Autosomal Recessive Inheritance

What are Genes?

Gene are the unique instructions that tell our bodies how to grow, develop and they make each of us an individual. There are thousands of genes in our body, each carrying its own specific instruction. Each gene contains a string of letters, arranged in a particular order, like the letters and words that make up a book.

If a spelling change happens in a gene, this can cause a genetic condition or disease. Sometimes doctors and scientists refer to the spelling changes in genes as a genetic mutation.

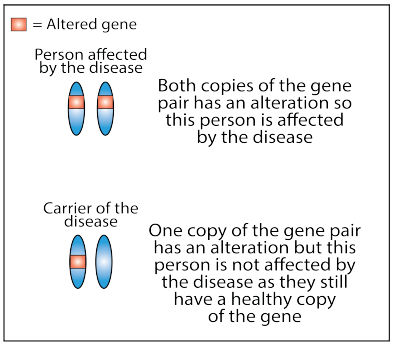
Each person has two copies of each gene. One copy is inherited from the mother and one copy is inherited from the father. When the couple has a child, each parent only passes on one copy of each gene pair. The child then has two copies of each gene (one copy inherited from each parent). This is how certain family traits (such as eye color) or genetic disorders can be passed through generations of a family.

What does Autosomal Inheritance Mean?

Genetic disorders that show autosomal recessive inheritance are caused by spelling changes in BOTH copies of a particular gene pair. In other words, there is a spelling change in the copy of the gene received from the child's mother and in the copy received from the child's father. Since there is no working copy of the gene the body is missing that instruction, and the person will be affected with the genetic disorder.

In autosomal recessive genetic disorders, individuals who have one normal copy of the gene and one copy of the gene that is altered by a spelling change, are healthy and not affected by the condition. This is because the person still has a working copy of the gene.

A person who carries one copy of a gene that is altered by a spelling change would be referred to as a carrier of the genetic disorder. Every person in the world is a carrier for different genetic disorders. It is not possible to tell if a person is a carrier for a particular disorder by looking at them.

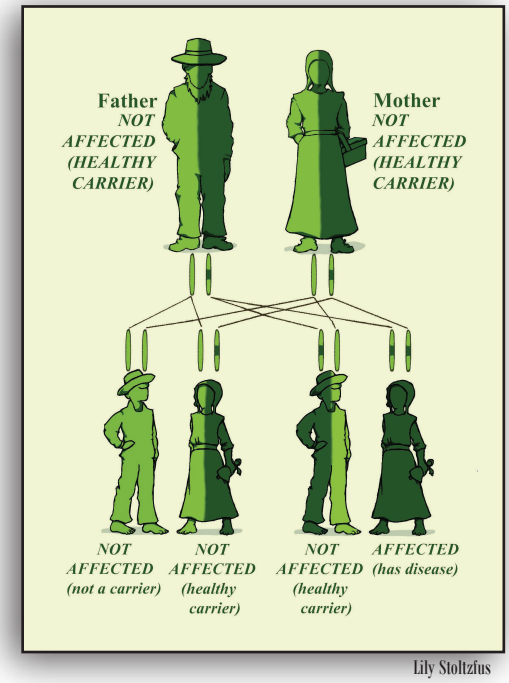


Having Children

If two healthy parents both carry a spelling change in one copy of the same gene, then each child they have has a:

- 1 in 4 chance of inheriting the altered gene from both parents and therefore being affected by the disorder
- 1 in 2 chance of inheriting the altered copy of gene from one parent and the normal copy from the other parent. The child would therefore be a healthy carrier (like their parents)
- 1 in 4 chance of inheriting the normal gene from both parents and being neither affected nor a carrier. When the child grows up, even if they were to have children with a carrier of the genetic disorder, none of their children would be affected by the condition

Overall, this means that for two parents that are carriers for the same genetic disorder, there is a 3 in 4 chance of having a healthy child and a 1 in 4 chance of having a child with the condition.



Lily Stoltzfus

If only one parent is a carrier of the altered gene, then each of their children has a 1 in 2 chance of being a healthy carrier, depending on which copy of the gene pair the carrier parent passes on.

These chances are the same for each pregnancy, and the same for boys and girls. There is no way to control which genes are passed on to a child, this occurs randomly for each baby.

There are no supplements or changes in our lifestyle which can influence or change this. Similarly, there are no medications currently available that can mend a gene if it is altered by a spelling change.

Genetic Testing

A gene test is required to make a diagnosis of an autosomal recessive condition. To arrange this, please see the contact information below.

This informational pamphlet is a project of New Leaf Center - a member of the Plain Community Health Consortium (PCHC)

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Midwest Genetics Network (Okemos, MI) **517-324-8300**
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WeCare Clinic - Medical Care for Special Needs (Pembroke, KY) **270-962-7383**