DECIPHERING THE TALK OF THE GENE

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What is a Pedigree?

A pedigree is a chart/diagram that shows how a particular trait or disorder is inherited across generations in a family.

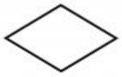
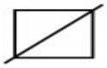
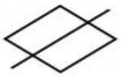
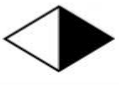
Useful in:

* Studying inheritance of genetic diseases

* Identifying carriers

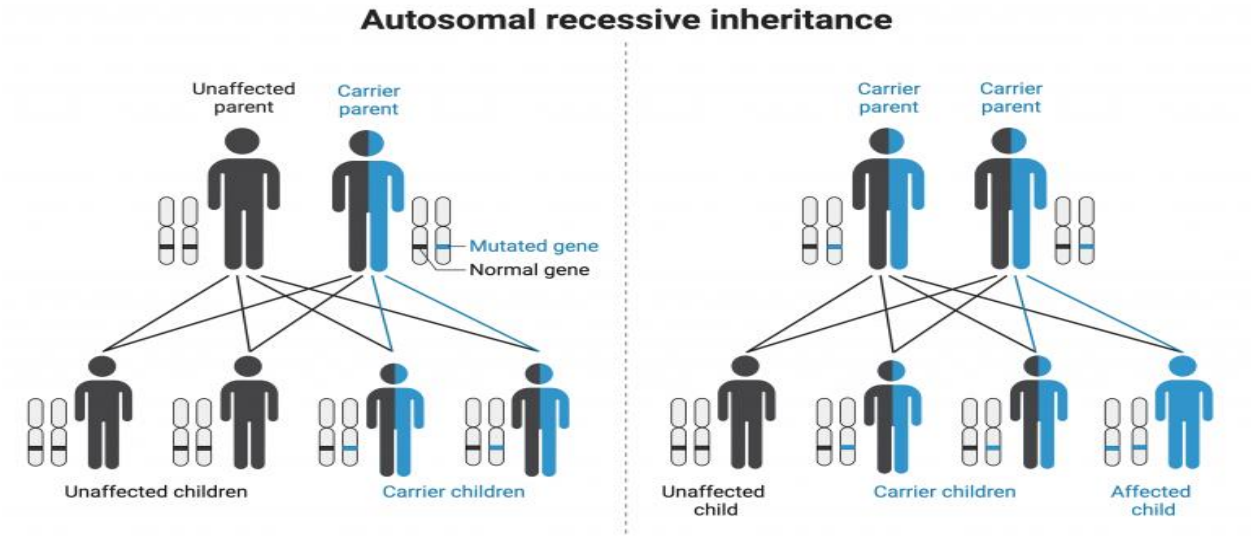
* Predicting risk in future generations

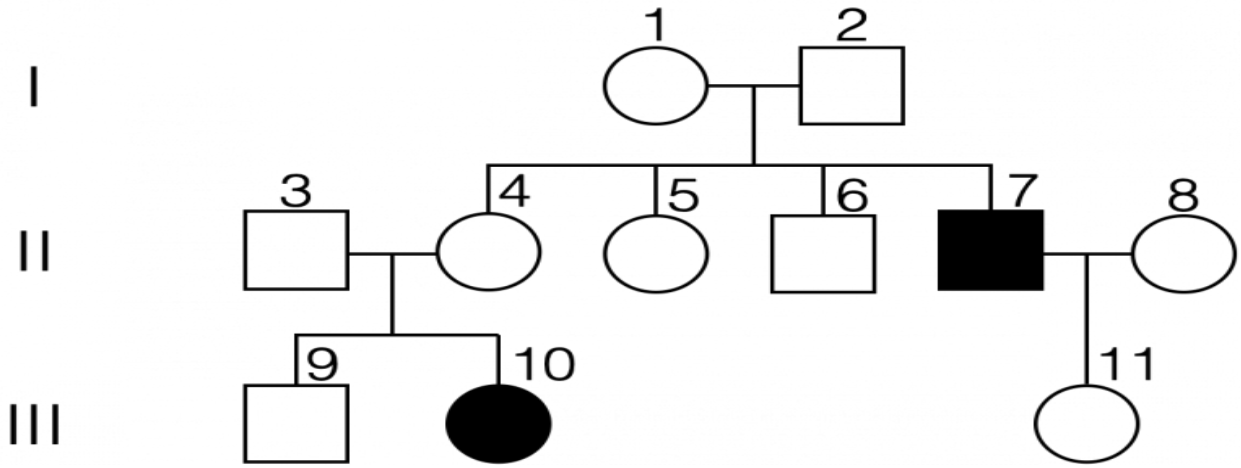
Pedigree Symbols (Universal)

