Muscular Dystrophy UK's lifestyle magazine
Summer 2016

Target MD

HRH The Duke of Edinburgh – celebrating 50 years of support

Looking back and looking ahead: the fight to beat muscle-wasting conditions

Remembering Lord Walton of Detchant – charity founder and eminent scientist

Target Research: news and updates

Photo © Kit Studios

Muscular Dystrophy UK
Fighting muscle-wasting conditions

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The magazine for supporters of Muscular Dystrophy UK, written and produced entirely in-house.

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On the cover
Tanvi Vyas, Trailblazers Project Manager, giving her speech at the reception at St James's Palace (see p8)

FundRaising Standards Board

Hello

Hello and welcome to the 2016 summer edition of *Target MD*. As always it comes to you with stories of the outstanding people who support Muscular Dystrophy UK. It's also something of a celebration of 50 years of royal support, and an opportunity to look ahead.

While we share some reflections following HRH The Duke of Edinburgh's anniversary event at St James' Palace in June, we also look ahead. Professor Dame Kay Davies does both, as she reflects on the progress in research and looks at the equally promising times ahead. There are also updates of amazing families doing great things to support the ongoing work of the charity, with their sights set firmly on a positive future for their children.

It's just about time for the Rio Paralympics to begin. Charity ambassadors and elite athletes, Stephen McGuire and Ollie Hynd will represent Great Britain in Boccia and swimming respectively. See what they have to say about sport and disability (page 4).

Meet some fantastic #TeamOrange members. See what motivated them to get involved in our events and challenges. Have a look at what's coming up too – we'd love to have you join in and fundraise for us. Get in touch with our events teams; they'd love to welcome you. They always do a great job of supporting you too.

As always, please keep in touch and tell us what you'd like to read about in future editions. We want to bring you the magazine you want to read.

Ruth

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About us

Muscular Dystrophy UK supports 70,000 children and adults with muscle-wasting conditions to live as independently as possible. We accelerate the pace in development of effective treatments and cures.

Helpline

If you'd like to speak to someone about living with a muscle-wasting condition, please call our friendly care and support team.

They are available from 8.30am to 6pm Monday to Friday, on **0800 652 6352** (Freephone helpline) or **info@musculardystrophyuk.org**

Join us online

Get all the latest news and updates by joining our social media networks.

Online forum
community.muscular-dystrophy.org
Join our friendly online community.

www.facebook.com/musculardystrophyuk
Join the **22,000+ community** on our Facebook page.

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

www.youtube.com/c/musculardystrophyuk
Watch our videos on YouTube.

instagram.com/musculardystrophyuk
Share our pictures on instagram.

Photo © Anne-Marie Briscombe

Thank you for your ongoing support

Everything we do, as you'll read in the pages ahead, relies on the support of generous people like you. People who understand what it will take to beat muscle-wasting conditions.

Join us – we can do this, together.

Please contact our fundraising team to find out about all the ways you can get involved. Call **0300 012 0172** or email **fundraising@musculardystrophyuk.org**

Our Paralympic champions

Photo © Kii Studios

While most of us will be armchair spectators of the Paralympic Games 2016, a group of elite athletes with muscle-wasting conditions will be representing Great Britain at swimming and Boccia in Rio. Between 7 and 18 September, take a look out for:

- Oliver (Ollie) Hynd (pictured above) – swimming
- Stephen McGuire (pictured right) – Boccia
- Scott McCowan – Boccia
- Jamie McCowan – Boccia.

Stephen McGuire (31), charity Ambassador from Hamilton, as Scottish, British and European champion and World Number 2, is the most successful British BC4 Boccia athlete. He has an undiagnosed form of muscular dystrophy.

"I'm absolutely delighted to have been selected for Rio this year, where I hope to build on my success from London 2012. I'm really proud to be representing Great Britain at the Paralympics, as well as people all over the world living with muscle-wasting conditions.

"At Rio, I want to show people that it is possible to achieve their dreams and hopefully bring home the gold for Britain."

Stephen encourages young people with an interest in sport to pursue their dreams.

"If you're interested in competing right up to Paralympic level, the opportunities are out there. Boccia is a very inclusive sport for individuals with neuromuscular conditions; it's also a great way to be competitive, meet friends and travel the world."

Ollie Hynd MBE from Nottinghamshire is a gold-medal-winning Paralympic swimmer, who holds British, European and world records across his events.

Now 21, Ollie was diagnosed with neuromuscular myopathy as a young teenager. He has been an ambassador for Muscular Dystrophy UK since 2012, and is keen to encourage more young people with muscle-wasting conditions to benefit from sport.

"I couldn't be more excited to represent Great Britain again at the Paralympics in Rio. Swimming for my country is such a great honour and the entire squad is training hard and eagerly anticipating the games. I am ready to give my all in Rio and show other young people with muscle-wasting conditions that they can achieve their dreams, just like I have."

Keep up-to-date with news of our elite athletes at www.musculardystrophyuk.org/RoadtoRio

Join in the conversation online using #RoadtoRio

Photo © Kii Studios

A vision for the future

The power of football

The Olympic and Paralympic Games often motivate and inspire young people to take up sport. Powerchair football has emerged as one of the most popular and fastest growing sports for people living with muscle-wasting conditions – more than half of all players in the UK have a muscle-wasting condition.

The Wheelchair Football Association (WFA) was formed in 2005 to govern the sport of Powerchair Football in England. Recognised by the Football Association (FA) as the sport's governing body, with its Laws sanctioned by them, the WFA works closely with the FA and networks to develop more playing opportunities.

The WFA focuses on developing the sport at grassroots level, and runs its own Coach Education and Referee Training courses.

As well as the national Premiership and Championship Divisions, the WFA also runs a number of regional league competitions throughout the season. Aspire PFC finished the 2015/16 season top of the Premiership table, with Northern Thunder and West Bromwich Albion close behind.

Finally huge congratulations to Trailblazer, Connor Colhoun (19), who has Duchenne muscular dystrophy. Scoring an incredible 55 goals for Queen's Park PFC this season, he was named the Scottish FA National Powerchair League Top Goalscorer 2015/16.

To find out more about powerchair football, visit **www.thewfa.org.uk**

All of us connected with the charity felt great sadness but also gratitude, respect and affection when we heard of the passing of John Walton. Many well-deserved tributes were paid to him and of course his loss has been felt most keenly by his family. We were fortunate that the young Dr Walton, with his mentor Professor Fred Nattrass, met Joseph Patrick and together they established the Muscular Dystrophy Group, as we were then known, in 1959.

Almost by chance, after meeting boys with muscular dystrophy at a hospital fête in 1962, the late Richard Attenborough generously committed his support for the charity. Together they set about raising huge sums for research and care. Tireless and innovative, they were a formidable combination and we are continuing to see progress today based on the firm foundations they laid.

For more than 50 years, John was a loyal, dedicated friend of the charity. He was committed first and foremost to his patients and all those living with muscular dystrophy. He recognised the charity could not only raise vital funds for research but also provide advice and support.

His expertise, knowledge and integrity were recognised in this country and internationally and many formal honours came his way. In his later years, as a widely respected member of the House of Lords, he made many telling contributions to debates around research, NHS care and muscle conditions.

In his last illness, John must have been delighted to hear the much delayed decision by NICE and NHS England to allow Translarna to be available for boys with Duchenne caused by an underlying nonsense mutation. It is a landmark decision and marks a key staging point in our determination to accelerate the path to effective treatments. The drug is not yet available in Scotland and our campaigning work there continues, supporting the small group of boys eligible for the treatment.

As a tribute to the immense contribution of John Walton, we have launched the John Walton Research Fellowship to support a young researcher. The most important tribute to John Walton will be the day when effective treatments are available for all types of muscle conditions. This is the vision that inspired him at the start of his career and we will continue to do all we can to bring this day forward.

Robert Meadowcroft, CEO

TRAILBLAZERS
Muscular Dystrophy UK
Fighting muscle-wasting conditions

MOVE THE GOAL POSTS

With so much media focus on the Rio Olympics and Paralympics, Trailblazers have revealed just how difficult it can be for disabled people to watch sport.

Following on from two of their previous reports that briefly touched on spectator sports (*Calling time* and *Game on*), a new report out, *Move the goal posts* found that sports venues were failing in their duty to disabled people. It showed that many sport venues were not making reasonable adjustments for disabled spectators, leading to an inferior spectator sport experience.

In a survey of more than 100 young disabled people, the **key findings** include:

- 85 percent feel they are at a substantial disadvantage over non-disabled people when booking tickets to sporting events
- more than half have had to sit in an unsheltered seating area at a sporting event
- more than half say attending events in groups is the most difficult part of accessing live sporting events
- more than half have had to sit away from friends and family, at venues, with more than 20 percent having had to sit alone.

"Visiting the venues makes a day of the whole thing, rather than just watching a game, you're taking in the whole occasion: the venues, the atmosphere inside the stadiums, as well as the rest of day (where in cricket there will be lunches, breaks, etc). For the players it's a team sport, but it's the same for the spectators; better enjoyed together."
Joe Richardson, London

What next?

Trailblazers have called on sports venues, clubs and teams and the Government to:

- put accessibility into the heart of any future venue designs throughout the UK, by working with groups such as Trailblazers to discuss best practice
- ensure all front-facing staff have adequate disability equality training
- work with Trailblazers to set up a sports fan access group to discuss issues and improvements
- create an access card (similar to the Cinema Exhibitor Association card) to enable online ticket booking for disabled spectators, including carers
- offer online booking, with a Freephone number as an alternative.

