Fetal Autopsy: Genetic Investigations Information for parents

Finding out the cause of a condition can be important for many families. Understanding the genetic basis of an affected pregnancy may help health professionals give you information about the chance of a recurrence in a future pregnancy.
Genetic testing is a type of medical test that identifies alterations in chromosomes, or genes. There are different types of genetic tests that can be performed. They are designed to identify gene alterations that may cause a genetic disorder.

Types of genetic testing that may be performed:

1. MicroArray Testing

If you did not have prenatal diagnosis with microarray analysis, the first genetic investigation we will perform at autopsy is a microarray. This test uses techniques to look at the chromosomes to see if there is extra genetic material (duplication), or some missing genetic material (deletion). These chromosome changes can sometimes explain the abnormalities found in your pregnancy.

If the microarray does not find an explanation for the abnormalities detected, your case will be reviewed at a multidisciplinary meeting.

In over half the cases no further genetic testing will be recommended as it is more likely to cause confusion than provide answers. A negative genetic test will not completely rule out a genetic cause for the abnormalities detected.

If there is clear indication, further genetic testing will be undertaken.
In this situation, gene panel testing may be requested.

2. Gene Panel Testing

Gene panel testing involves looking at several different genes that have been grouped together on a panel. This enables testing to be performed at the same time and, in some situations, can be helpful in making a diagnosis. Panel testing looks for alterations in genes that could explain the abnormalities found in your pregnancy.

Why do we need DNA samples from both parents?

We need DNA samples from both biological parents so that the result of the genetic test in the pregnancy can be compared to the results from both parents. This allows us to find new alterations that have occurred in the affected pregnancy or identify inherited gene alterations that can cause a genetic disorder.

If the father is not present, a member of the team will fill in a genetic lab request form with his details for the father to date and sign. They will give you a spit kit (saliva sample) which you can give to the father. He can enclose his sample and consent form in the free-post envelope and send it to the Genetics Department.

How is the test done and how long do the results take?

Testing is performed via a blood sample (red/purple top EDTA tube) and/or saliva sample and the results usually take several weeks/months to come back, due to the amount of analysis involved.

After you have read this leaflet, if there is anything that you would like to discuss, we would be very happy to speak to you.

Please contact us on 0131 537 1116 and ask to speak with the duty Genetic Counsellor.

Local contacts

Your local genetics service:

Duty Genetic Counsellor

SE of Scotland Clinical Genetic Services,

MMC, Western General Hospital,
Crewe Road South,
Edinburgh, EH4 2XU.
Telephone: 0131 537 1116
Monday- Friday- 9am-5pm

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