

NEWSLETTER

May
2020

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Hello,

Its nearly a year since we met with some of the familiar faces of our lovely friends and acquaintances at the MDSG Annual Conference, and had chance to make new friends. Not sure when we will meet again, but we are so looking forward to seeing everyone as soon as it is possible.

As the saying goes, 'Absence makes the heart grow fonder' so the next Conference will be extra special for all meeting up again.

We are generally the first faces (our apologies for this) that you see when arriving at the Conference. Before you've had chance to catch your breath you're being asked for your name and where you come from, its like going on a blind date! Maybe height, hair colour, eye colour may be included next time!!

We do hope that our smiles on arrival help, particularly as sometimes the queue to register can be long, and that reading this you are smiling now.

Our very best wishes, and please stay safe.

Linda, Judith and Rob



Margaret Bowler and the Trustees of MDSG send sincere warm wishes to you in these uncertain times.

We are all personally aware of self-isolating and, for many, shielding, and the inevitable impact it has on individuals and families.

Our thoughts are with you.

As volunteers we are continuing to work on your behalf, and thank all the contributors to this Newsletter who help us to feel 'connected' encouraged and reassured.

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Hello Everyone

The last few weeks have been very challenging for us all. I guess many members are in the 'same boat' as I am, restricted in going out, because of age.

The weather is bright and warm and that is helping the situation especially if you have an outside space to sit in. I do hope you have received the information I have sent out about people with Myotonic Dystrophy (DM) being vulnerable and to stay in the confines of your home.

Some members have received sanitisers with one of the letters. I am sorry I didn't have enough to send everyone one. The second letter I sent was also sent to the people who are not members (not paid subs) as I know they have DM and are still vulnerable and need the information.

I am fortunate to sit outside writing this letter. I can hear the birds singing plus the noise of the pigeons!

I am putting together the history of MDSG for the last 10 years, and hope to attach it to the history of the previous document of 20 years. So by 2021 there should be the history of 30 years of MDSG. As I write this I can bring to mind several people we are still in touch with who were members 30 years ago. I will not mention names but you know who you are! You will be older and grey like me!! There will have been lots of sadness in our families also during those years.

The Annual Conference has been cancelled this year, but some of the Medical consultants have written information for us so that is encouraging. We are privileged that they have given their time to keep in touch with us. Thank you. The office is not functioning normally, Mike Walker is taking the phone calls,

Elycia Ormandy is taking the Helpline calls and I am receiving the emails. Julie is going into the office 2 days a week and taking the work home. Greetings to you all, Keep Safe

Margaret Bowler

Letter from The Editor

Most of you will have experienced the addition of many new words and phrases that have crept into our vocabularies in recent weeks. I have ventured into areas of previously unexplored (by me!) technology in order to keep up with the many changes to our lives.

Overnight we have had to adapt and accept these changes to the way things 'have always been done'. Restricted access to GP surgeries, hospital clinic consultations by telephone or Skype, Zoom social gatherings, a new pace of life and a new approach to exercise. It is good to know that some things haven't changed. Namely the need of the Myotonic Dystrophy Family to keep supporting one another.

Cancellation of our 2020 Conference was a difficult decision for the Trustees.

To maintain the strong links between MDSG members and their families, and the dedicated clinicians and researchers who come to speak at our Annual Conference, Margaret Bowler invited each of them to write a short article for this Newsletter.

Lynne and I both send you our warmest best wishes. We especially send a smile and hello to the young adults who would normally be sharing our own get together in June. Keep in touch and keep reading!

Elycia

Update from the University of Glasgow

As with everyone else, we have been hit hard by the lockdown restrictions imposed to ensure we can contain the COVID-19 epidemic. The University closed for face to face teaching with students on 16th March, but teaching nonetheless goes on with lectures, tutorials and exams all moved online. The following week, the University laboratories were closed for all non-COVID-19 related research.

