

Some Information about your Appointment

This information has been prepared for patients and families who have an appointment to see a genetic specialist. The aim is to give you some information about why you have been referred to see the specialist, and what you can expect to happen during your appointment.

Why have you been referred to see the specialist?

Some of the reasons for being referred include:

- Because there is a genetic condition that happens in your family or your partner's family.
- You or your partner have a child with learning difficulties, developmental delay or health problems. Your doctor thinks there may be a genetic link.
- You or your partner have a genetic condition that might be passed on to your children.
- You have had another type of test that is done during pregnancy (such as an ultrasound, nuchal translucency scan or blood test). It shows that there is an increased risk that your baby has a genetic condition.
- You or your partner have had a miscarriage or stillbirth.
- Particular types of cancer have occurred in several close relatives.
- You and your partner are close relatives and would like to have a baby.
- Your doctor believes that a genetic specialist may be able to provide you with additional information about your current health.

How will you be helped?

There are several ways that the specialist may be able to help you. Some of these are:

- Explaining the types of test available that will help confirm a diagnosis.
- Making or confirming a diagnosis.
- Providing information about the condition and how it is passed on.
- Discussing the risk that you may be affected by the condition in the future.
- Discussing the risk that future children may be affected with the condition if it already occurs in your family.
- Talking about ways of living with the condition and the medical and social support available to you.
- Answering any questions you may have about the condition diagnosed.

Before your appointment

It can be helpful for the specialist to have medical details about other people in your family, including those affected by health problems which may have a genetic cause.

Please bring the following details of family members as far back as your grandparents generation. Write them in the spaces provided.

Relationship to you e.g. mother, uncle	Full Name	Date of Birth (and date of death if relevant)

If appropriate make a note of:

- **Their genetic diagnosis and details of any other health problems and the age at which they were diagnosed**

- **When and where they were diagnosed or treated**

For affected family members you may be asked for their address and their family doctor's name and address.

We understand that it is not always possible to give these details. Information provided by you will be treated confidentially and will only be shared with other relatives or health professionals involved in your care **with your permission**. Relatives will never be contacted without your permission.

Bring any questions or concerns with you to your appointment – write them down. You might also want to bring your partner or another relative or friend with you. If you need an interpreter, let the department know.

After the appointment it is likely that you will be sent written information that covers the topics that were discussed. This will help you to remember everything. You might also want to show the written information to other members of your family. Ask the specialist whether written information will be given to you and how soon you can expect it to arrive.

More information can be obtained from your local regional genetics centre (www.gig.org.uk/services.htm) or from these addresses:

The Genetic Interest Group

Unit 4D, Leroy House,
436 Essex Rd.,
London, N1 3QP
Telephone: 0207704 3141
Provides information about specific genetic conditions and contact details of support organisations.
Email: mail@gig.org.uk
Web: www.gig.org.uk

British Society of Human Genetics

Information for patients available at:
www.bshg.org.uk/for_patients/for_patients.htm

Orphanet

Free-access website providing information on rare diseases and orphan drugs, and links to support groups across Europe.
Web: www.orpha.net

EuroGentest

Free-access website providing information about genetic testing and links to support groups across Europe.
Web: www.eurogentest.org

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