



# NEWSLETTER

## Spring 2014

Issue 23

A MESSAGE FROM THE CHAIR

IN THIS ISSUE

**Liz Williams, Chair FSH-MD Support Group**



Dear Members, I am excited to bring you our Spring Newsletter with details of our Get Together on 28<sup>th</sup> June 2014 at the Daventry Court Hotel. We have news on a proposed research project from Christopher Graham PhD, Trainee Clinical Psychologist. Natalie Woodcock's Fundraising endeavours for the Muscular Dystrophy Campaign's Action on FSH Appeal. Libby Wood has written an article with an update on the FSH registry.

*FSH Registry update: Libby Wood*

*Fundraising: Natalie Woodcock*

*Louise Moyle, King's College London*

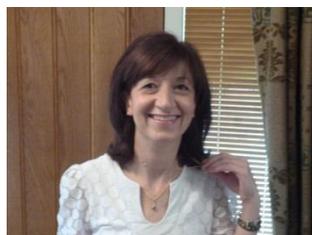
*Online Psychology Study: Christopher Graham*

*FSH Support Group, Action on FSH fundraising update.*

*MDC Advocacy Ambassadors*

*FSH-MD Support Group Information Day/Get Together.*

*Neil Withers Adventures in Bali*



**Natalie Woodcock:**

**FSH-MD Support Group UK Committee**

Hi everyone,

I'm Natalie and as I have just been appointed to the FSH-MD Support Group committee I thought I'd use this opportunity to tell you a bit about myself and why I have become involved with the group.

My journey here started about 7 years ago when our family of four received the news that one of our brood had FSH – a condition we had never even heard of at that point. Although we were all obviously devastated at the time, as parents we have had to take on board the amazingly positive attitude our daughter has shown and her determination to resolve the challenges of her condition in her own unique way – from inter-railing with her uni friends to travelling to the Cannes Film festival under her own steam she aims to do as much a travelling as possible for as long as she can.

With this in mind, once our youngest had flown the nest I was determined to raise as much money as possible for research into FSH, this determination was increased when I visited the Centre for Life in Newcastle and could see firsthand the dedication and enthusiasm of the

researchers working toward the development of new therapies for a whole variety of muscle wasting conditions.

So you can now guess one of my main interests is both raising awareness of FSH and fundraising for existing and future research projects. I hope to kick start both major funding initiatives involving as many willing members of the group as possible and also encourage the continuation of the vitally important smaller individual fundraising initiatives I've heard so much about – I feel these form the backbone of fundraising and awareness.

Over the past year I have held a variety of fundraisers at home. In fact it's a standing joke amongst my friends that you have to pay an entrance fee to get into our house – but in my opinion £5 for a glass of wine and some nibbles is cheaper than going to the pub!

I would love to get a Facebook conversation going to hear about your successful events and ideas for new ones. Over the past year I have held a 'pampered chef' party, wine tasting evening, Christmas party, carol concert, vintage stall and deposited a number of fund raising boxes in local pubs – all small in themselves but add up to a significant sum over the year. If you would like to contact me please feel free to do so through the FSH- MD support group page – I would love to hear your ideas.

**Natalie Woodcock, FSH-MD Support Group UK**



## FSH Patient Registry Update

The UK FSHD Patient Registry was launched at the Muscular Dystrophy Campaign's information day last May and now, less than a year on, over 360 people are registered. The registry is open to all people with a confirmed diagnosis of FSHD. It aims to make research easier and faster by helping to recruit people into clinical trials and research studies providing a link to the research community for everyone involved. The registry is funded by the Muscular Dystrophy Campaign (MDC) through the Action on FSH Appeal. Being part of the registry will ensure you are kept up to date with the progress being made into research and may provide an opportunity for you to be involved.

The easiest and fastest way to register is online at:

[www.fshd-registry.org/uk](http://www.fshd-registry.org/uk)

Alternatively you can request a paper copy from the curator, Libby Wood (0191 241 6840).

Professor Hanns Lochmüller is in charge of the registry at the International Centre for Life in Newcastle. He and the registry curator work closely with the support group and Andrew Graham, Stuart Watt and Suzanne Watt are part of the registries steering committee. This committee which also includes doctors and scientists from across the country is in place to help monitor the registry's activities and make decisions about how the data in the registry will be used.

It is important to mention that your personal data will never be disclosed to a third party. The registry may contact you about research that you may find interesting. The registry would provide you with the details of the researchers allowing you to choose if you would like to make contact or not. Being part of the registry does not mean that you have to take part in any research, this is always entirely voluntary.