You may have heard, one of the three national COVID-19 Testing Lighthouse Labs is located in Glasgow. The testing centre is hosted by the University of Glasgow at the Queen Elizabeth University Hospital Campus, and opened in collaboration with the Scottish Government and industry experts. Notably, the “PCR” diagnostic test that is used to test samples for the presence of the virus is very similar to the types of PCR tests that we routinely use to examine DNA from individuals with myotonic dystrophy and related conditions. Our laboratory has donated various pieces of equipment to the Lighthouse Lab and all of our team have volunteered to help the national cause and work in the testing centre. The sooner we can introduce mass-testing, the sooner we can control the epidemic and all of our lives will be safer, and we can get back to laboratory-based research. Although, as of 24th April, none of our team have yet started working in the Lighthouse Lab, the expectation is that several of them will be asked to help over the next few weeks.

You can find out more about the Glasgow Lighthouse Lab testing centre here:

https://www.gla.ac.uk/news/headline_720507_en.html
<https://www.bbc.co.uk/news/uk-scotland-52389064>

Meanwhile, the myotonic dystrophy research hasn't stopped completely though. Although we are no longer analysing myotonic dystrophy DNA samples in the laboratory, over the last few years we have generated a lot of genetic data from individuals with myotonic dystrophy type 1.

Much of this data has yet to be fully analysed and the research team are all still working hard at home analysing the data and writing papers for publication. This work is furthering our understanding of how genetic variation contributes toward the symptomatic variation that we observe, and in particular how the ongoing increases in the number of CTGs that occurs throughout life, relates to progression of the symptoms. These myotonic dystrophy data, and similar data that we have generated in parallel in a genetically similar disorder Huntington disease, reveal that increases in the number of CTG repeats throughout the lifetime of an individual is a key part of the disease process. Indeed, it may even be that in many individuals it is essential that the number of repeats increase beyond the number of repeats inherited to get any symptoms at all.

This therefore strongly suggests that if we could slow down, or ideally stop, the increase in the number of repeats, this would be therapeutically beneficial. We also now know the identity of some of the proteins that are responsible for making the repeat get bigger, and it is now increasingly recognised that if we could develop drugs that inhibited these proteins, they would likely be useful to treat, not just myotonic, but Huntington disease and several other conditions with a similar genetic basis.

This idea has garnered much enthusiasm from the pharmaceutical industry, and we are working with several companies to advance this area and hopefully generate some novel new treatments for myotonic dystrophy and related disorders.

Continued over

The other good news we have is that Dr Mark Hamilton has almost finished his training and has been appointed to a Consultant position in Clinical Genetics at the Queen Elizabeth University Hospital in Glasgow. We will be really pleased to have Mark back, hopefully in August, where, along with all of our other fantastic clinical collaborators in Scotland, Mark has lots of plans for how to involve even more individuals in clinical and basic research. This is an exciting time for myotonic dystrophy research in Scotland, and we are all keen to get back into the laboratory to take these ideas even further forward. Meanwhile, we are still working as hard as we can, and contributing where we can to help the country get through the crisis. In the meantime, stay safe everyone!

Darren Monckton
Professor of Human Genetics
University of Glasgow

Meals on Wheels



MDSG member Steve Hewitt, delivering a meal to Margaret who was following Government advice and staying indoors.

Disabled Discretionary Trust

Hello I hope you are all safe and well, and sorry that I won't get a chance to meet you at the Conference this year. I help my Clients by setting up Wills, Trusts, Lasting Powers of Attorney and Funeral Plans. I have had the pleasure of being able to help several MDSG members with Disabled Discretionary Trusts to protect their children.

This type of Trust can be used where a child or other relative has a disability. If this person inherits from a Will directly, the Local Authority will want to assess them to determine what help they are eligible for. If they hold assets over a certain threshold, the Local Authority may consider that they can now financially support themselves and cut the benefits that they receive. They may also struggle generally to manage their own money.

With a Disabled Discretionary Trust, you are able to leave them their share of inheritance in a Trust which is managed by a Trustee. The Trustee can ensure that the beneficiary can access their inheritance when they need it, without their benefits being affected. I know a lot of people that I speak to are concerned about what the future looks like in terms of social care.

I am offering appointments via Zoom (internet) or telephone. If you would like advice on any aspect of Estate Planning with no obligation, please get in touch on 0800 0029235/07852 308182 or info@binamenzestateplanning.co.uk.

