

# Autosomal Recessive Inheritance



**When your baby was about 48 hours old, a blood sample was collected from your baby's heel onto a blood spot (Guthrie) card. The blood was used to test for more than 20 rare metabolic disorders.**

Your child has been shown to have

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By now you should have discussed this condition with your paediatrician or the National Metabolic Service. You should know what the diagnosis is and how it affects your child. You should also know that it is a genetic disorder that has been inherited from both mum and dad in an autosomal recessive way.

This information sheet helps to explain this genetic inheritance pattern.

## What are genes?

Who we are, what we look like, and how we grow is determined by our DNA – the blueprint of life. The genetic material contained in our DNA is organised into thousands of individual units called genes, which are grouped together in units known as chromosomes that are found inside every cell in our bodies.

There are 46 different chromosomes (23 pairs); the sex chromosome (the pairing of X and X or X and Y) and 22 pairs of autosomes (any chromosome that isn't a sex chromosome). One member of each pair of chromosomes has come from each parent.

We each carry more than 20,000 genes in every cell in our body. Each gene produces a different protein and these proteins make



up how our cells and bodies function and they work together to maintain our health and wellbeing.

## How is a genetic disorder inherited?

We inherit our genes from our parents. Many variations in our genes are passed from generation to generation. A variation in the genetic code, called a 'mutation', can cause a gene to be faulty and not work properly, resulting in an adverse effect or disease.

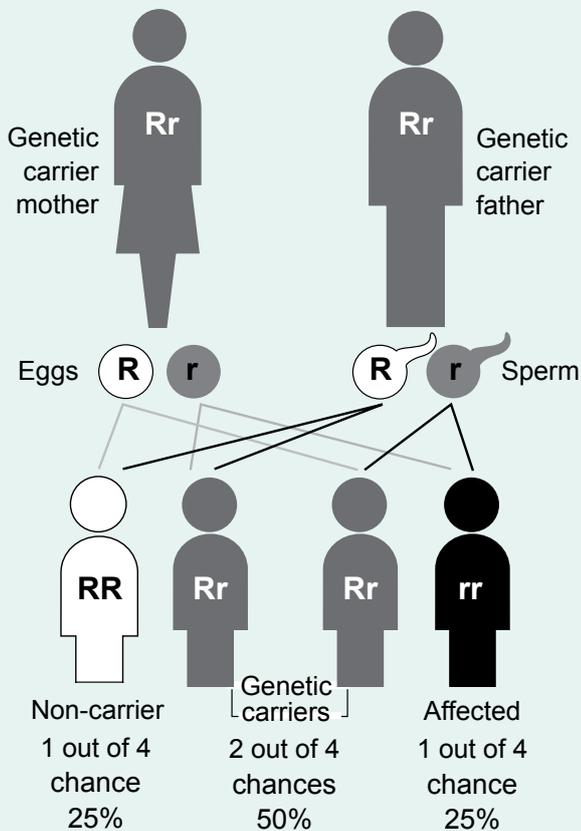
All of us are 'carriers' for mutations without knowing anything about them until our children, other family members or ourselves are affected by an inherited disorder.

Most genetic disorders diagnosed through newborn screening are inherited in an autosomal recessive way.

## How does autosomal recessive inheritance work?

When a baby is conceived, each parent passes on a copy of each of his or her genes to them. Therefore, the baby is a 'mixture' of their parent's genetic information.

There are four possible combinations of the genetic information that parents can pass on. You can see how that works in the diagram<sup>1</sup> below. In the diagram, the autosomal recessive 'faulty' gene copy is represented by the letter 'r', and the normal gene copy is represented by the letter 'R'.



R = normal gene r = faulty gene

As the diagram above shows, in every pregnancy, there is:

1. a 25% chance (1 out of 4 chance) that the baby will inherit a normal gene from both parents and will be unaffected by the metabolic disorder

2. a 50% chance (1 out of 2 chance) that the baby will inherit a faulty copy of a gene from one parent and a normal copy of the same gene from the other parent. The baby will be clinically 'normal' or 'unaffected' and will not experience a metabolic disorder. However, they will be a 'carrier' for the disorder (just like their parents)
3. a 25% chance (1 out of 4 chance) that the baby will inherit a faulty copy of a gene from both of their parents and will be 'affected' by or predisposed to develop a metabolic disorder, which interferes with chemical reactions that occur naturally in our bodies to help us live.

### Further information

Genetic Health Service New Zealand is New Zealand's provider of expert genetic diagnosis and advice. Clinics operate throughout New Zealand with a staff of clinical geneticists and genetic associates. Genetic Health Service New Zealand is part of New Zealand's public health system.

Genetic Health Service New Zealand is divided into three regional services. You can find your nearest region at:

[www.genetichalthservice.org.nz/about\\_us](http://www.genetichalthservice.org.nz/about_us)

You can find more information about metabolic disorders on the:

- National Screening Unit (NSU) website: [www.nsu.govt.nz](http://www.nsu.govt.nz)
- Screening Technology and Research in Genetics (STAR-G) website: [www.newbornscreening.info](http://www.newbornscreening.info)

<sup>1</sup> The information for this diagram was sourced from the Centre for Genetics Education, New South Wales Government, Australia.