Some of the information collected will help inform future research or help develop standards of care. In particular the registry collects additional data about pain, quality of life and scapular fixation. The data held within the registry may be analysed and presented at meetings, conferences and in scientific journals but this will never contain any information which could be used to identify you.

### Already registered?

If you are already on the registry it is really important that your genetic details are included. You are asked to nominate a doctor to provide this information when you first register. If your doctor is not listed then please get in touch with the curator (contact details below) providing the name and contact details of your neuromuscular specialists. Alternatively if you have a copy of your genetic report this can be sent directly to the registry in Newcastle. Please remember to update your details once a year, this ensures the registry contains the most up to date and accurate information. You will receive an e-mail advising you when it is time to update.

Prof Lochmüller and the registry team would like to thank everyone involved so far, we are confident we can reach 400 before the next FSHD meeting in June 2014. A significant amount of data has already been collected and some recent statistics are available in the [registry newsletter available on the website](#).

**Please contact the registry curator, Libby Wood, for more information.**

**Email:** [fshdregistry@treat-nmd.eu](mailto:fshdregistry@treat-nmd.eu)

**Telephone:** 0191 241 8640

# ACTION ON FSH



Update on my research, March 2014.  
Kings College, London.

Louise Moyle

My PhD aims to find out how adult muscle stem cells are affected in facioscapulohumeral muscular dystrophy (FSHD). FSHD is caused when a gene called DUX4 is turned on in muscle. Genes are the instructions needed for a cell to make a protein (a bit like a recipe). Therefore, when the DUX4 gene is turned on, the muscle produces DUX4 protein. People who do not have FSHD do not have DUX4 protein in their muscles and unfortunately, DUX4 protein in muscles is harmful. If DUX4 is made in the adult muscle stem cells, it can prevent them from doing their essential job; maintaining the muscle and repairing any damage. Over time, DUX4 production in the muscle stem cells of individuals with FSHD leads to the clinical symptom of muscle wasting.

I work in the laboratory of Professor Peter Zammit at King's College London, who specialises in growing muscle stem cells in a dish. To model FSHD in the lab, we compare healthy muscle stem cells to those that we add DUX4 to. By knowing how DUX4 protein is harmful to the muscle cell, we can have a better idea as to how to block it using drugs. Throughout my PhD we have discovered some ways to block the harmful effect of DUX4 in our model system. The next step is to see whether we can have the same beneficial effect in muscle stem cells taken from FSHD patient biopsies. To do this, we are planning to collaborate with another lab that is also researching into FSHD. One of

the great things about science is that people from around the world work together and combine expertise in order to help solve problems.

I am in the final year of my research now, and recently I have been writing up the results of my experiments for my thesis, and hopefully a future publication in a scientific journal. As you can see, we are at an early stage of research and there are many hurdles to overcome before any FSHD therapy becomes available in the clinic. However, it is an exciting time to be working in the field – so much has been discovered since I started in 2010!

The last three and a half years have been an amazing experience, and I have learnt so much along the way. None of this would have been possible if it wasn't for all the tireless fundraisers and campaigners pushing to get funding for research into this rare but devastating disease. Therefore, I would like to thank all of you for your dedication and commitment, and I hope that I can help push the research one small step towards a future cure.



**Picture: Adult muscle stem cells, satellite cells highlighted in red/green.**

**Louise Moyle**

<http://www.muscular-dystrophy.org/fshappeal>

## Now Recruiting: An online study investigating the importance of psychological factors over time

Along with medical professionals from across the UK, for the past four years I have been studying psychological processes in adjustment to muscle disease; in particular understanding why some people adjust well to life with muscle conditions (including FSH-MD), even with symptoms at their most disabling, while others struggle to adjust. The reason that we have been so interested in this is quite simple: If we know why some people adjust well to living with their muscle condition, then we can use this information to develop interventions to help people who are struggling to adjust. And this is what we are now beginning to do.

I would like to take this opportunity to discuss our research to date, and then to introduce our online study which is now recruiting.

### Previous studies

Our very first study involved administering questionnaires measuring mood, quality of life, and level of disability, alongside questionnaires measuring psychological processes (optimism, a person's beliefs about their illness and coping methods). The results returned a number of interesting findings. First, psychological processes were often more strongly related to quality of life and mood than was level of disability; and, second that of these psychological processes, people's beliefs about their illness were most important in explaining quality of life and mood. Together this suggests that how a person views their muscle condition is important in explaining their well-being.

