MUSCULAR DYSTROPHY UK | OUR MUSCLES MATTER

Campaign

The newsletter for our supporters

Autumn 2023

Inside:

Celebrity Sports Quiz 2023

The Celebrity Sports Quiz, one of our key fundraising events, was held on 14 June. Hosted by our President Gabby Logan MBE and TV presenter Kirsty Gallacher, this very special event took place in the Long Room at Lord's Cricket Ground – affectionately known around the world as the home of cricket.

In this competitively-charged evening, supporters were joined by celebrities from the sporting world as teams competed for the coveted Celebrity Sports Quiz title.

The evening included honest and heartfelt speeches, with Kiera Santry from our fundraising team sharing her personal journey of living with a muscle-wasting condition, which received a well-deserved standing ovation. This year's event raised a fantastic £88,000, which will go towards our work supporting people living with muscle-weakening and wasting conditions.

Kirsty Gallacher and Gabby Logan MBE

Kiera Santry

Monty Panesar

Rory Underwood MBE DL and Dame Jessica Ennis-Hill

Welcome

Welcome to this autumn edition of *Campaign* newsletter – written especially for our supporters.

On pages 4 and 5, you'll not only read about Professor Volker Straub's work at the John Walton Muscular Dystrophy Research Centre at Newcastle University, but also how he and his team have literally thrown themselves into fundraising for us! And on pages 6 to 8, we share some promising advances in research across a range of conditions.

You can also catch up with Josie Chubb on page 12, the face of our recent Great Muscle Raffle. Read about how we have been able to be there for five-year-old Josie and her family with support and practical advice when they needed it most.

I do hope you enjoy reading about the work your support makes possible, and the incredible difference you're making to people living with muscle-wasting and weakening conditions. Thank you.

Gabby Logan MBE
President, Muscular Dystrophy UK

PS: Our Christmas shop is now open! Be one of the first to browse this year's range of Christmas cards as well as homewares and gift ideas for all the family. Shop today at www.musculardystrophyuk.org/shop

On the cover: Professor Volker Straub and some of his team at the John Walton Muscular Dystrophy Research Centre

Muscular Dystrophy UK
32 Ufford Street, London SE1 8QD

0300 012 0172
reply@musculardystrophyuk.org
www.musculardystrophyuk.org

Find us on

Registered charity in England and Wales (205395) and Scotland (SC039445)

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Skydiving into research

Professor Volker Straub, Director of the John Walton Muscular Dystrophy Research Centre (JWMDRC) at Newcastle University, strongly believes in teamwork. He says it is an integral part of the research undertaken at the JWMDRC, and he finds building relationships with colleagues and collaborators to be very important. This is one of the reasons Professor Straub chose to do a skydive to raise funds for us.

Professor Volker Straub mid dive

"Our mission statement is about improving the diagnosis, care and treatment for people with neuromuscular conditions. We have a long-standing collaboration with MDUK, and it's great to support the charity in the same way the charity supports us when it comes to grants.

"With around 100 members of staff, the JWMDRC focuses on a wide range of neuromuscular conditions, including ultra-rare conditions, such as limb girdle muscular dystrophy, Pompe disease, congenital myopathies and more.

"To better understand any condition and to be able to find a treatment or cure, researchers start from scratch, performing basic research. Doing so, researchers can uncover the detailed processes underlying each condition and determine if something has gone wrong and how to fix it. This is the start of any pathway to treatments.

"Our mission statement is about improving the diagnosis, care and treatment for people with neuromuscular conditions."

"At the JWMDRC, there are both basic and clinical research teams. The clinical research team is involved in clinical trials and currently participates in around 30 trials for various conditions. The centre also provides a clinical

service and patient registries can help to recruit participants for clinical trials and research studies. It can also act as a link between patients and researchers.

"Collaboration is key to the successes achieved at the centre and will continue to be an integral part of our future work. Teamwork and dedication are helping us build the foundational understanding we need to develop new and better treatments for muscle-wasting conditions."

It is thanks to readers like you, our generous donors and supporters, that we have been able to award grants to the JWMDRC, and it is also a testament to Professor Straub's passion for his work that he felt inspired to join you in fundraising for us.

And as for the skydive. After a number of weather-related postponements, three members of the team have jumped and three are yet to complete their skydives so watch this space!

To find out more about the work of the JWMD Research Centre, visit https://newcastle-muscle.org

Professor Volker Straub in the John Walton Muscular Dystrophy Research Centre

Research updates

Thanks to your incredible support this year, we have been able to continue to fund innovative research. Here are two important breakthroughs.

New insights into Emery-Dreifuss muscular dystrophy

Emery-Dreifuss muscular dystrophy (EDMD) affects approximately one in every 250,000 people. It usually begins in childhood or adolescence, and is generally characterised by the early development of muscle contractures, muscle weakness and heart problems. Research into EDMD can be a challenge; family members with changes in the same genes can experience very different symptoms, while people who don't have any genetic similarities can have similar symptoms.

