Duchenne UK

Transforming our rare reality

Life with Duchenne muscular dystrophy in the UK

State of the Duchenne Nation | September 2024

Acknowledgements and about Duchenne UK

We thank the following people for their contributions to this report:

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About Duchenne UK

Duchenne UK is a charity that was set up in 2012 by Emily Reuben OBE and Alex Johnson OBE following both of their sons being diagnosed with Duchenne muscular dystrophy (DMD). They set up Duchenne UK to tackle some of the big challenges in drug development in their search for a cure for DMD.

In 13 years, Duchenne UK has raised more than £27 million and used this money to:

- Fund pioneering medical research into the development of transformative treatments for DMD, including clinical trials of medicines that are now showing promise for DMD.
- Create the DMD Hub, a DMD clinical research network of hospital sites across the country which, since its launch in 2016, has led to more clinical trials of DMD treatments coming to the UK than ever before, giving hundreds of children living with DMD access to new treatments.
- Set up DMD Care UK, a national programme for patients with DMD establishing best practice across all the disciplines involved in DMD clinical care, to ensure healthcare professionals and parents know exactly what treatments and medical care children and adults with DMD need and can access in the NHS.
- Develop innovative assistive mobility technologies, such as Elevex and the Dream Chair, to support the independence of people with DMD.
- Form Project HERCULES, a multinational collaboration set up by Duchenne UK to develop tools and evidence to support health technology assessments and reimbursement decisions for new treatments for DMD.

Contents

Chapter 1: Foreword by our founders Emily Reuben OBE and Alex Johnson OBE

Friends: it is with an immense amount of pride and gratitude that we are publishing this, our first Policy Report.

It will give you a complete picture of what it is like to live with DMD in the United Kingdom in 2024. It reports on the gaps in treatment, care and support, and sets out our vision and our roadmap for change. In it, we take stock of the progress that has been made in the 13 years since we set up Duchenne UK.

When our sons, Eli and Jack, were first diagnosed, we were bereft. Instead of being scooped up by a protective ring of health and social care, we realised, from the beginning, that we were going to have to fight for everything. A fight to access social care support, a fight to get access to the best standards of care, a fight to get the right equipment.

We were facing unimaginable odds and battling a relentless and horrible disease. But there was almost nothing to help us. Very limited treatments, little support and very little care – and no hope.

That was 13 years ago, and a lot has changed since then for people living with DMD in the UK. A child diagnosed with DMD has a far better prognosis because of improved treatments, better care, and more support.

Healthcare professionals covering many disciplines have played a key role in this progress. So too has an incredible community of people personally affected by DMD.

There are the amazing DMD Family and Friends Funds that raise hundreds of thousands of pounds to support our work.

There are the brave families and sons who sign up for research – who fill in questionnaires and who take part in clinical trials – many of which do not reach their primary endpoints of success.

Our partner charities: Joining Jack, which Alex and Andy Johnson set up after their son Jack's diagnosis with DMD. Many other brave families have done the same and have now joined up with us – because together we are stronger. Alex's Wish, which was set up by Emma Hallam, whose son Alex has DMD. The Duchenne Research Fund that was set up by Kerry and Doron Rosenfeld after their son Gavriel was diagnosed with DMD; Chasing Connor's Cure that Matthew and Emma Crawford set up following their son Connor being diagnosed with DMD. Duchenne Now, which Tony Levene set up when his son Joey was diagnosed with DMD.

And it's a community of driven parents fighting to change the world. And we are lucky to partner with those people.

Dr Janet Hoskin, whose son Saul has DMD, has written a book to guide people on DMD, and runs DMD education support programme Decipha with her husband Nick Catlin.

Fleur Chandler, whose son Dominic has DMD, set up Project HERCULES with us to build the case for approval of new DMD treatments.

Annie and Ilan Ganot, whose son Eytani has DMD, set up Solid Biosciences, a life science company focused on advancing genetic medicines for neuromuscular and cardiac diseases, and who helped kickstart the development of Elevex, which is our new technology to assist arm movements.

The wonderful Manoj and Sejal Thakrar, whose son Shiv had DMD. They set up the Family and Friends Fund 'Smile with Shiv', and Sejal, whose background is data analysis, undertook some of the research that went into this report.

Thank you for reading it, and we hope to work with you on our framework for change.

Emily Reuben OBE, Chief Executive, Duchenne UK, and founder of Duchenne UK

Alex Johnson OBE, Chief Executive, Joining Jack and founder of Duchenne UK

Chapter 2: An introduction to DMD and our report

DMD: the rare disease that causes so much pain

You may not have heard about DMD before you started to read this report. Or it may be a central part of your and your family's life, and you wish you had never heard of it.

That's the sad truth of a rare disease like DMD. It's devastating to those personally affected, but this devastation is poorly understood by most people.

The seldom seen pain of living with, and loving someone with a rare disease.

DMD's rarity means that there isn't the same level of awareness, understanding of the disease, care, treatment and support available that there is for many more common diseases.

A parent tells someone that their child has cancer. That person, whether they are a neighbour, the child's teacher, or even a stranger they meet in a park, will immediately understand the gravity of what the child and their family are facing.

A parent tells someone their child has DMD, and they are usually met with a blank expression. It is not that people don't care. They just don't understand.

More seriously, a parent tells a doctor or nurse in an accident and emergency department that their child has DMD, and they are too often met with a poor understanding of the disease and what it means for their treatment and care.

These are common scenarios in the first few years after diagnosis, when the parents, carers and family of a child with DMD are reeling from the diagnosis and are trying to adjust and rebuild their lives in their new normal.

When children with DMD have only just started to be affected by its progression, it might not be obvious to others that they are living with a very serious and life-limiting condition.

But their parents, carers and everyone who loves them know. They know what DMD is and what lies ahead.

And this is what they know.

DMD symptoms and progression

DMD causes muscle weakness and wasting. It eventually affects all the muscles in the body, including the heart and lungs. It is a genetic disease caused by a mutation in the dystrophin gene and it is typically diagnosed in childhood between the age of three and six. It is diagnosed with a blood and genetic test, and sometimes a muscle biopsy.

DMD is a life-limiting disease. Currently, people with DMD live into their twenties and thirties on average, with some living into their forties and fifties. There are five stages of DMD[i]:

Early ambulatory (walking) state

In the early ambulatory state, people with DMD can stand from lying down and can run/walk ten metres or more.

Additionally, people with DMD can bear weight while standing.

Late ambulatory state

In the late ambulatory state, people with DMD cannot stand from lying down but can still run/walk ten metres or more. They can still bear weight while standing.

Transfer state

People with DMD then progress into the newly identified state, the transfer state. This is a state that we recently identified through our Project HERCULES research (you can read about this work on pages 24 and 25).

In this state, people with DMD can no longer run/walk ten metres. They can still bear weight while standing, and remain standing for three seconds or more, to support transfers to a bed, sofa or toilet.

Early non-ambulatory state

At the beginning of this state, people with DMD can no longer stand for three seconds or more, but still have hand-to-mouth function. They are not on a ventilator. Following this, people with DMD move to one of two health states which present themselves in no fixed order:

- No hand-to-mouth function, no ventilator
- Hand-to-mouth function, night-time ventilation

People with DMD in both of those states eventually progress to the no hand-to-mouth function and night-time ventilation state.

Late non-ambulatory state

In this state, people with DMD have no hand-to-mouth function and require full-time ventilation.

Data suggests that people with DMD spend approximately:

9.5 years in the ambulatory states

1.5 years in the transfer state

The remainder of their lives in non-ambulatory states with a median predicted survival of 34.8 years

DMD mainly affects males

Prevalence of DMD

DMD is the most common and severe form of muscular dystrophy.

The global prevalence is 19.8 per 100,000 live male births[ii].

DMD can affect any ethnicity. DMD is very rare in females, but it can happen.

Women and girls can also be carriers of DMD. Some female carriers of the disease (approximately 2.5-7.8%)[iii] are 'manifesting carriers', meaning they show some milder symptoms of the disease. This is not the same as having DMD, but these girls and women will produce reduced amounts of dystrophin. Manifesting symptoms can include fatigue, mild muscle weakness, cramping, and they will have an increased risk of heart problems.

Cause of DMD

DMD is a genetic disease. Children can either inherit DMD from their mother if they carry the genetic mutation in the dystrophin gene, or it can be the result of a random genetic mutation.

DMD occurs when there is a fault in the dystrophin gene. This is the gene that the body needs to produce a protein called dystrophin, which is essential for maintaining muscle strength and function.

The dystrophin gene is the largest gene in the body

There are three errors, called mutations, that can prevent any or enough dystrophin being made by the body:

- Exon deletion – this means that one or more exons in the dystrophin gene is missing. This is the cause of around 70% of cases.
- Exon duplication – around 10% of cases are caused by one or more exons having extra copies in the dystrophin gene.
- Other small changes, such as tiny deletions of genetic code. These cause around 20% of cases.

