Duchenne UK

Research to transform lives:

our holistic approach to end Duchenne

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Foreword

It's not an overstatement to say that Duchenne UK has transformed the landscape of care and research for patients in the UK living with a diagnosis of Duchenne muscular dystrophy (DMD).

Alex and I met as two mothers fighting to save our sons. What we've built is a movement to end Duchenne.

Since 2012 we have invested £15 million in DMD research. We have funded clinical trials of drugs that are now approved by the Medicines and Healthcare products Regulatory Agency (MHRA) and the National Institute for Health and Care Excellence (NICE) and are being made available to patients.

Our latest impact report demonstrates the transformative impact of our work.

Everything we do is to get to our goal to end Duchenne.

Funding medical research into treatments that slow down and ultimately will stop the progression of the disease or will be curative, continues to be at heart of what we do, as is funding and supporting the vital infrastructure to deliver the clinical research that brings these innovative treatments to people with DMD.

We exist to accelerate research innovation that translates into transformative treatments and medical care reaching the clinic and people with DMD as fast as we can, and we won't stop until we will have made the lives of all people with DMD free of the devastating impact of this condition.

We will continue to play a central role in the landscape as a research funder and enabler, and patient advocate to ensure progress at pace, because people with DMD don't have any time to waste.

Underpinning our research funding are our flagship programmes: the DMD Hub, DMD Care UK, Hercules, and Duchenne UK Connect, a new exciting initiative we have launched in 2024 to deliver a new Duchenne patient data platform.

We work tirelessly with the research community to achieve our goal to end Duchenne. We could not do this without you. Together we will end Duchenne.

Emily Reuben OBE, Chief Executive and Founder of Duchenne UK
Alex Johnson OBE, Founder of Duchenne UK and Chief Executive of Joining Jack

Executive Summary

Innovation in medical research is core to our strategy.

Our priorities are informed by people living with DMD and their parents and carers, ensuring that we are sharply focused on the research innovations that will create the biggest impact for them.

Our focus today is informed and built on our successes to date, as a funder, innovator and enabler of positive change for our community. Our intention is to continue to fund, drive and enable the most effective research to develop new treatments and the best clinical care interventions, and ensure that people with DMD can access them as quickly as possible.

Underpinned by our flagship programmes (DMD Hub, DMD Care UK, HERCULES and Duchenne UK Connect), our research portfolio includes projects focussed on:

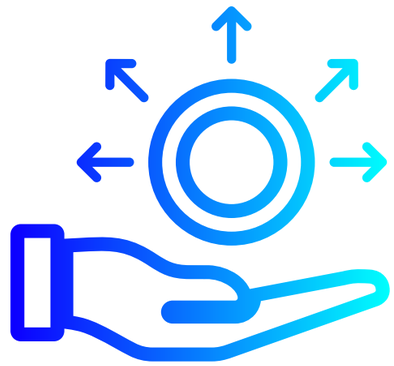
THERAPEUTICS
translational and clinical stage drug development programmes

CARE
clinical research studies to address evidence gaps to drive improvement in DMD clinical care and management

CLINICAL RESEARCH INFRASTRUCTURE
funding towards vital national resources to enable clinical trials in the UK

We collaborate with the DMD community

We collaborate with drug developers in industry and academia, with the clinical community, and with other patient organisations and key stakeholders nationally and internationally, to ensure that as a funder and a partner we bring the most value and make the biggest impact for people with DMD.



Smart funding

We are smart in how we fund research and drive its commercialisation to ensure the fairest return of investment to us, so that we can invest more funding into research to transform lives.

Our Vision

To end Duchenne and its devastating impact

Our Mission

To accelerate the development of transformative treatments and clinical care for DMD and ensure that people with DMD have access to them as quickly as possible

Our strategic priorities in Research

THERAPEUTICS

Accelerate the development of new treatments through funding of translational and early-stage clinical programmes in industry and academia

CARE

Drive the highest standards in DMD clinical care through research generating evidence to improve clinical practice

RESEARCH INFRASTRUCTURE

Support the clinical research infrastructure to deliver clinical trials in the UK and accelerate access to approved treatments through our flagship programmes

DMD HUB Expanding Trial Capacity

HERCULES

DMD CARE UK Best care for all

How we do it

Put the voice of people with DMD at the heart of everything we do

Invest in a focussed and targeted way, maximising impact and return on investment

