RETINA australia

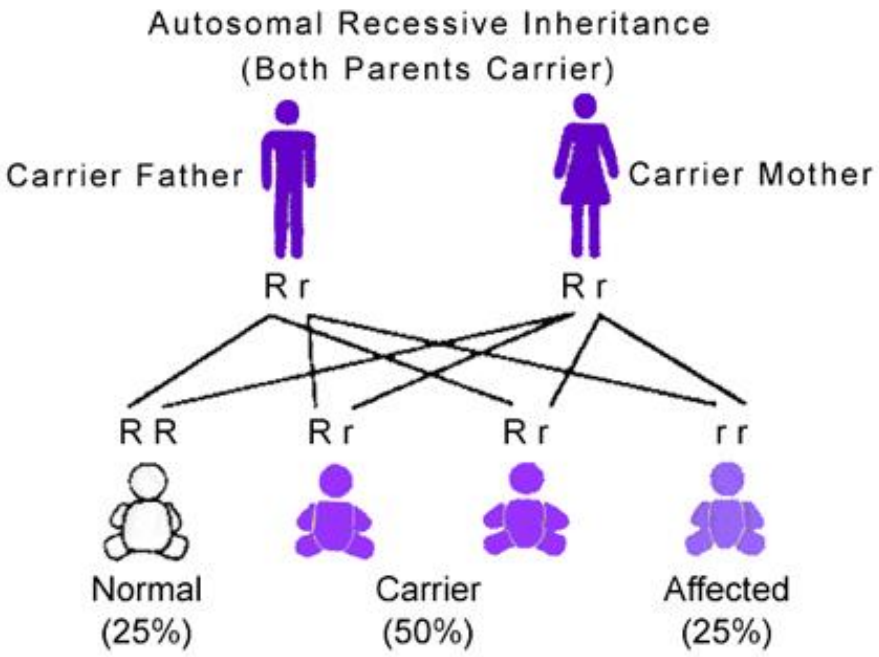
Inheritance Patterns

Some genetically transmitted diseases are the result of a single gene defect however inherited retinal diseases result from a large and as yet unknown number of gene defects.

In the case of retinitis pigmentosa, around a hundred gene defects have been found. However, there are three main genetic pathways through which a child can come to be born with an inherited retinal disease and the probability of this happening varies according to which of the genetic pathways applies.

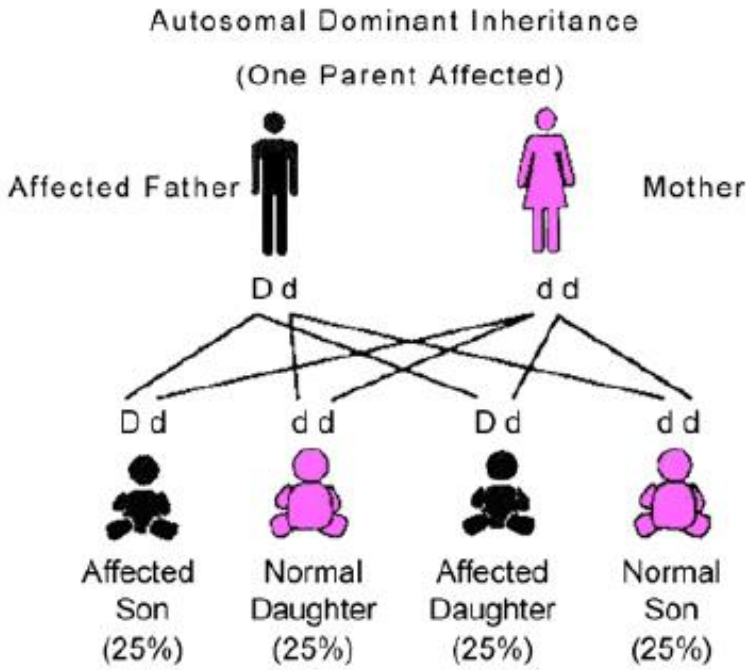
Sometimes the inheritance pattern of an inherited retinal disease can be identified through a recurrent family history of inherited retinal disease, but if the inheritance pattern is recessive, the inherited retinal disease can appear in families without any prior history.

In simple terms inherited retinal disease is inherited in three main ways:



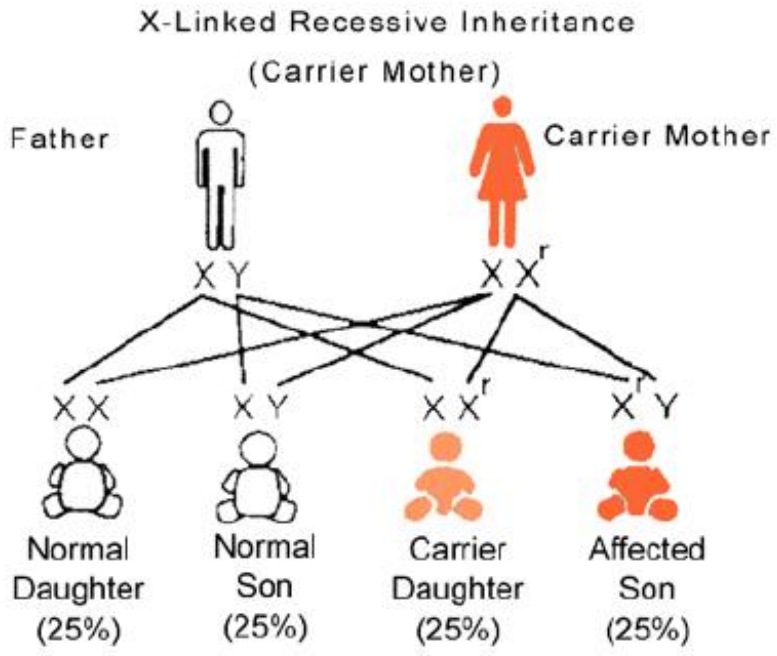
Autosomal Recessive Inheritance Pattern.

This is the most common way in which retinitis pigmentosa is inherited, and Leber congenital amaurosis is usually inherited in which way. This form means each parent is a carrier, but it 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 carried parents to their children. The mutant gene is represented by “r” and the normal gene by “R”.



Autosomal Dominant Inheritance Pattern.

In this type of inherited retinal disease 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 form is carried by the female and passed onto the male. There is a 50% chance that each son will have the inherited retinal disease. There is also a 50% probability that each daughter will be a carrier. Juvenile retinoschisis and choroideremia are inherited in this way, and retinitis pigmentosa is sometimes inherited in this way. The 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 an X-linked inherited retinal disease; the mutant gene on the X chromosome is represented by "r". the man can only convey normal genes.