Bina Menz, Managing Director
Bina Menz Estate Planning Ltd
www.binamenzestateplanning.co.uk

A word from that bloke who sits in the office

This really is a strange and difficult time. I hope everyone associated with the Support Group; patients with DM, families and Trustees, are all well, COVID-free and coping with the social isolation. I must admit I am finding it difficult to work from home. Like many of you I have become used to Teams meetings and Zoom but rarely feel a sense of satisfaction after a remote meeting. I do keep in contact with Anjani, Belle, Jake, Abdul and Sarah, but online chat is no substitute for



meeting in person and obviously we are unable to do any lab work to progress the search for a DM treatment. Hopefully we will be able to return to work soon and as I write the government has provided guidance on safe return to work, so let's hope it won't be too long before we are back.

Just before the shutdown I bought a greenhouse and a germinator. I have gone into overdrive growing tomatoes, courgettes, potatoes, garlic, cucumbers, gherkins, carrots, parsnips, leeks, sweetcorn and beetroot all in a space 6ft x 10ft. My crops are coming along nicely – mostly leaves at present but I am optimistic edible parts will follow. Please see the accompanying pictures (potatoes, carrots, beetroot and sweetcorn – left, and

tomatoes and courgettes - right). It's a pity there's no conference this year as I could have brought some produce for Phil to sell on his stall.

As you can see from the image below, I have also



taken the opportunity to learn new skills such as the production of PPE. My face mask from an old sock is simple but effective, just like me. Though it does pull my ears down a bit.

The big news from our lab is that after a monumental effort, Ami's paper was published in the journal Science Translational Medicine on the 29th April. If any of you would like to read it, a link to the paper is available via:



<https://thebrooklab.wordpress.com/recent-publications/>

Ami has written an account of the history of conducting and publishing this work during which time she has had 2 children (Harry and Maisy). Also included in this issue are pieces from Anjani, Belle, Sarah, and new-boys Jake and Abdul.

Please take care and stay safe and I hope we will all be able to meet up again soon, perhaps with fried eggs (and chips)?

BW David

Ami's paper – a potted history by Ami Ketley

On the 29th April 2020 myself, David, Chris and Becky met for a virtual celebratory coffee break to mark the publication of our paper in the journal Science Translational Medicine. The road to getting to this day has been very



very long with many ups and downs but I'm pleased to say we finally made it! The journal captioned the paper "Muscling in on myotonic dystrophy" but I think a more appropriate title could easily be "A story of perseverance!" I started working on the project in David's lab in February 2010. The plan was to try and find drug compounds that may be useful in the treatment of DM and I'm sure many of you have heard myself or David talking about how we look at 'spots' in cells and see if we can reduce or remove them with our treatments.

The work went well and we found some compounds that looked interesting but we didn't know how they were working. In 2011 we met with a group of scientists from the pharmaceutical company GSK. They were looking to work with university groups to develop drugs and so we put together a 'Dragon's Den' style pitch to hopefully get their investment. Unfortunately, they couldn't fund us in the way we had planned but they were so interested in the project

that they wanted to help and suggested I go and work with a department at their site in Stevenage to develop the project further.

So, in January 2012 I packed my case and spent my weeks living in Stevenage. Originally the plan was for a couple of weeks but this soon turned into 6 months! The time there was really useful and GSK gave us their time, experience and resources to understand things a lot more. When I came back to Nottingham we kept working with GSK and had regular contact with their different teams worldwide. I continued doing experiments too and by the beginning of 2014 we thought we had enough information to write a paper. We sent the paper to a few different journals who had different reasons for turning us down. Then in November 2014 we sent our draft to Science Translational Medicine. They sent a positive response saying they thought the work was interesting but suggested a few additional experiments would allow them to consider publishing it. We did these experiments with Charles Thornton's group at the University of Rochester, thankfully the results were really promising. This work continued through 2015 and in another major milestone I had my son Harry in October of this year.

