

Autosomal Dominant Inheritance

Dominant Inheritance

The following information will discuss what dominant inheritance means and how dominant conditions are inherited. However, it is first important to know about genes and chromosomes (see fact sheet ?).

Autosomal dominant (AD) inheritance refers to a condition that is passed on in a family in a dominant way due to a change (mutation) in a gene that is located on an autosome (one of the numbered chromosomes), which causes the gene to be faulty and not produce its protein product correctly. In AD conditions, a person inherits one normal copy, and one faulty copy of the gene. However the faulty copy is dominant (or overrides) the working copy and this causes an individual to be directly affected by or predisposed to a genetic condition.

Some AD conditions affect a person from the moment they are born, while others only affect that person during adulthood (e.g. Huntington's disease). Some gene mutations that are dominantly inherited will predispose people to develop a condition (e.g. Breast or ovarian cancer) but this also depends on environmental factors.

How are dominant conditions inherited? (See Figure 1.)

When a baby is conceived, each parent passes on one copy of each of their genes to the baby. If a parent has a faulty copy of a gene, that parent can either pass on their working copy, or the faulty copy of the gene to their child. This means that each child has a 50% (1 in 2) chance of inheriting the faulty gene and being affected by the condition. There is also a 50% (1 in 2) chance that the child will inherit two working copies of the gene and be unaffected and unable to pass the condition on to their children.

It is important to know that these possible outcomes are random and the chances remain the same for each pregnancy. AD conditions affect males and females equally.

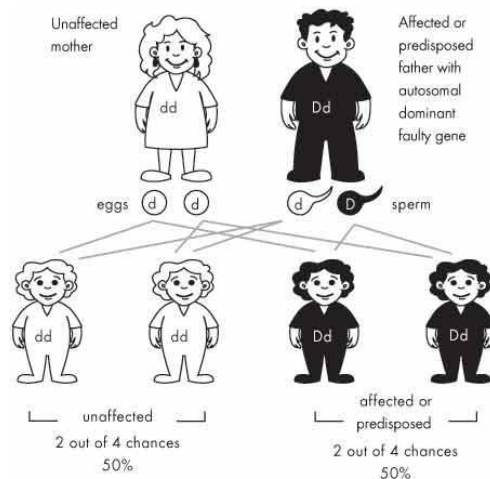


Figure 1. Autosomal dominant inheritance when one parent carries a copy of the faulty gene. The dominant faulty gene copy is represented by “D” and the normal copy by “d”. (Image from the Centre for Genetics Education website: <http://www.genetics.com.au/factsheet/fs9.asp>)

Why does the genetic condition sometimes appear to miss out a generation?

Some AD conditions can affect family members very differently. This is called variable expression. The condition doesn't actually miss out a generation but the symptoms may be so mild in an individual that they appear to be unaffected and may not even know themselves that they have the condition.

In condition that present later in life (adult onset conditions, e.g. AD Retinitis pigmentosa, inherited breast cancer and Huntington's disease), people may have died earlier of unrelated causes, leaving no time for the condition to appear, or may have been incorrectly diagnosed. However, the parents may have passed on the condition to their children.



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What if neither parent has the condition but they have an affected child?

Sometimes a child can be born with a AD genetic condition and be the first person affected in a family. This may happen because a change has occurred in a gene, for the first time (spontaneous mutation), in either the egg or sperm cell that went to make that child. When this happens, the parents of the child are not affected and the chances of having another child with the same condition are low, but this should be discussed with a genetic counsellor or doctor.

However, the child that is affected now has the faulty gene and can pass this on to their children.

Is it possible to test for a AD condition?

For some AD genetic condition it may be possible to test a pregnancy and see if the fetus is affected with the condition. Predictive genetic testing for some adult onset conditions is also available (testing an individual before any symptoms appear). However, in both these cases it is vital to first speak with a genetic counsellor or doctor to see if it is an appropriate and viable option.

For condition specific information please see other factsheets in this series or contact the Division of Human Genetics.

Sources of information:

The Centre for Genetics Education:

<http://www.genetics.com.au/factsheet/fs9.asp>

Eurogentest:

http://www.eurogentest.org/patient/leaflet/english/dominant_inheritance.xhtml