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NEWSLETTER

Liverpool Center for Genomic Medicine (LCGM)

JULY 2024

In this edition…

Welcome to this issue of the Genomics Newsletter

Welcome to the second edition of the Liverpool Centre for Genomic Medicine (LCGM) newsletter. Our quarterly newsletter provides you with information about our Genomics team, Genomics service developments, interesting patient stories, condition specific information, current research in Genomics and exciting Genomics in the news.

In this edition we aim to tell you about:

- The role of a Genetic/Genomic Counsellor
- The career pathway to become a Genetic/Genomic Counsellor
- A real-life patient story focusing on a patient with Apert's syndrome.
- Information about Apert's syndrome as a condition
- Current research updates
- Genomic charity events
- Exciting new Genomics stories in the news; OTOF gene and pioneering gene therapy restoring a patient's hearing.

Patient Story – Amelia, 17 years

1. To Amelia's mum, when did you notice the differences in Amelia? How did you feel once the diagnosis of Apert syndrome was made?

The differences in Amelia were noticed straight after birth in Liverpool Women's Hospital. The warning signs were the development of her hands and feet which were fused. The paediatrician was called in straight away and he checked Amelia over and informed us that he thought Amelia had Apert Syndrome. Amelia was taken to Special Baby Care Unit and her diagnosis of Apert Syndrome was confirmed later that day by a neonatal consultant. Once the diagnosis was confirmed we were extremely worried and concerned, we had never heard of Apert Syndrome before and neither had the midwife on duty. We were swiftly given a printout of information regarding Apert Syndrome to read which did nothing to allay our fears as it stated that children with Apert Syndrome had to undergo several operations including major craniofacial surgery at 1 year old.

The diagnosis was shocking as no problem had been detected during pregnancy and all scans had been clear. Our thoughts quickly turned to panic as we started to worry what life Amelia would have, if she would walk, talk, go to school and live an independent life. The thought of all the operations that Amelia would need was daunting and we started to wonder how our life was going to change, would we be able to manage, would we be able to eventually go back to work or would there be a financial impact upon us.

2. Amelia, living with Apert syndrome isn't easy. You have done very well and now you are a confident young lady. Can you share some of your experience growing up? Do you have any advice for others with a similar condition like you?

Growing up, I spent lots of time in hospital going to appointments and having operations. I went to mainstream school from the age of 4 to 16 16 and came out with GCSE qualifications and I am now at college doing Business Administration. I am now looking to move into doing a Health and Social Care course.

Patient Story – Amelia, 17 years - *Continued*

I would tell anyone in a similar situation that you can achieve many things with determination. Going through school was tough as nobody else had seen another child with Apert Syndrome, there was lots of staring, comments and questions from other children and it was sometimes difficult to make and keep friendships. Having Apert Syndrome sometimes makes me feel isolated and different because it is such a rare syndrome and not many others have the same condition. Some of the activities I've done is: drama, Brownies, swimming, St John Ambulance Cadets. My advice to others is to not let people tell you that you cannot achieve things, it's best to always try and have a go.

3. 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 and how did it go? What did you feel afterwards?

I had been starting to wonder if I had a family of my own, what the implications would be because I've got Apert Syndrome. I was a bit nervous going into the geneticist appointment but eager to gain as much knowledge as possible for my future, the geneticist then explained everything to me in detail. After the appointment, I was glad that there are some options available to me when the time comes if I decide to have a family of my own.

