



# NEWSLETTER

## Autumn 2015

Issue 26

A MESSAGE FROM THE CHAIR

IN THIS ISSUE

Liz Williams, Chair FSH-MD Support Group



Dear Members, this issue includes an overview of our FSH-Get Together held in Newcastle on 5<sup>th</sup> September, Natalie Woodcock has written regarding her latest fundraising endeavour for Action on FSH, a campaign run by Muscular Dystrophy UK along with her ideas for her next project. I have written an update regarding the Ironman event Chris Finister took part in to raise funds for FSH-MD Support Group

*FSH-MD Support Group UK Get Together Gill Penny*

*Ironman Lanzarote fundraising for FSH-MD Support Group UK, Liz Williams*

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*The Scapular Fixation updates will now be in the Spring Issue.*

## FSH-MD Support Group UK Get Together September 2015

### Newcastle upon Tyne

Gill Penny

This year's Get-together was held in Newcastle upon Tyne, the first time we have been to the North-East. The venue, The Beacon, was on a hill just outside the city centre, and our main room was large, light and airy, with windows and a terrace that gave a broad view over the city. Catering was good, facilities reasonable, and the venue staff looking after us responded very promptly and cheerfully to any of our requests.

Numbers were slightly down on last year, which was a little disappointing but there was a good mix of members living in the north-east and those who had travelled further. There were some familiar faces but many were attending a Get-together for the first time. Whilst there was a

programme of talks there were quite a few breaks that left people plenty of time to mingle and get to know each other, sharing stories and experiences. At the end of the day, everyone I spoke to felt that they had enjoyed and benefitted from attending.



The first set of presentations focused upon the work being carried out in Newcastle, with Professor Hans Lochmuller from the recently named Lord John Walton Centre for Muscular Dystrophy, providing the initial overview of the

team. The Centre brings together five areas of activity for neuromuscular disorders consisting of clinical care, clinical research, diagnostics, basic research and strategic partnerships and networking. It is also the base for the FSH Registry. Professor Lochmuller explained the nature of the research, the reason for the registry in terms of supporting clinical trials and the importance of standardising questions so information can be shared with other registries. He pointed out that the small numbers of people with FSH meant there was a need for researchers to collaborate globally in order to obtain sufficient numbers of suitable patients for their particular clinical trials. He emphasised the importance of networking for both service and research staff when dealing with rare diseases such as FSHD. In response to questions he touched upon the relevance of the blood bank where proteins are stored and explained about the use of muscle tissues for clinical research, for example when testing the efficacy of a drug through pre- and post-tests. Whilst genetic testing has taken over from muscle biopsies for diagnosing FSHD, they are important for research, as are blood and skin samples. The latter can be reverted back to stem cells which have the advantage of being live so that they can see what's happening more clearly.

Then Geraldine Bailey and Edwina Perkins, Specialist Physiotherapist and Regional Care Advisor for Muscle Wasting conditions respectively, spoke about their roles within the Service. Geraldine said that they had developed the Exercise Guidelines hosted on MD-UK website and emphasised that she was happy to work with physios elsewhere, including non-NHS physios, to advise on appropriate exercises for those with MD. She also mentioned that UK-MD was running an up-skilling event for physios. Edwina had brought with her a range of leaflets relating to benefits which she realised were a particular issue for those with MD and was happy to answer questions and concerns individually.

Libby Wood from the FSHD-Registry Newcastle Centre for Life then took us through some key points about the Registry, talking about her role, the nature of the information asked for, how it was used, and what was currently available in aggregated form. She emphasised the links with other registries, noting that they were in contact with 13 which all used the same questions. The registry has been growing steadily since its inauguration in 2013. It is receiving up to 20 registrations a month and currently has 560 people registered on the database. Some of the general patterns of the data show that more people are registered in urban areas, that the bulk of registrations are people with FSHD1, with only 9 having FSHD2;

the oldest person registered is 83 years of age with the majority being between 30 and 60. Most can walk independently up to the age of 40 and then begin to use some form of assistance. About 25% of people on the register had had scapular fixations. Fewer than 10% required ventilation and about 20% had some form of hearing loss, although this could be a feature of natural ageing. The average age of diagnosis was 29 years. The key uses of the registry were in planning clinical services, recruiting people for research, doing research and disseminating information on the findings for both care and therapies. Libby took us through a couple of examples of the use of the registry for research, from research into the effects of scapular fixation to the development of outcome measures to establish the effectiveness of treatment. She finished by emphasising the need to join the registry and, if we were already on it, to keep our entries up to date.

