

# CONGRATULATIONS

to

**Tolulope Oyeniya**  
Glasgow, with  
her winning  
poster



and **Dr Ami Ketley**  
Nottingham

on winning awards for their poster and presentation at the recent IDMC-11 Conference in San Francisco.

See their reports inside on pages 10 and 11

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2017**

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## National Co-ordinator

Margaret Bowler

## Directors

L Appleby  
M Bowler  
Dr H Brownlee  
E Cumming  
P Ferrari  
R Harrison  
E Ormandy  
M Walker

## Advisors

Prof D Brook (Patron)  
Dr I Bowler  
Prof P Harper  
Dr D Hilton-Jones  
Prof D Monckton  
Prof R Moxley USA  
Dr M Phillips  
Dr M Rogers  
Dr C Turner

Office Staff;  
Julie and Mike

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## Letter from National Co-ordinator Margaret Bowler

Happy New Year to all our Readers.

Looking back a few months, the research into Myotonic Dystrophy, was described at IDMC 11 – as our “Tomorrow.”

In many parts of the world so many Researchers are working for our families. Excellent results of research by our GB young researchers, who were financed by MDSG to go to the International Conference. (Reports in full are in this edition of the newsletter.)

During the coming year we will be hosting a training day for all Contact family leaders, this day will be a “must” for people wanting to organise an area meeting. It is so important that the leaders of the groups know all about the legal side of the charity as well as the medical and day to day running of the charity.

We do need more leaders for our area meetings, the aim 30 years ago was to have a leader for each county, and we have not yet managed to achieve this within the 30 years.

Could you organise an area meeting? Please contact me at the office to discuss the possibility.

**Margaret Bowler**  
National Co-ordinator

## Letter from Editor Elycia Ormandy

Hello Everyone,  
We hope that this edition may reach some households in time for Christmas. For others it may take a little longer and arrive in the New Year.  
Whenever you are reading this, we hope you will enjoy turning the pages to learn about the Sahara, San Francisco and reports of meetings closer to home. Thank you to each and every one of our contributors. All of these events are brought together in this edition so that we discover what is being done on behalf of our members to raise awareness, encourage research and to work hard on our behalf and for MDSG. Lots of our members and friends have appreciated being able to attend our Annual Conference year on year. In 2018 we are hoping to encourage more families who live in the South to come and join us at The Oxford Belfry near Thame. Put the dates in your new diaries, and seriously consider being there. Also check out the Early Bird offer and don't miss the deadline! Snow is on the way as I write. Trips and falls do happen, so best avoid treacherous weather. Stay in and cosy up with a fleece instead. It's also time for a reminder to get that essential annual 'flu jab, - if you have DM or are a Carer **you do qualify**. Have you had **your** Pneumonia jab? It is generally repeated every 5 years if you have a long-term medical condition. It is very important for people with Myotonic Dystrophy, who may have some respiratory issues. There are some nasty chest infections around. Take care and get any 'chestiness' and wheezes, checked out. No apologies for the reminders! Will we see you at the Conference? With warmest wishes that you will have a Peaceful Christmas and a Happy New Year

**Elycia.**

**29th Annual Conference  
15 to 16 June 2018  
Oxford Belfry, Thame, Oxfordshire OX9 2JW**



We have decided to move from Nottingham to the countryside of Oxfordshire.

Book **NOW** for our next Annual Conference and Annual General Meeting.

Fill in the booking form with this newsletter. To take advantage of the Early Bird offer for accommodation, please book your room before 23 March. MDSG are again offering you the chance of staying Friday evening at half the quoted rate, but that is only if you book the Early Bird offer. If you book after that date, your refund will only be up to the quoted rate.

The AGM will be on the Friday evening followed by refreshments. Saturday will consist of main speaker talks and several workshops.

The Oxford Belfry is a 5 minutes from junctions 7 or 8A of the M40.

