Muscular Dystrophy UK

Fighting muscle-wasting conditions

Campaign

The newsletter for our supporters

Inside:

Update from the Keenan family

Our new commitments

JPT – promoting independent living for 30 years

Spring 2017

A life-changing decision

With your support, Muscular Dystrophy UK has for some years funded research into mitochondrial IVF. Now, Newcastle University has received the first licence from the Human Fertilisation and Embryology Authority (HFEA) to perform the technique. It can prevent eligible women affected by mitochondrial disease from passing the condition on to their future children.

"This is wonderful news for the many women we know who have faced heartbreak while trying to start their own family. Newcastle was where we first funded Prof Sir Doug Turnbull and his research in this field more than 10 years ago, and so there couldn't be a better place for this technique to be trialled. We eagerly await news of the first children in the UK to be born free of this serious, debilitating condition."

Robert Meadowcroft, CEO of Muscular Dystrophy UK

Welcome

I'm pleased to bring you this spring edition of *Campaign*, giving you news of Muscular Dystrophy UK's exciting work to beat muscle-wasting conditions.

Inside, you'll read about some new international research projects we are funding into the rare condition, nemaline myopathy. You'll also see how we're pressing to get Duchenne drug, Exondys51, approved for use in the UK by the European Medicines Authority.

We're so grateful for your generosity at Christmas time, in response to Liz Keenan's letter. As well as this, you sent in over 300 Christmas baubles with messages of support to everyone living with muscle-wasting conditions – a truly overwhelming response. Thank you. On page four you can read what the Keenan family is doing now.

Please enjoy this edition of *Campaign*. Thank you for everything you do to improve the lives of people living with muscle-wasting conditions.

Sue Barker, OBE
President, Muscular Dystrophy UK

PS Our online shop is full of fun, meaningful and novelty gifts you can buy for friends and family – there's something to suit everyone. Shop now at www.musculardystrophyuk.org/shop

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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Join us online

Make sure you keep up-to-date by joining our social media networks.

www.facebook.com/musculardystrophyuk
Join the **25,000+ community** on our Facebook page.

@MDUK_News
Keep on top of our breaking news by following us on Twitter.

www.youtube.com/user/MuscularDystrophy
Watch our videos on YouTube.

instagram.com/musculardystrophyuk
Share our pictures on Instagram.

On the cover: Liz, James and Myles Keenan with supporters of the Family Fund 'I fight for James'.

Update on the Keenan family

On behalf of the Keenan family, we'd like to say a huge thank you to all our supporters for your generosity at Christmas. Over 1,200 of you responded to Liz's letter with gifts totalling more than £35,000.

Liz and Sean Keenan have three boys, James (10) and the twins, Myles and Joshua (who turned three just after Christmas) – all three boys live with Duchenne muscular dystrophy.

Christmas was a busy time for the Keenans. As well as the usual festivities, the Keenan family once again organised an annual Christmas Market in their local area – this year's was bigger than ever and even Santa made an appearance.

Liz says, "The Christmas Market was a huge success and everyone loved it. We couldn't have done it without all our supporters. So – with a drumroll please – I am overwhelmed to say that, with some matched funding, we raised £5,111 in total!"

And the Keenan family haven't stopped the fundraising there. In the coming months they have three big events planned through their Family Fund 'I fight for James':

- On **6 May**, a team of 17, including 10-year-old James and his cousin, Henry, will be taking part on the Velocity zip wire in Snowdonia – it's the fastest zip wire in the world reaching speeds of up to 100mph

Below (l to r) Myles, Liz, James, Sean and Joshua Keenan

- On **16 July**, they have a Move a Mile for Muscles Family Fun Day planned at the Torbay Velopark
- And in **September**, Dad Sean will be climbing Mount Kilimanjaro with a friend.

Liz says, "It's amazing to think just what can be achieved with all that funding for research. With lots of prayers and hopes for our boys' future, I truly believe that a cure is one step closer every day!"

With your support, and the hard work of families like the Keenans, Muscular Dystrophy UK can continue to invest in high-quality research to find effective treatments and cures for conditions like James', Myles' and Joshua's, as quickly as possible.

Update on GP training course

At Christmas Liz also told us of the importance of early diagnosis. GPs play a key role in ensuring early diagnosis by being able to pick up on the tell-tale signs of a muscle-wasting condition and refer onto a specialist. We spread the word about how to ask your GP to take Muscular Dystrophy UK's specialist training course on neuromuscular conditions and are delighted to announce that we saw a definite increase in the number of GPs taking the course in the months following Liz's Christmas letter.

