UK FSHD Patient Registry Newsletter

ISSUE 5
http://www.fshd-registry.org/uk
http://www.treat-nmd.org

Accelerating research and improving care in FSHD

Remember to update your details and tell your doctors about the registry – it's important for us all to work together.

Inside this Issue:

We attended the 2019 FSH Society International Research Congress in Marseille on 19-20th June 2019. Here is the registry curator presenting the UK FSHD Patient Registry and the research it has supported. For a copy of the poster please contact Ben Porter.

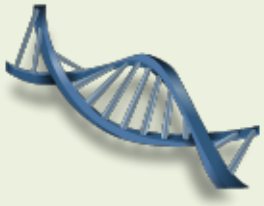
Welcome to the fifth newsletter of the UK FSHD Patient Registry.

As of April 2020 there are 987 participants registered with the UK FSHD Patient Registry. A huge thank you is in order for all of the patients, clinicians, caregivers and patient organisations who have supported and contributed to this superb achievement.

1. Please provide your genetic report where possible

Your genetic diagnosis is one of the most important pieces of information within the Registry. This is currently provided by your neuromuscular consultant if you see one. However, if you do not see a neuromuscular consultant (though we recommend you do), it is still important we have this information. If you have a copy of your genetic report this can be sent directly to the Registry curator. Alternatively you can speak to your neuromuscular consultant at your next appointment, they should be able to provide you with a copy of the report if you have been tested.

Most studies and clinical trials in FSHD looking for participants will only include people with **genetically confirmed** FSHD1 or FSHD2.



REMINDER - if your doctor does not appear on the registry as a selected healthcare professional then please update them about the registry at your next appointment or via email, and to contact the registry curator for further information.

2. New studies and research updates

Activity monitoring in progressive muscle diseases – ongoing ★

This study is being run from King's College London.

The purpose of the study is to measure active and resting behaviours in people with progressive muscle weakness, and to test the quality of measurement. Participants are asked to complete various questionnaires and to wear an activity monitor for at the start and end of the study. An optional part of the study is to provide participants with their own Fitbit activity monitor whilst they were monitored remotely.

The study recruited participants aged 18 years and above who had a diagnosis of a progressive muscle disease (such as FSHD).

Following the promotion of this study through the registry, 196 participants expressed interest in taking part, however, only 4 participants were included in the study. This was due to a high volume of interest. Results will be shared once the study has concluded.

Academic pain survey - complete ★

This survey was collected by the University of Southampton as a part of a research project studying the quality of registries that collect information on pain.

By collecting information about the registries such as the type of information collected and how often the data was updated, the study will help contribute to making a registry of all pain registries internationally. Through this the researchers hope to optimise the quality of pain registries therefore, improved patient health may come as a result of a good quality registry.

Sleep in FSHD survey – not yet recruiting ★

A survey will soon be distributed through the registry to understand more about sleep in FSHD, including sleep difficulties, strategies for coping etc.

The current research suggests that neither the psychology nor physiology of sleep has been examined in those with FSHD yet.

ReSOLVE FSHD natural history

A new FSHD research study, entitled, "**(ReSOLVE)",** is being conducted at 8 sites around the U.S, as well as at sites in Europe.

The study is designed to help drug developers successfully design clinical trials by validating two clinical outcome assessments and refining clinical trial strategies. All participants will be asked to undergo FSHD-specific functional rating scale tests and procedures and electrical impedance myography.

This study will enroll 150-220 people, aged 18-75, with **FSHD or FSHD1.**

For further information on the study protocol and requirements and please visit: https://clinicaltrials.gov/ct2/show/NCT03458832 or https://dx.doi.org/10.1186%2Fs12883-019-1452-x.

Natural history study collects health information in order to understand how the medical condition or disease develops and how to treat it.

Clinical outcome assessments measure a patient's symptoms or mental state. They are useful in showing whether a drug is beneficial or not.

FSHD SOCIETY ★

In February, the registry promoted a series of surveys created by the FSHD Society, one of the largest FSHD patient organisations in the world. These are to help communicate the patient voice for an upcoming meeting with the Food and Drug Administration in the USA, that will be used to evaluate future FSHD therapies. The surveys can be accessed from: https://www.fshdsociety.org/fsh-events/vopf/.

In February, there were already 50 participants from the registry who completed all five of the surveys. Thank you to all of those who have since completed these surveys, and to all those who plan to complete them.

For those interested, you can also view the latest FSHD Society newsletter here: https://www.fshdsociety.org/wp-content/uploads/2020/02/FSHD-Advocate-Magazine-2020-Issue-1.pdf. This contains lots of exciting updates in research and clinical trials.