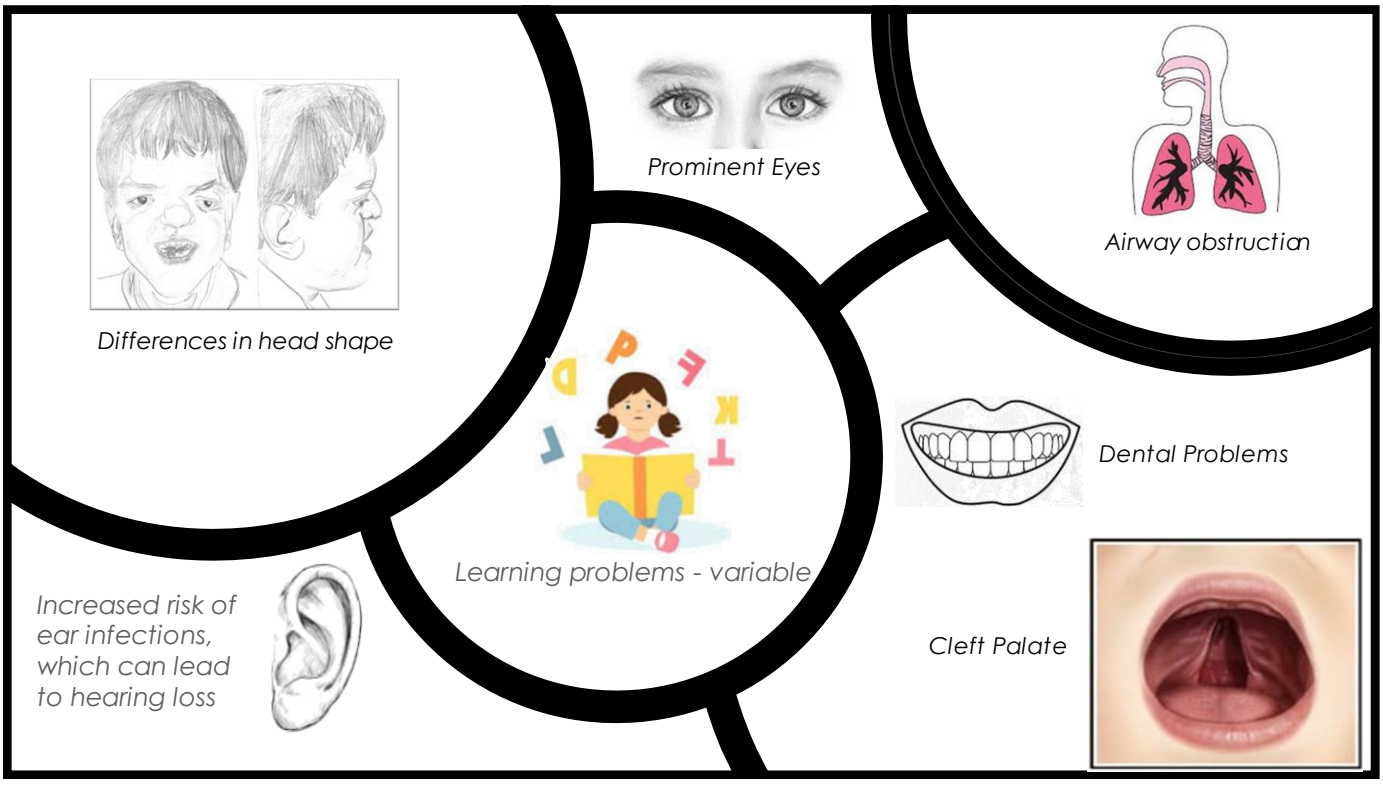
APERT SYNDROME

What is Apert syndrome?

Apert syndrome is a rare genetic condition characterized by premature fusion of the skull bones, also known as craniosynostosis. This prevents the skull from growing and affects the shape of the head and face, causing increased pressure around the brain. In almost all cases, Apert syndrome is associated with other complex differences in development, such as the fusion of the fingers and webbing of the fingers and toes.

What are the most common features?

The most common features include:



APERT SYNDROME - *Continued...*

What causes Apert syndrome?

Specific alterations in the ***FGFR2* gene** cause Apert syndrome. This gene produces a protein that stimulates cells to develop as bone cells while the baby is developing in the womb. The altered *FGFR2* gene causes bone development to occur more rapidly than normal.

In most cases, condition is caused by a **new genetic alteration that isn't inherited from either parent.** In these cases, the chance of the parents having another child with Apert syndrome is very small. In other cases, the altered *FGFR2* gene **is inherited from one of the child's parents.**

Can Apert syndrome be passed on to future children?

Apert syndrome is an autosomal dominant condition which means that an individual with this condition has a 50% chance of passing it on to future children.

What is the treatment?

