

Understanding
chromosome
disorders

Unique



A Clinical Genetics Appointment



rarechromo.org

This leaflet has been written to explain what you might expect if your family has been referred for a clinical genetics appointment.

What is clinical genetics?

Clinical genetics is a medical specialty and is a service that is available within certain hospitals. Most clinical genetics teams are based in a single regional teaching hospital but travel to other hospitals throughout the region to hold regular clinics. In 2016, when this leaflet was written, there were 33 clinical genetics centres throughout the UK and Ireland.

The clinical genetics team are specialists in genetics and see patients and their families who are thought, or known, to have a genetic condition or be at risk of one. They discuss and may arrange genetic testing. They also help to coordinate any medical care together with help from a multidisciplinary team who may include other medical specialists, nursing teams, and other health professionals such as physiotherapists and the GP.

Why have I been offered a clinical genetics appointment?

Your (or your child's) GP or specialist will have referred you to your local clinical genetics service. People are seen in genetic clinics for many different reasons:



You may have a family member who has been diagnosed with a genetic condition.

You may have a family member with medical and/or learning difficulties and your doctor thinks they may be due to a genetic condition.

You may have a known genetic condition in the family which is heritable.

You may be pregnant and there is some risk that your baby could have a genetic condition, either because of a known condition within the family, or because of findings on a scan or test during pregnancy.

There may be a known gene alteration in the family which can increase the risk of a particular medical condition such as heart disease or cancer.

Who will I see at my appointment?

You will see a member of the clinical genetics team. The team consists of clinical geneticists (doctors), specialist nurses and genetic counsellors. For some conditions, such as children with a chromosomal change, it is most likely that you will see a clinical geneticist (doctor) but in some UK genetic centres you may see a genetic counsellor.

Clinical geneticists are experienced medical doctors who have already undergone substantial training in adult or child (paediatric) medicine before further specialising in genetics. They usually see families in a hospital clinic, or occasionally on a hospital ward, and their role is to diagnose people with genetic conditions and help coordinate the care of families who have genetic conditions.

Genetic counsellors are healthcare professionals who are specially trained in genetics. They see people in clinic to talk about the family history where a diagnosis is known, to provide information, genetic risk assessment, support and to offer testing if indicated.

What will happen at my appointment?

Before the appointment

- Appointments will usually last between 30 and 60 minutes so that there is plenty of time for a full discussion.
- Before your appointment, you may be contacted and asked for some details about your family history. You may be asked to fill in a family history form. If so, please fill it in as fully and accurately as you can and return it in plenty of time so the genetics team can ask for any additional information or medical records that are needed before the appointment. Sometimes you may be asked to pass on consent forms for your relatives to sign if they are happy for the department to access their medical records for information that may be relevant for you or your family.
- Occasionally you may be asked to bring family photos with you. It may be helpful to write down any questions you have before your appointment and bring these with you (see 'Questions' section).



During the appointment

If you have a family member who is likely to need testing, or has been tested, the geneticist will ask questions about the family in general, and especially about the affected person. This is likely to include questions about the pregnancy and birth of the affected person, their development and schooling and their medical history.

- The geneticist is likely to wish to examine the affected family member. This is to perform a general medical examination e.g. listening to the heart. It is also to look for signs of a genetic diagnosis which can occasionally be found on examination e.g. skin markings. Sometimes they will ask to examine other family members as well, for example the parents or any brothers and sisters.

- The geneticist is likely to ask your permission to take photos of the affected person. This is so that there is a record of any special features and it is also most helpful for comparison, especially in children, if they are then seen again at an older age. The photos will be kept securely, alongside medical records.

- The geneticist should then discuss with you the findings so far. This may include the results of testing that other doctors have already done, e.g. your child's paediatrician. They should go through the findings with you, if you wish, and answer your questions.

- The appointment will also include details about the medical management of the condition, how likely it is to occur in other family members and what options are available for your family.

- A genetic test may be available. This may already have been carried out in some cases. It is most often done with a blood test. Testing with a mouth swab or a saliva sample may also be possible and this option can be helpful for children and adults who are needle-phobic. If testing is available, you will be asked if you are happy for you or your child to have testing and to sign a consent form.



Genetic tests sometimes need to be carried out on more than one member of the family. Sometimes, however, despite expectations of the family and the referring doctor, genetic tests cannot be offered, especially when the affected member of the family is no longer alive.

- If you are pregnant, there may be testing options that are available and these will be fully discussed with you and your family.
- The chances of a genetic condition happening again in the family will also be discussed with you, if this is relevant.

