

UK FSHD PATIENT REGISTRY NEWSLETTER

www.fshd-registry.org/uk

Issue1 • December 2013•



Welcome to the Registry!

The UK registry has been launched just 6 months and over 320 people are registered. This newsletter will hopefully explain why the registry is important and what you can do to ensure it will have maximum impact. We also summarise some of the data collected in the registry so far.

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1. Is Your Record Complete?

The registry is only as useful as the information it contains so it is important you keep your details up to date. You will be reminded to update your record once a year. If anything does change before then, e.g. your contact details, then remember to update them in the registry too.

While updating your details remember to check if you have:

Chosen a doctor; your doctor confirms the genetic details of your diagnosis. If your doctor is not listed on the registry then please contact the registry curator providing the details of your neuromuscular specialist. We are not able to include GPs on the registry, if you do not see a neuromuscular specialist then please contact the registry to discuss alternative ways we can obtain the details of your genetic diagnosis.

Completed all the questionnaires; the more information we have the more useful the registry can be. The details you provide may help to inform future research and improve standards of care.

Useful Contacts

The Registry:

E-mail: fshdregistry@treat-nmd.eu

Telephone: 0191 241 8640

Post:

TREAT-NMD Office
Biomedicine West Wing
International Centre for Life
Newcastle upon Tyne
NE1 3BZ

Muscular Dystrophy Campaign:

www.muscular-dystrophy.org

Email: info@muscular-dystrophy.org

Telephone: 0800 652 6352



Libby Wood is the registry curator. She works in Newcastle with the TREAT-NMD team. Libby is responsible for making sure all of the data in the registry is accurate and up to date. She is your point of contact for everything to do with the registry so please don't hesitate to get in touch if you have any questions.

2. Clinical Trials: Why we need a registry.

More is known about FSHD than ever before and research continues to move quickly bringing us closer to clinical trials. The primary aim of the registry is to make research, including clinical trials, easier and faster. Clinical trials are rigorously controlled tests of a new treatment or therapy using human subjects. These are important research studies that ensure a drug, treatment or therapy is safe and effective.

Rare diseases, like FSHD, face many challenges when developing a potential treatment or therapy from the laboratory to clinical trials. In particular knowing where people with the condition are and how many are available to take part. The patient registry can be instrumental in helping move past these obstacles not only as a source of information but more importantly as a recruitment tool; being able to contact hundreds of people quickly and efficiently.



3. How the registry works: Who has access to the data?

The registry allows researchers access to detailed information about patients with FSHD while protecting the identity of those involved. If a researcher would like to use the registry they must submit a proposal to the steering committee (find out more about the steering committee on page 3). This committee votes to allow the researcher to work with the registry.

Researchers may want to use the registry for a number of different reasons:

- Finding out how many people there are with a certain symptom or in a certain location. (e.g. how many people have hearing difficulties or how many people are over 35)
- Contacting patients about a clinical trial or other research they may want to participate in.
- Using the registry to send out research questionnaires or ask additional information of the people registered.

Researchers are then only provided with anonymous aggregate data and will never be able to contact you directly. If the data in the registry suggests you may be eligible for a study or trial then we will contact you, researchers will never be given your personal details. You will never be asked to contact a commercial company but may be directed to a doctor or nurse helping to carry out the study.

You do not have to participate in any research you hear about through the registry. Participation in research of any kind should always be entirely voluntary.

4. The Steering Committee

The registry was established by a team of researchers, doctors and patient representatives from across the country that now make up the Steering Committee. The responsibility of the Steering Committee is to monitor the registry's activities and to determine its strategic direction. Having a wide range of people overseeing the registry ensures that everyone's interests are looked after. The committee has helped make the registry possible and they continue to provide invaluable input and support. The members of the committee are listed to the right. More information about the committee members can be found on the registry website.

