Fact sheet **18**

WHEN PARENTS ARE RELATED – **CONSANGUINITY**

When parents share a common ancestor, this is called a 'consanguineous' relationship. There is a higher chance that they will both share DNA changes in common. Therefore they have a higher chance of having a child with a genetic condition.

IN SUMMARY

- We all carry certain DNA changes (variations) that do not usually impact our health
- If people are not related to each other, they are less likely to carry the same gene variation
- People who are blood relatives share more of the same genetic code because they have a common ancestor
- When parents are blood relatives, their children have a higher chance of having a genetic condition
- 'Consanguinity' describes a relationship between a male and female who are related by blood, and is a culturally acceptable and favourable practice in many societies.

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. In some societies, this may be more common.

The reasons for promoting consanguinity in certain communities are mainly social and include: strengthening family ties; keeping 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, people who are blood relatives will share more genetic code in common than unrelated people.

In *Table 18.1* the closeness of relationships are shown based on how much genetic code partners share. The closer the biological relationship is between relatives, the more likely that they will have a gene variation in common.

Table 18.1:
Proportion of genetic code shared between close blood relatives

Relationship to each other	Relationship type	Proportion of genetic code they have in common
Identical twins (monozygotic)		All (100%)
Brothers and sisters, non-identical (dizygotic) twins, parents and children	First degree relatives (1^o)	Half (½, 50%)
Uncles and aunts, nephews and nieces, grandparents and half-brothers and half-sisters	Second degree relatives (2^o)	Quarter (¼, 25%)
First cousins, half-uncles and aunts and half-nephews and nieces	Third degree relatives (3^o)	Eighth (⅛, 12.5%)

GENES AND CHROMOSOMES

Our bodies are made up of billions 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*).

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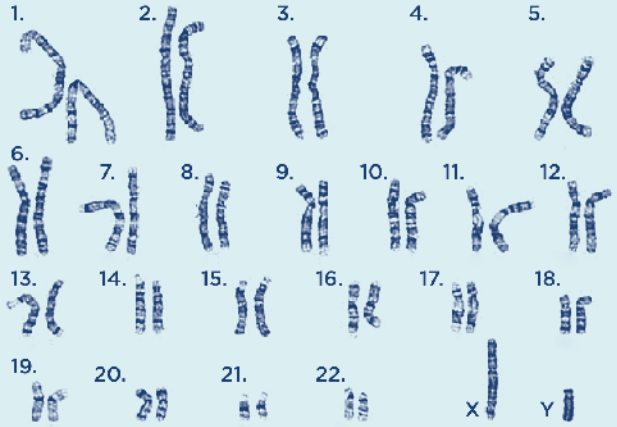
Figure 18.1:
Chromosome picture (karyotype) from a male 46,XY.



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 females differently.

The genes in our DNA provide the instructions for proteins, which are the building blocks of the cells that make up our body. Although we all have variation in our genes, sometimes this can affect how our bodies grow and develop. Generally, DNA variations that have no impact on our health are called **benign variants** or polymorphisms. These variants tend to be more common in people. Less commonly, variations can change the gene so that it sends a different message. These changes may mean that the gene does not work properly or works in a different way that is harmful. A variation in a gene that causes a health or developmental condition is called a **pathogenic variant** or **mutation**.

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 because of a higher chance that they will both carry the same **autosomal recessive** variant, passed down through the family.

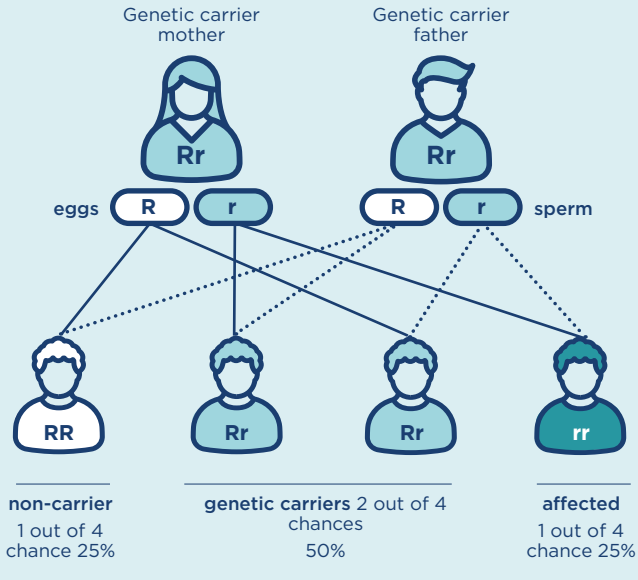
In general, when parents are consanguineous, they do not have an increased chance of having a child with genetic conditions that are due to X-linked or autosomal dominant gene variants.

There will, however, 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 variant** on an autosome (one of the chromosomes numbered 1-22). See *Figure 18.2*.

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



There are two copies of every autosomal gene. People, who have a recessive variant on one gene copy, and a working copy of that gene on the other partner chromosome, are **genetic** carriers for a particular condition.

Genetic carriers of autosomal recessive genetic conditions generally do not show any symptoms of the condition and do not develop health problems due to being a genetic carrier.

If both copies of a particular gene have a recessive variant, the gene product is not made in the right way and a person with this will have symptoms of the genetic condition.

It is these recessive gene variants which may be shared by consanguineous parents and can be passed on to cause a genetic condition in their children.

In societies with a tradition of first cousin marriage, couples may be more closely related than other couples who are first cousins. Their chance of having a child with an autosomal recessive genetic condition may be significantly higher.

GETTING 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. In addition, a genetic condition may be more common in some population groups than in others. If the ancestry of the parents is clear, genetic testing may be possible to work out if they are carriers of more common recessive conditions relevant to their particular background.

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 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 thalassaemia, a condition that affects the blood.

Reproductive genetic carrier screening is also available for couples who are planning pregnancy, or early in pregnancy.