To find out more about Trailblazers, MDUK's network of young campaigners, please visit www.musculardystrophyuk.org/trailblazers or call 020 7803 4800

Move a Mile for Muscles competition

Always wanted to watch a Premier League football match with an England and Arsenal star? Enter our competition and you and a guest could be heading to the Emirates!

Muscular Dystrophy UK Ambassador, Jack Wilshere, recently launched his Move a Mile for Muscles competition for the 'most creative mile'. It could be anything from hopping a mile on one leg, to whizzing around a race track in your powered wheelchair!

Jack will pick the winner after the competition closes on 30 September. You and a guest could then be heading to the Emirates to join Jack in his box for a Premier League football match.

Email your pictures and videos to volunteerfundraising@musculardystrophyuk.org Include your full name, address, contact phone number and email address, as well as a few words about what inspired your creative mile.

To find out more, visit **www.musculardystrophyuk.org/move-a-mile-for-muscles**

Photo © Kii Studios

go orange for a day!

Join in #TeamOrange's brightest day yet!

On Friday 27 January 2017, people in schools and workplaces across the UK will go orange for a day to help beat muscle-wasting conditions.

Get your school or workplace involved – it's quick and easy to register at **www.musculardystrophyuk.org/go-orange**

This promises to be an orange day to remember – be part of it!

Celebrating 50 years as royal patron

Photo © Kii Studios

On Thursday 30 June, our patron HRH The Duke of Edinburgh hosted a very special event at St James' Palace on behalf of Muscular Dystrophy UK. The majestic Queen Anne Room was the perfect setting for the 240 friends and supporters – including those who had applied through a public ballot – to celebrate 50 years of Prince Philip's support.

Prince Philip met Sir Alex Ferguson, Dame Mary Peters, and a number of Paralympian hopefuls. CEO Robert Meadowcroft introduced him to families living with muscle-wasting conditions, supporters, as well as scientists and healthcare professionals.

After a short welcome from Prince Philip, MDUK Chairman Bill Ronald talked about the charity's key achievements over the past half-century. In looking to the charity's future, he introduced Trailblazers Project Manager, Tanvi Vyas (pictured on the cover).

Tanvi, who was diagnosed with spinal muscular atrophy (SMA) when she was two, spoke of what the charity had done for her, her family and other families like hers.

"I joined the charity as a volunteer in October 2009, and soon became a member of staff. Here I am six years on a different person: I've gone from being a slightly nervous law graduate to being here in a palace, speaking to 240 of you!

"Working with MDUK has given me so much. An opportunity to help others, to see their confidence grow – as did mine – through having a platform, pushing my boundaries, finding my voice and speaking out for what I believe in.

"We're all here today because the charity has touched our lives in one way or another. My parents, as is the case for many of you I'm sure, want to know that we're at the cutting edge of research. Not just for me but for the next generation and many others to follow."

Photo © Kii Studios

"We met so many very interesting people and it was lovely to be there with the same deep rooted cause in all our minds. What an enormous privilege it was to meet HRH, I think everyone was touched by his interest and extraordinary charisma."

"Thank you for arranging yesterday. It was an honour to meet HRH Duke of Edinburgh – it was a truly memorable day."

"I met some amazing people, including some of the Trailblazers, who are an inspiration not only to people like myself but to everyone in society for the work they do."

Real hope of treatments – for the first time

Professor Dame Kay Davies CBE FRS FMedSci, Dr Lee's Professor of Anatomy, University of Oxford

Professor Dame Kay Davies is an award-winning human geneticist, renowned internationally for her work on Duchenne muscular dystrophy. An inspirational role model for women in science, Professor Davies is an MDUK Vice President, Deputy Chairman of the Wellcome Trust and Honorary Director of the MRC Functional Genomics Unit.

Significant successes

MDUK has funded Professor Davies' work at the University of Oxford over many years, and continues to do so today. In the 1980s, she developed a simple test to allow for the screening of pregnant women thought to be at risk of having a baby with Duchenne muscular dystrophy. One of her early successes, this continues to be extremely important and significant for families around the world.

Her work also led to the characterisation of the protein utrophin, a relative of dystrophin. Her research group showed that, in mouse models, increasing utrophin levels could prevent disease. More recently, Professor Davies has been pursuing this as an approach to treat Duchenne muscular dystrophy in humans. Unlike some existing therapeutics, effective only in a small number of patients with the condition, this strategy would be applicable to all patients.

Honours list

In 2008, Professor Davies was made a Dame Commander of the Order of the British Empire (DBE) in recognition of her achievements. She has published 384 peer-reviewed papers, won numerous awards and delivered special lectures at various institutions, and was awarded Honorary Membership of the Genetics Society in 2012. In 2015 she received the American Society of Human Genetics' annual William Allan Award, in recognition of her substantial and far-reaching scientific contributions to human genetics.

Neuromuscular research

Professor Davies first became involved in neuromuscular research in 1980. She had just returned to the UK from Paris, and was set to work in London on cystic fibrosis (CF) research.

"It was the early '80s, and the era of new genetics. The genes for most genetic diseases were unknown. We didn't know where the genes were in the genome. With CF, we knew nothing. For Duchenne muscular dystrophy (DMD), we knew that the causative gene was on the X-chromosome because primarily only males were affected. This was an important clue, and meant that developing technologies for DMD as a model system would help other disorders later."

Having heard of Duchenne muscular dystrophy but not knowing much about it, Professor Davies was gripped.

"It was at this point that I met Paul Walker (Chief Executive of what is now MDUK), then I met a patient with Duchenne muscular dystrophy, and since then I've never looked back.

Significant progress over the decades

"There have been four significant milestones in neuromuscular research since I've been involved: exon skipping, stop codon read-through, utrophin modulation, and AAV gene therapy.

"With all of these key developments, we are very close to getting effective treatments. The next step will be to think about combination therapy.

"We are going to see some significant advances not just for Duchenne muscular dystrophy, but in general. It's also exciting to see the big pharmaceutical companies involved too," said Professor Davies.

The impact of MDUK's funding and support on research

"MDUK got me into DMD research in the first place. My team and I could not have done it without MDUK, the networks they set up, and their help in getting patients on to clinical trials.

"MDUK's funding and support have had a huge impact on research. The charity has, from the outset, maintained a funding stream for basic science, which has underpinned the movement towards the promising technologies we are seeing today.

"This strategic approach gives scientists the opportunity to develop novel strategies to effectively treat the disease. We are seeing the rewards of that foundational research today.

"Over the years, patients and their families have become much more informed about muscle conditions because of the internet. MDUK's communication and information resources have grown in pace with that, and the charity has really been able to support families with helpful and valuable information.

"For any young scientists considering a future career in neuromuscular research, it is an exciting field. It is challenging and unbelievably rewarding to work with the charity, as well as the patients and their families.

"We are going into an era where progress is so fast. There is – for the first time – hope that we will be able to have effective treatments."

Read more about Professor Davies' and other research at www.musculardystrophyuk.org/research

Exon skipping: a therapeutic approach currently in clinical trial for Duchenne muscular dystrophy. It involves small pieces of DNA called 'molecular patches' which mask a portion of a gene where there is a mutation. The name comes from 'exon', which are the segments that genes are divided into, and the molecular patch causes the body to ignore or 'skip' over an exon.

