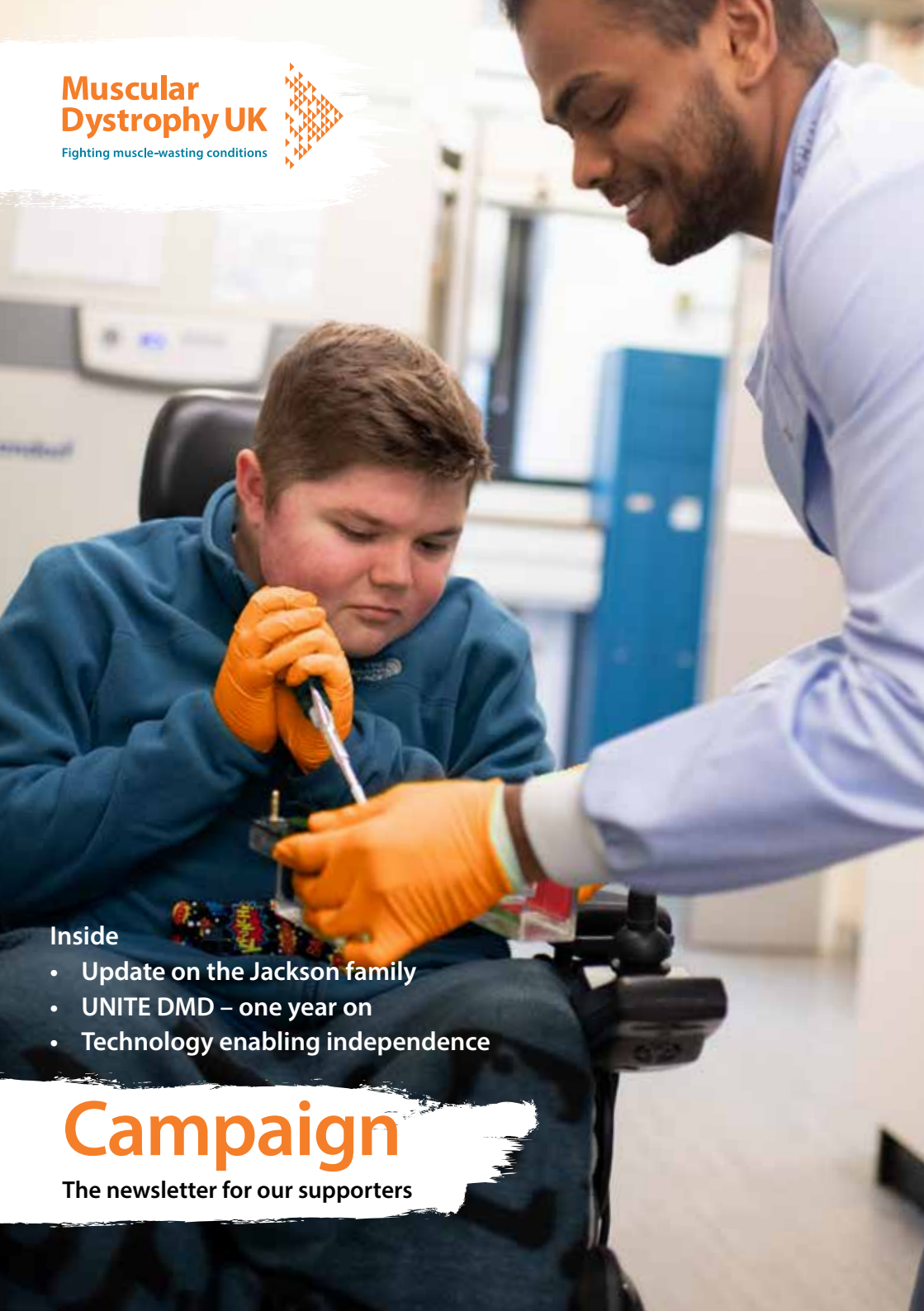
Muscular Dystrophy UK

Fighting muscle-wasting conditions



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Campaign

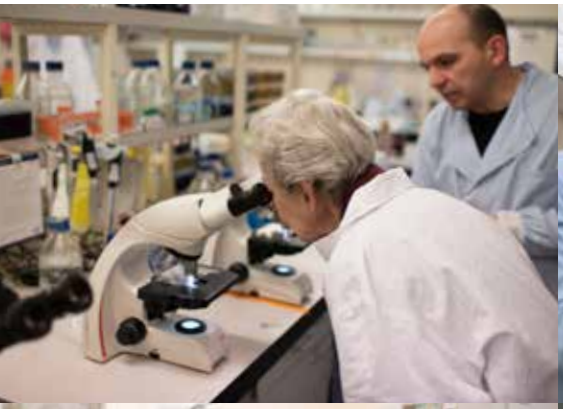
The newsletter for our supporters

Gifts in Wills Week event

In February, we invited a group of our supporters, who have chosen to leave a gift in their Will, to visit the research laboratory at Royal Holloway University. We wanted them to see first-hand how the charity invests in research and how their gifts might be used in the future.

They heard presentations by Professor George Dickson, who has studied gene therapy for over 20 years, and Dr Linda Popplewell, who is in the last year of her five-year MDUK-funded Lectureship. Supporters were given a tour of the laboratory, got to try out some of the equipment in the lab and speak to the team of researchers working there.

Gifts in Wills fund 30 percent of everything we do. Find out more about leaving a gift in your Will at www.musculardystrophyuk.org/giftsinwills



Welcome

As the new president of Muscular Dystrophy UK, I'd like to welcome you to this spring edition of *Campaign* – the newsletter for you, our valued supporters. I'm delighted and honoured to be continuing the excellent work of Sue Barker who was a dedicated and active President of the charity for 14 years.

