



# NEWSLETTER

## Autumn 2014

Issue 24

### A MESSAGE FROM THE CHAIR

Liz Williams, Chair FSH-MD Support Group



Dear Members. It's been a very busy year for me and our family with various things going on and it seems our members have been very busy too with various endeavours. I hope you find this edition of the Support Group Newsletter informative and useful.

If you have an article you wish to place in the next edition please contact me at [fshgroup@hotmail.com](mailto:fshgroup@hotmail.com)

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## ACTION ON FSH



### Christopher Finister London Marathon Run

In our previous newsletter my partner, Chris was about to run the London Marathon to raise funds for Action on FSH Campaign, an appeal launched by the Muscular Dystrophy Campaign in 2013. The Action on FSH appeal raises funds for the FSH Registry and for Research into FSH-MD.

Chris ran the London Marathon in a time of 3 hours 26 minutes, beating his previous time by just over an

hour. With the help and support of both members of the FSH-MD Support Group UK, friends and family, some of whom we haven't seen in 20 years and members of the Support Group we have never met, Chris raised £2,407,44. We were overwhelmed by the support given so I am taking this opportunity to thank everyone who donated online and offline for the Action on FSH appeal.





## Birthday in Paris

By Traceyanne Pilato

I had always hoped to spend my 50<sup>th</sup> birthday in Paris but unfortunately with one thing and another it just did not happen. So this year I was determined that I would get there and I would be able to tick off something that had been high on my "bucket List" for so long which was to have dinner and watch a show at the Moulin Rouge!

I flew to Paris with EasyJet and I certainly had no complaint regarding the service I received. Being a wheelchair user I was able to board the plane before the other passengers that way I didn't feel like all eyes were on me while trying to scramble across to my seat. The flight was 1 hour 20 and just flew by, excuse the pun!

On arrival in Paris I caught a train from the airport to Gare du Nord, which is the main station in the centre of Paris. The train was accessible. I was able to wheel straight on without the use of a ramp, which was just as well as there was a serious lack of station staff. Gare du Nord was horrendous. It is huge and the lifts were few and far between. It took us a good while just to find our way out of the station.

As we were in no hurry we decided to walk to our hotel using a map on our iPhone, not the best idea, but it did mean we got to see a lot of places we wouldn't have if we'd just jumped into a taxi.

The hotel, Novotel Eiffel Tower was very modern. My room was perfect. The only complaint was that, although there was a ramp into the hotel, the reception was on the first floor with no lift up to it. Wheelchair users were escorted to the rear of the

hotel where there was a staff/service lift. For a relatively new hotel I thought this was rather poor. However it didn't mar my stay.

Once settled my first stop was a leisurely stroll or roll as the case may be along the banks of the Seine to the Eiffel Tower. Pavements and paths were in good condition which was nice after my recent encounter with the pavements and cobbles of Rome! As a wheelchair user you are able to jump the queue at the tower. Although only allowed up to the second platform you are still rewarded with a magnificent view across the city. I was totally in awe of just how huge the tower was.

The following day we took a bus tour around the city, finally jumping off at Notre Dame. There was a step into Notre Dame so we were advised to enter by the exit which had a far smaller step, probably 2 inch, easily manageable. Inside there were a few steps at the far end of the cathedral but I didn't feel like I was missing out in any way not being able to go down them. We spent the rest of the day just wandering and soaking up the atmosphere of Paris's café life while drinking wine and eating escargot. Accessible toilets were certainly an issue. We didn't find one café that had a wheelchair accessible toilet; maybe we were just unlucky. There are the occasional outside, public loos dotted around the city all of which had very long queues and were used by abled and disabled alike.

Our wandering took us to the **Arc de Triomphe** and the Louvre although we were far too pushed for time to go inside. It just means that I will have to revisit someday.

Our final evening was spent at the Moulin Rouge. It is not accessible! I use a manual wheelchair and was lifted down a flight of stairs by 4 of the waiters. I didn't mind as this was something I was determined to do but unfortunately it does mean that if you are in an electric wheelchair you won't be able to visit and once inside there is no access to a toilet. It was a wonderful experience and for me very much worth the inconvenience.

I did fall in love with Paris and can't wait to return as there's just so much I missed.