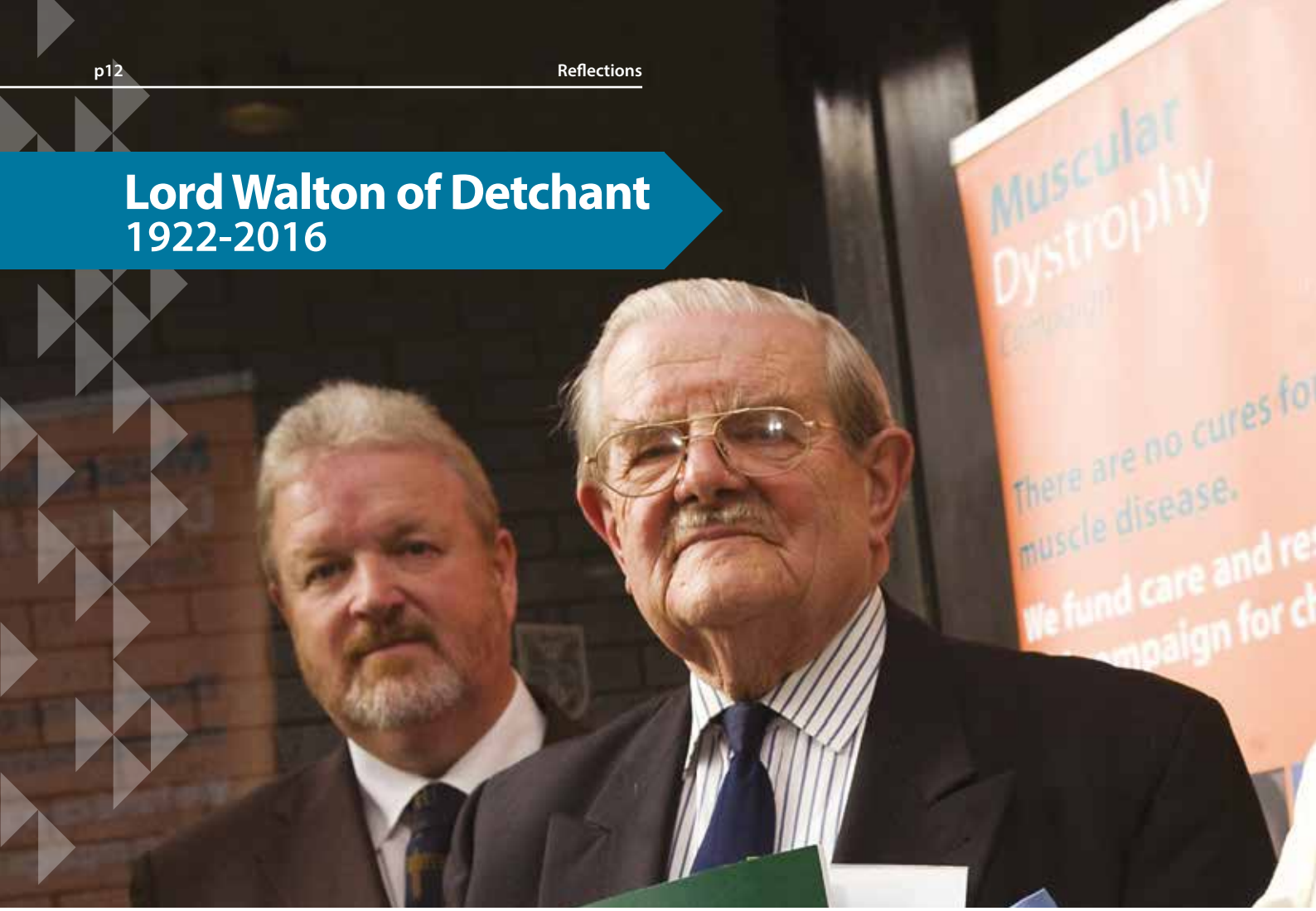
Stop codon read-through: cells read the DNA letters in sets of three (triplets), which are called codons. Within the DNA there is always a codon to tell the cell to stop 'reading' the gene (the stop codon). Some dystrophin mutations cause there to be a premature stop codon in the DNA sequence, so the cell terminates the process too early and the resulting dystrophin protein is unstable and degraded. Drugs like Translarna (ataluren) force the cell to continue 'reading through' the premature stop codon.

Utrophin modulation: tests new compounds to increase the level of utrophin protein, as a treatment for boys and men with Duchenne and Becker muscular dystrophies. Utrophin, a protein that occurs naturally in the body, is very similar to dystrophin in its structure, and has been shown to compensate for the lack of dystrophin in an animal model of Duchenne muscular dystrophy. Utrophin modulation is expected to be a disease-modifying approach in patients.

AAV gene therapy: Adeno-Associated Virus (AAV) is a small virus, which infects humans and some other primate species. It is not currently known to cause disease, and causes a very mild immune response. These features make AAV a very attractive vehicle for delivering genes into cells. To date, AAV vectors have been used in phase I and phase II clinical trials for the treatment of Duchenne muscular dystrophy, limb girdle muscular dystrophy types 2C and 2D, spinal muscular atrophy (SMA).

Through the dedication and generosity of our supporters, MDUK has been able to contribute significantly towards the ongoing development of each of these approaches. Find out how you can support pioneering research at www.musculardystrophyuk.org/research

Lord Walton of Detchant
1922-2016



theguardian.com
Newspaper of the year
Winner of the
Pulitzer prize
theguardian

Lord Walton of Detchant

Neurologist who improved treatment of muscular dystrophy

John Walton, Lord Walton of Detchant, who has died aged 93, was a neurologist who improved the diagnosis and treatment of muscular dystrophy. He headed several medical charities and raised substantial funds for them; and was a popular medical politician, as head variously of the General Medical Council, the Royal Society of Medicine and the British Medical Association. His publications included a classic textbook, Essentials of Neurology (1961), and books on disorders of voluntary muscle, brain haemorrhage, and the history of Duchenne muscular dystrophy, the commonest form of the condition. He also co-edited the Oxford Companion to Medicine (1986).

Walton was born in a Durham mining village, Rowlands Gill, to Eleanor (nee Watson) and Herbert, both Methodist teachers, who instilled in him the value of hard work. At medical school in Newcastle upon Tyne he was active in student politics, graduating in 1945 with a first and most of the prizes. After two years' national service as an army doctor, he returned to Newcastle, where he was inspired by two neurology greats, Fred Nattrass and Henry Miller, to study muscular dystrophy, at first thought to be a single condition that caused muscle wasting, but now known to be a group of linked genetic diseases.

Nattrass had been asked by the Department of Health to investigate why some patients diagnosed with muscular dystrophy recovered unexpectedly. Walton and he found that, rather than muscular dystrophy, some had polymyositis, a condition treatable with steroids. Walton's most important research publication was a 63-page landmark paper in the journal Brain in 1954, On the Classification, Natural History and Treatment of the Myopathies, which collated the research work to that date. Classifying neuromuscular diseases cleared the way to making the correct diagnoses in this complicated field.

Walton balanced a part-time neurology consultantship with medico-legal work and a substantial private practice. He was appointed professor of neurology at Newcastle University in 1968 and dean of medicine three years later. His reforms of the curriculum at Newcastle, integrating clinical medicine with basic sciences, were copied by other UK medical schools. In 1971 he was elected to the General Medical Council, on which he served for 18 years, the last seven as president. One of the few criticisms against him was that he failed to address the GMC's need for reform.

In 1959 he was a co-founder and chairman of what is now Muscular Dystrophy UK and for which he raised millions. He was also a patron of HealthWatch, which promotes evidence in healthcare.

Ennobled in 1989, he was an active crossbencher of the House of Lords for 27 years, taking the 6.30am train to London each Monday and returning to his home in Berwick on Thursday. He spoke in favour of using embryos in medical research and "three-parent" techniques, made major contributions to the legal frameworks for medical advances in rare diseases, neuromuscular conditions and mitochondrial research, and chaired scientific inquiries on topics ranging from antibiotic resistance to homeopathic treatments.

Walton was head of the General Medical Council, the Royal Society of Medicine and the British Medical Association Photograph: Muscular Dystrophy UK

In March this year, he had been speaking in the Lords on disability assessment and the junior doctors' dispute, shortly before he suffered two fits that resulted in the diagnosis of a brain tumour. Transferred to Royal Victoria hospital, Newcastle, he taught neurology to junior doctors from his bed.

Years earlier he had persuaded the Wolfson Foundation to fund an elderly care centre, Bell View, near Detchant, the Northumberland hamlet where the Waltons spent many summer holidays and where they retired. He spent his last weeks at home, supported by Bell View carers, and wrote to colleagues with his news. Six days before his death he had the pleasure of learning that the National Institute for Health and Care Excellence (Nice) had recommended ataluren, the first NHS drug to treat an underlying cause of Duchenne muscular dystrophy.

Soft-spoken, diplomatic, urbane and thoroughly nice, Walton was prodigious, punctilious and punctual. His wife said she had to make an appointment via his secretary if she wanted to discuss the children's education. His autobiography, The Spice of Life: From Northumbria to World Neurology, was published in 1993. He delighted in later life, when he took his former trainees round the House of Lords, in pointing out that his coat hook, labelled Walton of Detchant, adjoined that labelled Wales, Prince of.

He met Betty Harrison in chapel and married her in 1946; she died in 2003. He is survived by their daughters, Ann and Judy, their son, Chris, five grandchildren and 10 great-grandchildren.
Caroline Richmond

John Nicholas Walton, Lord Walton of Detchant, neurologist, born 16 September 1922; died 21 April 2016

Birthdays

Charlotte Bingham, novelist, playwright, 74; **Gary Busey**, actor, 72; **John Dawes**, rugby union player and coach, 76; **Amanda Donohoe**, actor, 54; **Baroness (Sally) Greengross**, chief executive, International Longevity Centre, UK, 81; **George Howarth**, Labour MP, 67; **Lord (Brian) Hutton**, Iraq investigator, former lord of appeal in ordinary, 85; **Katherine Jenkins**, mezzo-soprano, 36; **Prof Martin Jones**, archaeologist, 65; **Fran Kirby**, footballer, 23; **Michael McIntyre**, yachtsman, 60; **Rosa Mota**, marathon runner, 58; **Anne-Sophie Mutter**, violinist, 53; **Ian Paice**, rock drummer, 68; **Baroness (Usha) Prashar**, deputy chair, British Council, 68; **Mark Radcliffe**, DJ and musician, 58; **David Rudkin**, playwright, 80; **Nicole Scherzinger**, singer, 38; **Ruth Smeeth**, Labour MP, 37; **Matthew Weiner**, television writer, 51.

Announcements

DEATH NOTICES

BROWNE, Dr Elizabeth Foster, BM BCh 1949 Oxford; MRC Psych 1971; died on 16 June after a short illness, aged 91. A cremation service took place on 27 June in London. Service of thanksgiving at Holy Trinity Church, Grange in Borrowdale, at 3pm on Friday 1 July, followed by interment of ashes at St Andrew's Church, Borrowdale. Donations in Elizabeth's memory to Oxfam.