There are some exciting articles in this edition. Sarah Jackson, mum of six-year-old Louis, wrote to you before Christmas, and since then we've been overwhelmed with your generosity and kind words of support for her family. Thank you ever so much. On p4 and p5, you can read an update on the family and find out how they've been getting on.

You'll also read on p10 and p11 about the incredible assistive technology that we are able to fund thanks to supporters like you. You're helping to give greater independence to the individuals and families we support.

I hope you enjoy reading this edition of *Campaign*. We're so grateful for all you do to support MDUK.

Gabby Logan
President, Muscular Dystrophy UK

Our vision

A world with effective treatments and cures for all muscle-wasting conditions and no limits in life for individuals and families affected.

Campaign newsletter for supporters of Muscular Dystrophy UK, written and designed entirely in-house.

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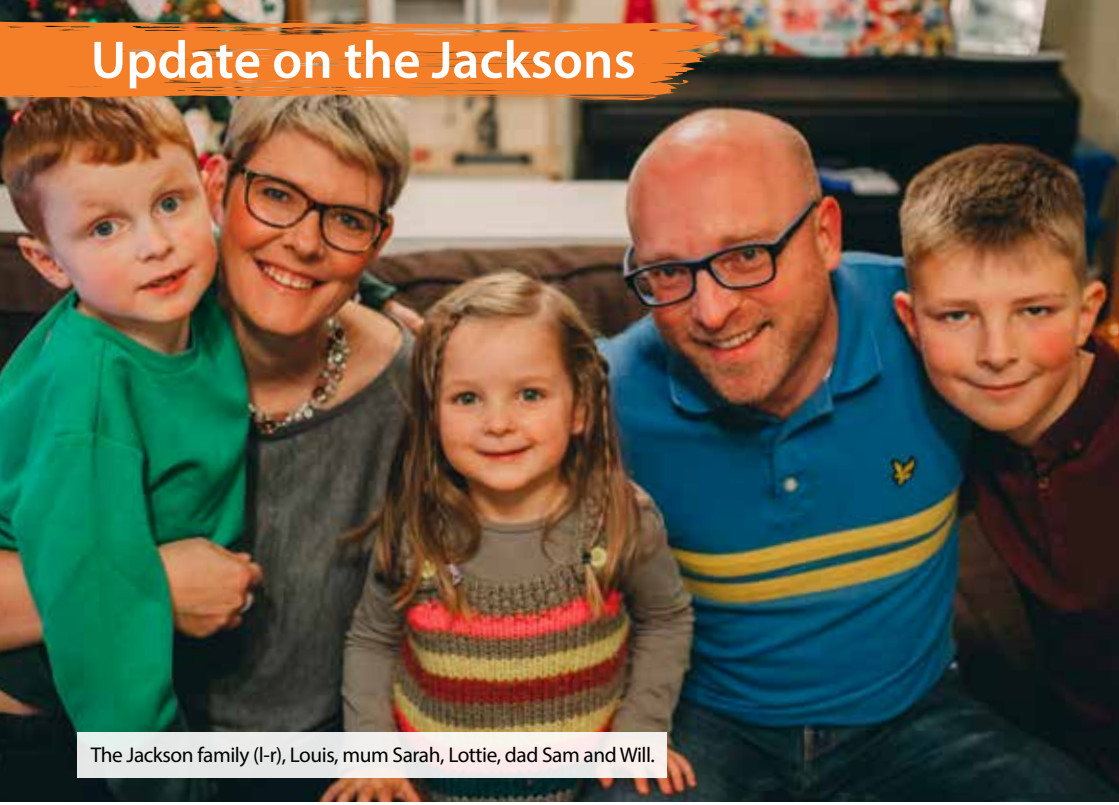
On the cover: Luke Taylor tries some of the equipment in the lab at Royal Holloway University.

