NHS
Liverpool Women's
NHS Foundation Trust

NEWSLETTER

Liverpool Centre for Genomic Medicine (LCGM)

Issue 1
April 2024

WELCOME

Welcome to the first edition of the Liverpool Centre for Genomic Medicine (LCGM) newsletter. We aim to deliver a quarterly newsletter providing you with information about our team, service developments, patient stories, condition specific information, current research in Genomics and exciting Genomics news.

In this First Edition

We aim to introduce you to our service, where we are located, a real-life patient story and current research updates

Firstly, let us tell you about where we are located and what we do

Where we are

Our regional Genomics Service covers, Cheshire, Merseyside and the Isle of Man. We are known as the Liverpool Centre for Genomic Medicine (LCGM) and we are located within the Crown Street site of the Liverpool Women's hospital. We deliver clinics across the region, such as Alder Hey, Liverpool Heart and Chest Hospital, Chester Hospital, Leighton Hospital, Warrington Hospital, St.Helens and Knowsley hospital and Ormskirk Hospitals.

What we do

Our main role is to diagnose and counsel families with possible or known inherited disorders. We receive referrals for families who have or at risk of a having a genetic condition. Our team of specialists support families by providing...

- An accurate diagnosis
- Information about how a genetic condition may develop
- Assessments on the risk of transmitting a genetic condition
- Support for families with understandable information and counselling

How we help individuals and families

There are lots of other ways we help individuals and families. In the main we..

Help patients to understand the types of genetic testing available

Help patients and their family adapt to the impact of a genetic condition for their future health

Help patients understand health information about a genetic condition

Help patients to understand the possible outcomes and implications of this genetic testing

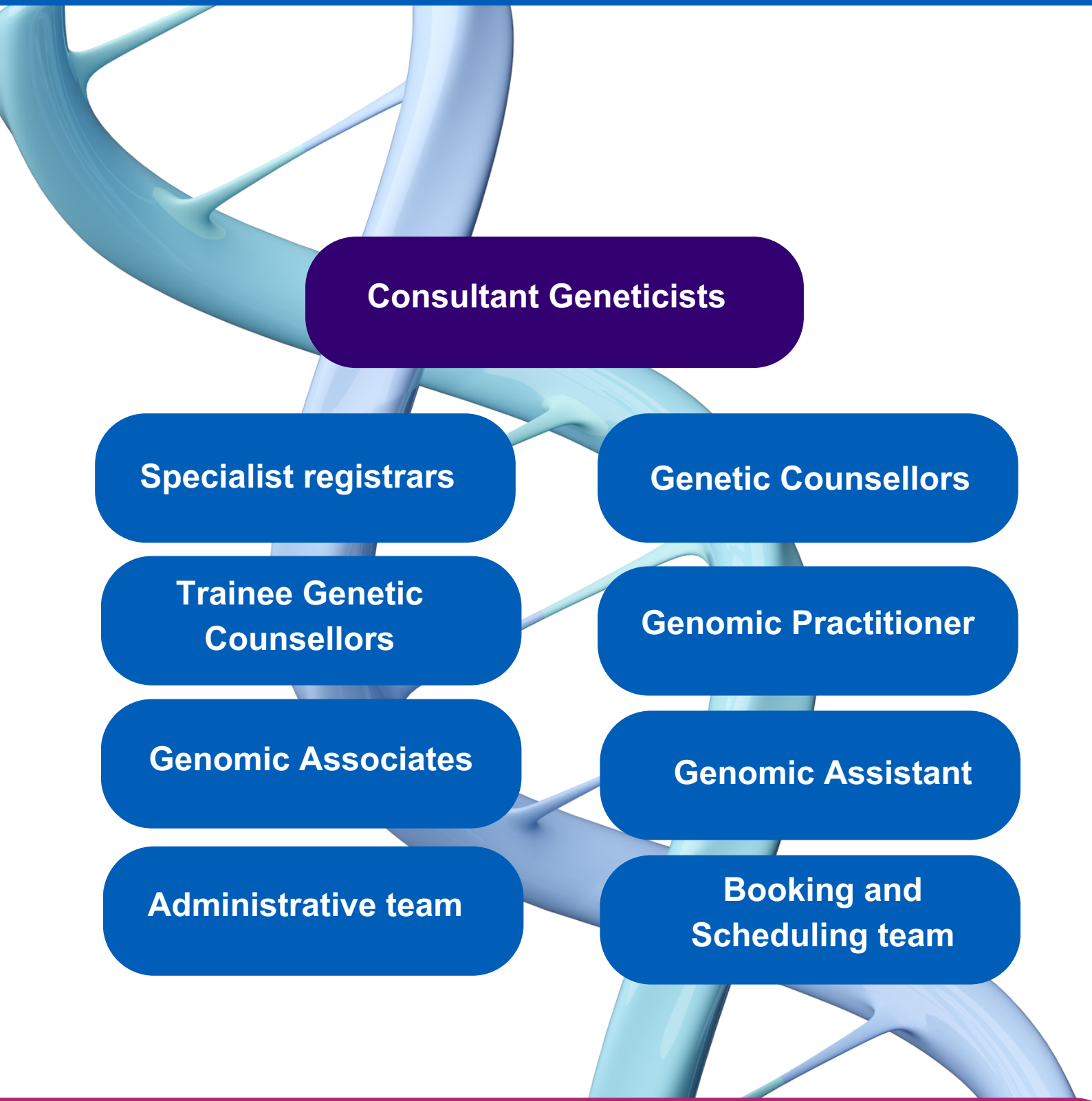
Help patients to understand the genetic risks for current and/or future children

