

# Our Impact in 2021



**66** Duchenne UK has given us light in our darkest hour **99** DMD parent

### Contents

- About Duchenne UK 3
- Letter from our founders 4
- Discovering new treatments for DMD 5
- Advancing gene therapy 6
- Trialling new treatments Clinical trial success! 7
- Changing Charlie's Future 8
- Harnessing existing treatments for DMD Tamoxifen 9
- Improving clinical trial data 10
- Driving innovative gene editing research 11
- Improving quality of life 12
- Changing the landscape 13
- The DMD Hub expanding access to clinical trials 14
- Project HERCULES continues its groundbreaking work 15
- Making our voices heard 16
- Ensuring the best care for all patients DMD Care UK 17
- Here for every family affected by DMD 19
- 21 Supporting young people growing up with DMD
- Building the DreamChair 22
- Thank you to our Duchenne Dashers and community fundraisers 23
- How we raise and spend our funds 25
- Thank you to our supporters 26



### About Duchenne UK

Duchenne muscular dystrophy (DMD) is a devastating musclewasting disease. It is the most common and severe form of muscular dystrophy. Usually diagnosed in early childhood, it mainly affects boys. Started by families affected by the disease, Duchenne UK has one clear aim – to end Duchenne.

#### What we do

Duchenne UK is going further and faster to fight DMD and end its devastating impact.

As the leading DMD charity in the UK, we're funding ground-breaking scientific research to find transformative therapies and technologies. We aim to not only improve quality of life, but stop the disease in its tracks.

We connect leading researchers with the pharmaceutical industry, the NHS, and patients to accelerate progress and bring treatments from the lab to those who desperately need them.

Thanks to the army of supporters who have joined our fight, we've made incredible progress already.

Our heart and soul, and reason for carrying on, are our boys and men, and rare girls, living with DMD.

## Join us, and together, we will end Duchenne.



Research to find new treatments

Accelerating access to treatments



Here for every family

## Letter from our founders

#### Dear friends,

It's been nine years since Alex and I first met. Both of our sons had recently been diagnosed with DMD and we were overwhelmed. Overwhelmed and terrified of a future that neither of us had planned or were remotely prepared for.

Over time, we rebuilt. And we have created not just a charity. But a movement: a communal endeavour to change the world - in the only way it can be changed. With grit. Determination. Resilience. Courage. And a fearless ambition to attain the unattainable.

We create. And we innovate. And you will see real life examples of our ground-breaking work throughout this impact report.

Charities like ours fund half of all medical research in the UK – to the tune of £1.7billion. These past two years have thrown into sharp relief the absolute necessity of medical research. Today, 1 in 4 people choose to support medical research charities like ours – and for that we are so grateful.

And as we enter the era of 'levelling up' and integrated care, Duchenne UK is pushing at the forefront of what is possible.

Through our flagship DMD Hub programme, we are creating a sustainable infrastructure to make sure all DMD patients can access and participate in clinical research.

You will read about our innovative DMD Care UK programme, which is putting research at the heart of care to make sure all DMD patients, regardless of where they live, will receive the best care – this has never been done before. In fact, a lot of what we do has never been done before! Look at the work we did to bring together competing pharmaceutical companies to work together in the space of health technology, creating the award-winning global collaboration Project HERCULES.

Duchenne

The research we have funded is bearing fruit – you will read about the successful Phase 2 trial results of Vamorolone, a steroid alternative that we funded in 2014.

And the exciting work in gene therapy continues, with the first DMD patient being dosed in a gene therapy trial in the UK this year!

We're going to do it **faster**, because this generation of people living with DMD can't wait.

The only way we can achieve this is **together**. We create a network of mutual support for families. We collaborate with scientists and the pharmaceutical industry, bringing them together to share knowledge. And we work with health services so that patients get the care they need.

So, thank you for supporting us on our journey, and our fight to end Duchenne.