- **If you'd like to ask your GP to take the course, you can get more information from Lloyd Tingley, MDUK's Neuromuscular Outreach Officer at l.tingley@musculardystrophyuk.org or 020 7803 4804.**

- **If you haven't yet read the Keenan family's inspiring story, please visit www.musculardystrophyuk.org/christmas**

Over 300 baubles with messages of support were sent in over Christmas. The baubles covered the MDUK Christmas tree from top to bottom and created a truly moving display.

Research

It is through your generosity that we are able to push forward groundbreaking research, taking us closer to finding treatments and cures for muscle-wasting conditions.

Exondys 51, the FDA and the EMA

The Food and Drug Administration (FDA) in the USA has given the green light to a new drug for Duchenne muscular dystrophy. While this is the first exon skipping drug to be approved, the decision doesn't apply to Europe.

Sarepta Therapeutics, manufacturer of the drug Exondys 51, has now applied to the European Medicines Agency (EMA). The application has been validated for assessment during 2017, and MDUK is working with other patient groups to push for fast access to the drug here.

MDUK has backed the development of exon skipping technology for over 20 years. Exondys 51 treats boys with a gene mutation amenable to exon 51 skipping, and it is designed to slow down the progression of Duchenne. Three years of clinical trials showed that boys on the drug could walk 151 metres more in six minutes than boys not on it.

Professor Matthew Wood, who is currently developing exon skipping approaches at Oxford University, said:

"Muscular Dystrophy UK has played a critical role over almost two decades supporting the

development of exon skipping drugs, which have real potential to bring significant clinical benefit to Duchenne patients. First-generation exon skipping drugs such as Exondys 51 are very important as they have helped to build knowledge and understanding of how such drugs can be successfully developed and evaluated in patients."

T-Jay Roe - who could benefit from Exondys 51

Nemaline myopathy

MDUK has invested £375k into two international research projects into the rare condition, nemaline myopathy.

Professor Coen Ottenheijm at VU University Medical Centre, Amsterdam is investigating whether blocking a particular cellular process could slow down muscle atrophy in mouse models of nebulin-based nemaline myopathy. His research could lead to a potential treatment that prevents, or at least slows down, muscle atrophy in people with nebulin-based nemaline myopathy. The treatment could also reduce their muscle weakness.

Dr Carina Wallgren-Pettersson and her team at the University of Helsinki, and Samfundet Folkhaelsan in Finland are developing techniques for a fast and reliable genetic diagnostic service for people with nemaline myopathy. This research will also give scientists a better understanding of how nebulin mutations cause the condition. This knowledge will be important for the development of treatments in the future.

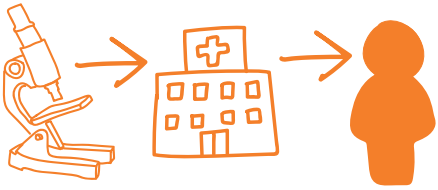
Families taking part in the project will also gain knowledge about how their condition has been passed down or inherited. This may be important for prospective parents who are thinking about starting their own family. The findings will also be the starting point for research into potential therapies, which could have important benefits for people with nebulin-based nemaline myopathy.

Keep up-to-date with all the cutting-edge research you're helping fund: www.musculardystrophyuk.org/research

New commitments

Thanks to your generous support over the years, we have been able to invest over £4.5 million into five new transformational commitments. These game-changing projects, aim to accelerate research into an ambitious new era for clinical trials and improve quality of life.

1. The Oxford Neuromuscular Translational Research Centre



By investing £1m into the development of a multi-million pound centre – the Oxford Neuromuscular Translational Research Centre – we aim to change the landscape for clinical trials in the UK.

Centres in the UK have been forced to turn down some clinical trials for muscle-wasting conditions owing to lack of capacity. To create additional capacity, we are investing in posts and other resources so more potential treatments can be tested in clinical trials.

We're working with Oxford University to establish a world-leading centre aimed at bringing new treatments to patients more quickly. The Centre will not be a physical building but situated in existing sites in Oxford.

We have always funded world-class research in Oxford – including the work of our Vice Presidents, Professor Dame Kay Davies and Professor Matthew Wood. Their work alongside that of Professor Kevin Talbot, and the rest of the clinical care team, has truly moved forward the treatment options for neuromuscular patients. With this new investment, we are able to drive this work out of the laboratory and into the clinic.

