You should be informed of the follow-up arrangements prior to discharge. A genetics appointment usually involves one of the doctors taking a family history, asking questions about your pregnancy and delivery, finding out about how your baby has progressed since birth and examining your baby. The examination will usually involve measuring the baby’s head, weighing the baby and doing a full examination to see whether there are any subtle clues to an underlying genetic diagnosis.

If you have any further questions please do not hesitate in speaking to the neonatal team looking after your baby or contact:
East Anglian Medical Genetics Service
Box: 134
Level 6, Addenbrooke’s Treatment Centre
Cambridge University Hospitals NHS Foundation Trust
Hills Road,
Cambridge,
CB2 0QQ

Tel: 01223 216446
Fax: 01223 217054

Further information:
UNIQUE Website for information about chromosome disorders and microarray
Contact a family Website for information regarding genetic disorders
www.cafamily.org.uk

This leaflet was given to you by:

If you would like this information in another language or audio, please contact
Interpreting services on telephone: 01223 348043, or email:
interpreting@addenbrookes.nhs.uk
For Large Print information please contact the patient information team:
patient.information@addenbrookes.nhs.uk

We are now a smoke-free site: smoking will not be allowed anywhere on the hospital site. For advice and support in quitting, contact your GP or the free NHS stop smoking helpline on 0800 169 0 169.

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<tr>
<th>Authors</th>
<th>Dr Ruth Armstrong</th>
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<tbody>
<tr>
<td>Pharmacist</td>
<td>N/A</td>
</tr>
<tr>
<td>Department</td>
<td>Cambridge University Hospitals NHS Foundation Trust, Hills Road, Cambridge, CB2 0QQ</td>
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<td><a href="http://www.cuh.org.uk">www.cuh.org.uk</a></td>
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<tr>
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Why have I been given this leaflet?
The doctors caring for your baby think that a genetic test or a referral to clinical genetics may be helpful. This leaflet addresses some of the questions that many parents have when ‘genetics’ is first mentioned in relation to their baby’s health.

What is a genetic condition?
The human body is made up of billions of individual cells. Most of these cells contain 23 pairs of chromosomes. The chromosomes contain the 21,000 genes which instruct the body how to develop and work properly. Genetic conditions can occur either because of a problem with the chromosomes such as Down’s syndrome, or with the individual genes for example, cystic fibrosis.

Why is my baby having a genetic test?
Your baby may be offered a genetic test for a number of different reasons. For example:
- a family history of a genetic condition
- problems identified on antenatal ultrasound scan or blood tests
- concerns about a baby’s health, growth or development.

What types of genetic tests are there?
Chromosome analysis (karyotype) involves looking down a microscope at your baby’s chromosomes to see whether the correct number of chromosomes is present and whether they are in the right place. This type of test detects conditions such as Down’s syndrome.

Microarray studies look at the chromosomes in much greater detail to see whether small pieces of chromosome are missing or duplicated. This technology sometimes identifies chromosome abnormalities but we are uncertain as to whether they are causing your baby’s problems. Sometimes we need to obtain a sample of blood from the parents to help clarify this. Rarely, we identify abnormalities that are unrelated to a baby’s current problems but may have implications for their future health. Further information can be obtained regarding microarray analysis from the UNIQUE website.

Molecular (DNA) studies involve looking at specific genes. The type of test depends on the problems your baby is experiencing. Each specific test will be discussed with you. It is usual practice for a sample of DNA to be stored so that if further testing is available in the future, your baby does not have to have another blood test.

What does the genetic test involve?
Genetic tests usually involve a blood sample being sent to the laboratory for further analysis. Some genetic tests may be available within a few days; others may take weeks and some several months. The doctor ordering the test will tell you when to expect the results and how the result will be given to you.

When is the right time to have a genetic test?
Some families are offered genetic testing during a pregnancy whilst others ask for testing after birth. It may be helpful to have a genetic test during the first few weeks of life as the results may help determine treatment. However, if there are no immediate treatment issues, some families may wish to delay genetic testing until they see the neonatal team or geneticist in the outpatient’s clinic (usually a few months after discharge).

What does a referral to genetics involve?
Some babies may benefit from referral to clinical genetics for further evaluation. Some are seen whilst they are still on the Neonatal Unit whilst others may be offered an appointment in clinic following discharge. This should not delay your baby going home as clinics are held throughout East Anglia. If you are outside of the East Anglia catchment area, arrangements will be made for you to be seen by your local genetics service.