Muscular Dystrophy UK

Fighting muscle-wasting conditions

Inside

- Spinraza update
- New tools to help beat UCMD
- Achieving life goals with Powerchair football

# Campaign

**The newsletter for our supporters**

# Ready and able

**In May this year, Trailblazers – MDUK's 800-strong national network of young disabled campaigners – held a reception at the House of Commons hosted by Heidi Allen MP to launch their new *Ready and able* report.**

The report was a result of speaking to young disabled people and employers over an 18-month period to identify what needed to change to improve things in the future for disabled job-seekers. As well as highlighting the barriers that make it difficult for young disabled people to find and stay in work, the report calls on Government and employers to take action to remove these barriers.

On the day, Ms Allen spoke about her work with the All Party Parliamentary Group for Young Disabled People and her commitment to getting more disabled people into work.

**Go to page 10 to find out more about the Trailblazers' valuable campaigning work.**

MDUK Employability Officer, Emma Vogelmann with Heidi Allen MP

# Welcome

I'm delighted to welcome you to the autumn edition of *Campaign* – the newsletter that shows you what your kind support of Muscular Dystrophy UK's work makes possible.

On page 8 and 9, you'll read about how England Powerchair football team member, Marcus Harrison discovered the sport and what it means to him now.

You can also read about a momentous landmark in MDUK's fight to beat muscle-wasting conditions and how our tireless campaigning work has helped ensure more people than ever will have access to Spinraza – the first treatment for spinal muscular atrophy (SMA).

I hope you enjoy reading about all the ways that your incredible support is making a difference to those living with muscle-wasting conditions. We really are so grateful.

**Gabby Logan**
**President, Muscular Dystrophy UK**

**PS** Visit the MDUK online shop to see the range of fun and festive Christmas cards and gifts – you're sure to find something you love!
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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On the cover: Marcus Harrison (left) of West Bromwich Albion PFC and Jon Bolding (right) of Aspire PFC go head-to-head in an MDUK Premiership match.

# Spinraza set to be available for patients with SMA Types 1, 2 and 3

Zac, (now four) who has SMA Type 1, with his mum, Amy

**Spinraza – the first treatment for the rare condition spinal muscular atrophy (SMA) – is set to become available on the NHS for children and adults with SMA Types 1, 2 and 3. This could be life-changing for the 1,300 people believed to be living with SMA in the UK.**

NICE, NHS England and Biogen (the pharmaceutical company who produce the drug), have agreed to make the treatment available through a scheme known as a Managed Access Agreement (MAA). This means that patients will be able to access Spinraza while more long-term data on its effectiveness is gathered. Without the hard work and collaboration between MDUK, clinicians, SMA charities, MPs and, above all, people with the condition and their families, this important progress would not have been possible.

Amy Cameron, mum to four-year-old Zac (pictured opposite) who has SMA, lives in Scotland where Spinraza is already available for people with SMA Types 1, 2 and 3.

Amy says:

**"Spinraza gives Zac the best possible quality of life that he can have right now. For us it really has been a miracle to have access to that treatment and families in other parts of the UK need this drug now."**

But our work isn't finished. MDUK and Spinal Muscular Atrophy UK are calling for Biogen and NHS England to urgently implement the MAA so patients can access the drug quickly and will work to make sure Wales and Northern Ireland follow the approach taken by Scotland and England in approving Spinraza.

**To find out more about our campaigning for faster access to Spinraza, visit: www.musculardystrophyuk.org/spinraza**

## What is SMA?

SMA is a rare, inherited muscle-wasting condition caused by a lack of an important protein called survival motor neuron (SMN) protein, which is essential for keeping the cells that instruct our muscles to move (our motor neurons) healthy.

There are four main types of SMA. In Types 1, 2 and 3 symptoms begin in childhood, while Type 4 symptoms begin in adulthood. Type 1 is the most severe form of SMA and, without treatment, children with this type are unlikely to reach their second birthday.