"This is a very important time in the history of research and therapy into muscle conditions. MDUK has been a major contributor to the basic research for more than two decades. The funding of a new neuromuscular translational centre at Oxford ensures that they can build on this investment and deliver on effective therapies for paediatric and adult disorders. I am very excited to be part of this initiative."
Professor Dame Kay Davies, Professor of Anatomy at the University of Oxford

2. NorthStar Programme

NorthStar is the world's largest natural history study for Duchenne muscular dystrophy, which we helped to establish in 2004. With our investment of £1.45m, NorthStar will be upgraded significantly to improve the collection of data for clinical trials, and to gather information from adults with Duchenne. It also offers a unique platform to influence best practice in the care of Duchenne patients nationally.

Emerging trend-data from the study is already helping to improve quality of life. By showing the benefits of steroid treatments, NorthStar led UK clinics to use steroids in treating Duchenne patients.

As a result, children with Duchenne are now able to walk for around 3.5 years longer than children in the previous decade. This tangible benefit of a treatment was identified as one of the most impressive achievements ever demonstrated at a national level.

3. UNITE-DMD Trial

By investing £1.6m into UNITE-DMD, an international collaboration working on gene therapy, we aim to advance treatments for muscle-wasting conditions. Once preclinical studies are complete, UNITE-DMD will assess the safety of gene therapy for Duchenne muscular dystrophy in a phase I/II clinical trial.

To date, no gene therapy trials for muscle-wasting conditions have been undertaken in the UK. Although this particular gene therapy will be designed to treat Duchenne muscular dystrophy, its development will refine and improve the technique generally. This will help towards the development of gene therapies for other muscle-wasting conditions in the future.

UNITE-DMD is a four-year project taking place in the UK and France. While we are funding the UK side of the project, in partnership with Action Duchenne, the French Muscular Dystrophy Association (AFM-Telethon) is funding the French arm of the project.

4. Here for you: clinic support

Working alongside healthcare professionals, our new Here for you: Clinic support programme will address a growing need for support in local communities. We will have information and advocacy officers working alongside healthcare professionals in clinics, to offer

practical and financial support to people living with a muscle-wasting condition.

By offering tailored, region-specific support to clinics, our teams will also call on the complementary services we offer. This will ensure access to all the resources and community support as well as the best of specialist healthcare.

We have already established posts in clinics in London, Wales and Northern Ireland. This investment will see four further roles at Great Ormond Street Hospital, Oxford, Newcastle and Scotland.

"The role here in Newcastle would provide essential support to our patients and their families, ensuring they receive the highest quality health, education and social care services."
Dr Chiara Marini Bettolo, Consultant Neurologist at John Walton Muscular Dystrophy Research Centre

5. Here for you: mental health matters

Living with a rare, progressive muscle-wasting condition can be incredibly challenging. But when you're denied – or not offered – basic support, for simple things like emotional wellbeing, it can leave you feeling isolated and alone. And that's hard.

Long-term prognosis is improving and there are exciting treatments on the horizon. But muscular dystrophy and its wide-ranging impact can affect every area of life, including mental health. If left unsupported, these negative experiences can have a detrimental effect and can ultimately lead to devastating consequences.

Professional psychological support is essential to improve emotional wellbeing. It doesn't make sense for this support to be forgotten and ignored. But we know this is happening, leaving many out in the cold to support themselves or seek information from non-professional sources including the internet.

Driving for access to specialist psychological care and support from the moment of diagnosis to end-of-life, this investment is our commitment to making tangible changes to the current offering for all individuals and families living with muscle-wasting conditions.

To find out more and how to support these projects, please visit www.musculardystrophyuk.org/new-commitments

Become a Friend of MDUK

Here's a brand new scheme that you can join for just £15* a year. You and your family will get a whole range of discounts, and you'll be helping fight muscle-wasting conditions at the same time.

Anyone can become a Friend of MDUK, and here's what you'll get:

Inspiration and information

Each year, we'll send you two editions of our lifestyle magazine, *Target MD*, one *Target Research* round-up on groundbreaking research, and the charity's impact report, outlining how you're helping beat muscle-wasting conditions.

Peace of mind

Our partnership with specialist insurers, Fish Insurance, will give you and your family members: 10 percent discount on independent living cover, 15 percent on travel and home insurance, £50 off the cost of your car insurance and much more.

New and stronger connections

You and everyone in your household will get 50 percent discount on MDUK's National and Scottish Conference tickets. At the conferences you'll get to meet and connect with other families living with muscle-wasting conditions, and hear updates on research and care.

Accessible breaks for less

Handiscover does all the research for you into accessible holiday accommodation. They check all of the accommodation listed on their website to ensure it meets your needs. You'll get £40 off one holiday booking with Handiscover each year.

