

Happy Birthday Dear Margaret.

At the beginning of February, Margaret Bowler celebrated a Special Birthday.

Margaret, we all send you our love and very best wishes for a very happy birthday, and trust that your birthday party with family and friends has given you lots of moments to treasure.

A wonderful celebration for a very special lady.



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Letter from National Co-ordinator Margaret Bowler

Welcome to another newsletter for families who have myotonic dystrophy.

I would like to start by saying a big thank you to many people who have sent Christmas greetings to us in the office. Your greetings are appreciated.

For each of us the past year has some sad and some happy memories. On a positive note, we are always pleased to hear from you and share your stories.

As you will have read in previous newsletters there is encouragement from the researchers as they work hard to find treatment for the Myotonic Dystrophy condition.

Thank you for the many donations that arrive in the office either for Research or for the ongoing work and general running of the Myotonic Dystrophy Support Group.

People are fundraising in so many ways. It is encouraging to hear how much effort goes into raising money.

THANK YOU

Letter from Editor Elycia Ormandy

Hello Everyone,
A warm welcome to all our Readers new and not so new.

The Trustees of MDSG are looking forward to another year of working alongside you to best represent you all in our endeavours for the families that are affected by Myotonic Dystrophy.

Last year we had an excellent attendance at our Annual Conference. This coming year we are hoping that

many of you will be able to join us again; in Nottingham but at a different venue.

**Jubilee Conference Centre,
Triumph Road,
Nottingham University,
Nottingham NG7 2TU**

The Trustees all had the opportunity to visit the Jubilee Centre last year, and feel confident that you will be able to combine relaxation in lovely surroundings while attending the Friday evening AGM and the following day at our Annual Conference. Time now to look at your new calendar/diary and enter the dates.

**Annual All Day Conference 24 June 2017
AGM Friday 23 June 2017, 7.30pm**

There will be a good mix of members who belong to the Myotonic Dystrophy Family and have attended many Conferences, and new families attending for the first time.

We want you to take this chance to have time to meet with others at this very special event organised for you. It is so important to keep up to date with latest developments so that you can make informed choices and be best helped in the management of your condition. Our Conferences also enable delegates to listen and learn from some top specialists in Myotonic dystrophy

Do read all the information in this Newsletter, and try to take part in this year's Conference.

At this early planning stage times and programme details still have to be confirmed, but we will keep you up to date with any changes in the next few weeks.

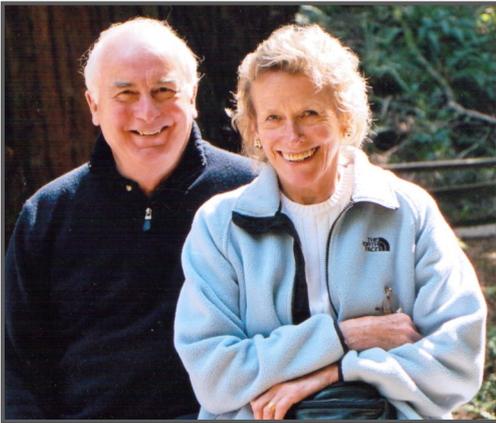
Lastly, please note that our office hours are Tue Wed Thur mornings 9am -1pm. Many thanks. Best wishes to you all,

Elycia.

Trustees

Sue and John Kelly

Whilst on holiday in America last year visiting their son Jeremy and family, Sue had a stroke. Thankfully, she is now back home, recovering and receiving the medical care she needs. Owing to this, Sue and John have now resigned their positions on the Board as Trustees and as Contact people for the Hampshire Area. We wish to thank them for their invaluable contribution over several years and for their time in helping MDSG. We send them both our best wishes and for love and prayers for Sue to make a full recovery.



Euan Cumming

At the last Trustees meeting, we were pleased to co-opt Euan Cumming onto the Board. Euan lives in Southwell, Nottinghamshire.

Greetings

A lovely Christmas greeting was received from **Phillipa Harpin** a former MDC (now MDUK) Occupational Therapist, who wished to be remembered to any member that remembers her from the past.

28th Conference and Annual General Meeting 23 and 24 June 2017

We are keeping the same format as last year, as from the returned evaluation forms, the majority of those that attended, appreciated the three sessions of workshops. This gave a wider scope of topics for people to attend and glean useful information. Many of the speakers will be familiar faces, who give their time and expertise to MDSG and the Conference.

In recent years we have had a speaker from overseas and this year we are pleased to announce a researcher from France, Dr Mario Gomes-Periera has accepted an invitation to come and speak at the Conference and take part in a workshop.

