Liverpool Women's
NHS Foundation Trust

NEWSLETTER

Liverpool Centre for Genomic Medicine (LCGM)

JUNE 2025

In this edition…

Welcome to this issue of the Genomics Newsletter

Welcome to the Liverpool Centre for Genomic Medicine (LCGM) newsletter. Each quarter, our newsletter aims to provide you with information about our Genomics team, Genomics service developments, interesting patient stories, condition specific information, current research in Genomics and exciting Genomics information in the news.

In this edition we aim to tell you about:

- The role of a Clinical Research Practitioner within Genomic Medicine.
- A patient story of a child diagnosed with Duchenne muscular Dystrophy (DMD) and the family's experience with Genomic Medicine.
- Information about DMD as a condition.
- Upcoming genomics events.
- Genomics in the news

Patient Story – Arlo's story (from his mum)

Duchenne Muscular Dystrophy DMD

Arlo's 5 yrs old - Arlo has a deletion in the DMD gene, associated with Muscular Dystrophy which results in a lack of the protein essential for muscle repair. This affects his muscle function, leading to reduced energy levels, rapid fatigue, and difficulty keeping up with his peers.

Arlo is a happy and affectionate boy, always ready for a smile or cuddle. Despite the daily challenges he faces, he continues to adapt well. As he grows, we are noticing an increase in the symptoms as expected. for his condition. The journey to a diagnosis was incredibly challenging. Initially, my concerns had been with comments such as "boys are just lazy," and I reassured that there was nothing to worry about.

However, I knew that something wasn't right. I persisted, seeking advice and referrals across various departments until we were finally seen by a paediatrician who took our concerns seriously. From there, genetic testing was conducted, which confirmed Arlo's diagnosis.

Has having a genetic diagnosis been helpful?

Absolutely. Receiving a diagnosis confirmed that my concerns were justified and allowed us to begin accessing appropriate support and resources. It has meant that we can give Arlo the best chance of managing his condition.

Can you share some of your experience with clinical genetics?

Our experience with clinical genetics was very positive. Although we waited three to four months for an appointment, the geneticist was incredibly kind and informative. She took the time to explain everything thoroughly and answered all our questions. I underwent genetic testing, which confirmed that I am a carrier. Subsequently, my younger son has been tested as well, to which he is unaffected. We were fortunate to have the same person throughout our genetic journey, even when she moved from the clinic to the hospital.

Do you have any other thoughts you would like to share?

My advice to other parents is to trust your instincts and do not give up as you know your child best. Genetics are amazing. There's so much you can find out through a genetic test.

Condition

What is Duchenne Muscular Dystrophy (DMD)?

Duchenne muscular dystrophy (DMD) is a condition that causes **progressive muscle weakness** and wasting. It stops the body from making dystrophin, a protein needed for muscle strength and function. DMD mainly affects boys and typically starts in early childhood and eventually leads to a loss of mobility. Over time, the condition also affects the heart and breathing muscles, which can cause life-threatening complications.

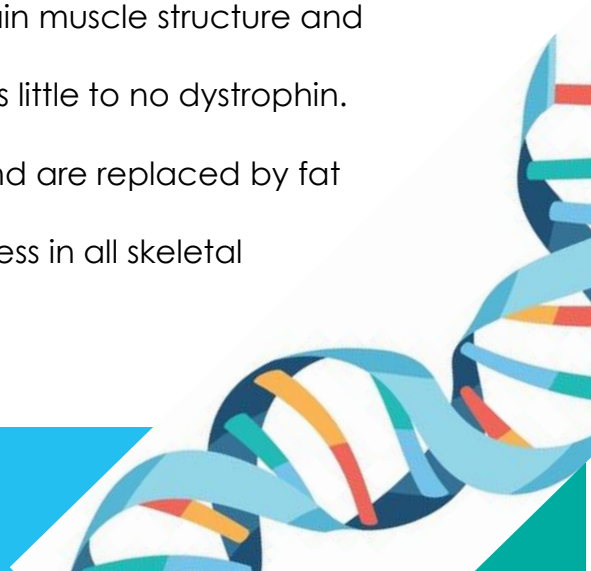
While there is no cure for DMD, research is ongoing, and new treatments are being developed. DMD shortens life expectancy but with improved medical care, more people are living into their 40s and 50s, and their quality of life is improving.

What are the symptoms?

Muscle weakness is the most noticeable and early symptom of DMD, which worsens over time. As the condition progresses, it affects the heart, respiratory system, and other organs, including the gastrointestinal system, and leads to a wide range of challenges. Some people may also have learning difficulties, behavioural problems, and speech delays.

What causes Duchenne Muscular Dystrophy?