I spent a year on maternity leave trading my science hat for mummy duties, although I was never far away from updates and developments in the lab. I came back to work part time at the end of 2016 and finished off experiments and wrote the next version of the paper. We sent this back to the journal in June 2016. We were optimistic we had addressed their original comments but they still wanted more! Cue more experiments, head scratching and more versions written than we can keep track of and we submitted another manuscript in May 2018, 2 weeks before the start of my maternity leave with my daughter Maisy.

Continued over

Unfortunately, the response again wasn't what we wanted to hear and the reviewers asked for more experiments and explanations. I had just enough time to put a plan together before Maisy was born and, in my absence, Marzena and David carried out further experiments in the lab. Throughout my maternity leave I was modifying the paper, squeezing in time when babies and toddlers were sleeping to answer the reviewer's queries!

Fast forward to August 2019 and the final roll of the dice! The journal sent the paper to reviewers who replied with the single sentence: "The authors have addressed all of my comments". WOW! David called me as soon as he saw the email and I don't think either of us dared to believe we might have done it! The journal doesn't actually say a paper has been accepted until you have dealt with further editorial issues but the magic word appeared on their website of 25th February 2020, **ACCEPTED!** I have to admit I had a few tears of happiness when I saw this.... followed by a celebratory glass of wine or two!

Further edits and comments were dealt with and on 29th April 2020 at 7pm the paper was published on their website. We'd finally done it!

It's been a long time coming but we got there in the end! I feel really proud of this piece of work and more than anything I hope it has a positive impact on myotonic patients and their families. Margaret and the MDSG have always been so supportive of me throughout my time working on this project and for that I am very grateful.

I hope you're all keeping well in this strange and difficult time.
Take care and stay safe, Ami

Anjani Kumari

Being stuck inside isn't all that bad!

My progress at the lab is 'on ice' -- literally -- so in the meantime I have been focusing on the literary arts of my PhD. Reading papers and grading scrutinizing other people's work for once!

I have my little desk at home, my favourite food stocked up and weekly meetings, which usually I would grumble about, but they have been the driving force behind my days. I miss the office and I miss the being around people.

The crowning jewel of my lockdown adventure has been getting to go to the animal lab once a week for some animal work. Hope to see everyone soon for another tumble in the mud :D



Sarah Buxton

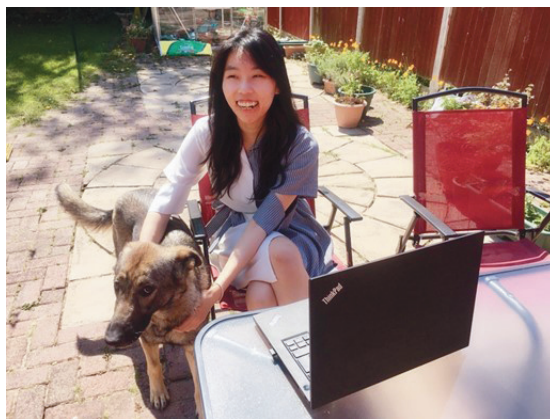
I'm in lockdown with my partner Scott and our 2 children Holly (5) and Dexter (3). During the week my partner is at work so it's just me and the kids most days. We've been keeping busy trying the Joe Wicks P.E. tutorials, home-schooling, going for walks, and gardening. We've also tried some new methods of keeping touch with family and friends using apps such as 'house party' and 'zoom' with varying levels of success!!



in

I've gone to the lab a few times to genotype our transgenic mouse lines. Including the HSALR mice (the Myotonic model mouse containing 250rpts) and the Cdk12 mice. This has been strange as we have used a new lab and had to maintain social distancing! But was refreshing to see other people (Anjani and David) and get a break from the kids!

Xiaomeng Xing (Belle)



In the early February, I was told that I had to renew my visa in my home country China, which indeed gave me a headache. Not only because a novel virus COVID-19 was spreading rapidly in China at that time, but also because my study would have to be interrupted for 4-6 months. However, just before the lockdown got started when I had just adjusted myself to accept and handle this "hassle", somehow a dramatic shift happened. I was told that for my case, I could possibly make my visa application inside the UK. This

great news did make me relieved as it's no exaggeration to say that the flight tickets from London to China were going through the roof (about 2k-10k for a single journey). Also due to the global breakout of COVID-19, people from abroad have to be quarantined for 14 days once they land in China, which could be a really unpleasant experience.

