

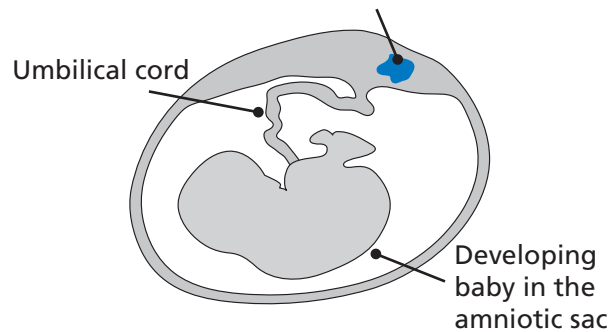
## Mosaicism and prenatal testing

Sometimes it is possible to offer tests for genetic or chromosomal alterations in a developing baby during a pregnancy. One way of doing this is to look at a small sample of the placenta in a Chorionic Villus Sampling (CVS) test (see separate CVS leaflet for more details about how this test is carried out).

Cells from the placenta can sometimes show mosaicism. If mosaicism is found on a prenatal test it can mean:

- 1) the mosaicism may be confined to a small area of the placenta with the rest of the baby's cells being normal. This is called confined placental mosaicism (or CPM) and is found in about 1% of chorionic villus sampling tests, see diagram below.

An area of confined placental mosaicism - some cells have altered genes or chromosomes and some are normal. If the CVS sample is taken from here it will show a mosaic result.



- 2) the baby may actually have a mosaic result having combination of normal cells and cells with altered genes/ chromosomes.

## If a test shows a mosaic result what happens?

If a baby is mosaic for a particular gene or chromosome problem then they may have some medical problems as a result. Alternatively, they may be completely healthy. The extent of any problems will usually depend on the proportion of normal cells and which genes or chromosomes are altered in the other cells. However there is no direct correlation between the level of mosaicism detected at CVS and any problems that may result. The doctor or genetic counsellor you see will be able to discuss this with you in more detail.

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**Birmingham Women's NHS Foundation Trust**  
Edgbaston, Birmingham, B15 2TG  
Telephone: 0121 472 1377  
Fax: 0121 627 2602

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Author: Clinical Genetic Unit (Group 10)  
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Birmingham Women's   
NHS Foundation Trust

# Mosaicism

## An information leaflet for parents and families

If you need more advice about Mosaicism please contact:

**Clinical Genetics Unit**  
Birmingham Women's  
NHS Foundation Trust  
Mindelsohn Way  
Edgbaston  
Birmingham  
B15 2TG

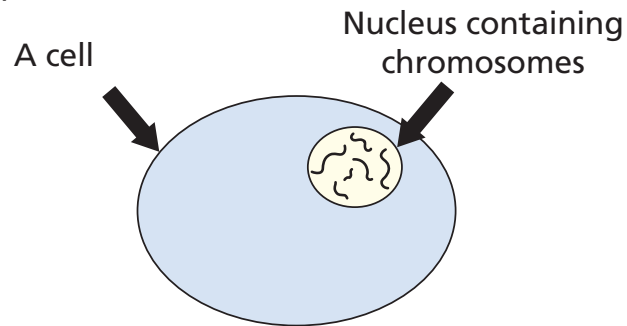
Telephone: 0121 627 2630  
Fax: 0121 627 2618

Email: [Clinicalgenetics.info@bwhct.nhs.uk](mailto:Clinicalgenetics.info@bwhct.nhs.uk)

## Introduction

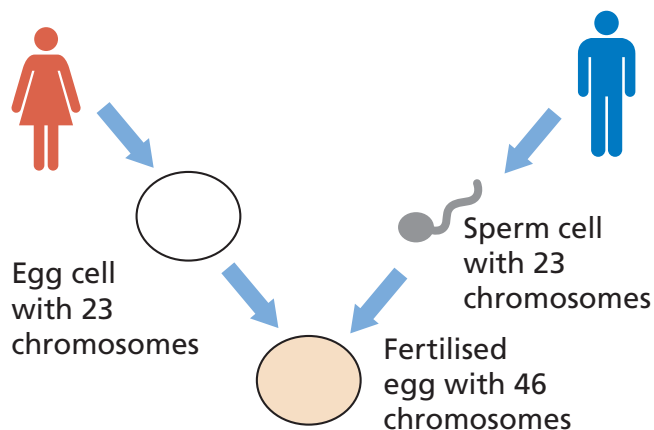
Our bodies are made up of cells. Within each cell is a complete set of our genetic material; genes are the instructions that make each of us an individual.