Dr Faraz Mughal, a GP from Solihull, West Midlands, who wrote a case study dealing with a case of myotonic dystrophy and it was published in the British Medical Journal. We are delighted he has accepted our invitation to the Annual Conference.

Lisa Smith and Becky Smith, Play Specialists at City Hospital Campus, Nottingham Hospital Trust, have offered their services to run an official crèche, (ages 0 to 9). Places must be pre-booked. We cannot take bookings on the day.

If you have never been to a Conference because of travel costs etc, please discuss this with me at the office, telephone 0115 987 5869.

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Local meetings

Essex

We had a lovely meeting with Margaret Bowler. There were about the usual number there. 22. Margaret gave a talk about how the support group started and then chatted to everyone in turn. They all thought it a lovely afternoon, which included some lovely refreshments made by my faithful friends in the kitchen.

Gwen Mumby

Northampton

A good number of almost 20 turned up for the local meeting in Yardley Hastings, near Northampton. We had three new families turn up, which was great, especially when two of the families discovered that they were related and the two gentlemen were cousins. **Michael Walker**

Nottingham

Unfortunately, owing to a lack of support, this event was cancelled.

Knaresborough

We met on Saturday 29 October. Although we had a smaller group, six families in all, it gave time for everyone to talk from a more personal perspective. Our guest Sue Manning who is a Neuro-Muscular Care Advisor was able to speak on a number of issues to each individual family.

A current theme for those with MD is anxiety and for some not to be able to articulate difficulties either verbally via their doctor or are unsure about access to the internet, is very common. Also it became clear that deterioration in a loved one had changed quite significantly over the last twelve months. Carers were able to share their feelings especially about feeling so helpless.

Whichever way we look at it MD is a hard, complex, medical disorder with

a need to see a wide range of doctors, often at yearly intervals. Families can see the benefit of joining a group to feel they are not alone. Penny attended in her role as Occupational Therapist. We enjoyed a light lunch and hope to meet again in the Spring 2017. **Roberta Elliott**

Croydon

Over 40 members, armed with their questions, got together on Saturday 29 October to ask Dr Chris Turner his advice on a variety of issues concerning Myotonic Dystrophy. These ranged from individual concerns to those which affected almost everybody, such as day-time sleepiness, night-time breathing, depression, help available and current research.

Dr Turner, in his friendly and relaxed way, began the proceedings by answering questions on a number of topics:

Diagnosis for MD:

Pre-implantation genetic diagnosis (PGD);

Modafinil;

Masks;

The Heart;

Depression:

At this point, it was suggested that we should pause for a 'water break'; which was actually much more than this! Lesley Smith, who had provided the venue for us, also organised tea, coffee, scrumptious cakes..and strawberries. This break is always a good time to catch up with other members, of what now seems to be like a big family, and share experiences. Many of those who were slightly less forthcoming in the Q & A session took the opportunity to talk to Dr Turner during the break to take advantage of his expertise and ask him more personal questions.

Croydon (continued)

Research:

After being suitably refreshed, we reconvened for a shorter concluding discussion which focused on current research and hopes for the future. Dr Turner discussed some clinical trials which are taking place at the moment.

One trial, by Ionis using the drug “DMPKRx” developed by Charles Thornton and his team in Rochester, New York has recently entered Phase 1/2a Trials. DMPKRx dissolves the accumulation of the mutation in the nucleus of cells. Following the meeting, it has become apparent in early January 2017 that IONIS are not taking the drug DMPKRx forward as it does not reach sufficient levels in muscle to be effective. Although this is initially disappointing, Ionis reports that small but encouraging trends in biomarker and splicing changes were observed during the trial, and that this study provided a much better understanding of how future clinical trials and improved clinical endpoints may be used. Ionis are now working on a new muscle-targeting LICA chemistry.

The second trial discussed was PhenoDM1 - a natural history study. One of the problems of finding a treatment for DM1 is knowing when a drug is effective over short period of time (6-12 months). Developing “biomarkers” is the primary objective of the trial and will hopefully enable researchers to know if their drug has helped. The good thing about performing trials within the NHS is that it is one body, rather than disparate organisations, can work effectively together. A number of people present were already helping with

this project and Dr Turner emphasised the importance of being on the DM research registry and offering to be part of the research.

A third trial is a GSK3 beta inhibitor, in Newcastle, which is looking into slowing down the effect of DM on the brain and muscle in DM patient with the congenital form. In Dr Turner’s view, it is possible that this might slow down the effect of DM but it is unlikely to reverse it.

It was good to be able to close the meeting on such a positive note and to know that there is so much research going on to try to find medication for DM. The whole group heartily thanked Dr Turner for taking the time to come to talk to us on a Saturday afternoon, and we all felt that we had learned a great deal from him.

