

MD NEWSLETTER

of Myotonic Dystrophy Support Group

February
2022

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Myotonic Dystrophy
SUPPORT GROUP



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Printed by
L & T Press Ltd.,
Bedford
07831 347143

Hello to all members of MDSG

Another New Year!
Will we remember 2022, and why?
Many of us have so much to be thankful for. Vaccination development and distribution has depended on many researchers and volunteers and we have all benefitted from access to the protection offered.

MDSG meetings face to face may be possible as time goes on. We plan to have our annual 'Rare Diseases Day' meeting in Nottingham at the end of the month. Other area contact families may feel able to arrange local meetings in the near future.

Because we haven't had our Annual Conferences and regular meetings, there has been a significant reduction in the amount of paper work in the office. It was decided not to appoint new staff until we have a need as we return to the usual pattern of office routine. In the meantime we have been joined by Kirstie who is currently assisting us with our office admin.



In recent weeks we have been streamlining various office systems by storing data electronically. Our A4 files are no longer needed, and we are happy to give them to you free if you can make use of them and live locally, to avoid postage cost. Michael Walker continues to work on all MDSG finance matters. The next Trustees meeting will be by telephone conferencing.

My family has had various medical issues to deal with this year. Like all families they have had to cope with Covid restrictions including isolation when necessary.

Last December I was again invited to switch on the Christmas lights in the laboratory at Nottingham University. Photo shows me with Professor David Brook on this annual occasion. Keep safe, have your jabs

Margaret Bowler

Letter from The Editor

Hello Everyone,
"Keep in touch with one another!"
During the first Lockdown, that message was repeated time and again. Similarly people spoke about the importance of detail, the 'little things' that made the days brighter.

This week I had a phone call from a friend, a letter from another and some flowers left in our porch. Each was totally unexpected, and made me feel good. According to recent reports, Her Majesty the Queen is asking us to use the theme of KINDNESS to celebrate her Platinum Jubilee. A thank - you, a nod of appreciation, a wave costs us nothing. How inclusive is that!

So, to all our readers whoever you are, we would like to show OUR appreciation of YOU for being a part of our MDSG Family.

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For you, we send our thanks and a front page springtime flowers. It is a tiny gesture but sincere and heartfelt. Go on. Feel valued, feel good!

Now a request... Please update your address books. Every now and again mail arrives for MDSG addressed to our *old address*. It is only thanks to an eagle-eyed postman/woman that the letter is correctly delivered. Surprisingly this mail is often sent by subscribing members. Please check.

Lots of folk are busy spreading awareness of Myotonic Dystrophy. Some fundraising stories are in this issue. We do thank you all for your enthusiasm and creativity. Read about latest news from the Nottingham based Myotonic Dystrophy research team as we thank Prof David Brook for generating and inspiring interest in the ongoing search for answers to further our understanding of DM.

Very best wishes, **Elycia Ormandy**

Christmas Cards

The Christmas cards we produced for sale were a great success. We printed 200 sets of 10 and sold 170. Originally we were only going to do 100 sets.

Thank you to all those who purchased them as that meant 1700 cards were sent to people, spreading awareness of not just the Myotonic Dystrophy Support Group, but also the condition of myotonic dystrophy.

Subject to approval by the Trustees, we intend to produce them again for this Christmas, with a different design. We know many of you buy your cards early, so we will have them available much earlier, possibly late Summer.

Brochures

With this newsletter, you will have noticed we have sent you copies of our latest brochures, **Painkillers and Myotonic Dystrophy**, written by Dr Ian Bowler and **Lungs and Breathing** written by Lynn Hewitt. Extracts were included in previous newsletters.

If you wish to have additional copies or any of our other publications please contact the office by phone or email.

