

Craniofacial Team

Why is Genetics Important?

This leaflet only gives general information. You must always discuss the individual treatment of your child with the appropriate member of staff. Do not rely on this leaflet alone for information about your child's treatment.

This information can be made available in other languages and formats if requested.

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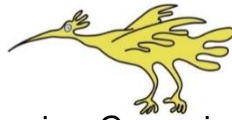
Information for patients

Why are genetics so important?

Each person has about 20,000 pairs of genes. They work like instructions that determine things such as the colour of our eyes, how many fingers and toes we have and how tall or short we are. Genes are arranged along strands, rather like beads along a string. These strands are called chromosomes and are inside most of the cells in our body. Most people will have changes or alterations within their genes or chromosomes. However, sometimes these changes change the instructions and cause a medical condition such as a craniofacial condition.

These gene/chromosome changes are sometimes passed through families and from parent to child. At other times the gene change starts for the first time in that person. Some people with gene/chromosome changes have other medical problems when they are born, as well as a craniofacial condition.

When should I go to a Genomic Medicine clinic?



There is no set time in your life that you need to be seen in a Genomic medicine clinic. You do not have to arrange an appointment if you do not want one. However, you may want an appointment to talk about your personal and family information before you think about having children of your own, to discuss the options available to you. The genomic medicine clinicians see individuals at any age and are there to support you by providing information and helping you make decisions throughout your life.

What happens at a Genomic Medicine appointment?

Your appointment will be with a genomics clinician who will either be a Genetic Counsellor or Doctor specialising in Clinical Genomics. At your appointment, the clinician will discuss your personal history and then review your family history (if you have previously been seen in a Genomic Medicine you may have had your family pedigree drawn). This enables the consideration of other family members who may also be affected with the condition.

The clinician may offer some genetic testing to try and confirm a diagnosis, as they will then tell you how likely it is that other members of the family, including any children you may have in the future, could be born with a craniofacial condition. If genetic testing has already taken place, then the clinician can discuss with you what has been found and help you to interpret and understand the information. They may talk to you about any additional investigations that could be useful. They may also discuss any options available to you if you are planning on having children of your own. Don't worry if you can't remember everything that was said, you will receive a letter after your appointment.

If I plan to have children in the future, will they have a craniofacial condition?

If you have been diagnosed with a craniofacial condition, it is a possibility that when you go on to have your own children, they can inherit the condition from you. If/when you start planning your own family in the future, you may want to come to Genomic Medicine clinic to talk about reproductive choices. There is now a specialised IVF technique called Preimplantation Genetic Diagnosis (PGD) used to find out whether an embryo has a genetic condition before pregnancy. This is done in a fertility clinic using IVF technology. There are also tests that could be offered to a lady during pregnancy to see whether the baby has a genetic condition. To discuss your prenatal options, ask your GP or consultant and ask to be referred to your local Clinical Genomics service.

How do I get an appointment in clinic genetics?



Ask your GP or the healthcare professionals involved in your care to refer you to your local Genomic Medicine clinic.

If you have any questions and would value a telephone appointment, please ask the craniofacial team to refer you to the Genetic Counsellor. If following the telephone consultation, it is thought that a clinic appointment is beneficial, then they can refer you to your local Genomic Medicine clinic.