Help patients with, or at risk of, a genetic condition by referring them for appropriate ongoing screening

Help patients to find support through the NHS, or associated charities, on living with a genetic condition

If you are worried about your family history, in the first instance, we would recommend you discuss this with your GP. They will be able to offer advice and decide if a referral to our service is indicated.

Our team consists of...



- Consultant Geneticists
- Specialist registrars
- Genetic Counsellors
- Trainee Genetic Counsellors
- Genomic Practitioner
- Genomic Associates
- Genomic Assistant
- Administrative team
- Booking and Scheduling team

With each edition, we will aim to give you an overview of the different roles within our service.

Research studies

The GENROC Study

In this edition, we have summarised information about two on-going research studies. We have included details about the aim of these research studies, who can take part and what taking part in the research study would involve.

The GENROC Study: Improving the treatment of people with GENetic Rare disease: an Observational Cohort study

GENROC is a research study that aims to work with families to increase understanding of growth and development of children affected with rare genetic conditions. With improved understanding of rare genetic condition, this study aims to improve medical care for children affected. Involvement within the study includes consent to collection of medical and educational information about the child diagnosed with a rare genetic condition and for some of this non-identifiable information to be uploaded anonymously to DECIPHER- a world-wide website used by health professionals to help provide a better understanding of these rare genetic conditions.

If you are interested in this study, please contact your genetics healthcare professional.

Aim of the Study?

Improve understanding of rare genetic conditions that affect children to improve their medical care

Who can take part?

Children with rare genetic conditions included in the study

What does it involve?

Consent to medical and educational information & completing online questionnaires about your child

For more information about GENROC research study, please visit https://redcap.link/GENROC

Research studies

The Precision-HBOC study

Precision Hereditary Breast and Ovarian Cancer (HBOC) Precision-HBOC study is a research study looking at personalised cancer risks for women undergoing predictive (pre-symptomatic) genetic testing for a known gene variant in their family linked to breast and ovarian cancer. The aim of the study is to assess whether providing women personalised risk estimates for developing breast and ovarian helps making decisions regarding risk medical management e.g. health surveillance and risk reducing surgery

In this study, women are either randomised to the intervention arm or control arm. In the intervention arm, women are estimated personalised risk of breast and ovarian cancer based on additional analysis of their DNA sample. While, in the control arm women are estimated typical breast and ovarian cancer risk estimates. Participation involvement includes completing some questionnaires over the course of a year. Some participants may be approached for interviews.