By Traceyanne Pilato

**If you have been on your travels and feel you would be able to share your holiday experiences, please email Liz Williams at:**

**fshgroup@hotmail.com**

# Accessible Motorhome Holiday

by Liz Williams



This year, after a great day at the Tour de France Yorkshire, we pondered the possibility of getting to the Tour de France in France. We deliberated whether this was at all feasible, with the complexities of my mobility issues being the paramount concern. After a spot of googling, we happened upon a site offering a disabled access motorhome for hire. After initial enquiries and having not booked a holiday this year we decided to plump for a few days away in Tenby. We reserved the motorhome and booked a pitch on the Haven site. I had 3 weeks to get used to the idea of camping, well sort of!

The motorhome company could deliver this to your door but we opted to pick it up from their premises in Tamworth. On arrival we were given a whistle stop briefing of the gadgets and gizmos the motorhome had to offer, it boasted a wheelchair lift, wet room, loo and two double beds. Along with the usual table, sink, oven and microwave, there was a television, fridge, a small wardrobe space and lots of cupboards above the bed in the main bed area. Our children's double bed was above the driver and passenger seats in a clever compartment which we referred to as the bat cave for the rest of the holiday.

The driver and passenger seats both swiveled to face the interior of the motorhome, these were quite heavy to move so if I needed to turn around, I had to commandeer help. The wheelchair lift was very clever. Just looking at the vehicle from the side, you would not imagine it had a lift. This appeared by a mechanism that stores this under the door and step. It extends out and is a large platform operated by a handset. This gets a big thumbs up from me as it was discreet, and there was also a manual override in the event of it breaking down (which didn't happen by the way).

The wet room and toilet were hidden behind bi-folding doors which open out into a larger space. The main floor area of the motorhome had a slight recessed area with plughole sized drains to take the water from the shower away. It was certainly making the most of the available space, but, I would only really want to use this if there was no other option, and most sites have disabled facilities these days. I did use this though and managed to get a shower and wash my hair which is all you can ask really isn't it? The site we stayed on had great disabled facilities; I have to say that the wet room the camp site provided was one of the best I have ever seen outside residential properties. There was a wet room with adjustable shower chair and disabled loo. My only gripe would be this was also used as a baby changing area and though it was spotlessly clean, the nappy bin did hum on one visit. That aside, it has to be praised.

My review of the accessible motorhome would be this. The innovation of the lift, giving access is absolutely fantastic, and I would certainly recommend having a bash if there is somewhere you really want to go. If you are in a wheelchair full time maneuvering in living spaces is going to be problematic. I entered the accommodation via the lift but I would get out of the wheelchair once I was inside and fold the chair up. I think ultimately our Tour de France dream may be quashed in a motorhome. It is cramped and 5 days was long enough for me, but had we had better weather, my answer may have been different. However, if I wanted to go to, for example, Glastonbury, or another festival, I would certainly not hesitate to book this as a more luxurious option to camping as a tent would be a no no. It is very important for me to add that this motorhome was a demonstration, made available to hire primarily for people who are perhaps interested in buying their own adapted motorhome. The company we hired from stated that they had never adapted two vehicles in the same way, they are bespoke and as a demonstration hire vehicle, this one was always going to be a compromise. With that in mind, as it was not adapted for me, it came pretty close to as good as it gets.

## Tips:

**Consider cost of diesel when booking a motorhome holiday.**

**Don't pack suitcases as there is nowhere to store them once emptied, especially if you have to accommodate a wheelchair.**

**I would recommend booking a short hire in the first instance to see if it is viable and I am sure the company I used would be very happy for you to have a tour of one before booking anything.**

**If anyone wishes to hear more about the company used and access in Tenby in general, please do not hesitate to contact me at [fshgroup@hotmail.com](mailto:fshgroup@hotmail.com) , I would be happy to help with any queries.**

