NEWSLETTER

Autumn 2013

Issue 22

A MESSAGE FROM THE CHAIR

I would like to take this opportunity to thank Traceyanne Pilato for her commitment and dedication over the past 3 years as Chair of the FSH-MD Support Group UK. I am reassured that Traceyanne is not going far and will remain on the Committee as a Members' Rep and I will certainly be needing her advice and support as I take up the role of Chairperson. I am really excited about my new role. My first event as Chair will be manning the FSH Support Group Desk at the MDC National Conference along with Gill Penny (Secretary) and Liz Orme (Members' Rep) so looking forward to seeing some new faces on October 12th 2013. Details can be found on The MDC website at www.muscular-dystrophy.org. A big thank you to all who contributed to this Newsletter, if anyone wishes to submit an article for the next Spring 2014 Issue could they send submissions to lizfsh@hotmail.co.uk. Many thanks, Liz Williams, Chairperson, FSH-MD Support Group UK.

IN THIS ISSUE

Aniko's Swimming Challenge

Sadie's Fundraising

Laboratory Visit to Centre for Life

An Italian Adventure

Update on FSH Research

Aniko's Swimming Challenge

By Annamaria Kazinczy

London, 03 September 2013

My name is Annamaria Kazinczy. My friends call me Aniko. I am 42 and was diagnosed with FSHD Muscular Dystrophy when I was 24. I had symptoms since I was born, but the more visible ones started when I was about 17.

I used to do acrobatics, ice-skating and swimming regularly when I was a teenager.

I was shocked when I was diagnosed with FSHD, especially as it is a genetic condition and I was worried that I will not be able to have a family. The diagnosis changed my lifestyle as I did sport really intensively (2 hours in 5 times a week). I learnt that this is too much as it can actually speed up the dystrophy.

I started to see a physiotherapist. She was great. We started to read and learn more and more about this condition together. I learnt that I need to ensure that I am not overdoing sport, however keep doing it to keep me mobile. I frequently had pains when I overdone something, so we learnt a technique how to apply a massage on some muscles when stretching that helped to release the pain.

I learnt to live with this condition. Not long after this I met my husband. After a few dates I told him about my condition and all he needed to expect if we continue dating and later living with me. He accepted me with my condition. There was no way to test babies during pregnancy for this condition at that time, so we decided to wait with starting a family. Worrying about passing on the condition I started to focus on my career and did less and less sport.

About 6 years later we were told that a test is available during pregnancy. I started to have a hope again. We had the required blood tests and it was confirmed that my baby can be tested once I will be pregnant. I was in the middle of my studies of my MBA degree and my father was critically ill, so we decided to wait a little bit longer.

My dad encouraged us to have a baby before he passes away. So when I finished my degree we gave a try. I am sure God was with us as I got pregnant quite quickly despite my endometriosis and my baby was tested negative during pregnancy.

I enjoyed the pregnancy and had no particular problems relating to my condition. I joined the lessons for pregnant ladies of a physiotherapist who took my condition into consideration and helped me to get and keep fit during my pregnancy. She also prepared me mentally to give birth. My gynecologist, the midwife and me agreed that we will give a go for a normal delivery but we also considered a caesarean. My baby was finally born with a normal delivery after a relatively short labour. I was really proud of myself.

The breast feeding period was much harder; I felt I am losing my muscles. I had pains as it was hard lifting, carrying and looking after my baby. I knew that I need to do something to keep myself fit. So I started to take aqua fitness lessons. Obviously I could not use the weights but I learned few exercises that helped me. However I found that aqua fitness was too hard so I started to do swimming instead.

Not much later my husband got a job in Norway so we spent a year there. Due to the too much snow and my difficulty to cope with it I had to give up my swimming. As I could not cope with the snow we had to leave Norway. My husband got a job in London so we moved to Eltham. Shortly after we moved I found out about the Lions Swimming Club

The Lion Swimming Club is a charitable organisation based in The Eltham Centre, Eltham, London and is for anyone with a permanent physical, mental or sensory disability. Age does not matter. Members can enjoy the sessions to swim, exercise or just have fun. The club is affiliated to NASCH (The National Association of Swimming Clubs for the Handicapped). I have been using this swimming club for 6 years now.