Partner and collaborate with the DMD research community nationally and globally

Leverage our flagship programmes DMD Hub, DMD Care UK, Hercules and Duchenne UK Connect

Be a thought-leader and advocate to influence the research priorities

Our Impact in Research So Far

From research to patients, how we helped vamorolone become a reality for the DMD community

Phase 1- Funding

Funding by Duchenne UK, Joining Jack and Duchenne Research Fund bridged the valley of death in clinical research by funding Phase 1 of research into vamorolone

Phase 2 - Clinical Development

We worked with the company to select the outcome measures for the Phase 2 and Phase 3 trials, which gave clinical meaningfulness to the data

Phase 3 - Clinical Development

We supported Professor Michela Guglieri, from the John Walton Muscular Dystrophy Research Centre at Newcastle University, who was the study chair of Phase 2b, and the UK hospital sites which delivered it

Phase 4 - Collaboration

We helped Newcastle University and ReveraGen, the company developing vamorolone, win a 6 million euro investment from the European Union to undertake Phase 2 research into it by supporting their application

Phase 5 - Value Assessment and Access

Our Project HERCULES provided crucial evidence about the impact of Duchenne muscular dystrophy to build the case to National Institute for Health and Care Excellence (NICE) for patient access to it

Phase 6 - NHS Adoption

Through DMD Care UK, we are supporting vamorolone's implementation in the NHS, to ensure that every patient who wants it can access it in a timely and safe way. We are publishing a new DMD Care UK family guide on steroids and there will be a presentation about it at our New Horizons conference in March 2025

VENTURE PHILANTHROPY

We made money from our investment and all of the money we receive goes into research for new DMD treatments

We have supported DMD treatment vamorolone at every stage of its development

Our Investment in Research to date

How much funding?

£15M of which **£3.3M** towards research infrastructure and the rest largely towards therapeutic development

How many projects?

106

How many posts in the DMD Hub?

34

The impact of our funding

How many new therapeutic approaches have reached clinic thanks to Duchenne UK?

7

How many treatments have been approved thanks to us?

2

How many people with DMD have had access to clinical research?

574

How many trials have taken place in the UK?

60

How we use our programmes to support our research

Duchenne UK and the John Walton Muscular Dystrophy Research Centre at Newcastle University work together to improve the lives of people with DMD through driving clinical research and care excellence nationally.

Established in 2016 in partnership between Duchenne UK and the John Walton Muscular Dystrophy Research Centre at Newcastle University, the DMD Hub is the UK clinical research network for Duchenne muscular dystrophy. The DMD Hub exists to enable and accelerate delivery of clinical trials in the UK for the benefit of patients, clinical sites and industry.

DMD Care UK is a collaborative initiative between the John Walton Muscular Dystrophy Research Centre at Newcastle University and Duchenne UK, embedded in the UK North Star Network. It is funded by Duchenne UK, Duchenne Research Fund and Joining Jack.

