NHS Genetic testing for Recurrent Miscarriage

If you have had three or more miscarriages, you are eligible for genetic testing on pregnancy loss tissue. This is to understand if there is a genetic reason that might help to explain why. This leaflet should be read alongside a discussion with your clinician. It does not replace the advice and guidance of the clinical team caring for you.

What do we mean by recurrent miscarriage?

Recurrent miscarriage means the loss of three pregnancies before 24 weeks' gestation. This includes any chemical pregnancies (very early miscarriages that usually happen in the first five weeks), but does not include ectopic or molar pregnancies.

Your losses don't all need to be with the same partner, and you may have had a successful pregnancy in between.

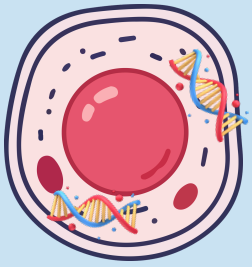
It is important to know

Genetic testing might not give you the reason for your miscarriage

It may give you information that you weren't expecting

You may still need other tests or scans

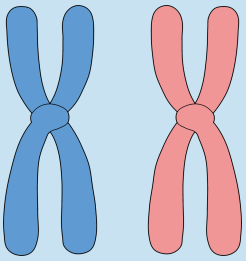
What do we mean by genetics?



We have 2 metres of DNA in almost every cell in our bodies.



Genes are small sections of DNA. We have around 23,000 genes which dictate who we are, what we look like and how our bodies work.



Genes are arranged into large structures called chromosomes. We inherit 23 chromosomes from each parent.

A note about language

This leaflet explains what genetic testing means and how it works. We've created this leaflet in partnership with women who have had personal experience of multiple miscarriages and genetic testing. We have chosen our language very carefully but we recognise that everyone will have very personal preferences.

NHS South East Genomic Medicine Service

supported by Miscarriage UK

Association of Early Pregnancy Units

Am I eligible for genetic testing?

Research shows that up to 60% of early miscarriages are caused by a genetic change in the pregnancy.

Miscarriages caused by a genetic change in the pregnancy are usually due to a change in the number of chromosomes. The most common are Down syndrome, Edwards syndrome & Patau's syndrome which are caused by an extra copy of the chromosome 21, 18 & 13 respectively. Changes to the number of chromosomes often lead to early pregnancy loss.

Although we know genetic changes are common causes of miscarriages, they are usually random events with no increased risk to future pregnancies. More information here.

Genetic testing is available on the NHS for women who have had recurrent miscarriages.

There are currently two genetic testing pathways available on the NHS:

1. **Genetic testing on pregnancy tissue:** is offered if you have had **three or more** miscarriages and pregnancy tissue is available for testing. This leaflet will talk you through this pathway.

2. **Genetic testing on parents:** is offered if you have had **five or more** miscarriages and pregnancy tissue is not available for testing. We have more information on our website www.southeastgenomics.nhs.uk

What genetic testing are you being offered?

The genetic test being offered looks for changes in the number of chromosomes in the pregnancy tissue. The test is carried out by specialists in a laboratory. If a chromosome change is found, this is likely to explain the reason for your miscarriage. If no change is found, the test will not be able to explain why your miscarriage happened.

How does the testing happen?

This test will require a sample of the pregnancy tissue to be collected and sent to a specialist genetic laboratory where it will be treated with sensitivity and respect.

Most genetic causes of miscarriage are due to random chromosomal changes in the pregnancy, and not a change that has been inherited from you or your partner. Therefore, testing of the pregnancy is more likely to give you a cause for your miscarriage, compared to testing you and your partner.

It can be difficult to collect a pregnancy sample, but support should be available to you from your care team. A sample from the pregnancy is essential to enable genetic testing to take place.

We have more detailed information on our website www.southeastgenomics.nhs.uk

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What are the possible results?

1 Chromosome change identified in pregnancy

- Testing could identify that the pregnancy has a chromosome change such as too many, or too few, chromosomes which would have caused a miscarriage. Depending on the specifics of this change, you and your partner may be offered further genetic testing. Your clinician will explain more.
- This is usually a random event and does not mean that any future pregnancies are more likely to miscarry. More info is on our website.
- The test can also reveal the sex of the embryo if you decide you want to know

2 No chromosome change identified in pregnancy

- Testing could identify that the pregnancy had the expected number of chromosomes (23 pairs)
- If there are no changes to the chromosomes, this test won't be able to determine the cause of your miscarriage
- It will not exclude any non-genetic causes such as hormonal or anatomy changes
- It will not detect smaller genetic changes such as changes to single genes.

3 No result

- Testing may be unable to identify the number of chromosomes in the pregnancy. There are a number of reasons for this, including too little DNA from the pregnancy tissue to get a reliable result, or too much DNA from the mother making it difficult to interpret the result.

When should you expect results?

The specialist genetic laboratory will be working to get your results back as quickly as possible. They aim to do this within 6 weeks, but delays are possible. A delay does not indicate any problems, but do contact your clinical team if you are concerned.

What happens next?

- This will depend on what the results show. Your team will advise you.
- You may decide that you want your pregnancy tissue to be returned to you. This can be arranged by your care team so do let them know before the genetic test is done.
- If the result shows the pregnancy has an additional full copy of chromosome 21, 18 or 13, you will be eligible for a specialist screening test called Non-Invasive Pre-natal Test (NIPT). This test will be offered if you become pregnant again and are over 10 weeks gestation. More information on our website.

Questions & support

Miscarriage UK miscarriageuk.org Tommy's tommys.org

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