Liz Williams

## TRAVELS WITH A ROLLATOR – TALES FROM A ROOKIE USER

This year I finally accepted that I needed some support with walking beyond a walking stick in one hand and my husband in the other. So, in June I took delivery of a rollator. I wanted to be more independent so I'd searched around and bought one of the lightest on the market at 7.5kg. I also wanted one that folded as small as possible, both sideways and down, so we could fit it in the car going off on holiday to France. There aren't many that fold down compactly so my choice was very limited. However, it duly arrived and we unpacked it and took it out for a couple of trial runs from home, before setting off for France. First disappointment was that, whilst the rollator did fold up to a small[ish] size for fitting in the car, having to lay it all out on a solid surface (OK in the house, not so good on the ground outside in the rain) whilst you fitted and screwed in four bolts each time was not conducive to a quick getaway. As we were touring we really needed something that was easy to set up and fold down and that would fit in the car with the rest of the luggage. In the end we just half folded it and put it on the back seat, holding it securely with the seat belt. Second disappointment was that, light as it was, I still couldn't lift it in and out of the car on my own, so my dreams of being fully independent weren't realised - I still needed help. However, it gave me lots more freedom to move around, and certainly trying it out in Milton Keynes gave me a wholly different and more enjoyable walking experience. Not only could I move much more easily, but I could sit down and rest when tired or in queues for the till when shopping.

So how did I fare on holiday in Northern France? I found that, like most other countries, it's not that great for people with limited mobility. I only saw one other rollator user throughout the three weeks we were there and that may well be related to some of the problems I experienced. Many of the town centres have cobbles or granite setts, and I quickly found that rollators and cobbles/setts don't mix. It needed a fair bit of force to push the rollator over them, and when it suddenly hit a higher cobble or went down a rut, the front wheels would turn, the rollator would stop and I would nearly, though fortunately never did, go over the 'handlebars'. Turning front wheels are great when you want to turn, but not when you are trying to navigate uneven ground. French kerbs seemed to have few slopes off, not even at pedestrian crossings; they were often six inches plus, and the gutter was often slightly lower than the road itself. So getting the front wheels of the rollator off the pavement was bad enough, but then trying to lift them from the gutter whilst trying to get the back wheels off the pavement was nigh impossible on my own. And if anyone can keep their balance whilst pushing down on either of the small levers near the back wheels that are supposed to help you lift the front, could they give me lessons please. French pavements also have gullies across them to enable the rainwater down-pipes on the buildings to drain into the road, meaning that every few metres the front wheels had to be lifted over a dip. All this meant that I really relied on my husband to help me navigate the difficult parts.

However, problems were not always physical ones, as exemplified by our experience in the National Archaeological Museum in St Germain in Paris. It's located in a fine stone castle with the exhibits on the first and second floors. At reception we were told that they would get the key-holder to take me up in the lift. Sounded a little strange, and so it turned out to be. A short wait for a member of staff, and two unlocked doors later we were in what appeared to be a service lift. We stepped out, and the lift closed behind us. After an hour or so exploring the exhibits we went back to the lift and asked the nearby attendant how we summoned it. He would have to get the keyholder to bring the lift

up, so off he went to phone her. He returned with bad news – she had gone to lunch and so we would have to wait until she came back. And when would that be? In about an hour and a quarter. The French enjoy their lunches! And couldn't another member of staff manage the lift? No. And neither he nor the other attendant on the floor could do it because the regulations state that there have to be two attendants on the floor at all times. So, what to do? The only other alternative was a very long, hard, wide stone castle staircase, with very steep steps and no hand rail. Not something I was willing to try, and anyway by now, I was into making a point. Whoever goes up must come down, and you can't just have one person responsible without making alternative arrangements for when they are not there. The only seats on the floor were in the video room at the other end of the building to the lift so we walked back and waited there for well over half an hour. The other attendant came to check we were OK and to apologise. We waited for another quarter of an hour. And then the attendant came back to announce that a dispensation had been granted and she had been authorised to use the lift. Did we want to go to the second floor? Fortunately, Roger had already been up to the second floor via the stairs where the exhibits weren't ones that interested me so 'no', we would go back to the ground floor – I'm not sure either of us would have wanted to risk going anywhere else in the lift. In hindsight, an amusing interlude, but it was somewhat irritating at the time – especially as it was the regulations that were the problem, not the actual facilities. Fortunately this experience was a one-off. Most of the other museums that we visited in our travels across the country were fine and staff helpful and supportive.