## What is Spinraza?

Spinraza (also known as nusinersen) works by increasing the body's ability to produce the SMN protein, which is missing in people with SMA.

So far, clinical trials of the drug have shown significant improvement in children's motor function. Many babies with SMA Type 1 are living longer, and some children who would never have been able to sit independently have been able to crawl and even walk.

# Research blocking myostatin shows promise

Prof George Dickson and Dr Linda Popplewell who led on the research at Royal Holloway, University London

**Researchers worldwide are looking at whether blocking myostatin could allow muscles to grow bigger and stronger in people with neuromuscular conditions.**

Myostatin is an important protein produced naturally in our bodies. It stops our muscles from growing too big and helps keep their size and strength within healthy limits.

A team at Royal Holloway, University of London, led by researchers Dr Linda Popplewell and Prof George Dickson, has recently published a study testing this approach in a mouse model of oculopharyngeal muscular dystrophy (OPMD) – a genetic condition causing muscle weakness in the eyelids and throat.

In the study, the scientists used a 'monoclonal antibody'. The antibody has the ability to recognise myostatin, attach to it and block its activity. Mice with symptoms of OPMD were treated with the antibody for 10 weeks.

### What did they find out?

After this period, the researchers found a number of positive effects. Muscle mass and muscle fibre size had increased. The treated mice were also stronger than those who had not received the antibody.

OPMD is caused by a genetic fault in a gene called PABPN1. This produces a mutant protein that accumulates and forms clumps inside muscle cells. These clumps are thought to interfere with muscle function and have a key role in the progression of OPMD.

The researchers found that blocking myostatin did not have any effect on the number of clumps in the muscles. This demonstrates that the treatment does not need to target the genetic cause of the condition in order to have beneficial effects.

Dr Pradeep Harish

Based on these results, the scientists believe that blocking myostatin is a promising therapeutic option for OPMD that is worth exploring further.

Dr Pradeep Harish (pictured left), one of the lead authors of the study, said:

**"Inhibition of myostatin has the potential to be used as a generic therapy for muscle-wasting conditions, either independently or in combination with other therapeutic strategies. While this method does not correct the underlying cause of the disease itself, it may be beneficial in order to stabilise the muscle and improve quality of life of patients. We are excited by future prospects."**

*We are currently supporting the work of Dr Linda Popplewell as she completes her last year of a five-year MDUK-funded lectureship.*

**To find out more about research projects supported by MDUK, visit: www.musculardystrophyuk.org/current-grants**

# Powerchair football: a successful partnership in action

Marcus Harrison (second from left) with teammates from West Bromwich Albion Powerchair football club

**In its third year, MDUK's partnership with the Wheelchair Football Association (WFA) continues to go from strength to strength.**

Over 50 percent of the UK's Powerchair football players have some form of muscular dystrophy. This partnership aims to ensure that anybody, no matter their level of disability, can take part in the nation's favourite sport.

We spoke to a player from the England national team to find out what the sport means to him. Twenty-one-year-old Marcus Harrison (pictured) from Liverpool began playing Powerchair football in year 7 at school. Though he started playing for fun, almost 10 years on he now plays competitively for his club – West Bromwich Albion PFC – and has travelled around the world, representing England in the sport.

As well as being one of the eight players to bring England to victory in the 2019 European Powerchair Football Association Nations Cup, Marcus has also been voted 'Player

of the Year' by fellow players and coaches across a number of teams within the Muscular Dystrophy UK Premiership. This league consists of the country's top 12 teams, who battle it out to become National Champions.

"From starting at an early age I had just played for fun but as the years went on it became more and more competitive and intense," explains Marcus.

**"Powerchair football allows me to play sport at the highest level and this has meant I have managed to make new friends all over the country, and around the world. The sport has helped me grow as a person and I've definitely made friends for life!"**

MDUK has partnered with the WFA in England and the Scottish Powerchair Football Association to sponsor the three main national leagues in the UK: the MDUK Premiership and MDUK Championship in England, and the MDUK Premiership in Scotland.

