

Parent Information: Genetic Testing

What is genetic testing?

Genetic tests are used to see whether a baby's condition might be associated with the genes and chromosomes (the so-called genetic blueprint) that the baby has received from his or her parents.

Why has your baby's doctor suggested this type of test?

There is a possibility that your baby has a condition that might be associated with his or her genetic make-up. The genetic tests may help in deciding whether this is the case with your baby.

What types of tests are used?

In general two types of genetic tests are used:

- 1. Chromosome analysis** – Chromosomes are the structures within our cells that contain many genes – the packets of genetic material that we inherit. Chromosome analysis will look for abnormalities of chromosomes. If such an abnormality is present it *may* help explain some or all aspects of your baby's condition. It is also possible that we may find abnormalities in the chromosomes unrelated to your baby's condition. In either case the results will be discussed with you.
- 2. Detailed analysis of a single gene** - Just one of the 30,000 genes in your baby is analysed in detail. This sort of test is highly specific and generally only gives information about a single genetic condition.

Your baby's doctors will discuss which type of test (or tests) is being used with your baby. The laboratory will store some of the blood that is taken in case other tests are required later.

What will this involve for my baby?

A small sample of blood will be taken for testing. If any other type of tissue is needed this will be discussed specifically with you.

When will the results be available?

The length of time it takes to get the results will vary. This can range from a few days to many weeks. Your baby's doctors will keep you updated and discuss the results with you at the earliest opportunity.

Will further tests be needed?

If a diagnosis is discovered through genetic testing it may sometimes be appropriate to test other family members such as parents and siblings, to determine whether the problem has been inherited. The doctor will discuss this with you if it becomes relevant for you and your baby.

Consent

Your doctor will be happy to discuss any questions you may have about genetic testing. If you have further questions staff from the genetics service can also be contacted to give you information and support. It is

important to understand that genetic testing may have implications for members of your family other than your baby.

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