	Female	Male	Not specified
Individual			
Affected Individual			
Deceased Individual			
Carrier (note that carriers are not always marked on a pedigree; often they are simply shown as unaffected)			

Autosomal Recessive Inheritance

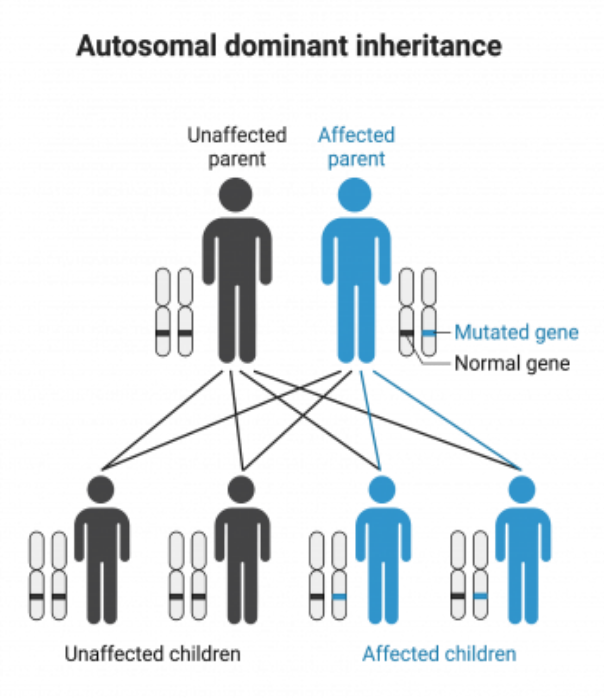
Some genetic conditions are autosomal recessive, meaning that the gene involved is found on an autosome, and affected individuals have two copies of the allele that causes the condition. If an affected individual in a pedigree has two unaffected parents, the condition is most likely recessive. Additionally, if daughters in the pedigree have two unaffected parents, the condition is most likely autosomal recessive (unlike X-linked recessive conditions, in which an affected daughter will have an affected father). With autosomal recessive inheritance, males and females are equally likely to be affected. Autosomal recessive conditions in humans include cystic fibrosis, sickle cell disease, Tay-Sachs disease, and phenylketonuria.

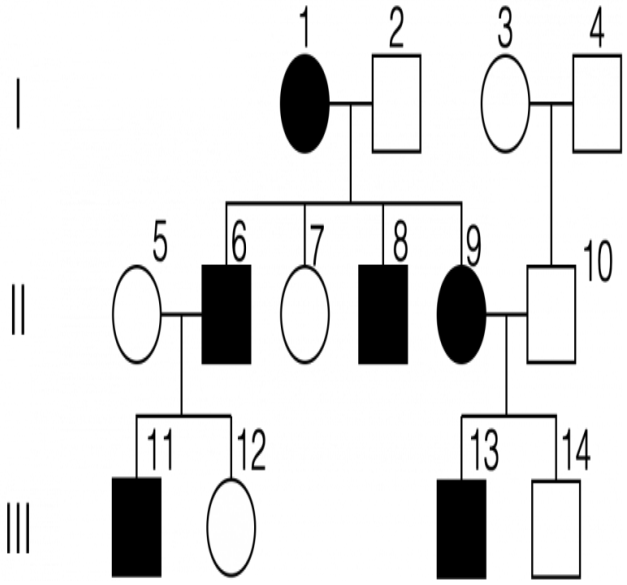




Autosomal Dominant Inheritance

Genetic conditions can display autosomal dominance. In this mode of transmission, a single mutant allele is sufficient to cause the condition because the mutant allele is dominant over the normal allele. Affected children generally have at least one affected parent (although not always, because some conditions display incomplete penetrance, meaning that not every individual with the allele will display the phenotype). Males and females are equally affected. Autosomal dominant conditions in humans include Marfan syndrome, Huntington's disease, and achondroplasia.