Apert syndrome is a lifelong condition with no cure. Treatment is given to improve symptoms and prevent complications. Early diagnosis can ensure that appropriate surgical intervention is carried out.

A child with Apert syndrome will need a coordinated programme of care and will see a number of specialists including speech therapists, orthodontists, psychologists and ophthalmologists. In the UK a number of specialist craniofacial centres are funded to provide coordinated care for children with Apert syndrome.

What is the outlook?

The outlook for children born with Apert syndrome is variable depending on the severity of their symptoms and the impact it has on some functions such as breathing, vision and hearing. They require long term monitoring, particularly during period of growth in childhood and adolescence, but surgery tends to be completed by the time the child is in their late teens to early twenties.

Information and support group:

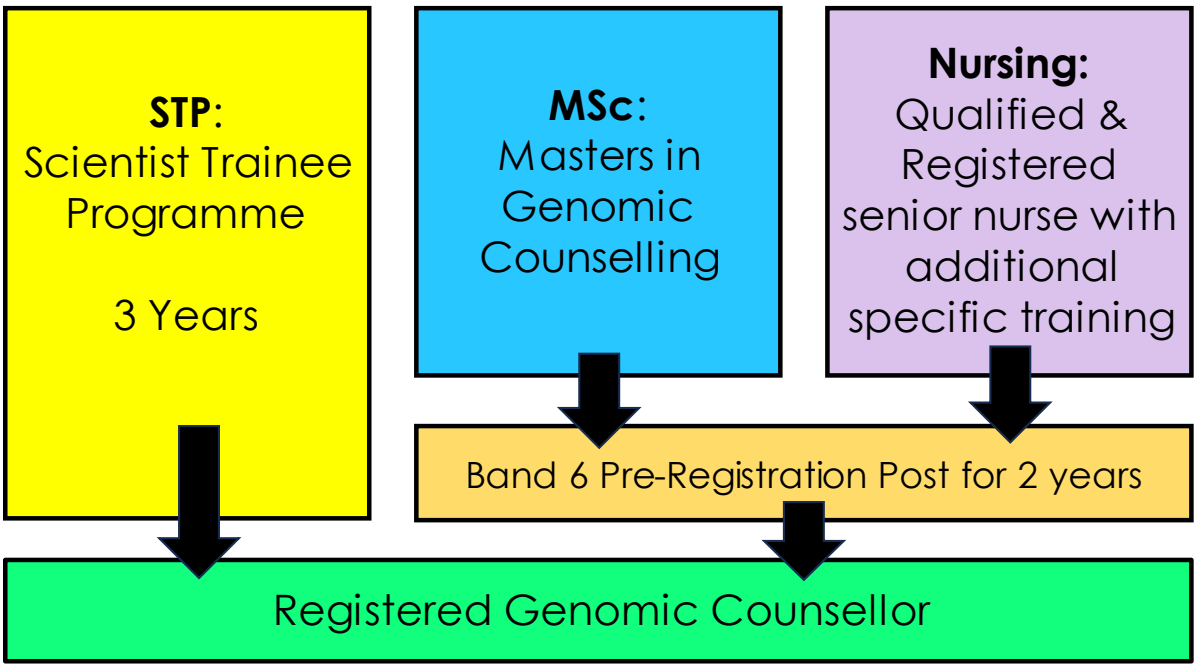
https://www.headlines.org.uk/

CAREERS

Genomic Counsellors

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.

Routes to becoming a genomic counsellor:



Further information about each route can be found via the following link:
Genetic counsellor job profile | Prospects.ac.uk

CAREERS

Genomic Counsellors - *Continued*

A massive thank you to Louise Dubois, Principle Genomic Counsellor at LCGM, for sharing information about her role.

How long have you been a GC?

14 years

Which route did you take to becoming a GC?

Nursing, I am a Registered Paediatric nurse. I completed relevant science and counselling courses prior to obtaining GCRB (now HCPC registration). I have also completed a masters in Genomic Medicine.

Were there any particular ways you gained knowledge, skills, or experience to help you toward your goal of becoming a GC?

In my nursing career I worked with many families who had children diagnosed with genetic conditions.