SHAW, Professor Ronald (Ron), of Cottingham, died on 21 June aged 86. Distinguished mathematician, tennis player and much loved father of Lizbie and grandfather of Jackie Ronald. He will be very much missed. Private committal, commemorative occasion to be arranged later. Donations in Ron's memory for Dignity in Dying may be sent along with inquiries c/o H Kemp and Son Ltd, hkempandsonltd@gmail.com.

Lord Walton, one of the founders of Muscular Dystrophy UK, passed away in April 2016. He was an outstanding Chairman of the charity from 1970 to 1994, after which he became an Honorary Life President.

He remained actively involved; a loyal and constant guiding influence on the development of the charity over the last 50 years.

"As a young doctor, John Walton was moved by the impact of muscular dystrophy on the children and families in his care. In later years, he often cited with real sadness, the words of the mother who had said to him, 'I watch my son die a little every day', as the boy's muscles inexorably wasted.

"He knew funds were desperately needed for research, and this led to his becoming a founder and driving force behind the charity. John would have been delighted to learn just weeks before the onset of his final illness, that Translarna, the first treatment to target an underlying cause of Duchenne muscular dystrophy had been approved by NICE."

Robert Meadowcroft, CEO

Lord Walton Fellowship Fund

Throughout his career, Lord John Walton gave great encouragement to many young doctors and researchers who recalled with warm recognition the part he had played in helping them get a first step on their career ladder.

In tribute and in honour of his significant contribution to the charity and to neuromuscular research, Muscular Dystrophy UK has launched the Lord Walton Fellowship Fund.

The Fellowship provides an important step for an outstanding young scientist or clinical researcher to advance their career. It has a clear focus on muscle-wasting conditions and will be located at one of the country's leading neuromuscular centres. The Fellowship is intended to help to accelerate the path to treatments.

To find out more or to contribute to the Lord Walton Fellowship Fund, please visit: **www.musculardystrophyuk.org/waltonfellowship**

Huge campaign win

After months of campaigning, driven by the support of families, access to treatments for muscle-wasting conditions is becoming a reality.

Muscular Dystrophy UK is thrilled to bring you the final 'yes'! NHS England and PTC Therapeutics reached an agreement in July, and NICE has signed off its final guidance: NHS England will fund Duchenne muscular dystrophy drug, Translarna.

This is history in the making: Translarna is the first-ever drug to target an underlying genetic cause of a form of muscular dystrophy. It will be used to treat children whose Duchenne muscular dystrophy is caused by a 'nonsense mutation', who are aged five and over and who are able to walk.

Sue Barnley, whose son, Harry (7), is eligible for the treatment, said: "After two years of uncertainty, Harry will finally be able to receive Translarna on the NHS. Giving our beautiful and cheeky boy extra time to play like other children his age is truly priceless."

What happens now?

The drug has now finally been approved for funding on the NHS in England. As part of the agreement, NHS England has waived their usual three-month waiting period, meaning the drug could be available within weeks.

As we go to press, MDUK has learned that some boys are about to get their first NHS treatments.

What about the rest of the UK?

We have been working with health ministers and officials in Northern Ireland (NI) and Wales. They have agreed, in NI, to follow NICE's guidance, and it is expected a similar decision will follow in Wales.

In Scotland, the process is entirely separate. The Scottish Medicines Consortium rejected the drug for overall approval. Along with families in Scotland, we are campaigning to secure NHS funding.

The drug has also not been approved for funding in the Isle of Man. Working with a family there, we're putting pressure on the Isle of Man government to reverse its decision.

For more information, please contact Peter Sutton on **020 7803 4838** or email **p.sutton@musculardystrophyuk.org**

www.musculardystrophyuk.org/translarna

Watching the changing landscape

Honorary Life President – J Alexander Patrick CBE DL

Alexander Patrick has been integrally involved in Muscular Dystrophy UK right from the start. His father, Joseph, was a founder of the charity; he established the Joseph Patrick Trust (JPT) in 1986, and their own Patrick Trust has been a generous and dedicated funder of the charity's work.

Inspired by his brother, Andrew, who had Duchenne muscular dystrophy, and named in memory of his father, Joseph, Alexander established the JPT to promote independence and quality of life for all people living with muscle-wasting conditions. JPT is Muscular Dystrophy UK's welfare trust.

"My younger brother, Andrew, had Duchenne muscular dystrophy and died in 1962, at the age of 13. While Andrew bore his condition very well and very cheerfully, there was little if any support or equipment available for him.

"During Andrew's life in the 1950s and early '60s, there was almost no support outside of the family. My father had to have an electric wheelchair made for him, because there was simply nothing available.

"Along with Lord John Walton and Professor Fred Nattrass, and with Andrew as his motivation, my father set up what is now Muscular Dystrophy UK, in 1959."

"Looking back, I remember when Richard Attenborough went to one of the charity fêtes and met a group of boys with Duchenne muscular dystrophy. That was in the early '60s and the start of his lifelong commitment to the charity.

"Watching the charity grow and develop over the years, it feels like we have gone from having great hope, to a lack of hope (when research appeared not to be fruitful) and back to great hope again.

"This is a result of the perseverance of all of our researchers and everyone who works hard to fundraise towards this groundbreaking research. It's encouraging to see how it's all starting to bear fruit now.

"Aside from the promise of this exciting research, the landscape has changed and improved tremendously for people living with muscle-wasting conditions.

"Today, we see many youngsters with Duchenne muscular dystrophy living into their 30s and 40s. When Andrew was a child, that hope was far on the horizon. There is also 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.

"I've been impressed with the charity's media coverage. There is something in the media about Muscular Dystrophy UK almost every day, all over the country, as well as some excellent national coverage.

"As a charity, we have certainly played our part to advance neuromuscular research, internationally, to where it is today."

Read more about JPT at **www.musculardystrophyuk.org/jpt**

An Honorary Fellow of Green Templeton College, Oxford, Alexander Patrick received a CBE in 2012 for charitable services.

Taking Stock of 52 years of support

Photo © Kit Studios

David Jackson MBE and his wife, Ann, (pictured far left and second left above) have been dedicated volunteers for Muscular Dystrophy UK since hearing Lord Attenborough speak at an event in 1965. Lord A's legendary enthusiasm for the charity he loved and supported drew them both in. The couple from Stock in Essex have never looked back.

"Soon after we were married, Ann and I went to a Round Table dinner in Rayleigh. 'Dickie' Attenborough was there to receive a cheque on behalf of the then Muscular Dystrophy Group. We were enthralled by his impassioned after-dinner speech – thanking the guests for the cheque, and telling us what the funds would mean for families living with muscular dystrophy. Ann and I turned to each other and decided there and then to help this charity in any way we could."

David and Ann established the Redbridge Branch in 1965 where they served as Chair and Secretary, respectively. In 1969, they moved to Brentwood, and remained with the Redbridge Branch until 1974 when they set up the Brentwood Branch.

They both remain involved as Chairman and Secretary of this Branch.

David has served on the charity's finance committee, as a trustee, and now as Chair of the Grants Panel for the Joseph Patrick Trust (JPT). Ann sat on the MedQuest committee, which selected merchandise and Christmas cards for the charity to sell.

With no family connection to muscular dystrophy, David and Ann have dedicated more than half a century to the cause. Their dedication and hard work have not gone unnoticed by the charity. Nor, it would seem, by the nation.

"In 2009, I received a letter from 10 Downing Street, advising me I was to receive an MBE (Member of the Order of the British Empire) in the Queen's birthday honours list. This was for voluntary service to Muscular Dystrophy UK. It was a real surprise, and an honour. I've always said it was awarded to both Ann and me," said David.

Later that same year at the National Conference, charity President Sue Barker OBE presented David with a Lifetime Achievement Award.

"Ann and I have remained involved in the charity ultimately to help others and to help raise funds so that groundbreaking research can continue. There have been tremendous advances over the 52 years.

"I have learnt a tremendous amount through my work with JPT. Receiving letters of thanks from the families we help is worth a million pounds to me. It's fantastic to know we are able to do that.

"The JPT was set up to provide equipment for people with muscular dystrophy. We meet three times a year as a panel to make the awards.

"We now also have some funds available for assistive technology too. We're keen to encourage more applications for these grants."

JPT grants – the impact on families

"It's made a massive difference. I'm now able to live a comfortable and independent life thanks to the JPT."

"The JPT has helped me fund a wheelchair to allow me to stand up, for the first time in my life. Nobody will ever understand how special this is!"

"My electric wheelchair is basically my legs, without it I would have to rely on people pushing me around in a manual chair. My electric wheelchair gives me so much more independence."

To find out more about the JPT's grants towards specialist equipment, please visit **www.musculardystrophyuk.org/jpt**

Muscular Dystrophy UK

Fighting muscle-wasting conditions

Gifts in Wills

For many children living with muscle-wasting conditions a simple smile and a comforting hug soon become impossible.

Muscular Dystrophy UK has been funding research to find effective treatments and cures, and helping families live with muscle-wasting conditions for over 50 years. Our work depends almost entirely on charitable donations, including gifts in Wills, voluntary donations and family fundraising.

If you would like to find out more about leaving a gift in your Will to Muscular Dystrophy UK, please contact Catriona Parker for more information:

020 7803 4834
legacy@musculardystrophyuk.org

Target Research

Welcome to the research pages of *Target MD*. My name is Jenny and I am the editor of *Target Research*. I have recently finished my PhD at University College London, where I was studying core myopathies.