Aim of the Study?

Assess if personalised breast and ovarian cancer risks help women who carry certain gene variants associated with increased breast and ovarian cancer risk make decisions about their medical risk management

Who can take part?

Women who are undergoing predictive genetic testing for a known gene variant linked to breast and ovarian cancer

What does it involve?

Additional analysis of the woman's DNA sample & completing various questionnaires about medical risk management

For more information about precision HBOC research study, please visit https://www.isrctn.com/ISRCTN15331714

Patient Story

Grayson

When did you notice that Grayson was different?

Grayson was a good birth weight despite being a month early, he fed well and grew, maintaining a steady curve between the 25th and 50th centile. My original concerns were when Grayson started attending pre-school around age 3. It was easily noticeable that he was shorter than his peers and this seemed to become more and more obvious as time moved on. My original thought to this was that he may have a growth spurt a little later than the other children his age and soon catch up. Once Grayson started to attend reception year and he continued to not grow very much (around 3 to 4cm a year) my concerns became greater and towards the middle of 2021 when Grayson was 5 I contacted our GP to express my concerns. At this stage Grayson had now fallen into the 2nd centile for his height.

What has your experience been with regards to medical investigations before genetics consultation?

Testing for Grayson began very quickly with the GP understanding my concerns given his drop on the centile chart. Original testing was to indicate any issues with iron levels and thyroid function. After these came back as negative, we were referred to the local hospital paediatrics. At our first appointment with paediatrics further tests were carried out to measure Grayson's bone age using an x-ray and further blood tests. At this appointment the doctor also noted a possible heart murmur, so we were referred to cardiology for that as well. Results from these tests showed that Grayson's bone age was for aged 3 he was 5 and 4 months at the time, he also showed low IGF1 levels, his cardiology appointment showed that his murmur was innocent. From these results the doctor then referred Grayson for a hormone stimulation test. The doctor also noted some skeletal facial features that prompted her to also refer us to genetic testing. The results from the stimulation test showed Grayson's IGF1 levels were persistently low.

My overall experience with all the different blood tests and appointments Grayson has attended has been incredibly positive. The tests carried out were sometimes difficult for Grayson but all the nurses and doctors we have dealt with along the way have been fantastic. I have always been kept informed of results from any appointments and anything I was unsure of has always been fully explained to me.

Genetics consultation is not very common, so most people will not be aware of what it entails. What were your thoughts prior to the genetics appointment?

My original thoughts for the genetic appointment were that family history would be discussed and blood samples taken. I didn't really believe that there would be any abnormal genes found given that there has been no family history of anything concerning height or growth.

How did the genetics appointments go? What did you feel afterwards?

The genetics appointment was incredibly informative and thorough. After the initial appointment it became more apparent that there was obviously a gene that Grayson and myself must share. Given how well it was explained to us Grayson didn't just resemble me because I was his mother but because we both had similar characteristics of Noonan syndrome. Previous to the appointment I had never heard of Noonan syndrome despite it being reasonably common. **Once the diagnosis for Noonan syndrome had been confirmed it was a relief that we had a reason for Grayson's short stature and to also be able to monitor him for any other factors associated with Noonan**. Grayson's father and myself were then tested to see if one or both of us carried the gene and it came back that I carried the gene, this was not a shock given the information and explanations we had been given at the previous appointment. My feelings after the appointment were very positive as I had reached 36 years of age and had no idea, I had Noonan. I felt that the diagnosis would not impact Grayson very much and that we could just continue to do what we could with his short stature. I know there will always be information available for Grayson as he gets older and would like to understand more about the condition but for now, **he is a happy, healthy boy who is just enjoying his childhood.**