What this report has investigated

This report is based on a wide range of evidence sources including direct patient experiences and surveys, opinions from healthcare professionals and data on service availability direct from Integrated Care Boards, Health Boards, Trusts and other parts of the NHS throughout the UK. It includes:

A survey of those working to help people with DMD

We surveyed 40 people who are working to help those with DMD. The respondents included five neuromuscular consultants, one endocrinologist, five nurses, two physiotherapists, two occupational therapists, four medical research scientists, and four people who work in a school.

Personal experience of DMD

People's experience of DMD care within treatment and support is the driving force behind the work we do. The picture of the DMD experience in the UK that we've developed is informed by what are our founders are living through, a survey of 200 people personally affected by DMD that we ran in April 2024, and a workshop for adults with DMD and parents/carers of children with DMD that we ran in May 2024. Their experiences have framed the priorities laid out in this report.

Freedom of Information requests to healthcare services

To understand more about service availability across the country, we asked all NHS Integrated Care Boards in England and all Health Boards in Wales, Scotland and Northern Ireland about the care they offer to people living with DMD in the form of Freedom of Information requests (FOIs), collected between April and June 2024.

Expert feedback from clinicians

In May 2024, we held a roundtable of experts working across DMD care and research to ask them what areas they thought needed further development, and what was working well.

Evidence review

We undertook an evidence review to understand who is living with DMD in the UK, what DMD care, treatment and support looks like at the moment, and what the impact of DMD is in the UK.

Chapter 3: Executive summary and recommendations

Two families a week are given the devastating diagnosis that their child has DMD.

The impact of the people living with DMD is huge on those who love and care for them - seriously affecting the lives of thousands of people.

They all live with this pain and struggle largely unnoticed by most of the UK.

That is mainly because it is a rare disease – and the DMD experience tells us a lot about what it is like to live with a rare disease.

And while DMD is a rare disease, rare conditions are individually rare but collectively common, with over 3.5 million people in the UK living with a rare condition. Seven in ten rare conditions affect children[iv].

Multiply that impact by the number of people who love and care for the 3.5 million people in the UK with a rare disease.

That's a lot of suffering. It's unacceptable that so much of it is not recognised in our society, not only in healthcare but also in key areas such as research, social policy and education.

Framework for action on rare diseases

This report sets out the state of the DMD nation in the UK.

It outlines the current landscape of care, treatment, research, technological innovation, and awareness of DMD. It shows the progress that has been made and the role of our incredible community in driving forward much of this progress, and the obstacles that still need to be overcome.

More than anything, we hope that it can be used as a template for other rare diseases to show what can be done to overcome our shared problems. It is also an opportunity for rare charities to work together on common challenges that can be addressed through policy changes and lobbying.

We give a framework for change that we want to see in each of these areas, and many of these policy solutions are applicable to other rare diseases.

We look at five key areas for people with DMD.

DMD: researching a rare disease

- The UK Government should look at how trial capacity can be increased at existing Centres of Excellence.
- We encourage the National Institute for Health and Care Research (NIHR) to work with charities to help provide seed funding to enable Trusts to recruit posts to facilitate clinical trials and develop expertise – these posts can become self-sustaining through income from the pharmaceutical industry. This has been proven to work in our model: the DMD Hub.
- We encourage the NIHR to work with charities to further streamline site set-up and support the development of a resource registry.
- The UK Government should work with National Institute for Health and Care Excellence (NICE) and the Medicines and Healthcare products Regulatory Agency (MHRA) to produce best practice in clinical trials to ensure the information regulators need to make assessments is collected from the start. They can draw on our Project HERCULES experience.
- We invite opportunities to collaborate on the development of advanced therapies and gene therapy innovation, building on our investment in research in this area, and opportunities to facilitate academic led research.

Ensuring access to promising treatments: the role of advocacy

- DMD treatments should be appraised via the Highly specialised technology (HST) route instead of Single technology appraisal (STA). HST committees are used to dealing with uncertainty – they can approach these diseases with the pragmatism, sensitivity, and flexibility which the HST route affords, and which a STA does not. To do this, NICE should replace the new criteria for HST with a system that recognises challenging, paediatric, life-limiting, progressive diseases that are complex and need to be treated differently to more common and less complex diseases, which would enable diseases like DMD to qualify for the HST route.
- Carer QoL must be included in health technology assessments. It's currently sometimes considered in a qualitative way but this means there is uncaptured value in a product which can't be translated into anything that is useful for a quantitative-focused quality-adjusted life year (QALY). If a caregiver quality of life (QoL) metric was included, the cost effectiveness arithmetic of medicines for rare diseases and complex paediatric progressive life-limiting conditions would change dramatically.
- The disabling nature of diseases such as DMD adds a significant physical and mental burden and requires significant support from the care system as well as the health system. The costs associated with social services and social care such as Personal Independent Payments, Education, Health and Care Plans, Disability Living Allowance, should be included in health technology assessments.

A scandal - the tech deficit in DMD

- The Minister of State for Social Security and Disability should invite a coalition of disabled people and patient advocacy groups to create a quality standard for equipment provision, in the same way that NHS England has commissioned the National Wheelchair Alliance to devise a quality standard for wheelchair services in England.
- The Government should require each Local Authority to find out how many people in its region have DMD and will need its services, and assess the equipment they will need to live well with it. The Government should use this information to pool and ringfence a national budget which Local Authorities can draw on.
- The Personal Wheelchair Budget should be extended to assistive technology so that people have individual budgets to cover the cost of their assistive technology but to which they can add their own finances if they chose.
- The Government should review how this could be integrated with payments for care such as Direct Payments and Carer's Allowance to avoid duplication in the system.
- Train and recruit more NHS Occupational Therapists to help reduce waiting time for assistive technology assessments. Review and address Occupational Therapists' retention within the NHS.

Standards of Care in DMD - no more postcode lottery

- NICE to include the DMD Care UK clinical guidelines as part of its treatment guidelines for DMD.
- The establishment of clinically driven regional networks of key neuromuscular stakeholders including doctors, allied health professionals, established North Star neuromuscular clinical centres, NHS managers, commissioners, patients, carers and relatives of patients. The network would aim to develop and establish world class and equitable care for people of all ages living with rare neuromuscular conditions in the region, and to improve their quality of life and overall experience of NHS services. This would involve working with health and social care professionals to develop the appropriate services and increase the knowledge and skills required to manage these complex conditions. It would be modelled on the South West Neuromuscular Operational Delivery Network, which is the only Neuromuscular Operational Delivery Network in the UK.

Duchenne UK

- An NHS awareness campaign aimed at increasing understanding of DMD and associated care guidelines, targeted at healthcare professionals that deliver specialist care to DMD patients. This would include the development of three training modules on DMD and the care guidelines, one each for primary, secondary and tertiary care.
- Establish neuromuscular diseases as paediatric sub-specialist training for doctor and health care professionals to ensure the next generation of experts.
- Introduce neuromuscular care co-ordinators or care advisors at each specialist centre to support the delivery of care.
- NHS in England, Scotland, Wales and Northern Ireland to work with Duchenne UK on developing a Centre of Excellence badge that can be awarded to centres delivering DMD care and running DMD clinical research trials to a very high standard.
- We invite opportunities to collaborate on supporting the next generation of neuromuscular clinicians by providing fellowships to attract healthcare professionals into the specialism.
- We invite the NHS to work with us on a pilot for a new model of care, a virtual hospital ward. It would support the joining up of people involved in a patient's health and virtual care. It wouldn't replace in-patient care or in-person appointments but could complement it and allow different members of a patient's multidisciplinary care to work together.

Seeking compassion: the importance of awareness in supporting patients

- The governments in each of the four nations should fund awareness campaigns to improve understanding of the range of neuromuscular conditions. This should be led by input from the neuromuscular community, with the relevant government department and public health body in each nation, in close collaboration with local health systems, clinicians, and other relevant specialists.
- The awareness campaigns should be targeted at healthcare and education professionals, along with materials published online and in print for the public.
- Parents and carers should be offered emergency trauma support following the diagnosis of their child with DMD.
- People with DMD and their parents and carers should be offered psychosocial support from someone trained in DMD when they enter each new stage of DMD.

Chapter 4: DMD - researching a rare disease

There is a lack of treatments for DMD, which is in part a result of a historical dearth of research into the disease. But, in the last ten years research into DMD has stepped up and we are seeing an ever-increasing number of therapeutic programmes in the preclinical and clinical pipeline.

Current research

A big focus of DMD research currently is into developing treatments that target its causes. DMD is caused by a genetic mutation that means the body is unable to produce the dystrophin protein. Dystrophin is essential for strengthening and protecting muscle fibres. This research is looking at ways of replacing the missing dystrophin in people with DMD.

Gene therapy

One of the new potential treatments for DMD that is being researched is gene therapy. This involves delivering new genetic material to cells to overcome errors (or mutations) on the dystrophin gene. DMD gene therapy aims to deliver a working version of the dystrophin gene, so that the body can produce dystrophin.