**Emily Reuben and Alex Johnson** Founders of Duchenne UK Research

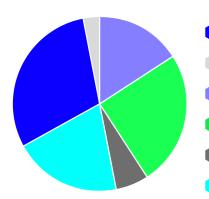


## Discovering new treatments for DMD

We are funding the most cutting-edge science to find life-changing DMD treatments. The following represents just some of the progress this year towards our ultimate goal: ending DMD's devastating impact for everyone affected.

We look forward to launching a new research strategy in 2022 in response to a changing landscape with increasing opportunities to innovate.

#### Our ongoing research in 2021



- **Repurposing drugs** Technology to improve lives Dystrophin replacement
- Improving muscle function
- Nutraceuticals
- Profiling the disease

New studies funded: hydrotherapy clinical trial, heart protection follow-up study, three projects focussed on overcoming the challenges of gene therapy delivery

More than £9M invested in **DMD** research to date

£2.3M invested in repurposing existing treatments for DMD

£3.1M invested in research into treating the root cause of disease to date. including £1.7M invested in gene therapy

## Advancing gene therapy

Gene therapy is the key that could unlock a future where children with DMD have muscles that stay strong as they grow, so they can live near normal lives.

It works by delivering new genetic material to muscle cells. This allows the body to produce a dystrophin-like protein which could help muscles function normally.

Duchenne UK invested in pre-clinical gene therapy studies and together with clinicians, industry and the NHS, has fought to bring those trials to the UK. We are so proud to be able to tell you about what we are doing in this area:

• FIRST BOY DOSED... In a clinical trial of Pfizer's gene therapy treatment in Newcastle, one of the DMD Hub sites. The phase 3 trial will look at the efficacy and safety of it in boys who are able to walk, aged between 4 and 7. This is a huge milestone in the treatment of DMD, and our support has been vital to making it happen. We invested in critical preclinical research to help pave the way to the clinic, and through the DMD Hub, we've invested in the infrastructure needed to make the UK 'gene therapy' ready.

• VIRAL VECTOR RESEARCH... Harmless viruses, called AAVs, are being used as a vector or carrier to get gene therapies into patients' bodies, but in some patients they can be blocked by antibodies in their immune system. This precludes these patients from being treated with these therapies. Together with US charity PPMD, we have provided funding of £260,000 for research into preventing this immune response. If successful, it could mean that more patients would be able to receive the treatment, and there would be potential for booster treatments.

• LIPID DELIVERY RESEARCH... We have given more than £280,000 to fund early-stage research into the use of lipid nanoparticle technology, successfully used in the recent Moderna and Pfizer/BioNTech COVID-19 vaccines, for DMD gene therapy. It is thought that these naturally occurring fatty molecules could carry genetic material, and be less likely to be rejected by the body than viral vectors.

ENGAGING THE PHARMACEUTICAL INDUSTRY... We remain determined to harness the benefits of gene therapy for DMD. To that end we are bringing together the regulators MHRA and NICE, NHS commissioners, clinicians and patient representatives for an industry day early in 2022 to ensure these transformative treatments can get to patients quickly.





## Trialling new treatments – Clinical trial success!

While driving forward our ambition to end Duchenne, we are also pushing hard to improve the lives of people living with the disease today.

Duchenne UK is delighted to say that a trial into a potential steroid substitute, Vamorolone, has shown positive results. While steroids keep children physically active for longer, they have serious side effects such as poor bone health, delayed puberty and weight gain. We were pivotal in providing funding for a successful phase 1 trial, and are delighted to report this year that a second phase trial run by Santhera Pharmaceuticals and ReveraGen BioPharma has also shown good results.

It still needs approval from regulators, but we are pushing hard for it to be made eligible for the Early Access to Medicine Scheme so people living with DMD can receive its benefits faster.

## Changing Charlie's Future

"Charlie loves music, dancing, dinosaurs, superheroes and is often found wearing a variety of dressing up costumes and hats! He is a happy little soul and, at present, unless you know to look for a few subtle differences, Charlie is just a normal little boy.

However, in January 2021, after an agonising Christmas and New Year waiting for test results, he was diagnosed with Duchenne muscular dystrophy and our universe imploded.

