UK FSHD PATIENT REGISTRY NEWSLETTER

ISSUE 2
July 2014
www.fshd-registry.org/uk
www.treat-nmd.eu
www.muscular-dystrophy.org

Accelerating research and improving care in Facioscapulohumeral dystrophy

Remember to update your details: The registry is only as useful as the information it contains.

Inside this Issue:

The UK FSHD Patient Registry is funded by the **MUSCULAR DYSTROPHY CAMPAIGN**. 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 currently have an "Action on FSH" helping to fund 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

Welcome to the second newsletter of the UK FSHD Patient Registry.

It has been one year since the launch of the FSHD patient Registry. Over 400 people are now registered. This has surpassed our expectations and we would like to thank everyone involved. This our second newsletter discusses how and why it is important to keep your details update. It will also introduce some FSHD research and provide an update on the information within the registry. You can find previous newsletters under the newsletter section of the website.

1. Update your details

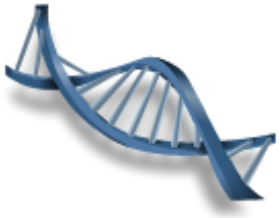
Every year you will receive a reminder to login and update your details. This is so we always have the most accurate information about you in the Registry.

Contact details: If we want to contact you about a study we think you might be interested in or provide updates like this newsletter it is important your e-mail, postal address and telephone number are up to date. Please note that your personal details are only available to a limited number of Registry staff and will **never** be given to a third party.

Your Condition: This information will inform researchers if you are eligible to take part in a clinical trial or study. It may also be important for researchers wishing to look at how FSHD progresses in different people. Please check this information is up to date at least once a year and report and changes in circumstance, e.g. if you start using a wheelchair.

Pain and Quality of Life: We hope the information in these sections will help inform research in the future and potentially help develop standards of care, too. If we can collect this information at different time points, evenly spaced apart (once a year) this will provide more information to show how FSHD can progress.

2. We need to know your genetic diagnosis!



Your genetic diagnosis is one of the most important pieces of information within the Registry. This is currently provided by your neuromuscular consultant. However, if you do not see a neuromuscular consultant (though we recommend you do!), it is still important we have this information. Most studies and trials looking for participants will only include people with genetically confirmed FSHD. If you have a copy of your genetic report this can be sent directly to the Registry. Alternatively you can speak to your consultant (neuromuscular specialist) next time you have an appointment and they should be able to provide you with a copy. If you have any questions, or are unsure if we have your genetic details please contact the Registry curator, Libby Wood, fshdregistry@treat-nmd.eu

3. A new study recruiting in Newcastle.

Who are CINRG?

CINRG is an international consortium of medical and scientific researchers from academic and research centres whose common goal is to positively impact the lives of neuromuscular disease patients. The network includes 28 centres and Newcastle is the first UK site within the CINRG network. This study in FSHD will be the first Newcastle is taking part in. You can find out more about CINRG on their website: www.cinrgresearch.org.

What is a natural history study?

A natural history study collects information about a group of people with the same condition. It does not involve a drug, treatment of therapy. Understanding the "normal" progression of disease is important when testing for new therapies. We need to know what the normal characteristics are before we can know if they are changed by treatment or therapy.

A new observational study will start recruiting in Newcastle this summer. The study is led by a group called CINRG (Cooperative International Neuromuscular Research Group) who are based in the United States. The CINRG network is made up of 28 sites across the world and 17 are taking part in this study. At the moment Newcastle is the only site in the UK. The study is looking at the infantile onset form of FSHD, in particular people who experience symptoms **before the age of 11 years old**. The aim of the study is to learn more about infantile onset FSHD and provide a better understanding of how FSHD affects people differently. The study will also see if these differences can be seen in the genetics of this person and aims to create standardised methods for testing muscle weakness in children and adults with infantile onset FSHD. This will all be important for future clinical trials.