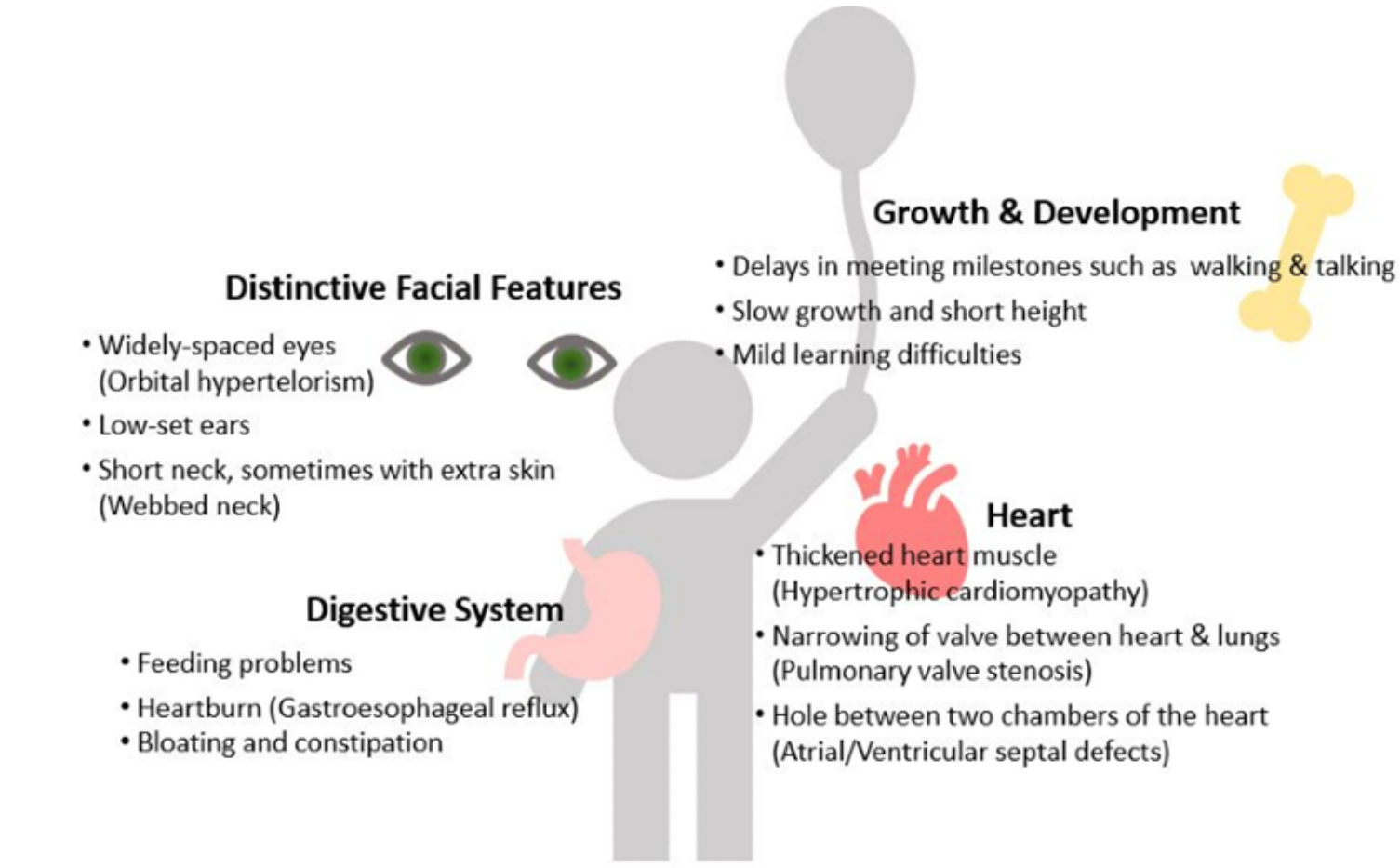
NOONAN SYNDROME

What is Noonan syndrome?