"Researchers are one step closer to a world where EDMD is no longer a mystery."

We fund research, led by Professor Eric Schirmer at the University of Edinburgh, which is investigating the way cells work in people with EDMD. The researchers analysed cells from 10 people with EDMD who had different changes in seven EDMD-related genes. They found that these genetic changes affect various cellular processes, leading to variations in how the body produces energy; how scar tissue forms; and how genes work between these 10 people.

By understanding how these differences vary amongst people with EDMD, the researchers hope to better predict how EDMD will progress in each person who has the condition. This will allow doctors and scientists to develop new treatments to target these differences, as well as provide more personalised care for people with EDMD.

With this new understanding of the complexities of EDMD, researchers are one step closer to a world where EDMD is no longer a mystery but a manageable condition.

Producing mini muscles in the laboratory

Significant steps have been made towards developing pre-clinical muscle models to potentially speed up the availability of new treatments for muscular dystrophy. Researchers at University College London and The Francis Crick Institute set out to identify the necessary laboratory steps to convert stem cells into skeletal muscle.

Skeletal muscles are the muscles we use to move our bodies. They are made up of long fibres, capable of producing the physical forces required for movement. These muscle fibres are supported by an intricate range of other cells, each with their own unique and essential function. Recreating this intricate system in the laboratory is invaluable to scientists researching skeletal muscles and their associated conditions, such as muscular dystrophies. Every person has stem cells in their tissue. They are generally taken from blood or bone marrow for research purposes. Stem cells are unique in that they can be converted into the

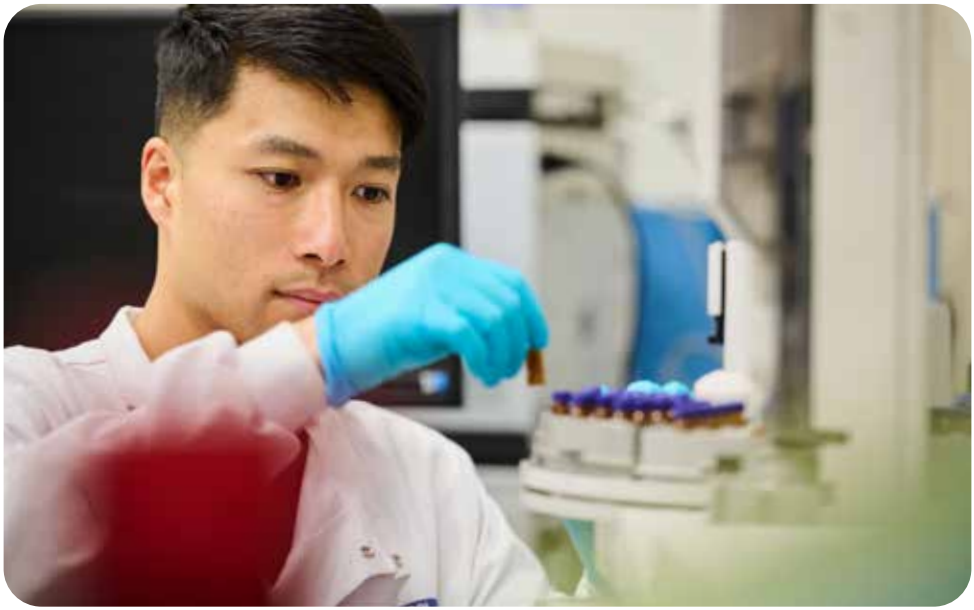
"Recreating this intricate system in the laboratory is invaluable to scientists researching skeletal muscles and their associated conditions, such as muscular dystrophies."

cells of any organ or tissue, in this case, those that make up skeletal muscle. The researchers placed the stem cells into three-dimensional water-based gels, generating the arrangement of cell types that make up skeletal muscle, including important blood vessels and nerve cells.

Some mini muscles were created from cells donated from muscular dystrophy patients, allowing the team to generate a model of the condition which could be studied and compared with healthy muscle. These mini muscles were then successfully used to investigate different types of therapies for treating muscular dystrophies, highlighting how this approach could be used to develop new treatments. Generating muscles in this way is also an important approach to replace and reduce the use of animals in pre-clinical research.

These new strides in research are only possible because of your support. For more research news, visit www.musculardystrophyuk.org/research where we publish up-to-date breakthroughs, projects, and research strategies.

These updates have been adapted from pieces written by Dr Larissa Ferguson, who is a postdoctoral researcher at the MRC Laboratory of Molecular Biology in Cambridge; and Ben Futcher, who is studying for a doctorate (DPhil) in oncology at the University of Oxford.