I spent time listening to family stories and learning about genetic conditions. I volunteered to shadow the genetics service following a patient journey through the service. I also completed a counselling course.

How would you describe your role?

I have clinical line management and service development responsibilities within my role. Within my clinical role I have patient facing capacity to support patients and their families in the face of a genetic diagnosis or genetic risk to understand and adjust to this information and make informed decisions about their health and communicate this with the family. In my line manager role, I support staff with their career development, HR issues, education and continuing professional development and health and wellbeing. Within service development I am the lead for the digital development of the service and have recently contributed the implementation of a new Electronic Patient Records.

CAREERS

Genomic Counsellors - *Continued*

What do you enjoy most about your job?

Diversity in the role each day. The ability to have a hybrid role encompassing clinical face to face patient facing, education, research, service development and line management. The most rewarding part of my role is hearing patient stories and helping patients understand complex information.

What do you find most challenging about the role?

Giving bad news is always a challenging part of my role. However, it's rewarding in knowing we have the knowledge and skills available to support families through these results.

Are there many opportunities for progression?

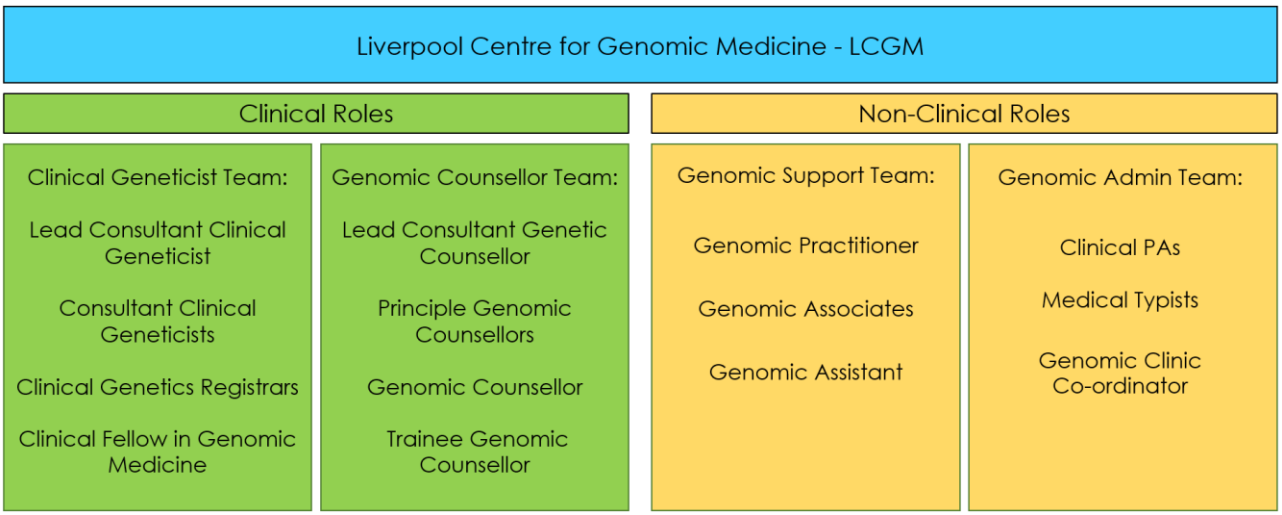
Yes plenty, we have areas of specialism e.g. cancer genetics, cardiology. We work with the Genomic Medicine Service Alliance undertaking projects such as embedding genomic testing into the mainstream and there are plenty of opportunities to be involved in education and research.

Do you have a special area of expertise/interest within your role?

Cardiology genetics

Next edition: Genomic Associate

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.



RESEARCH STUDIES

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.

Early Development in Neurofibromatosis Type 1 (EDEN)

Many children with NF1 have difficulties such as Autism Spectrum Disorder (ASD) and Attention Deficit Hyperactive Disorder (ADHD). EDEN is a research study that aims to work with families to increase understanding of social and cognitive development of babies either with Neurofibromatosis Type 1 (NF1) or have a parent with NF1. By improving our understanding, this study hopes to speed up the discovery of treatments that can be used to boost learning and social development in NF1.