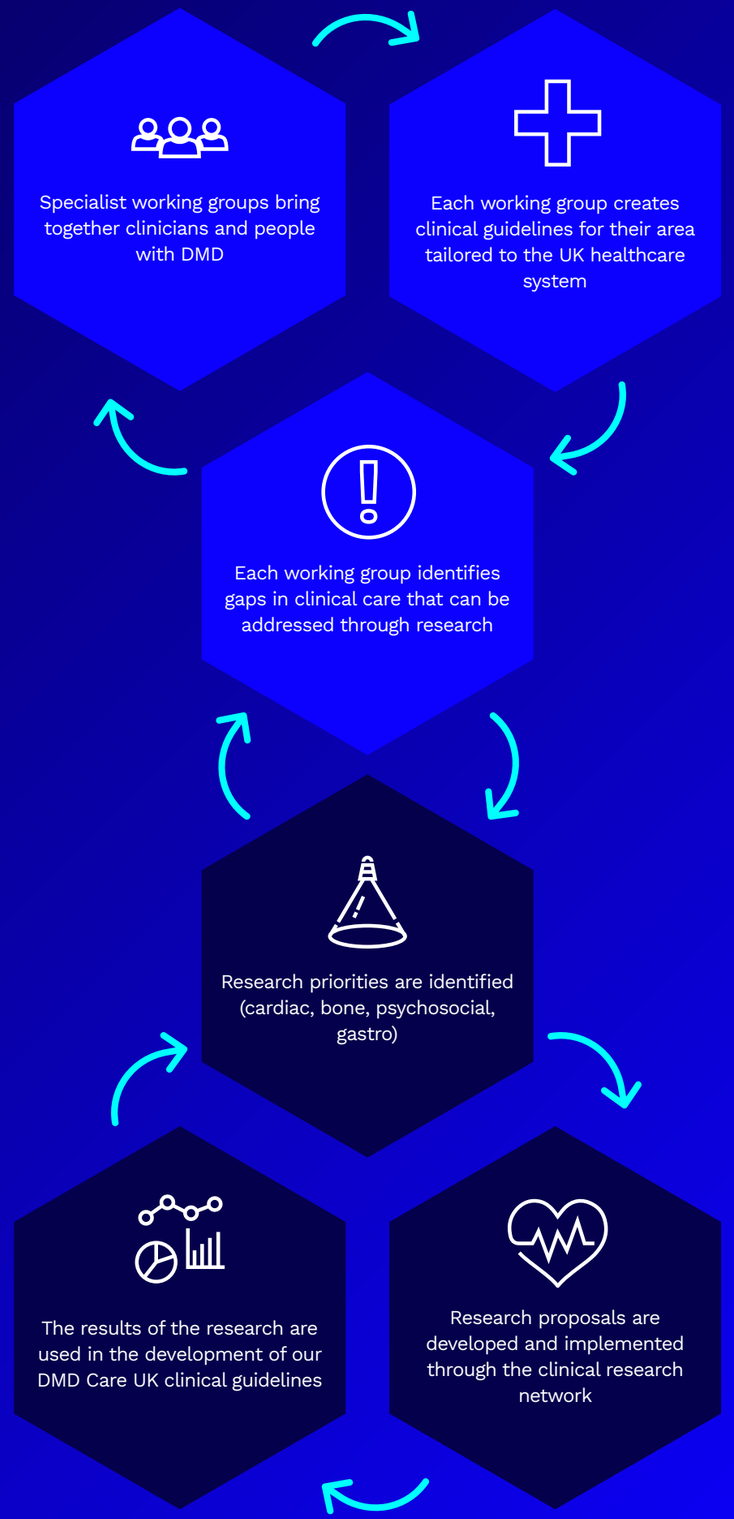
DMD HUB

DMD CARE UK
Best care for all

The John Walton Muscular Dystrophy Research Centre

Duchenne UK

DMD CARE UK
Best care for all



DMD HUB
Expanding Trial Capacity

Where we are today

THERAPEUTICS

We are at an exciting time in DMD therapeutic development, and we have never before been more hopeful.

Two new treatments have been approved in the UK in 2024/2025 - Vamorolone and givinostat. They are not curative, but they are effective at slowing down disease progression.

The first gene therapy for DMD has been approved in the US. While this is an encouraging step forward, we are acutely aware that more effective and safer gene therapies will be needed to make this approach transformative for people with DMD.

CARE

Through our DMD Care UK programme, driving the highest standards of clinical care in DMD nationally, we have identified several areas of DMD clinical care that we need to improve, to drive better clinical outcomes and quality of life.

We have been using this platform, which brings together people with DMD and clinicians, to drive consensus on what research needs to be done to support the generation of evidence to achieve this.

We are excited to play a critical role as a funder to enable this research, and then drive its translation in better care pathways and treatment strategies in the clinic.

RESEARCH INFRASTRUCTURE

Through the DMD Hub we have transformed the clinical research landscape for DMD in the UK. There have never been so many clinical trials and people with DMD participating in research before. But delivering clinical research in the NHS can be challenging due to capacity and resource constraints, and even more so, to deliver newly approved treatments ahead of NICE review.

OUR ROLE IN THE LANDSCAPE

Therefore, our role in today's research landscape, nationally and globally, will be to continue to address these challenges as a funder, enabler, and advocate, while staying sharply focused on what matters to our community - accessing new, better treatments, as quickly as possible and accessing the best clinical care.