There had been a lot of hope for gene therapy, particularly as it has been able to halt the progression of other conditions such as spinal muscular atrophy (SMA). However, DMD is a complex condition. In order to work, gene therapy needs to reach the skeletal muscles and the heart. Skeletal muscle is the largest organ in the body and grows with time, and the current limitations of gene therapies mean that they can't be as effective as needed to have a transformative impact on the disease. Results of a major phase three trial showed that the gene therapy didn't achieve a statistically significant difference in the key measure chosen to test its effectiveness (primary endpoint)[v], the North Star Ambulatory Assessment, which measures motor function in people with DMD. However, other measures of the trial (secondary endpoints) were met. In June this year, another trial of gene therapy to treat DMD showed that it didn't meet its primary or secondary endpoints[vi].

We are funding research to overcome the challenges of gene therapy in DMD, with projects looking at reducing the patient's immune response, which can prevent gene therapy from entering the body, and exploring alternative delivery methods for gene therapy that could carry a larger dystrophin gene.

Stem cells

In recent years, considerable research effort has been directed to developing cell therapy as a new therapeutic option to treat DMD. Stem cell-based therapies' goal is to replenish the muscle cell pool with cells containing dystrophin that are able to promote muscle regeneration.

Gene editing

Gene editing, or genome editing, is a technology that enables the editing of parts of the genome (the body's set of genetic instructions) by removing, adding or altering the DNA sequence. This is a new area of research that has the potential to make precise, targeted changes to correct the mutations that cause diseases like DMD.

Researchers have developed techniques that use enzymes (called endonucleases) that work like a pair of molecular scissors to cut the DNA at a specific location. A guide RNA (gRNA) ensures that the enzyme cuts the DNA in the right place. Once the DNA has been cut, the body tries to repair it. This removes the deletion that prevents dystrophin being produced, and allows for the genetic sequence to be read. This approach has been mainly tested in animal models. As with gene therapy, there are several challenges to delivering it as a treatment.

Exon skipping

The active part of the dystrophin gene is made up of 79 pieces called exons. These exons link together to form a code that is read in the cells so that the protein dystrophin can be made. In DMD, some of the exons are not readable. The result is that very little or no dystrophin is made. Exon skipping drugs hide or 'patch' the missing piece so that the exons fit together again and can be read. This means that a functional, although shorter, dystrophin protein can be produced by the body.

There are four approved exon skipping treatments in the US. However, since exon skipping drugs are designed to skip over a particular exon, they are mutation specific and can only be used in the patients carrying that mutation.

Repurposing drugs

Research is taking place to explore whether existing treatments for other conditions can be repurposed to treat the symptoms of DMD.

This is an important area of research. Not only for the therapeutic possibilities, but repurposed drugs also have a significant advantage in decreasing the development cost and time to market over standard discovery.

We have experience in this area of research. Along with our charity partners, Alex's Wish and Joining Jack, we funded a trial called TAM-DMD in 2017 to assess the safety and efficacy of tamoxifen.

We are funding a clinical trial of a muscle stem cell therapy to look at its potential to promote muscle regeneration in the feet of boys with DMD. This research is a stepping stone towards progressing to larger trials expanding the use of this approach to larger muscles in the arms and the legs.

This drug, which is currently used to treat breast cancer, had shown promise for DMD in preclinical research studies. Unfortunately, the trial did not show evidence for tamoxifen's effectiveness as a DMD treatment.

We believe that there is a significant research opportunity in this area for cardiac treatments. This is particularly important as cardiac care is so important for people with DMD. That's because the heart is a critical determinant of survival in DMD.

Funding for DMD research

Despite a marked increase in DMD research over the last ten years, it remains a critically underfunded topic of research.

The Rare Diseases Research Landscape Project Report[vii] published in September 2023 analysed the number of industry projects in rare disease research and development in the UK between 2016 and 2021, and found that there were less than a third of projects focused on DMD compared to those on pulmonary arterial hypertension, and less than half compared to uveitis, and idiopathic pulmonary fibrosis.

The NIHR, the UK's largest public funder of clinical research, has put significant funding into supporting research into rare diseases and making advances in developing new tests and treatments over the last few years. As part of the three Rare Diseases Action Plans for England that were published in 2022[viii], 2023[ix] and 2024[x], they have invested £790m for the NIHR Biomedical Research Centres. The NIHR, along with the Medical Research Council, has also set up the UK Rare Disease Research Platform to drive collaboration in research. While LifeArc announced in April this year that it was setting up four new LifeArc Translational Centres for Rare Diseases[xi].

However, families of people with DMD have had to drive and raise funds for a significant amount of DMD research. We have committed millions of pounds to DMD research since we launched in 2012.

We launched a grant call in 2024 aiming to invest up to £1.5 million to fund research into transformative treatments for DMD. This includes research to support the repurposing of new treatments for the DMD heart.

We joined forces with The Patrick Trust, who is committing £200,000 to it over three years, and our partner charities Joining Jack and Alex's Wish.

DMD trial infrastructure

The Government's UK Rare Diseases Framework acknowledged that very few rare diseases have established treatments, but where they do exist, they can be life-changing, significantly improving prognosis and/or quality of life.

It recognised the need to research into rare diseases and stated that it was essential that the UK can offer an environment that will attract substantial investment in high value life science products of the future, and that will attract discovery scientists from global pharmaceutical companies to the UK. This aligned with its ambitions to attract and retain global investment, science, research and innovation talent to the UK that are set out in the Government's UK Life Sciences Industrial Strategy published in 2017[xii] and reiterated in its 2020 update[xiii].

A key part of attracting research is having a robust trial infrastructure. A lack of trials and trial infrastructure is what our two co-founders found when their sons were diagnosed with DMD in 2011. They were shocked to learn how little research into DMD treatments was taking place globally and how limited the opportunities to take part in DMD clinical research trials were for people living with DMD in the UK.

Research is not only important for increasing understanding and developing treatments, it is a chance to access potentially life-changing treatments when no effective treatments have been approved. People living with a progressive and life-limiting condition such as DMD need early access to innovative treatments that could improve, extend or even save their lives.

To address the issue of a lack of DMD trials in the UK, we created the DMD Hub in 2016 in collaboration with The John Walton Muscular Dystrophy Research Centre at Newcastle University.

Case study

DMD Hub – providing DMD clinical research infrastructure and access to clinical trials for people living with DMD in the UK

The DMD Hub is a network of clinical trial sites that have dedicated, trained staff, funded to carry out research studies for DMD.

The DMD Hub's network includes eleven trial sites, coordinated by a team funded by Duchenne UK at the John Walton Muscular Dystrophy Research Centre at Newcastle University, which undertake trials for people with DMD from across the UK. By the end of 2023, the DMD Hub has helped run 51 trials and 793 people with DMD have taken part in a DMD trial (interventional, open label extension, observational and natural history) since it was set up in 2016.

To help ensure children and adults with DMD experience fair and equitable access to clinical trials, we have created a central recruitment database as part of the DMD Hub. The database allows people with DMD who are interested in taking part in research studies to register their interest. Information within the database is used to support clinical sites to identify potentially eligible candidates for research studies.

It also provides clinical research sites with the contact details for the individual's main neuromuscular centre and clinician to facilitate a joined-up approach. It means people with DMD are now able to take part in research studies, regardless of where they live, and clinical trials can recruit to time and target, helping ensure the UK remains an attractive place for companies to run clinical trials.

While our DMD Hub has been very successful in delivering and recruiting to clinical research trials for DMD, it is not operating in a vacuum. The health of the UK's life sciences sector impacts its ability to attract commercial clinical research trials including DMD research trials.

In May 2023, Lord O'Shaughnessy published a report following an independent review into commercial clinical trials in the UK[xiv]. He emphasised the importance of clinical trials in product research and development and said that "for a therapy, device, diagnostic or digital tool to reach patients, a long, often laborious process of translating insights into products and then testing their safety and efficacy through clinical trials is required".

Worryingly, his report pointed out how the UK is falling behind in the number of trials initiated, particularly for phase three trials, with the UK's relative ranking against other countries dropping from 4th to 10th best in the world in the same timeframe.

To change this and make the UK one of the best places in the world to conduct clinical trials, Lord O'Shaughnessy recommended that the Government creates clinical trial acceleration networks (CTANs) that would be designed, funded and equipped to deliver genuinely best-in-world clinical trial services in areas of critical strategic interest for the UK's health and life sciences sectors.

The Government responded to the report by announcing its commitment to a set of measures that would support clinical research trials in the UK, including establishing support to emerging place-based knowledge clusters to ensure that research delivery is spread both nationally and at a local level[xv].

The CTANs are very similar to the DMD Hub and we welcome their development by the Government. We have a set of policy recommendations for how DMD research and clinical trials in the UK can be supported further.