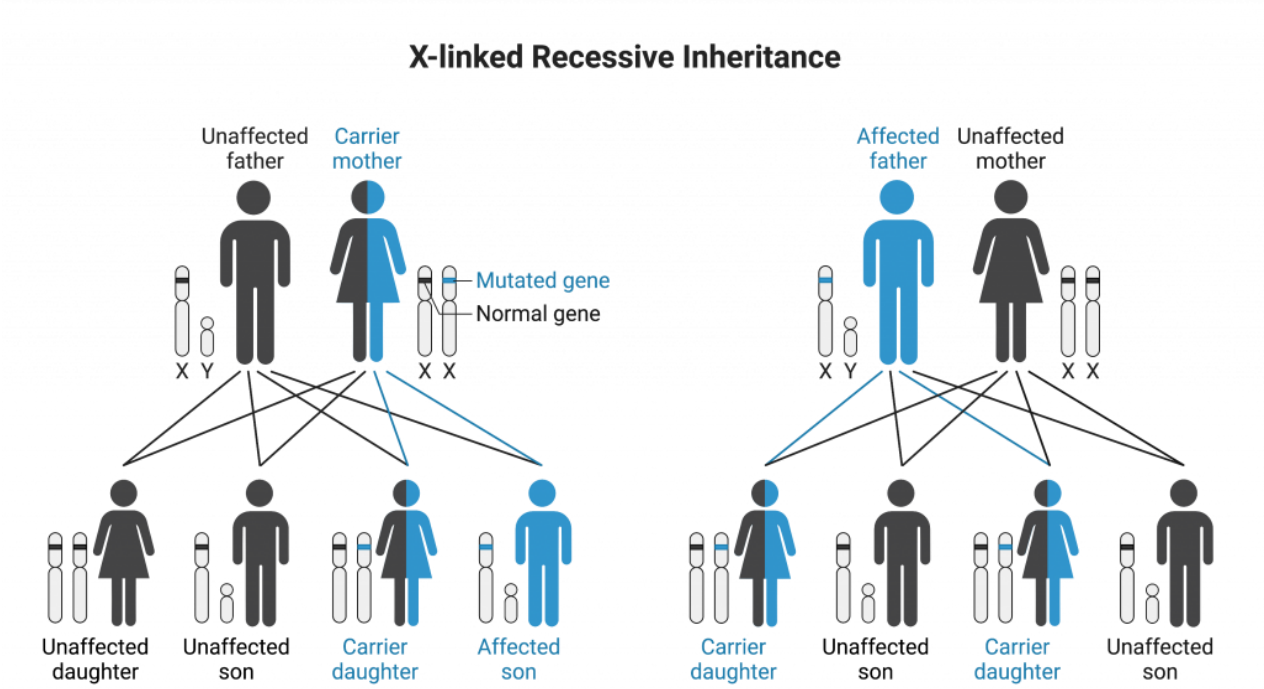


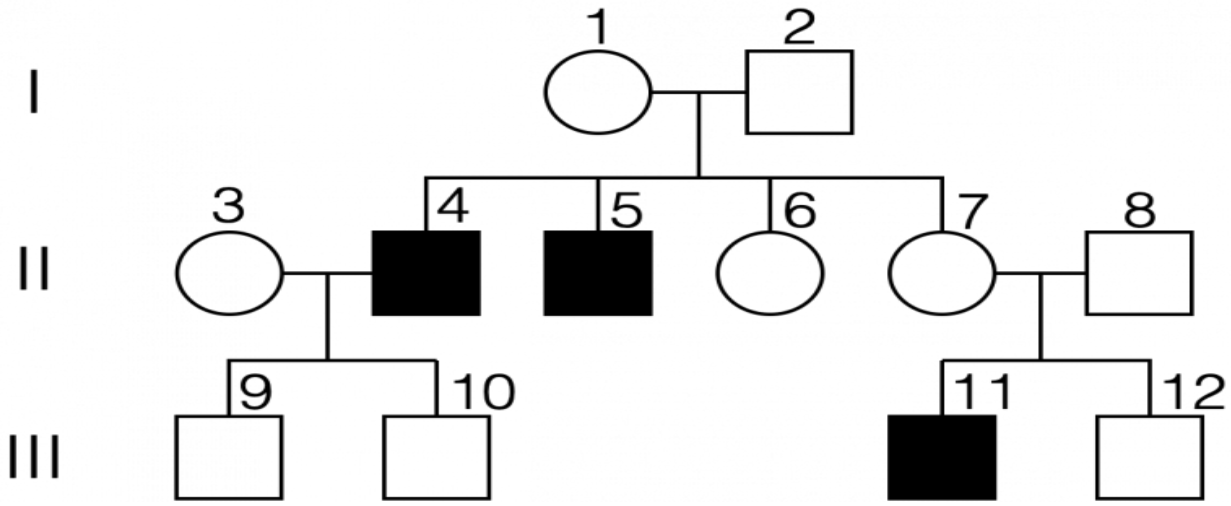


X-linked Recessive Inheritance

Some conditions are sex-linked, meaning the gene that causes the condition is on one of the sex chromosomes. In mammals, this is usually the X chromosome, because the X chromosome is much larger and has many more genes. X-linked recessive conditions are much more common in males, because they have only one X chromosome. Therefore, if they inherit an X chromosome with the mutated allele that causes the condition, they will display the condition. Females must inherit two copies to display an X-linked recessive condition. Males cannot be