DMD is caused by a change in the **dystrophin gene**, which provides instructions to make the dystrophin protein. This protein helps maintain muscle structure and function. In DMD, the change means the body produces little to no dystrophin. As a result, muscle cells become damaged over time and are replaced by fat and fibrous tissue. In time, this causes progressive weakness in all skeletal muscles.



Duchenne Muscular Dystrophy - *Continued...*

There are different types of DMD, known as variants, depending on the specific genetic change. Some variants may have different treatment options, but all types of DMD can be slowed with steroids, which are commonly used.

DMD is not like Becker Muscular Dystrophy (BMD), which is also caused by a change in the dystrophin gene but allows for some functional dystrophin to be produced, which results in milder symptoms and slower progression.

- Inheritance

DMD is inherited in an **X-linked recessive pattern**, meaning the genetic change is on the X chromosome. Since the male sex has only one X chromosome and the female sex has two, males are typically more significantly affected by DMD, while females may be carriers. The condition can be inherited from a carrier mother, but it can also be caused by a new change in a child's genes. **Mothers of children with DMD should be offered genetic counselling and a test to find out whether they are carrier for the DMD mutation. Many different reproductive options are available for couples at risk of having a child affected by DMD.**

- Manifesting carriers

Affected males cannot pass the changed gene to male children, but all their female children will be carriers. Most female carriers don't experience symptoms because they have a second X chromosome, from which the dystrophin protein can be produced. However, a small number of female carriers can have muscle weakness and some associated symptoms of DMD. This means they are a manifesting carrier of DMD.

How is it diagnosed?

DMD is **often diagnosed around the age of three**, when symptoms such as **muscle weakness** or difficulty with physical activities are noticed. The initial test is usually a blood test to check for raised creatine kinase (CK) levels – an enzyme released when muscles are damaged. The liver enzymes (ALT and AST) may also be raised, and this indicates muscle damage and breakdown, rather than liver problems. If CK levels are high, genetic testing is done to confirm the diagnosis by identifying a change in the dystrophin gene. In rare cases, a muscle biopsy may be done if genetic testing is inconclusive, but this is becoming less common.

Genetic counselling and testing for other family members should be arranged soon after diagnosis and can be organised by a clinician or GP.

How is it treated?

Treatment to help reduce the symptoms, improves quality of life and prolongs survival. DMD needs a **multi-disciplinary approach** for effective management, which involves a team of specialists working together to address the wide range of symptoms caused by the condition.

To prevent the progression of DMD treatment with **glucocorticosteroids** (a type of steroid), is routine. These can help slow the progression of muscle weakness and maintain function for longer.

New treatments, such as Translarna (ataluren), are available for specific genetic changes known as 'nonsense variants' and may slow disease progression. Access to Translarna varies in different parts of the country and it can be harder to access in some parts than others. Ongoing research is focused on developing more treatments that target the underlying cause of DMD.

A regular programme of **physiotherapy** is essential to reduce contractures and promote mobility.

CAREERS

Job title: Clinical Research Practitioner (CRP)

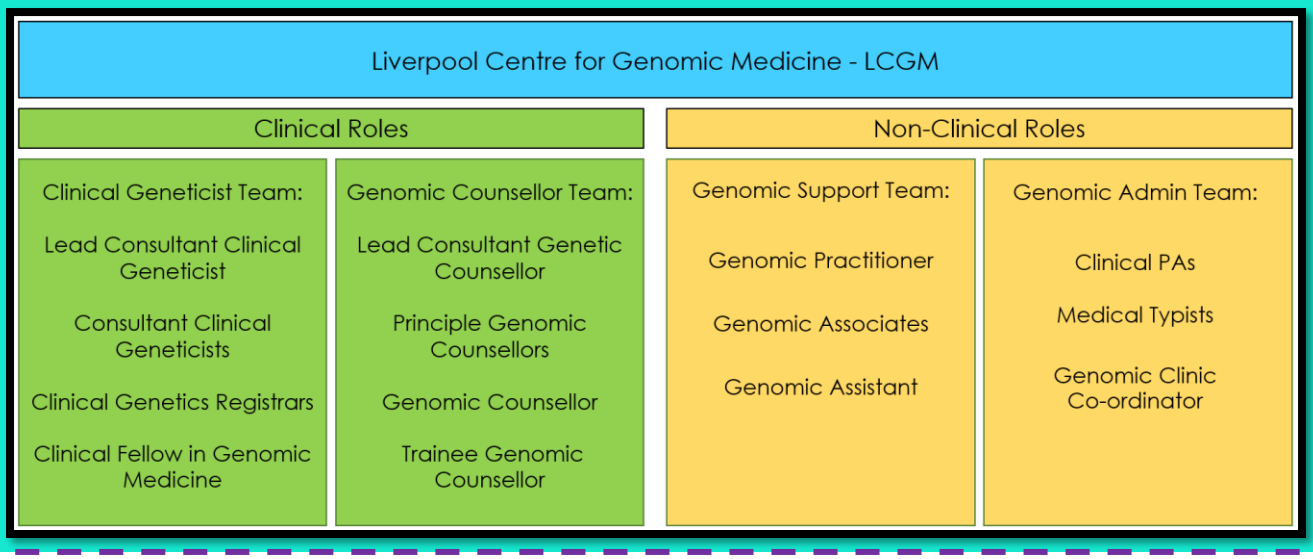
How would you describe your role?