- Mark Busby, Bradford
- Andrew Graham, Patient Representative
- David Hilton-Jones, Oxford
- Hanns Lochmüller, Newcastle
- Cheryl Longman, Glasgow
- Peter Lunt, Bristol
- Fiona Norwood, London
- Richard Orrell, London
- Marita Pohlschmidt, Muscular Dystrophy Campaign
- Mark Roberts, Manchester
- Stuart Watt, Patient Representative
- Suzanne Watt, Patient Representative
- Tracey Willis, Oswestry



The MUSCULAR DYSTROPHY

CAMPAIGN was founded in 1959 and is an important contact for people with a muscular dystrophy or related neuromuscular condition. They fund world class research and provide practical information, advice, and emotional support to people affected by all types of muscular dystrophy and related neuromuscular conditions. They are funding research into FSHD at the University of Nottingham and King's College London, being carried out by Professor Jane Hewitt and Dr Peter Zammit respectively. They currently have an "Action on FSH" helping to fund this important research and the registry itself. Dr Marita Pohlschmidt is Director of Research at the charity and also sits on the registry's steering committee; more information: www.muscular-dystrophy.org



FSHD-MD Support Group is run by members for members and exists to improve the quality of life for those who have the condition as well as their carers, families and friends. They offer support and information through their website and newsletters. The support group also has a private, very active forum page on Facebook where members can ask for advice or just exchange information that might improve member's quality of life. They are a point of contact for the newly diagnosed and are able to facilitate contact between members. The group also runs annual themed "Get Togethers", Information and research days and social events across the UK. They work closely with the Muscular Dystrophy Campaign and have been very important in helping raise awareness of the registry. For more information: www.fshd-group.org

FAST FACTS

79%

Of people registered experience weakness in their facial muscles.

97%

Of people registered report shoulder weakness, the most commonly reported symptom.

75%

Of people registered report foot weakness.

80%

Of people registered report weakness in the hip girdle muscles.

56%

Of people require some assistance with walking

19%

Of people registered reported experiencing hearing loss

5. Registry Statistics

The registry has been launched for six months and there are now over 320 people registered. The data collected provides a great insight into the FSHD community in the UK. This data may help inform future research and can be used to help develop nationally agreed standards of care. There is a broad age range represented (from 2 to 82 years), with slightly more women (55%) than men (45%) registered (fig 1). There is a wide distribution of people registered from all over the country, as far north as the Shetland islands and as far south as Cornwall (fig 2).