Now, apart from managing to work from home, keeping myself motivated and productive is another thing I need to learn. Meanwhile, people might have found the social distancing suffocating, but this is the kind of sacrifice that we have to make if we want to win the battle against COVID-19. If you think we're at the darkest moment, that's because the darkest hour is just before the dawn!

Abdulkadir Abakir

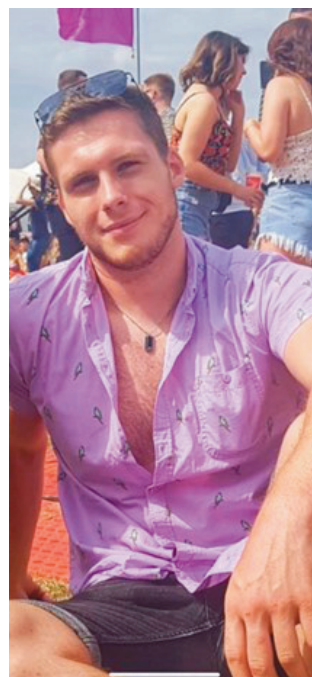


My name is Abdulkadir, a final year PhD student in the School of Medicine, University of Nottingham. In my PhD research project, I have discovered the presence of methylated adenosine on the RNA component of R-loops. R-loops are nucleic acid structures formed by an RNA:DNA hybrid and unpaired single-stranded DNA that represent a source of genomic instability in mammalian cells. Since a key molecular characteristic in DM1 patients is the presence of an unstable trinucleotide (CTG) repeat in the 3' untranslated region (3' UTR) of dystrophin myotonic protein kinase (DMPK) gene on chromosome 19q13, I want to find out if R-loops play a role in the molecular pathogenesis of DM1 disease. I had worked for one week in the lab before the shut-down. During my thesis write-up, we have made plans to conduct experiments to

study R-loops presence and function in DM1 but, unfortunately, the Covid19 lockdown meant none of the planned experimental work could be carried out. To make things more complicated, the thesis writing was also disrupted as I have to home-school my son – Mohammed and take care of our daughter Mawadda (both shown in the picture above, along with Abdul's shadow). Though I am adapting to working on my thesis from home, and plan to defend my thesis online, I hope things can go back to normal so I can get into the lab.

Jake Brown

My name is Jake, I'm a new member of the lab in the first year of my PhD. I graduated from the University of Nottingham last summer with a Master's degree in Biochemistry and Genetics after doing my final year project as part of this same lab (so I'm not a total newbie). After my degree I was lucky enough to find a place on a BBSRC funded PhD programme which, to my surprise, meant me returning to Nottingham within months of leaving. Outside of study my hobbies include rugby and going to the gym. Although the lockdown has meant that I was able to be in the lab for only around 2 weeks before being sent home, it has meant I've had the time to teach myself some new skills such as computer coding that will be useful to me during the rest of my PhD. I'm currently looking forward to seeing the end of lockdown so I can get back to the lab and officially begin my research.



The corona virus pandemic – how it affects us all

By the time you will be reading this, we will hopefully be coming out the other side of the biggest health crisis of the last 100 years. Sadly, we will all know friends and family who have been severely affected or killed by COVID 19, and others who have struggled financially because of lockdown. I would like to share with you how this virus has affected my family, including some unexpected positives, and how to make the best of it.

As you know, I work in a hospital and for Wales Air Ambulance, and once it became apparent that the UK was going to be affected as severely as everywhere else in the world, there was a few weeks of hurried planning and preparation everywhere in the NHS. In my hospital, similar to most others in the UK, outpatients and clinics were closed down, operating theatres were converted into intensive care wards, and lots of staff were quickly retrained to cope with new roles and how to care with people while wearing protective equipment. I started telephone clinics,

which worked well, and we started taking trauma victims to fill wards that used to house elective surgical patients.