Rare Diseases Day is usually 29 February as that is a rare day.

In your area, please try to have a get together near that date and let your local radio station know they will announce it and that will spread awareness of Myotonic Dystrophy. MDSG will pay the expenses if notified in advance of any planned meeting.

The Nottingham Rare diseases Day meeting will be on Tuesday 28th February. It will be held in St. Paul’s Church, Carlton Hill, Carlton NG4 1BJ The venue has had to be changed as we had so many people come and the room was over crowded. The Church is almost opposite the usual meeting place of the Methodist church.

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Fundraising Concert

In March 2016 we said our good-byes to Jennifer Margaret Scott who sadly passed away with Myotonic Dystrophy. At her funeral, monies were collected in her memory for the Myotonic Dystrophy Support Group (MDSG).

Shortly after, a committee was formed to continue the fundraising efforts, led by cousins Janice and Trevor. Teresa and David Thomas of KeyNote Concerts kindly offered to sponsor MDSG for their annual charity event.

On 3 December 2016 they presented a charity night with award winning cabaret and dance music by Saremma. As well as being a highly successful fundraiser, this event also became a celebration of Jennys life. The evening also featured close-up magician Martin Hinchliffe who donated his fee to MDSG. Refreshments were available during the evening.

Over 170 people attended the fundraiser. One of the highlights of the night was the raffle in which over 50 prizes donated by local businesses and individuals were given away. Her husband Richard later said, "Unfortunately due to ill health I was unable to attend but I'm grateful for everyone who stood up and made the event a success."

In attendance were Dr. Jeremy Rhodes and Dr. Helen Brownlee (far left) who kindly gave a short presentation on Myotonic Dystrophy. They were also available to answer questions that people had about the disease whilst they manned an information table throughout the evening. This was a significant part of the

event because although many people knew and loved Jenny, few truly understood the nature of her illness. There is no cure for the disease that Jenny suffered from, but the family was able to turn to the Myotonic Dystrophy Support Group for assistance. Richard said, "I've always been able to ring them at any time. When I needed support I knew they were there. They know all about the disease because they know about it through personal experience. It's not something that everyone knows about, few people do, and they are able to offer family support during a very difficult time."

On a personal note, I learned a lot from the Scott Family about Myotonic Dystrophy with Mr. Scott giving me pamphlets and a book full of information to read so that I could learn more. I know that I very much enjoyed volunteering to help the Scott family for my scout DofE. Little things like helping to peel veg for Sunday dinner, walking the dog, and especially watching football with Mrs. Scott will be memories I will treasure forever. I would encourage people to help out in any way they can with someone who has the disease just like my community did by raising £1,826 (and still counting) for the Myotonic Dystrophy Support Group.

By Freedom Scott G.R. Tansley
Age 14
Brookville, Norfolk



The power of print: We had a positive response for a member who wanted a 1994 copy of the newsletter. Someone read this and sent back the original copy. See what happens when you read the newsletter.



Unfortunately owing to poor response, the planned contact training day was cancelled. Another date will be rearranged.



Our awareness programme, this year, includes information stands at

Institute of Physicians Annual Conference, Manchester in March;

Royal College of General Practitioners, seminar on neurology, London, March;

Primary Care Exhibition at NEC Birmingham, in May;

Association of Anaesthetists 50th Conference for Trainees, Cardiff, July;

RCGP Annual Conference, Liverpool in October.



Thank you to those members, their family and friends who have organised fundraising events. Please let us know, so that we can acknowledge your efforts. We receive money on a regular basis, but unfortunately, Just Giving or Virgin Giving do not break this down by event. They send us whatever has been raised from all events as one payment.



Government Warm House Scheme: offered by some suppliers in some areas to claimants in receipt of some benefits. An enquiry to your supplier to see if you are eligible may result in

a reduction of £140 off your bill.

New Contact Person

We welcome **Lesley Smith** to our contact team. Lesley is supporting Penni Cotton in the South East London/Surrey area. Penni is looking after the Kingston/Twickenham area and Lesley, the Croydon area. Following a request to tell us a bit about herself, she writes;

My husband Jonathan was diagnosed with MD about 6 years ago. He is now 61 and retired early as he could no longer cope with the daily travelling. We have two daughters aged 24 and 21. The eldest has had genetic testing/counselling and been told it is very unlikely that she has the condition. The youngest is not at a stage where she wants to know. We believe that Jon inherited the condition from his Dad but as his parents divorced when he was very young and his father has now died we cannot confirm formally. His Mum does not have MD. Jon has two brothers, neither of whom are affected with the condition, nor any of his extended family.

