

- Parents who are **first cousins once removed** or **2nd cousins** have an **additional risk** of about 1 in 100 (1%) and therefore a **total risk** of about 3 in 100 (3%).

This means that when there is no family history of a recessive disorder, most children of first cousins and more distant relatives will be healthy (95% for first cousins, and 97% for first cousins once removed and second cousins).

However, certain couples may be more closely related if there is a family tradition of cousin marriages going back generations. In this situation, the couple will have a higher risk of having a child with problems.

Can these abnormalities be detected in pregnancy?

About half or 50% of these severe abnormalities are thought to be detectable by specialized ultrasound scanning at around 18 weeks of pregnancy. These scans can be easily arranged by a midwife, genetics department or GP.

Which autosomal recessive disorders may be tested for?

If a couple has a known family history of an autosomal recessive condition, it is recommended that they contact their GP or midwife who can refer them to their local genetics centre where they can discuss their situation and the tests that may be available.

Certain autosomal recessive disorders are particularly common in some populations. Therefore the following screening is available for couples from the following ethnic backgrounds:

- Sickle cell screening for black couples
- Thalassaemia screening for Asian and Mediterranean couples
- Cystic fibrosis screening for white couples
- Tay Sachs disease for couples of Jewish origin

How will the testing be done?

For some couples at risk of passing on an autosomal recessive disorder, screening may be available. This screening involves:

- An initial blood test of both parents to determine whether or not they are carriers for a particular disorder.
- If the couple are **both** found to be carriers, a test **can be** offered in pregnancy to find out whether or not the child is affected by the disorder.
- If the child is found to be affected, the parents then can decide whether or not they wish to continue with the pregnancy.

For more information

[If you need more advice about any aspect of consanguinity, you are welcome to contact:](#)

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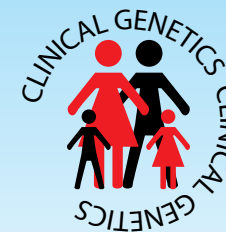
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Consanguinity



A leaflet for parents
who are related

What is Consanguinity?

Consanguinity comes from the Latin words: *con* meaning shared and *sanguis* meaning blood. Therefore consanguinity refers to a situation in which a couple are 'blood' relatives, i.e. they share a common ancestor. An example is a couple who are first cousins. Consanguinity is common in many cultures and can have a number of social benefits.

In genetic counselling, consanguinity is important because it increases the risk of certain genetic disorders called autosomal recessive disorders.

These are disorders which only occur if a child has a change (known as a mutation) in **both** copies of a particular gene.

What are genes and autosomal recessive disorders?

Our bodies are made up of millions of cells. Inside each cell is a set of 'instructions' telling our bodies how they should be made.

These 'instructions' are divided into about 30,000 pairs of genes.

Each gene:

- is a different instruction and determines a different characteristic e.g. eye colour.
- comes in a pair with one copy inherited from our mother and one from our father.

Gene Changes

Sometimes a change can occur in a gene which can affect the instruction produced by that gene. This could mean that a particular component required for the body to function normally is not made or is made incorrectly, causing health problems.

Healthy Carriers

Because our genes come in pairs it often doesn't matter if we have one changed copy because the other copy is normal and can compensate for the changed gene. A person with **one** changed copy is therefore called a '**healthy carrier**'.

Affected Individuals

For an individual to have an autosomal recessive disorder he/she must have **two changed copies** of a particular gene. For example, an individual with cystic fibrosis (a common autosomal recessive disorder in Europe) has two changed copies of the cystic fibrosis gene.

Parents of affected individuals

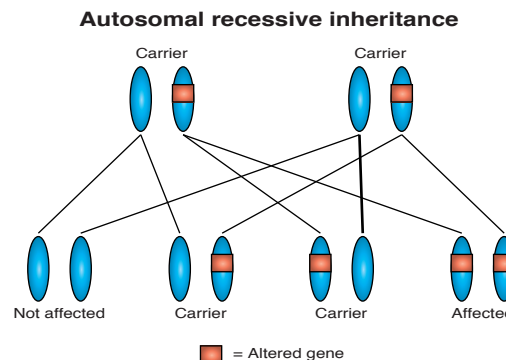
Because one copy of each gene comes from our mother and one from our father, both parents of an individual with an autosomal recessive condition must have at least one changed copy of the gene causing the disorder. Therefore if two carriers have a child together there is a risk that their child could be affected by that disorder.

What are the possibilities for children of parents who are both carriers?

As mentioned previously, each parent only passes on one copy of each of his or her genes. So for parents who are both healthy carriers of, for example cystic fibrosis, there are several possibilities for each of their children:

- A 1 in 4 (25%) chance that the child could be **affected** by cystic fibrosis.
- A 1 in 2 (50%) chance that the child could be a **healthy carrier**.
- A 1 in 4 (25%) chance that the child could have 2 normal copies of the CF gene and therefore would **not be a carrier or affected**.

With every pregnancy this chance stays the same, a bit like tossing a coin or throwing a dice.



Why are related couples more likely to have a child with an autosomal recessive disorder?

Everyone is thought to carry several changed gene copies, without affecting their health or development, because they have a normal gene copy to compensate.

We all carry thousands of possibly harmful changed genes. However, unrelated couples generally have alterations in **different** genes so their children usually only receive one changed copy of any particular gene. This means that an **unrelated couple** are at low risk of having a child with an autosomal recessive condition.

A **related couple** are more likely to have an alteration in the **same** gene because they have both inherited some of their genes from their shared relatives. In the case of first cousins, both of them could have inherited the same changed gene from one of the grandparents that they share.

So for first cousins, if one of the grandparents they share is a carrier of CF (i.e. has one changed copy of the CF gene), there is an increased risk that both cousins will also be carriers of CF.

The more distantly related a couple is, the fewer gene copies they will share and therefore the lower the risk that they will have a child with an autosomal recessive disorder.

If a couple are related, what are the risks of their child having severe abnormalities?

- To put things into context **unrelated parents** have a risk of about 2 in 100 (2%) of having a child with a severe/lethal abnormality.
- Parents who are **first cousins** have an **additional risk** of about 3 in 100 (3%), giving them a **total risk** of about 5 in 100 (5%).