



**Birmingham Women's
and Children's**
NHS Foundation Trust

Information leaflet for patients and families

Chromosome Inversions



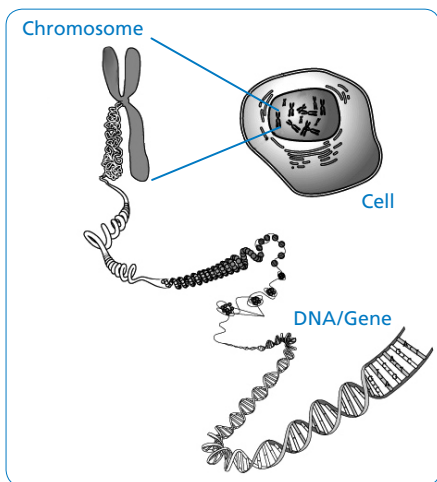
By your side

What is a chromosome inversion?

A chromosome inversion is a rearrangement of one of the chromosomes (see below) that are found in the cells of our bodies. Chromosome inversions rarely cause any health problems but they can have implications for pregnancy and fertility.

What are chromosomes?

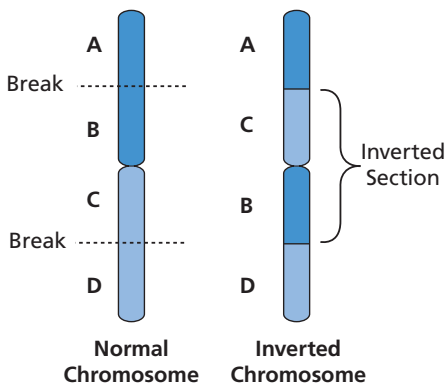
Chromosomes are microscopic packages of genetic information and are found in most cells in our bodies.



This information is organized along the chromosomes in the form of genes. Genes instruct our bodies how to grow, function and develop. There are thousands of genes on each chromosome. We normally have 46 chromosomes. These are arranged in 23 pairs: one of each pair is inherited from each parent.

What happens in a chromosome inversion?

Occasionally, chromosomes break and rearrange themselves. In an inversion, a chromosome breaks in two places (see diagram, below). It is repaired but with the broken fragment having been 'flipped round', or inverted.



Does
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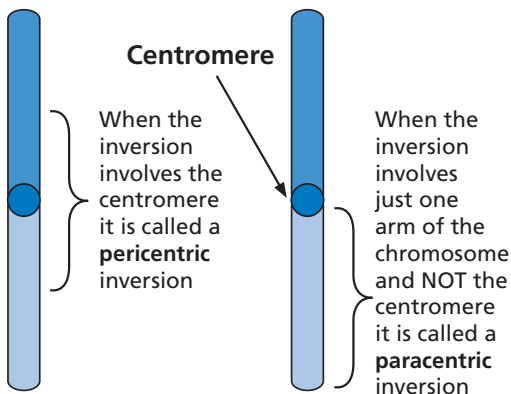
any health problems?

Not usually. Mostly, the body is still able to read the genes on the inverted piece of chromosome, even though it is 'backwards'. Occasionally the chromosome breaks in the middle of an important gene. This can cause different problems, depending on the particular gene(s) involved.

What are the effects on pregnancy and fertility?

This depends on the type of inversion involved. There are two types:

- Pericentric
- Paracentric



Pericentric inversions

This means that the 'middle' part of the chromosome called the centromere is involved. Pericentric inversions can lead to mistakes being made when the eggs or sperm are made. Some may get extra chromosome material and some may have missing chromosome material. This can lead to an increased chance of miscarriage (usually fairly early in pregnancy) or, less commonly, babies being born with a variety of problems, such as physical and/or learning disabilities. It is difficult to predict these as each inversion tends to be unique.

Paracentric inversions

This means that the inverted fragment is on one side of the centromere. The chance of having a baby born with problems (physical and/or learning disabilities) is very low compared to pericentric inversions. The chance of miscarriage is similarly increased due to missing or extra chromosome material in the sperm or eggs.

Do inversions cause fertility problems?

Not necessarily. Some studies have found that a small number of individuals with fertility problems have either peri or paracentric inversions. However, the chance of an inversion causing fertility problems is likely to be small.

What tests are available?

- Blood test. Inversions can be detected with a simple blood test and the results take up to 6 weeks.
- During Pregnancy. If you are a 'carrier' of an inversion you can have a test during pregnancy to detect if the baby has any extra or missing chromosome material. (See leaflet 'Tests for you and your baby during pregnancy.') This is a complicated subject and your genetic counsellor or doctor will be happy to discuss any questions you may have about chromosome inversions.

For more information Clinical Genetics Unit

www.bwhct.nhs.uk/genetics-wmrcgshome.htm

If you need more advice about any aspect of

Chromosome Inversions please contact:

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