I am very grateful for the PhD funding I received from Muscular Dystrophy UK and hope that I can put my neuromuscular research background to good use as your new editor.

In this issue, you will hear about some exciting new research projects that the charity is funding. There is also a roundup of the latest research news, including updates from clinical trials and summaries of new scientific research articles that have been published.

I very much hope that you enjoy reading these pages.
Jenny Sharpe,
Editor, *Target Research*

Muscular Dystrophy UK-funded research

New Duchenne research project

We are pleased to announce that through our Duchenne Research Breakthrough Fund, we are funding a new project in the laboratory of Professor Matthew Wood (on page 19) at Oxford University. Professor Wood and his team are investigating the use of molecular patches to prevent heart muscle disease (cardiomyopathy) in Duchenne muscular dystrophy. With cardiomyopathy being a major contributor to death in people with this condition, tackling this aspect will offer invaluable benefits.

To find out more about the Duchenne Research Breakthrough Fund, visit **www.musculardystrophyuk.org/duchenne-research-breakthrough-fund**

Clinical research training fellowship

In partnership with the Medical Research Council (MRC), we awarded a four-year clinical research training fellowship to Dr Claire Wood at Edinburgh University. Dr Wood is examining bone development in mouse models of Duchenne muscular dystrophy and testing whether bone growth can be improved by treating these mice with growth-promoters. This research could potentially address the growth problems and the weakening and thinning of bones (osteoporosis) that are common in people with Duchenne muscular dystrophy.

Clinical study on ACT

Muscular Dystrophy UK is co-funding a new clinical study called ACTMuS, which aims to find out whether a psychological treatment called Acceptance and Commitment Therapy (ACT) can enhance the quality of life of people affected by a muscle-wasting condition. The study is being led by Dr Michael Rose and Professor Trudie Chalder from King's College London and will focus on people with inclusion body myositis, limb girdle muscular dystrophy, facioscapulohumeral muscular dystrophy and Becker muscular dystrophy.

For more information please get in touch with Emilie Shore at e.shore@musculardystrophyuk.org

SMA research study published

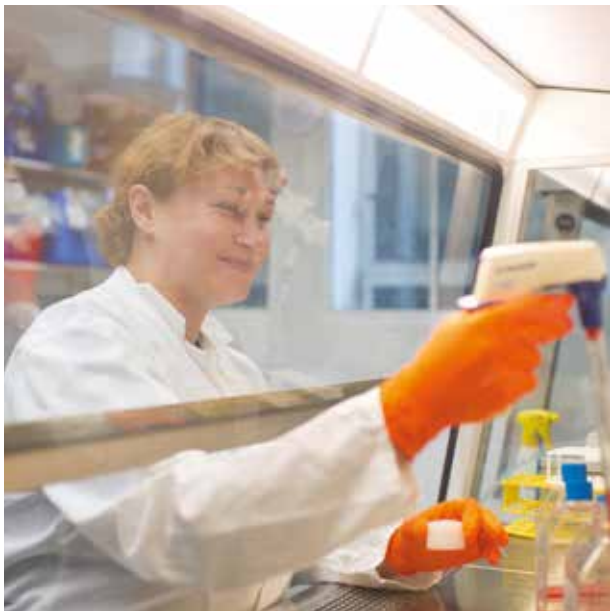
Muscular Dystrophy UK-funded researcher Professor Tom Gillingwater has published a scientific research study on spinal muscular atrophy (SMA). The study suggests that restoring levels of Survival Motor Neuron (SMN) protein in a group of cells that support the nervous system can significantly improve neuromuscular function in a mouse model of the condition. This finding suggests that targeting these types of cells, called Schwann cells, alongside other cells of the nervous system that are affected in SMA, may help to improve symptoms associated with the condition. This may be an important point to consider when developing new potential therapies for SMA.

Read more about research we are funding at:
www.musculardystrophyuk.org/current-grants

Update on clinical trials

SMA drug nusinersen

Biogen and Ionis Pharmaceuticals have announced that infants with SMA type 1, who have received nusinersen as part of the ongoing phase 3 trial called ENDEAR, have already experienced a statistically significant improvement in the achievement of motor milestones. The companies now intend to file for marketing approval with regulatory authorities in the coming months. Biogen is also working to open a global expanded access programme, which will allow all eligible infants with SMA type 1 to take nusinersen. We are seeking further information from the companies on implications for the UK and will share more details as soon as we have them.



Positive phase I results for myostatin inhibitor

The pharmaceutical company, Acceleron, recently announced the results of its phase I clinical trial testing ACE-083 in healthy people. ACE-083 is an investigational protein therapeutic that binds and blocks the action of a family of proteins that negatively regulate muscle growth (including the myostatin protein). The trial results showed that ACE-083 was safe, and significantly increased the volume of the healthy muscle that it was injected into. According to their recent press release, Acceleron now intends to advance ACE-083 into a phase II clinical trial for people with facioscapulohumeral muscular dystrophy (FSHD).

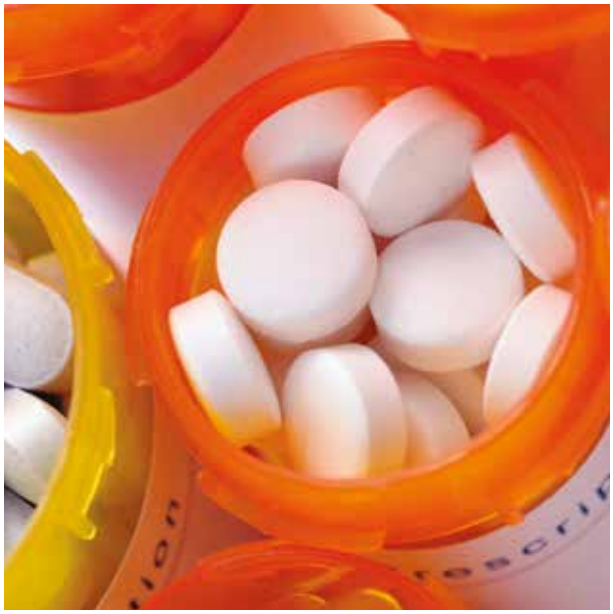
Raxone reduces respiratory complications in Duchenne muscular dystrophy

Santhera Pharmaceuticals have recently published new data from their phase III trial (called DELOS) testing Raxone in people with Duchenne muscular dystrophy, who were not taking steroids. The data showed that the group treated with Raxone had a reduced risk of respiratory complications compared with those in the placebo control group. Respiratory problems become more common as people with Duchenne muscular dystrophy get older so tackling this aspect of the condition will offer invaluable benefits. Although Raxone was only tested in patients not taking steroids, Santhera are planning another phase III study (called SIDEROS) to test its effectiveness in patients using steroids. The active substance in Raxone is called idebenone, which improves energy production in mitochondria within our cells and also has anti-oxidant properties.

Following the promising results from their phase II (DELPHI) and phase III (DELOS) studies, Santhera have submitted a Marketing Authorisation Application (MAA) to the European Medicines Agency (EMA) for Raxone (idebenone). The company expects an opinion from the EMA's expert review committee in the first quarter of 2017.

BioMarin withdraws application to license exon skipping drug in Europe

We are sorry to report that BioMarin is discontinuing the clinical and regulatory development of drisapersen (Kyndrisa), as well as three other exon skipping drugs, BMN 044, BMN 045 and BMN 053. This decision follows negative discussions at the May meeting of the EMA's Committee for Medical Products for Human Use, as well as the rejection of drisapersen by the US Food and Drug Administration (FDA) in January.



The company has confirmed that they will continue to develop second-generation exon skipping drugs for Duchenne muscular dystrophy, though this research is at the early stage. We are following up with the company to see what their plans are in relation to the discontinued drugs, and whether trial participants could continue to access them through compassionate use programmes.

Fast track designation for CMD drug

Omigapil, which is being developed by Santhera Pharmaceuticals as a potential treatment for congenital muscular dystrophy (CMD), has received 'fast track' designation from the FDA. This fast track status will help accelerate the process of getting the drug to patients. Omigapil inhibits a cellular signalling pathway that leads to the death of muscle cells. It is currently being tested in a phase I clinical study called CALLISTO.

Update on eteplirsen

Sarepta Therapeutics recently announced that the FDA had requested to see data already obtained from eteplirsen's ongoing confirmatory study (called PROMOVI). This request is part of the FDA's continuing assessment of eteplirsen as a treatment for Duchenne muscular dystrophy. The FDA want to know how effective the drug is at increasing dystrophin levels in the muscle and so have asked to see muscle biopsy data measuring dystrophin levels.

MDUK strongly supports FDA approval of eteplirsen. We are in contact with Sarepta on their future plans to seek approval for the drug here in Europe.

More efficient exon 51 skipping drug taken forward into clinical trial

We are pleased to report that WAVE Life Sciences are taking their exon 51 skipping drug candidate forward into clinical trial. The trial is expected to start in the second half of 2017. The company reports that their compound is 25 times more efficient than other exon 51 skipping drugs, drisapersen and eteplirsen. The compound also showed a broader tissue distribution in animal models of Duchenne muscular dystrophy, reaching critical tissues such as skeletal muscle, heart and diaphragm. This is exciting news and we are delighted that the work has been conducted in close collaboration with Professor Matthew Wood (pictured below) from the University of Oxford. Muscular Dystrophy UK has supported Professor Wood's research into exon skipping for many years.