A second study was then undertaken. It aimed to establish which ways of viewing one's illness are either helpful or unhelpful. Here a profile of beliefs which were based

around acceptance, as opposed to optimism, emerged in people who reported a good quality of life and mood. Later interview studies confirmed this pattern in people who do well, and we trialled a small intervention study of a psychotherapy which was designed to improve acceptance. This small trial returned encouraging results, it appeared to improve acceptance and improve quality of life and mood in some people. However, the research design did not allow us to establish if this was a real effect or simply the result of a placebo effect or chance. Therefore we have now applied for a grant to undertake a larger trial of this intervention, to understand if it makes a real difference. We do not know yet if this study will be funded, but we are hopeful.

### Current online study

To help refine this intervention, we have developed a new study which is operates fully online. You can participate now if you wish; please have a look at the following link:

[http://www.muscular-dystrophy.org/research/news/7318\\_online\\_psychology\\_study\\_opens](http://www.muscular-dystrophy.org/research/news/7318_online_psychology_study_opens)

Or search "online psychology study" using the Muscular Dystrophy Campaign's website: [muscular-dystrophy.org](http://www.muscular-dystrophy.org).

This study is more ambitious than the previous studies as it is designed to investigate how important certain aspects of acceptance are in predicting mood and well-being over time. Acceptance consists of a number of different facets; this study will hopefully help us pin-point more clearly which aspects of acceptance are most important for well-being.

**Dr Christopher Graham is a Trainee Clinical Psychologist at the University of Edinburgh and NHS Lothian. Last year he completed a PhD funded by the Muscular Dystrophy Campaign.**

# ACTION ON FSH



As you are aware the FSH-MD Support Group UK have been supporting the Action on FSH fundraising campaign launched by the Muscular Dystrophy Campaign last year.

Upcoming events include:

London Marathon Run (Christopher Finister) Sunday 13 April 2014.

This is my partner and I conned him into taking on this task to help our cause. He has previously run the marathon in 2009 and is hoping to improve on his previous time by 1 hour to add an extra pressure to finishing. He has a Just Giving page: <http://www.justgiving.com/Christopher-Finister> and all donations go straight to the Action on FSH campaign by the magic of the internet.

A table top sale has been organised (Charlotte Daniels) on 3<sup>rd</sup> May 2014 at Colmworth Village Hall (postcode MK44 2JX), between 1pm and 4 pm.

Our members have also been fundraising with Craft Events (Liz Orme), Vintage Sales (Natalie Woodcock). Traceyenne Pilato has also donated postcard prints of some of her original artwork. Our total as FSH-MD Support Group UK at time of typing including Marathon Money is over £3000. A big thank you for everyone who has raised money, or donated so far, your help is greatly appreciated.

You can keep up to date with future Action on FSH Fundraising Events via the Action on FSH page on Facebook, just search Action on FSH using the Facebook Search box. There is also a donate button on the FSH-MD Support Group UK website at [fsh-group.org](http://fsh-group.org). If you wish to have a collecting tin for Action on FSH please contact me by email at [fshgroup@hotmail.com](mailto:fshgroup@hotmail.com).

Liz Williams, Chair FSH-MD Support Group UK



## Muscular Dystrophy Campaign Advocacy

### Ambassadors Network

The Muscular Dystrophy Campaign have launched their new Ambassadors Network. This will provide support for people in their area with Neuromuscular Conditions. As FSH-MD Support Group Chair I have volunteered to be part of the network and following further training will be a fully-fledged Ambassador for the Muscular Dystrophy Campaign but also for the FSH-MD Support Group.

I went to the first Training Session in February this year and it was great to see so many enthusiastic people who had signed up to the network, all eager to help others navigate the complex care and benefits system. The service will be invaluable to those that choose to use it but the first step is asking for the help, which can be the hardest.

#### What is Advocacy?

An advocate will listen to you in times of need and speak on your behalf should you be unable to do so yourself. If you're struggling to access services, benefits or equipment that you're entitled to, working with an advocacy service can help lighten the load in times of great stress and be of great help in creating a better quality of life and greater independence for you. If you can advocate for yourself in times such as these, it can reduce your dependence on outside assistance and can be incredibly empowering.

Advocacy Ambassadors are trained peer advocates who will offer advocacy to their peers and support or mentor others interested in developing their advocacy skills. From forming links to local people with neuromuscular conditions, to liaising with their regional neuromuscular care advisors and other local health and social care professionals. Advocacy Ambassadors will be drawn from various backgrounds and areas around the country, and all need to have personal experience of living with a neuromuscular condition, directly or indirectly.

Ambassadors will be available at your local Muscle Group Meeting, I will be attending the Milton Keynes meetings but other Ambassadors will be attending their local Muscle Group meetings in other regions.

The advocacy service is free of charge so if you are struggling to get the care and support you are entitled to, please contact the Muscular Dystrophy Campaign on 020 7803 4800. Alternatively, you can send an email to

[info@muscular-dystrophy.org](mailto:info@muscular-dystrophy.org).