Despite the swimming I was still missing mentally the feeling to do group exercises. I searched for the local groups having lessons after working hours (as I work full time). After a long search I found a lady who runs Pilates lessons in a nearby church hall. I contacted her and updated her about my condition. We agreed that I will try one of her lessons. I loved to be in a group again. With my acrobatics background we worked out how we can adapt the exercises for me. I do my Pilates once a week since then and Mandy Brown my trainer supports and motivates me since then.

I found that attending these swimming and Pilates lessons helped me to control all my muscle pains. My muscles got weaker and weaker over the past years obviously but I can manage all the stiffness and pains. Any back and neck aches goes after few length of backstrokes. And any back and neck related headaches go after my Pilates lessons/exercises. I also found that mentally I feel much better.

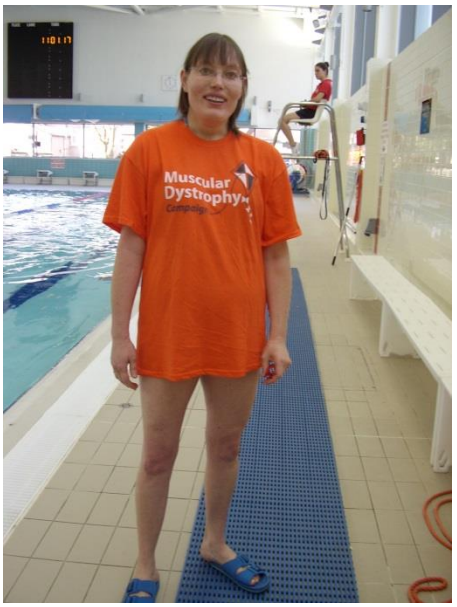
My legs started to be impacted more and more last year and I felt quite low so I decided that I need to do something that keeps me motivated. I also felt that I would like to give back something for my swimming club and support any research to find a treatment for FSHD so I decided that I am doing a swimming challenge.

I discussed my idea with Dr Norwood and Jo Reffin physiotherapist at Kings College hospital during my yearly follow up and we agreed that I should not overdo so I am better of doing my challenge in smaller bits.

Following that I started my swimming challenge in February this year and committed myself to swim 10k in blocks of 500m within six months.

I am happy to report that I completed 12k by the 21st July. I managed to raise GBP 1072. As the company I work for kindly offered to double this up both the MD Organisation and the Lions Swimming Club will receive this amount shortly.

I received so much help, support and kind words during my challenge from the people at the Lions Club, at my Pilates lessons, at work and from friends as well. It was truly a rewarding experience. I hope all the money collected will help to move on the research on FSHD; and allows the Lions Club to offer the possibility of swimming for other disabled people and children.



Aniko

Remember if you are seeking advice from other FSH members the easiest and quickest way to do this is to find us on Facebook. Just search FSH-MD Support Group UK and ask to be added.

You will usually get answers to your questions on the same day or even within minutes. It's a great way of getting feedback or even helping someone else with your experiences.

FSH-MD Support Group UK

facebook

Sadie's Fundraising

By Michelle Goldthrite

Sadie had wanted to do a charity walk for a long time as she was inspired by a family friend Danielle Woolcock who does lots of charity work. I, their Mother have FSHD as did my Mother. Sadie was diagnosed with FSHD in September 2011; Abigail has now been tested and does have it too. (This is not information we have shared with many people) We contacted the MD Campaign and we received some guidance from Andy Putner who told us about the South West Wales branch where we found Ray Thomas who has been a massive support and the group were a great help. Initially the girls were going to walk alone but soon friends and family were asking to join them and the sponsored walk got bigger as the day approached. The head-teacher Mrs Claire Dineen from the girls' school, St. Josephs Primary in Neath very kindly donated chocolate eggs for the children who took part. On 30th March Easter weekend it was freezing but still around 70 people arrived to take part including several members of staff from the girls' school. One teacher came dressed as a sheep. We also had Minnie and Mickey Mouse, Cat in the hat and a Banana come on the walk.

The walk took place along Aberafan beach, The Children carried, buckets, banners and wore the orange t-shirts and caps, everyone had a lovely time and £106 was collected along the walk in buckets. Sadie and Abbie are both very special. Only 3 weeks before the walk Sadie was in a foot splint as she had broken her foot. The photos supplied were taken by Mike Davies a friend of Joel's from his metal detecting group. Joel Is American and his family contributed to the fundraising by sending over money. Bethan Jenkins AM also attended the walk. On 12th April a Disco was arranged by the girls which took place in the Naval club which was given to us free of charge but we did have to pay £50 for the DJ. The night raised £180, The Rogers/Mason family sold sweets and decorating the hall was done by Jane Corse and Ray Thomas. The walk raised almost £1,500 and the girls are now planning their next fundraising venture.