After the morning break for coffee, Professor Lochmuller took us through the talk that Debbie Smith, Higher Specialist Trainee Scientist at Bristol had been scheduled to give, but had withdrawn through illness. She had kindly sent up her slides, and Hans more than effectively stepped into the breach. He described the work carried out at the Bristol Genetic Testing Laboratory and the key aspects of testing for FSHD. The description of the genetic basis of FSHD, the different pathways involved in FSHD1 and FSHD2 and the way in which our understanding of the disorder has developed historically was one of the clearest I've heard. At the end of the talk he answered questions about inheritance patterns and the relevance of IVF for those with FSHD.

Teresa Evangelista then updated us on the recently published guidelines and recommendations for FSHD patients by the American Association of Neurology [AAN]. The authors had read and analysed over 60 published articles, reviewed them and condensed their findings into these guidelines which allocate a value from A to D for each recommendation, comment on the quality of evidence and provide a definition. The Guidelines are published in the journal *Neurology* and are available on the TreatMD website –

<http://www.treat-nmd.eu/about/network/>

The last of the morning slots was filled by Maria Field, Senior Manager and Helen Leuchars, Co-ordinator of Pinpoint Health and Home Care. This is a private company that works with people with neuromuscular disorders to help them define and then reach their personal goals. They described how they worked with their clients, helping them to clarify what they wanted

to achieve and then providing practical and emotional support to enable them to do so. Support could involve dealing with practical issues such as when services don't respond, or where different services are not working together to provide an effective service, or the transfer from child to adult services, and helping people at any stage of life to develop skills, make contacts, and get the support they needed. They have helped people to achieve a wide variety of goals, with examples ranging from going to the cinema down the road right through to going to Australia. Whilst they said that they can't always guarantee to achieve everything a client wants they will keep trying to open doors to make things happen for them. Whilst Maria and Helen are based in Gateshead, the company is a national one, so is available in areas as far afield as Aberdeen in Scotland down to Southampton on the south coast.

After lunch we had two talks, the first from Professor George Dickson of the School of Biological Science, Royal Holloway, University of London who spoke of the work being carried out in their laboratory which they hope will lead eventually to the development of drugs to suppress the effects of DUX4. He took us through a range of issues to do with muscles and muscle cells and how a drug might work to reduce the effects of the disorder. Professor Dickson stressed that this was a long term project and we should not expect drug therapies in the immediate future, but he was positive showing that there had been developments in our understanding of FSHD, which at least suggested ways in which treatments might be effected.

The final speaker was Peter Sutton from MD-UK who provided an update on the Advocacy Team, Action for FSH Fundraising, and Muscle Group Meetings, all of which confirmed the increasing degree of support being offered by the charity for those with muscle disorders.

Altogether I found the day to be a highly informative and enjoyable experience where I increased my understanding of the work being done for those with FSHD, caught up with old friends and acquaintances and met new ones. Very well worth the trip 'Up North'.

Gill Penny.

## Christopher Finister's Ironman Lanzarote Fundraising for FSH-MD Support Group UK May 2015

Back in October 2014 Christopher Finister, my partner of 24 years decided to take part in Ironman Lanzarote. For those who are not aware of what an Ironman entails this is a swim of 3.8 km swim, a 180.2 km bike ride and as if that is not enough a 42.2km marathon. In old money ; a 2.4 mile swim, 112 mile bike ride and a 26 mile run.



### Back at the hotel tired but alive!

As a part time wheelie, the logistics of even going to watch Chris filled me with more than a little apprehension but after extensively looking at reviews of hotels on the island, a bit of research with holiday companies who specialise in holidays for people with disabilities and scouring Trip Advisor for clues we decided it would be possible for us to support Chris' challenge as a family like anyone else could.

Chris paid all costs to enter so all funds raised were donated to the Group directly. We (Liz Williams, Ralph and Ronnie) wanted to see him complete this challenge and we decided to make this a family holiday too (again to make it clear all costs were paid by us). This posed it's own challenges as I rely on the use of a wheelchair outside of the home and Chris for that matter. Chris was clearly going to be indisposed so after the initial puzzle of finding an accessible hotel with roll in shower/wetroom, the next issue was how was I going to be able to watch this event without help? We hired a powered wheelchair which was delivered directly to the hotel and was there when we arrived 2 days before the Ironman event took place. I had two days to master the new wheels and wow were they a God send. We certainly looked a sight at the airports with a bike case, 3 suitcases, a wheelchair and two children in tow. I think we were more worried about the bike than the wheelchair in the hold of the plane. We also hired a car (VW Caddy) to carry us all to our hotel and to use to explore once the big day was done.

Our Hotel was lovely, completely accessible ... all areas and even a ramp into the swimming pool. There was easy access via various ramps to the main street in Puerto Del Carmen. It also happened to be on the route of the cycle part of the Ironman and the final marathon run, so I could get to see Chris 4 times on the course and then get down to see him at the finish line. On the morning of the Ironman the Hotel put on a special Ironman breakfast at 4:30 am for the guests taking part.