Update on the Jacksons



The Jackson family (l-r), Louis, mum Sarah, Lottie, dad Sam and Will.

Thank you to all our supporters who gave so generously to our Christmas appeal. Mum Sarah and the whole Jackson family have been so moved by the outpouring of support they received following her letter, and the heartfelt messages that were sent. Your kind donations have now raised an incredible £31,000!

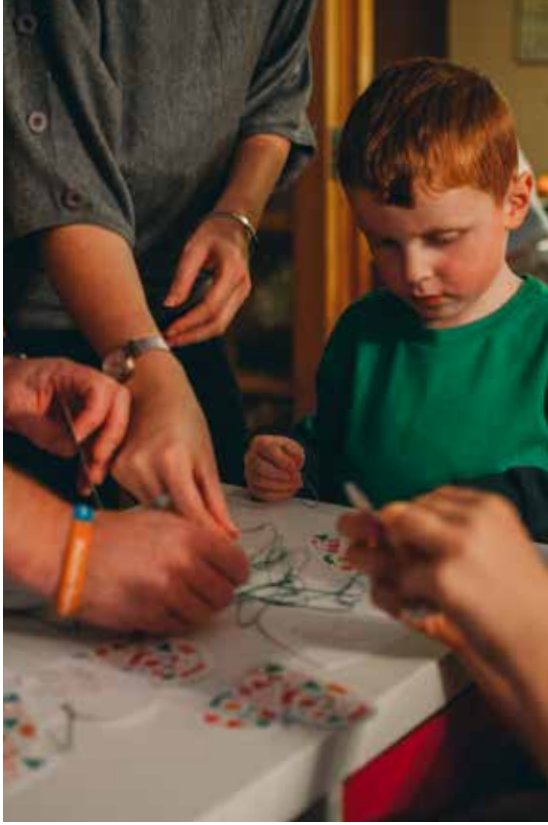
Three years ago, Sarah and her husband Sam found out their son, Louis (now seven), had Becker muscular dystrophy.

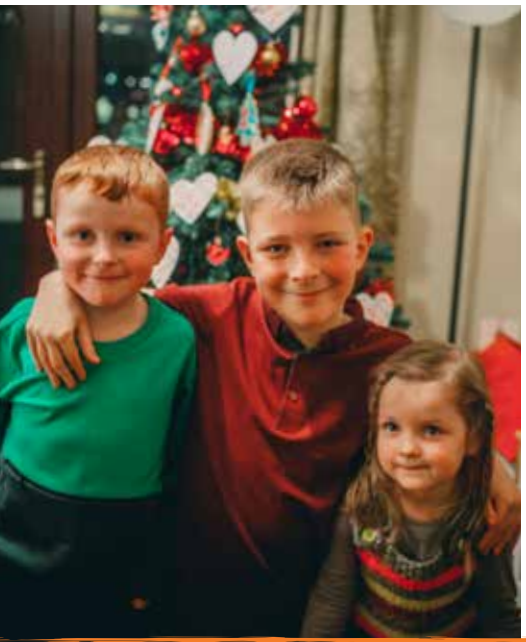
When Sarah wrote to you, she shared her experience since this diagnosis: the overwhelming sense of worry she feels for Louis and the uncertainty about what lies ahead for him as his condition progresses.