Laboratory visit to Centre for Life, Newcastle.

The visit was organised by the Institute of Genetic Medicine at Newcastle University and the Muscular Dystrophy Campaign on Saturday 1st June

By Cath Mitchell

My Daughter and I attended this event; I'm always interested to know what's happening in the way of research, especially at the Centre for life as it's where I have my check-ups and yearly MOT.

I certainly wasn't disappointed as the talks were excellent. Sadly Katie Bushby wasn't able to be there due to illness.

The day began with Marita Pohlschmidt (MDC) explaining the work done by MDC and what was currently going on by way of research at 'Centre for life' and at various other places. Two students gave excellent presentations: Callum Kirk from Newcastle Uni regarding Limb Girdle (funded by the Muscular Dystrophy Campaign) and Louise Moyle from Kings College on FSH.

I had to admit I didn't know much about Limb Girdle MD, so it was interesting to learn how that people who have this form of MD have similar problems with their shoulders as people with FSH do.

I could never imagine how actual cures for FSH MD and Limb Girdle MD would come about, but listening to people who are making it their life's work and for it to be happening in the City where I live was fascinating.

Louise Moyle's presentation on FSH was excellent and explained so well in 'laypersons terms' , I found out things about on-going research that have swayed my opinion into thinking

a cure will be possible one day! Her study is on-going and several people (myself included) expressed an interest in eventually being able to read about all her findings.

My Daughter and I had an interesting chat with a lady from the bio bank about the legal and ethical side of samples; research etc. There's divided opinion about this in society, but our questions were answered very thoroughly.

Libby Wood (Newcastle University) gave a talk on the recently launched FSH Patient registry. She spoke of how important this is with regards to research and strongly recommended signing up to it. The uptake so far is very promising.

Katie Mitchell then spoke on action on Fundraising. She reminded us of the ways in which the Muscular Dystrophy Campaign funds so many issues and how important it is to keep the fundraising alive.

There was a tour of the Labs at Centre for life to view at first hand the research currently going on this took place in small groups at allocated times. The researchers were there to answer any questions.

The members of Newcastle's clinical team were available to explain their services in more detail and answer questions. This was a good opportunity for anyone recently diagnosed or anyone who cares for a person with either Limb Girdle or FSH MD (they certainly had a queue!)

The event was to end with closing remarks and questions to John Bourke (Newcastle Uni.) and Marita Pohlschmidt.

I had to leave before the last session as my Daughter had to complete her assignment towards her podiatry degree. It was an excellent event; I enjoyed the whole day and came away quite a lot wiser.

The buffet was excellent and although I didn't have any of the cake (honest!) I was informed by others that it was delicious! So many thanks for a great day go to The Centre for life and the Muscular Dystrophy Campaign.

Action on FSH Appeal Update.

By Liz Williams

Action on FSH Appeal.

The Muscular Dystrophy Campaign's Action on FSH Appeal has been established to support two critical research projects that will further our understanding of FSH. The MDC are seeking donations to enable them to complete this vital work in the year ahead.

Project one - Understanding the molecular causes of FSH

Project two - investigating the role of muscle stem cells in FSH

FSHD patient registry

The Muscular Dystrophy Campaign has been working with TREAT-NMD to set up a FSHD patient registry. The establishment of patient registries is vital as they focus on the information that is needed to find patients eligible for clinical trials, and help clinicians develop care standards. Patients can link to the research community and have the opportunity to access information directly relevant for their condition.

For more information about the FSHD patient registry, including how many people have registered so far, and to join yourself, visit fshd-registry.org/uk.

A curator based at the Institute of Genetic Medicine in Newcastle has been in post since the beginning of 2012 to establish the registry and £25,000 is now needed to take this project forward: to implement, manage and maintain the UK patient registry for FSHD.

The team in Newcastle has unique expertise in setting up patient registries. Professor Hanns Lochmuller, one of the Centre's directors, has developed national and international registries within the scope of TREAT_NMD and is a leading expert in this area.

There are already registries for FSHD in the US, Netherlands and Italy and other countries. A national registry is urgently required in the UK so that patients will not be left out of studies that could provide early access to treatment.