Noonan syndrome is a genetic condition that is part of a larger group of conditions called RASopathies. RASopathies arise when something goes wrong in a pathway involved in the way the cells of our bodies communicate the information they need to function properly. Since the cells of our bodies make up our tissues, organs and organ systems, RASopathies can have widespread effects on growth, development and health.

What are the most common features?

The specific features each person has can be different, even within the same family. Below is a summary of the most common features seen in Noonan syndrome.



What causes Noonan syndrome?

Noonan syndrome is caused by a fault in one of several genes. The first gene to be discovered was the **PTPN11 gene** but it is now known that several genes can be involved.

In some cases, the faulty gene associated with Noonan syndrome **is inherited from one of the child's parents.** The parent with the faulty gene may or may not have obvious features of the condition themselves.

In other cases, the condition is caused by a **new genetic fault that isn't inherited from either parent.** In these cases, the chance of the parents having another child with Noonan syndrome is very small.

Can Noonan syndrome be passed on to future children?

Noonan syndrome is usually inherited in an autosomal dominant pattern which means that an individual with this condition has a **50% chance of passing it on to future children.**

What is the treatment?

There's currently no single treatment for Noonan syndrome. The treatment and management are based on the individual's specific needs. **Growth hormone** may be used to help with growth and **severe heart problems may need an operation.**

What is the outlook?

The outlook **can range from being very mild to severe and life-threatening**. Almost all children with Noonan syndrome reach adulthood and most can lead normal, independent lives. Most people with Noonan syndrome will need to have their heart monitored regularly throughout their life.

information and support groups:

https://www.nhs.uk/conditions/noonan-syndrome

https://www.noonansyndrome.org.uk

Noonan Syndrome Association

LATEST NEWS

News Health

Breakthrough Alzheimer's blood test could detect disease 15 years before symptoms emerge

Simple procedure could lead to a national screening programme for everyone aged over 50, say experts

A simple blood test can detect Alzheimer's up to 15 years before symptoms emerge, with experts claiming it could "revolutionise" early diagnosis of the disease.

Swedish trials found the test to be as accurate at detecting the signs of Alzheimer's as painful lumbar punctures, and better than a range of other tests currently being worked on.
Experts say it could pave the way for a national screening programme for the over-50s, and that current treatments could work better with the cases picked up earlier.

The test works by measuring levels of a protein called p-tau217 in the blood that indicate biological changes taking place in the brain during Alzheimer's disease.

It could identify patients as likely, intermediate, or unlikely to have Alzheimer's disease and potentially rule out the need for further, more invasive investigations.

Although the article states that it could pave the way for a national screening programme for the over-50s, this is not something that is available currently but may be available in the future

There are different types of Alzheimer's disease. In most cases, Alzheimer's does not have a single genetic cause. Instead, it can be influenced by multiple genes (genetic risk variants) in combination with lifestyle factors (non-genetic factors). Consequently, a person may carry more than one genetic variant or group of variants that can either increase or reduce the risk of Alzheimer's. Due to **the complex risk factors that lie behind the more common form of Alzheimer's disease, the NHS does not offer a genetic test for it.** The Familial Alzheimer's disease is a type of Alzheimer's that is caused by a single-gene change and genetic testing is available within the NHS. It is very rare. If several of your family members have developed dementia over the generations, and particularly at a young age (<55 years), you may want to seek genetic counselling for information and advice about your chances of developing Alzheimer's disease when you're older.

Prof David Curtis, honorary professor, UCL Genetics Institute, University College London, said: "Everybody over 50 could be routinely screened every few years, in much the same way as they are now screened for high cholesterol.