Chris went down to the start with his bike, wetsuit and various potions of drinks he'd made up to replace salts he would lose through sweat etc. When I managed to persuade the kids to get up and have a leisurely breakfast, Chris had already completed the 2.4 mile swim and was out on the cycle ride through the Volcanic plains.

It was very windy so this probably added at least half an hour to his bike ride of 112 miles which he completed in 6 hours 36 minutes. We had driven the course the previous day to look at the descents he would have to navigate and these were hair raising. I have to admit it made me worry. He knew what he was in for and we saw him come in to Puerto Del Carmen before 3 pm more than a little relieved. The atmosphere was lovely, people were running for all sorts of charities as you can imagine and it was such a lovely experience to watch especially after I initially thought I might not be able to due to my lack of walking skills. Also a member of Chris's running club and his wife came out to watch, by that I mean added two days to their planned holiday specifically to help me if I needed it. (I had never met them before and the sentiment was quite overwhelming really, what a lovely gesture). We met up near the finish. They were so generous by helping me navigate the crowds, assisted in getting family photos and even took our children down to find Chris once he had crossed the finish line. That would have been pretty much impossible for me to do. We were overwhelmed at the support we received either via donations or other acts of kindness. A huge thank you to all who donated for Christopher's Ironman Lanzarote. Chris completed his Ironman in 12 hours, 32 minutes and 12 seconds. I am pleased to announce that he raised £576 in total for the FSH-MD Support Group UK. An experience we will never forget for all sorts of reasons, all good ones.



**All together at the finish!**



**The finish line 12 hours, 32 minutes and 12 seconds**

## Fundraising for FSH-MD Research

### Natalie Woodcock

As many of you already know, along with my family, I have run several events over the past three years to raise money for research into treatments and hopefully ultimately a cure for FSH Muscular Dystrophy. These events have ranged from car boot sales and 'Chelsea' afternoon teas to the more adventurous auction held by Nottingham City Transport at the Album Bus Industry conference. In late 2014 we made the decision to formalise our fundraising efforts and set up a fundraising group within Muscular Dystrophy UK – named 'Muscular Dystrophy Campaign Worksop' and our stated aim is to try and raise £10,000 per year towards essential research into FSHMD.

Our first major event came when we were able to secure the exclusive use of Lindrick Golf Club for a charity golf day on 26 June 2015. It was quite a coup as Lindrick holds a special place in golfing history, in that it was the last venue of the Ryder Cup in which Great Britain and Ireland, captained by Dai Rees, won the only post-war victory. Thus Rees became only the third and last British captain to lift the trophy.

After several months of planning and persuading we secured the support of not only two major sponsors, Scania (Great Britain) Limited (see photo) and Keltruck, but a further 30 companies and individuals who generously sponsored holes, raffle prizes, competition prizes, food and drinks. Due to the prestigious nature of the golf course we quickly filled the 32 team slots and even ended up with a waiting list! The day went exceptionally well and even the weather, despite a very short period of rain in the early morning, was good for the golfers. In all we raised £15,000 which has been invested in FSHMD research.

Quick to follow was the 'Pop-up' café at the Thorpe Salvin Garden Trail where I was banned from the toasted tea-cake station as I burnt the first three orders of the day – things quickly improved and we sold out of cakes by the end of the first day and had to rustle up a selection of crispy cakes and pavlovas for the Sunday customers. Coupled with more car-booting and the collection tins placed in our local pub I am just about to donate a further £500 which brings our three year fundraising total to £30,000. This has

made us realise that slowly but surely every little bit helps towards our target amount and definitely spurs us on to do more.



And so after a few weeks 'holiday' from fundraising we are throwing around ideas for next year's challenge. Our thoughts are ranging from a May Ball or possibly another golf day but I am currently trying to work out the logistics for launching a '100 day challenge' in January 2016. The idea is for 100 people or companies to sign up and commit to raising £100 in 100 days with a special prize for the first person to raise their target amount. This is where I would love your help - please can you post any ideas you have for how to raise £100 – it's much easier to launch a challenge like this if we can give people ideas as to how they can raise the money within the timescale! You can tag me (Natalie Woodcock).

So watch this space for an update and I hope to hear from you soon with your ideas. Natalie

## ACTION ON FSH



Please like the Action on FSH page on Facebook to receive updates of Natalie's next challenge. All the money Natalie raises via this challenge goes direct to FSH Research and it would be great if we could share this challenge far and wide.