Aside from that, we had a really enjoyable holiday touring northern France, exploring many small and large French towns and villages, watching a couple of stages of the Tour de France, wandering round lakes and churches in Champagne, war memorials in the Ardennes, mountain villages in the Vosges. And overall, what did I think of my travels with a rollator? In general it was liberating and increased my confidence. I certainly walked further than I would have been able to do without it, although it wasn't all plain sailing and I did need a fair bit of help at times. And there were other advantages too. My right shoulder didn't ache from the pressure of using the walking stick. I could take a handbag with me because it sat in the basket of the rollator and so didn't dig into my shoulder. I didn't need to rely on my husband to carry things - I could put guide books and gifts in the basket, so it was far more convenient and less tiring. And when I did get tired I had a very handy seat to sit on.

So definitely, the decision to use a rollator was the right one, though I think I was rather naïve in my expectations. My holiday experience has also taught me to appreciate the benefits of living in a modern city like Milton Keynes. Now for my next holiday all I want is an upgrade – a version 2. So, if you know of a very light rollator - under 5kg please - that I can manage on my own, that I can lift in and out of the car, that folds down as quickly as a child's buggy and without lots of screws, that has some way of enabling me to get on and off pavements safely, that has front wheels that allow me to navigate in tight spaces but can also lock for going straight ahead on rough ground when needed, possibly with knobby tyres – ie a 'cross country' rollator, do please get in touch!

Gill Penny.

## THE MDC MUSCLE GROUP MEETING – MILTON KEYNES, 24 October 2014

I've been attending the MDC Muscle Group meetings in Milton Keynes now for about 4 years and each time I come away having learned something new. The latest was on Friday 24 October at the Holiday Inn. Organised and chaired by Peter Sutton from MDC we had a number of items on the agenda. Usually around 12-20 people attend but this time we had around 24 plus MDC staff and speakers. Just over half of the attendees had a neuro-muscular disorder [NMD] and the others were family members, carers or friends. There was a core of 'regulars' plus some who attend occasionally and others who were there for the first time. A range of disorders were represented including four with FSH-MD. Many of the issues we face are common to all NMDs and it's interesting to hear of others' experiences.

This time we had three speakers from the MDC. Peter Sutton gave us an update on the 'Bridging the Gap', a programme set up by the MDC supported by funding from the Department of Health to establish and manage patient led neuromuscular forums to help shape the future of neuromuscular services in England. The forums bring together clinicians, NHS commissioners, patients and hospital trusts across England and Peter reported on several positive outcomes including a new NMD oriented physiotherapist role in Yorkshire and Humber and an increase to full-time for Anna Kent's part-time post of Neurological Conditions Clinical Specialist in the Milton Keynes Community Health Service. The MDC has also developed an online training manual on NMDs for GPs nationally, and is now extending it to other roles such as physiotherapists. Staff will be able to include the module in their annual mandatory staff development and it should lead to a better understanding of NMD patients in their care.

Neil Bennett, Research Communications Officer at the MDC, talked to us on 'Paving the Road to Treatment', describing the work of his team of research scientists and current progress in research. The team organises the peer review programme of MDC funded research into NMDs to select the most promising research projects, and supports communication between scientists working in the field of NMDs through arranging conferences and providing weekly online updates of ongoing research and research findings on NMDs. He mentioned the MDC funding for the development of a mouse model of FSH-MD to enable researchers to more fully understand the disease process. Neil also briefly went through some exciting developments in clinical trials of potential therapies for a number of muscular dystrophies. Duchenne MD was at the forefront, but as Neil explained, findings in one disorder can be relevant to a number of others. The therapy of exon skipping, i.e. providing a molecular patch that enables cells to "skip" over faulty sections of genetic code but still allows for a functional protein to be produced, which was first used in DMD has been extended to a number of other dystrophies including FSH-MD. Neil also outlined the New Research Strategy developed by the MDC which takes account of the increasing pace of research developments in the area of muscle wasting disorders. He also recommended the research pages on the MDC website if we wished to follow through on these matters - <http://www.muscular-dystrophy.org/research>.