**The Joseph Patrick Trust (JPT), MDUK's welfare fund, awards funding towards different kinds of specialist equipment including adapted computers, electric beds and wheelchairs specifically for Powerchair football. To find out more about JPT grants, and the difference your support makes, visit: www.musculardystrophyuk.org/jpt**

England's Marcus Harrison (left) looking to get past Mohamed Ghelami of France in a tight battle.

# Survey launched into disabled women's access to healthcare

Trailblazers Manager, Lauren West

**Access to healthcare should be available to all, and no-one should miss out because of their disability. That's why Trailblazers – MDUK's 800-strong national network of young disabled people – is campaigning to ensure equal access to cervical health screenings for disabled women.**

MDUK has heard first-hand accounts of women in wheelchairs struggling to get contraception, regular cervical screenings and mammograms. This can be because of physical barriers, such as a lack of hoists in healthcare settings, or perceptions that disabled women are not sexually active and therefore don't need contraception.

Trailblazers have launched a survey on disabled women's access to healthcare, which will run for up to six months and will aim to gather people's experiences of trying to access cervical screenings.

The survey will also highlight the experiences of those who are missing health screenings, often over many years, simply because medical centres lack hoists. Findings from the survey will help establish whether reasonable adjustments (like home visits or referrals to hospitals) are being made to overcome this.

Without access to regular health screenings, the reproductive and sexual health of disabled women is at risk. Trailblazers are campaigning for equal access so that disabled women can lead full and healthy lives.

**To stay up-to-date with the latest Trailblazers news, visit www.musculardystrophyuk.org/trailblazers**

# Ambulance action: an update

**It's vital that paramedics called to help anyone with a muscle-wasting condition have the knowledge to provide the right support at times of crisis. MDUK's emergency care campaign aims to ensure this happens, so that people affected by muscle-wasting conditions receive the vital care they need in emergencies.**

As part of the campaign, MDUK ran a survey, findings from which will be included in a report launching later this year in Parliament, to help improve emergency care for people with muscle-wasting conditions.

The campaign centres around:

- working with supporters, health professionals and ambulance services to improve emergency care for people with muscle-wasting conditions across the UK
- working in partnership with paramedics so they can better support the emergency care needs of people with muscle-wasting conditions
- creating and distributing condition-specific alert cards that advise health professionals of the key dos and don'ts required for their care.

**"I have often been asked repeatedly by medical staff why I should not have oxygen, how to spell my condition and to explain what it is.**

**"My alert card takes the pressure off me so that I can focus on staying calm and getting better,"** says Michaela Hollywood (pictured) who has SMA.

**To find out more about MDUK's emergency care campaign, visit www.musculardystrophyuk.org/ambulance-action**

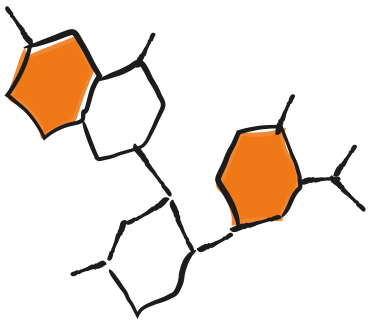
# Genetic therapies: new tools to help beat UCMD

**MDUK-funded researchers have corrected a common mutation causing Ullrich congenital muscular dystrophy (UCMD), using two genetic tools.**

UCMD is caused by mutations in genes that produce collagen VI protein. This protein acts as a scaffold to support our muscle cells.
In someone with UCMD, the collagen scaffold is faulty or absent, which means that their muscle cells are not supported properly and become damaged over time, leading to increasing disability.

The study, led by Professor Carsten Bönnemann at the NIH National Institute of Neurological Disorders and Stroke, USA, focused on a mutation in a collagen VI gene that creates an extra, unwanted piece of DNA within the gene.

The researchers wanted to test whether removing this unwanted DNA could restore the function of the collagen VI protein in cells taken from UCMD patients.



They did this by testing two different genetic tools:

- **molecular patches** – these are small pieces of genetic material that can mask or hide DNA so that it is ignored by the cell
- **genome editing** – this technique uses engineered molecular 'scissors' to cut out specific bits of DNA.

