University Hospitals of Leicester
NHS Trust

Autosomal recessive inheritance of genes

Genetics Department

Information for Patients

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What are genes?

Genes are the unique set of instructions inside our bodies which make each of us an individual. Our genes are bundled into 23 pairs of chromosomes. 22 chromosome pairs are autosomal, meaning relating to the body, and 1 pair is our sex chromosomes, which define our sex. We all have more than 22,000 genes, and each carries a different instruction. These genetic instructions tell the body how to do a particular job, for example, make this person have blue eyes or make this person have curly hair.

If a gene is changed, it may cause a genetic condition or disease. Changes to a gene are sometimes known as a variant, a mutation or an alteration.

We have 2 copies of each gene. We get (inherit) 1 copy from each of our parents. When we have children, we pass on only 1 copy of each of our genes.

What does recessive inheritance mean?

Some conditions are inherited in an autosomal recessive way. In recessive inheritance a person must have 2 altered copies of a gene to have the disease or condition. People with 1 altered copy and 1 normal copy of a gene are known as **carriers**.

In recessive conditions, **carriers** are usually healthy and do not have the condition. Their normal copy of the gene usually keeps them healthy and stops the effects of the altered copy of a gene. In some rare conditions having 1 copy of an altered gene may affect the carrier.

Who is at risk for recessive conditions?

Everybody has some altered genes. Parents who are closely related to each other, such as first cousins, are more likely to have children with recessive conditions. This is because they are more likely to have inherited the same type of altered gene.

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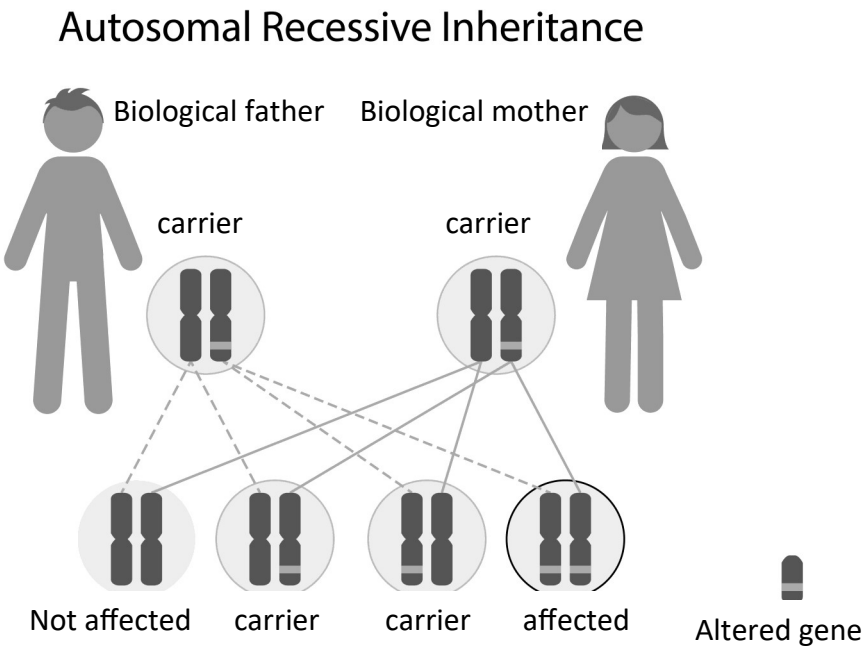
What happens when 2 carriers have children?

The recessive inheritance risk is the same for male and female children. If both healthy parents carry the same altered recessive gene, then each child they have has a:

- 25% (1 in 4) chance of having 2 normal genes
- 50% (1 in 2) chance of being a carrier of the altered gene, like their parents
- 25% (1 in 4) chance of inheriting the altered gene from both parents and having the condition



If only 1 parent is a carrier of the altered gene, each of their children has a 50% (1 in 2) chance of being a carrier, but will likely not be affected.

Where can I learn more?

A helpful charity that gives advice and support to families with disabled children is called "**Contact".** Please feel free to have a look at their website or call their helpline, listed below.

- www.contact.org.uk
- Free helpline 0808 808 3555

Contact details

Please discuss with your GP if you think you need an appointment with UHL Clinical Genetics. Please note that we only accept electronic referrals (ESR).

University Hospital of Leicester Clinical Genetics (Leicester Royal Infirmary, LE1 5WW)
Please call 0116 258 5736 with any questions.

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