

When parents share a common ancestor, it is termed a ‘consanguineous’ relationship and there is an increased chance that they will both carry the same faulty gene variation. If both parents have the same gene variation, there is an increased chance of having a child with a genetic condition.

**In summary**

- **We all carry certain genetic variations and faulty genes which do not usually impact our health**
- **Two unrelated people are less likely to carry the same faulty gene variation**
- **People who are blood relatives share a greater proportion of the same genes because they have a common ancestor**
- **When parents are blood relatives, children have a slightly higher chance of having a genetic condition.**

**WHAT IS CONSANGUINITY?**

The word **consanguinity** comes from two Latin words: **con** meaning shared and **sanguis** meaning blood. The most common form of consanguineous relationship is between first cousins and in some societies, can account for a large proportion of relationships.

The reasons for promoting consanguinity in certain communities are mainly social and include: strengthening the family ties; retention of property within a family and ease of finding a suitable marriage partner.

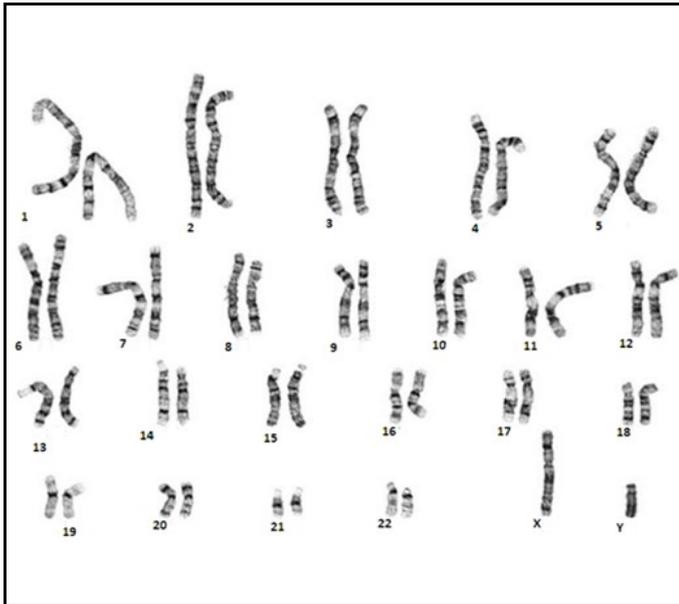
**RELATIONSHIPS BETWEEN BLOOD RELATIVES**

Since our genetic information is passed down to us from our parents and grandparents and so on, people who are blood relatives will share a greater proportion of the same genes than unrelated people.

Genetic specialists have classified how *close* relationships are considered, based on the proportion of their genes that they share (*Table 18.1*). The closer the biological relationship is between relatives, the more likely that they will have a gene variation in common.

Relationship to each other	Relationship type	Proportion of genes they have in common
Identical twins (monozygotic)		All (100%)
Brothers and sisters, non-identical (dizygotic) twins, parents and children	First degree relatives (1 <sup>0</sup> )	Half (1/2, 50%)
Uncles and aunts, nephews and nieces, grandparents and half-brothers and half-sisters	Second degree relatives (2 <sup>0</sup> )	Quarter (1/4, 25%)
First cousins, half-uncles and aunts and half-nephews and nieces	Third degree relatives (3 <sup>0</sup> )	Eighth (1/8, 12.5%)

**Table 18.1:** Proportion of genes shared between close blood relatives



**Figure 18.1:** Chromosome picture (karyotype) from a male 46,XY.

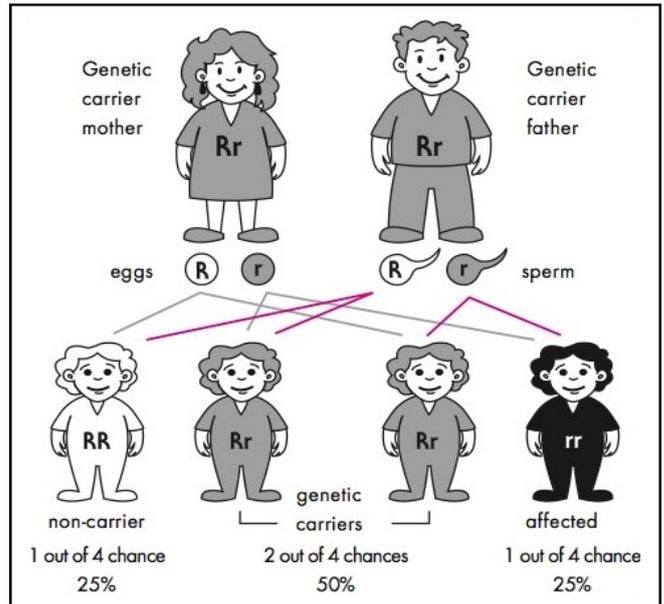
### GENES AND CHROMOSOMES

Our bodies are made up of millions of cells. Each cell contains a complete copy of a person's genetic instructions, or genes, packaged on chromosomes. Chromosomes come in pairs, giving two copies of each gene. The exception to this rule applies to the genes carried on the sex chromosomes called X and Y (*Figure 18.1*).