> ***"The DMD Hub is a leading example in the UK of clinical research infrastructure."***
> Lord O'Shaughnessy

DMD HUB
Expanding Trial Capacity

DMD Hub Sites

DMD Hub Coordination Centre

Recommendations for supporting DMD research

- The UK Government should look at how trial capacity can be increased at existing Centres of Excellence.
- We encourage the National Institute for Health and Care Research (NIHR) to work with charities to help provide seed funding to enable Trusts to recruit posts to facilitate clinical trials and develop expertise – these posts can become self-sustaining through income from the pharmaceutical industry. This has been proven to work in our model: the DMD Hub.
- We encourage the NIHR to work with charities to further streamline site set-up and support the development of a resource registry.
- The UK Government should work with National Institute for Health and Care Excellence (NICE) and the Medicines and Healthcare products Regulatory Agency (MHRA) to produce best practice in clinical trials to ensure the information regulators need to make assessments is collected from the start. They can draw on our Project HERCULES experience.
- We invite opportunities to collaborate on the development of advanced therapies and gene therapy innovation, building on our investment in research in this area, and opportunities to facilitate academic led research.

Chapter 5: Ensuring access to promising treatments - the role of advocacy

> ***"I'm scared. My son is two and a half, and thinking about putting him onto steroids is something I am really struggling with."***
> Parent of child with DMD

It is only in the last few years that treatments for DMD have been developed and re-purposed treatments prescribed as part of the Standards of Care for DMD.

That is because DMD and its cause (dystrophin deficiency) were only identified in the late 1980s[xvi]. An increased understanding of dystrophin structure and function since then has enabled the emergence of therapeutics to treat it.

This meant that, until very recently, as their young child was diagnosed, parents were told that there were no treatments. They were told that there was nothing that could be done to prolong their child's life, let alone cure DMD.

Current treatments

There is no cure for DMD but currently available treatments can improve quality of life and longevity to a varying degree.

This, combined with the improved DMD Standards of Care, can have a significant positive impact on the lives of people with DMD.

Steroids

Steroids (also called corticosteroids or glucocorticoids), are used as a routine part of DMD treatment. These are anti-inflammatory drugs which can slow the progression of weakness in the muscles, reduce the development of scoliosis (curvature of the spine) and delay breathing and heart problems. On average, children who take steroids walk for over two years longer[xvii] than those that don't.

Unfortunately, steroid treatment also has significant side effects, including weight gain, changes to mood and behaviour, weak bones, delayed growth and puberty, and stomach irritation. This can be very hard for children who are taking them, and some don't tolerate them at all.

Cardiac treatments

The heart is a muscle and dystrophin is needed to make it work properly. DMD inevitably causes a type of heart disease, called cardiomyopathy, where the heart muscle does not pump blood around the body efficiently.

Several drugs are currently available to help protect the heart in DMD patients and delay the onset of cardiomyopathy. The latest cardiac care guidelines agree that they need to be given early – before heart damage is detected, but more research is needed to back this up. These treatments currently include angiotensin converting enzyme (ACE) inhibitors and angiotensin receptor blocks (ARBs), beta-blockers, diuretics, anti-arrhythmic therapy, and left ventricular assist devices. There is also evidence that steroids can delay onset of cardiomyopathy.

But we still need to understand more about cardiac involvement in DMD so that more effective treatments can be developed to keep the hearts of people with DMD healthier for longer, or even to prevent the development of cardiomyopathy in DMD altogether.

Bone protection

People with DMD often have weak bones or reduced bone mineral density, caused by decreased mobility, muscle weakness and the use of steroids. This can lead to the weakening and thinning of bones, called osteoporosis, which makes fractures (broken bones) more likely. Children with fragile bones can be given bisphosphonate treatments, such as zoledronic acid. Doctors may also prescribe supplements such as Vitamin D and calcium to improve bone health.

Ataluren

Ataluren (brand name is Translarna), was approved by NICE in January 2023 to treat DMD in people who are two years and over, and who are ambulant (can walk). Ataluren can only be given to patients who have a specific type of genetic defect called a nonsense mutation, which is approximately 13% of people with DMD[xviii].

While it is still available in the UK, the European Medicines Agency (EMA) announced in January 2024 that it would not be renewing its previous authorisation of ataluren[xix]. This followed the full re-evaluation of its benefits and risks during the renewal of its marketing authorisation, which concluded that its effectiveness had not been confirmed.

Non-medicinal treatments

There are also several non-medicinal therapies that can help manage DMD. These include physiotherapy, moderate exercise, spine surgery, foot splints, and a biphasic positive airway pressure or continuous positive airway pressure machine (a ventilator) to assist with breathing at night.

People with DMD need a healthy, well-balanced diet and can take nutritional supplements. This is to support health and manage weight as weight gain is common in DMD due to reduced mobility and the side effects from steroids.

Some children and young people with DMD have associated learning and behavioural difficulties, as dystrophin is present in the brain as well as muscle cells. For example, people with DMD have higher rates of autistic spectrum disorders, anxiety, dyslexia and attention deficit hyperactivity disorder. Appropriate and timely educational support is needed in order that people with DMD can achieve their full potential.

Treatments undergoing regulatory approval in the UK

Vamorolone

Vamorolone (brand name is Agamree), a synthetic corticosteroid, was developed to keep or improve the efficacy profile of steroids, but with the aim of having fewer side-effects.

Vamorolone was approved by the MHRA in January 2024. It is currently being appraised by NICE.

Case study

Supporting vamorolone – seeking better treatment for people with DMD

Duchenne UK played a major role in the development of vamorolone in the hope of finding an alternative to steroids.

In 2015, Duchenne UK, Joining Jack, and Duchenne Research Fund invested £750,000 towards a phase 1 clinical trial in boys with DMD, which then enabled ReveraGen to win a £6 million grant from Europe's Horizon 2020.

Two years later we helped to set up the Phase 1 trial through the DMD Hub, offering ReveraGen assistance and support. The data showed the drug to be safe, and the company was able to go to Phase 2 trials, again supported by Duchenne UK, which were also successful.

Now produced by Santhera Pharmaceuticals, it is the first treatment for all DMD patients approved by the MHRA.

Duchenne UK has an interest in the technology as a result of this grant funding agreement. Duchenne UK received a milestone payment when the commercial rights of the technology were sold. Duchenne UK received $130,000 in Oct 2020, and Duchenne UK will receive further milestone payments based on future net sales of the product.

Givinostat

Givinostat is an 'HDAC inhibitor'. It blocks enzymes called histone deacetylases (HDACs), which are involved in turning genes 'on' and 'off' within cells. It works by targeting pathogenic processes to reduce inflammation and muscle loss.

Results from the Italfarmaco Pivotal Phase 3 EPIDYS Study of givinostat in DMD were published in The Lancet Neurology in March 2024[xx]. The EPIDYS clinical trial met its primary endpoint: change in four-stair climb assessment from baseline to 72 weeks, which demonstrated the potential of givinostat to delay disease progression when added to corticosteroid treatment. Givinostat-treated boys also showed favourable outcomes on key secondary endpoints assessed in the study.

The challenges facing new treatments for DMD

Although there are currently no transformative treatments that are able to dramatically change the trajectory of the disease, new medicines that have been proved to slow down its progression have been developed.

The key issue for these treatments is for them to be approved as quickly as possible. Time is of the essence when it comes to a progressive and life-limiting condition such as DMD, and people living with it cannot afford to wait. They deserve the chance to keep doing all the things they love for longer.

There are some specific challenges to the reimbursement of DMD treatments in the UK. This is reflected in the significantly smaller number of drugs approved for use in the NHS in the UK by the relevant health technology assessment bodies compared with the US. The United States Food and Drug Administration estimates that just 5% of rare diseases have an approved treatment[xxi], and many treatments for rare disease that are approved in other countries are not approved in the UK. Only 59% of treatments approved by the EMA between 2018-2021 had been approved in the UK, compared to 86% in Germany[xxii]. A key part of the problem is the changes to NICE's health technology assessment pathways. In 2022, after a multi-year consultation, NICE introduced four new routing criteria to replace the previous system used to determine whether a treatment would be assessed through the Single Technology Appraisal (STA) route, or the High Specialised Technology (HST) route.

These new criteria do not favour rare diseases such as DMD, because they determine that DMD is -too common-. They introduced a requirement that a treatment with more than 500 possible patients must be assessed through the standard STA, and not the more flexible HST.

Duchenne UK objected to these changes during NICE's consultation, and flagged that they would disadvantage conditions like DMD. These new criteria were introduced anyway, and we believe they should be repealed as soon as possible, and replaced by a system based on complexity, need, and the availability of data.

- A STA is not able to fully assess a complex disease such as DMD. Most rare diseases are complex, progressive and disabling, and there are often huge uncertainties in relation to the long-term outcomes and the impact of new treatments on them. Most (75%) affect children[xxiii] and as the child is growing and maturing, their bodies are changing. As a result, there is a constantly

moving baseline which makes it harder to assess the impact of a treatment. A system designed to evaluate traditional medicines for the general population is too rigid to accommodate the challenges posed by rare diseases such as DMD in children.

- In STA, the same process for measuring treatments for adults is applied to measuring treatments for children. However, there are significant differences between measuring a treatment for an adult than one for a child. For example, there will be inconsistency and bias when it comes to measuring functional outcomes for paediatric treatments as a child is more susceptible to external factors (parental pressure, impact of change of routine, upset at being taken out of school for the day) that can influence how they behave during the measurement. The STA system isn't flexible enough to take this into account.

