

Information for patients and carers

Cytogenetic testing after recurrent early pregnancy loss



What is cytogenetic testing?

Cytogenetic testing looks at genetic material (DNA) that could help us find an explanation for why you have had recurrent early pregnancy losses. It checks for missing, extra or rearranged chromosomes. When this testing is carried out for recurrent early pregnancy losses, it can be performed on pregnancy tissue or parental blood samples.

Testing of pregnancy tissue is offered after a couple's third or subsequent pregnancy loss.

Testing of parental blood samples is offered after a couple's fifth or subsequent loss, where it has not been possible to obtain pregnancy tissue or the testing has been declined.

Why is cytogenetic testing offered?

Performing a test to look at the DNA contained within tissue from a pregnancy loss can help to give us answers as to why it has happened. We think that genetic causes are the most likely reasons for miscarriage to occur. Identification of any genetic abnormality can help us guide future treatment, look for reasons why miscarriages are happening, and assess the risk of it happening again in future pregnancies. It can help you make important decisions regarding any future pregnancy.

Testing is only carried out with express permission of the mother. In the case of parental blood samples, both parents need to agree.

Most samples sent for cytogenetic testing come back with a normal result.

Testing of pregnancy tissue

Testing of the pregnancy tissue itself gives us the most useful information about why the pregnancy loss might have happened.

Pregnancy tissue can be obtained either by having surgical management of miscarriage, including manual vacuum aspiration, or by retrieving pregnancy tissue following expectant or medical management of miscarriage. There is approximately a 20% chance that the test fails as DNA cannot be extracted, and around a 20% chance of contamination from the mother's cells. For this reason, a blood sample from the mother is always sent alongside pregnancy tissue to help the laboratory interpret the results.

Only a small amount of pregnancy tissue is sent for cytogenetic testing. Once it is processed, it is unlikely that any tissue will remain for sensitive disposal. Your care team will talk to you about any other testing that is required, and your wishes for sensitive disposal of any remaining pregnancy tissue – a separate leaflet explaining your options will be provided to you.

Testing of parental blood samples

Testing parents' blood is of limited utility compared to testing pregnancy tissue itself, as it cannot give us reasons for a specific pregnancy loss. However, it can identify other underlying genetic issues that could give useful information for future pregnancy care.

Sometimes the laboratory will request blood samples from both parents to help them interpret results from pregnancy tissue.

If parental testing is undertaken, typically the laboratory will not accept further testing of pregnancy tissue from any losses in future.

What are the risks of cytogenetic testing?

Emotional and psychological: Waiting for results can cause anxiety and distress. If a genetic abnormality is detected, this can also cause

worry and upset. If an abnormality is identified, you will typically be referred to a specialised genetics counsellor.

Limited predictive value: Most cytogenetic testing does not give an answer for why recurrent pregnancy losses happen. Sometimes it can reveal an abnormality for which the significance is unclear. It may not give any definite answers regarding future pregnancy care.

Contact details

If cytogenetic testing of pregnancy tissue is offered to you, you will need to inform your care team at the time if you wish to proceed with this.

Testing of parental blood samples is usually offered via the Recurrent Pregnancy Loss clinic. Contact details will be included on any letters sent from here.

Sources of further information

www.lancsteachinghospitals.nhs.uk

www.nhs.uk

www.accessable.co.uk

www.patient.co.uk

www.lancsteachinghospitals.nhs.uk/veteran-aware

<https://bepartofresearch.nihr.ac.uk/>

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patientexperienceandinvolve@LTHTR.nhs.uk

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