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## Marathon des sables 2017

This April I completed the 250km of this multistage, self supported desert ultra-marathon. I am still thrilled when I think back on this once in a lifetime (probably) experience and I can't quite believe that I, a 45yr old mum of three, actually did it.

I only started running in my thirties as a way of combatting stress, but it gradually got under my skin and became an important part of my life. The idea to run the Marathon des Sables crept up slowly on me and when a friend whose family lives with MD asked me if I might run an event to raise awareness for the condition I decided to go for it and I entered!

The race occurs annually in the Sahara in southern Morocco. 1200 competitors from around the world take place in what has been called the toughest foot race on the planet. There are six consecutive stages, (the longest of which totals 90km), soft sand, rocky jebels, seemingly endless dunes and temperatures reaching 50 Celsius. You are completely, apart from a communal tent, self sufficient for the week, so all your food, sleeping bag, mat, torch, medical supplies etc. are on your back!

Despite the obvious discomfort it was the most brilliant experience; the desert is a beautiful, daunting, magical, inspiring place. The views are awesome and varied and most importantly distracting!

When all you have to think about is running, refuelling and getting some rest, the worries of normal life are temporarily banished. It was in many ways a very restful time. My seven wonderful tent mates, of whom my husband was one, completed the experience for me. Comparing blisters and rehydrated rations of an evening is bizarrely pleasant.

I set aside 18months to train for this as I wanted to not just survive it but run it well. My husband and I organised our training around work and kids and tried to run as many of our long run sessions together. But the reality was hours and hours of solitary running. The worry that I might fail and let people down was constantly at the back of my mind, but it pushed me on to train harder.

My children were enormously supportive and accommodating throughout our training, revealing to me that they could actually cook and clean up after all, which made things so much easier!

I have just enjoyed a summer of only running when I feel like it, and I do feel a great sense of relief that it is successfully over. My family and friends and have been hugely supportive and generous with their time and money. Auction nights and sponsored walks and non-uniform days are just a few of the community activities that took place. I have raised over £5000 for Myotonic Dystrophy Support Group and created more awareness about the condition so am very satisfied. But I can no longer face "bombay mix" and definitely don't recommend cold, rehydrated food!

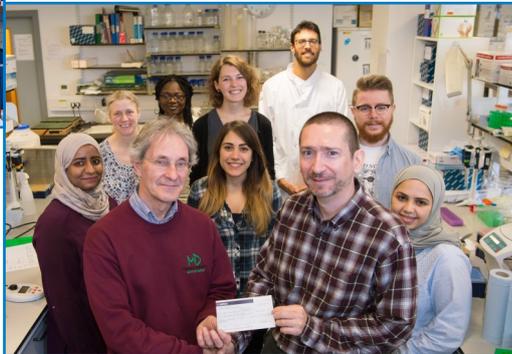
**Johanna Sutton**



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Michael Walker presenting cheques for £1250 each to Professor David Brook (left) and Professor Darren Monckton (below). The money was from donations made at the funeral of his son Nathan.



## Every Little Helps....

A great way to help with our fund raising effort is through the use of our distinctive green Collecting Boxes. A number of our members are using them to



prompt the deposit of spare change, and the occasional spare notes, from local meetings, fund raising events and family gatherings. They can also be used to collect funds raised from the sale of merchandise, with all proceeds sent in together.

If you would like to join in, just contact the office and we will arrange to send a Box to you. If you would like two or more, we have plenty in stock, so just let us know. From there, simply add up the amount you have raised from time to time, and send a cheque into the office (payable to Myotonic Dystrophy Support Group). If you have a story to go with the cheque, feel free to let us have this so we can put this into the newsletter - maybe to inspire other members to try something similar. For more information, please contact the office. **Peter Ferrari**, Trustee.