- Another issue is that 'Quality of life' (QoL) – a key concept in assessing the cost effectiveness of a medicine – is measured in paediatrics like in adults, despite the huge differences between the two. Research by ISPOR, the Professional Society for Health Economics and Outcomes Research, has found that there are no adequate QoL instruments in paediatrics[xxiv].

- It is also important to consider that some therapies developed to treat DMD may not be applicable to all people with DMD, so should therefore be considered for the HST route. For example, mutation specific therapies or gene therapies that are only applicable to a narrow age range and boys who do not have immunity to the vector. The costs of these therapies will be prohibitive if not awarded HST status.

In 2017, we set up Project HERCULES, a collaborative global project to increase the chances of patients with DMD of accessing innovative treatments. Through it, we have gained a very good understanding of the health technology assessment system in the UK, and have a set of policy recommendations for how the issues with STAs can be addressed and that would support a more appropriate way of assessing rare diseases such as DMD.

HERCULES

Case study

Project HERCULES – developing disease level evidence to support the case for approving new treatments

Project HERCULES (HEalth Research Collaboration United in Leading Evidence Synthesis) is a collaborative global project set up by Duchenne UK in 2017 to increase the chances of patients with DMD of accessing innovative treatments.

We set up Project HERCULES to address many of the issues we found with health technology assessment and reimbursement. We brought the main stakeholders together: patients, clinicians, academics, advisers, the regulators and industry in a non-competitive collaborative environment.

Together we developed tools and evidence to inform decisions about new treatments and support their case for the reimbursement of new treatments for DMD.

Since 2017, Project HERCULES has produced[xxv]:

- A new natural history model for DMD that identifies a new stage, the Transfer Stage, and new mortality data analyses.
- DMD-QoL, a new bespoke Quality of Life measure for DMD, along with 26 translations/linguistic validations to ensure it can be widely applied in clinical trials and practice.
- A cost of illness study to map the impact of DMD on patients, families and carers in the UK.
- Analysis of patient data from Clinical Practice Research Datalink, to inform the natural history and burden of illness workstreams.
- A core economic model, adaptable by individual companies, to support health technology assessment and reimbursement decisions.
- A systematic review of Quality of Life measures for DMD carers and families.
- A review of the burden of side effects from long-term corticosteroid treatment.

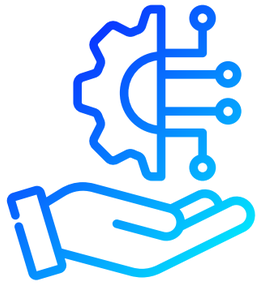
Recommendations for improving health technology assessments for rare diseases such as DMD

- DMD treatments should be appraised via the HST route instead of STA. HST committees are used to dealing with uncertainty – they can approach these diseases with the pragmatism, sensitivity, and flexibility which the HST route affords, and which a STA does not. To do this, NICE should replace the new criteria for HST with a system that recognises challenging, paediatric, life-limiting, progressive diseases that are complex and need to be treated differently to more common and less complex diseases, which would enable diseases like DMD to qualify for the HST route.

- Carer QoL must be included in health technology assessments. It's currently sometimes considered in a qualitative way but this means there is uncaptured value in a product which can't be translated into anything that is useful for a quantitative-focused quality-adjusted life year (QALY). If a caregiver QoL metric was included, the cost effectiveness arithmetic of medicines for rare diseases and complex paediatric progressive life-limiting conditions would change dramatically.

- The disabling nature of diseases such as DMD adds a significant physical and mental burden and requires significant support from the care system as well as the health system. The costs associated with social services and social care such as Personal Independent Payments, Education, Health and Care Plans, Disability Living Allowance, should be included in health technology assessments.

Chapter 6:
A scandal - the tech deficit in DMD



As well as treatments for DMD, assistive technology is an important way to help people with DMD manage as their muscles waste and they lose limb function.

Assistive technology is any item, piece of equipment, software program, or product system that is used to increase or maintain the functional capabilities of people with disabilities.

A wheelchair is one of the most common pieces of assistive technology equipment.

The life-changing potential of assistive technology

Assistive technology has the potential to help people with DMD continue to do things for themselves for longer. This can be everyday tasks such as cooking to brushing teeth and eating without needing someone else's help. The right assistive technology equipment can also enable people with DMD to keep doing the things they love for longer, from playing musical instruments to painting.

The stages of DMD progression where there could be most benefit from assistive technology equipment are from late ambulatory onwards. Being able to look after yourself through young adulthood can considerably increase overall physical and mental wellbeing. Teenagers and young adults with DMD want to maintain their independence and to be able to make the multiple choices (when to have breakfast, to read a book or play the guitar) that we make every day without having to rely on someone else.

Unfortunately, despite it being so fundamental to the lives of people with DMD, there are serious issues with accessing even the most basic assistive technology equipment as well as a lack of development of new assistive technology in areas of unmet need. As a result, too many people with DMD are missing out on the life-changing potential of assistive technology.

Access to assistive technology

This is part of a bigger issue for people with physical disabilities. The 2022 Global report on assistive technology by the World Health Organization and United Nations Children's Fund[xxvi] found that the majority of the people who need assistive technology do not have access to it.

In the UK, research by Newlife, the charity for disabled children, found that only two fifths of disabled children are provided with the specialist equipment they need to live a full life[xxvii].

This research identified a lack of local authority funding and very long waiting times for assessments as the main obstacles to access. It found children in almost a quarter of local authorities must wait over a year to be assessed for specialist equipment, with some having to wait three or more years. Added to this, nearly half of local authorities cut their equipment budgets in the last year. As a result, more families are turning to charities or self-fundraising to meet their child's basic equipment needs.

Meanwhile, the Parliamentary and Health Service Ombudsman has recently written to AJM Healthcare, the leading provider of NHS wheelchair services in the UK, after a significant increase in complaints from wheelchair users[xxviii]. Most related to people not receiving new wheelchairs or the correct parts, with the wait ranging from a month to two years.

Lack of innovation in assistive technology

There is also a serious lack of innovation in the assistive technology sector.

Although upper arm assistive devices have the potential to improve quality of life for people with DMD by enabling them to continue performing daily activities, most have been designed for rehabilitation to regain strength and motor control. Few have been designed to assist people with DMD during activities of daily living[xxix].

With the rapid pace that technology has developed over the last few decades, for example the transformation of the smart phone, you would expect that there would be technology to help people with disabilities such as DMD to stay independent for longer.

Unfortunately, and shockingly, there isn't. As people with DMD discover, as their muscles weaken, there is technology to help them organise and host an international webinar via their mobile phone, but nothing to help them lift a spoon or a fork for longer.

As for the long-standing technology that people with physical disabilities have relied on, such as wheelchairs, these have seen only minor innovations over the last decade and more.

The impact of this lack of innovation and how it isn't meeting the needs of people with physical disabilities is reflected in the fact that people who need assistive technology often give up on using it, with up to seven in ten people abandoning provided assistive technology equipment. A key reason identified was the design and function, including lack of user and therapist involvement in the design process, lack of options for customising it, high purchase and maintenance costs, and not being easy to use. Some technologies were too big or heavy for the user to carry, or to use in their home. Others could not be used in certain environments.[xxx]

Revolutionizing the assistive technology sector

Frustrated by the lack of technology to help people with DMD, we have embarked on two major technology projects.

Re-thinking the wheelchair

In 2018, People's Postcode Lottery funded a collaboration between WhizzKidz and the University of Edinburgh to develop the first prototype of the Dream Chair. The outcome of the project was the mark 1 prototype, which had lots of unique features. In 2021, we took on the Dream Chair project and explored manufacturing options for the mark 1 design concept to see if it could be made using existing manufacturers and production lines.

We explored what's possible using current production methods by working with a wheelchair manufacturer to create the mark 2 prototype. We invested our own money in doing this. We then took the prototype back out to the community and clinical stakeholders for review in January 2023.

The feedback from the DMD community was mixed, so we decided to dig deeper into the underlying issues and commissioned some user research into powered wheelchairs. This research included observing people with DMD using their powered wheelchairs in different environments and speaking to them about their experiences of being a powered wheelchair user. We learnt about the challenges of stability over rough and uneven terrain, the annoyance of the beeps from the controller, the importance of how it looks and much more.

We are now taking these learnings forward into three concept visions – opportunities for innovation that would transform the market and answer the expectations of the community. We'll conduct a benchmarking exercise to ensure that our chosen vision for the project is transformative when compared to current wheelchairs on the market. We will be engaging the community at every stage, and working in collaboration with organisations who share our vision and values.

Duchenne UK

Developing an arm-assist for upper body mobility

We are also developing an arm-assist, Elevex. We initially contributed funds in 2019 towards a Solid Biosciences project that used Twisted String Actuators (TSA) to create a product to help people with DMD use their arms. TSA technology had been created by the Stanford Research Institute to use in a lightweight, wearable garment – a soft exoskeleton suit.

And while there are other exoskeletons available, most aren't appropriate for people with additional needs related to their health condition. Whereas the Elevex exoskeleton could be life-changing for many people with DMD – and potentially other health conditions.