## Disabled Ski Session

### Mandy Altoft

So, 2015 was upon us and me & my partner (who is blind) wanted a new 'thing' to do. Inspired by the In-Laws who were going skiing at the end of February we started looking about and we came across Knockhatch Ski Centre in Hailsham that hosts a disabled ski session on the first Saturday of the month from 6pm-8pm. They also offered a come & try session for 50p, so we thought why not, let's give it a go. If we don't like it, it's only cost us a quid!

After only a couple of emails we were booked in for Saturday 7th February, each assigned our own instructor & trainee so off we went!

Knockhatch was very easy to get to from Brighton, about 25 minutes' drive. It has a huge carpark but it's not very well lit and isn't Tarmac, so beware of the bumps! Sign in at the little hut, get a high vis which has a symbol on the back denoting your kind of disability (Lorna's had an eye on it) then off to the main cabin to collect skis, boots & helmets and meet our instructors.

Everybody was friendly and welcoming and went out of their way to help us, though if I'm honest I think Lorna's guide dog Mac got the most attention!

As we had Mac with us they'd arranged for Lorna to go first from 6-7 then I'd have my turn 7-8 so Mac didn't have to be left alone, nice touch.

The slope is huge & has a nursery slope too so that's where Lorna went with her instructor, I'll just mention at this point that the slope is outdoors so if you want to spectate bring your blanket and put thermals on! I managed 10 minutes of spectating till I gave in and went back to the warmth of the main cabin/cafe.

After her hour session Lorna came back saying how good it was so I couldn't wait to get out there! My instructor Paul went and got me a helmet, pink of course! He happily wheeled me out to the bottom of the slope where our trainee, Martin, was waiting with a sit ski. Paul explained everything and talked me through what would happen, how I'd be strapped in etc. I can still walk a bit so I was able to transfer to the sit ski easily, it is very low to the ground but Paul lowered me down into it no problem. Then I embarrassed myself, whilst strapping me in he asked if I was wearing a bag, my reply, no, I left my handbag in the car! Then it clicked what he really meant!!



Once I was strapped in securely we did a practice of how to fall correctly, Paul would shout brace & I had to cross my arms in front of me, easy peasy! "Don't worry though Mandy, I've never fallen so you'll be fine" Paul cheerfully tells me, "first time for everything" says Martin, oh how we laughed!

The sit ski has several ropes in the front and a loop to attach it to the drag lift We get in line and then it's our turn, Paul said we're going all the way to the top of the slope, arghhh!!!

We get hooked on to the drag lift and we're off! But then halfway up it stops, the safety cut off has kicked in and we have to wait till they get it going again. 2 minutes later and we're ready to try again. Unfortunately, the lift jerks as it sets off again. Paul is pulled forward, loses his balance and both of us fall into the ditch at the side of the slope whilst the lift is still moving. Thought he said he never falls, but after the bag incident, I'll put that at one all! I wasn't hurt and neither was he. Several people rush to our aid and I'm soon back in an upright position and we're back on our way up the slope. At least I got to practice a fall early on in the process!

We get off at the top and he shuffles us over to the edge of the 110m slope. He says "ready?", I say "ready!" and off we go! The wind is freezing but it's worth it. He steers us from side to side and I almost touch the surface at the bottom. As we were going so fast he had to brake hard (when stand up skiers swoosh to the side to slow down!) and the sit ski went very low! After our fall incident earlier we only had time for one more run but I think I got my 50 pence worth!

The sit ski I was using can be used without a pilot if the user has a reasonable amount of upper body strength as it can be steered using special ski sticks called Outriggers. They look like a tiny crutch with a tiny ski on the end. Once I've had a few more goes I'll give it a try and see if my winged scapular can handle it. If not, they have a brand new piece of equipment called a Kart Ski, which has a bar in front of the user that is attached to the skis and is used to steer. This can be used easily by people with poor upper body strength.

So, that was our taster session at skiing and we both really enjoyed it, our aim now is to get well practiced and get to the stage where Lorna could be my sit ski pilot and we can go on the real slopes. Was Paul phased when I mentioned this was our aim? Certainly not! But hey, he never falls right?

Mandy

## Useful Links and numbers



**Email:** [fshgroup@hotmail.com](mailto:fshgroup@hotmail.com)  
**www:** <http://fsh-group.org/>  
**Tel:** 01280 840456



**Email:** [info@musculardystrophyuk.org](mailto:info@musculardystrophyuk.org)  
**www:** <http://www.musculardystrophyuk.org/>  
**Tel:** 020 7803 4800



**Neuromuscular Centre Midlands**  
**www:** <http://nmc-midlands.co.uk/>  
**Tel:** 02476 426084

Neuromuscular Centre  
Cheshire

**www:** <http://www.nmcentre.com/>

UK FSHD Patient Registry

**www:** <https://www.fshd-registry.org/uk/>