I will never forget the utter blackness and grief we felt. Our beautiful little boy had just had his future stolen from him by some cruel twist of genetics. Everything you hope and dream for your child, your family, just ripped from you.

In the dark days and weeks that followed, we would read what we could about DMD in our stronger moments, and Duchenne UK began to offer a shard of light. A factual, pragmatic, streamlined and hugely pro-active charity with one clear aim – to end Duchenne. We drew strength from reading about all that has been achieved since two hugely inspiring mothers of Duchenne boys set it up nearly ten years ago and all they aim to achieve.

Duchenne UK has given us light in our darkest hours and real hope that the dawn will soon break on more effective treatment for all those affected by Duchenne." Jennifer, Charlie's mum







for DMD – Tamoxifen progress

Using medicines already approved for other conditions to treat DMD is an approach being pioneered by Duchenne UK. It's known as drugrepurposing and is attractive because it means potential medicines can be tested in clinical trials in far shorter timeframes.

Duchenne UK has funded a clinical trial into the use of Tamoxifen – a breast cancer drug – for people with DMD. This year, this trial, known as TAM-DMD, has reported the first preliminary data. It showed that Tamoxifen is safe to use as a treatment for DMD. However, the difference between the placebo group, which did not receive the drug, and the group taking Tamoxifen was not as marked as we'd hoped. Overall, patients in the Tamoxifen treated group showed better outcomes, and there was a trend for less

9 Duchenne UK

disease progression in the Tamoxifen group, but the children in the placebo group also declined less than would normally have been expected.

In collaboration with partner organisations, we are continuing to analyse the data from the trial to determine the effectiveness of Tamoxifen in DMD. We will share any updates as soon as we can.

All findings from clinical trials are important to help deepen our understanding. We are incredibly grateful to the boys and families who took part. Clinical trials are complex - but we are committed to exploring every option for children with DMD, because we know that this is how we can change their lives for the better.

## Improving clinical trial data

Families living with DMD are fighting against time, so we want to make sure every trial we invest in helps in our journey to find effective treatments. We are funding several projects to improve clinical trial data and how we collect it, so that we can push forward the most promising research and enable more successful trials in DMD.

#### Developing less invasive ways to measure disease progression

We are working with Duke University and other clinical partners in the US and Newcastle NHS Trust in the UK to develop non-invasive and less expensive ways of measuring how DMD progresses. The aim is to try to develop a urine test which shows the state of children's muscles. This could prevent the need for painful muscle biopsies and expensive scans.

Thank you to our partners MDA and PPMD who are part funding this research with us.

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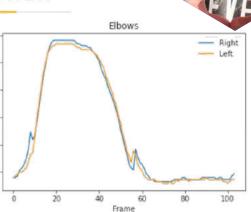
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#### LIFT MOTION



#### • Using digital technology to improve assessment of disease progression.

Duchenne UK is working with tech company Aparito to develop more sensitive measures of DMD progression that could be used in clinical trials. Currently, assessments like the North Star Ambulatory Assessment and the 6-minute walk test are used to measure whether treatments are slowing down the progression of the condition. However, Aparito's approach uses video technology and powerful analytics, capturing more detailed, accurate and objective data while the patient is going about their everyday life. We are working with Aparito and other partners to validate this approach and drive its adoption in clinical trials.



- Vertical location of elbows throughout the video. Arms lifted together, with continuous movement up and then down

## Driving innovative gene editing research

Gene editing is a promising approach that could lead to the next generation of innovative treatments for DMD. These therapies aim to correct the genetic faults which cause DMD by editing DNA sequences, allowing the production of a shorter but functional form of the dystrophin protein.

We supported US biotech company, Myogene Bio, to secure a \$3,4M grant to carry out pivotal preclinical research to test this approach in DMD. This work will help Myogene to make a case with the FDA, the US drugs regulation agency, to take the therapy into clinical trials.

**66** By enabling early studies into gene editing, we are helping to accelerate its development and paving the way for the therapy to arrive in clinics.