Stanford Research Institute built the first prototype of the suit in 2020. It also formed a company to commercialise the technology for use by people working on production and assembly lines. That company went on to become Seismic Powered Clothing. As the project neared its end, Solid Biosciences decided to focus on another area of research, and Seismic Powered Clothing focused on developing the suit for assembly workers.

ELEVEX
POWERING ARM MOBILITY

Case study

Elevex – giving people the freedom and confidence to move through life and embrace whatever comes their way

Determined to develop technology that could help people with DMD, and people with other conditions that affected upper body mobility, we took over the project in 2021. Knowing that we would need additional funding to get started, we applied to and won £1.25 million from People's Postcode Lottery's Dream Fund. Since then, we've raised another £800,000 through our partner charities, major donors and Family and Friends Funds to support the development work.

Building an innovative partnership

We are working on the Elevex project with the charity Spinal Muscular Atrophy UK and the University of Liverpool's Inclusionaries Lab. Our partnership of two leading UK charities and a top UK academic institution driving the development of a new product is almost unheard of in the world of design. But, as non-profit organisations, the difference is that we are innovating for impact over profits.

By coming together and collaborating with other organisations who have the expertise we need, we can accelerate the development process and get the product to market sooner. And we are doing so with a user-led product design strategy.

Developing an inclusive and life-changing exoskeleton

Since taking on the project in 2022, we've conducted in-depth research and built four working prototypes that we've tested with individuals who have either DMD or SMA.

A big focus for our latest prototype was to improve the lift mechanism to help reduce the loading on the shoulder joint, and also integrate a more intuitive control system. It's currently controlled by a switch, but we hope to use something like motion detection in the final product.

Showcasing development progress

We showcased the prototype this year and demonstrated it to an international audience at South by Southwest (SXSW), a technology and entertainment conference and festival that took place in Texas in March. Our co-founder and Chief Executive, Emily Reuben OBE, her 16-year-old son Eli, who has DMD, and Hayley Philippault, our Head of Technology, took part in a panel discussion about assistive technology. They stressed the need for a massive step change in how technology helps people with disabilities such as DMD. Awareness of this issue was raised further in a BBC Click documentary about Elevex and how it has the potential to help Eli keep his dreams of a music career alive[xxxi].

We are now currently refining and further co-developing the design and are on schedule to have the final device ready for manufacture by the end of 2025. We are excited and hopeful about launching a revolutionary piece of assistive technology to reenable meaningful arm movement for people with DMD.

Duchenne UK

While we aim to become the go-to partner for people with DMD by supporting them with life-improving products we all love, there are major issues with access to assistive technology equipment and the assistive technology industry. We have a set of policy recommendations for how to reduce barriers to assistive technology and how to unlock innovation in the sector.

Recommendations for increasing access to and innovation in assistive technology

- The Minister of State for Social Security and Disability should invite a coalition of disabled people and patient advocacy groups to create a quality standard for equipment provision, in the same way that NHS England has commissioned the National Wheelchair Alliance to devise a quality standard for wheelchair services in England.

- The Government should require each Local Authority to find out how many people in its region have DMD and will need its services, and assess the equipment they will need to live well with it. The Government should use this information to pool and ringfence a national budget which Local Authorities can draw on.

- The Personal Wheelchair Budget should be extended to assistive technology so that people have individual budgets to cover the cost of their assistive technology but to which they can add their own finances if they chose.

- The Government should review how this could be integrated with payments for care such as Direct Payments and Carer's Allowance to avoid duplication in the system.

- Train and recruit more NHS Occupational Therapists to help reduce waiting time for assistive technology assessments. Review and address Occupational Therapists' retention within the NHS.

Chapter 7: Standards of Care in DMD - no more postcode lottery

DMD is a progressive multi-systemic condition, affecting many parts of the body, which results in deterioration of the skeletal, heart, and lung muscles. DMD also has an impact in the brain. Care for DMD patients is complex.

This has resulted in many parents of children with DMD and adults with DMD having to try and navigate the care system for themselves, and almost project manage their own care.

However, the care for people with DMD has been transformed over the last 15 years by the improved care standards for DMD patients.

Care Considerations for DMD

First of all, general recommendations for medical care in DMD were published in 2009 by the DMD Care Considerations Working Group, under the auspices of the United States Centers for Disease Control (CDC)[xxxii]. These recommendations, that were published in The Lancet Neurology in 2010, were the first comprehensive set of clinical care recommendations for the diagnosis and management of DMD. They focused on diagnosis, and pharmacological and psychosocial management and the implementation of multidisciplinary care.

Standards of Care

This was followed by the publication, in three parts, of updated Care Considerations for DMD (also called the Standards of Care) by the CDC in *The Lancet Neurology* in 2018[xxxiii] which was a landmark moment for the care of patients with DMD.

The new Standards of Care reflected how clinical care for DMD had evolved in the space of a few short years. They were focused on improving patient care and aimed at addressing the needs of patients as they lived longer, to give guidance on assessment and treatment options, as well as considering the genetic and molecular therapies for DMD.

They were also more comprehensive, and covered diagnosis, neuromuscular, rehabilitation, endocrine, gastrointestinal and nutritional management, respiratory, cardiac, bone health and orthopaedic management, primary care, emergency management, psychosocial care, and transitions of care across the lifespan.

Adult North Star Network guidelines

Now with a growing number of adults with DMD in the UK, the Adult North Star Network of neuromuscular specialists recognised the need for adult-focused standards of care where the Care Considerations for DMD and Standards of Care primarily addressed care for children with DMD.

It published a Consensus Guideline for the Standard of Care of Adults with Duchenne Muscular Dystrophy[xxxiv] in 2021 to provide guidance for health professionals to ensure adults with DMD get best-practice care. It also published a therapist-specific guideline[xxxv] in 2022 to support therapist healthcare professionals who were not familiar with DMD in adults or managing the complexity of care it involves.

Development of care standards for UK healthcare system

While the development of care standards for DMD has been a seminal stage in the understanding of how best to treat people with DMD, there was concern across the DMD community in the UK about the extent to which they would be implemented here.

The international Standards of Care were issued in 2018, but rather than streamlining care in the UK, they led to confusion. Some of the recommendations (such as annual cardiac MRI) proved to be unworkable in the UK.

To ensure UK care delivery was improved and harmonised in line with the latest guidance, our co-founder Alex Johnson collaborated with Centre of Excellence, the John Walton Muscular Dystrophy Research Centre (JWMDRC) in Newcastle, to lead together the launch of our DMD Care UK programme at the beginning of 2020. DMD Care UK is a new nationwide initiative to ensure every person living with DMD in the UK has access to the best care, no matter where they live.

DMD Care UK brings together expert clinicians and the patient community to agree, communicate and implement DMD standards of care recommendations for the UK. Each guideline is developed and agreed by its own working group of specialists in that area and patient representatives after wide consultation across UK specialist care centres in the North Star network. All recommendations have been based on the 2018 Standards of Care but are more specific, practical and relevant for the UK healthcare system. Working closely with relevant professional bodies, each guideline is endorsed and, where appropriate, published in peer reviewed journals. Accessible versions of the recommendations are produced for patients and families.

Case study

DMD Care UK – driving better care for all DMD patients in the UK

DMD Care UK is a joint Duchenne UK and JWMDRC initiative, co-funded by Duchenne UK, Joining Jack and the Duchenne Research Fund. It is run in collaboration with the North Star Network.

Since 2020, DMD Care UK has produced[xxxvi]:

- Guidelines for healthcare professionals on bone and endocrine care for DMD patients (endorsed by the British Society for Paediatric Endocrinology and Diabetes) – and a guide for patients and families.
- Information for patients and families on managing adrenal insufficiency (caused by long-term steroid treatment) and how to prevent adrenal crisis.
- Guidelines on cardiac care for DMD patients (endorsed by the British Cardiovascular Society) – and a guide for patients and families.
- Guidelines on respiratory care for DMD patients (endorsed by the British Thoracic Society) – and a guide for patients and families.
- Guide for patients and families on managing pubertal delay and testosterone therapy for people with DMD.
- Guidelines for healthcare professionals on what to be aware of when a person with DMD presents in hospital during an emergency or unplanned visit – and a webinar for patients and families.

Adoption of care for DMD patients in the UK

Although the international Standards of Care were issued in 2018 and DMD Care UK has been developing them for the UK healthcare system over the last four years, far too many patients in the UK are not receiving the care they need.

As implementing the Standards of Care results in a lower burden on the NHS, this is a missed opportunity to optimize NHS resources.

However, it also means that there is very much a postcode lottery across the UK as to which DMD patients receive the best standards of care, and which don't. This is despite the Government's UK Rare Diseases Framework that was published in 2021 identifying increasing awareness of rare diseases amongst healthcare professionals as a priority. It highlighted the need to ensure that those involved in patient care are provided with appropriate education and support.