Anaesthesia and Sedation for patients with Myotonic Dystrophy

Basic Information for Midwives

Bowel problems in Myotonic Dystrophy

Congenital Myotonic Dystrophy

Excessive Daytime Sleepiness and Myotonic Dystrophy

Facts for patients, family members and professionals

Just Diagnosed

Myotonic Dystrophy and the Brain

Myotonic Dystrophy and the Eye

Myotonic Dystrophy Support Group Information

Myotonic Dystrophy Type 2

Relatives Information

Swallowing Difficulties in Myotonic Dystrophy

The Heart and Myotonic Dystrophy

Why do we get new families with myotonic Dystrophy?

Plus;

Tips for diagnosing Myotonic Dystrophy

100 Helpful Hints

How to join the Registry

Facts Book (priced £5)

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Giving Blood

Is a person with Myotonic Dystrophy a suitable blood donor?

Answer;

You can look at this in two ways.

You could ask, is the blood from someone with myotonic dystrophy going to be suitable for someone else?

The answer is, it is perfectly suitable and will not cause any problems at all.

You could also ask, is giving blood going to be harmful for someone with myotonic dystrophy? If someone has myotonic dystrophy mildly there should be no problem. However, if someone has the condition more severely it would probably be unwise. This is because taking a pint of blood has an effect on the circulation and if someone should have heart problems it is best not to do anything which might cause difficulty.

While we are on this subject, a person with myotonic dystrophy, wanting to give blood may get turned down because their blood pressure is too low. People with myotonic dystrophy have a lower than average blood pressure and that applies not only to those affected badly but also those who have it very very mildly indeed.

It has been suggested that this could be one reason why the gene has built up in the population. There are less incidences of blood pressure problems in pregnancy and for those who have the condition mildly it might actually be an advantage.

*This advice was given by the
Late Professor Sir Peter Harper*

Trustees Meeting 22 February 2022

The Trustees, Margaret, Elycia, Claire, Euan, Peter and Michael held their Trustees meeting via a telephone conference call.
Some of the decisions were;

It was felt by a majority that it was still too early to arrange an annual conference on the scale that we were having before the pandemic. With the scientists saying we need to learn to live with it, many people are still worried and concerned. Having a conference for 200 people it would be nigh on impossible to adhere to social distancing.
However, we have decided to restart the local contact group get togethers as a number of you have commented on how you have missed getting together and sharing. With fewer numbers and large village halls and being sensible, we should be able to stay safe.

Many organisations are returning to face to face conferences and exhibitions. We have already booked a stand at the Primary Care Conference in Birmingham in May and the Midwives Conference in Newport, Wales in October. We have also been invited by the Association of Anaesthetists to the annual conference in Belfast in September, with them offering us the stand free of charge.

Claire is working on an idea of producing a card that, if a person has to go into hospital, can be displayed above their bed.

The aim is to highlight how the condition affects you the patient, whether it be walking, grip, speech, swallowing, cognitive or other symptoms.
Despite the lack of fundraising due to the pandemic, we can report that the end of our financial year 31 December 2021, saw us in a healthy financial state.

Professor David Brook new student recruits

We have recently appointed several new PhD and MRes students to work on the myotonic dystrophy project. They are Jessica Petts, Gareth Williams, John Fox and Aiden Cranney who will be working on various aspects of the project from chemistry to computing and from molecules to mice.

They have all received Margaret's Mantra "WORK HARD!!!!!!" and I'm pleased to report that they are all doing well. Best Wishes David

My PhD project focuses on examining the understudied cognitive and neurological features of Myotonic Dystrophy and understanding how expression of the mutation in the brain is leading to the development of cognitive symptoms.

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Hi, I'm Jess and I am a masters student working in David Brook's lab on myotonic dystrophy. I started working here in October after completing my undergraduate degree in biology at Sheffield Hallam. I am currently investigating the potential role of a

group of enzymes called protein phosphatases within myotonic dystrophy and trying to identify which specific protein phosphatase is involved.