Supporting the Action on FSH Appeal

The FSH-MD Support Group UK supports the Action on FSH Appeal and we are in the process of finalising some exciting fundraising opportunities. A Marathon place has been made available by the MDC to raise funds for the Action on FSH Appeal and we hope to announce a willing volunteer shortly. If you know someone who would love to run the London Marathon and raise funds for the Action on FSH Appeal please contact Liz Williams on lizfsh@hotmail.co.uk.

The MDC needs £20,000 to fund the two vital research projects in their final year, as well as £25,000 to fund the FSH registry. Please help us reach the £45,000 target by making a donation.

www.muscular-dystrophy.org/fshappeal

The current amount raised for the Action for FSH Appeal is £18,000.

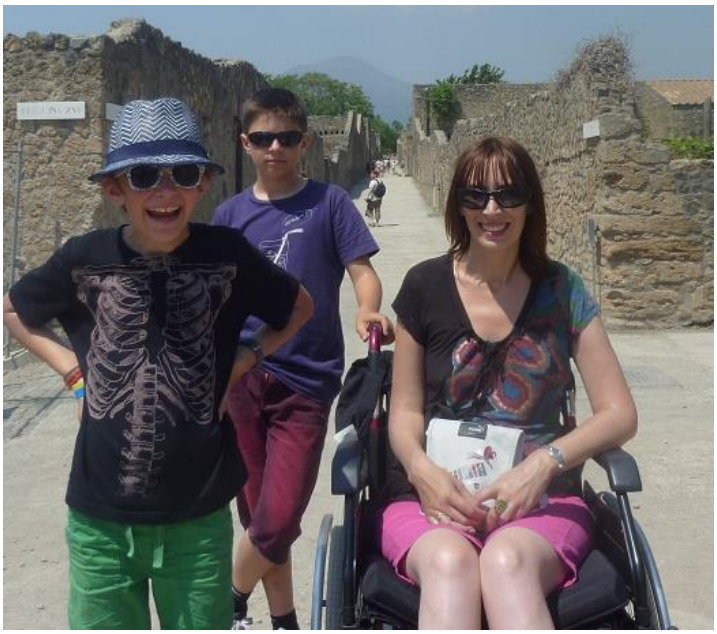
The FSH-MD Support Group UK is currently working on ways to fundraise so please keep visiting our website at

fsh-group.org

There are also updates on our Facebook page. We now have a donate button on our website which goes directly the Action on FSH Appeal. The target the Group aim to raise is £10,000.