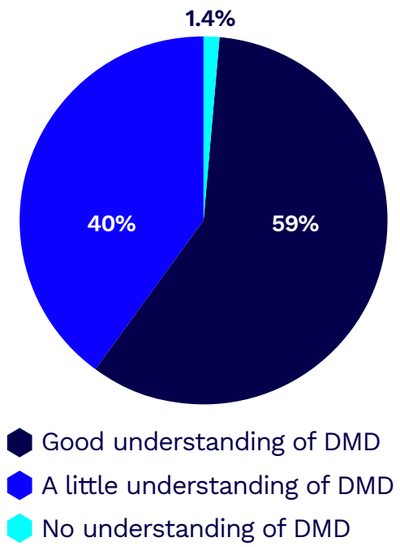
We ran a survey of DMD parents and caregivers of children and adults with DMD[xxxvii] in 2021 to understand how Standards of Care are delivered through UK healthcare sites and measure awareness of them amongst those personally affected by DMD in the UK. It found that satisfaction with care is highest for specialist neuromuscular care (67%), cardiac care (65%) and therapy (physio and occupational) (64%) and lower for emotional support (22%), emergency care (26%), gastrointestinal and nutrition (23%), and transition of care (33%).

We ran two further surveys[xxxviii] in 2024 to ascertain care standards six years on from the issuing of the Standards of Care in 2018. One survey was for people personally affected by DMD and the other survey was for those working to help people with DMD.

The survey of 200 people personally affected by DMD found that:

- Only 31% of people thought that healthcare professionals generally had a good or very good awareness and understanding of DMD.
- When it comes to healthcare professionals treating DMD, only 59% of people thought they had a good understanding of DMD, 40% thought that had a little understanding of DMD, and 1% thought they had no understanding of DMD.
- Only just over half (53%) thought that they and their family have been given the information by their healthcare practitioners that they need to understand DMD and its treatment.
- Only 20% of people thought that their healthcare professionals treating DMD were very familiar with the best practice guidelines on treating it, 37% said that they were familiar, 28% somewhat familiar, 14% unfamiliar, and 1% very unfamiliar.

Perceptions of awareness of DMD among healthcare professionals treating their DMD



The survey of 40 people working to help people with DMD found that:

- Only 19% of people thought that healthcare professionals generally had a good or very good awareness and understanding of DMD. This was not much higher than their perception of members of the general public's good or very good awareness and understanding of DMD (16%).
- When it comes to healthcare professionals treating DMD, only 65% of people thought they had a good understanding of DMD, 31% thought they had a little understanding of DMD, and 4% thought they had no understanding of DMD.
- Only 19% of people thought that healthcare professionals treating DMD were very familiar with the best practice guidelines, 35% said that they were familiar, 27% somewhat familiar, 19% unfamiliar.

As well as a perceived lack of awareness and understanding of DMD Standards of Care among individuals, it was almost always absent from NHS policies, guidance and plans.

Freedom of Information requests to all 42 Integrated Care Boards in England, 14 Health Boards in Scotland, seven Health Boards in Wales, and six Health and Social Care Trusts in Northern Ireland, found that:

- Only 35 have any DMD-specific (or muscle-wasting disease-specific) policies or guidance.
- Only 15 of their organisation's strategies, objectives, plans, performance indicators, audits, inspections, or similar, cite DMD (or any muscle-wasting disease) or use either as a metric.

This is disappointing. The introduction of the new Integrated Care Systems in England in 2022 was an important opportunity to provide a more consistent delivery of neuromuscular care across NHS England.

People with multi-system diseases such as DMD should be able to see the professionals treating them in one place and those healthcare professionals should be in contact with each other about the care of their individual patients. This would typically be a tertiary neuromuscular centre that facilitates a multi-disciplinary team of healthcare professionals to work in tandem with a patient's Neuromuscular Consultant and Care Advisor.

Implications for lack of awareness of Standards of Care

DMD is a devastating and life-limiting disease with very few treatment options. For people with DMD in the UK, to not be receiving the highest standards of care is very serious. While rare, there are 2,500 people in the UK living with DMD and 110,000 living with rare, muscle-wasting conditions[xxxix].

For DMD and muscle-wasting conditions to be absent from NHS policies, guidance and plans is very serious for the lives of people with these conditions and their families.

It is particularly hard to understand for DMD as there are already agreed care standards established.

We have a set of policy recommendations for how this can be addressed and that would support dissemination and adoption of the guidelines.

"I am especially sad and angry when health professionals don't understand, and in my grandson's case, completely missed it. For three years he was misdiagnosed and treated for something completely different."
Grandparent of child with DMD

Recommendations for improving standards of DMD care across the UK healthcare system

- NICE to include the DMD Care UK clinical guidelines as part of its treatment guidelines for DMD.

- The establishment of clinically driven regional networks of key neuromuscular stakeholders including doctors, allied health professionals, established North Star neuromuscular clinical centres, NHS managers, commissioners, patients, carers and relatives of patients. The network would aim to develop and establish world class and equitable care for people of all ages living with rare neuromuscular conditions in the region, and to improve their quality of life and overall experience of NHS services. This would involve working with health and social care professionals to develop the appropriate services and increase the knowledge and skills required to manage these complex conditions. It would be modelled on the South West Neuromuscular Operational Delivery Network, which is the only Neuromuscular Operational Delivery Network in the UK.

- An NHS awareness campaign aimed at increasing understanding of DMD and associated care guidelines, targeted at healthcare professionals that deliver specialist care to DMD patients. This would include the development of three training modules on DMD and the care guidelines, one each for primary, secondary and tertiary care.

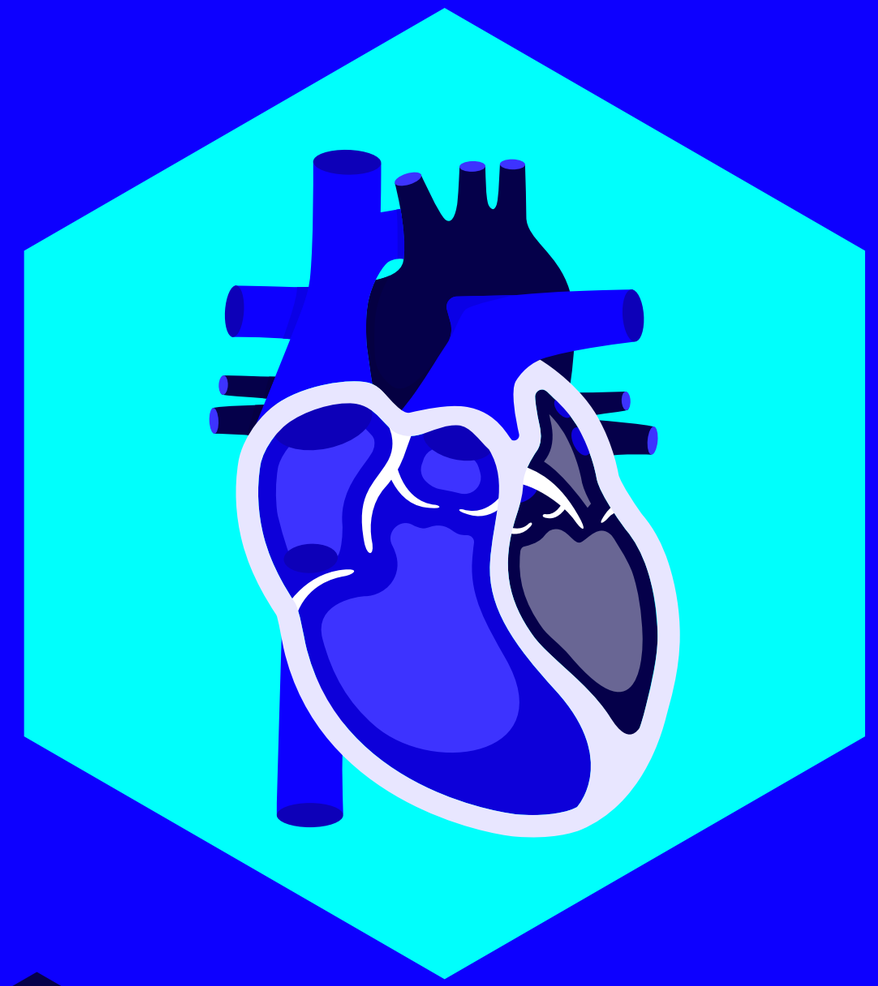
- Establish neuromuscular diseases as paediatric sub-specialist training for doctor and health care professionals to ensure the next generation of experts.

- Introduce neuromuscular care co-ordinators or care advisors at each specialist centre to support the delivery of care.

- NHS in England, Scotland, Wales and Northern Ireland to work with Duchenne UK on developing a Centre of Excellence badge that can be awarded to centres delivering DMD care and running DMD clinical research trials to a very high standard.

- We invite opportunities to collaborate on supporting the next generation of neuromuscular clinicians by providing fellowships to attract healthcare professionals into the specialism.

- We invite the NHS to work with us on a pilot for a new model of care, a virtual hospital ward. It would support the joining up of people involved in a patient's health and virtual care. It wouldn't replace in-patient care or in-person appointments but could complement it and allow different members of a patient's multidisciplinary care to work together.