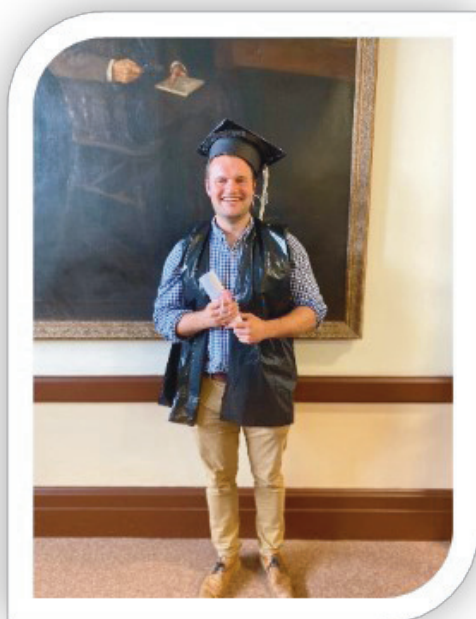
My wife, who works in a school, firstly saw her school close, and then started work again in a “Hub School” – the children of key workers from 8 schools in the area all in one place. Trying to socially distance 4- and 5-year olds is very difficult!

My daughter found that she could not ride her horse – this was non-essential exercise apparently – and had to go to the stables according to a strict timetable, to avoid other owners.

And, of course, my mum, who is well into her 80's, was told to isolate for 12 weeks. This meant we couldn't see her, and she couldn't go out to even do food shopping. Fortunately, she has great friends who shop for her, and we could talk to her via video-messaging on the Portal device we bought for her, which we have found really useful.



However, the people who saw the biggest changes due to the coronavirus outbreak were my two sons. Both were in their final year at University; Chris doing medicine and Nick doing a paramedic degree. At first, both got sent home from University, and we had a couple of weeks where 7 of us were all in one house, which was busy but actually quite fun! Both had completed all of the written parts of their courses, and were due to do their final placements – one in Sri Lanka that sadly had to be cancelled – but they were then asked if they would be willing to start working for the NHS 3 months early to help out in the crisis. Of course, they both said yes, and though their mum and dad were anxious about them putting themselves at risk, we were immensely proud of their decision.



Chris immediately started work at a hospital in Swansea as a Medical Student Assistant, in the Haematology ward where he had done a previous placement. Within a couple of weeks, the GMC announced that the new doctors would be registered early, and though they didn't have an official graduation, some of the consultants at the hospital staged an impromptu one for them with bin-liner robes and cardboard hats! He will continue to work as Dr Bowler on the ward until August, when he would have originally started work in a normal year.

Nick had several job offers and decided to work for the South Central Ambulance Service in Oxford, which is where he trained. He had to wait several weeks for a space on an induction course, because I had become unwell with a high temperature while everyone was at home with us, so he had to isolate with us for 14 days. He had a week's induction and then 2 training weeks in which he passed everything first time, and started work as a paramedic in the second half of May.



As the lockdown continues, we have noticed lots of things have become normal that 6 months ago we would have never dreamed of – queueing to get into the supermarket, having GP appointments by phone or sending a photo, not being able to go to the pub, doing a lot of gardening! But we have found that being forced to change the way we do things has had some positive effects, especially at work. I have been working 12 hour shifts instead of my normal hours, which has given me more time at home; telephone or video consultations are here to stay, people only go to hospital if they really need to.

We need to continue to follow the NHS advice and stay home to keep safe. This is vitally important for everyone with Myotonic Dystrophy, as you are in a higher risk group. Its also important to keep as fit as you can, even if it is just in your garden or from your chair, and to keep eating a good diet. Key workers like my family and many others involved in the group are working hard to get the country through this crisis, it really helps us if people with health conditions support us by keeping to the advice given by the NHS and the government. I look forward to seeing you all again at the next Annual Conference in 2021.

Ian Bowler



Scientists edge closer to treatment for myotonic dystrophy

Scientists at the University of Nottingham have taken a step closer towards developing a treatment for the long-term genetic disorder, myotonic dystrophy.