What does the study involve?

This study would involve one visit to the Royal Victoria Infirmary in Newcastle upon Tyne. This will be a full day visit which will involve a number of different assessments carried out by the doctor and physiotherapist. This will include physical assessments, hearing and eye tests, cognitive testing, strength testing and speech assessments among others.

How can you take part?

This study is recruiting through the Registry and you will have been contacted separately if based on the information you have provided we think you might be eligible to take part. This is a small study and only a limited number of people are expected to be recruited to the Newcastle site. You can find more information at www.clinicaltrials.gov, just search for FSHD.

4. Research Update

NeuromOmics : improving diagnosis and looking for treatments

Neuromics is an EU funded project looking at different neuromuscular (for example muscular dystrophy) and neurodegenerative (for example Ataxia and Huntingdon's disease) conditions. FSHD is one of the named muscular dystrophies this project will be trying to find out more about. This is a laboratory based research project looking to improve diagnosis and develop new treatments using the latest –omics technologies.

What is –omics technology?

–omics has been added to previously existing fields in research in order to indicate that the research involves a lot of data and is on a very large scale. For example: genomics or proteomics. This technology is possible due to the ability to sequence our genes. This started with the human genome project revealing a 3 billion long combination of bases (the simplest bacteria has just 600,000 bases in its genome). We are now able to sequence these 3 billion bases faster and cheaper than ever before. This allows us to carry out this data-rich research.

How will Neuromics help people with FSHD?

Neuromics hope to discover more genes involved in FSHD and determine how and why they might affect individual patients differently. The project will also identify and confirm new biomarkers. Biomarkers are substances in the body than can be measured accurately to see how a disease is progressing. This will be important for future clinical trials. If a drug changes the progression of disease this should show in the level of biomarker. These substances are most useful if they can be found in the blood or urine as these are easy to test.

The Neuromics project will take place over five years and is funded by the European Commission until September 2017. You can find more information and sign up for the Neuromics Newsletter at www.rd-neuromics.eu.

Research funded by the Muscular Dystrophy Campaign: Action on FSH Appeal

The Muscular Dystrophy Campaign (MDC) is funding two PhD students and their important research into potential targets for treatment of FSHD. Below is a brief summary of the projects, they are both in the early stages and more information can be found on the MDC website www.muscular-dystrophy.org.

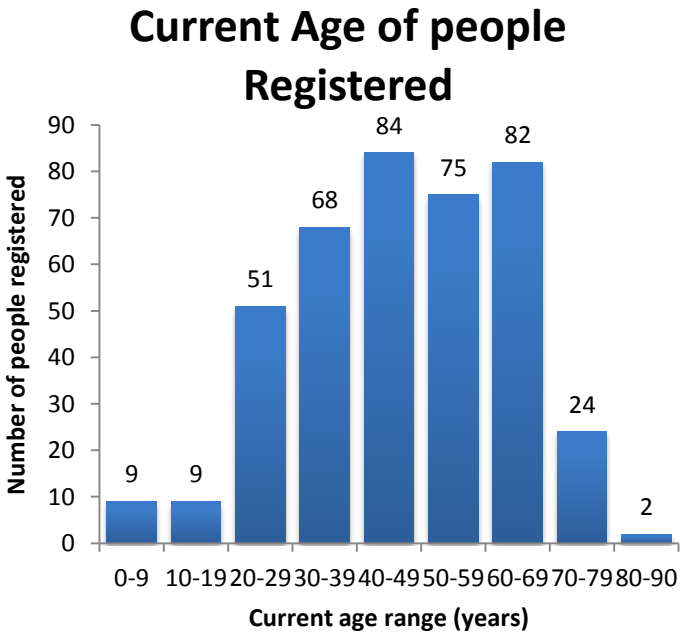
Christopher Banerji (University College London) is using a unique approach to research. He is carrying out his PhD at Centre for Mathematics, Physics and Engineering in the Life Sciences and Experimental Biology at University College London. He is using maths and computer technology to sift through publically available information about the genetic profiles of FSHD. This method has produced a set of biological networks that might be uniquely associated with the disease process in FSHD. One of these networks centres on a gene called beta-catenin which is part of the Wnt signalling pathway. This pathway is being looked at by a number of groups as a potential area for therapeutic development.