An Italian Adventure

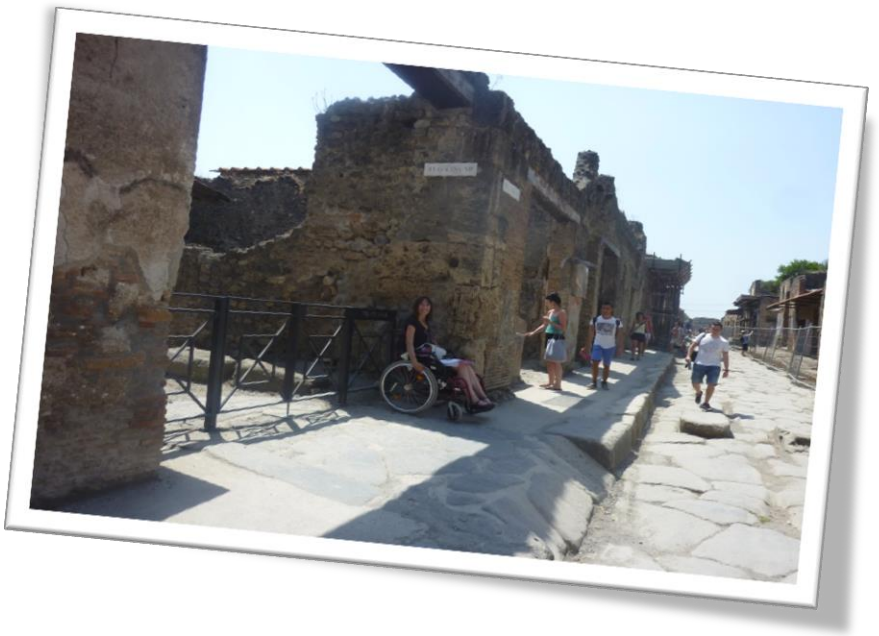
By Liz Williams



I recently took the plunge for the second time as a part time wheelchair user, attempting travelling to Italy for a holiday. We are a family of 4, two adults (one wheelie, me) and two children Ralph and Ronnie aged 12 and 8. The obstacles were daunting, air travel, accessible room etc., but after many years of staying in various holiday cottages in the UK and enduring the recent summer weather we had yearned for guaranteed sunshine. Our list of requirements was a beach (accessible) a walk in/wheel in shower and a hotel with a pool for the kids. I was not particularly thinking of me using the pool or swimming in the sea but at the very least being able to be on the beach. I contacted a disabled holiday company and went from there. We started with Sorrento as ideal destination (hilly I know) and the holiday agent came up with a suitable hotel with a guaranteed walk in shower and ramped access to the hotel and lift to the rooms. The pool wasn't accessible for me but as a first time attempt at ticking the boxes this was pretty good, the kids could have a swim while we availed ourselves of the bar etc. So to the airport. We flew with an Italian airline and arrived at Heathrow in good time to check in. We used Heathrow's short stay car park which is right outside the terminal and although expensive, just removed any worries of getting on a bus at the other car parks at Heathrow which are further away. We arrived in the pre-booked car park 100 yards from the door for the Terminal, we made our way inside. We saw the queue for the desk for our airline and decided to make ourselves known to the check in staff by sneaking down the fast track queue, this was a good move as they directed us to check in at Business Class desk, no queue and then we booked the assistance for boarding the aircraft. We made our way to the gate (in wheelchair). Security was the next experience, you remain in the chair while the security staff give you a frisk, this was a thorough frisk, you are touched almost everywhere and as my shoes were wedged I had to remove those too. Trouble free we went on to the gate. The staff took us to the aircraft prior to boarding the other passengers. Once at the plane, I was transferred to the airport wheelchair and we folded our chair and held on to our cushion. They loaded the chair into the hold so that they could get it out when we arrived at the destination. We kept the cushion as there is no way of securing it to the chair so it would be very easy for them to lose this in transit so we just held on to it and put it in the hand luggage compartment. The plane wheelchairs are narrow to navigate the aisle to your seat so if your limbs are likely to fall to the side they strap you in ala Hannibal Lecter. At this point we congratulated ourselves, we were going on holiday, just like an everyday family, yay. Once in Italy the plane disembarked on the runway so the other passengers got off to use the bus and we waited for assistance to arrive. The staff remain on the plane so you are not left on your own and then a vehicle with a lift is raised to take you from the plane. I was transferred to the Hannibal wheelchair, then once in the lift I was reunited with my wheelchair which had miraculously appeared. We boarded a further flight from Milan to Naples in much the same fashion, only the Italian airport staff escorted us to make sure we were where we were supposed to be for our connecting flight.

Once at Naples we were assisted off the plane again and the staff at the Italian airport helped all the way to the luggage collection point so I really was looked after and I am sure they would have seen me to the car if necessary.. We went to the conveyer belts……..no luggage appears; we filled out a form and were told it would be sent on to the hotel. As you can imagine, we were a bit unsure as to whether we would see our luggage again but once we found our taxi to Sorrento we forgot all about the fact we had no clothes to wear and enjoyed the fantastic views of the coast and Vesuvius looking ominous in the background. The heat was phenomenal, a nice 36 degrees so an air-conditioned taxi was a relief. We arrive at the Hotel we are to spend the next 7 days. Ramped access as promised and when we got to our rooms it couldn't have been better the boys were next door and there was an internal linking door which they unlocked so the kids didn't have to leave their room to get to us. The bathroom was as promised, walk in shower and so just what we had asked for. The original plan was, arrive, get the kids in the pool and mum and dad sit by the bar, however as we had no clothes we decided we had to get some food and then attempted some emergency shopping. Now Sorrento is hilly so if you are a wheelie you will need a fit carer to navigate the cobbled streets, but once you are at the main square it is fairly level and of an evening they close the main streets to traffic so as night falls the center is much easier to navigate in a chair. We found food at one of the many restaurants and cafes, sitting outside with easy access for the chair and enjoyed the fact we had arrived on holiday. We returned to the Hotel and no luggage but we had managed to get toothbrushes etc. so we could clean ourselves up. We decided it would be better to go to Pompeii the next day as a contingency for our luggage not being there in the morning. The next morning, still no luggage, we set out to Pompeii.