Since retiring I try to keep him active, making sure he walks every day and he is a regular Bridge player and a Crystal Palace season ticket holder. He has leg supports and a stick to aid walking but tries to be as independent as possible and is happy to get about on the bus as he can no longer drive. We live in Croydon and I work in a local Independent School as a Data Manager. Prior to working in schools I was a Manager with a Commercial Lending Company where my Law Degree was particularly relevant for the land law and funding processes. The location of my school to home is useful should I be called upon to help Jon out at any time.

Mobile is 07814 501787 and home is 020 8777 6587 and my email is lsmith09@hotmail.co.uk



Heavyweight collection: an amazing £33 was collected by Mr Graham Collinson. It was far too heavy to carry as it was all in copper. He found wheels to do the job. Thank you Mr Collinson for a wonderful fundraising effort.



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Myotonic Dystrophy Research Support

Myotonic dystrophy is part of a group of inherited disorders called muscular dystrophies. It is the most common form of muscular dystrophy that begins in adulthood. The Myotonic Dystrophy Support Group (MDSG) was fortunate enough to receive a very generous bequest from a family who had been diagnosed with this disease. Marion Heller was a long-term supporter of MDSG and had the disease along with her son who sadly died when he was 12. Marion left £147,000 in her will to MDSG, who were able to donate £50,000 of this to work being undertaken at the University.

A small research team, led by Professor Darren Monckton, is working on

They have also just initiated a new study investigating the effects of myotonic dystrophy on the brain, in particular correlating structural brain changes and genetic diversity with the excessive daytime sleepiness that can have such a major impact on quality of life plus are also developing new approaches to population level screening of the mutations that cause myotonic dystrophy types 1 and 2.

Darren Monckton says “support from the individuals and families with myotonic dystrophy are absolutely critical to our research. Without the generous participation of affected individuals in our research studies and the donation of biological samples, we would not have been able to make the considerable progress that we have. Likewise, donations from supporters have been absolutely critical



in allowing us to explore new ideas, purchase essential equipment, and retain key expertise, providing considerable added value to additional sources of funding”.

Many thanks to Myotonic Dystrophy Support Group

understanding the genetic disease Myotonic Dystrophy and were the recipients of the bequest. Their research is centred on understanding how genetic variation in the gene associated with myotonic dystrophy, and other genes in the genome, contribute toward the wide diversity of symptoms observed in families with myotonic dystrophy.

and to the Heller family for such a generous gift. Pictured are members of the MDSG touring the lab and presenting the cheque to Prof Monckton and his team.

Greetings from the Brook lab!

Having just returned from my maternity leave (Ami) I was keen to find out what has been happening on the DM research project and catch up with all the developments in the lab. Under David's direction and supervision there are currently 5 members of staff and 2 PhD students working on various parts of the project, with the majority of us based in the QMC lab but also a couple of people working on University Park in the Chemistry lab.



Following the successful grant application to try and identify drug compounds that could treat DM, Zhefeng and Philippine have been working with a drug discovery company and Matt, our resident chemist, testing compounds in DM cells. They have been looking to see if they can identify any compounds that reduce or remove the DM spots (foci) and testing to see if the cells remain healthy. Their results are then sent back to the company who use the information to make more compounds and to make changes to improve compound activity.

Marzena is working in the lab to identify new proteins and genes that are different in DM cells compared to non DM cells with the hope that they will act as markers to help diagnose DM1 and monitor its progression and response to drug treatment.

Naveed and Marta are both using a new technology to make a model system to help us to understand the effects of the repetitive DNA sequence found in DM and Paulina is looking at

ways to introduce small pieces of DNA sequence into cells to test if this is a possible option for treatment of DM.

All the work in the lab aims to give us more information about what is happening inside a DM cell to direct

the development of possible treatment options.

We would all like to thank the MDSG for your continued support towards our research and wish all members a very Happy New Year.

Dr Ami Ketley



With the donations that were sent to Myotonic Dystrophy

Support Group following Peter Bowler's death Professor David Brook bought a Liquid Nitrogen Container, seen here in his laboratory.

Professor David Brook and Margaret Bowler in the picture with the Canister and label with Peter's name on it.

Thank You all for the donations and to David for his kind thoughts in remembering Peter.



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Dr Mario Gomez Pereira Researcher from France

Guest Speaker at our next Annual Conference in June



I have a fifteen-year experience in research. During this period I have built a solid scientific background in areas of human genetics, inherited disease, molecular biology and neuroscience.