Then Tess Adams gave a comprehensive talk on the 'adaptations' process. Tess is Lead Community Occupational Therapist at Milton Keynes Community Health Services and she provided an excellent insight into the way in which requests for support and 'adaptations' are assessed and reviewed. Apparently, in Milton Keynes people can refer themselves without having to go through GPs or social workers, etc. Tess explained that 'adaptations' are anything that involves a fixture to the home – something that can't be packed up and taken with you when you move. She also said that their emphasis is on resolving the difficulty the person is experiencing in the simplest way possible, so applications need to focus on the behavioural problem. After that she went on to describe the process of assessment, in terms of visits to assess the difficulty, the way in which recommendations are processed through the various committees, how quotes for the work are obtained, the difference between minor works and major ones, the different levels of funding available, the basis of mean's testing, the timescales and the difference between urgent and non-urgent work, and the importance of not starting work before all aspects of the work and funding are agreed, because grants are not paid retrospectively. There was much more, as well as a very lively question and answer session. I think we all felt that we had a much clearer understanding when she'd finished. However, one aspect Tess did stress was that different councils can organise the process differently, so it's important to check you understand your own area's system before applying.

After that, there was just time for Sebastian Jenkins, Volunteer Fundraising Manager at the MDC to give a quick review of fund raising across the country and mention a few local projects. For example, staff in Santander, MK held a 'dress-down Friday' and raised around £1,500, which the company matched, increasing it to £3,000. They are planning several more. Teresa, a member of our muscle group placed an MDC collection goblet in a local pub for customers to put in their change from buying drinks, and raised nearly £1,500.

As you can see, the agenda was varied and informative. The seating was laid out in a circle to encourage interaction, and speakers were happy to be interrupted. The agenda included 'social interaction' time at the end but the talks overran a little, so this was limited. However, although the meeting room was only booked for a two hour meeting nobody came to throw us out, and many stayed on for nearly an hour after the formal part of the meeting just chatting. In this way, many of us have got to know others with NMDs who live near to us and have also arranged to meet at other times. Sometimes people will meet up for coffee or lunch before the meeting as it starts at 1.00pm.

I hope that if you've not yet attended a Muscle Group Meeting in your region, this gives you a flavour of what they can offer. Speaking personally, they have given me the opportunity to meet staff at the MDC, hear about a range of relevant regional and national activities and developments and find out about service provision and personnel within my region. But perhaps even more importantly, they've also enabled me to make contact with a range of people with similar disorders and to hear and learn from their experiences. Some of them are now firm friends.

**Gill Penny**

# ACTION ON FSH



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

In our last newsletter, our very own Charlotte Daniels organised a table top sale at her local village hall.



Charlotte (pictured above) kindly invited me along as Chair of the Support Group. Charlotte worked incredibly hard on creating this event. The day was well attended with a variety of stalls including, cakes, jewelry, bric a brac, tombola and raffle. A personal thank you to Charlotte and all her family members who made this event so special. Charlotte's parents were busy in the kitchen making bacon sandwiches and refreshments. Charlotte and her family raised a fantastic **£491.50** on the day.

My children Ralph and Ronnie, (top right) came along too and helped with a sweet stall. We had a lovely day to remember and really enjoyed being a part of such a fantastic event.



Other fundraising events have been organised by Liz Orme (Vice Chair) who has been busy crafting and holding tea parties over the last year.

Natalie Woodcock (Treasurer) has organized many events including a carol concert and vintage stalls and has raised over £7000 for the Action on FSH appeal.

Other funds raised include Muscular Dystrophy Campaign Collection Tins being placed in our members' local shops and a steady stream of donations have been arriving on our my donate page via our website [fsh-group.org](http://fsh-group.org) via this button



Email newsletter subscribers can copy this link to access the page directly:

<https://mydonate.bt.com/fundraisers/mdcfshsupportgroupuk>

This is a really easy way to donate, clicking the button from the [fsh-group.org](http://fsh-group.org) page. You can click this button on the right hand side of our home page which will take you to our online total. This does not include donations sent directly to the Muscular Dystrophy Campaign, but I am delighted to announce that our Group's fundraising as a whole totals over £17,000.

Please do not hesitate to contact me if you wish to organise your own event at [fshgroup@hotmail.com](mailto:fshgroup@hotmail.com)

Liz Williams

# Summer Get Together 2014

Thank you to all who attended the FSH-MD Support Group Summer Get Together in Daventry on 28<sup>th</sup> June 2014.



A meal was arranged for delegates to meet each other on Friday 27<sup>th</sup> June, before the main event on the Saturday. We were entertained as a School Prom was being held in the function room at the hotel so we watched as the youngsters turned up in London buses, horses and carriages, limousines, convertibles and it was rumoured one set of guests turned up in a helicopter. As always, it was great to meet everyone and see old friends and new.