Our cost of living grants

As the cost of living continues to affect us all, it places an additional financial burden on our community living with a muscle-wasting or weakening condition. Fuel, energy, and grocery prices continue to rise and it's difficult to see when things will improve. Earlier this year, we were pleased to offer 470 one-off grants of £150 each. These were in the form of a supermarket voucher and were limited to one grant per household.

These grants were made possible by the generosity of the Patrick family. The Joseph Patrick Trust (JPT) was established in 1986 by Alexander Patrick CBE in memory of his father, who was a founder of Muscular Dystrophy UK. Inspired by his brother Andrew who had Duchenne muscular dystrophy and died at age 13 in 1962, the JPT was set up to promote independence and a quality of life for people living with muscle-wasting conditions. The JPT is a subsidiary of MDUK through which we provide grants for mobility equipment.

Kyri Photiou, Head of Information, Helpline and Support, received some really encouraging feedback from one recipient:

"Please pass on my thanks to everyone involved in sorting them out and to the generous person(s) who donated the money."

"I'd just like to send you a big thank you for the cost of living grant voucher, it is greatly appreciated. Please pass on my thanks to everyone involved in sorting them out and to the generous person(s) who donated the money."

We know that the available grants were a small amount compared to people's needs and only a limited number were available. We have therefore established a cost of living hub on our website – available at www.musculardystrophyuk.org/costofliving – to provide information and support and to signpost people to other assistance for which they may be eligible.

We have also produced a detailed cost of living report and policy recommendations, written in collaboration with our community.

You can read the key findings and recommendations here: www.musculardystrophyuk.org/col

Progress in research and campaigning for SMA

Researchers identify new cause of muscle weakening in SMA

A research project funded by us has identified a new cause of muscle weakening in spinal muscular atrophy (SMA) during early muscle development. Dr Melissa Bowerman's team at the University of Keele has been investigating the mechanisms that drive skeletal muscle wasting.

SMA is a rare muscle-wasting condition caused by genetic changes that reduce the levels of a protein in motor neurons – the nerves that connect the spinal cord to the muscle. The protein is called survival motor neuron (SMN). SMA is characterised by the loss of motor neurons, leading to weakness in the skeletal muscles.

There are three treatments available for people living with SMA in the UK – Spinraza (nusinersen), Zolgensma (onasemnogene abeparvovec) and Risdiplan (evrysdi). But even though these are available, some people will still experience muscle weakness. So to develop fully effective therapies, it's essential that we advance our understanding of the processes that cause muscle wasting.

A group of researchers led by Dr Bowerman investigated a molecular pathway that has been linked to muscle wasting, called TWEAK/Fn14. Despite having a clear role in regulating muscle health, its involvement in SMA and early muscle development is yet to be understood.

By studying mouse models of SMA, the study found TWEAK/Fn14 is less active during SMA progression, contrasting with findings from previous studies which reported

increased TWEAK/Fn14 activity in wasting adult muscles. This suggests it is responsible for different things during development and in adulthood.

The scientists were also able to show that TWEAK/Fn14 and SMN likely work together in SMA mouse models and that SMA muscle symptoms were improved using a molecule that boosts TWEAK activity in young SMA mice. This suggests that restoring TWEAK/Fn14 signalling in SMA muscle might be therapeutically beneficial. These findings could eventually lead to effective combination therapies to increase muscle strength in people living with SMA.

This article has been adapted from a piece written by Bethany Evans who is a Gene Therapy Technician at King's College London.

Newborn screening for SMA

The developments in research coincide with progress towards the approval of newborn screening for SMA in the UK. In May this year, a report was published based on the findings from an inquiry into newborn screening for rare conditions by the All-Party Parliamentary Group for Muscular Dystrophy.

The inquiry heard powerful testimonies about the importance of newborn screening, with a focus on SMA, given the announcement in November 2022 by the UK National Screening Committee (UK NSC) that it has started reviewing whether newborn babies should be tested for SMA.

In July 2023, the UK NSC announced a significant step towards recommending SMA as a condition to be added to the Newborn screening programme. They recommended that plans for a UK-wide pilot (called an 'in service evaluation') of newborn screening for SMA should progress at the same time as the development of an economic model. Although this isn't a recommendation to start newborn screening in the UK, it means the UK NSC will be able to collect the information it needs to make a decision faster than originally planned. This is a great step forward and one that we welcome.

We hope that by highlighting these recommendations to UK NSC and other key decision makers, we are one step closer to the UK having a newborn screening assessment process that meets the needs of people with rare conditions.

You can read the full report here: www.musculardystrophyuk.org/newborn

I'll just do it the Josie way!