"It is possible that currently available treatments for Alzheimer's disease would work better in those diagnosed early in this way. However, I think the real hope is that better treatments can also be developed.
"The combination of a simple screening test with an effective treatment for Alzheimer's disease would have a dramatic impact for individuals and for society."

You may wish to look at
https://www.independent.co.uk/news/health/alzheimers-diagnosis-treatment-blood-test-b2482854.html for the full article.

Further information on P-Tau 217: A Promising Blood-Based Biomarker for Alzheimer's Disease visit:

https://www.quanterix.com/p-tau-217-a-promising-blood-based-biomarker-for-alzheimers-disease/

Biomarkers are biological measurements that can be used for a variety of purposes, including identifying individuals who are at **high risk of developing a disease**, **detecting disease early** at a stage when it is treatable and diagnostic classification for **personalized treatment** based on a biological characterization of the disease of each individual patient. Examples include the Prostate-specific antigen used in the diagnosis and monitoring of prostate cancer, HbA1c used to diagnose and monitor diabetes and the HER2 which is a protein that is overexpressed in some types of breast cancer and therefore may predict the response to specific therapies. The p-tau 217 is a promising new biomarker for early detection and monitoring of Alzheimer's disease. The sequencing of the human genome has provided an important body of information for the development of biomarkers for all of the purposes mentioned.

Events

In this edition, we have summarised some events upcoming in the UK. These events have been organised to provide support for families affected by such rare diseases as well as help promote awareness. These events are either held online or in person.

EVENT	WHEN	WHERE
Noonan Syndrome Association: Conference & Families weekend	Friday 26th – Sunday 28th April	Lichfield, Stafford

More information click link: noonansyndrome.org.uk/events

Haemochromatosis UK Social Iron Brew	9th May 2024 (+ occurs monthly!)	Online

More information click link: haemochromatosis.org.uk/events

In conversation with HDBuzz: Information on the latest Huntington's disease research	22nd May 2024	Online

More information click link: hda.org.uk/events

Behçet's UK Friends and Family Day 2024	22nd June 2024	Llandegfedd lake, South Wales