For more information on these clinical trials, please see the breaking news in research page on our website: www.musculardystrophyuk.org/progress-in-research/breaking-research-news

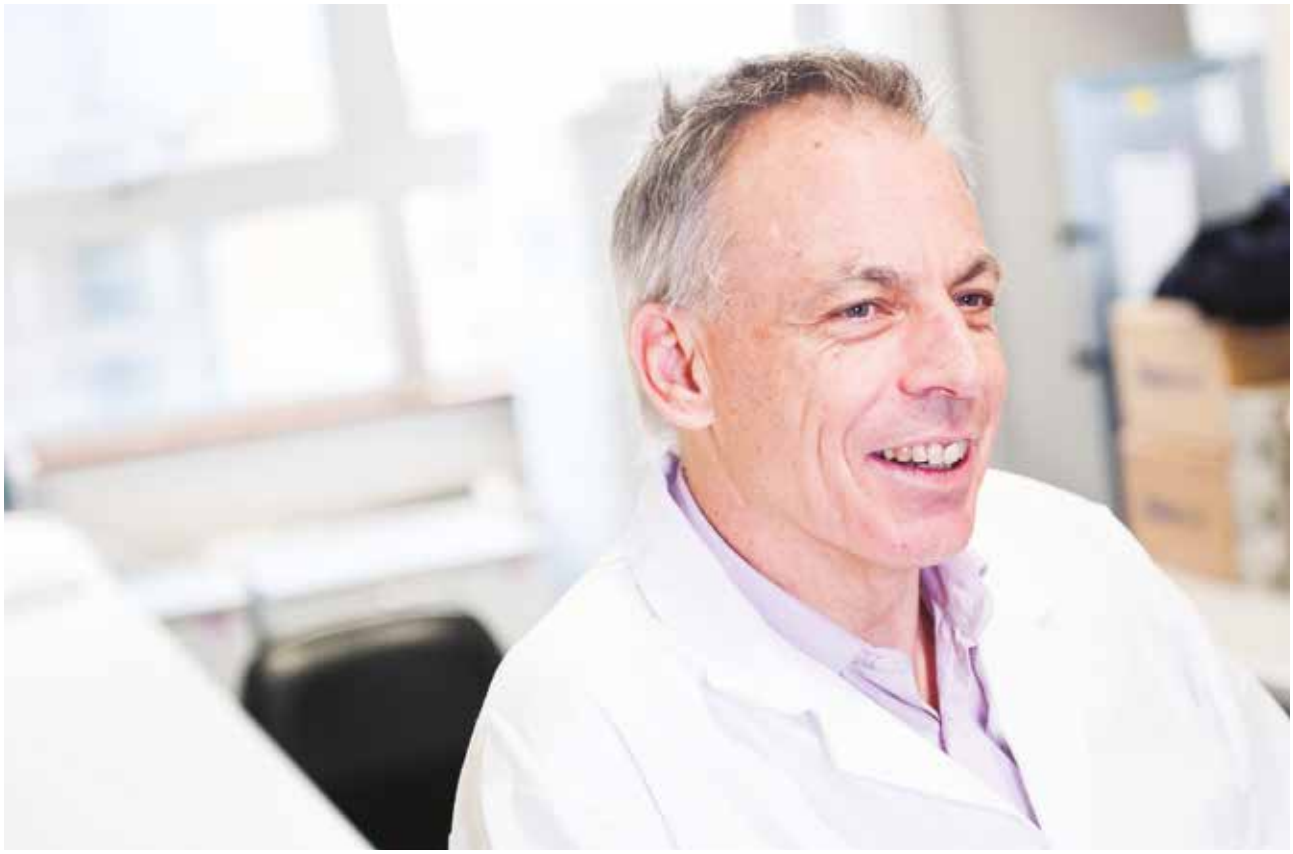


Photo © Kii Studios

Scientific research articles

Pain in young people with Duchenne muscular dystrophy

A new study carried out by researchers at the University of Central Lancashire, and clinicians at the Royal Preston and Alder Hey Hospitals, has made progress in the understanding of pain in young people with Duchenne muscular dystrophy. Interviews with 12 boys and young men with the condition, along with a survey of their parents or guardians, revealed that many of the young people experienced pain. However, they often keep this to themselves or only share this with close family members. The study also showed that participants who experienced more frequent or severe pain had a reduced quality of life.

These findings highlight the need for medical teams to be more proactive in enquiring about pain in people with Duchenne muscular dystrophy. To help raise awareness within the clinical community, we have circulated the results of this study to our network of health professionals.

New insights into Emery-Dreifuss and limb girdle muscular dystrophies

Researchers at the University of Edinburgh have discovered that nuclear membrane proteins play a role in the development of the muscles in the body. These findings help give a better understanding of what is going wrong in muscular dystrophies where the nuclear membrane is affected, such as in Emery-Dreifuss muscular dystrophy and some limb girdle muscular dystrophies. This knowledge is important for the development of future treatments for these conditions, which currently have very limited treatment options.

Mitochondrial donation IVF

Researchers at Newcastle University have published a new study on the safety and efficacy of mitochondrial donation IVF (see Figure 1). They have significantly refined the procedure and shown that it can reduce the risk of mitochondrial disease being passed from mother to child. Although mitochondrial donation IVF does not guarantee the prevention of mitochondrial disease, understanding of the risk involved is vital for helping prospective parents to make informed reproductive choices.

One of the researchers involved in this study is our Vice President, Professor Sir Doug Turnbull, who has recently been awarded a knighthood for his contribution to mitochondrial disease research. With the help of our supporters, Muscular Dystrophy UK funded Professor Turnbull's early research to develop mitochondrial donation IVF. We are very pleased that his outstanding work has being recognised in this way and look forward to future advancements of mitochondrial donation IVF.

Figure 1: In vitro fertilisation (IVF)

Therapeutic effects of new drug on IBM

New research shows that a drug called arimoclomal may have therapeutic effects for people with sporadic inclusion body myositis (sIBM). Arimoclomal aids protein folding inside our cells, which is thought to be disrupted in sIBM. In collaboration with a group of scientists in the USA, researchers from the MRC Centre for Neuromuscular Diseases in London showed that arimoclomal reduced the signs of sIBM in rat muscle cells grown in the laboratory. The researchers also carried out a small-scale clinical trial and found that the drug was safe and well-tolerated by people with sIBM. The researchers are now planning a larger trial to assess the effectiveness of the drug.

Measuring the amount of utrophin in muscle

UK scientists have developed new staining and imaging techniques to measure utrophin levels in muscle biopsies from people with Duchenne and Becker muscular dystrophy. With utrophin-modulating compounds already in clinical trial, it is crucial to have reliable tools to assess how well these compounds work. These new tools will be invaluable in assessing utrophin modulator activity in future clinical trials.

Read more at: www.musculardystrophyuk.org/progress-in-research/news

It is your generous support that enables MDUK to fund pioneering research. We cannot do this without you. Find out more at www.musculardystrophyuk.org/get-involved

Take a leap and Make Today Count

Come and skydive in Muscular Dystrophy UK's Make Today Count event in 2017. By the time you land, you'll have funded nine hours of groundbreaking research.

Our 2017 event got off the ground in early August. There are dates in late February and March 2017, at a range of sites across the country. When you sign up, you'll join hundreds of supporters across the UK also jumping out of planes for MDUK.

The early bird registration offer of just £29 (for a limited time only), along with the £399 we ask you to fundraise, will make the day count. It will fund nine hours of groundbreaking research into muscle-wasting conditions.

And you'll join our Make Today Count yearbook: since 2012, an incredible 760 brave skydivers have taken part and raised a huge £485,905. That's enough to fund over 11,000 hours of research!

"I decided to sign up for Make Today Count 2016 for my friend Ryan, who has Duchenne. A group of us got together to skydive as Team Ryan. The experience was exciting and nerve-wracking, and I'm so pleased I did it. I'd never been on a plane before, and I was jumping out of one for charity!" Thomas, from West Yorkshire

Sign up today at **www.musculardystrophyuk.org/maketodaycount**

Photo © Kay family

The adventures of Bertie's Buccaneers

Alison and Matt Kay live in Southport, near Liverpool, with their children Archie (10), Emmeline (7) and five-year old Bertie (pictured above). The children loved seeing the Giants when they visited Liverpool recently. They watched the huge puppet Sophie and her dog walk through the streets, and then get on a boat to sail away.

So it made sense that when setting up a Family Fund earlier this year, the family chose to call it Bertie's Buccaneers.

"We may not like it but our son has a rare condition for which there is no cure. It's a feeling we liken to being adrift in a vast grey ocean that's going to overwhelm us.

"In this situation, you either build a raft together and paddle for land like there is a shark behind you, or you give up and sink.

"Bertie's Buccaneers is our raft, and this is our adventure," said Alison.

Diagnosis

"Bertie was diagnosed with a collagen VI-related myopathy at the age of two. He uses a wheelchair to get around, although he can still walk inside the house.

"He has hyperlaxity (as well as weak muscles), which means his fingers and joints are super-bendy. Pushing buttons, opening things and holding a pencil are all really hard for him. He's doing well, though, and always has a great day at school. He's just finished his Reception year.