In a paper published today in the journal *Science Translational Medicine*, scientists from the Schools of Life Sciences and Chemistry at the University, have discovered that by inhibiting a molecule in patients' cells called CDK12, they can potentially develop a therapy to alleviate some of the symptoms, and help treat this incurable condition.

Myotonic dystrophy is a long-term genetic disorder that affects muscle function. It is the most common form of muscular dystrophy in adults and affects about one in 8,000 people. *There is currently no treatment available.* Symptoms include gradually worsening muscle loss and weakness. Muscles often contract and are very slow to relax. Other symptoms may include cataracts, intellectual disability and heart conduction problems.

Some patients have a very mild form and others have severe form, where they are congenitally affected from birth. This is due to the molecular underpinning of the condition, which is caused by a dynamic mutation; a triplet repeat expansion, in which three base pairs of DNA are present in different copy numbers. In the general population people have 5-30 copies of this DNA sequence. In patients with myotonic dystrophy—this particular segment of DNA becomes bigger than it is in the general population, often with hundreds of copies of the triplet repeat. The faulty gene produces a faulty RNA which contains the expansion

sequence, (RNA is a macromolecule essential for all known forms of life which transfers information from DNA in the nucleus to the cytoplasm of a cell where it makes proteins). The faulty RNA gets stuck in the nuclei of myotonic dystrophy patients' cells, resulting in disruption to many cellular processes. In this new study, scientists have discovered that through the inhibition of the molecule CDK12 – the additional faulty RNA disappears, and so reduces the symptoms of the condition.

David Brook, Professor of Human Molecular Genetics at the University, is the lead researcher on the study. He said: "Through our research we now understand a key molecular component in the pathway of the condition and that's a target for us to try to inhibit this particular CDK12 protein which will then have beneficial effects in terms of developing a treatment.

"Transcription is the process by which RNA is made from DNA and this can require CDK12. When the repeat sequence is transcribed, it makes the faulty expansion RNA – but we think that the myotonic dystrophy patients' cells struggle to make the faulty RNA and they increase their levels of CDK12 to keep ploughing through the expansion sequence and make more of this RNA because the cell doesn't know this is toxic.

"What we've found is that our inhibitors affect the function of CDK12 and so prevent the transcription of the faulty RNA which offers a possible route to a treatment of the condition.

"We are now at the stage where we know if we can inhibit CDK12 selectively – then it's going to be a potential therapy – and now we are trying to work out how to do that."

Link to the paper on:
<https://thebrooklab.wordpress.com/recent-publications/>

How can a Speech and Language Therapist help you?

Hello all, I'm Jodi, a Senior Speech and Language Therapist based at The National Hospital for Neurology and Neurosurgery in London. I'm sorry we



couldn't meet together at the MDSG conference this year but we are of course very aware how important staying at home is at the moment to minimise risk of catching and spreading the Coronavirus.

As you already know, Myotonic Dystrophy affects several different aspects of the body. Some of these changes mean people living with Myotonic Dystrophy are considered more vulnerable in the current pandemic. Those who experience difficulty swallowing are considered to be more vulnerable and swallowing difficulties are common in Myotonic Dystrophy. Speech and Language Therapists support people living with swallowing difficulties (known as dysphagia) to minimise the risk of these difficulties causing problems later on in life. For this reason, we encourage people living with Myotonic Dystrophy to be aware of their swallowing and symptoms that may suggest some difficulty.

Swallowing problems cause a range of symptoms including challenges with chewing hard food, food sticking in the throat, and coughing and spluttering. Over a period of time these difficulties can cause weight loss and chest infections from food and drink perhaps going down the wrong way. Speech and Language Therapists use their expertise to accurately diagnose the nature of the swallowing problem and search for practical management advice. We work very closely with our multidisciplinary colleagues such as the dietitian, occupational therapist and physiotherapist in order to find the best possible solutions as well as address other problems that Myotonic Dystrophy may cause such as challenges with speech, mobility and completing day-to-day activities.

I would encourage you to be vigilant to any symptoms of swallowing difficulty and seek a referral to a Speech and Language Therapist for assessment and advice if you experience any problems. You can read more about swallowing difficulties in Myotonic Dystrophy in the MDSG leaflet pictured below.