CRPs play an important role within the delivery of research. Like research nurses and midwives, we work alongside principal investigators (PIs) to deliver safe, ethical and high-quality clinical research care. We currently have 4 CRPs working at LWH, spread across gynaecology, maternity and genetics.

How long have you been in your role?

I have been in my current role as a Clinical Research Practitioner (CRP) at Liverpool Women's Hospital for the last 4 ½ years, but I have been working within research for 9 years now.

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.



CAREERS

Clinical Research Practitioner - *Continued*

A massive thank you to Charlotte Stanley at LCGM, for sharing information about their role.

What route did you take to becoming a Clinical Research Practitioner?

I originally gained my BSc and MSc in Biomedical Science with the intention of becoming a histopathologist. However, I quickly came to realise I did not want to be in the labs full time and wanted to be more involved in patient care.

I got a job as a research assistant in IBS studies in Wythenshawe hospital before coming to work as a genetic CRP here at LWH. In 2022, I became a registered CRP.

CAREERS

Clinical Research Practitioner - *Continued*

What do you enjoy most about your job?

My favourite aspect would have to be the variety within my job role. Working across a wide range of research studies means that no two days are ever the same. I can be screening clinics, approaching and recruiting participants to research studies, taking and processing research samples, and dealing with data queries all in one day.

What do you find most challenging about your job?

The most challenging part of my job is time. I am the only researcher within our trust who works on recruiting for the genetic portfolio, and we currently have 28 active studies recruiting. I also assist on gynaecology and maternity research studies, so I am normally quite busy!

What areas of research are you involved in?

The research portfolio that I lead on focuses on both rare genetic diseases and cancer genetic studies. This broad research portfolio gives many patients the opportunity to take part in research.

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.

The identification and characterisation of inherited predispositions to colorectal tumours study - 2 (CORGI2)

We know from research that there are genetic factors present in our DNA that can affect our risk of developing tumours in our bowel. This includes precursor lesions, known as bowel polyps, as well as bowel cancer.
In this study, the researchers will look at the genes of a large number of people with a history of bowel cancer, bowel polyps or a related cancer, to identify common genetic factors that may increase or reduce the risk of developing bowel cancer.

Aim of the study	Who can take part?	What does it involve?
To identify new susceptibility genes for colorectal cancer (CRC), bowel polyps and other cancers genetically related to CRC, such as endometrial cancer.	Any individual over 6 years with a colorectal tumour or with a cancer or disease strongly aetiologically related to colorectal tumours (e.g. endometrial cancer) **OR** Individuals of any age with ≥10 polyps (where previous genetic testing has not found a cause for the polyps)	Participants will complete a questionnaire about their medical history. Genetic testing will be performed using a sample of the participants DNA.

For more information about this research study, please visit https://www.hra.nhs.uk/planning-and-improving-research/application-summaries/research-summaries/corgi-2/

LATEST NEWS

Blood test developed that could speed up the diagnosis of rare disease in babies

Scientists say that new approach means effects of many genetic mutations can be analysed at once and yield results in days

Guardian article

The Guardian published an article that outlined how a new blood-based tests could speed up diagnosis for children born with a genetic disorder. You can read the full article at:

https://www.theguardian.com/science/2025/may/23/blood-test-could-speed-diagnosis-rare-diseases-babies

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
International Conference for Ectodermal Dysplasia	10th June – 12th June	Austin Court, Birmingham, UK

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

ACGS Summer Meeting 2025	12th – 13th June	ICC, Birmingham, UK

For more information, click the link: https://www.acgs.uk.com/events/acgs-summer-meeting-2025/

Curating the Clinical Genome. 9th *Curating the Clinical*	11th –13th June 2025	Welcome Genome Campus UK and Virtual

For more information, click the link: https://coursesandconferences.wellcomeconnectingscience.org/event/curating-the-clinical-genome-20250611/

Genetics Society Anniversary Day 2025	25th June 2025	Biffen Lecture Theatre, Department of Genetics, University of Cambridge

For more information, click the link: https://genetics.org.uk/events/genetics-society-anniversary-day-2025/

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SCAN ME

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

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