"Diagnosis was a very drawn-out affair for us, with numerous referrals, long periods of waiting and uncertainty. It felt like we were lost in the system.

Let down by the system

"The bottom had fallen out of our universe. Instead of the process we expected for a sick child, we were left hanging, and we felt really let down and abandoned by the NHS. I don't think I could describe in words what that period of our life felt like. It would be just too hard.

"We appreciate that the system is under considerable strain. Nonetheless, we don't believe the diagnostic experience we had was acceptable.

Invaluable support

"It was around this time that we discovered MDUK on the internet. We were trying to find out what muscular dystrophy was, and what it would mean for Bertie and for us as parents.

"The support we've had from the MDUK advocacy service has been invaluable. We had a very negative experience trying to access the Disabled Facilities Grant (DFG) for Bertie through our local authority. They advised us to install an unsuitable lift that in the end wouldn't fit, so eventually we moved house. We found the perfect house for a ground floor adaptation, started the DFG process and again hit a brick wall.

"We tried to reason our way through this bureaucratic process but it was all very stressful. The advocacy team at MDUK sought some clarity on our behalf and lent us a listening ear.

Photo © Kay family

"Ultimately, our MP had to come on board before our ground-floor adaptations were approved.

"We started this process when Bertie was two. He's now five, and we hope he'll have an adapted bathroom and bedroom by Christmas. Without MDUK, we probably still wouldn't be any nearer to getting the adaptations Bertie needs.

Fundraising for rare conditions

"Initially Bertie's Buccaneers was just a Facebook page, to help us raise awareness of Bertie's condition and to keep friends and family informed. Over time, as we became aware of what a shockingly small portion of the overall UK charity pie went to muscular dystrophy research, we decided to fundraise.

"Muscular dystrophy is rare; hardly anyone we shared Bertie's diagnosis with seemed to have heard of it. If they had, it was usually Duchenne. We found ourselves having to explain the difference a lot.

"We heard about Abbi [Bennett] and Dan [McLellan] and their fantastic Family Funds. So we decided to join them and do what we could to support the Ullrich CMD fund too.

Personal touch, national charity

"Our regional development manager helped us set up Bertie's Buccaneers. He has a wealth of experience and insight, and is always happy to share with us. This personal touch is very important; ultimately we're still juggling working/family life and a child with a disability. It's not a laid back lifestyle!

"And, on a practical level, having a national charity behind you is very helpful for eye-catching branding and validating your fundraising. Being a Family Fund helps in giving structure to what we're doing and we feel part of a broader team mission. MDUK gains too because the personal, local story is always going to attract more empathy.

"We work with MDUK, who makes sure all fundraising supports meaningful research and clinical trials. And that's the only way to fix Bertie's muscles."

Parallel London

"We were so excited when we heard about Parallel London on social media! It's the world's first fully-inclusive fun push/run. How fabulous to have a fundraising event that Bertie can take part in himself!

"It is going to be such an amazing experience for Bertie to visit the Queen Elizabeth Olympic Park. He'll meet other children and adults with disabilities in a totally positive and empowering environment. Bertie won a place in the open ballot, but we're fundraising for MDUK – he's hoping to raise £1,000."

A Family Fund is a great way to fundraise for Muscular Dystrophy UK, as a family or group. We look after the admin and paperwork for you, so you can concentrate on fundraising. Contact us on **volunteerfundraising@musculardystrophyuk.org** or **0300 012 0172** to find out more.

www.parallellondon.com/get-involved

Support Bertie's fundraising here: **www.justgiving.com/fundraising/Bertiesbuccaneers**

Bertie's condition

Collagen VI-related disorders

This is a spectrum of conditions, including Bethlem myopathy (at the milder end of the spectrum) and Ullrich congenital muscular dystrophy (at the more severe end). These conditions are caused by mutations in genes that carry the instructions to produce collagen VI protein, leading to a deficiency of this protein.

Collagen is the main structural protein making up connective (supportive) tissue. There are several different types, found in various parts of the body, including the skin, tendons, hair and nails. The type of collagen found in muscle tissue is called collagen VI and this acts as a scaffold to hold and support muscle cells.

Photo © Kay family

Fundraising updates

To 100 collections and beyond

Our resident superhero, Phil Grant has raised over £16,000 towards the work of the charity. Dressed as a St Trinian, Ginger Spice, Snow White, a cowgirl, a ballerina, a fairy, a nurse, and his trademark Wonder Woman, Phil has done collections, fun runs and faced his fears to raise funds. The results are clear: his outfits draw attention, get people talking and then donating.

The inspiration behind all these superhero feats? Phil's son, Chris (29), who was diagnosed with Duchenne muscular dystrophy at the age of three.

"When Chris was little, we had noticed he wasn't at the same level, physically, as other children his age. It took a year to get the diagnosis," said Phil, who lives in Oxfordshire with his wife, Debbie, and younger son, Michael (28).

"We got involved with MDUK, as we wanted to do something to help raise funds and awareness. We started off doing car boot sales, fun runs, table top sales.

"The first two fun runs I did in ordinary clothing, and raised some money. The third time, I ran as a St Trinian and raised a lot of money. Because I was wearing a costume, people would notice me and ask me questions. I definitely raised more money this way."

And so began his run of events dressed in fun and colourful outfits. A few years ago, he started doing collections at the annual Comic Book Convention, and in May this year, Phil did his 100th collection – at the Oxford Town and Gown 10k.

"It's great to do collections at the Town and Gown events. I can get loud, egg people on, get people motivated at the starting line, move through the crowds and high-five as many people as I can!"

Phil never misses an opportunity to leap out of his comfort zone. Earlier this year, he took on his scariest challenge yet.

"I'm really afraid of heights. I could never jump out of a plane, but when I heard about indoor skydiving, I thought I'd give it a try. If I do something that really pushes my limits, it's easier to ask people to support me. It was really scary."

"I still have about 10 more collections to do before Christmas. I have already raised over £2,500 this year – bringing my total to more than £16,000."

For his sterling efforts, Phil received a Charity Champion Award 2014 from President Sue Barker OBE, and met Prince Philip at the celebration event in June.

Chris, who Phil says 'enjoys life', is happy that Phil raises the money for the charity. It seems this superhero won't be hanging up his cape any time soon.

We rely on the support of fundraisers like Phil to fund our vital work to beat muscle-wasting conditions. Find out how you can take on similar challenges, or do collections in your local community, to raise funds and awareness. Contact us at volunteerfundraising@musculardystrophyuk.org or 0300 012 0172.

Oxford Town and Gown 10k

This year's Oxford Town and Gown 10k on 15 May, saw over 4,000 adult and 200 junior runners take on the 35th annual Town and Gown 10k. Thanks to their hard work and dedication, our #TeamOrange runners raised over £151,000. This will help fund groundbreaking research and vital care and support for people with muscle-wasting conditions.

Dutch physicist, Luke Metselaar, won the race in 31:35. "I thought there might have been a chance that I would win, so I am very pleased. It's good to encourage people to get outside and get into running. It's good for the charity too."

How to join #TeamOrange:

- register for the 2017 event in Oxford
- sign up for the 2016 Cambridge Town and Gown 10k on Sunday 16 October
- keep a look out for our brand new Town and Gown 10k in 2017 too.

You'll find all the info you need on our website www.townandgown10k.com

London Marathon

Congratulations and thank you to the 110 incredible #TeamOrange runners who took on the London Marathon on 24 April. Thanks to your epic fundraising efforts, you raised a massive £290,000.

Volunteers and supporters, along with MDUK staff, cheered the runners on at Tower Bridge and Embankment. After crossing the finish line, our runners were treated to a well-deserved massage and shower at their VIP post-race celebration.

"I am so proud to say I was a part of this year's #Team Orange! The reception I got after the race nearly had me in tears!"
Geraldine

"What a day. What a team. What a charity. Incredible effort all round, incredible support from all MDUK runners and staff. Buzzing right now!"
Alex

Apply for the London Marathon 2017 at **www.musculardystrophyuk.org/londonmarathon**

Running for Evan

Over 140 runners took on the Belfast Marathon for MDUK in May, 100 of whom ran in memory of Evan McCrotty. The eight-year-old boy from Derry lost his life, alongside members of his family, at Buncrana Pier in March.

Evan had Duchenne muscular dystrophy. His mum, Louise, and Davitt Walsh (pictured right), who saved Evan's baby sister, Rioghnach-Ann, were among family, friends and members of the community running together as Team Evan. The Team ran in specially designed T-shirts featuring Evan and his big brother, Mark (12), who also lost his life.

Team Evan raised £14,500 towards our Duchenne Research Breakthrough Fund, which funds the best scientific research into the condition.

"I want to thank every single person who took part to remember Evan, whether you ran the race or put your hand in your pocket to donate. You did something remarkable in his name and to celebrate his life. I know he would be proud.

"Duchenne muscular dystrophy is a heart-breaking condition. For the hundreds of young people who live with it today, we need to raise awareness. We need to help make sure that in the future, children and their families don't face the same fears and challenges we did."
Louise, Evan's mum

MDUK would like to say a huge thank you to Louise for this wonderful and generous gesture.