"Hello, my name is John Fox and I'm from Leamington Spa. I studied for my undergraduate degree in chemistry at the University of Nottingham and now I'm

doing a PhD also at Nottingham with Professor Chris Hayes and the David Brook group. My research focuses on the synthetic chemistry reactions used to make potential drugs that inhibit CDK12 proteins and potentially treat DM. My work focuses on making the molecules efficiently and quickly and to a high standard so they can then be tested for their effectiveness"

My name is Gareth, I am a first year PhD student at the University of Nottingham. I completed my undergraduate degree at Cardiff University studying Biomedical Science.



'My name is Aiden and I'm originally from Preston, Lancashire. I am a Chemistry PhD student, and I am currently using simulations and machine learning to study potential new treatments for Myotonic Dystrophy'



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H**This Day 30 years ago
6 February 1992**

As I heard the date announced at 7am on Sunday morning February 6th, the date 'rang a bell'.

It was on February 6th 1992, the world was told that the researchers had discovered the gene that causes Myotonic Dystrophy.

This was in the early days of MDSG. I had been informed about this exciting development earlier, but the news was officially embargoed until February 6th.

The researcher who headed the research team that made this genetic breakthrough was Dr David Brook who was then working in America. He later returned to UK to work in the Medical School at Nottingham University, and we now know him as our Patron, Prof David Brook.

I had a phone call from Dr Peter Harper from Cardiff. (He later became Prof Sir Peter Harper)

Dr Harper asked if I could find someone who had myotonic dystrophy who lived in London and someone from Wales. They need to be available to be interviewed by the BBC on the 1pm news today!!

Ken Ware from Cardiff offered and he was interviewed at his office desk. Beverley Bass from London had the BBC in her kitchen, following the hairdressers visit! What a wonderful Day.

A few years later when Prof. Brook was working in Nottingham he said, "In 10 years time we will have a cure"
Many researchers worldwide are working in Laboratories to find, first a treatment for people with Myotonic Dystrophy and then a cure.

The word to all the researchers is **WORK HARD!**

Thank you to all researchers across the world.

Consensus Guidance Meeting

On January 27th 2022, Michael Walker and I joined a zoom meeting to discuss Care recommendations.

This discussion started over 2 years ago, but of course we couldn't meet as a group, because of the Covid 19 infection. Today was the day that Dr Chris Turner arranged for many medical specialists to talk about the specific interests that people had in connection with DM1.

It is very encouraging that so many Specialists are interested in families with Myotonic Dystrophy. The subjects that the specialists people had an expertise in were; Anaesthetics and pain, Respiration, Cardiovascular condition, Rehabilitation, Myotonia, Pregnancy, Sleepiness, Gastro, Eyes, Tumours, Endocrine, Neuropsychiatric, Psychosocial aspects, Palliative care, Emergency care.

The discussion lasted 2 Hours.

Genetic research is continuing. The Researcher's the MDSG support, are Dr Jeremy Rhodes at the University of East Anglia, Prof Darren Monkton, at Glasgow University and Prof David Brook at Nottingham University. All the researchers have teams of people working with them.

A real team of enthusiastic people working for our families with Myotonic Dystrophy!

**International Myotonic Dystrophy
Conference — IDMC 13**

IDMC Osaka, Japan 22-25 June 2022

We have recently had confirmation that this meeting will go ahead.

For those who can travel there will be the usual face to face conference, and the organising committee are arranging to have online access. They anticipate that the majority of delegates will be joining the sessions online.

For the latest details please view on www.idmc13.org

It has been decided that there will not be an official representative from MDSG attending in Japan, but, as always, we will be sending a donation towards the costs of this important scientific meeting.

Robin's Venture for Gaining Sweet Success!!

Robin, together with his carer Hannah, decided to turn Christmas into cash for the Myotonic Dystrophy Support Group. Having many ideas they decided to make a 'clean sweep' by making various different guest soaps including lavender (grown by his parents) and other floral infusions.

It was trial and error but their endeavours were rewarded and the first few bars of soap ran off a very small production line and ready for selling to friends and family and eventually a stall at a Christmas Fair made a small profit. During the times of Covid and shielding quite a few organisations cancelled their events but any profit however small was a profit for the cause.