Genes are packaged in structures called chromosomes, which are found in the nucleus of each cell. Chromosomes come in pairs; we inherit one copy from each parent.



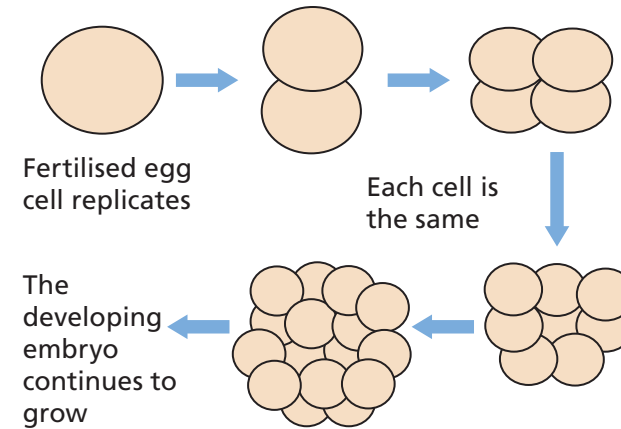
## How are genes and chromosomes inherited?

At conception the egg and sperm both bring half a set of chromosomes together. The fertilised egg has a complete set of chromosomes.



## Early development

A fertilised egg grows and replicates. Each time the cells divide, the chromosomes, and the genes they carry, are copied so that each new cell has an identical set of chromosomes.



Later, the cells begin to differentiate into different types of cell e.g. skin cells and muscle cells; they all contain the same genes and chromosomes but different genes are switched on or off.

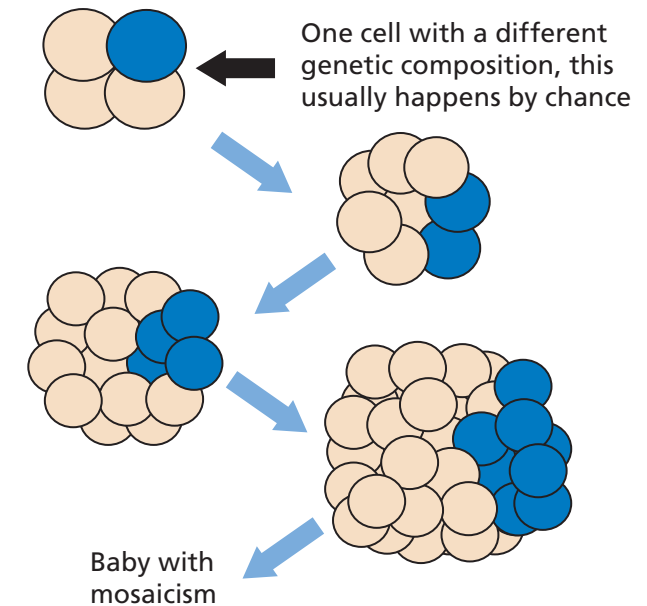
## What is mosaicism?

Sometimes when a cell divides the chromosomes do not copy or separate correctly. This can lead to an altered cell or cells that contain too few or too many chromosomes or a spelling mistake in a gene may occur which makes a gene not work properly in that cell.

Altered cells may continue to replicate resulting in an embryo which has "mosaicism" (also called a mosaic embryo). This simply means that it is composed of different populations of

cells; some being normal and some having altered chromosomes or genes.

## Diagram of mosaicism



## If a test shows a mosaic result what happens next?

It is difficult to predict the implications of a "mosaic" result as we do not know for definite what proportion of cells in the rest of the body have altered genes or chromosomes and what proportion are normal.

It may be possible to compare results from different types of cells (e.g. blood cells and skin cells). However no test can look at all the cells in a body.

People with a mosaicism may have symptoms of a disease caused by the altered genes or chromosomes.