Since Sarah's letter, the family have been successful in getting a Blue Badge – a parking permit that allows the drivers of passengers with severe mobility problems to park close to where they need to go. One of Muscular Dystrophy UK's Care and Advocacy team helped them through the application process, and getting the badge has had a big impact:

“Having a Blue Badge has been absolutely invaluable and has enabled us to do so much more. Knowing we can park at a venue means Louis can use his energy there having fun rather than just walking to it,” says Sarah.

The Jackson family have also taken on a fantastic fundraising effort. They got many of their local schools and nurseries to take part in our annual national fundraiser, Go Orange for a Day, to raise money for MDUK, and even persuaded their town to light up the town hall and fire station in orange for the day. The family were delighted that so many people showed their support and joined their fight to beat muscle-wasting conditions, raising almost £2,500!





Your kind support of MDUK could have a life-changing impact on families facing a muscle-wasting diagnosis like the Jacksons. With your help, MDUK can make sure they don’t feel alone and have all the information and support they need.

If you haven’t read the Jacksons’ story, please visit: www.musculardystrophyuk.org/christmas

Top image: Louis looks at messages of support.
Left: Louis with big brother Will and little sister Lottie.

Investing in gene therapy UNITE-DMD – a year on

With your valued and continued support, MDUK is proud to be a long-standing supporter of research into gene therapy. This technology has advanced significantly over the years and we're pushing it forward to test its safety in people living with muscle-wasting conditions in the UK.

An international collaboration, UNITE-DMD is a four-year project that we're funding in partnership with Action Duchenne and the French Muscular Dystrophy Association (AFM-Téléthon). One year into the project, we thought we'd bring you an update on its progress.

Background

This project will help to refine and improve gene therapy generally, and though it focuses on Duchenne muscular dystrophy, it will be helpful for other muscle-wasting conditions in the future.

The overall aim with gene therapy for Duchenne is to replace the faulty dystrophin gene with a healthy copy. But the dystrophin gene is one of the largest genes in our bodies, which makes it difficult to incorporate into a gene therapy. Scientists have managed to get around this by creating smaller but similar genes called microdystrophins.

The UNITE-DMD researchers have developed a microdystrophin gene called MD1. This has been packaged into an adeno-associated virus (AAV), which acts like a delivery vehicle. Once injected into the body, the AAV carries the gene into the muscle cells.

Following promising results in animal models, the AAV8-MD1 gene therapy will now be tested in boys with Duchenne as part of the UNITE-DMD trial and will focus on evaluating the safety of this technique.

