

chronic villus sampling (CVS). Separate leaflets are available which explain these two tests in more detail.

For more information

If you or any other member of your family want to talk more about the chromosome inversion or any of the points raised in this leaflet, then please do not hesitate to contact:

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Seen in clinic by

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Chromosome Inversions



Information Leaflet

Introduction

This leaflet is written for individuals who have a chromosome inversion and for members of their family. It is intended to explain what a chromosome inversion is and what this means for individuals and members of their family.

What are genes and chromosomes?

Genes are tiny 'packages' of vital information which influence our growth and development. Each person probably has about 30,000 genes, which all do something different. They work rather like special computer programmes, and determine such things as the colour of our eyes, how many fingers and toes we have, how tall and short we are, and so on. Genes are so small that they cannot be seen, even under a powerful microscope. They are arranged along little threadlike strands, rather like the way that beads are arranged along a string. These strands are called **chromosomes**, and are inside most of the cells in our body. Unlike genes, chromosomes can be seen under a microscope.

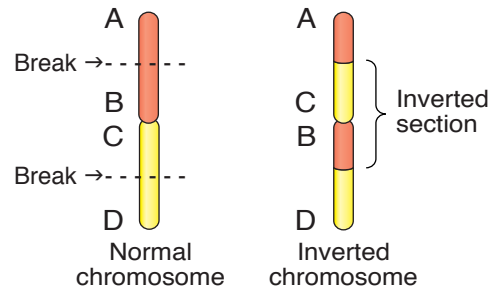
Every cell in a person's body contains 23 pairs of chromosomes (46 in total), which can be seen down the microscope.

To help differentiate between chromosomes, each pair has been given a number. The number 1 chromosomes are the largest, the number two are the next largest and so on right down to the number 22 which are the smallest. The remaining pair (X and Y) are called the sex chromosomes and determine what sex a person is.

What are chromosome inversions?

Sometimes several members of a family will have a chromosome which looks a little different to normal down the microscope. This is because occasionally a chromosome will break in two places and the piece which is broken rejoins UP SIDE DOWN.

This is called a CHROMOSOME INVERSION and is shown in the diagram below.



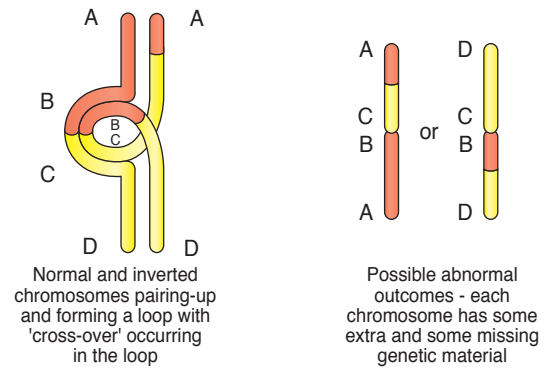
A person with a chromosome inversion will have one normal chromosome and a chromosome with an inversion.

People who have a chromosome inversion are completely normal individuals because they have no extra or missing genes. It does not really matter which order the genes are in as long as they are all present. If a person has a chromosome inversion it can, however, cause problems when the individual comes to have children. Each person can only pass on half of their chromosomes to their children. This means that an egg or sperm will only contain one of each of the pairs of chromosomes.

Normally before each chromosome pair splits up they line up closely together and sometimes exchange genes in a process called a crossover.

When there is an inversion in a chromosome the genes are in a different order compared to the other chromosome. In order for each of the genes to pair up with its partner on the opposite chromosome, one chromosome needs to form a loop.

If there is a crossover between the chromosomes within the loop, an imbalance in the number of genes will occur. When they separate, both chromosomes will carry extra copies of some genes while other genes will be missing. This is illustrated in the diagram opposite.



What happens when a person with a chromosome inversion has children?

- One of three things might happen:

1 - The parent with an inversion passes their normal chromosome on in the egg or sperm. Thus the baby will have a completely normal chromosome pattern with the correct number of genes in the correct order.

2 - The parent with the inversion passes their chromosome with the piece which is inverted on in the egg or sperm. The baby will inherit the inverted chromosome. However, because the correct amount of genetic material will be present the baby will be perfectly normal and healthy.

3 - If a loop and crossover have occurred whilst the egg or sperm were being formed, a baby could inherit a chromosome which is carrying an incorrect number of genes. This may result in an early spontaneous miscarriage or problems with infertility. A child who has inherited either too much or too little genetic material will usually have physical disabilities and learning difficulties.

Unfortunately, we are unable to predict which of the three events is likely to have occurred in any particular pregnancy. However, tests are available in pregnancy that enable us to see if the baby has inherited the correct amount of chromosome material or not. The two tests are called amniocentesis and