On the day Andy Findlay was our colourful compere and introduced our speakers. Louise Moyle, PhD Student, Kings College London was our first speaker giving an update on the research project investigating the role of muscle stem cells in Facioscapulohumeral muscular dystrophy.

Christopher Banerji, PhD Student, Kings College London presented his findings regarding FSHD networks, a new approach for drug discovery.

Libby Wood, Registry Curator, International Centre for Life, gave an insight into the data collected from over 400 registered FSH patients on the FSH Register. These findings can be found viewing the FSH Registry newsletter by following this link [http://www.treat-nmd.eu/downloads/file/registries/FSHD/FSHD\\_Newsletter2\\_July2014.pdf](http://www.treat-nmd.eu/downloads/file/registries/FSHD/FSHD_Newsletter2_July2014.pdf)

I will take this opportunity again to encourage members to sign up to the FSH Registry online by using this link: <https://www.fshd-registry.org/uk/>

I was really excited about the data given, and as you can see if you read the newsletter this information is invaluable for all sorts of reasons. For instance FSHers who are contemplating scapular fixation can see the data given from other FSHers who have undergone the procedure.

We were very excited to introduce, Liz Cowgill, Chairwoman of the FSHD Europe family interest group all the way from Holland, to give an overview of FSHD Europe.

Here is a link to the FSHD Europe website: <http://www.fshd-europe.org/>

Liz very kindly donated 50 copies of the book You're not alone with FSHD, for the Action on FSH Appeal and our members will have the opportunity to donate to obtain a copy at future events.

Our very own FSH-MD Support Group, Members Representative, Emma Orme gave a personal account of Access to University as an FSHer and went down a storm. This young lady will go far.

Maddy Rees, Advocacy Development Officer, Muscular Dystrophy Campaign, gave an overview of the Ambassadors Network and the web facility that is under construction giving a map of services for Neuromuscular patients. This is a very exciting project the Muscular Dystrophy Campaign are undertaking and will enable patients to quickly locate services and facilities in their local area, perhaps long term with a list of accessible leisure facilities which have equipment available to access their services, eg: swimming pools which have hoists or accessible ramps. This will develop over time and should be an invaluable resource. The Ambassadors Network goes from strength to strength with volunteers giving peer to peer support for all Muscular Dystrophy Campaign members should they need help with benefits, equipment and services. We will update our members when the map of services goes live and I for one will be clicking to see what is available. If you are interested in becoming an Ambassador for the Muscular Dystrophy Campaign, please contact Maddy Rees by email: [m.rees@muscular-dystrophy.org](mailto:m.rees@muscular-dystrophy.org)

Then it was my turn to take the microphone, a somewhat daunting prospect. I will take the opportunity again to thank the Committee and perhaps a few things I missed too. We do need Committee Members, we meet on average twice a year to discuss future plans and present projects and need new members. If anyone has the time to Volunteer please contact me via email: [fshgroup@hotmail.com](mailto:fshgroup@hotmail.com) for further information. If there is a particular area you wish to pursue then this is a great opportunity to do so with the backing of the Committee. All our members are volunteers and give up their time and experience to help others and we really need more members to be involved with the day to day running of the Group. If you are unable to travel we will accommodate this. Please do not hesitate to step up, you will be supported.

Please do not hesitate to contact me. I can email the slides from our respective speakers and supply further information on any of the subjects discussed. [fshgroup@hotmail.com](mailto:fshgroup@hotmail.com) or find us on facebook **search FSH-MD Support Group UK and ask to join.**



Liz Williams

# Karyomapping: researchers develop a new pre-implantation genetic test

You may have seen or read in the news that researchers and clinicians in London have developed a new test that can be used for pre-implantation genetic diagnostic. Using a new IVF-based technique called karyomapping allowed clinicians to select embryos not affected by a condition before implanting them into the womb of a mother at risk of having a child with a genetic condition. The first person in Europe to conceive after using this technique has Charcot-Marie-Tooth disease.

The news that the first person in Europe is now pregnant following use of a new IVF-based technique called karyomapping will be exciting news for many families affected by a muscular dystrophy or related neuromuscular condition. Carmen Meagu has Charcot-Marie-Tooth disease and was told that she had a fifty percent chance of passing the condition to her children. Clinicians at the Centre for Reproductive and Genetic Health in London used the new IVF-based technique to identify unaffected embryos before they were implanted into the womb, and Carmen is now 17 weeks pregnant.