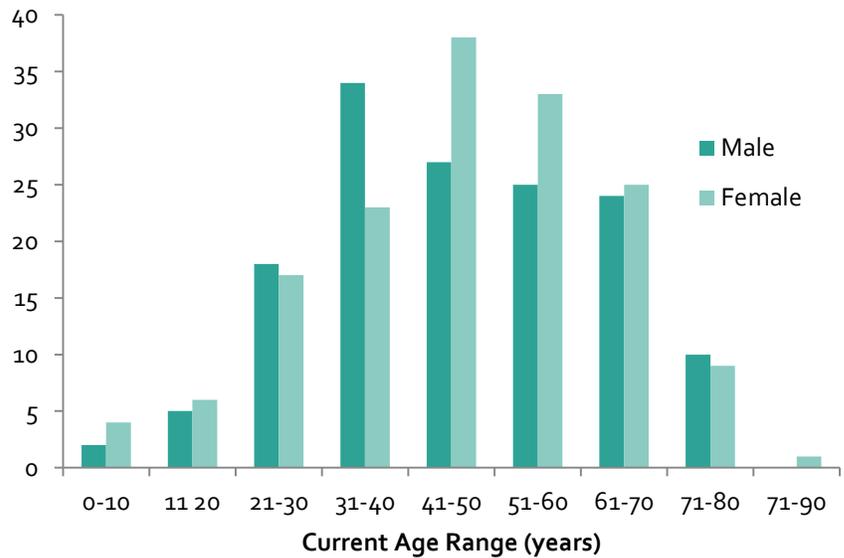


Figure 1: Current Age range (years) stratified by gender

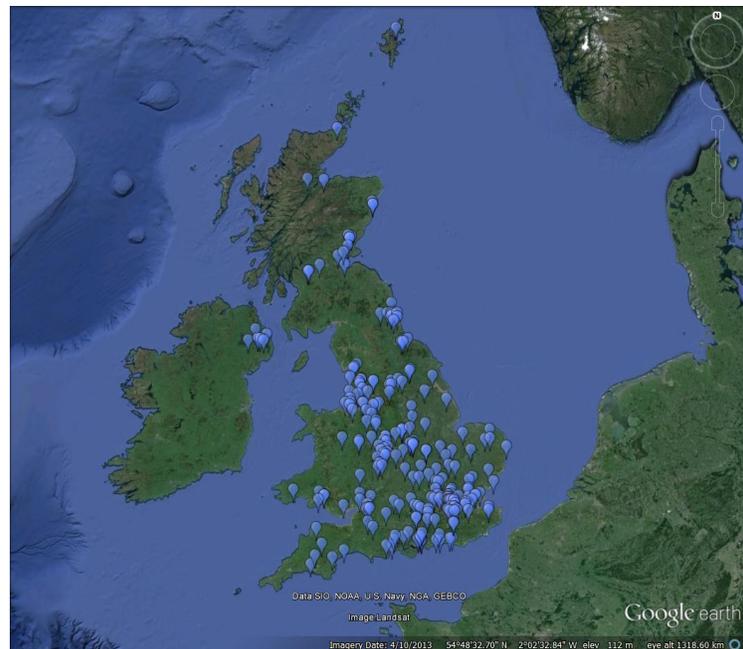


Figure 2: Map of people registered (October 2013)

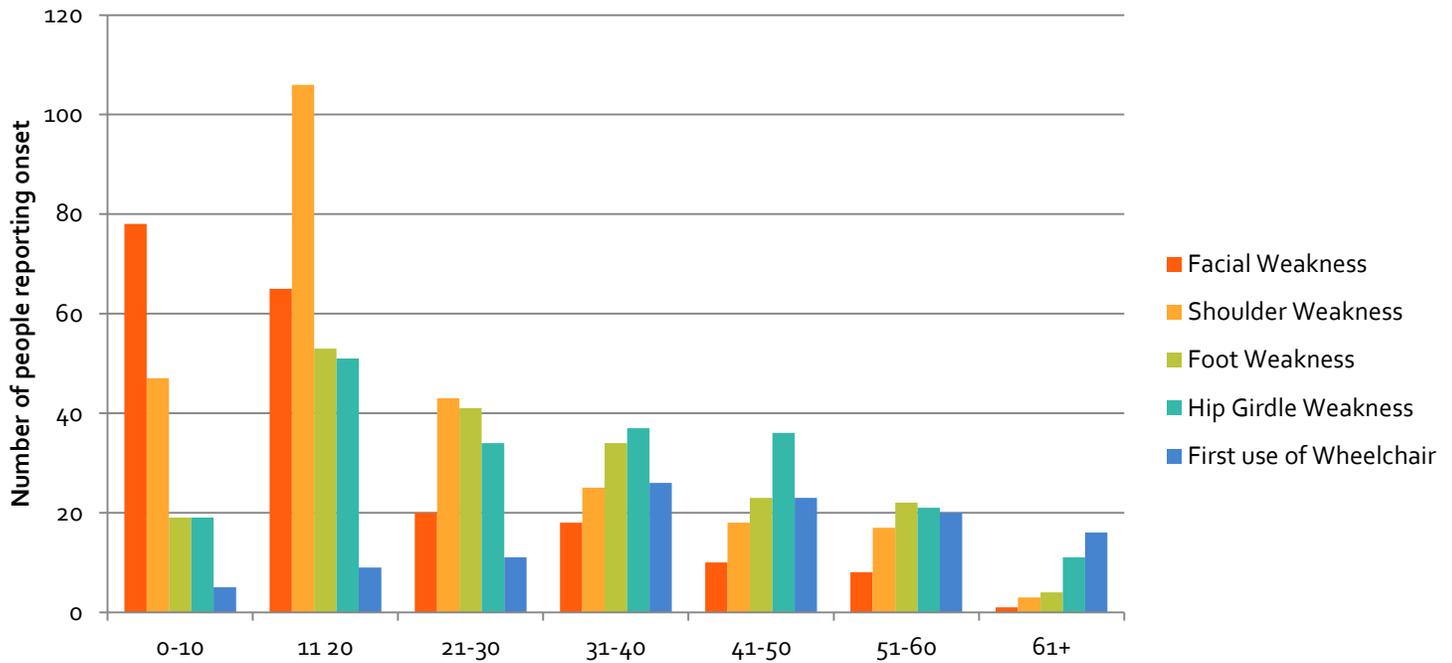
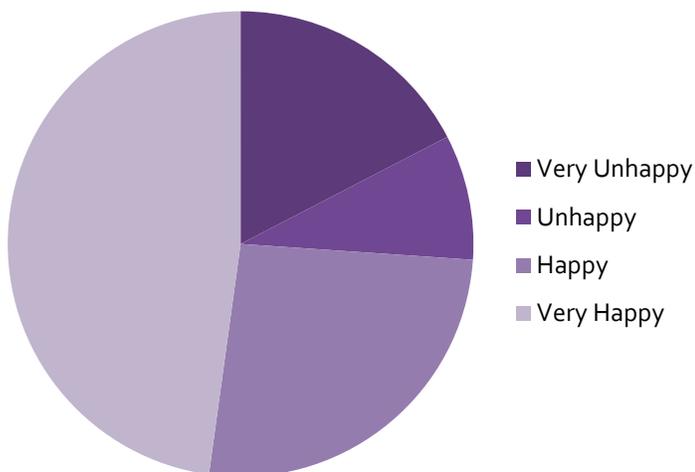