Chapter 8: Seeking compassion - the importance of awareness in supporting patients

DMD is a devastating condition with few treatment options, woefully inadequate technology, and inconsistent adoption of care standards across the UK healthcare system.

Compounding these challenges is a lack of awareness or understanding of the disease which further adds stress and anxiety to families. It's very difficult to fight for your child if they have a condition that no one has heard of.

We've shown in section Chapter 7 that people living with DMD believe that even within the healthcare system there is low awareness of DMD. And there's certainly a lack of general awareness in society and education – which adds extra burden to their lives.

Lack of awareness among the public

Our survey of 200 people personally affected by DMD found that they are constantly having to explain to others what DMD actually is.

Almost a third of people (31%) surveyed reported having to explain it once, or more than once a week. 29% had to explain it every few weeks. It's hard enough living with DMD. Even harder to keep having to explain it to people and relive the challenges every time you do it.

This takes a huge toll on people with DMD and their families as it is not only frustrating to have to keep explaining something very painful but this is usually within the context of having to fight for help and support to which they are entitled but which hasn't been forthcoming.

This is what some of the people who took part in the survey told us about the impact of people not being aware or understanding DMD:

"It's very hard to get the right support for my son and that makes me very sad, frustrated and feels very lonely and difficult to have to constantly explain."
Parent of child with DMD

"Lack of empathy and understanding. People stare at you when in public, as you struggle to get out of a seat and walk around. It's okay when in a wheelchair, but when still walking it is incredibly noticeable and they look at you like there's something wrong with you. Lack of support, example includes falling over and being unstable on your feet. For my son who is 17 years old, they think he's drunk or something is very wrong. There is a lack of understanding or knowledge about muscular dystrophy in general."
Parent of teenager with DMD

"It can be upsetting for my son and me. There are a lot of stares when I help him to do things that another child his age can do. It can be emotional explaining it to different people."
Parent of child with DMD

"It's upsetting having to go over it all with people."
Parent of child with DMD

"Explaining it makes me relive the issues I felt when I found out my grandson had DMD."
Grandparent of child with DMD

"Because the diagnosis happens a long time before many noticeable physical differences are apparent, it can be difficult for people to grasp the seriousness and limitations of the condition."
Parent of child with DMD

"My little boy is only 18 months so I guess things aren't as impactful at this moment. However, it can be difficult in public, say at a play park. Where other parents are shocked to hear his age and realise he isn't mobile yet. Seeing other children his age, running around and playing in a different way can also be quite challenging. It's almost too much to say 'oh he has Duchenne' because you're then faced with more confusion, little understanding, awkwardness or upsetting questions."
Parent of child with DMD

Understanding of the severity of DMD

As well having to explain what DMD is, parents of young children with DMD struggle to explain the seriousness of the disease when their child is young and the impact of DMD isn't obvious or is only starting to be.

"In everyday life people assume my son is a normal six year old. For example, at the supermarket it can be stressful when he is grabbing things off all the shelves."
Parent of child with DMD

Lack of awareness at school

The survey found that people with DMD and their families were struggling with a lack of awareness of DMD at school, with only 10% thinking that those who worked in education had a good understanding of DMD.

This is important as many children with DMD have learning or behavioural difficulties and will have special educational needs and disabilities. DMD is not just a physical muscle wasting disease, it can impact the brain. It is critical that there is wider recognition and understanding of this, particularly among education professionals.

A lack of awareness and understanding among those working in a school also means that they don't have the knowledge to help the school population understand DMD. This leaves pupils with DMD vulnerable when at school, and many experience bullying by other pupils.

To support children with DMD and their families get the help they need from their school, we fund the Decipha 'Roadmap for Life' programme that provides free educational support.

"Our son's mental health is affected when his teachers don't understand or have any empathy even though they have had his neuromuscular care advisor go in to school to talk about DMD."
Parent of child with DMD

Case study

Decipha – helping parents advocate for their children at school

The Decipha 'Roadmap for Life; programme delivers:

- Support for families in finding solutions to problems facing the young person at home and in school.
- An assessment of the young person's skills and Person Centred Planning at school with the young person, their parents and school staff.
- A review of current interventions used in school or college and home with recommendations for best practice.
- Advocacy support for mediation and tribunal appeals.

Support for people with DMD and their families

Despite the impact of living with and loving someone with a rare disease of which there is very little awareness, people personally affected with DMD are rarely offered professional support to help with this impact. Only a quarter (27%) of people surveyed have been offered support, and even then it is inconsistent. There is no automatic offer of psychological support for parents when their child is diagnosed despite the trauma that parents experience when they are told that their child has DMD.

To understand the best interventions for people with DMD and their families, we funded research by Dr Isabella Vainieri and Andriani Papageorgiou of the Clinical Intervention and Practice Research Group, at the School of Psychology in the Faculty of Health and Medical Sciences at University of Surrey[xl].

Their research sought to understand what are the most common emotional and behavioural problems experienced by children and young men with DMD and the impact of such challenges on their daily life. They also explored the current support received for such challenges, and what kind of support is needed by these families in relation to such behavioural and emotional challenges. They found that:

- The children reported that the most prominent difficulties they are experiencing include; high levels of anxiety and worry, the feeling of being angry at themselves and annoyed with others and sleeping problems due to excessive negative thinking or sensory overstimulation leading to extreme tiredness.
- The impact of these difficulties was reported to be evident in the family's quality of life, specifically affecting the mental health of the parents, relationships with siblings and family outings.
- The current support received is deemed limited by parents and a "postcode lottery" that mainly focuses on physical abilities, whilst neglecting the emotional and behavioural difficulties. A big theme that came up was the never-ending battle with the system regarding receiving any form of psychological and educational support, and for those who managed to get help, the approach by the clinicians and different services was perceived as disjointed, confusing, and frustrating.
- Parents and children agreed that further psychoeducational support is needed within the family setting but also in school and clinic environments. Having a space where they can share experiences and be heard by others who are in similar situations and a clearer pathway for receiving psychological support.

We believe that it is possible to live a happy and fulfilling life with DMD and more children and young people than ever are planning positive futures as they grow into adulthood. However, it is clear that support is needed for people with DMD and their families and we fund a programme that aims to build the case for more psychosocial support for people with DMD and create guidelines for how their care should best be delivered.

Case study

Psychosocial support - developing guidelines on best practice and assessments for psychosocial care

Through our DMD Care UK programme, we, along with Duchenne Research Fund and Joining Jack, have funded a three-year research project that began in 2022 to carry out research and develop guidelines on best practice and assessments for psychosocial care in the UK.

The project will also keep track of the unmet need from the DMD community – looking at how many referrals for help are requested and the reasons for them. This information will be used to show the level of need to the NHS and make a better case to fund these kinds of posts in different centres around the UK.

This research will lead to a better understanding of the needs of people with DMD and their families and therefore more effective identification, support and treatment across the whole of the UK. We hope that this will reduce the impact of psychosocial risks in DMD and significantly improve quality of life.

The funding includes clinical psychologists who are now part of the neuromuscular multi-disciplinary teams at Newcastle and at University College London. Alongside the research in the project, they are also able to see patients at those centres as well as support and advise colleagues locally and nationally.

Two psychiatrists are also funded (in London and Exeter) and are exploring a more national approach. They hold open advice sessions for clinicians every month to help answer questions about psychiatric support. They accept online referrals from anywhere in the UK as well as in-person appointments for local patients.

Duchenne UK

Based on our research, and the experience of Decipha and our psychosocial programme, we have a set of policy recommendations for improving awareness and understanding of DMD, and support for people affected by it.

Recommendations for increasing awareness and understanding of DMD, and support for people affected by it

- The governments in each of the four nations should fund awareness campaigns to improve understanding of the range of neuromuscular conditions. This should be led by input from the neuromuscular community, with the relevant government department and public health body in each nation, in close collaboration with local health systems, clinicians, and other relevant specialists.
- The awareness campaigns should be targeted at healthcare and education professionals, along with materials published online and in print for the public.
- Parents and carers should be offered emergency trauma support following the diagnosis of their child with DMD.
- People with DMD and their parents and carers should be offered psychosocial support from someone trained in DMD when they enter each new stage of DMD.

Chapter 9: Conclusion

This report reveals an improving picture of DMD care, treatment and support across the UK. However, the opportunities for further advancement are significant. It is crucial that they are not missed because the impact they could have on the lives of people with DMD is immense.

Healthcare professionals and policy-makers alike have the power to make a dramatic improvement in the lives of people with DMD across the country. In this report we have set out what we and other experts believe needs to change.

The landscape is changing, and this report is the beginning rather than the conclusion of a conversation about DMD. There is much to do if the recommendations set out here are to become a reality.

Many of these recommendations for DMD are also transferable to other rare diseases and neuromuscular diseases.

If you or your organisation would like to discuss our recommendations or any of the issues outlined in this report, please get in touch by emailing **communications@duchenneuk.org**. As we move forward, we want to work with as many people and organisations as possible to make excellent DMD care, treatment and support in this country a reality.

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