

Genetic Testing after a Pregnancy Loss

A pregnancy loss can be very difficult. This can be a time of emotional distress and disappointment. Sometimes it can be helpful to understand why a loss occurred. Physicians often order genetic testing to try to determine the cause of a pregnancy loss. Knowing the cause for a pregnancy loss can help predict the chances that such a loss will occur again in the future. This is especially true if the same person has had more than one pregnancy loss. Genetic testing can look for differences in an individual's DNA, genes, and/or chromosomes.

What is DNA? What are genes and chromosomes?

- DNA is genetic material we all have.
- DNA provides information our body needs to grow and develop.
- DNA is packaged into genes. Many genes together are packaged on a chromosome. Most people have 46 chromosomes.
- If there are too many or too few chromosomes or if there is extra or missing information on a single chromosome, a pregnancy loss may occur.

What does the SNP microarray test look for? Approximately half of all first trimester miscarriages are due to chromosome abnormalities.

- Chromosome studies can find extra or missing chromosomes, large pieces of missing genetic material, or large extra pieces of genetic material. In order to do this test, cells have to grow in a dish.
- Sometimes the chromosome test does not work because the cells do not grow.
- A Microarray test can find most large chromosome changes and also smaller pieces of extra (microduplication) or missing (microdeletion) genetic material (DNA). This test does not require cells to grow in a dish.
- SNP microarray testing may help to provide answers after a pregnancy loss.

What does the SNP microarray test NOT look for?

- There is no guarantee that this test will find the reason for your miscarriage.
- Some rearrangements of genetic material do not result in missing or extra pieces of DNA. These balanced rearrangements will not be detected with microarray testing.
- Genetic conditions are not always caused by extra or missing genetic material. Often a small change (point mutation) in a single gene can cause a health problem. These small changes cannot be found using chromosome or microarray studies.
- If only a small number of cells carry extra or missing material this is called mosaicism, which will not be detected if the number of cells with these changes is too small.
- These tests do not detect every possible genetic change.

When will I receive the results?

Test results can be expected within 14-21 days after the start of the test. The test may be delayed if insurance coverage clarification is needed.

How will I receive the results?

You can expect to receive results from the healthcare professional who ordered this test for you.

Who can I call if I have questions about the test results?

You can contact the healthcare professional who ordered this test. If you feel that your questions have not been answered, it may be helpful to speak with a prenatal genetic counselor.

What results can I expect?

- Possible results are normal, abnormal, or a result of unknown significance.
- An abnormal result is an indication of a deletion or duplication of genetic material. The loss or gain of genetic material may lead to one or more broken or missing genes.
- If the results are uncertain, we may recommend testing both parents.
- This test may find out if the parents who had a miscarriage are related by blood.
- This type of genetic testing is new. There is still a lot to learn about the effects of very small losses or gains of genetic material. This is why some results are not clear and are reported as uncertain clinical significance.

What is my risk of having a future pregnancy loss?

Risk is largely dependent on the cause for a pregnancy loss. Your healthcare professional can refer you to a genetic counselor to discuss risk of recurrence.