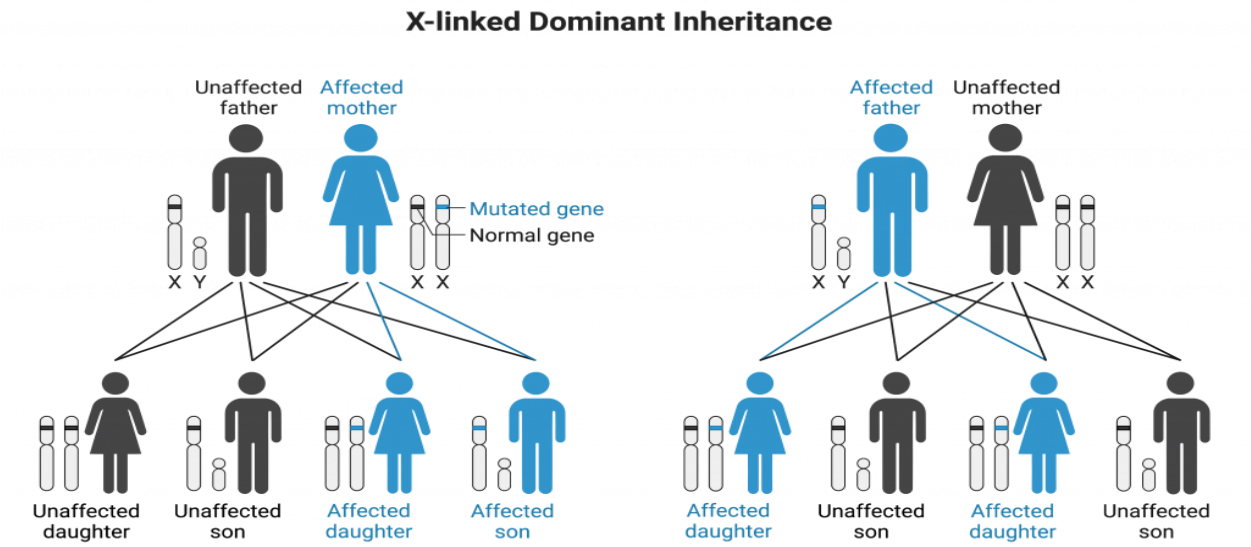
carriers; they will either be affected or unaffected, depending on which allele they inherit on the X chromosome. Examples of X-linked recessive conditions include Duchenne muscular dystrophy, the most common form of color blindness, and hemophilia A and B.

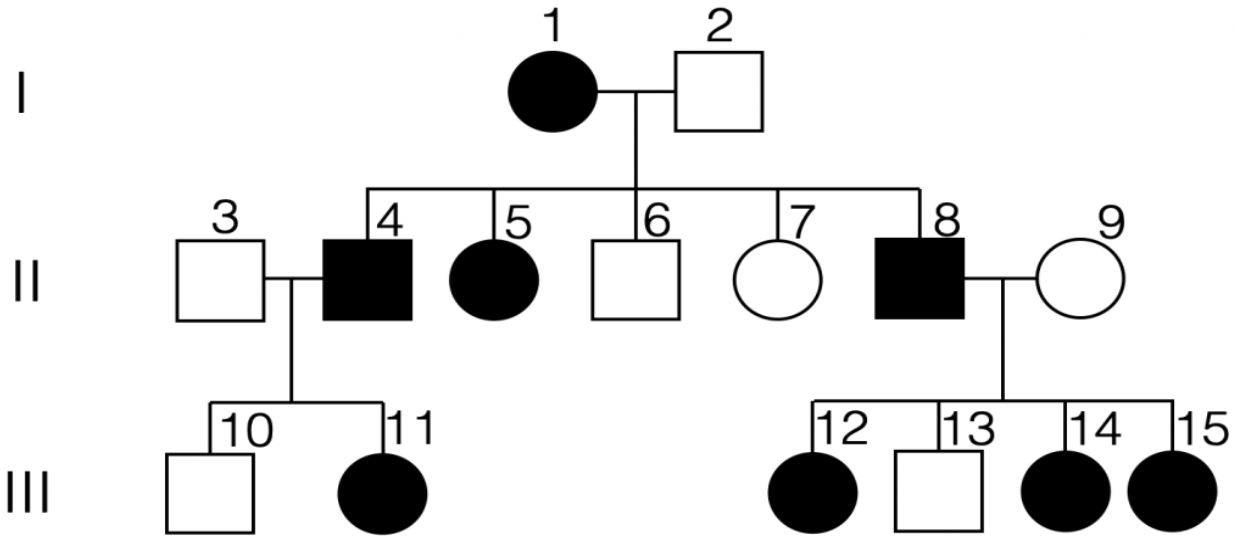




X-linked Dominant Inheritance

Conditions that display X-linked dominant inheritance are less common than X-linked recessive conditions. For this mode of inheritance, affected fathers will pass on the allele and condition to all of their daughters. Sons of an affected, heterozygous mother have a 50% chance of being affected. Examples of X-linked dominant inheritance include Rett syndrome and most cases of Alport syndrome.





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