

Autosomal Recessive Inheritance

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Each person has two copies of every autosomal gene. Both copies of the gene send a message to the cells to produce a particular product such as a protein. If a person has one faulty gene on one chromosome, and a correct copy of that gene on the other chromosome, they are said to be "carriers" of the faulty gene for a particular condition. They are therefore genetic carriers of the condition.

In **Autosomal Recessive** conditions it takes two "faulty" genes to cause the condition, with one faulty copy inherited from each parent (each parent being a carrier). Neither of the parents generally have the condition because they have one normal copy to compensate for the faulty copy. Autosomal Recessive disorders affect men and women equally and it can be passed on to a son or a daughter.

If both parents are carriers:

If both unaffected parents are carriers of one copy of the altered gene there is a **25% risk for each child** of inheriting an altered gene from both parents, therefore having two altered genes and being affected with an Autosomal Recessive disorder. There is a **75% chance of each child being unaffected**. If the child is unaffected, that child has a 66% risk of being a carrier. Figure 1 illustrates the inheritance pattern when both parents are carriers (thus not affected, but carrying one faulty gene) of Autosomal Recessive disorders. **These risks apply to every pregnancy.**

When parents are related (consanguineous marriage), there is an increased chance of both parents carrying the same mistakes in their genes (because they could have inherited these mistakes from the same ancestors) and this increases their risk of having children with autosomal recessive conditions.

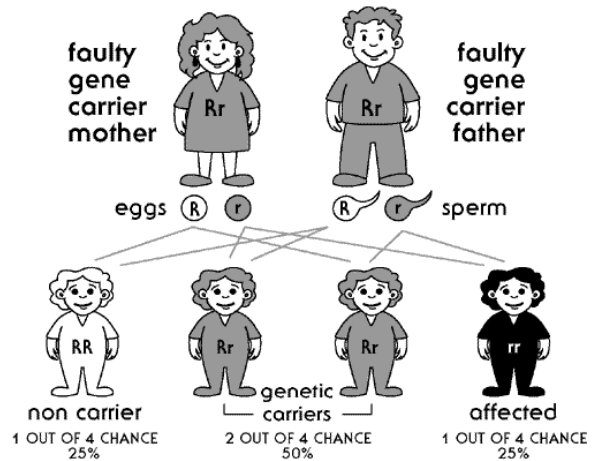


Figure 1: The faulty copy of the gene containing a recessive mutation is shown by "r". The correct copy of the gene is shown by "R". (Picture from the Centre for Genetics Education website: <http://genetics.com.au>)

If one parent is a carrier:

As shown in Figure 2, the couple will not have a baby affected with the particular condition. There is, however, a 50% chance that the baby will be a carrier of the faulty gene, just like his/her parent.

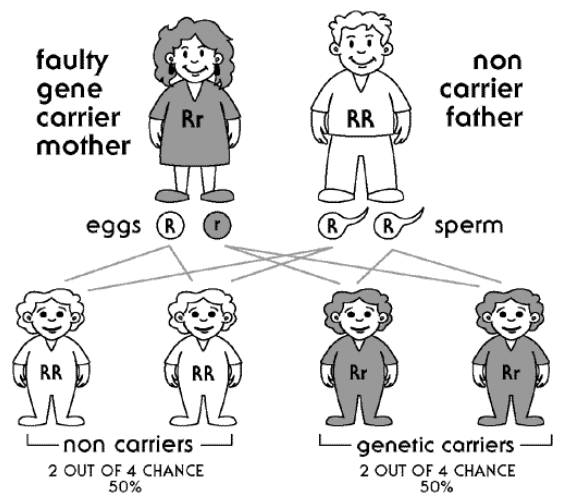


Figure 2: The faulty copy of the gene containing a recessive mutation is shown by "r". The correct copy of the gene is shown by "R". (Picture from the Centre for Genetics Education website: <http://genetics.com.au>)