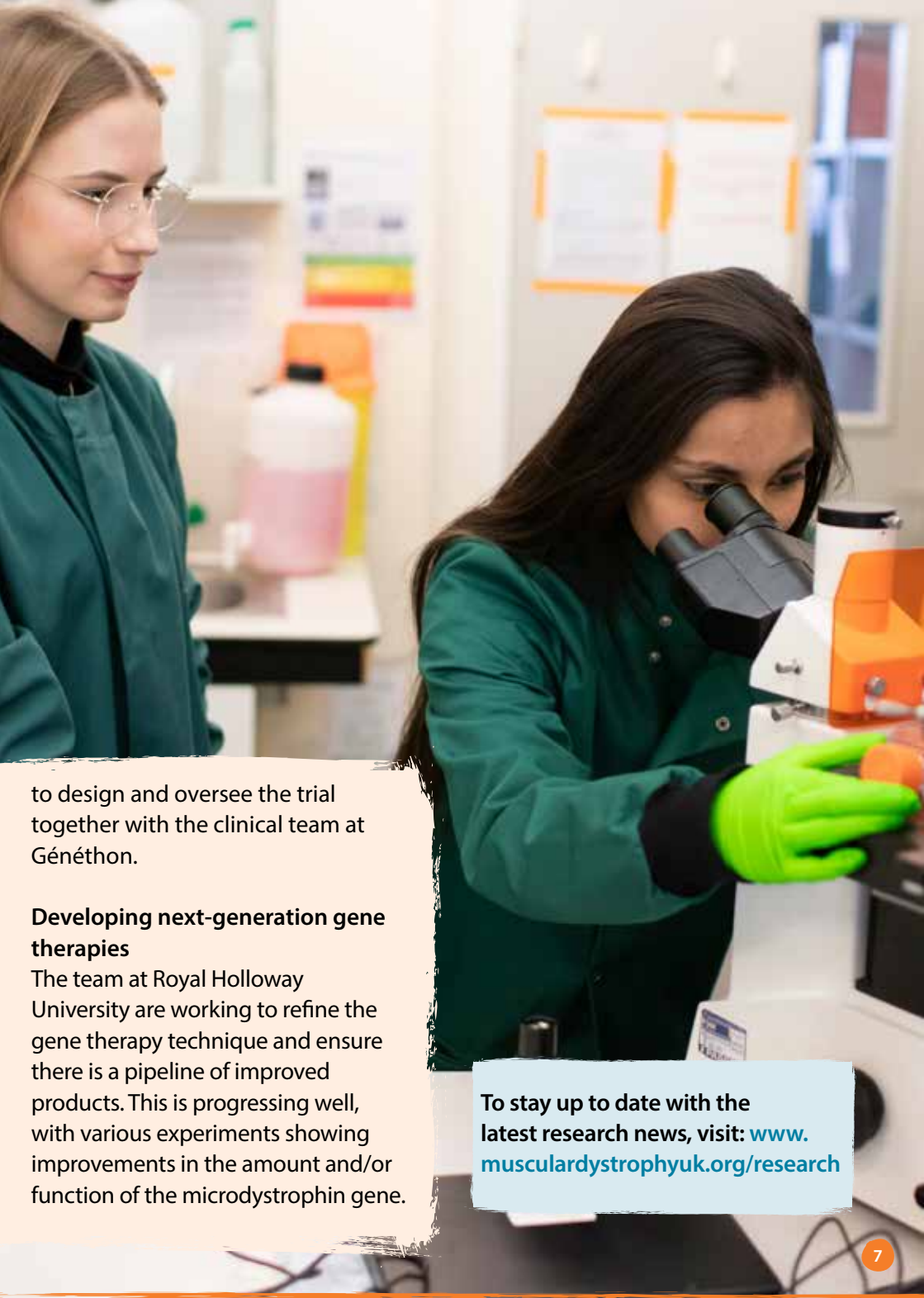
At the end of the first year

Producing the virus

Our French partners AFM Téléthon, and its not-for profit biotech, Généthon, are 'scaling up' the manufacturing process to produce large amounts of the AAV8-MD1 at clinical grade quality.

The UNITE-DMD clinical trial

Preparatory work for the clinical trial is underway in London and Newcastle, where researchers have started recruiting the clinical teams



to design and oversee the trial together with the clinical team at Généthon.

Developing next-generation gene therapies

The team at Royal Holloway University are working to refine the gene therapy technique and ensure there is a pipeline of improved products. This is progressing well, with various experiments showing improvements in the amount and/or function of the microdystrophin gene.

To stay up to date with the latest research news, visit: www.musculardystrophyuk.org/research

Positive results from myotonic dystrophy study

The first-ever trial for adolescents and adults with congenital or juvenile-onset myotonic dystrophy type 1 was held at the John Walton Muscular Dystrophy Research Centre in Newcastle.

The trial was to investigate the safety and effectiveness of tideglusib (AMO-02) – a drug that limits a particular enzyme found to be over-active in people with the condition. Research in animal models had shown that inhibiting this enzyme can increase muscle strength.

There were 16 participants, aged between 13 and 34, in this phase 2 study. Every day for 14 weeks, the participants took an oral dose of either 400mg or 1000mg of the drug, or a placebo. Doctors and caregivers monitored them over the 14 weeks.