Aim of the study	Who can take part?	What does it involve?
Improve understanding of the early social and cognitive development of infants with Neurofibromatosis Type 1.	Infants under 14 months who have NF1 or have a parent with NF1.	Visits to the research centre (funded), questionaries, interviews and obtaining a cheek swab.

For more information about EDEN research study, please visit
https://nervetumours.org.uk/news/eden-study

RESEARCH STUDIES – *Continued*

Epidemiological Study of Familial Breast Cancer (EMBRACE)

Embrace is a research study that is looking to recruit individuals who have a have gene variant in the family known to increase the risk of developing breast cancer and/or ovarian cancer. The aim of EMBRACE is to improve our understanding of:

- How many people develop cancer
- Other factors that play a part in the development of cancer
- How cancer risk may be reduced
- How we can diagnose cancer early
- How we can improve care for people who carry these gene variants.

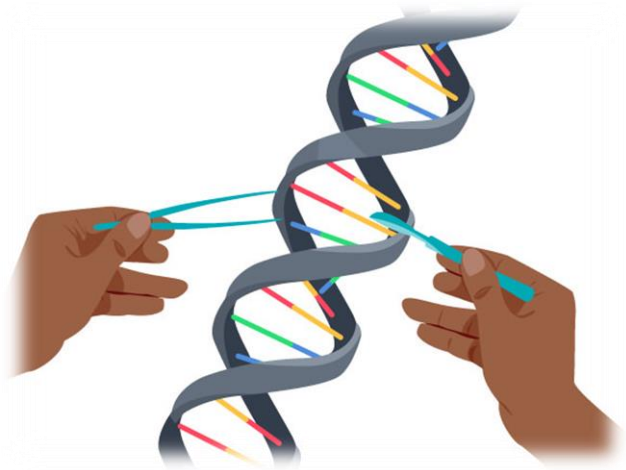
Participation in this research study involves filling in a questionnaire and obtaining blood samples.

Aim of the study	Who can take part?	What does it involve?
To create a register of families who have gene variants that increase risk of developing breast and/or ovarian cancer.	Individuals of any gender aged 18 and above with a gene variant in the family known to cause increased risk of developing breast or ovarian cancer, who have had already had genetic test or plans to in the future.	Filling in a questionnaire and obtaining blood samples

For more information about EMBRACE research study, please visit https://www.cancerresearchuk.org/about-cancer/find-a-clinical-trial/a-study-of-people-who-have-faulty-breast-cancer-genes

LATEST NEWS

Gene Therapy in the spotlight



Pioneering gene therapy restores UK girl's hearing

9 May

A UK girl born deaf can now hear unaided, after a groundbreaking gene-therapy treatment.

Opal Sandy was treated shortly before her first birthday - and six months on, can hear sounds as soft as a whisper and is starting to talk, saying words such as "Mama", "Dada" and "uh-oh".

Given as an infusion into the ear, the therapy replaces faulty DNA causing her type of inherited deafness.

Opal is part of a trial recruiting patients in the UK, US and Spain.

You may wish to look at *https://www.bbc.co.uk/news/health-68921561* for the full article.

This is the first time gene therapy has been used on a deaf child in the UK.

LATEST NEWS

Gene Therapy in the Spotlight

What is gene therapy?

Gene therapy is when DNA is introduced into a patient to treat a genetic condition. The new DNA usually contains a functioning gene to modify the effect of an altered gene that's causing a genetic condition.

Trials in Cambridge and London are testing out gene therapy for children who have a particular type of genetic deafness caused by lack of a protein called Otoferlin (OTOF), leading to severe or profound deafness from birth. Otoferlin encodes sounds within the inner ear and allows nerve signals to be sent from the cochlea to the brain. The gene therapy developed by biotechnology company, Regeneron, inserts 'normal' genes into the cochlear hair cells so that Otoferlin is produced, enabling signalling between the ear and the brain.

The **gene therapy will only be effective for people who have a variation in the *OTOF* gene**. If you are deaf because of a different gene, then you wouldn't be eligible to take part in the trial. Only a small percentage of children are deaf because of a variation in the *OTOF* gene.