## Fundraising



Samantha Hawkes (pictured here with funny ears!!) a friend of Trustee Michael Walker, is running the London Marathon in April 2018, in memory of his son Nathan who died earlier this year. She will be running with a picture of Nathan on her vest. Anyone who wishes to make a donation and support her, go to her virgin giving site <https://uk.virginmoneygiving.com/samanthahawkeslondonmarathoncharity>

In April 2018, Steve Welsh will be doing a sponsored walk for Myotonic Dystrophy across Hadrian's Wall dressed in Roman armour. It took the Romans 16 years to build the wall. Hopefully it won't take Steve that long to walk it. Steve's brother-in-law, Andy Smith, died of the condition this year. Andy's son Iain also has the condition. Steve is hoping that Iain, his nephew, can walk some of the way with him. He will be calling at the Centre for Life in Newcastle towards the end of the journey where he'll hopefully meet the research staff. Steve also has a virgin giving site.



## Speech and Language Conference Glasgow

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On 27<sup>th</sup> and 28<sup>th</sup> September Mike Walker (Director MDSG) and Ann Cobburn (A MDSG contact person for Scotland), attended the Speech and Language Conference held at the Conference Event Campus in Glasgow. The objective was to raise awareness of MD. It is a difficult condition to diagnose and takes time and many hospital appointments before the condition is diagnosed.

We had a stall set up with all our brochures, including the new one "Swallowing Difficulties in Myotonic Dystrophy" written by Jodi Allen and relevant for this particular conference.



We handed brochures out during the breaks in the conference to as many delegates as possible (hardly anyone escaped us as Mike was handing brochures to people queuing for lunch, no escape, hard task master! )

I considered the exercise a success as there were many Speech Therapist there who had treated patients with MD and were aware of the condition. Some of the other professionals were not aware but did take on board what we are all about.

After conversations with professionals who were interested in the condition, they took away various brochures for themselves and took even more booklets to pass on to colleagues who could not manage to come to the conference. Quite a number took brochures and said they would place on

their Noticeboards at their workplace or place around their clinics. Very encouraging indeed.

The Events Manager of Conference made a special visit to meet us and expressed how much interest had been raised by the delegates to our cause and invited MDSG to attend another event in London in January 2018.

The organisers of the event mentioned that from time to time they have space in the SALT Journal and would be willing to offer us an advert free of charge, whenever they have a slot. The circulation of the journal is 18000

I would like to say a big thank you to organisers of the event and for giving us the opportunity to get our message about MD out there to professionals.

**Ann Cobburn**

## GP Conference

Elycia and Lynne Ormandy and Michael Walker raised awareness at the GP Conference in Liverpool by having an Exhibition stand. Many GP's thanked us for being there and making ourselves known in a good way. There were over 1200 delegates and we were delighted when Dr Jane Fenton-May a good friend of mdsdg came across and had a word.

## Belfast

Margaret Bowler and Michael Walker arranged a get together for the families in Northern Ireland who are affected by Myotonic Dystrophy. Prof Darren Monckton, Dr's Richard Petty, John MacDonald and Ian Bowler kindly gave up their time to talk to the 21 delegates. Most of the families had not met anyone with the condition before.

## Wales Neurological Association

Margaret Ware, the contact person for Cardiff Area, represented MDSG at the WNA meeting held recently.

**South London Get together,  
Croydon  
Saturday 28<sup>th</sup> October 2017**

On a lovely sunny day, over 20 of us got together in our usual Croydon venue which was kindly organised by Lesley Smith. It was really good to meet so many regular members and also to welcome four new friends into what now seems like our MD 'family'. Lesley supplied badges for us with first names in larger text, which greatly helped when chatting to each other.

