

Germline mosaicism is usually only confirmed if parents have more than one child with the same genetic or chromosome disorder when both parents are not affected or carriers of the condition.

Can you test a future pregnancy if germline mosaicism is a possibility?

Please discuss this issue with a doctor or genetic counsellor at your local genetics clinic. In some situations testing is possible. This test can only be done if the genetic alteration that affected the previous baby is known. Usually a test requires either an amniocentesis at around 16 weeks of pregnancy or a CVS at around 11 weeks, in order to obtain genetic material from the baby for testing.

The genetics clinic can give more information about these tests and have separate leaflets giving more details.

It is important to remember that germline is rare, and that usually, when healthy parents have a child affected by a genetic disorder, their risk of having a further affected child is very low.

For more information

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Birmingham Women's 
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Germline Mosaicism

An information leaflet for
parents and families

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Germline Mosaicism please contact:

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Introduction

Germline mosaicism, also known as gonadal mosaicism, can explain why healthy parents sometimes have more than one child with the same genetic or chromosomal disorder.

Germline mosaicism is uncommon, and is only found in a few genetic conditions and some chromosomal problems.

In order to understand more about germline mosaicism, it is important to know what genes and chromosomes are and what dominant inheritance means.

What are genes and chromosomes?

Genes are the instructions that make each of us an individual; a complete set of our genes is found in each cell of the body. All our genes come in pairs and we inherit one copy from each parent.

Each gene is an instruction for making a protein. Genes are packaged in structures called chromosomes. Each chromosome contains thousands of genes.

Humans have 46 chromosomes in most of the cells in the body, arranged in 23 pairs numbered 1-22 according to their size. The two remaining chromosomes, X and Y, are called the sex chromosomes. Females have two X chromosomes (XX). Males have one X chromosome and a Y chromosome (XY).

How can genes or chromosomes cause disease?

If a gene is altered then the instruction that it carries may not be read properly. This can cause disease. Usually, if someone has a gene alteration then this is found in every cell of the body.

If some chromosome material is missing or extra this can cause an imbalance of genetic messages. This can affect normal development. If someone has a chromosome alteration this is usually found in every cell of the body.

How are genetic and chromosomal diseases usually passed on?

Many genetic and chromosomal diseases are passed down through families. For this to happen one parent must be affected (dominant conditions), or both parents must be carriers (in recessive conditions). In other conditions the altered gene is on the X chromosome so these diseases are described as being X-linked. Separate leaflets give more information about dominant, recessive and X-linked inheritance.

Sometimes a genetic or chromosomal disease happens 'de novo'. This means it occurs for the first time in a family when a baby is conceived and is not due to altered genes or chromosomes in either parent.

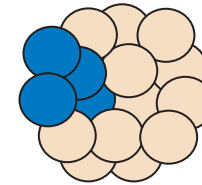
The chance that this random event will happen again is usually very low. There

may however be a small possibility of having another affected pregnancy if one parent has germline mosaicism.

What is germline mosaicism?

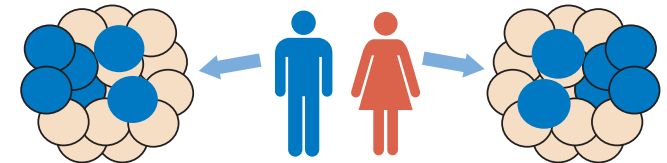
This term can be broken into two words. "Germline" refers to germline cells which are egg cells in women and sperm cells in men. These are the cells that fuse together when a baby is conceived.

"Mosaicism" means a combination of cells. Some have normal genes and chromosomes. Some will have altered genes or chromosomes.



Mosaicism - a collection of different cell types. Some cells have normal genes or chromosomes, some cells have altered genes or chromosomes.

If someone has germline mosaicism this means that some of their egg or sperm cells are normal and some contain a specific genetic alteration or chromosome problem.



The gene or chromosome alteration is only present in germline cells (egg or sperm cells) so would not be seen on a blood test. We cannot test egg or sperm cells for mosaicism.