Louise Moyle (King's College London) is looking at muscle cells grown in a dish to find new ways of stopping the toxic gene DUX4 from being expressed. The mutation that causes FSHD allows DUX4 to express when it is not supposed to. Her work focuses on a particular enzyme (a molecule that speeds up reactions in cells) called Ret. Louise is still working to see if Ret is a major factor in FSHD, but if Ret signalling turns out to play a role blocking it could be a treatment strategy. This pathway is also involved in some cancers and some drugs to inhibit Ret are being developed for cancer treatment.

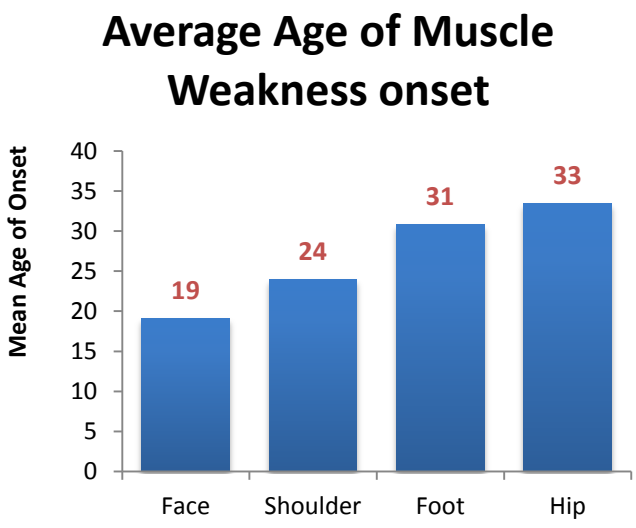
5. Registry Update

Some of the data in the Registry was recently presented at the FSH- MD Support Group UK annual meeting in Daventry. You can find out more about the support group and what they do on their website www.fsh-group.org. As many of you may not have attended the meeting the Registry data that was presented is summarised for you here.

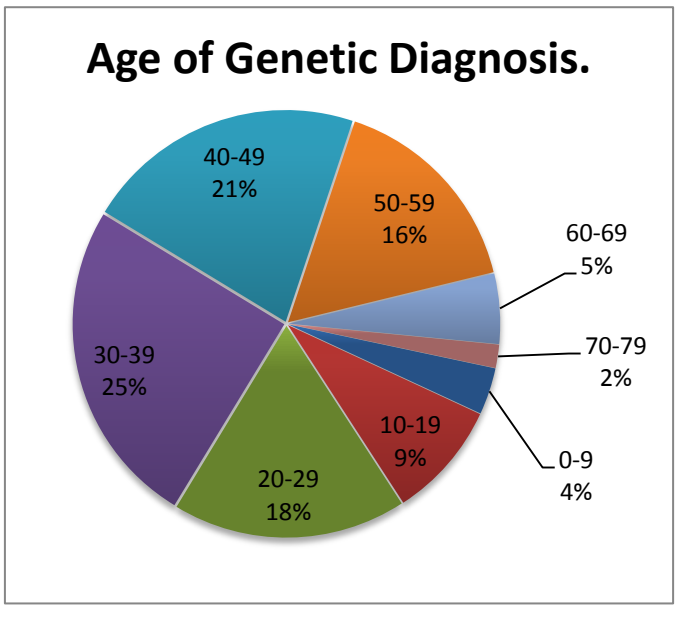
Location: People have registered from all over the country, with Wales, Scotland, Northern Ireland and England all represented.