> Dr Alessandra Gaeta, Director of **Research at Duchenne UK**



## Improving quality of life

Advances in medicines are important, but we studies into its benefits, so health providers are believe we can also make an impact on quality reluctant to pay for it. So we've funded a new of life through other types of interventions. study from experts at the Royal Preston Hospital and Manchester Metropolitan University involving Thanks to better care, people with DMD are 44 boys and young men, both ambulant and living longer, but more can be done. That's why non-ambulant, to gather evidence. We hope that we've invested almost £300,000 into researching the findings will help more people living with the benefits of hydrotherapy. DMD access this important therapy.

Hydrotherapy means using water for health benefits. For people with DMD, this often involves doing gentle exercises in the buoyant environment of a special pool which relieves stress on the muscles. This not only helps the muscles, but helps people to enjoy the many benefits of exercise like stress relief, better sleep, and improved wellbeing.

Hydrotherapy is often recommended as a treatment for DMD, but there are few reliable



Acceleratating Research

## Changing the landscape

The research we fund is only one part of getting new treatments to the people that need them. We need high-quality data, the right infrastructure, and the best care, to make sure that all DMD patients can access the benefits of research. We are committed to making this happen by collaborating with the NHS, regulators, clinics and the pharmaceutical industry. And we are making huge strides:



## The DMD Hub – expanding access to clinical trials

We are so proud of the pioneering work of the DMD Hub. We want to make it possible for anyone living with DMD to take part in clinical trials if they want to, allowing them to access treatments they wouldn't otherwise receive.

In 2015, when we created the concept of the DMD Hub there just weren't the facilities to make this happen. Each year, we extend our outposts and staff to make this a reality. Thanks to the DMD Hub, 437 more boys have been recruited onto clinical trials so far. 11 hospitals are now running 10 trials into potential treatments for DMD. This year we are delighted to have:

Extended the contract for the DMD Hub Manager, Emma Heslop, for another four years. She is the vital contact point between clinicians, industry partners and patients. She helps select sites and recruit patients and makes sure best practice is shared between Hub sites

• Welcomed Katie Pegg and Philip Cammish to join the core Hub team. They will be looking at improving the recruitment of patients to trials, and the training of Hub staff to prepare for more gene therapy trials in the UK.

 Funded five new posts across the different Hub sites, employing people like research nursers and physiotherapists to help run clinical trials.

£3M committed to the DMD Hub since 2015

> 34 posts funded

437 more boys on clinical trials since 2015

> **11 DMD** Hub sites

20 trials ongoing in the UK, with 8 new trials opening in 2021

> 9 more trials in scoping

Acceleratating Research

## Project HERCULES continues its groundbreaking work

## HERCULES

DMD-QoL

Ducher UK

The UK is a leading innovator of new medicines and research. Yet when new treatments are approved, patients in the UK are often some of the last to access them due to challenges in reaching agreement on the cost of new medicines.

According to the Association of British Pharmaceutical Industry (ABPI), patients in France and Germany are five times more likely to receive a new medicine than patients in the UK.

This is why we launched Project HERCULES, so that companies developing treatments for DMD have the evidence they need to speed up these negotiations, giving their medicines the greatest chance of being paid for.

Our groundbreaking international collaboration has brought pharmaceutical companies together with leading academics, clinicians and patient organisations to develop the evidence needed to get newly approved treatments to patients as quickly as possible.



The DMD-QoL is a new quality of life measure developed by the University of Sheffield in collaboration with Project HERCULES and involving patients at each stage of its development. This improves on existing measures, which didn't capture the reality of living with DMD and aspects of most importance to patients and families. This measure can be used when assessing the impact of new treatments, giving effective treatments a better chance at approval.

#### What's next for Project HERCULES?

We continue to collaborate with clinicians, pharmaceutical companies and academics to deepen our knowledge about DMD and address existing gaps. Two areas of focus going forward will be the impact of long-term steroids use, and assessing the impact of DMD on families and caregivers.