The new IVF-based technique is called Karyomapping. It relies on the fact that the DNA of any two individuals varies - often tiny changes where a single letter is used instead of another. Since these changes are specific to an individual, if DNA from different members of the same family is analysed, researchers can build a family tree identifying which copy of a chromosome is inherited from which parent and even grandparent. In families affected by a genetic condition, by comparing the inheritance of chromosomes to family history of disease,

researchers can identify the copy (or copies) of the chromosome that hold a copy of a mutated gene that causes the condition.

Having built a family tree, clinicians can take a single cell from an embryo and identify which chromosome was inherited from each parent and whether the chromosome carries the mutated or functional copy of the gene causing the condition. This allows clinicians to choose embryos that are not affected to be implanted into the mother's womb.

The same karyomapping technique can be used for any genetic condition. The researchers believe that this will be more efficient than traditional pre-implantation genetic diagnosis which relies on testing embryos for the presence of a specific mutation found in one or both of the parents. However, like current PGD tests, karyomapping will only be useful when the gene causing an individual's condition is known.

Importantly, clinicians hope that the new test will be available for people on the NHS and, like current PGD tests, will give carriers or individuals affected by genetic conditions the chance to choose to have an unaffected child.

This article was sourced via the Muscular Dystrophy Campaign and the full article can be viewed by following this link:

[http://www.muscular-dystrophy.org/research/news/7735\\_karyomapping\\_researchers\\_develop\\_a\\_new\\_pre-implantation\\_genetic\\_test](http://www.muscular-dystrophy.org/research/news/7735_karyomapping_researchers_develop_a_new_pre-implantation_genetic_test)

Alternatively search Karyomapping on the muscular-dystrophy.org website.



## FORTHCOMING MUSCLE GROUP WORKSHOPS

Muscle Group meetings are a great opportunity for people living with muscle-wasting conditions, as well as their family and friends, to meet others affected and share advice and support.

Our forthcoming meetings will be adjustments and adaptations. This will be your chance to find out more information about these processes, and where you can get advice and support. There will also be an opportunity to discuss and share your own experiences of making adjustments or going through an adaptations process.

To join us at one of our workshops and for more information, please contact:

Peter Sutton on 020 7803 4838 or at or at

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

| Muscle Group workshop                            | Date of upcoming workshop | Time and venue  |
|--|---------------------------|---|
| South West Muscle Group meeting in Exeter        | Saturday 29 November 2014 | 2pm to 4pm, , Living Options, Units 3 - 4, Cranmere Court, Exeter |
| London Muscle Group meeting in Southwark         | Wednesday 3 December 2014 | 6pm to 8pm, Muscular Dystrophy Campaign Offices, London           |
| West Midlands Muscle Group meeting in Birmingham | Wednesday 3 December 2014 | 7pm to 8.30pm, Holiday Inn Express Birmingham NEC                 |



We really hope you find our newsletters interesting and informative. We have a thriving Facebook Group, set up by Karen Bayliss which now has 398 members. Our members are very generous with their comments and knowledge and this is a fantastic way to pick the brains of others in the FSH community. Please be assured that your posts can not be seen on your friends' newsfeeds. Posts may appear on your newsfeed but if you choose not to receive notifications from the group, they should not appear unless you click on the group to look at our page. This provides a safe environment for people to discuss FSH related issues in private.



Find us by searching FSH-MD Support Group UK in the search box on Facebook.

FSH-Registry

We will take this opportunity again to ask everyone to register with the FSH-Registry at the International Centre For Life, Newcastle upon Tyne. You can register at:

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

Alternatively please email: [fshdregistry@treat-nmd.eu](mailto:fshdregistry@treat-nmd.eu)

Or by post by writing to:

FSH-Registry

Institute of Genetic Medicine  
Newcastle University  
International Centre for Life  
Central Parkway  
Newcastle upon Tyne  
NE1 3BZ

Finally, if anyone would like to make a donation to the FSH-Support Group UK please could you make cheques payable to FSH-Support Group UK and send to:

**FSH Support Group UK**  
C/oLiz Williams  
27 Lawyers Close  
Evenley  
NN13 5SJ