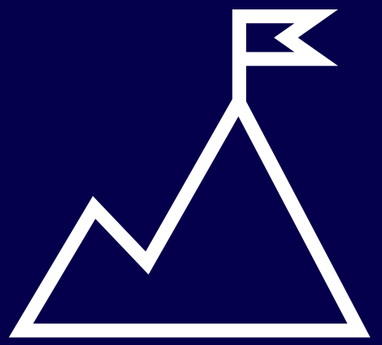
Our impact in research comes to life through our policy work

On World Duchenne Awareness Day we launched our first ever policy report, *Transforming our rare reality*. The report addresses some of the biggest challenges in DMD in the UK and offers practical and productive solutions – building on the work the charity has already set in place. Recommendations include asking NICE to include the DMD Care UK clinical guidelines as part of its treatment guidelines for DMD, and for the UK Government to look at how trial capacity can be increased at existing Centres of Excellence.

Our goals

Our ultimate goal is to end Duchenne, and this means a life free of the impact of the condition for the people living with it and their families.

Finding curative treatments is no longer a dream - research innovation continues to advance at pace to make these a reality, and we are determined to play our part.



Short term (2-5 years)

THERAPEUTICS

Significant progress has been made in recent years leading to a healthy pipeline of new therapeutic approaches for DMD in preclinical and clinical development. These include second generation dystrophin restoration approaches, including both improved gene therapies and exon skipping, gene editing, and cell therapies, and also novel anti-inflammatory and antifibrotic strategies. We will continue to support and encourage this progress, to accelerate the development of effective treatments for all people living with DMD.

CARE

We will also support the development of treatments that address the cardiac and psychological involvement in DMD, which are both areas of unmet need and have a huge impact on mortality and quality of life respectively. We will accelerate the validation and translation in clinic of drug repurposing approaches, working closely with our DMD Care UK programme.

CLINICAL RESEARCH INFRASTRUCTURE

We will also continue to invest in the clinical research infrastructure through our DMD Hub to ensure the right resources are in place to deliver trials and newly approved treatments to patients.

Long term (5-10 years)

Over the coming years, we will continue to partner with clinicians, industry and others in the community, to drive forward innovative research for a cure - this will include driving genetic approaches that can treat the muscles, the heart and the brain, and that can be given as early as possible from birth. We will continue to bring the voice of people with DMD into the research agenda, setting priorities that reflect their priorities.

Together we will end DMD.

Our priorities

THERAPEUTICS
Accelerate the development of new treatments through funding of translational and early-stage clinical programmes in industry and academia.

We will continue to use our response-mode grant funding approach to fund therapeutic programmes advancing novel and disruptive drugs to the clinic, with a focus on:

- next generation gene therapies- delivering full length dystrophin
- other approaches that can be used in combination with gene therapies and test novel mechanisms of actions
- cardiac treatments and in particular clinical trials of repurposed drugs
- psychopharmacological interventions addressing brain involvement in DMD

We will support both small biotech and academic projects where our funding is helping them to translate to the clinic within two years. We are also interested in clinical stage programmes ready for phase 1b/2 trials in DMD.

We will maximise impact and return on investment.

CARE
Drive the highest standards in DMD clinical care through research generating evidence to improve clinical practice.

We will continue to work with DMD Care UK to identify gaps in evidence to drive improvements in standards of care and translate these into research priorities. We will look at all clinical domains of DMD, with a focus on bone health, gastrointestinal health, and pain management.

RESEARCH INFRASTRUCTURE
Support the clinical research infrastructure to deliver clinical trials in the UK and accelerate access to approved treatments.

We will continue to invest in the DMD Hub, the UK national clinical research network for DMD, to ensure that people with DMD can participate in clinical trials.

We will also work with the North Star Network, the NHS and other stakeholders to accelerate translation of approved treatments in the NHS, leveraging the DMD Hub knowledge, expertise and network to drive innovative and cost effective translational pathways.

Our approach

- **Focus on what matters to people with DMD and their families**
- **Collaborate with patients, clinicians and industry and bring them together to co-produce**
- **Drive translation - bridge the gap from the lab to the clinic, and from research and development to access so that translation can happen faster**
- **Partner with others to amplify our impact**
- **Maximise return on investment so that we can do more**

How You Can Help

If you are a researcher and want to work with us or you have an interesting idea that you would like to pitch to us, you can reach us at **research@duchenneuk.org.** **We welcome applications all year round from any interested parties.**

If you believe in our mission and want to join our fight to end DMD, you can help us raise vital funding to support our portfolio through the following fundraising initiatives

- **Dash**
- **Duchenneber**
- **Family and Friends Funds**

You can also donate directly to us here: **duchenneuk.givingpage.org/donate**

Duchenne UK

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