## Making our voices heard

We work at the highest level with regulators and industry leaders to speed up the drug development process and create meaningful outcomes for patients. Because we want results now. We WILL have our voices heard, and we will use our voices in the most effective way we can.

We are excited to announce that our CEO Emily Reuben became a member of the



#### Patient Advisory Council of the

pharmaceutical trade body The Association of the British Pharmaceutical Industry. She is one of eight leaders from across the health and charity sectors to give advice to some of the leading pharmaceutical companies in the world. She will be championing DMD among the people with real power to change the course of this condition.



## Ensuring the best care for all patients - DMD Care UK

It is our mission that every patient with DMD – wherever they live in the UK – should be able to access the same quality of care, and that that level of care should be excellent.

We have brought together the DMD community of patients and families with neuromuscular experts and specialist clinicians to try to make this a reality with the DMD Care UK project.

DMD Care UK is such an important project for raising and harmonising standards of care for ALL people in the UK living with DMD. It works because we are a collaboration between neuromuscular expert centres, specialist clinicians and the patient community - we are all working towards the same goal!
Dr Michela Guglieri, John Walton Muscular Dystrophy Research Centre in Newcastle; PI of DMD Care UK

We have been carrying out a comprehensive review to get a benchmark of how six areas are managed across the country. Psychosocial

Bone and endocrine health





#### Psychosocial care

We want every patient and their family members to receive the appropriate psychosocial evaluation, care and treatment throughout their lives. We are therefore partnering with the Duchenne Research Fund who awarded a grant of £640,000. This will fund two research clinical psychologists posts, a neuropsychiatrist and additional support for three years at Newcastle University and the National Hospital for Neurology and Neurosurgery in London. They will make recommendations about how to improve this area of care.

I am so thankful for Duchenne UK's webinar on stress dosing of steroids. This critical part of caring for our boys when they are poorly had never been mentioned to me before by any of Tom's consultants and it is only because of the new recommendations that I was promptly able to get a potentially life-saving hydrocortisone pack. S Rebecca Burnett, DMD parent

#### Bone and endocrine health

The endocrine system is a network of glands in the body which make the hormones that help us all function. We are working with the British Society of Paediatric Endocrinology and Diabetes to find an effective way to monitor the health of the endocrine system, and also bone health. Steroids have a noticeable effect on these areas so we are producing guidance for families and health professionals.

One particularly challenging area is delayed puberty, so we have produced leaflets giving advice on this subject.

We have also produced detailed advice on steroid stress dosing – when extra steroids are needed for patients who become ill or injured. We are working on updating checklists in clinics across the country to educate parents and medical staff.

Our steroid wristbands were developed to alert medical staff in Accident and Emergency departments that the wearer needs special treatment. So far 240 wristbands have been ordered by families. We are also developing an emergency care app, which is currently in the testing phase.

Educating parents is key so we held a wellattended webinar on steroid stress dosing, informing parents and caregivers on what to do if their child is unwell and injured while taking steroids.



Here for every family affected by DMD



This year, we...

Distributed 76 DMD Family Folders to newly diagnosed parents Launched a new guide to adolescence and adulthood – 158 orders and 167 digital downloads so far

Welcomed 100 parents and caregivers to our Parent Information Day - 100% agreed/strongly agreed that they were better informed to make decisions about their child

Ran four new webinars, which have been viewed 991 times

C Thank you again for holding an amazing, informative day. Being in a room with people going through the same thing is so empowering and uplifting. 
Parent Information Day 2021 attendee

While we fight for treatments, our children need to be able to live their lives and have the same opportunities as their peers. Many children and young people with DMD have specific educational needs, so we back projects to help them at school and beyond.

We continue to provide funding for Decipha, an educational organisation which offers invaluable support to families. Decipha's RoadMap for Life programme has helped hundreds of families navigate the tricky waters of their Education and Health Care Plans (EHCPs), supporting appeals and working with SENCos (special educational needs coordinators) and teachers.