After initial re-introductions, Sarah Price (*Respiratory Nurse Specialist at the Royal Brompton Hospital*) joined us to share her knowledge and expertise. She had brought along some breathing kit to illustrate her talk and was happy to answer our questions as she went along. Due to the fact that most people in the group had had a great deal of difficulty in using breathing equipment, the conversation was very lively and Sarah pointed out that many new masks are now available. She began her talk by explaining why breathing and ventilation are so important for MD sufferers because respiratory failure is a leading cause of premature death. She covered the topic under four headings

**Respiratory Problems and Diagnosis:  
Treatment and Benefits:  
Problems and Solutions:  
Emergency Planning and Preventative Measures:**

Following an extended tea/coffee break, with delicious cakes and biscuits supplied by Lesley, we began to share some of the 'tips' and 'concessions' for disabled

people that are available in the London area. It was amazing how many of these we managed to discover. Group members shared tips on concessionary theatre and cinema tickets; reductions for National Trust, English Heritage, Kew Gardens; reduced or free travel including Greenwich ferry; Blue Badge exemption from congestion charge; Motability; *Please Offer Me a Seat* badge; hospital free parking; help with PIP and other benefit forms; the government's Warm House Scheme which is available for all disabled people and can be obtained through gas and electricity providers in October/November.

Our spring meeting, will be held on **Saturday 7<sup>th</sup> April, 2018** at the Hawker Centre in Kingston upon Thames, and we are delighted that Dr Mahalekshmi Desikan from Dr Chris Turner's MD Centre has agreed to talk to us and answer our questions.

**Penni Cotton**

**For Sale**

MDSG have been given a number of wheelchairs and mobility aids. If you would like one *free of charge*, although you would need



to collect, please ring the office and we can explain what we have.

**There are still some 2018 Calendars available**

**12 pictures of researchers and their teams from around the world.**

**Size A4 with hangers, Price £6 plus postage £1**

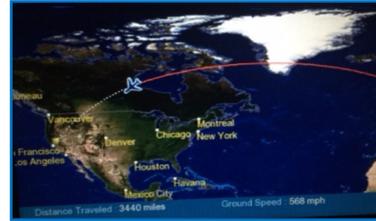
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**International Myotonic Dystrophy Consortium Conference – IDMC 11**  
**5 – 8<sup>th</sup> September 2017**  
**San Francisco, USA**

Margaret Bowler, the National Coordinator of MDSG, was again invited to attend the only world-wide scientific conference for research purely into Myotonic Dystrophy. This is a high profile event, where all the leading research groups – those looking at basic science, potential drug treatments, and therapy based treatments, all meet together to present their findings, discuss research priorities, and network with colleagues from around the world. MDSG was one of the sponsors of this event, and also gave travel bursaries to several UK researchers from centres in Nottingham, Glasgow and Newcastle.

This year, the conference was run concurrently with the Myotonic Dystrophy Foundation (MDF) conference. MDF is a USA charity that has similar aims to MDSG in the UK – increasing the profile of Myotonic Dystrophy, raising money for research, and supporting families with myotonic dystrophy.

The reason for the invitation was that Margaret had been invited to do a presentation to the researchers – some of whom had never met a patient with Myotonic Dystrophy. This part of the conference format was suggested and pioneered by Margaret in previous IDMC conferences. Subsequently, I had been asked to do a talk on anaesthesia to the patients and carers conference of the MDF. So, after an early bus trip to Heathrow..



and an 11 hour flight....we arrived at our hotel which was also the base for the conference delegates. We had a few hours on the first day to do some sightseeing.



And then we were invited to the conference opening ceremony at the Herbst Theatre, a very beautiful venue.

The next day the conference proper started, and we heard many talks from researchers from around the world, including work on: tumours – Patients with DM1 have an increased risk of some benign tumours, including salivary gland, uterine fibroids, and colorectal polyps. There was no increased risk of cancer in congenital or childhood onset DM1 but this may be because of earlier death from other causes. There was an increased risk of certain cancers in classical onset DM1, particularly Thyroid, Endometrium and Melanoma. Patients with late onset DM1 seemed to have a similar incidence of cancer as the general population.

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Brain changes – there are specific changes seen on brain scans, particularly in the frontal lobes of the brain and in the thalamus, which seems to correlate with disease severity.

Sleepiness – there were several talks on this – sleep tests show that the sleep disturbances in DM1 are complicated, are not the same as Narcolepsy, and may be associated with a particular receptor in the brain which is oversensitive.