SRA 004-2018: A Patient Focused Survey to Assess a Proposed Clinical Study Design in Patients with FSHD ★

Background

In February 2019, the registry circulated an online survey for Fulcrum Therapeutics, about a proposed clinical study design, to FSHD patients who met the eligibility criteria they provided. The survey included information about various aspects relating to the clinical trial and it was felt that before running this, it was very important to capture the patient voice, to ensure that patients with FSHD would actually enrol in the clinical trial.

Results

Fulcrum Therapeutics approached **6** different sites from **North America** and **Europe**, and the UK FSHD Patient Registry provided the greatest recruitment out of all of the sites. Out of the total number of participants recruited, the registry provided **41%** of those taking part.

Thank you to all those who participated! This survey provided extremely valuable information to the company and they too were highly appreciative of those participants, and the registry itself for being such a useful research project. We hope that with this relationship established with Fulcrum Therapeutics that the UK could be involved in future clinical trials.

REMINDER - If the registry is used to promote or assist with the recruitment for a clinical trial or research study, all eligible patients will be contacted by the registry curator via email.

3. Therapy development and how to get involved in clinical trials

Losmapimod is a tablet developed by Fulcrum Therapeutics. It is a selective inhibitor of p38α/β mitogen-activated protein kinases (MAPK), a class of enzymes involved in acute inflammation and cells' response to injury. Losmapimod was bought by Fulcrum after the company discovered its ability to reduce the activity of the DUX4 gene, the root cause of FSHD, during preclinical studies using patient-derived muscle cells.

Fulcrum Therapeutics is currently conducting phase II trials in North America and Europe, investigating the safety, tolerability, and efficacy of losmapimod to treat the root cause of FSHD1. See: https://clinicaltrials.gov/ct2/show/NCT04003974, https://clinicaltrials.gov/ct2/show/NCT04004000 and https://clinicaltrials.gov/ct2/show/NCT04264442.

Acceleron Pharma announced that they have stopped the development of their drug **ACE-083,** in FSHD.

The company said that as ACE-083 did not show any meaningful functional benefits as well as not being able to grow muscle, that the drug showed no benefit.
ACE-083 was in development in a phase II trial. See the company press release: http://investor.acceleronpharma.com/news-releases/news-release-details/acceleron-announces-topline-results-phase-2-trial-ace-083.

Pre-clinical research

There is also a wide range of pre-clinical research occurring in FSHD, aimed at suppressing the **DUX4** protein. The expression of DUX4 is thought to be responsible for the inflammation and muscle wasting seen in FSHD. The DUX4 protein is encoded by the DUX4 gene.

For further information on potential therapies in development please visit: **https://www.fshd-registry.org/uk/media/images/FSHD_Drug_Development_Pipeline.pdf**

Pre-clinical research is important laboratory work done using cell models and animal models to firstly identify a new drug and to then test it to see if it is safe and effective before being tested in humans. This can take between 1-5 years.

How do I take part in a clinical trial?

You can search the **Be Part of Research** site to find trials relevant to you, and you can contact researchers yourself to potentially get involved - https://bepartofresearch.nihr.ac.uk/. Using this site enables you to search for trials in the UK near to where you live, and based on your condition. Alternatively you can ask your doctor or a patient organisation about any ongoing clinical trials that you may be eligible for.

Clinical trials often involve comparing the effects of one treatment with another and can involve patients, healthy people, or both. To learn more about clinical trials and the process of getting involved in research see: https://www.nhs.uk/conditions/clinical-trials/

4. Previous and upcoming events

In December the registry was presented at the **6th TREAT-NMD International Conference** in Leiden, where there were many academics, patients, carers, patient advocacy organisations, clinical specialists and industry. Following directly on from this meeting, the registry was discussed at the **TREAT-NMD Global Database Oversight Committee (TGDOC)** meeting with registry curators across the globe, to discuss best practice and future plans.

Upcoming events (that will take place online due to COVID-19)

- **27th Annual FSHD Society International Research** – this will take place online on June 25-26th 2020.

Coronavirus: information and advice for people with muscle-wasting conditions

For the latest information and advice on COVID-19, please see the following information provided by Muscular Dystrophy UK: https://www.musculardystrophyuk.org/get-the-right-care-and-support/coronavirus-information-and-advice-for-people-with-muscle-wasting-conditions/

Thank you for being a part of the UK FSHD Patient Registry.

Please remember to log into the registry and update your details and data if you have not already done so within the past 12 months.

If you have any questions, feedback or suggestions or you would like to share your story, please contact below. Also please feel free to promote the registry in any support groups you may be a member of. We are always welcome to more UK patients joining the registry.

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