An **autosomal** gene is a gene located on a numbered chromosome and usually affects males and females in the same way.

An **X-linked** gene is located on the X or Y chromosome and affects males and female differently.

A variation that makes the gene faulty is called a **mutation** or a **pathogenic variant**. Genes code for the proteins our body needs to function. A mutation in a gene will affect the body differently depending on how much it changes the resulting protein, how critical that protein is to the body and how much of that protein is needed in the body.



**Figure 18.2:** Autosomal recessive inheritance when both parents are unaffected genetic carriers for the condition. The faulty copy of the gene containing a recessive mutation is represented by 'r'; the working copy of the gene by 'R'.

### WHAT IS THE CHANCE OF HAVING CHILDREN WITH A PROBLEM WHEN PARENTS ARE CLOSE BLOOD RELATIVES?

If parents are unrelated, their chance of having a child with a birth defect or disability is between **2% and 3%**.

If parents are first cousins, the chance is a little higher at **5% to 6%**. This is due to the increased chance that they will both carry the same **autosomal recessive** mutation, passed down through the family.

In general, when parents are consanguineous, they do not have an increased chance of having a child with genetic conditions that are due to X-linked or autosomal dominant gene mutations.

However, there will be some increased chance of a child inheriting conditions that are due to a number of different genes acting together (polygenic) or where there is an interaction between genes and the environment such as spina bifida and some forms of congenital heart disease.

## AUTOSOMAL RECESSIVE INHERITANCE

This type of inheritance refers to the inheritance of a **recessive gene mutation** on an autosome (one of the chromosomes numbered 1-22). See *Figure 18.2*.

There are two copies of every autosomal gene. Both copies of the gene send a message to the cells to produce a particular product such as a protein. Individuals who have a recessive mutation on one gene, and a working copy of that gene on the other partner chromosome, are said to be **genetic** carriers of the mutation for a particular condition.

Although only one of the gene copies is correctly sending the instructions to make the gene product, the cell can usually still work with this reduced amount.

Genetic carriers for the great majority of autosomal recessive genetic conditions do not show any symptoms of the condition and do not develop health problems due to being a genetic carrier. As mentioned above, we all have a number of recessive gene mutations which are hidden due to the fact that we have a backup working copy of the gene and therefore the correct gene product is made.

If a person has both of their genes for a genetic condition containing a recessive mutation, they do not have the ability to make the correct gene product and will have symptoms of the genetic condition.

Therefore, for autosomal recessive conditions, having one gene mutation does not usually cause a health problem. You will only get symptoms of the genetic condition if both of your genes have a mutation.

It is these recessive gene mutations which may be shared by consanguineous parents and can be passed on to cause a genetic condition in the children of some couples.

**In societies with a tradition of first cousin marriage, many couples are often more closely related than first cousins are and consequently their chance of having a child with an autosomal recessive genetic condition may be significantly higher.**

## GETTING CORRECT AND UP TO DATE INFORMATION

It is important for couples who are blood relatives and thinking about becoming parents, to seek genetic counselling.

Some relationships are complex, involving a number of generations where parents are closely related. The specialised knowledge that the genetic counselling team has is required to estimate any possible impact on children resulting from the couple's family relationships.

In addition, a few genetic conditions are more common in some population groups than in others. If the ancestry of the parents is clear, genetic testing may be possible to determine if they are carriers of more common recessive conditions.

For example, people where their background is from:

- Europe (including people from the United Kingdom), have a 1 in 25 chance of being a carrier of the faulty gene that causes cystic fibrosis, a condition that affects the respiratory and digestive system
- People from Southern Europe, the Indian sub-continent, the Middle East, Africa and Asian countries have a similar chance of being a carrier of the faulty gene that causes one of the forms of thalassaemia, a condition that affects the blood.

For information about where genetic counselling services are available in Australia, see [www.genetics.edu.au](http://www.genetics.edu.au)