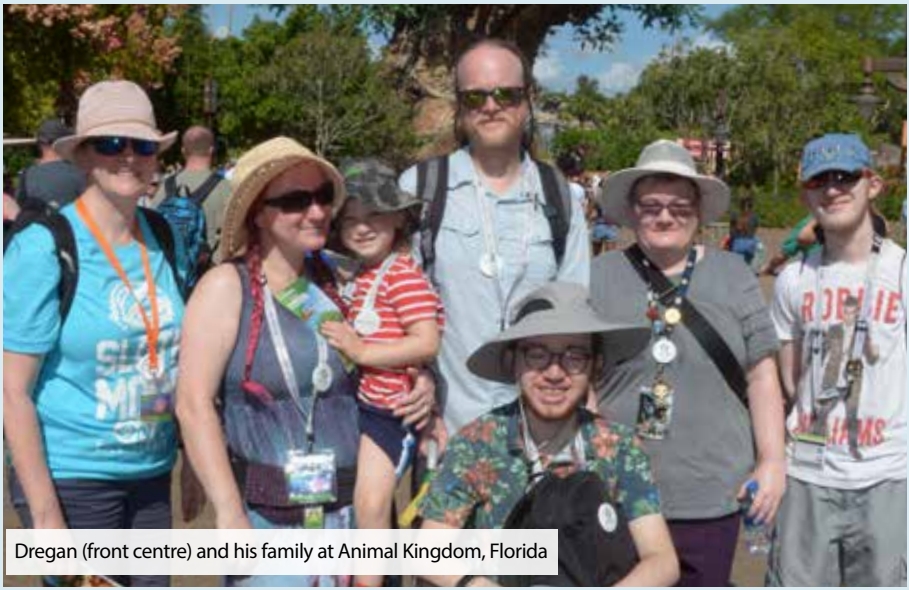
Most participants who received the drug had improved brain function and reported feeling less tired. They were better at performing day-to-day tasks and activities and several participants showed improvement in autistic symptoms.

The higher and lower doses of tideglusib were both found to be safe and well tolerated, and the higher dose appeared to more beneficial. The study was described as a 'landmark trial' by Dr Michael Snape, Chief Executive Officer of AMO Pharma, the company that conducted the study.

"These significant data are an important step in the development of AMO-02 as a potentially safe and effective treatment option for many patients living with congenital and childhood onset myotonic dystrophy type 1."

AMO Pharma is planning to further evaluate the efficacy of tideglusib in larger multi-site clinical trials in the US, Canada and the UK.

To find out more about research and clinical trials, get in touch with the MDUK Research Line on 020 7803 4813 or at research@musculardystrophyuk.org



Dregan (front centre) and his family at Animal Kingdom, Florida

Meet Dregan

19-year-old Dregan Turner is one of the 16 people who took part in the trial.

Dregan was diagnosed with congenital myotonic dystrophy in 2014 when he was 15 years old, after having been diagnosed with a learning disability and autism.

Dregan's mum, Emma-Jayne, felt the trial was very successful. She said it made a real difference to Dregan's ability to interact with others.

"It seemed to literally open up his brain, allowing him to interact with people and the wider world. He started to pay attention to the family and join in, he became more empathetic and interested in the world around him and his speech improved.

"Now that the trial has ended, the symptoms of Dregan's autism and learning disability are increasing again. It's really hard for us to see, as it makes us realise there is more inside him, it's just stuck".

She added that they hoped the drug could be made available in the future, because of the potential it offered to people like her son.

Technology enabling independence

For more than 30 years, and with thanks to generous supporters like you, MDUK has helped more than 5,000 children and adults with muscle-wasting conditions to purchase specialist mobility equipment not available on the NHS.

This is done through the Joseph Patrick Trust (JPT) – the welfare fund within Muscular Dystrophy UK, which provides grants towards the costs of this specialist equipment.

JPT also awards grants towards the costs of different kinds of assistive technology - cutting-edge technology that helps individuals with muscle-wasting conditions to maintain or improve their ability to communicate independently and maintain greater control of their immediate environment.

Adaptive switches and switch interface

These switches allow you to control a computer, phone or tablet by using large, easy-to-press buttons, or existing wheelchair controls. They are very useful for people with muscle-wasting conditions because they don't require precise movement in your hands and so are often easier to use if you have less dexterity in your fingers or weakness in your hands and arms.

Robotic Neater Eater

This robotic eating-aid (pictured right) offers greater offers greater independence to those who face difficulties when eating. The self-levelling spoon and rotating plate can be controlled using a tablet. Switches can also be used with the robotic Neater Eater so that it can be controlled by a person's head or eye movements for example.

Communications software

This uses text-to-speech technology, which translates what you type into audio. The software ranges from programmes with a simple vocabulary to programmes that allow for more detailed sentence construction and include predictive text to increase the speed of communication. This can help those who struggle with speech, by acting as a substitute voice.