Age Range: The majority of people registered are in mid to late adulthood, with 59% between 40 and 70 years old.



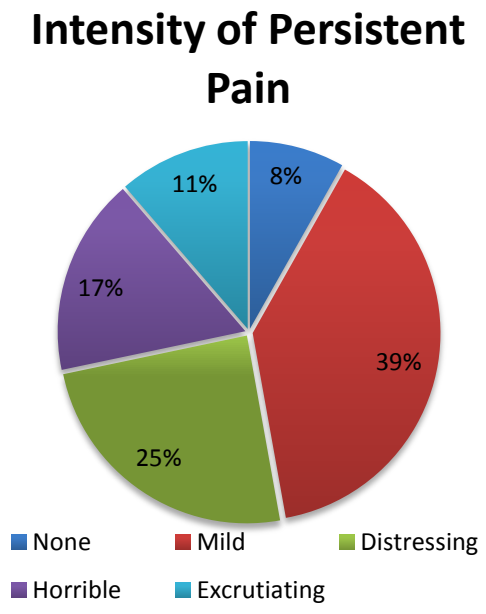
Muscle Weakness: The chart above shows the mean (average) age of onset of muscle weakness in different areas of the body. It is clear that most people report weakness in their face and shoulders before in the lower body.

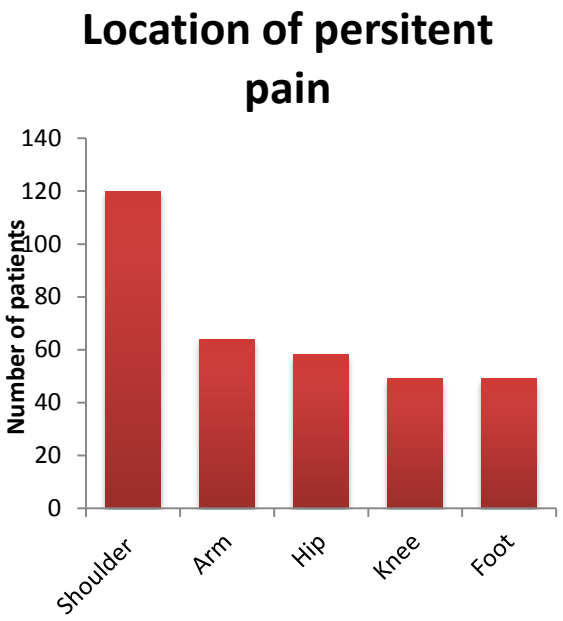


Genetic Diagnosis: We do not have genetic diagnosis for everyone on the Registry- we will be working hard to get this over the next few months. Of those we know most were diagnosed after 30 years old. The mean (average) age of diagnosis is 38 years old.

Pain

The registry contains a number of questionnaires about pain; of the 400 people registered 350 have completed at least some of these questionnaires. We include information about pain as our steering committee (including patient representatives, doctors, scientists and the Muscular Dystrophy Campaign) wanted to know more about the pain associated with FSHD. We hope this can lead to better understanding about the level and type of pain experienced and find better ways to help manage this pain.