Currently I am investigating the molecular neuropathogenesis of myotonic dystrophy, a devastating triplet repeat disorder. My work led to the identification of molecular synaptic abnormalities that mediate brain dysfunction. These new and exciting data will feed translational research and help identify molecular targets for rational therapeutic strategies. I am in contact with pharmaceutical companies to explore these opportunities.

My scientific curiosity extends beyond my research topic, and it has helped me establish a solid background over the years in genetics, neurobiology and molecular biology. I have extensive experience in paper and grant peer-reviewing and in

lecturing to both specialised and lay audiences.

At a personal level, the international and multicultural diversity that I have experienced as a result of my stay in various countries is an important asset in current times, when science is characterized by an ever-increasing international and multicultural nature.

My post-doctoral experience at Glasgow University working with Professor Darren Monckton and his team, helped me consolidate the scientific and technical skills acquired during my PhD, and complete some projects. During this period I gathered compelling evidence to definitely reject long-standing mutational mechanisms of microsatellite repeat number mutation. I proposed a valid alternative based on experimental data gathered in a cell model system, which I developed and characterised.

Open Day

The Neuromuscular Centre Midlands situated at the Hereward College Campus Bramston Crescent Tile Hill Lane Coventry CV4 9SW had its first Open Day last month.

NMC Midlands
warmly invites you to our
very first Open Day!
Saturday, 28th January 2017
12 – 5 pm

An afternoon dedicated to information and connection featuring:

Taster Sessions
Assistive technologies & equipment
Care, advocacy and support resources

Taster sessions begin at 12:30 and again at 3:30 and include:

- Seated Pilates & Assisted Stretching
- Seated Relaxation techniques
- Gentle and passive exercises to improve breathing and circulation
- Nutrition for those with MD

Also featuring **Myotonic Dystrophy UK**
Afternoon Tea with East & West Midlands Muscle Groups

Register for FREE at <https://nmcopenday2017.eventbrite.co.uk>
Or call Roxanne at 02476 100770 and upon sign-in on the day, you will be automatically entered into a prize draw to win a **Breville Hot Cup!**

nmc
neuromuscular centre
midlands
inspiring independence

We are rebranding!
Come see the unveiling of
our new name and logo

Christmas Concert

Family and friends gathered at the Methodist Church in Radcliffe-on-Trent for a really lovely and meaningful Carol Service just before Christmas. I didn't count the people, but the church was full. The quality of the music and the singing was really good led by the Rhythm and Rhyme Choir whose pianist was Christine Smith. Following the service, refreshments were served including lots of mince pies. It was pleasing to see one of our own members there. I was presented with a cheque by Mrs Thelma Akers for £278.13. The church had chosen MDSG as their charity for donations.



As you can see from the poster, they have many activities planned. Register and pop along to see the good work they are doing for people with Myotonic Dystrophy and other neuromuscular conditions. Let us know if you were able to go.

Flowers

St Saviours Nurseries in Guernsey, Channel Islands have regularly sent donations to mdsG for many years and have provided complementary bouquet of flowers at our conference.

Have a look at their website www.stsavioursnurseries.com for the beautiful displays and gifts and nominate MDSG as your chosen charity with your order, for us to receive a donation from them.
Telephone 01481 265521

Forms

Membership forms are now distributed annually from the date you first became a member. If you have received one with this newsletter, that means your annual membership subscription is now due.

Please re-join as your membership is important. The membership fee is the same as last year £15 per family address.

Conference Booking form

Although it may seem a few months off, it is helpful to the office and administration if you are able to send the booking form back as soon as possible. Thank you, look forward to meeting you once again.

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.

Aberdeen
Jenny Watt
(contact office)

Bedford
Kath Dixon
01234 708520

Cardiff
Margaret Ware
02920 869277

Croydon
Lesley Smith
020 8777 6587

Cumbria
Neil Braiden
01228 512385

Dorset and Somerset
Ruth Harrison
01425 626133

Essex
Gwen Mumby
01245 601343

Glasgow
(vacancy)

Kingston-upon-Thames
Penni Cotton
(contact office)

Leicester
(vacancy)

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

Northampton
Michael Walker
07831 347143

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

North Yorkshire, Knaresborough
Roberta Elliott
01423 868455

Nottinghamshire
Margaret Bowler
0115 987 5869

Pontefract
Jack Lawrence
01977 705496

Sheffield
Rachel Reeson
0114 230 6177

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

Wakefield
Pauline Ferrari
01924 377986

Wiltshire, Hampshire
(vacancy)

Jaine Meridith-Kite who has DM2 is willing to be a contact person for people who have a DM2 diagnosis. Her contact details are 01732 841652