You may remember five-year-old Josie Chubb from our Great Muscle Raffle earlier this year. She is a creative, adventure-loving little girl who loves to be active, especially swimming – even in the sea! Josie was born with Ullrich congenital muscular dystrophy (UCMD) but this doesn't stop her from getting involved in anything and everything, and she doesn't consider her disability as a limitation.

Charlotte, Josie's mum, says: "If something gets in Josie's way, she gets more determined and just says 'I'll just do it the Josie way!'

"Josie's diagnosis journey started in 2018 when she was about 18 months old. We noticed that her walking hadn't really progressed but thought it could be a balance issue. We enrolled her in ballet lessons which she loved, however, she was still falling and struggling to get up.

"Eventually, just before Josie turned two, we were referred for tests including a muscle biopsy, but all the results came back inconclusive. Then Covid-19 hit, and things ground to a halt. In all, we waited two years for a diagnosis which was hard.

"Before the diagnosis, nothing was available to us. I applied for a blue badge, for example, but because we had no diagnosis, I was explaining a condition to people that nobody sees. But, once we had a diagnosis and were put in touch with MDUK, it felt like the support doors really opened up.

"This is where talking to MDUK really helped. Speaking to people who really understood and had either been through a similar diagnosis or had supported others in our situation, reassured us. Knowing that we were not alone was a great comfort.

"MDUK's Care, Information and Advocacy team gave us invaluable advice on any adaptations we could make to our house. They also provided information on access

to financial support and Disability Living Allowance as well as helping us in talking to Josie's school. They connected us with other parents so we could exchange advice and support. I honestly do not know where we would be without them."

Your support means we can continue to be here for families like Josie's. Read more about the work you are making possible here www.musculardystrophyuk.org/get-support

The Chubb family – dad James, little brother Harry, Josie and mum Charlotte

Our brand refresh

You may have noticed something a little different in this edition of *Campaign*...

We've changed our look. Refreshing our brand so that we can reach and support more of the over 110,000 people living with a muscle-wasting or weakening condition in the UK. We're still orange, we're still called Muscular Dystrophy UK, but we're now clearer, bolder, and stronger.

Having reviewed our brand last year we found that, although it had some great elements, it didn't present a clear picture of who we are today and where we want to go. We also knew that our brand colour, font and logo weren't as accessible and inclusive as they could be, particularly online.

Our community makes us stronger and that's why we decided to work together to refresh our brand. So that people could know who we are, what we do, and in turn be able to reach out for support and become part of our community through fundraising, campaigning, and volunteering.

Get involved!

Could you be our next big winner?

Our Christmas Raffle is a great way to support our life-changing work AND to be in with a chance of winning our top prize of £3,000, just in time for Christmas! What would you do with a prize like that? At only £1 per entry, it's a fun and easy way to support our work.

Visit www.musculardystrophyuk.org/raffle from early October to enter our Christmas Raffle. Best of luck!

Congratulations to our Great Muscle Raffle winners, drawn in June 2023: **1st prize £3,000** Jackie Cooper, Slough. **2nd prize £250** Mrs Lungley, Colchester. **3rd prize £50** Mrs Larkin, Bexhill-on-Sea.

Bake a Difference like Charlotte on Thursday 9 November 2023

Last year, Charlotte (pictured right with her dad and baby son) organised her baking fundraiser as her dad lives with Becker muscular dystrophy.

"Taking part gave me an opportunity to raise awareness about muscle-wasting conditions and how they can affect people and their families. And it was perfect for me as I love baking!

"Having a nice catch up with those you love, eating some yummy cakes and treats (shop-bought counts too!) while helping raise money for a very important cause - what's not to love?"

Sign up to receive your free fundraising pack and Bake a Difference this November: www.musculardystrophyuk.org/bakeadifference

MDUK shop

Head to our online shop to discover this year's selection of Christmas cards, homewares, gifts and 'Team MDUK' supporter wear. With stocking fillers starting at just £1, you're sure to find something to suit every taste and budget. And remember, all the proceeds go directly to helping us support people with a muscle-wasting or weakening condition through every stage of their life.

Shop online at:
www.musculardystrophyuk.org/shop

Leave a lasting legacy

Did you know that Gifts in Wills fund a quarter of all our work? These special gifts enable new research to find effective treatments and cures.

You can be a part of these discoveries with a gift in your Will. If this is something you are considering and would like to know more about, please contact Grace Moran on **0207 803 4845** for an informal chat or email on **legacy@musculardystrophyuk.org**

You can also request our **Guide to leaving a Gift in your Will** brochure which shows the impact your gift will make, as well as practical information on writing your Will.

To find out more, visit:
www.musculardystrophyuk.org/giftsinwills

MUSCULAR DYSTROPHY UK | OUR MUSCLES MATTER

Weekly Lottery

Scan here to join our lottery today!

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 – and play our 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

£10,000 JACKPOT