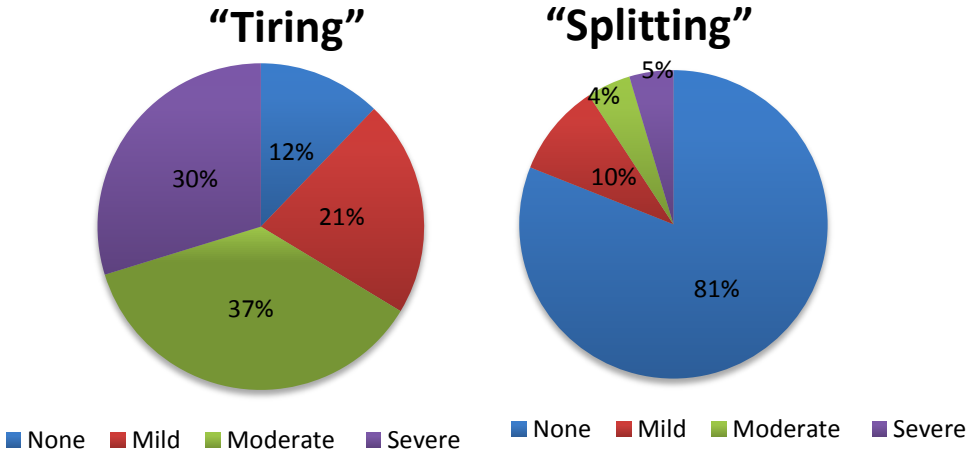




Persistent pain: Persistent pain is pain that is experienced for at least 3 months of the year, either constantly or intermittently. Sixty one percent of people registered experience pain like this with the majority describing it as distressing, horrible or excruciating

Location of pain: The area that pain is reported coincides with areas of muscle weakness, but is reported more in the shoulders than anywhere else.

Types of pain experienced

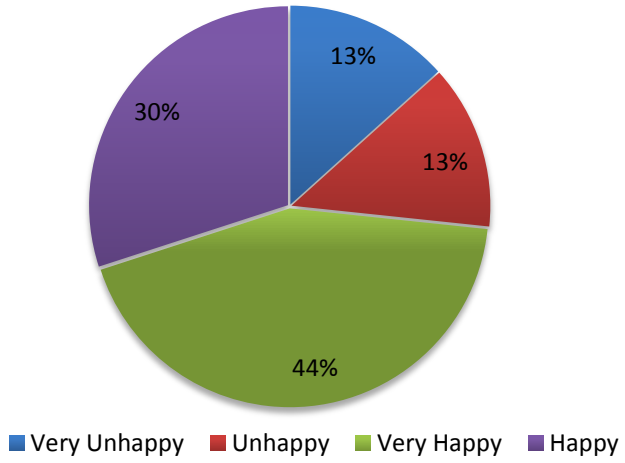


Types of Pain: One of the questionnaires within the Registry asks about the different types of pain experienced in the last seven days. A long list of words that could be used to describe pain is presented. The words that are most commonly selected are tiring (above), heavy and aching. With words like Splitting (see above) and stabbing being reported the least frequently.

Scapular Fixation

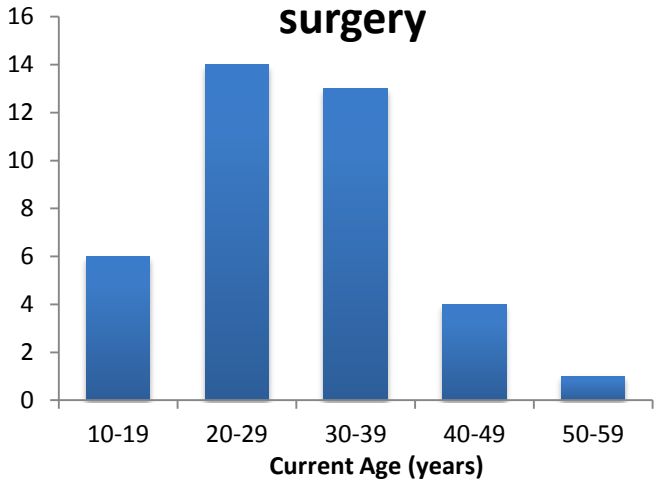
A questionnaire about scapular fixation (or thoracoscapular fusion) is included in the Registry. Forty out of the 400 people registered have had this operation with 30 completing the additional questionnaire. Some of the results of are summarised here.