They found that both strategies successfully corrected the gene message, resulting in improved assembly of the collagen VI scaffold outside the cells.

Although these experiments were carried out in cells in the laboratory, further research could lead to the development of a potential treatment for UCMD.

**It's your support that enables us to continue to fund research projects like this. Find out more about our current research projects at: www.musculardystrophyuk.org/current-grants**

# Meet Dan

Dan McLellan is one of fewer than 250 people in the UK living with Ullrich congenital muscular dystrophy (UCMD).

After he was born, his mum, Debra became concerned Dan wasn't walking or crawling and he wasn't reaching the milestones he should. Following visits to physiotherapists, GPs and hospitals, at just two-and-a-half years-old, Dan was diagnosed with UCMD.

As time went on, he struggled with climbing stairs, running and his balance. UCMD is a progressive muscle-wasting condition and Dan, now 10, uses a wheelchair full-time to get around and does physio exercises every day to keep his muscles as active as possible.

"To hear about this kind of development in research is the most amazing news. The next stage in the research journey has been successful and Dan's specific mutation has been corrected in a lab. We're so grateful to every person who has helped make this research possible," says dad, James.

10-year-old Dan McLellan who has Ullrich congenital muscular dystrophy.

**"When we started there was no hope. Now we have it."**

# Get involved

## The MDUK Christmas shop is now OPEN!

Visit the MDUK online shop for some special gifts for your loved ones this Christmas. Discover our range of beautiful Christmas cards, as well as charming gifts and stocking fillers – stocking fillers, including our new range of Christmas homewares featuring a charming design of The Nutcracker ballet, and beautiful scented candles.


Plus, anything you buy from the MDUK online shop will help us continue to be there for people living with muscle-wasting conditions.

**Shop online at www.musculardystrophyuk.org/shop**



## Congratulations to our Great Muscle Raffle winners!

**1st prize – £3,000**
E Morris, DOLGELLAU

**2nd prize – £250**
V Osborne, TORQUAY

**3rd prize – £50**
K Hughes, LLANON

Thank you to everyone who took part. Keep an eye out for the MDUK Christmas Raffle – with a top prize of £3,000 – which will be opening soon!

**If you'd like to receive tickets for the Christmas Raffle, either to buy yourself or sell to your family and friends, let us know by calling 01628 201 289 or emailing raffle@musculardystrophyuk.org**

## BAKE A DIFFERENCE
**14 to 20 October 2019**

Be the secret ingredient that turns an ordinary tea party into one that changes lives!

Your cake sale at school, work or even at home with friends is a fantastic way to raise vital funds and to help beat muscle-wasting conditions. Why not host your own this October?

**You can take part in Bake a Difference by calling us on 0300 012 0172 or emailing bakeadifference@musculardystrophyuk.org**


## Support in your own way

If you have a great idea of your own about how to fundraise for MDUK, let us know what you're planning and we can support you with advice, tips and materials.

For example, Marion Bloomfield (pictured) from South London knits adorable teddies which she sells to raise money for MDUK.

A huge thank you to Marion and all our other fundraisers for all the ways you help support our work.

**If you have a great fundraising idea, let us know by visiting www.musculardystrophyuk.org/support-your-way or by calling 0300 012 0172.**

Muscular Dystrophy UK

Fighting muscle-wasting conditions

Weekly Lottery

# Play our Weekly Lottery and you could win £10,000!

Here's an exciting way for you to join the fight against muscle-wasting conditions and help people like Luke – play the MDUK Weekly Lottery!

Playing is easy – from just £1 a week you can have the chance to win some great cash prizes from £5 to £1,000 – and a whopping first prize of £10,000!

Enter today at **www.musculardystrophyuk.org/lottery** or by phoning our Weekly Lottery hotline on **01628 821 983** (Mon-Fri, 9am-5pm)

Luke de Bruin who lives with Duchenne muscular dystrophy

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