Not leaving it there they made other goodies, a firm favourite of the family who bought the lions share were chocolate stirrers to stir into hot milk to make a delicious hot chocolate. Perfect for a cold winter's evening.....yummy.



With costs of setting up and buying ingredients Robin and Hannah made a healthy profit of £111 and if the Covid situation subsides they say next Christmas there will be more to come...watch this space.

From very proud parents
Steve & Jayne Ashman.

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Neuromuscular Complex Care Centre Queen Square, London

We know many of you have and are attending the clinic run by Dr Chris Turner and his Team at The National Hospital for Neurology and Neurosurgery.

We asked Dr Turner if there was anything that we could buy that would help the team and the patients. They came up with four items, which the Trustees agreed to buy.

These were, a large fridge freezer, a garment rail with 20 hangers, a Soda Stream with a years gas plan and a Respiratory Pressure Meter with consumables.



Above: NMCCC Team with fridge freezer and garment rail

*Left to right: Standing:
Mary, Reynard, Cara,
Simran, Naj
Sitting:
Sr Mayumi; Dr Deshikan*

*Right:
Members of the team, full of*





Speech and language Therapist Jodi Allen (left) and Physiotherapist Charlotte Massey (right) with their new equipment

Exercise Therapy and Respiratory Pressure Meter

The Physiotherapy and Speech and Language Therapy team at The National Hospital for Neurology and Neurosurgery are offering an exercise therapy, known as Expiratory Muscle Strength Training (EMST). This therapy aims to maintain swallowing and cough function in patients living with neuromuscular disease. This includes patients living with Myotonic Dystrophy Type 1 (DM1).

EMST does not currently have a research base for use with patients with DM1; however we have seen some promising outcomes in patients who have used the trainer. We have written up our findings with one patient living with DM1 in a journal called *Neuromuscular Disorders*. The study demonstrated that cough flow generation in DM1 may be increased by a programme of EMST but further research is warranted. To offer EMST to patients with DM1, the therapy team need access to a respiratory pressure meter. This meter measures the inspiratory and expiratory breathing pressures before and after EMST. It also helps therapists to set the EMST device at the right level for the patient. **We would like to say a huge thank you to MDSG for funding the respiratory pressure meter. It will help therapists accurately measure breathing pressures and build the evidence for using EMST in this patient group.** Jodi Allen (Highly Specialist Speech and Language Therapist)

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Y**New Registry Curator**

My name is Helen Walker, I am the new Curator and Manager of the

[Myotonic Dystrophy Patient Registry](#)

based at the John Walton

Muscular Dystrophy Centre (JWMDRC) at Newcastle University. I come to this role with several years' experience working as the Global Registries Network Manager for the international neuromuscular network TREAT-NMD, and I'm proud to now be using my skills to support Myotonic Dystrophy research in the UK and beyond. I am looking forward to working closely with the patient community and to support and engage with individuals living with this disease as much as possible, so I hope to become a familiar face! Please feel free to reach out if you'd like to chat about joining the UK Myotonic Dystrophy Patient Registry or would like to learn more about what we do and how we can help support research – you can reach me via email myotonicdystrophyregistry@newcastle.ac.uk.

**Facebook**

I recently wrote a few words about Myotonic Dystrophy to put onto the MDSG Facebook page. The idea was that any members who see the post can "share" it with their Facebook friends i.e. send it on to their friends to read who may, in turn, share it with their friends. I am absolutely delighted to say that it reached 10,657 people – and hopefully that includes a few thousand people who had never previously heard of DM.

This is what I posted:

"Can you spare me a few minutes to read about the disease Myotonic Dystrophy [DM] which affects me and my family? Myotonic Dystrophy is a type of muscular dystrophy which is genetic, there is a 50/50 chance of passing it on to your children. It's a strange disease because no two people are affected in the same way – there may be muscle problems [myotonia, irregularities in heart beat], intellectual disability [concentration, organisation], excessive daytime sleepiness, cataracts, problems with swallowing...the list really does go on. The most severe type of DM, called Congenital DM, causes severe birth defects and can be fatal. There is no cure for DM so research is aimed at finding drugs to treat the symptoms of DM. Many families are unaware that they have the disease until a particular issue becomes problematic enough to visit a medical professional and even then the disease may be mis-diagnosed as it is so rare. Those of us who are not in medicine or research can do our bit by raising money but, more importantly, raising awareness of this cruel disease. So, I'm not asking for your cash, just that you share this post as much as you possibly can. Thank you.