People with DMD deserve the opportunity to live independent lives. This year we have funded a project with Pathfinders Neuromuscular Alliance, a charity run by and for adults with muscle-weakening conditions, to look at what stops them from doing so. We discovered that many adults with DMD were unable to live independently because of the shortage of accessible housing. We are now helping

19 Duchenne UK

Pathfinders work with campaigning groups in Parliament to address this, and aim to improve the information and support in this area.

While life has been conducted remotely for many months during the pandemic – we reached out to the DMD community through webinars. These allowed us to give parents access to vital information and support structures. This year, topics covered included gene therapy, home-schooling during lockdown and the COVID-19 vaccines.

We know the value of a friendly face when your world is collapsing after a diagnosis of DMD, and the power of arming yourself with information - that's why we continue to run our Parent Information Days. Thank you to everyone who came to our event this year in Manchester. Our talks on the condition, wellbeing for children and families, and advancements in research were very well received, and we were pleased to run a session for dads and male carers for the first time. Support

## Supporting young people growing up with DMD

Better treatments and support mean people with DMD are living longer. This is fantastic and a key aim of the charity. But quality of life is as important as longevity. We want our youngsters to dream big and fulfil these dreams – like any other child. The teenage years – when children transition to adults – can be overwhelming for DMD families. So this year – following on from the huge success of our Family Folder for newly diagnosed families - we have launched the DMD Adolescence Folder, a comprehensive new guide to support families of teens and young adults with DMD. It provides help with life skills, education, hobbies and relationships. Already, more than 160 copies have been downloaded and 150 print versions have been ordered. Our next step is to get this valuable resource out to all clinicians who treat DMD families.



It's already becoming my 'go-to' source and my 'to do' list is growing the more I read (in a positive way!). It's also a fantastic resource to look further into the future to help consider the longer term. As they say 'knowledge is power' and I can already see this being added to my new DMD bible collection! Thank you Duchenne UK and all who contributed to create such a great resource for parents. \$\$

Kerry, mum to George, age 13

## Building the DreamChair

## F1 Innovators join the wheelchair project

Williams Advanced Engineering, the technology spin off of Williams F1, have joined forces with Duchenne UK and Whizz-Kidz to input on the design of our dream wheelchair.

Paul McNamara, WAE Technical Director:

"We are delighted to be part of this project and welcome the opportunity to contribute to a programme that will be making a genuine difference to people's lives. Our experience with past projects has shown us the importance of prioritising the needs of the individual and we look forward to delivering some new and innovative design ideas."

We've always believed that it isn't DMD that disables our children. It's the barriers that society puts up that prevents wheelchair users from fully participating. Take Cop26 in Glasgow, 2021, when an Israeli minister was unable to access the conference venue because she was in a wheelchair. If this was allowed to happen to a senior government figure attending a global conference with the eyes of the world upon us, what chance do our children have and the thousands of other wheelchair users in this country!

That's why we partnered with Whizz-Kidz and PPL to develop a wheelchair to overcome some of these barriers. We unveiled the chair at our Parent Information Day and the response was incredible.



I really loved the wheelchair. I'd never seen anything like it before and I think I would actually enjoy using it. I can't wait to have one myself! ??

Joey, age 16



## Thank you to our **Duchenne Dashers**

#### **Duchenne Dash:** Around the World

While COVID once again prevented us from cycling from London to Paris, we didn't let this stop us. 164 Duchenne Dashers joined us at Goodwood Motor Circuit in July 2021 where they collectively cycled 40,000KM – the total distance around the world!



#### **Duchenne Dash AT HOME**

We brought back the Dash AT HOME so that anyone, anywhere, could set their own KM challenge and achieve it in their own way. 175 people walked, ran, biked, or wheeled in their local area over six weeks to raise an amazing £60K!

## Duchenne DASH

## £552,000 raised!



## And to our community of fundraisers!



#### Start Stop

We asked you to start something new this October and help bring a stop to DMD, with challenges ranging from 5K a day runs to learning to

#### Donate a day

We asked you to swap your daily coffee, takeaway or commute for a donation to Duchenne UK this February, and we really felt the love!