There is a station at Sorrento which has a lift up to the platform and then a ramp but, there is a big step up onto the train. We asked the ticket booth staff if it was accessible at Pompeii to which the gentleman made a gesture which my partner took to mean flat, but on arrival at Pompeii we realised this was lost in translation and actually



Not very accessible Pompeii

meant no access whatsoever. So after an eventful journey with 'entertainment' including a rendition of la bamba, we got off the train at Pompeii to find the only way off the platform to the exit was via a flight of stairs down and a flight of stairs up to the other platform. Mild panic set in. Fortunately, a very, very kind English man saw our predicament and even though we were trying to get the attention of the guard on the other platform my partner and this willing volunteer carried my chair down and up the stairs. I felt very much like the King of Siam and was rather embarrassed at putting myself in that position but we could see that this wouldn't happen on the return journey as the other platform was accessible. In Italy it is quite common for there to be a ramp to nowhere so this was knowledge gained, and over the course of the holiday Chris did a few dummy runs to see where they took us before pushing me up them. We got to the entrance to Pompeii, massive queue and Chris went to the front to enquire about access and they directed us to another entrance, about a mile away. However, we trogged along to this entrance and there was no queue and because Pompeii is clearly not wheelchair friendly they let us all in free of charge. There was a more accessible path to the main street of Pompeii and we did see quite a bit. It was not easy and really hard on my wheelchair pusher and I would definitely recommend not visiting for the limited access on one of the hottest August days ever, but you can get a feel for the vastness of the place in the shadow of Vesuvius. We returned to the station and although there is a high step on to the train we managed to navigate this and returned to our hotel. Yes, our luggage had returned and really lifted our spirits; the kids could finally go for a swim.

The next day we went to the beach. We got to the cliff top where there is a lift, a proper lift, down to the beach. You have to pay and this is discounted for wheelies with carer and you can pay for a return journey. There are queues at peak times but this is a means to an end. We arrived at one of the paid private beaches. Chris has a look and there was a ramp down to the beach and the attendants gave us a brief tour of the facilities, which included bar/café, disabled loo with a shower, and ramp to the beach and then announced they had a wheelchair to get me in to the sea. WOW. I had no swimming costume but that was my next task. We had a lovely time I was on the beach with the kids, with coffees and a loo so perfect. I procured a swimming costume for a princely sum but I didn't even have to try it on , the lady in the shop had a very good eye for sizes so picked out exactly what I needed, one that tied at the back of my neck. The next day I took the plunge. The attendant at the beach asked me "swimming costume?" I said "yes" he said "brilliant". When I was ready we called the lifeguard, he brought the floaty wheelchair and I got on and he pushed me while Chris helped me into the sea for the first time in 20 years. Well what a feeling! I think I was laughing for the first half an hour, much to the amusement of the rest of the people on the beach, I laughed so much I cried, at first my legs wouldn't go down so I was a bit like a turtle on my back, and the reflexes I don't have really were apparent. However, once my brain got on board to concentrate on how to get my legs to sink rather than float I got a bit more confident so Chris could let go of me. I could only really tread water or swim on my back, I tried to float forwards, on my stomach, but I think my shoulders inhibited me and I just felt like I would drown, whether I needed to relax again I don't know but with waves I didn't feel confident to test this theory in the sea. Very quickly my brain and body got together which enabled me to turn myself, and even got from floating on my back to standing, I could kick my legs, without any great strain and it really was like being free, an experience I will never forget and one I really want to repeat. We spent 7 days in sunshine, in and out of the sea, with great food, a bespoke family holiday.

I think I just wanted to say that although lots of things were denied me due to access issues, the things that I could do totally outweighed the negatives and there were a lot of negatives because everything outside my own home is difficult, but I have decided with the support of my family that this will not stop me going where I want to go. The experience has given me the shove to get to a swimming pool here before the year is out too.

Update on FSH Research at King's College London

By Trevor Thomas

This was my third annual visit to King's College London to learn about progress there with research on FSHD. Again I was representing the Lay Panel of the Muscular Dystrophy Campaign. As previously, I met Professor Peter Zammit, whose group specialises in the study of muscle stem cells, and Louise Moyle, whose PhD research project is part-funded by the MDC. This time they were joined by Christopher Banerji, a mathematical biologist based at University College London, who has been collaborating with Prof Zammit for about a year.