Figure 3: Age range (years) of onset for multiple symptoms

The registry asks about the different symptoms experienced and the age at which they first developed (fig 3). Problems with facial muscles (e.g. trouble smiling or whistling) occurred before the age of 20 in more than 70% of people registered. The largest proportion of people experiencing weakness in the shoulder muscles first developed this symptom between 11 and 20 years old. The use of a wheel chair shows an opposing trend with the largest number of people beginning to use a wheelchair between 31 and 40 years old.

Scapular Fixation



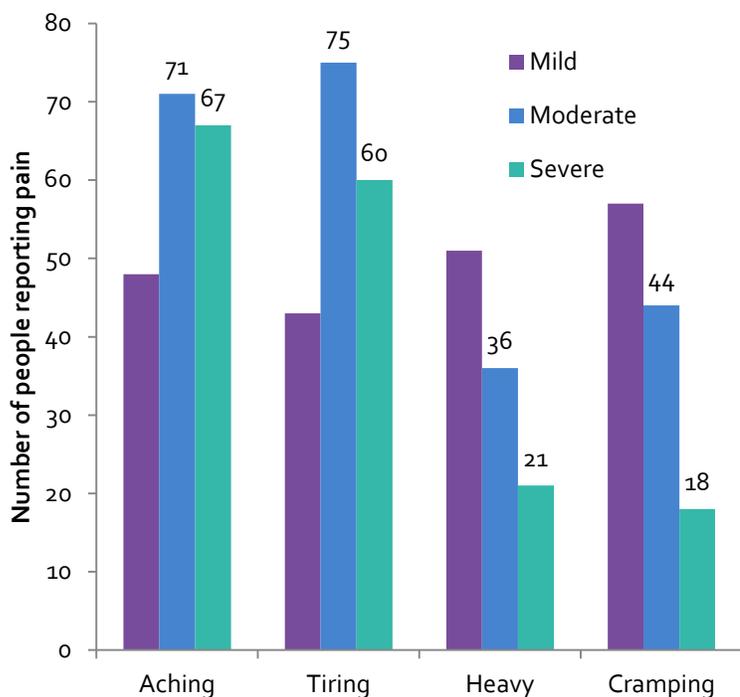
As part of the registry an additional questionnaire is asked about scapular fixation. 28 people registered indicated they have undergone scapular fixation and 23 have completed the questionnaire. None of these people have had the fixture removed and 71% would recommend the procedure to others.

Around 40% of those answering still feels some pain as a result of the operation and a similar amount have some muscle wasting in their hand as a result of the procedure.

Figure 4: Are you happy you underwent scapular fixation?

Pain

The registry contains additional questionnaires about pain. These questionnaires were added after consultation with patient organisations, researchers and doctors who thought that there was not enough information available about the pain experienced as a result of FSHD. The questionnaires ask about the different types of pain experienced, its intensity and also the different methods used to help alleviate it.