#### Duchenneber

We dedicated the whole month of December to raising awareness and funds for DMD, and the community shared some wonderful festive artwork with us

#### **Family & Friends** Funds

Our Family & Friends Funds took on challenges, hosted events, sold their creations and much more to raise money for Duchenne UK

## More than raised!

dance on rollerskates!



#### Captain Tom 100

To commemorate what would have been Captain Tom's 100th birthday, fundraisers across the UK set their own '100' based challenges in April. We were delighted by your innovative challenges for Duchenne UK, including 75-year-old Alan who walked the 1.3 mile Southend Pier 100 times!

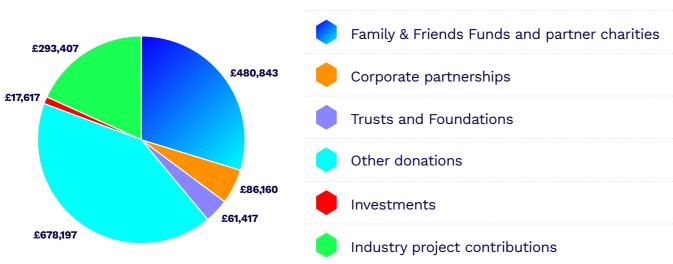
# £438,000



...and many more brilliant fundraisers joined our mission to end Duchenne!

## How we raise and spend our funds

As an ambitious and dynamic charity, we work to ensure every pound you donate makes a difference in our mission to end Duchenne, whether by funding research or helping us to raise even more.



#### How we raise our funds

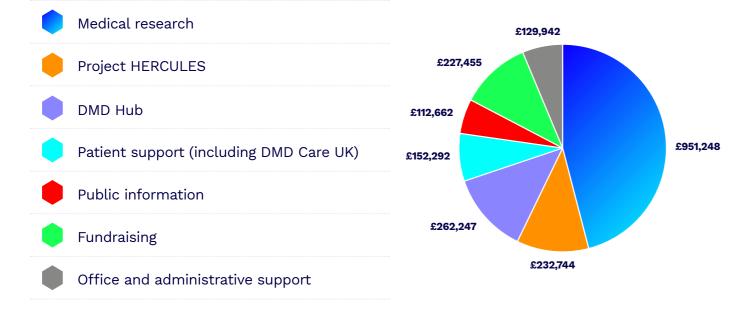
## Thank you to our supporters

Thank you to every individual, family, company and charity who supported our mission this year, including but not limited to:

#### Family & Friends Funds and partner charities

Access to Life Action for Arvin Action for Zach Alex's Wish Archie's Army Archie's March **Backing Jack** Ben vs Duchenne Changing Charlie's Future Chasing Connor's Cure Cure4George Defending William Against DMD Duchenne Research Fund Joe's Journey to end Duchenne Edward Steam Team Elliot's Endeavours

#### How we spend our funds



#### Funders and corporate supporters

Astellas Bunzl Clarke Wilmott **Courier Post Logistics** Foodbuy Franke Coffee Systems UK Freightliner FUEL10K Glaceau Smartwater

Family Saul Following Felix For Felix Help Harry Helping Hayden Henry's Hurdles Hope for Gabriel Hope for Harry Jack's Aim Jack's Mission Jacobi's Wish Jayden's Army Joining Jack Lifting Louis Love for Leon

Lygo Family Fund Mission Jensen Moving Muscles for Marcus Muscles for Mitchell Pathfinders Neuromuscular Alliance Project GO Ralphy's Fund Smile with Shiv Standing with Jack Strength for Stanley Team Dex Team Felix Team Oscar Together for Rhys William's Fund

**ICE Trading** Invesco James Tudor Foundation LCH Little Steps LXA Marshall Wace Asset Management Memery Crystal Pearson **PTC** Therapeutics

**RC** Components ResMed RPC Sale Sharks Saracens Strategic Dimensions **UCC** Coffee Wheelhouse Commercial WPP Health



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