Louise has one year to go in her PhD project. An article by her appeared recently in Target Research (Issue 2, 2013) where she explains how in FSHD a protein called DUX4 is produced in the muscles and can interfere with normal maintenance and repair processes. The maintenance and repair of skeletal muscle rely on the "satellite" cells, which are stem cells living inside muscles. Satellite cells lie dormant until triggered by muscle damage, due to injury or normal wear and tear, when they fuse together into new muscle cells. At the same time they also multiply to keep their numbers up to repair future damage.

A dedicated team: Peter Zammit, Louise Moyle and Chris Banerji.

The internal workings of muscle cells are complex. Everything happens in accordance with instructions held in the DNA, in the nucleus of the cells. The DUX4 protein can interfere with other parts of the DNA, turning genes on or off, so causing wrong instructions to be given. Louise has been trying to reveal the chains of events, or "pathways" between the genes, which lead from the unwanted production of DUX4 to the poor fusion of stem cells into new muscle. The aim is to find ways of blocking the effects of DUX4, which could be developed into potential treatments

Since my last visit Louise has continued to investigate of the role of a gene called RET. Last year she had already confirmed that DUX4 causes the RET gene to become overactive and had shown in a separate experiment that overactive RET prevents the satellite cells from fusing together efficiently, i.e. having a similar effect as DUX4.

Since then she has studied the role of RET in healthy satellite cells, finding that when DUX4 is absent RET has a normal role in regulating the rate of fusion. She has now successfully performed a "rescue" experiment in which the harmful effects of DUX4 were reduced by manipulating the RET pathway, resulting in more muscle being made. This raises hope that an existing drug, which targets the RET pathway, might achieve a similar result. Louise has already started experiments with the drug in cells in the laboratory.

Louise is now also studying how DUX4 causes muscle cells to die, having taken over this work from Janet Davies who left KCL last year. For practicality, satellite cells are used in this work too, because DUX4 kills them as well as full-grown muscle cells and researchers believe the processes to be similar. During my previous visit, I learned that DUX4 kills muscle cells by triggering a process which normally removes unwanted cells. Janet had already been studying another gene and had concluded that it sits on a

pathway linking DUX4 to this process. Louise has now started "rescue" experiments to try and block this pathway. She has also been investigating the possible role of RET in this process and is planning further rescue experiments if a link is proved.

The arrival of Chris Banerji on the FSHD research scene has opened another line of attack in unravelling the effects of DUX4 inside muscle cells. Chris is tackling a big problem with this research: where to start looking in the mass of information now available on gene activity. In the case of Louise's research, she started with the results of a previous study at KCL, in which the effect of DUX4 on a large number of genes had been measured, showing which genes become more active and which less active. It was not known which of these genes were actually connected with the symptoms of FSHD. RET was chosen for detailed study because it was known to interact with the surroundings of a cell and with many other genes inside the cell, and was therefore likely to play an important role. The question remained which other genes were also worth studying in such depth.

Chris started his career as a mathematician in the banking sector. When he discovered that he had FSHD himself, he decided that his mathematical expertise was being applied in the wrong direction, so he went back to university and is now a mathematical biologist. Chris has developed a powerful new mathematical method for analysing large numbers of sets of data, to identify cross-connections between things that are changing. Using his computer programme he has analysed all the publicly available data on gene activity changes from biopsies of people with FSHD, with the aim of identifying the interactions between different genes. The result is a map of the signalling network between genes which exists in muscle affected by FSHD and not in healthy muscle. Further mathematical analysis has revealed that only a relatively small number of genes are involved in a high proportion of the disturbed signalling in FSHD. One gene has been identified as a central hub in the network and is now being studied in the laboratory by Louise, who has already validated that DUX4 has a big effect on its activity. Now she is studying how the activity of this gene affects satellite cells, with the aim of evaluating it as a possible drug target for FSHD. In previous meetings with Peter Zammit and his team I was struck by various things: by their enthusiasm and commitment; by the complexity of the problems being tackled and by the amount of careful work needed for each step forward. After this visit I was reassured that the team is applying a very powerful combination of techniques, bringing together state-of-the-art mathematical analysis and exploration of FSHD biology. While practical treatments for FSHD are certainly not just round the corner, progress towards that goal seems to have gathered pace.

Diary Dates:

Visit www.muscular-dystrophy.org and search 2013 National Conference for booking information.