Eye gaze (pictured below)

This incredible technology allows a user to control other devices using only eye movements. The devices have built-in cameras, which track eye movement and translate it accurately to the movement of the mouse cursor on screen.

Smart home and environmental controls

With home-based assistive technology, you can control the lighting, temperature and other electrical equipment in your home, either by using an app on your smartphone or tablet, or by voice control.

This makes everyday tasks easier – from being able to turn on the light, to boiling the kettle to making phone calls.

To find out more about JPT grants, and the difference your support makes, visit: www.musculardystrophyuk.org/jpt

New online module for teachers

MDUK has launched a new module aimed at teaching professionals, with the aim of helping them provide better care for the 2,500 pupils across the country who have a muscle-wasting condition.

The free online module takes just 15 minutes to complete. It provides up-to-date information about inclusive education for young people with muscle-wasting conditions.

Developed with the support of specialist clinicians, it gives an overview of some of the problems associated with having a muscle-wasting condition and looks at how secondary schools can identify the challenges that affect the ability of these pupils to learn.

Nigel's 12-year-old daughter Emily has Charcot-Marie-Tooth disease (CMT). He writes:

"As a parent of a daughter with CMT who is at secondary school, I am so pleased that MDUK has developed this new module. It is really important that teaching staff are able to support children with the extra challenges they have to face as a pupil with a muscle-wasting condition. If teachers had known about even the basics of my daughter's condition, it would have been a massive help."

Bobby Ancil, Head of Outreach, Commissioning and Professional Development at MDUK, said:

"Schools can play a vital role in supporting and encouraging a child with a muscle-wasting condition to live and think independently. We would encourage all teaching professionals to make use of this free tool so they can be better equipped to help the children in their care."

If you'd like to let a teacher or education professional know about this online module, please contact David Stephenson at d.stephenson@musculardystrophyuk.org or on 020 7803 4826

Giving patients a voice in drug development

Members of the PREFER project group at the 2018 annual meeting in Leuven, Belgium

Drugs are developed for patients, so it is essential that their views and preferences be incorporated into the development process. For example, pharmaceutical companies need to know what patients value, in order to develop drugs that are suitable for patients' needs.

As we continue to strive to find treatments for muscle-wasting conditions, it's important that those involved in drug development know how to collect this information so the patient voice can be heard.

MDUK is working with pharmaceutical companies, academics, patients, health technology assessment bodies and regulatory authorities in a five-year international project called PREFER. The project looks at how and when it's best to Capture and include patient preferences in decision-making during the drug life cycle.

The PREFER project, which started in 2016, aims to find out patients' views and preferences on different treatments, both real and hypothetical, across three different health areas: rheumatoid arthritis; cancer and neuromuscular conditions; and to gain insight about how patients made decisions about treatments. It also advises on how to collect patients' preferences and how decision-makers should use this information.

Together with Newcastle University, MDUK is helping to lead the neuromuscular patient preference study, focusing on myotonic dystrophy type 1 and mitochondrial disease.

Watch this space for updated information about the study: www.musculardystrophyuk.org/research

Get involved

The Great Muscle Raffle is now open!

If you would like to receive raffle tickets in the post to buy or sell to family and friends, please contact us at **raffle@musculardystrophyuk.org** or **0300 012 0172**

Every ticket you buy will help us continue to be here for individuals and families living with muscle-wasting conditions in the UK.

To play online, visit **www.musculardystrophyuk.org/raffle**

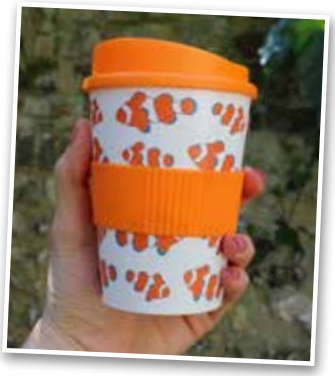
Congratulations to our Christmas Raffle winners!

First prize: £3,000	R. Wynne, Rochester
Second prize: £250	R. Watts, Burton on Trent
Third prize: £50	C. Davis, Leicester

Thank you to everyone who took part

Find something you love at the MDUK shop

Visit our online shop and see the exciting new items that have arrived – including the brand-new MDUK travel mug (pictured), a new addition to our fabulous clownfish range of gifts.



