

GENETIC INHERITANCE

Whereas other genetically transmitted diseases are the result of a single gene defect, RP results from a large and as yet unknown number of gene defects. So far around a hundred gene defects have been found to cause RP.

However, there are three main genetic pathways through which a child can come to be born with RP and the probability of this happening varies according to which of the genetic pathways applies.

Two of them are usually identifiable through a recurrent family history of RP but the third and by far the most prevalent one is not.

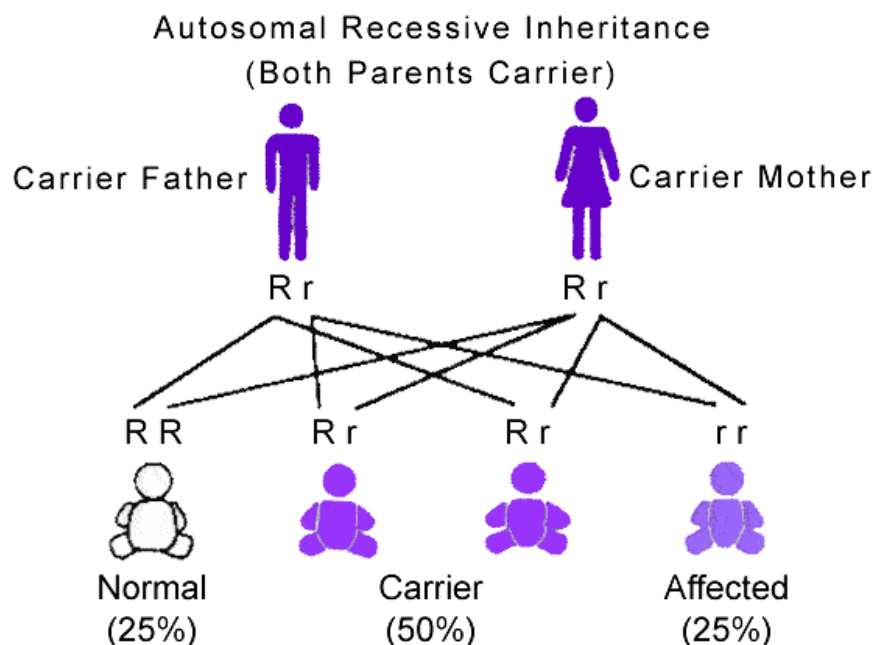
In simple terms RP is inherited in different ways: -

Autosomal Recessive Inheritance Pattern

This is the most common form of RP. This form means each parent is a carrier (have the gene present, but DNA testing is still unavailable) but is not affected by the disease themselves. Each of their children will have a 25% chance of being affected, with males and females having an equal chance of being affected.

The diagram indicates how the recessive genes are passed from the two carrier parents to their children.

The mutant RP gene is represented by "r" and the normal gene by "R".

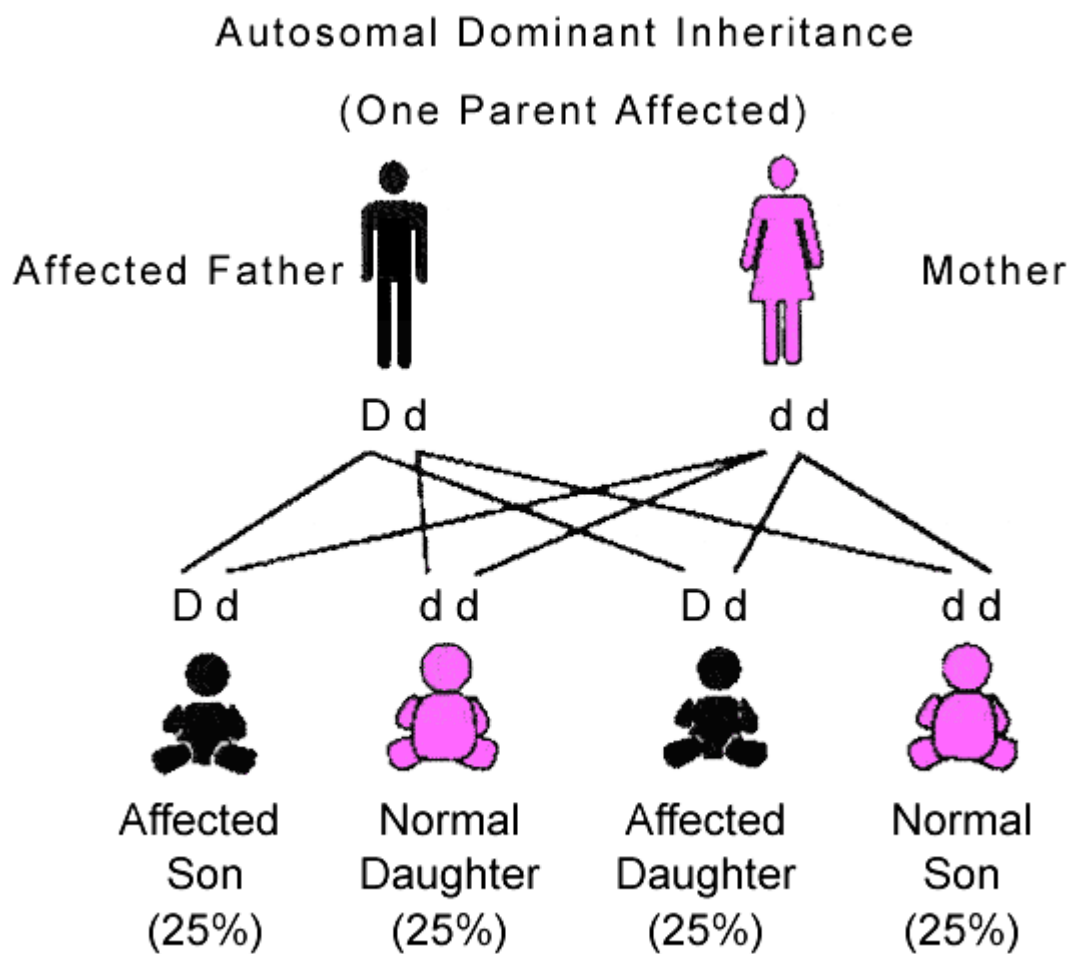


Autosomal Dominant Inheritance Pattern

In this type of RP, one parent is affected and each pregnancy has a 50% chance that the child will be affected. Males and females are equally affected.

The diagram indicates the four possible combinations of genetic information that may be passed on by the parents.

The mutant dominant gene is represented by "D" and the normal gene by "d".



X-linked Recessive Inheritance Pattern

This is the least common form of RP. This form is carried by the female and passed onto the male, there is a 50% chance that each son will have RP. There is also a 50% probability that each daughter will be a carrier. X-linked RP is the one form in which carriers can often be detected.

This diagram depicts the four possible combinations of genetic input from a couple, each contributing a copy of each of their genes. The woman is a 'carrier' of X-linked RP; the mutant gene on the X chromosome is represented can only convey normal genes.