Question: Please describe your pain during the last week.

When asked to describe the pain experienced over the last week most people used the words aching (75%) and tiring (72%). A number of people also used the words heavy (44%) and cramping (48%). The severity of the pain reported varied with the type of pain described with more people describing pain as aching also finding it severe and those people describing cramping pains reporting this as mild. The same types of pain were reported across all ages and gender.

Figure 5: Description of pain during the last week

Question: How would you describe the level of chronic pain you have experienced?

As part of the pain questionnaires we ask about different types of pain including chronic or persistent pain. This is described as daily pain that can be intermittent or constant experienced for at least 12 weeks of a year. 59% of people completing this questionnaire experienced this kind of pain. Over half (53%) of them describe this type of pain as distressing; horrible or excruciating (fig8). This type of pain is most often felt in the shoulder (75%).

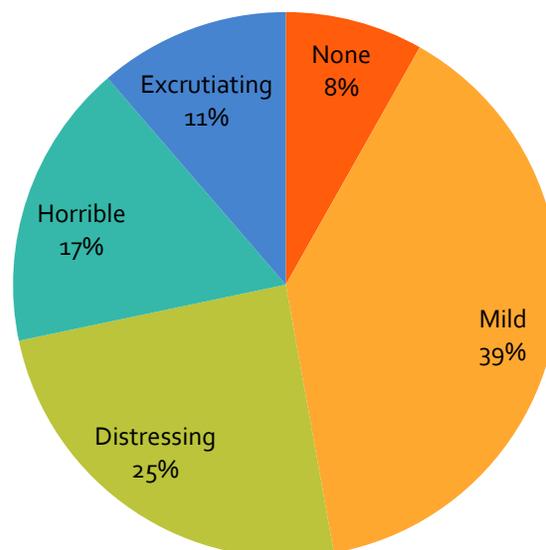


Figure 8: Severity of chronic pain described.

Question: Please describe the greatest level of pain you have experienced in the last seven days because of your FSHD in the following locations.

People answering this question were asked to express the severity of pain (mild, discomforting, distressing, horrible or excruciating) in various different areas of the body (arms, hands, hips, legs and feet). More people reported experiencing pain in their legs than anywhere else with least number of people reporting pain their hands. In all areas mild pain was experienced the most frequently with less than 17% of people describing pain as distressing, horrible or excruciating for any single area.