I know that Facebook gets much bad press but I'm sure you'll agree that for us folk with Myotonic Dystrophy this post can only help our battle to raise awareness. If any of you have other ideas of how we can raise awareness please let us know – all suggestions considered! If you have access to a computer, tablet or Smart phone please take a look at our Facebook page and join in. You will find us at Myotonic Dystrophy Support Group NB not Myotonic Dystrophy and Muscle Support Group – this is not us!!

Claire McAlonan

Members Stories

I was diagnosed with Myotonic Dystrophy in the early 2000's and as we know the condition can get worse as we get older.

Two and a half years ago I moved into an assisted living accommodation which consists of 54 flats.

Upon arrival, the Occupational Health Therapist will assess your needs. For example riser/recliner chair, raised toilet seat, hoists etc. Each flat has a kitchen, living room, bedroom, wet room, emergency cords, as well as a personal alarm which gives you access to the carers on site in an emergency, who also visit to help with my domestic chores and shower needs. I tend to fall regularly and am unable to get up without assistance and have used the onsite carers several times for help with this.

The block has guest rooms which you can book for family and friends when they want to stay. There are also social gatherings if you wish to socialise. There is a kitchen and communal dining area which provides meals for residents six days a week and a laundry room with washing machines and dryers. There's even a buggy room to store and charge up my mobility scooter with direct access outside, though I could take it into my own flat if I wished.

Overall I am very independent and feel secure in this accommodation, which I found through a housing association with the assistance of my social worker. It may not be for everyone, but those with MD should consider it.

John Cahill

Peter Ware spoke with the local family advisor about hydrotherapy pools, or lack of them in Wales. He asked why swimming pools couldn't be heated to a higher temperature for people with a disability. There are few hydrotherapy pools currently available. Peter loves swimming but public pools are far too cold for him, and he can't feel his limbs when he gets out of the water.

When he gets out, she said she'd not thought of that as she is able bodied and that she would "follow" it up.

Margaret Ware

I was diagnosed with myotonic dystrophy about 20 years ago when I was living with my family but in the last three years my situation changed and I had to live on my own which necessitated using carers regularly every morning. The care agency assessed my needs and have provided carers who have the skills and compassion to carry out daily tasks such as personal care without which I wouldn't be able to live in the private rented flat enjoying the independence that I do at the moment. They also help with the household duties which I am unable to do.

The care agency have provided a carer every morning without fail for the last three years and now due to the nature of myotonic dystrophy I need a carer in the evenings which the care agency is diligently supplying. I am lucky enough to have some really efficient people who go the extra mile ensuring my physical condition is as good as it can be. Also a cheerful smile and positive attitude from what may be the only person I see that day.

Andy

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

Kent
Kirsty Blount and Amanda Scott
01622 764824/01622 764347

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

Cardiff
Margaret Ware
02920 869277

Kingston-upon-Thames
Penni Cotton
(contact office)

Sussex
Michele Wilmshurst
01424 421013
Rita Clarke
(contact office)

Croydon
Lesley Smith

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

West Yorkshire
Pauline Ferrari
01977 799565
Jack Lawrence
01977 790886

Cumbria
Neil Braiden
01228 512385

Northampton / Milton Keynes
Michael Walker
07831 347143

Devon
Josephine Holmes
(contact office)

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

Dorset and Hampshire
Ruth Harrison
01425 626133

Nottinghamshire
Margaret Bowler
0115 987 5869

Glasgow
Michael Kneafsey
01360 311440

Somerset
Lucy Howard
07748 636122



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