RideLondon

Twenty-eight heroic cyclists tackled the Prudential RideLondon-Surrey 100 event on Sunday 31 July for MDUK. Alongside 25,000 other cyclists, they completed the demanding 100-mile route, starting at the Queen Elizabeth Olympic Park and finishing on the Mall. After conquering the infamous Leith and Box hills along the way, the team enjoyed a special celebration picnic at the end with friends and family.

This #TeamOrange, all of whom are all affected by muscle-wasting conditions in some way, are set to raise £25,000.

"This was such a brilliant event. Being new to cycling events, it was tough for me. But the crowds of people kept me going. You can see why they call it the London Marathon on wheels!"
Krishan Solanki

"What a feeling! It was tough, but knowing what Sophie has to deal with every day, for such a little girl, spurred me on."
Tony Caruso

Register for RideLondon 2017 at
www.musculardystrophyuk.org/ridelondon

www.medicotech.co.uk

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Charity updates

Ambulance action

"My respiratory problems can be frightening. It's a huge relief to know that a medical team will have full access to my medical records in an emergency."

Ravi Mehta (26) has Duchenne muscular dystrophy. He's relieved to know that Muscular Dystrophy UK has launched an exciting new partnership with the London Ambulance Service. The 'Ambulance Action' scheme will support people like him – living with muscle-wasting conditions – in cases of emergency.

In the scheme, neuromuscular specialists will use the 'Co-ordinate My Care' database to provide the London Ambulance Service with names and contact details of their patients who have muscle-wasting conditions. This means that if Ravi has to call emergency services (999 or 111), his call will automatically be 'flagged' as a high priority case. Paramedics will also have access to his individual care plan, which means he'll get the emergency care he needs in relation to his condition.

"It's difficult in emergency situations to tell paramedics about my complex and specific medical needs. With this new system, I won't have to do that and paramedics will know what I need.

"I hope this scheme will make a difference to people like me," said Ravi.

The scheme is currently only available in London. There is a plan, however, to work with regional ambulance trusts to roll it out across the UK.

The 'Ambulance Action' scheme is part of Muscular Dystrophy UK's work to improve emergency care. This includes the launch of 11 condition-specific alert cards, which outline key aspects of specific muscle-wasting conditions, for people to share with health professionals.

To find out more, contact Lloyd Tingley at **l.tingley@musculardystrophyuk.org** or on **020 7803 4804.**

Brexit – what does it mean for research?

"There are many questions that need to be answered regarding the implications of the EU referendum. What does this mean for Muscular Dystrophy UK, and the fight to accelerate treatments and ultimately cures for muscle-wasting conditions?"
Robert Meadowcroft, Chief Executive of Muscular Dystrophy UK, following the UK's vote to leave the EU in the recent referendum.

"It goes without saying that during this period of uncertainty we will continue to put the interests of people living with muscle-wasting conditions first. It is essential we maintain and accelerate progress towards effective treatments, improved care and support; this remains our priority. The fight goes on as before."

Potential risks

Other important considerations we'll be making include:

- continued access to the EU's Research and Innovation Horizon2020 funds and future funding programmes after Brexit
- whether European Medicines Agency (EMA) conditional approval for new treatments would still apply to the UK
- the harmonising of regulations and data-sharing initiatives for clinical trial development and patient registries
- the position of the UK in relation to European Reference Networks
- how to retain and attract the best researchers.

Potential opportunities

We'll be exploring:

- funding opportunities and collaborations across the rest of the world
- welcoming the best researchers from the rest of the world
- maintaining the UK's definitive position on innovative research.

For more information, please email **info@musculardystrophyuk.org** or call the Muscular Dystrophy UK freephone helpline on **0800 652 6352.**

Charity updates

New networks of excellence

Following the first national audit of managed clinical neuromuscular networks, Muscular Dystrophy UK has awarded Network of Excellence status to the Scottish Muscle Managed Clinical Network and the South West Neuromuscular Operational Delivery Network.

A neuromuscular network is a group of hospital trusts and healthcare providers who work together to improve NHS services across a region or country. By pooling together resources and teams, they can share expertise and close any potential gaps in care. In this way, centres of all sizes can effectively promote and develop best practice in the diagnosis, care and management of patients with muscle-wasting conditions.

By awarding Network of Excellence status, we are for the first time recognising the important role of networks within neuromuscular provision. We are also congratulating those that provide a full and comprehensive service to a wide range of people with muscle-wasting conditions.

This builds on MDUK's work to recognise Centres of Excellence across the UK. In setting a marker for clinical excellence, we are helping to raise standards of neuromuscular services and recognising the networks and centres that are leading the way forward. This all serves to improve standards of care for individuals and families with muscle-wasting conditions.

Info and resources

Help with PIP

Michelle's PIP assessment last year left her without access to a Motability vehicle. With the support of MDUK's advocacy team, her case was reviewed – she'll now get both a vehicle and her independence back.

"Receiving my PIP decision that I had 'won' my appeal initially confused me. They had decided at the very last minute that I didn't even need to attattend! They awarded me the enhanced rates for both care and mobility, which was what I had hoped for. But to have the term determined as 'indefinite' was a pleasant and very welcome shock!"

Are you going through the process of claiming for PIP, or challenging a decision you've received? For help and advice, contact our friendly advocacy team.

Resources for recently diagnosed families

"It's at times like this, I am so grateful there are people like you to call. Thank you again."
Anne called the MDUK helpline soon after her four-year-old nephew had been diagnosed with Duchenne muscular dystrophy.

Our care and support team chatted to her on the phone, emailed her a Duchenne factsheet, and posted her a pack of useful info for families facing a new diagnosis.

If you or your family are facing a new diagnosis with a muscle-wasting condition, don't do so alone – call our care and support team. They have time, they have info you can trust, and they have networks of people to put you in touch with.

Alert cards

"The alert card is one of the best things Muscular Dystrophy UK has done. It's a perfect summary of my condition and makes it so much easier for not only doctors to understand my needs, but friends and family too."

Margaret Wyman, from Dorset, who has Charcot-Marie-Tooth disease (CMT), received one of the more than 8,000 alert cards MDUK has sent out to

people with muscle-wasting conditions, as well as health professionals. Visit our website to find out which cards are available.

These cards, which can fit in your wallet, are currently available for 13 conditions, including undiagnosed muscle-wasting conditions. They have important info on them about the conditions to help non-specialist health professionals know what to do, in an emergency.

To order an alert card for yourself or someone you know, contact us.

Free information

MDUK offers support and free information to anyone living with a muscle-wasting condition. New information publications include:

- A guide for parents: children with muscle-wasting conditions
- A guide for parents: children with Duchenne muscular dystrophy
- Financial and practical support: find out what you're entitled to

Our Information Service publishes a wide range of information factsheets on muscle-wasting conditions, as well as welfare and benefits. These have been awarded the Department of Health's Information Standard – a quality mark that means it is information you can trust.

Call Freephone **0800 652 6352** or email **info@musculardystrophyuk.org**
www.musculardystrophyuk.org/alert-cards

National Conference

Victoria Plaza, London

Saturday 1 October 2016

This year's National Conference programme is full of research presentations, interactive workshops, the annual President's Awards and an all-day crèche for children up to the age of 16. Add in the opportunity to meet other families living with muscle-wasting conditions, and you won't want to miss this.

You'll also get to hear about:

- progress in gene therapy research and exon skipping
- psychological support
- exercise
- physiotherapy
- accessible tourism
- emergency care.

Professor George Dickson, from Royal Holloway, University of London, will explain the concepts of gene therapies for neuromuscular disease.

"I'll also talk about the amazing technical and practical progress we've made towards getting these powerful new medicines into the hospital pharmacy, and safely to the bedside for doctors to test and use.

"My gene therapy research relates specifically to Duchenne, Becker and OPMD. But we hope the discoveries and technical advances will be applicable to many other conditions, such as limb girdle muscular dystrophy and FSHD.

"Never before have there been so many prototype medicines in clinical trial. It is a time of great endeavour in clinical research, and of great optimism and hope."

Specialist equipment – what's best for me?

Kirstie Spencer, a care advisor based at the Nottingham University Hospitals NHS Trust, will lead a workshop on specialist equipment. A trained occupational therapist (OT), Kirstie will be on hand to chat to you about specialist equipment for your specific needs. She'll also tell you how to go about getting funding for it.

The Young Person's Conference

On the same day, and at the same venue, the Trailblazers' conference is geared specifically for young people. You'll have the chance to meet other young people living with muscle-wasting conditions, and talk about employment, education, working with the media, and accessible tourism.

Adults – £15 / Concessions and children – £5 / Family ticket – £35
Costs include lunch, morning and afternoon refreshments

To book, visit **www.musculardystrophyuk.org/nationalconference2016**
Contact Lyn at: **lyni@musculardystrophyuk.org** or **01132 301313**

Scottish Conference

Beardmore Hotel and Conference Centre, Glasgow

Saturday 22 October 2016

At the Scottish Conference, you'll get to meet up with old friends and make new ones, and hear about the latest in research and clinical care.

Adults – £10
Concessions and children (over 12) – £5
Family ticket – £20
Costs include lunch, morning and afternoon refreshments

The day includes:

- neuromuscular care advisor and advocacy surgeries
- research, clinical care and daily living workshops
- a full programme of children's activities
- the presentation of the Inspire Awards.

To book, visit **www.musculardystrophyuk.org/scottishconference2016**
Contact Lyn at: **lyni@musculardystrophyuk.org** or **01132 301313**

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