To find out more, give us a call on 0300 012 0172 or visit www.musculardystrophyuk.org/friends

£15 a year when you join by Direct Debit. £18 a year when you join by debit/credit card or cash

Joseph Patrick Trust

Adam in his new school uniform

Promoting independence for over 30 years

You've helped around 6,000 children and adults with muscle-wasting conditions to live more independently. With your generous support, the Joseph Patrick Trust (JPT), our grant-awarding arm, has awarded £6m worth of grants since its establishment in 1986. These grants go towards the cost of wheelchairs, mobility aids, and some forms of assistive technology.

Adam, who has Duchenne muscular dystrophy, needed a powerchair for getting around his new secondary school and for sitting comfortably in lessons. He was turned down by the NHS for wheelchair funding, so his family applied successfully to the JPT. The grant they received went towards the cost of a Quickie Salsa chair, designed specifically for children.

"Adam had a fantastic first day at school and was extremely comfortable in his chair during all of his lessons; he is settling in well. Without the help of the JPT part-funding his wheelchair, starting school would have been an even more daunting prospect for him. Adam is able to be independent and integrate fully with his friends."
Toni, Adam's mum

The JPT was established by Alexander Patrick CBE in memory of his father, Joseph, a founder of Muscular Dystrophy UK. Inspired by his brother Andrew, who had Duchenne muscular dystrophy and who died in 1962 aged 13, Alexander established the JPT to promote independence and quality of life for people living with muscle-wasting conditions.

"When Andrew was a child, hope was far on the horizon. There is now greatly improved availability of equipment and care. All of this underlines the importance of Muscular Dystrophy UK's role in funding research and providing support, along with JPT providing equipment to help maintain mobility and independence."
Alexander Patrick, Honorary Life President of Muscular Dystrophy UK

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


Alexander Patrick and Libby Smalley

Ways to get involved

The winner of our Christmas card competition

A huge thank you to all the children who entered our 2016 Christmas card design competition. The standard of entries was very high, which once again gave our Chief Executive, Robert Meadowcroft, a very difficult choice in deciding a winner.

However, we are very pleased to announce that 10–year-old Archie Kay has won the competition with his entry *The Polar Express*. **Congratulations to Archie!**

- **Archie designed his entry in honour of his brother Bertie, who has Ullrich congenital muscular dystrophy. Keep an eye out for Archie's card in our 2017 Christmas card range, available from September.**

Raffles - a fun way to support

Thank you to everyone who took part in our Christmas Raffle. Our warmest congratulations to Ms Carter who won the first prize of £3,000 and also to Mrs Young and Mrs Jenkins who won second and third prizes respectively.

Our Great Muscle Raffle 2017 has now launched and there is still time to buy some last-minute chances before it closes on 8 May 2107. Taking part is a fun way to support individuals and families across the UK who are living with muscle-wasting conditions.

- **If you'd like to buy chances in the Great Muscle Raffle, please phone our Raffle Hotline on 01628 201 289 or visit www.musculardystrophyuk.org/raffle**

Run in our 36th Oxford Town and Gown 10k

Run in our 36th Oxford Town and Gown 10k

Join **#TeamOrange** on Sunday 14 May and help us turn Oxford orange in this chip-timed race to fundraise for Muscular Dystrophy UK.

The unique and scenic route through Oxford makes it perfect for all levels, from experienced runners wanting to beat a personal best to beginners taking on their very first 10k run.

Book your place today – it costs just £25 for adults and £12 for juniors.

- **Find out more at www.townandgown10k.com/oxford or email townandgown10k@musculardystrophyuk.org**

Spring catalogue – out now!

With some brand new items as well as some old favourites, our spring catalogue is sure to have something to tempt you.

And it's shopping that really makes a difference, because when you shop with us, you'll know that all the proceeds will go directly towards beating muscle-wasting conditions.

- **So treat yourself or a loved one today. You can order using the form in the enclosed catalogue or view the range and purchase online at: www.musculardystrophyuk.org/shop**

Muscular Dystrophy UK

Fighting muscle-wasting conditions

Move a Million Miles for Muscles

This summer, scoot, swim, cycle, wheel, walk or even hop a mile in a Move a Mile for Muscles event!

You can also join our ambitious challenge to Move a Million Miles, and track your miles. By turning your miles into money, you'll be taking strides towards treatments and a cure.

Find out more at **www.musculardystrophyuk.org/move-a-mile-for-muscles** or call **0300 012 0172**

www.musculardystrophyuk.org/move-a-mile-for-muscles

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#MusclesMatter