Heart – a long term study showed that pacemaker and internal cardioverter / defibrillator (ICD) insertion was more common in patients in DM1, that Cardiac Sudden Death occurs about 12-18 times more commonly in DM1 patients than the general population, and that Atrial Flutter was very common, and this was associated with CSD. The risk factors for heart problems were conduction abnormality on ECG, increasing age, younger age of onset, and being male.

Bowel problems – both upper and lower GI problems are common in DM1, and delayed gastric emptying occurs even if patients do not have any GI symptoms. It was suggested that a structured questionnaire was used for bowel symptoms.

DM2 – a new MD2HI scoring system was presented, similar to the one used for DM1.

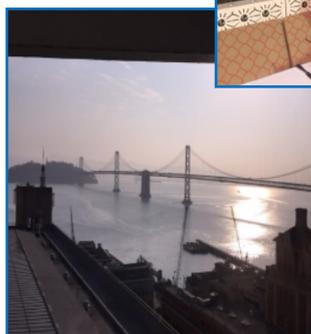
There were also presentations about new drug and physical therapies for DM1, which are in the research



phase at present. We also saw presentation from the Myotonic Dystrophy Foundation, the Myotonic Dystrophy

Patient's Group of Japan, and from a lady from Nottingham who insisted on wearing a green hat!

At the end of the week we were treated to a closing dinner, looking



out over San Francisco Bay, which was very special.

The Myotonic Dystrophy Foundation meeting followed on from the IDMC conference, this was a patient group meeting which included prizes that were given out to the best presentations by young researchers. Three out of the four group winners were from research groups in the UK, and two of those researchers were sponsored by MDSG to go to the meeting – this is a great result for UK research and shows the benefit of MDSG's continuing support of the researchers. I think both Margaret and I learnt a lot from our trip, both from the researchers, the other patient groups, and the very friendly people of California.

The next IDMC meeting will be in Sweden in 2019.

I would like to thank MDSG for paying my expenses to attend the meeting as I thought it was a fantastic experience and has promoted more ideas and international partnerships in research and understanding of Myotonic Dystrophy.

**Dr Ian Bowler**

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**IDMC-11 Meeting**  
by *Tolulope Oyeniyi (Glasgow)*

It was such a great experience to be at the most significant scientific forum for myotonic dystrophy: the 11<sup>th</sup> meeting of the International Myotonic Dystrophy Consortium (IDMC-11) took place 5<sup>th</sup>-9<sup>th</sup> September 2017 in San Francisco, USA.

At IDMC-11, clinicians, researchers and investigators, representatives from the pharmaceutical industry and patient groups, gathered together with a central focus on understanding the effects of DM expanded repeats at the molecular level: the ultimate aim of which is to find an effective target for a cure.

Reflecting upon my enriching experiences at the week-long meeting, there were many sessions with several talks and presentations: insightful and informative talks on improving care and medical management, brain-related effects of DM, and new therapeutics, clinical trials and outcome measures amongst others.

Of particular interest were the fascinating talks from Richard Finkel (USA) and Francesco Muntoni about recent experiences in drug development for separate neuromuscular conditions, including Duchenne muscular dystrophy (DMD) and spinal muscular atrophy (SMA). This provided the DM1 research community with insights into drug development and clinical trials. There was an encouraging update from IONIS pharmaceuticals regarding the DM1 antisense oligonucleotide drug (DMPKRx). This drug had recently completed a small clinical trial, in which results were somewhat not satisfactory: although the drug is safe and tolerable, the drug delivery to muscle was not as effective as was hoped. The IONIS representative

outlined the company's plans to develop a new drug which is more effective in its delivery to tissues of the muscle.