Satisfaction after surgery



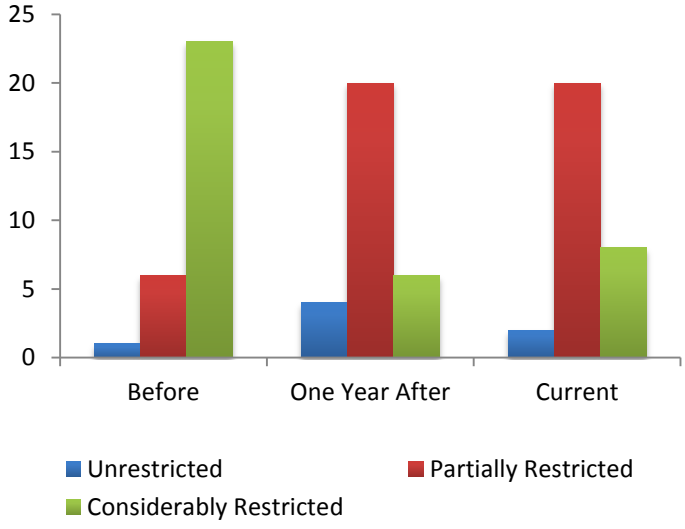
Satisfaction: The majority of people, 23 out of 30 were happy with the outcomes of their scapular fixation.

Age at which people had surgery



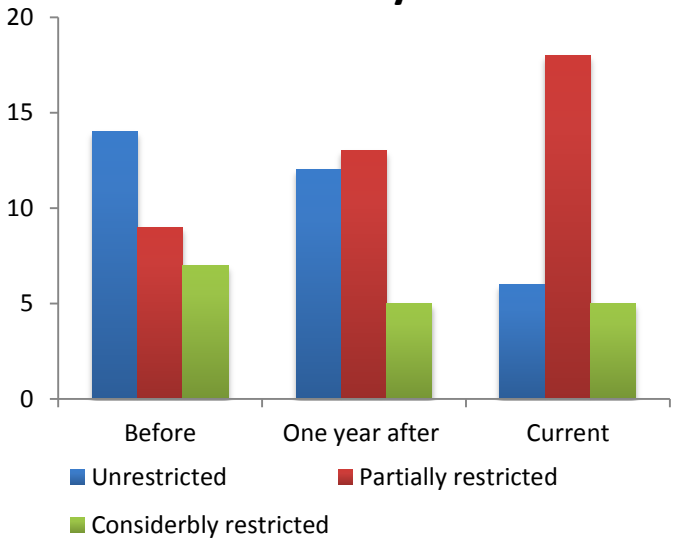
Age of surgery: People underwent scapular fixation at a broad range of ages but most often between 20 and 40 years old.

Change ability to raise arm



Arm Raise: The ability to raise the arm changes before and after surgery. Most people found this range of movement improved one year after surgery and that this is still the case now (ranging from 1-48 years after surgery , on average 15 years later.)

Change in rotational mobility



Rotational Mobility: Change in the ability to rotate the upper body has been reported after surgery. Most people report little change within the first year but restrictions seems to increase over time for some of the people completing this questionnaire.

The UK FSHD Registry recognised internationally.

In addition to the FSH-MD Support Group UK meeting the Registry is also being presented across the world. In order for the Registry to have the biggest impact we need to spread the word among the global patient and research community. Increasing interest is important so that the Registry can be used to its full capacity. In May this year a poster about the Registry was presented at the European Conference for Rare Diseases and Orphan Drug products (ECRD), our poster was ranked in the top 20 by the judges. A summary of this poster (also known as an abstract) will be published in the Orphanet Journal of Rare Diseases (OJRD) later in 2014. The Registry will also be presented later this year at the 3rd International Workshop on Rare Disease Registries in Rome, Italy. The Registry will be presented as an example of an advanced rare disease Registry.

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.

E-mail : fshdregsitry@treat-nmd.eu
Telephone: 0191 241 8640
Post:
TREAT-NMD Office
Biomedicine West Wing
International Centre for Life
Newcastle upon Tyne
NE1 3BZ

www.fshd-registry.org/uk