After the appointment

- You will receive a summary letter of your appointment and a copy of any relevant information leaflets. It may also be possible for the clinical genetics team to put you in touch with other families who have a member with the same condition or with a specific support group, such as *Unique*.
- Your genetics team member should have discussed with you whether you wish or need to be seen by the genetics team again. Sometimes families are just seen once by the genetics team and then further care is transferred to the paediatrician, GP and/or other specialists. However, sometimes families are followed up at certain time periods by the genetics team, or when an affected child is reaching the age when they may be able to have children themselves.

In this last case, you will usually have to ask your GP for a referral back to genetics at the appropriate time.

- If testing has been organised in the clinic, your genetics team member will discuss how you would like your results given to you (for example, by telephone or letter). Occasionally another appointment will be arranged for you to receive the results. The genetics team member will let you know how long the testing will take, as genetic testing is complex and often takes longer than other blood tests.
- Your GP and other specialists should receive a letter from the geneticist, giving information on the condition and outlining appropriate care. Remember that most genetic conditions are very rare, so you or your child's GP, medical specialists (or indeed the geneticist!) may not have seen it before.

Research and teaching

Genetic services are usually involved in medical teaching and research. At your appointment the healthcare professional may discuss any research that is relevant to you or your family. This would be offered as an additional option to your routine clinical care and it is completely your choice whether you wish to take part. It will not affect routine medical care if you or your family do not wish to take part.

If the genetics department is part of a teaching hospital, you may be asked if other healthcare professionals or students may sit in in your appointment to observe.

Questions

It can often be helpful to write down any questions you would like answered and bring them to the genetics appointment. Please let your doctor or genetic counsellor know that you have a list of questions at the start of the appointment, so they can plan the appointment to include answering them.

Questions that other Unique families have found helpful to ask have included:

Questions about the appointment/testing:

- 🔴 Why have we been given a genetics appointment?
- 🟢 Why was my child tested/ why do you feel our child needs testing?
- 🟡 How long will test results take to come through?
- 🟡 Can we have a discussion without my child being present?
- 🔴 Please can you go through the genetic report with us? What do all the numbers / letters mean on the microarray/sequencing results?
- 🟢 What genes are affected in my child's condition and what do those genes do?
- 🟡 Please could you explain what genes and chromosomes are?
- 🟡 My child is needle-phobic. Are there any alternatives to a blood test?

Questions about the family:

- 🔴 What does the future hold for our child / me/ my partner?
- 🟢 Do I and my partner need testing?
- 🟡 What is the chance of passing on the condition to further children?
- 🟡 If my affected child were to have a child of their own, would their child also be affected?
- 🔴 Will other members of the family need testing? Can brothers and sisters be tested? Why are brothers and sisters sometimes not offered testing?
- 🟢 Are there any alternative reproductive options so we do not have another affected child?

- 🔍 How (and possibly when) do we explain the disorder to our child?
- 🔍 How do we tell their brother and sister and other close family members like grandparents?
- 🔍 Is there any emotional counselling available to cope with the diagnosis?
- 🔍 What happens after the genetics appointment?

Questions about short and longer term care:

- 🔍 Have you had experience of a child with the same condition as mine?
- 🔍 Will we see a geneticist again in the future? Does the genetics department give follow up appointments?
- 🔍 Are there any associated medical conditions? Will my child need further screening/testing/follow up appointments for these?
- 🔍 Will my child be under the care of a paediatrician (children's doctor)?
- 🔍 What services am I likely to need?
- 🔍 Is my child likely to attend a mainstream or a special school?
- 🔍 Can I have a letter for the school/ my GP detailing my child's condition and the services/resources they will need?
- 🔍 What should I do if my child wants to know more about their diagnosis in the future? Can we come back to the genetics service?
- 🔍 How can I obtain more information on this condition? Are there any medical papers on it? Is there any family-friendly information?
- 🔍 Is there information in languages other than English?
- 🔍 Is there a specialist or specialised centre for this condition?
- 🔍 Will my child make friends? Will they be able to play with their brother and sister?
- 🔍 Could my child have a family of their own later in life?
- 🔍 Where can I go for support? e.g. support groups/parent groups
- 🔍 Can my child be cured? (e.g. stem cell therapy/gene therapy)
- 🔍 What happens when my child is an adult?
- 🔍 Is my child likely to have a normal life span?
- 🔍 Is there anyone I can speak to if I have follow on questions after this appointment?

Inform Network Support



Rare Chromosome Disorder Support Group

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Join Unique for family links, information and support. Unique is a charity without government funding, existing entirely on donations and grants. If you can please make a donation via our website at:

<http://www.rarechromo.org/html/DonateNow.asp>

Please help us to help you!

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