I hope this has provided a little information about how a Speech and Language Therapist may be able to support you. If you already experience swallowing difficulties and would like to share your experiences for learning and research purposes please do take a look at the flyer within this magazine which will give you more information.

Stay well and stay safe everybody, I hope to see you all at next year's conference! **Jodi**

See insert with this newsletter to take part in a survey on swallowing.



The UK Myotonic Dystrophy Patient Registry is a research database that aims to recruit any individual, from anywhere within the United Kingdom, with a diagnosis of myotonic dystrophy, who may be interested in becoming involved in future planned clinical trials and upcoming research.

As of April 2020 there are 773 participants registered with the UK Myotonic Dystrophy Patient Registry, with five new registrations per month. The average age of participants is 44 years.

Most participants on the registry have myotonic dystrophy type 1 (DM1), with approximately 13% having a diagnosis of congenital DM. A small number of participants have DM2. Almost half of all participants on the registry have genetic confirmation of DM1. **We welcome all those with myotonic dystrophy to join the UK Myotonic Patient Registry.**

The most commonly reported symptoms are day-time sleepiness or fatigue (78%), and myotonia (75%).

Almost 20% of participants report using medication for day-time sleepiness or fatigue

Nearly 10% report using medication for myotonia

Some participants report having a heart condition and 20% of these report using medication to help.

Benefits to joining the registry

It can help:

Identify participants for **clinical trials**

Help develop **care standards**, to help improve the care people receive

Support specific **research** questions for **doctors and scientists**

Contribute to understanding the **natural history** of a rare disease

Provide a **link to the research community** enabling people to receive information directly relevant to their condition (for example, newsletters or updates on standards of care).

The registry has helped recruit for various clinical trials and research studies such as the OPTIMISTIC clinical trial, the PHENO-DM1 natural history study, the AMO Pharma tideglusib phase II clinical trial, the PREFER project, as well as **18** other projects.

If you would like to join the registry or if you have further questions, please visit <https://www.dm-registry.org/uk/> or email ben.porter@newcastle.ac.uk.

Thank you for the continued support from all patients and patient organisations!

Ben Porter -

UK Myotonic Dystrophy

**Patient Registry Project Manager
and Curator**



Please see the insert inviting MDSG members to participate in the PREFER survey regarding future help for people with myotonic dystrophy. This project is being supported by MDSG and organised by the team in Newcastle, who look after the UK Registry.

Regional Group Contacts

The numbers below enable you to make contact with someone else in your region. Phone to ask about local meetings, provision of services in your area for families with neuromuscular conditions, or to discuss concerns. Is your area represented?

Aberdeen
Jenny Watt
01224 580559

Cardiff
Margaret Ware
02920 869277

Croydon
Lesley Smith
020 8777 6587

Cumbria
Neil Braiden
01228 512385

Devon
Josephine Holmes
(contact office)

Dorset and Hampshire
Ruth Harrison
01425 626133

Glasgow
Ann Cobburn
01389 381246
Michael Kneafsey
01360 311440

Kent
Kirsty Blount and Amanda Scott
01622 764824/01622 764347

Kingston-upon-Thames
Penni Cotton
(contact office)

**Manchester, North West and
North Wales**
Elycia Ormandy
0161 445 5844

Northampton / Milton Keynes
Michael Walker
07831 347143

**Northumberland, Tyne and
Wear, Teesside**
Peter Bodo
01740 620707

Nottinghamshire
Margaret Bowler
0115 987 5869

Sheffield
Rachel Reeson
07544 353076

Somerset
Lucy Howard
07748636122South

**Birmingham and
Worcestershire**
Gillian and Chris Stock
01527 64988

Sussex
Michele Wilmshurst
01424 421013
Rita Clarke
(contact office)

West Yorkshire
Pauline Ferrari
01977 799565
Jack Lawrence
01977 790886



National Co-ordinator

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Helpline: Freephone 0808 169 1960 - Office Telephone/Fax Number: 0115 987 5869

Email: contact@mdsguk.org - Website: www.myotonicdystrophysupportgroup.org

Editor

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