## Upcoming Events



I am delighted to announce our next Get Together will be held on Saturday 28<sup>th</sup> June 2014 at the Daventry Court Hotel, Daventry, NN11 OSG, from 10 am – 4 pm registration from 9:30 am.

I do hope as many of you as possible can attend, we will have updates on the FSH Registry, information about the Muscular Dystrophy Campaign's Ambassador's network, an update on Action on FSH Campaign. Information on Research Projects into FSH-MD along with stands for products and services and talks from Holidays for the Disabled and Can Do Holidays.

Unfortunately, this year we have to charge for this event. The delegate rate will be **£25.00 per person**, this is a heavily discounted rate as the Muscular Dystrophy have been very kind in negotiating a discounted rate for us this time.

Attached is our booking form so please if you can attend you need to book your place before 28<sup>th</sup> May 2014.

Liz Williams, Chair FSH-MD Support Group UK

[fshgroup@hotmail.com](mailto:fshgroup@hotmail.com)



Search FSH-MD Support Group

## Naidex 2014

**Tuesday 29 April 2014 - Thursday 01 May 2014**

Halls 17 - 19

Car Parking: £10.00 per day

Trade Show

Naidex National is the home of the UK independent living market. It is the largest UK exhibition and conference of its kind, showcasing a comprehensive range of products and services that enable people to live more independently.

### Opening Times

29th April: 10am -5pm

30th April: 10am-5pm

1st May: 10am-4pm

Please visit [naidex.co.uk](http://naidex.co.uk) for more information



**National Conference  
and AGM**

**This year's National Conference and Annual General Meeting (AGM) will be held on Saturday 18 October 2014 at the Hilton Hotel in Coventry**

Tickets are available to [buy online](#). Once you have booked your place, we will be in touch with further information about the day. For more information please contact Lyn Inman at:

[lyni@muscular-dystrophy.org](mailto:lyni@muscular-dystrophy.org)

or Tel: 01132 301 313.

# Neil Withers

## Bali Adventure

Hi all, I'm back from what has been a massive adventure with a few new battle scars and some life lessons learnt.

Some of you will know that last month I decided to jump (or should that be roll... well actually in certain parts of the journey heaved) on to a plane and do a bit of travelling. Two reasons really, one was to test myself and the other was to visit a penpal (for want of a better word), so on the 1st of Jan I booked myself a flight to Bali... well actually it's two flights, one 12hr to Singapore and then a 2hr short hop to Ngurah Rai International Airport (Bali). All this on my own and in a wheelchair, from my front door, back to my front door 2 weeks later.



Loads of research went into the booking but as with anything it's the unpredictable that catches you out. Like being stuck for 2hrs at Heathrow check-in desk while they decided if I could even take my chair on the plane! Or that when my front leg hanger went through the Bali security check they caused the security to pull their guns on me, ask me to raise my hands (doh) and then move towards them slowly (how can I roll my chair with my hands in the air?).

I must say flying with one carrier the whole journey was great and the guys & girls of Singapore Airlines really know their stuff... pity the same can't be said for the ground staff at

Singapore where their disabled help almost caused me to miss my connecting flight in both directions.



Now that's  
a ramp!

Anyway... much of what I had planned for toileting on the plane didn't quite go to plan, sure the Imodium stuffed me up good, a little too good, but the Conven urisheath leaked first try, luckily I wore trousers with zip off legs and without going into too much graphic detail i did a Paula Radcliffe (google Paula Radcliffe Marathon 2005 Pee).

Bali was amazing but totally wrecked my Invacare XLT manual wheelchair, luckily my friend was there to help and also the maid from the Villa I was staying at... Two beautiful ladies helping me - my idea of heaven!!

The Villa over promised and undelivered a bit on the Disabled front... the toilet was too low to transfer to and the bed was way too high (and I'm 6'). Also I came unstuck with the pool hoist because it didn't go low enough into the pool to make getting out easy. Plus the hoist was so slow and going up and down I felt more at danger than less - probably from dying of boredom. This lead me to not being able to find a good place to stand - so for almost two weeks I was off my legs, which has caused a lot of problems since I've been home but now 5 days later they are getting back to normal (useless as that is).

I must say it was all worth it, I've seen some amazing things and appreciate what I have and how I'm treated in the UK as a disabled person a little bit better. I will be going back, call me a glutton for punishment... but there are some things I can't bring home in my duty free.

I guess I'm saying I conquered my fears and short comings with bravery and ingenuity - don't let FSH stop you, for every problem there is a solution, trust your own ability to deal with the world.