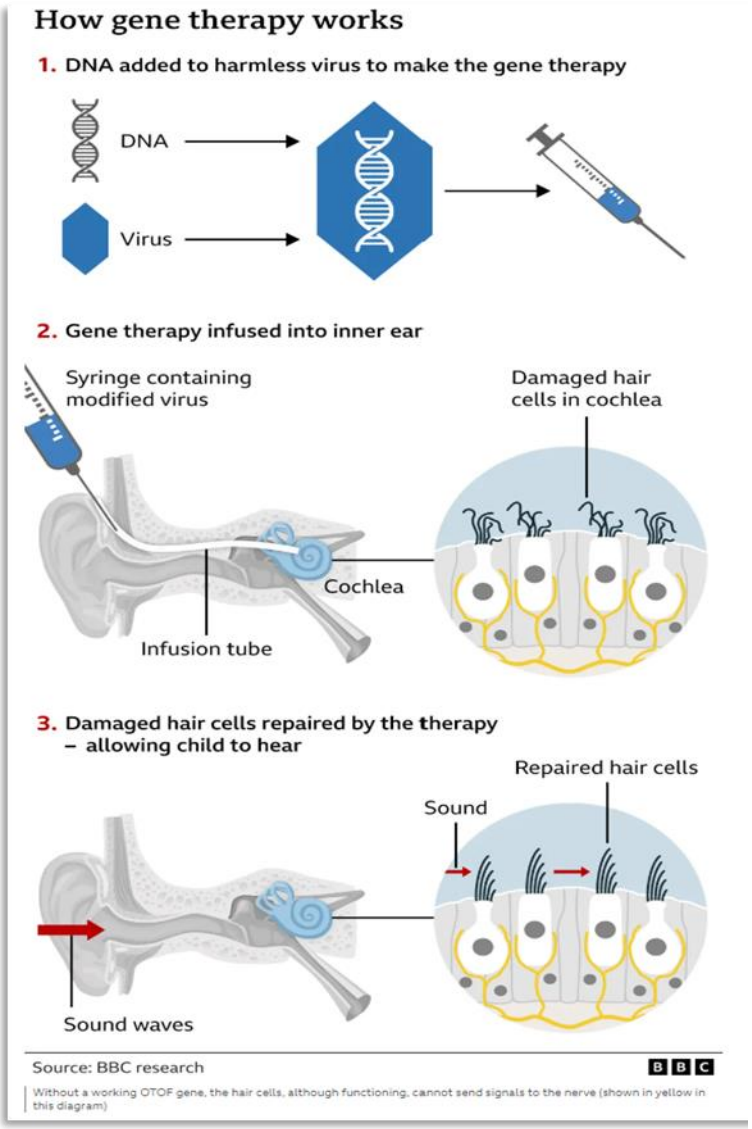


Image from the same BBC article: *https://www.bbc.co.uk/news/health-68921561*

If you know that your child is deaf because of a variation in the *OTOF* gene, then you can get in touch with one of the clinical trial sites for more information.
See https://clinicaltrials.gov/study/NCT05788536?cond=OTOF Gene Mutation&rank=3#contacts-and-locations for contact details.

EVENTS

In this edition, we have summarised some events upcoming in the UK in the table below. These events are either in person or on online.

EVENT	WHEN	WHERE
Weekend for adults with Tourette's syndrome	9-11th August 2024	Staffordshire

For more information, click the link: https://www.tourettes-action.org.uk/

Wolfram Syndrome Support and Information Day	21st September 2024	Daventry

For more information, click the link: https://wolframsyndrome.co.uk/

Rett UK: Family Weekend 2024	18th - 20th October 2024	Coventry

For more information, click the link: https://www.rettuk.org/

Oesophageal Atresia (OA) & Tracheo-Oesophageal Fistula (TOF): Adult and family information seminar	16th November 2024	Warrington

For more information, click the link: https://tofs.org.uk/

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

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SCAN ME

www.liverpoolwomens.nhs.uk/our-services/liverpool-centre-for-genomic-medicine-lcgm/

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