The Leeds Teaching Hospitals NHS Trust

Leeds Clinical Genomics Service

Autosomal Recessive Inheritance

Information for patients

Leeds Clinical Genomics Service

What is a gene?

Our DNA is a long line of letters which is split into sections called genes. Genes are the unique instructions for how our bodies develop and function to make each of us an individual. There are thousands of different genes, each carrying a different instruction. We have two copies of most of our genes, because we get one copy of a gene pair from each parent.

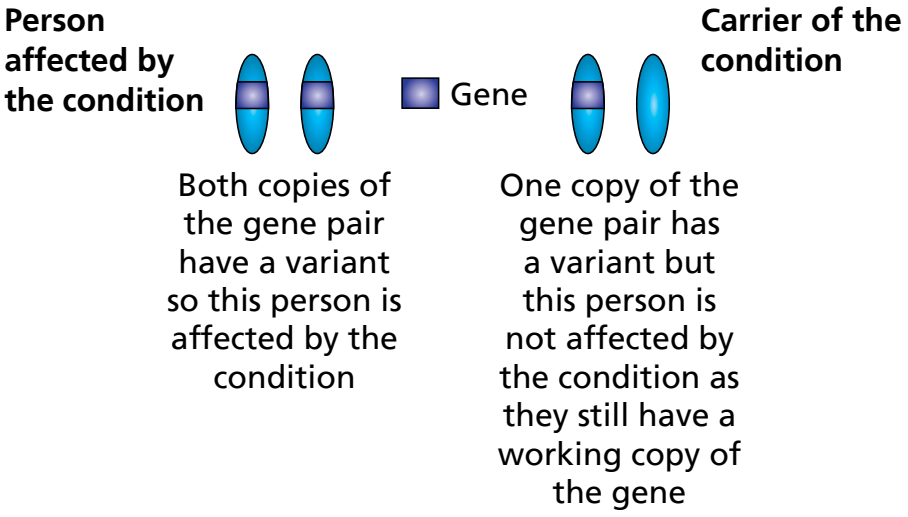
What is a gene variant?

We all have spelling changes (which are called variants) in our genes. Most variants do not affect our health, they simply make us different from each other. Some variants (called pathogenic or disease-causing) can stop a gene from working as it should which can result in a genetic condition or health problems.

What does autosomal recessive inheritance mean?

Genetic conditions that show autosomal recessive inheritance are caused by having a variant in **BOTH** copies of a particular gene pair. This means that a person will not have a working copy of the gene and will be at risk of developing the condition.

The diagram on the next page shows two gene pairs. The left pair shows both copies of the gene pair have a variant, and so the person will be affected by the condition. The gene pair on the right shows one non-working copy, and one working copy, and so the person will be a carrier of the condition. Usually, a carrier of an autosomal recessive condition will not be affected, as one working copy of the gene is enough to avoid health problems.

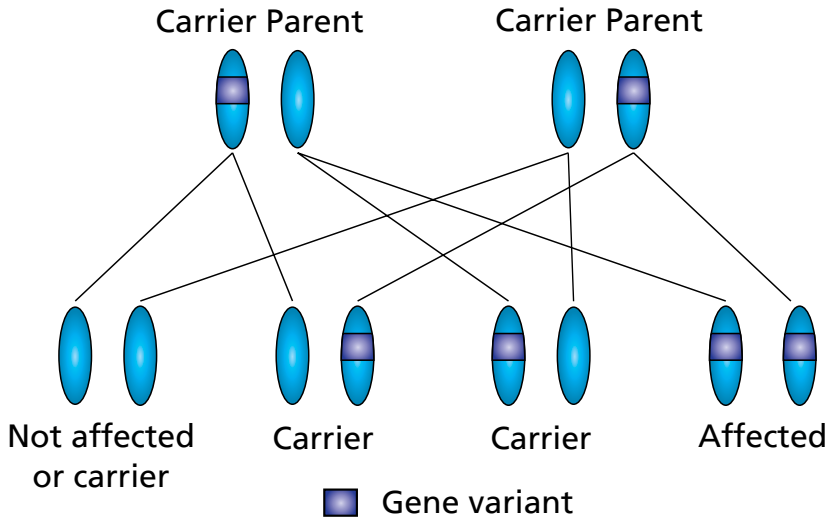


How are autosomal recessive conditions inherited?

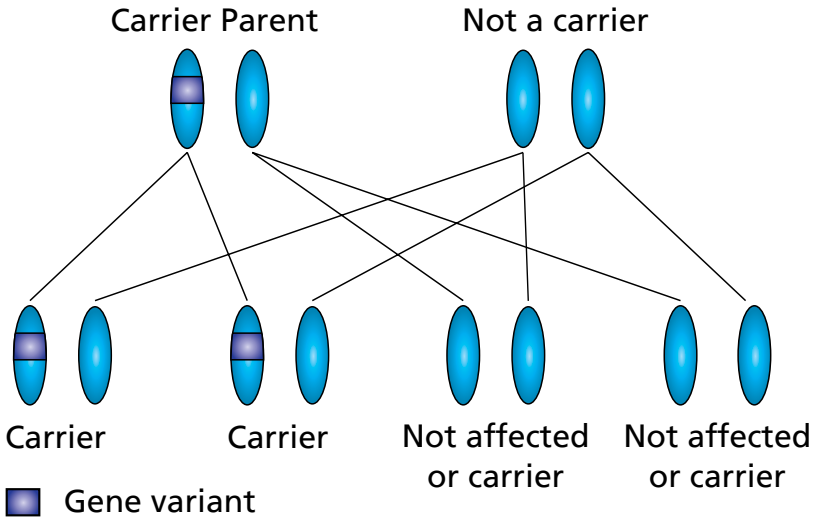
If two parents are both a carrier of the same genetic condition, there is a 50% (or 1 in 2) chance that they will each pass on their non-working copy of the gene. This chance is the same for each child, regardless of their sex.

The diagram below shows how for two carrier parents, then each child they have together has:

- a 25% (1 in 4) chance of inheriting the non-working gene from both parents and therefore, being affected with the condition.
- a 50% (1 in 2) chance of inheriting one copy of the non-working gene and one normal copy. If this happens, they are carriers of the condition.
- a 25% (1 in 4) chance of inheriting two normal copies of the gene and therefore having no problems relating to the specific condition.



If only one parent is a carrier of the condition, then each of their children has a 50% (or 1 in 2) chance of being a carrier. The diagram below shows how there is an equal chance of the carrier parent passing on their working and non-working copy of the gene.



Parents who are closely related to each other (for example cousins) are more likely to have children with an autosomal recessive condition. This is because they have a higher chance of both being carriers of the same genetic condition.

For more information:

If you need more information, please contact your local Genetics Department. If you live in the Yorkshire region, please contact:

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Notes

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With thanks to The Department of Clinical Genetics at Guy's & St Thomas' Hospital

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Produced by: Medical Illustration Services • MID code: 20250307_006RC

LN002379
Publication date 07/2025
Review date 07/2028