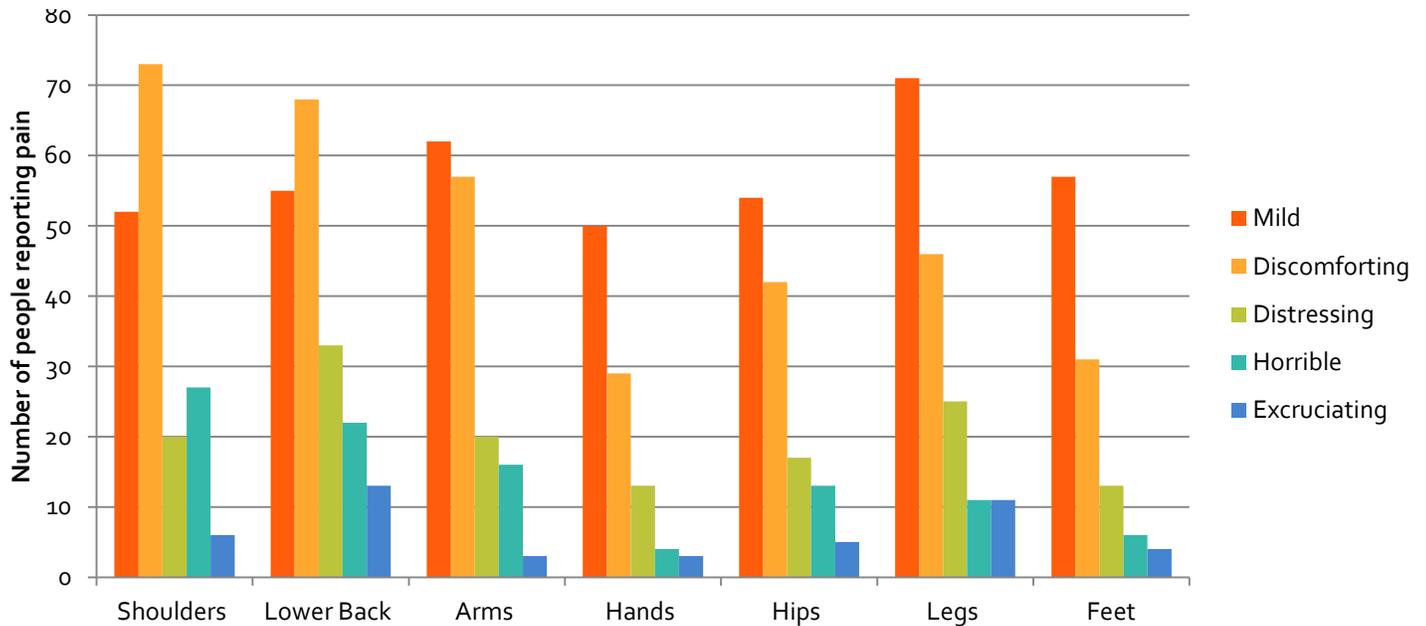


Figure 6: Severity of pain experienced in different locations.

Physiotherapy

As part of the registry information is collected about the treatments and therapies used to help alleviate pain this includes physiotherapy. A total of 109 (41%) of patients completing the pain questions have received physiotherapy to help them deal with pain. The majority of people undergoing physiotherapy did not have to pay privately for the treatment. Of those having had physiotherapy 52% attended 1 or 2 sessions a month with 60% attending less than 10 sessions in total. 42% of people reported at least some reduction in pain.

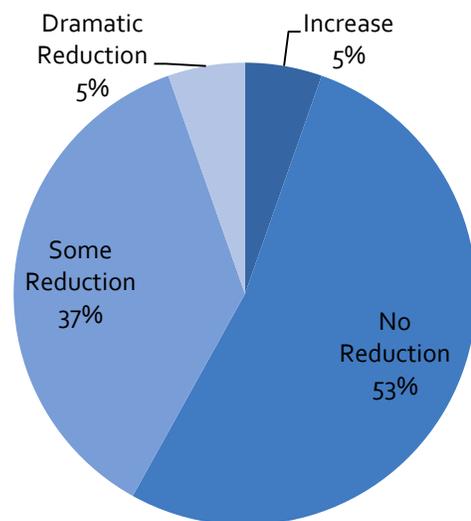
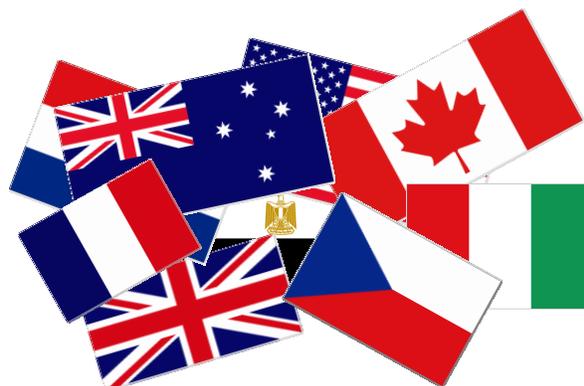


Figure 9: Did you see a reduction in the pain you experienced due to physiotherapy sessions?

Nation	Number Registered (approx.)
United Kingdom	320
United States	750
Czech Republic	70
Italy	> 1000 patients
Egypt	6 (newly launched)
Canada	60
France	150
Netherlands	Launching soon
Australia	Launching soon.

6. FSHD Registries: An international effort



The UK registry is part of an international network of TREAT-NMD registries collecting information about people with FSHD. All of these registries collect similar information so they can be utilised as a single data source for trial recruitment and planning. Italy and the United States have the largest and most established registries with many more getting ready to launch soon.

In rare diseases no one country is enough and it is important to collaborate and share information with people all over the world.

Thank you for reading this newsletter, if you have any feedback or suggestions for the next issue then please contact the registry curator, Libby Wood.

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