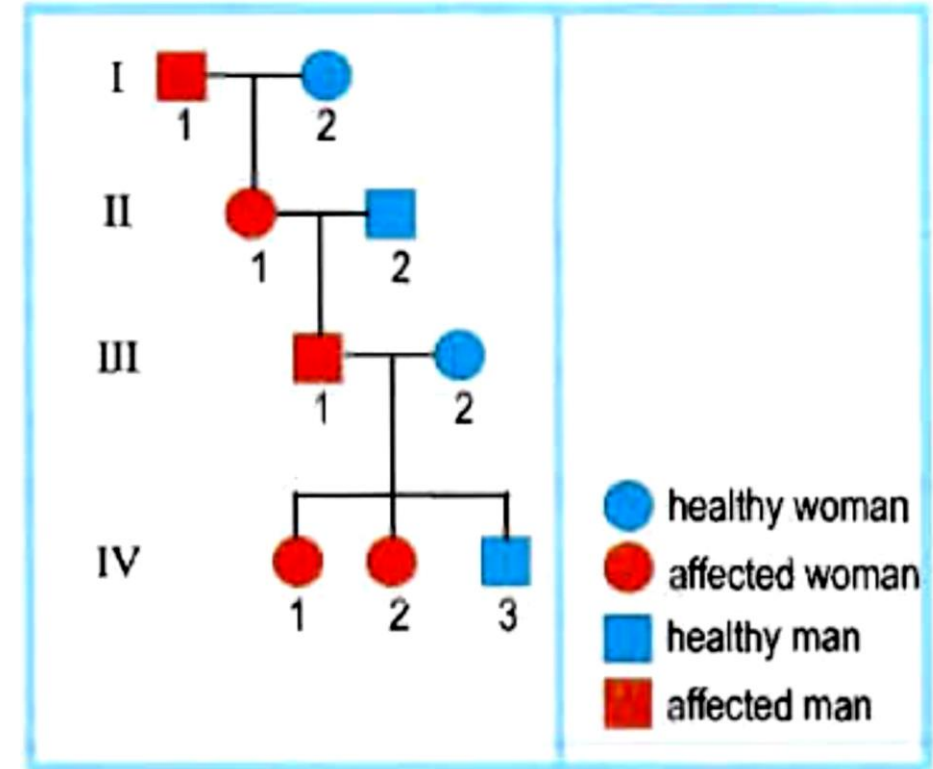


## 4 – Vitamin resistance Rickets, a dominant disease

Sex-linked dominant diseases are rare. Examples of these diseases are insipid diabetes and vitamin-resistant rickets. The latter is manifested by deformities in the skeleton following an insufficient calcification of bones. Unlike other types of rickets, the disease cannot respond to vitamin D treatment.

4- Explain why vitamin-resistant rickets is a dominant disease

The vitamin-resistant rickets disease is coded by a dominant allele because each affected member of the family has an affected parent. The number of affected persons in this family is very high, although it is a rare disease. .



*Doc.c* Pedigree of a family affected by vitamin-resistant rickets.