**Dr. Ami Ketley (Professor Brooks group, Nottingham) won an award for her presentation on the identification of a potential key therapeutic target in DM pathophysiology and my research which described a method for population-based screening in DM won the award for best poster presentation.**

Also of note were presentations from the Europe-wide OPTIMISTIC research study: Dr Baziel Van Engelen (Nijmegen) who was honoured with the senior investigator award for his collaborative research and Cecilia Jimenez-Moreno (Newcastle) who won an award for her presentation on cardiac magnetic resonance imaging assessment of the natural progression of cardiac phenotype in DM1. Dr. Mark Hamilton (Glasgow) updated about efforts in the UK to overcome the challenges of measuring brain-related symptoms such as excessive sleepiness and difficulty with aspects of thinking. Dr. Sarah Cumming (Professor Moncktons group Glasgow) shared some insights about careful screening for unusual genetic sequences within the DM1 repeat tract especially for clinical studies and drug trials.

The last day of the IDMC-11 meeting was jointly organized by the myotonic dystrophy foundation (MDF). During this joint meeting people affected by myotonic dystrophy described their own experiences with the condition, with emphasis on the impact of sleep problems on their quality of life.

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I could appreciate the important role of DM patients and care givers in helping clinicians, researchers and therapeutics developers to fully understand the brain-related impacts of DM from the patients' perspective. This underscores how the patients' continuous input will aid in the selection and development of new endpoints for clinical trials.

In summary, the conference was an excellent opportunity for me to interact with researchers, clinicians and families involved. It was greatly encouraging to hear first-hand from such a large number of researchers from around the world who continue to work dedicatedly on myotonic dystrophy every day to gain a better understanding of the mechanisms of the condition and translate the insights gained into better care for people with myotonic dystrophy today, and into new therapies in the near future. Lastly, I would like to express my heartfelt gratitude to MDSG for their generous sponsorship of my participation at the IDMC-11 meeting.

*Tolulope is a doctoral candidate in Professor Darren Monckton's lab at the University of Glasgow.*

### **IDMC-11 by Ami Ketley, Nottingham**

As reported in the previous newsletter, myself (Ami), Marta, Naveed and David flew to San Francisco for the IDMC-11 meeting in September. This is an international conference of DM researchers and clinicians and is a valuable experience in finding out what is happening around the world in DM research as well as sharing our recent results with the field. We travelled by various routes with Naveed winning the prize for the longest detour... via Iceland!! Having arrived safely we had a day to settle into our surroundings

and do the standard site seeing activities such as seeing the Golden Gate Bridge and Pier 39. The conference started with a keynote speaker, describing new methods for manipulating genes and giving the audience ideas of how we could use these methods in DM. This was followed by a welcome reception and then the following day the full program of the conference began.

The key themes of the conference included therapeutic targeting of RNA, disease mechanisms, clinical, ethical and social issues, new models that are helping us to study DM and information about patient registries and trial designs. I was lucky enough to be invited to present my work as an oral presentation. This was a great opportunity for me as we have some interesting data that we have been working on for the past few years. Naveed was selected to give a 'flash' poster presentation; a 3 minute introduction to the work he had been doing and Marta presented her work as a poster presentation and had lots of interest during her session with people asking lots of questions.

**I have attended the IDMC meetings since 2011 and have seen the promising developments in the work presented and the drive of the research field towards clinical treatments and approaches.**

The week is an inspiring experience and we all agree this has given us new ideas and motivation for our work back in Nottingham.

We are so grateful to the MDSG for providing us with the funding to attend this conference and the support it gives to the lab.

Thank you all for your continued support! Best Wishes,

**Ami (Dr Ami Ketley), Nottingham**

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## 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  
01224 249591

**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**  
Ann Cobburn

**Kingston-upon-Thames**  
Penni Cotton  
(contact office)

**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

**Nottinghamshire**  
Margaret Bowler  
0115 987 5869

**Pontefract**  
Jack Lawrence  
01977 705496

**Sheffield**  
Rachel Reeson  
07544 353076

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

**Wakefield**  
Pauline Ferrari  
01924 377986

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

