Notes

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This information guide is not a substitute for personal medical advice. Families should consult a medically qualified clinician in all matters relating to genetic diagnosis, management and health. Information on genetic changes is a very fast-moving field and while the information in this guide is believed to be the best available at the time of publication, some facts may later change. Unique does its best to keep abreast of changing information and to review its published guides as needed. This booklet was compiled by Unique (AP) and Dr Emma Baple, Consultant in Clinical Genetics and Makaela Jacobs-Pearson, Genetic Counsellor, Royal Devon and Exeter NHS foundation trust.

2022 Version 1 (AP)

Rare Chromosome Disorder Support Group
Registered in England and Wales

Charity Number 1110661
Company Number 5460413

Understanding Chromosome & Gene Disorders

Autosomal Recessive Inheritance

rarechromo.org

This guide aims to provide some introductory information about autosomal recessive (AR) genetic disorders associated with developmental delay and learning disability.

Unique also provides a more detailed guide to autosomal recessive disorders for those who wish to read a bit more in depth information.

What are genes?

Genes are the unique instructions in our DNA which make each of us an individual. Each gene contains a long string of letters arranged in a particular order, a bit like the letters of an alphabet that are arranged to form words. Genes tell our bodies how to grow, develop and function throughout life.

We have many thousands of different genes, each carries a different instruction.

We have two copies of most genes. One copy is inherited from each parent. When we have children, we pass on one copy of each gene.

What are genetic variants?

If a gene is altered, a bit like a spelling change in a word, doctors call these gene alterations genetic variants.

Most genetic variants do not change how the body reads the instruction and do not affect our health and development.

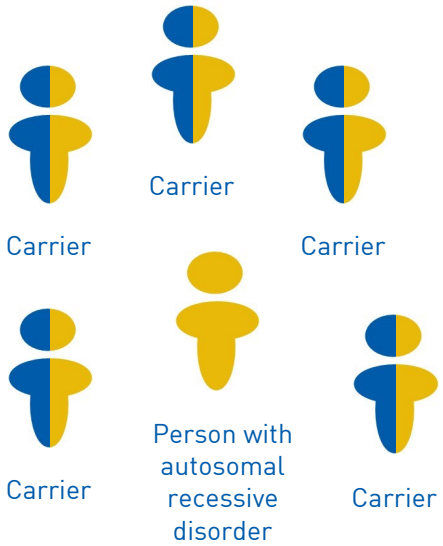
A genetic condition can occur when a genetic variant does alter the way the body reads the instruction. These are known as pathogenic (meaning disease causing) gene variants.

What does autosomal recessive inheritance mean?

An autosomal recessive condition occurs when there is a pathogenic gene variant in both copies of a gene. There is no unaltered copy of the gene to provide the expected instructions so the individual will be affected with a genetic condition.

In autosomal recessive genetic disorders, individuals who have one unaffected copy of the gene and one copy of the gene that is altered by a pathogenic variant, are healthy and not affected by the condition. This is because the person still has a copy of the gene that functions as expected. A person who carries one copy of a gene that is altered by a pathogenic variant is known as a carrier of the genetic disorder.

We are all carriers of many different genetic variants.

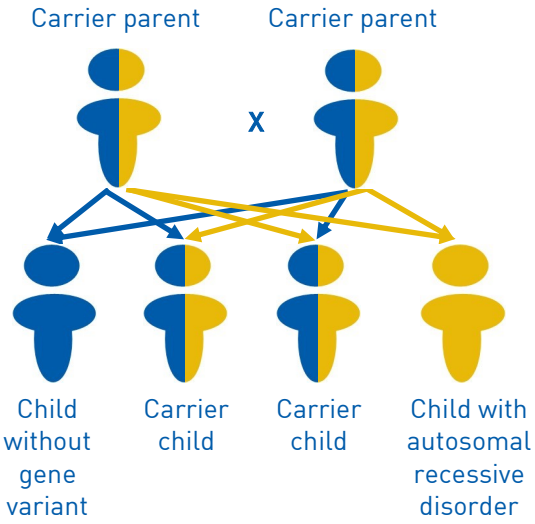


Will my child have a genetic condition if I am a carrier of a pathogenic gene variant?

The possibility of having a child affected by an autosomal recessive condition depends on whether both parents are carriers of the same gene altered by a pathogenic variant.

If both parents are unaffected carriers for the same genetic condition each child they have has:

- a 1 in 4 (25%) chance of inheriting an altered copy of the gene from both parents and being affected with the condition.
- a 1 in 2 (50%) chance of inheriting one altered copy and one working copy of the gene. If this happens, they will be an unaffected carrier.
- a 1 in 4 (25%) chance of inheriting two working copies of the gene and they will not be affected nor be a carrier of the condition.



A clinical geneticist or genetic counsellor can provide more specific advice for your family. It is important to remember that genetic variantshappen randomly. Nobody is at fault, they are not due to anything a parent did or did not do.