Whether you're hoping to find the perfect gift or a treat for yourself, you'll be sure to find something you'll love at the MDUK shop and best of all, all proceeds will go towards beating muscle-wasting conditions.

Shop online at www.musculardystrophyuk.org/shop

Remember someone special with MDUK

Fundraising in memory of a loved one is a wonderful way to come together with friends and family to celebrate their life.

"My eldest son Paul loved life and lived it to the full. He was bright, accomplished and his positivity left a mark on the people lucky enough to know him. When Paul passed away, we found out that he wanted us to hold a disco for his wake. He loved a party.

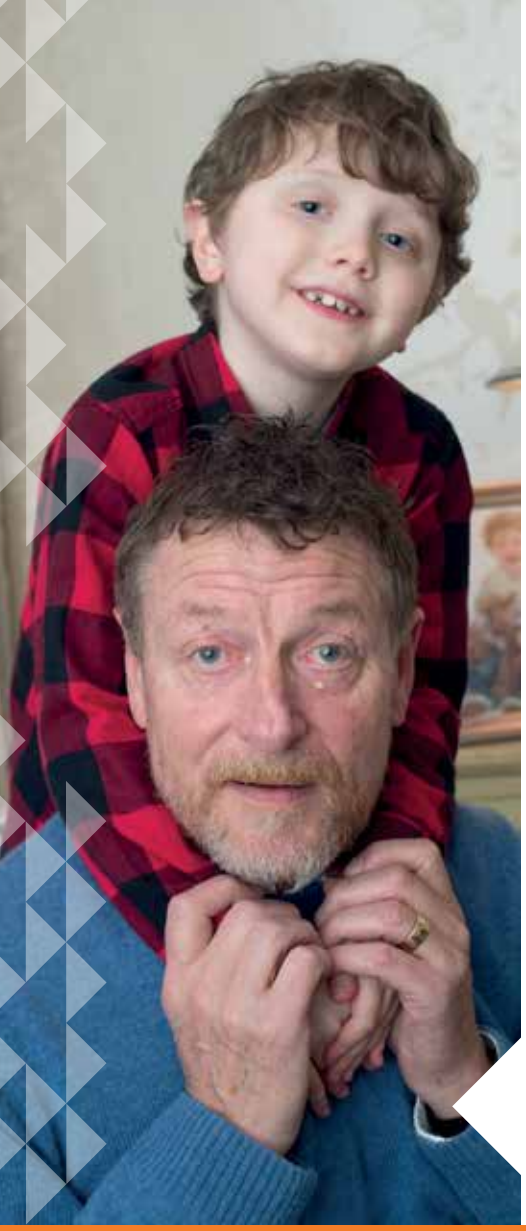
"When we started fundraising in Paul's memory, a disco felt like the perfect way to start raising money for Muscular Dystrophy UK. It's a way for us to keep Paul's memory alive and to help MDUK continue their work to find treatments for muscle-wasting conditions like his." Anne Peterson, whose son Paul (pictured) had Duchenne muscular dystrophy.

Visit www.musculardystrophyuk.org/get-involved to find out more about how you can fundraise for Muscular Dystrophy UK and help beat muscle-wasting conditions.

Thank you for giving us your feedback

Thank you to everyone who completed our recent survey to let us know your thoughts on *Campaign* newsletter. We'll continue to fill *Campaign* with the latest news, stories and developments in research that you enjoy reading about, and and will also aim to bring you articles with a greater range of topics and conditions.

The survey has now closed, but you can always email us at reply@musculardystrophyuk.org if you'd ever like to let us know your feedback or thoughts.



LEAVE A GIFT IN YOUR WILL TO CREATE A future free from the limitations OF MUSCULAR DYSTROPHY.

WITH YOUR HELP the next generation of RESEARCH scientists will FINISH WHAT WE'VE STARTED.

"We were devastated when we found out our beautiful grandson, Jack, had Duchenne. All we wanted to do was take this horrible condition away. We can't do that, we know. But your gift in your Will could make that happen one day." **Steve Gauder, Jack's granddad**

Muscular Dystrophy UK

Fighting muscle-wasting conditions

For more information about gifts in Wills, call Catriona Parker on **020 7803 4834** or email **legacy@musculardystrophyuk.org**

www.musculardystrophyuk.org/giftsinwills

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