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 CVS test (see our separate 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 and the rest of the baby's cells are normal. This is called confined placental mosaicism (or CPM) and is found in about 1% of CVS tests (see diagram below).

2. The baby may actually have a mosaic result and have a combination of normal cells and cells with altered genes/chromosomes.

If a CVS test shows a mosaic result what does this mean?

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. An amniocentesis may be offered to give more information.

Feedback

We appreciate and encourage feedback. If you need advice or are concerned about any aspect of your care or treatment please speak to a member of staff or contact the Patient Advice and Liaison Service (PALS):

Freephone: 0800 183 0204
From a mobile or abroad: 0115 924 9924 ext. 65412 or 62301
E-mail: pals@nuh.nhs.uk
Letter: NUH NHS Trust, c/o PALS, Freepost NEA 14614, Nottingham NG7 1BR

www.nuh.nhs.uk

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Our bodies are made up of cells. Within each cell there 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 (centre) of each cell. Chromosomes come in pairs, and 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 then has a complete set of chromosomes.

Early development
A fertilised egg grows and copies itself. 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 cells 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. Alternatively, a ‘spelling mistake’ may happen in a gene which stops it working properly in that cell.

Altered cells may continue to be copied, resulting in an embryo which has mosaicism (also called a mosaic embryo). This simply means that it is made up of different populations of cells: some are normal and some have altered chromosomes or genes.

Diagram of mosaicism

If a test shows a mosaic result what does this mean for a person?
It is difficult to predict the implications of a ‘mosaic’ result as we do not know for definite what proportion of cells in 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.