More information please visit: behcetsuk.org/events

To find more information about events please visit geneticalliance.org.uk

Awareness Days

Upcoming dates

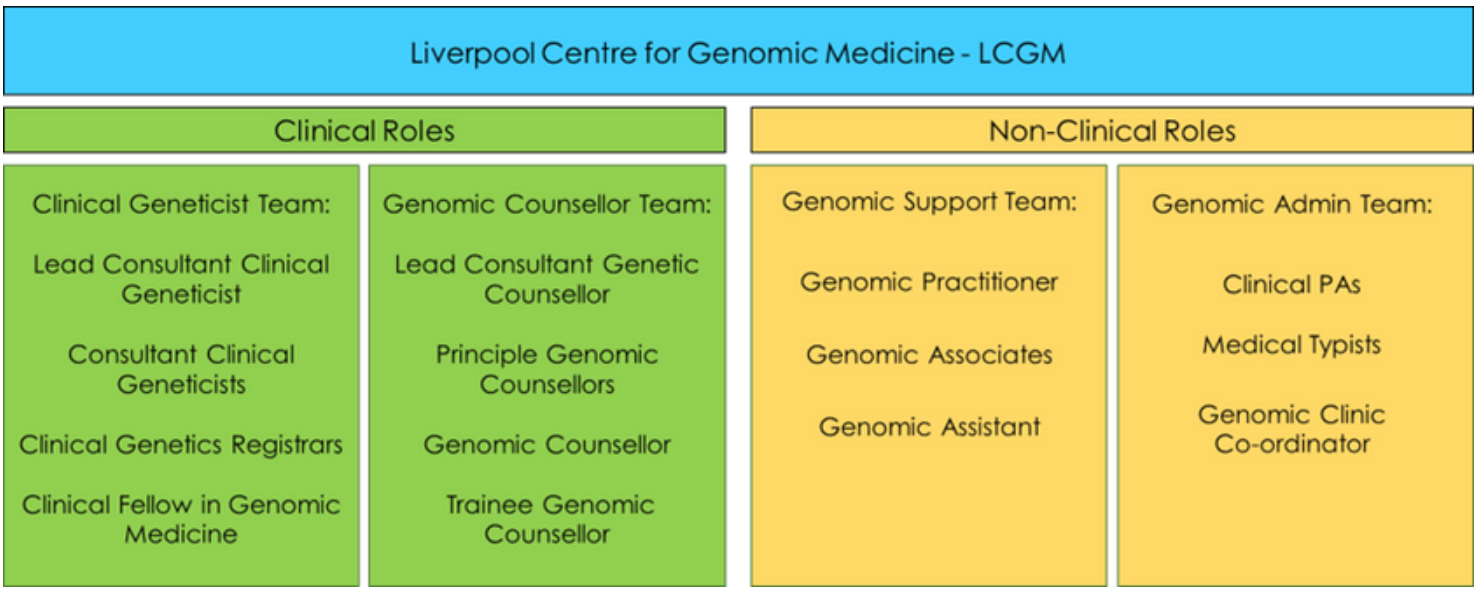
DEAF AWARENESS WEEK

Deaf Awareness Week

6-12 May 2024

CAREERS IN GENOMICS @LCGM

In this edition we summarise the roles within our department. We will delve into more detail about these roles in upcoming editions. The genomic medicine service provided by the **LCGM** involves the collaboration of many different roles across clinical and non-clinical teams. Some patients may encounter one or more members of each team throughout their patient journey.



Clinical Geneticist Team:

Consultant Clinical Geneticists, Clinical Genetics Registrars, Clinical Fellow in Genomic Medicine.

The clinical geneticist team is made up of doctors who have completed further specialist training in genomics. Each step up this career path comes with additional responsibilities and further training. Clinical geneticists have many responsibilities, including assessing and diagnosing patients. Evaluating whether patients are eligible for certain genetic tests and delivering the results. Collaborating with genetic counsellors to develop treatment plans for patients who have genetic conditions. Some complex results require a referral to a genomic counsellor to support patients and their families. Clinical geneticists can specialise further into an area of interest within genomics, such as cardiac conditions or dysmorphology. Responsibilities may include contributing to research and development, such as conducting studies into genetic conditions or hereditary illnesses.

CAREERS

Genetic Counsellor Team:

Lead Consultant Genetic Counsellor, Principle Genomic Counsellors, Genomic Counsellor, Trainee Genomic Counsellor.

Genetic counsellors have many responsibilities, including analysing family history information, assessing the risks of inheriting or passing on a medical condition, ordering and interpreting genetic and genomic test results and explaining these to the individual patient and their relatives. GC's use counselling skills to help patients adjust to having a genetic condition and support and empower them as they incorporate this information into their lives. GC's can specialise further into an area of interest within genomics, such as prenatal or cancer genomics.

Genomic Support Team:

Genomic Practitioner, Genomic Associates, Genomic Assistant.

These supporting roles have been developed as the Genomic Medicine Service has grown. These roles are key to facilitating the collection of information, consent, and test results to enable genomic counselling and testing or screening for patients with rare diseases and their family members. These roles involve communicating with patients and their families by phone, documenting health information and sometimes using software to create family histories for the clinicians. They also support patient events, professional education events and MDT meetings.

Genomics Admin Team:

Clinical PAs, Medical Typists, Genomic Clinic Coordinators.

The admin team are essential to the running of the department and to patient care. This team are responsible for booking appointments and coordinating the genomics clinics across all of the satellite sites. They are also responsible for dealing with enquiries, the reviewing and prioritising patient correspondence, processing of medical correspondence to and from clinicians, arranging meetings and taking minutes, and managing orders for necessary department resources.

Coming soon…

Next edition will cover the role of Genomic Counselling