UK FSHD Patient Registry Newsletter

ISSUE 6
http://www.fshd-registry.org/uk

Accelerating research and improving care in FSHD

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

Inside this Issue:

Welcome to the sixth newsletter of the UK FSHD Patient Registry. We hope you are all staying safe!

As of March 2021 there are **1,035** participants registered with the UK FSHD Patient Registry. This makes the UK registry one of the largest national FSHD registries in the world! A huge thank you is in order for all of the patients/caregivers, clinicians, researchers and patient organisations who have supported and contributed to this 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 (NM) consultant/neurologist (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 NM consultant/neurologist at your next appointment, they should be able to provide you with a copy of the report if you have been tested. **Please remind your NM doctor/neurologist about the registry and updating this aspect of it on your behalf.**

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, and to contact the registry curator for further information.

2. New studies and research updates from the registry

Digital endpoints in neuromuscular disease (NMD) survey – complete ★

An online survey was developed by Parent Project Muscular Dystrophy and a French company, Sysnav.

The aim of the survey was to collect patients, families and caregivers' opinions, expectations and concerns regarding the use of the wearable device, ActiMyo in NMD clinical trials and the clinical meaningfulness of its potential outputs.

128/403 patients who took part had FSHD and were recruited partly through the registry. There were also 8 FSHD caregivers who took part.

Swallowing difficulties in NMD survey - complete ★

An online survey was developed by The National Hospital for Neurology and Neurosurgery and University College London.

The aim of the survey was to understand the experiences and priorities of individuals (and their caregivers) living with an NMD, and swallowing difficulties in the UK.

An update should be provided in 2021.

Genetic Alliance patient and caregiver survey – complete ★

An online survey was developed by Genetic Alliance to understand the experience of the rare disease patient or caregiver, so that they can detect future changes brought about by the new UK Rare Diseases Framework (formerly known as the UK Strategy for Rare Diseases).

A summary of the new UK Rare Diseases Framework can be found here.

Pregnancy and fertility in NMD survey - complete ★

An online survey was developed by Newcastle University.

The aim of the survey was to explore the challenges faced by women with an NMD for conceiving, during pregnancy and delivery, and in caring for young children.

130/453 patients who took part had FSHD and were recruited through the registry. A further update should be provided in 2021.

Sleep in FSHD survey – complete ★

An online survey and sleep diary were developed by the University of Surrey.

The aim of the project was to explore the experience of sleep and factors that influence sleep in individuals with FSHD, using a questionnaire and sleep diary. This is an understudied area of research, particularly in the FSHD population.

84 patients from the registry took part in this study, with an average age of 54 years old. A further update should be provided in 2021.

Voice of the patient survey – complete ★

A series of online surveys were developed by the FSHD Society in preparation for a landmark Voice of the Patient Forum on drug development for FSHD.

The registry contributed to most of the 88 UK patients and caregivers who completed the surveys. The full Voice of the Patient report can be found here.

To see a list of the research projects and clinical trials that the registry has supported since 2013, please visit the new Projects supported section of the website.

3. FSHD drug development

There are currently a number of drugs in development for the treatment of FSHD. There are **5** drugs in clinical trials, and potentially **20** in pre-clinical development.

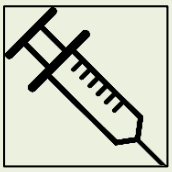
You can find these compiled on the UK FSHD Patient Registry website with further details below on a number of these *(the registry does not endorse any drugs listed).*

Fulcrum Therapeutics – *losmapimod* (phase II)
Fulcrum Therapeutics' phase II ReDUX4 study of losmapimod for FSHD is now complete. Please see: https://clinicaltrials.gov/ct2/show/NCT04003974. The study assessed the safety and efficacy of losmapimod, given as two tablets, twice per day over 48 weeks.

The company has previously announced some early study results suggesting that losmapimod may be most effective in those who have a greater expression of DUX4. Fulcrum Therapeutics plan to report on the full study results in the second quarter of 2021.

What is DUX4?

DUX4 is a toxic protein that researchers have found to be overly expressed in those with FSHD.



Companies close to clinic trials
(click on the company name to go to their website)

Avidity Biosciences

- Developed biological therapy (injection) that aims to target the DUX4 messenger RNA (this is what helps give the DNA instructions to turn into actions).
- Plans to begin clinical trials in **2022**.

Dyne Therapeutics

- Developed biological therapy (injection) that aims to reduce DUX4 expression and increase the distribution of the drug to the muscles.
- Plans to file a drug application in the USA and enter clinical trials **within the next two years**.

Myocea Inc

- Repurposing a small molecule (oral therapy) to target DUX4 via multiple mechanisms. This drug, has previously been studied in other conditions.
- Plans to begin phase I clinical trial in **2022**.

HealX

- Plans to use artificial intelligence (AI) to identify an existing drug to help treat FSHD.
- Using AI to identify existing drugs will reduce the time, cost and risk compared to traditional drug discovery.

Recent UK developments

Dumonceaux laboratory

- Developed a new therapeutic approach to treat FSHD by targeting DUX4, which is toxic and leads to cell damage.
- The new approach works by trapping and stopping DUX4 during its journey inside the cells, thus preventing its toxicity.

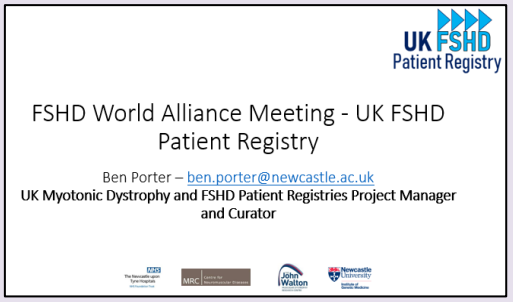
REMINDER - If the registry is used to promote or assist with the recruitment for a clinical trial or research study, all eligible patients will be contacted by the registry curator via email.

4. Previous and upcoming events

In June 2020 - a poster on the registry was presented at the virtual **FSHD Society International Research Congress**. This outlined a summary of some of the registry data and the research impact the registry has had, by highlighting all of the registry enquiries we supported to date. To view this click **here.**

In September 2020 - the registry was invited by the FSHD Society to present at their inaugural **FSHD World Alliance Meeting**.

- Attendees included patient advocacy representatives from across the globe, including countries such as Turkey and Argentina, and nations who did not yet have a national registry for FSHD.
- The aim of the presentation was to provide some insights into how the UK registry functions and works with patients, as well share best practice.
- At the end, a series of useful questions were asked and the overall feedback from the presentation was overwhelmingly positive.



In September 2020 – the registry Steering Committee met for their annual meeting.

- Most members were in attendance (virtually) and this was a very productive meeting.
- We discussed the current registry operations and the plans for 2021. This also involved discussing a vacancy on the Steering Committee which was filled by Consultant Neuromuscular Physiotherapist, **Robert Muni Lofra** who works at the John Walton Muscular Dystrophy Research Centre.
- The registry also welcomes **Dr Emma Matthews**, a Consultant Neurologist at St George's Hospital who joined the Steering Committee earlier in the year too.

In December 2020 - 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.

The **UK Neuromuscular Translational Research Conference 2021** will be held online on 25th and 26th March 2021. The registry plans to present a poster at this meeting.

Upcoming events

The **28th Annual FSHD Society International Research Congress** is currently scheduled for Leiden, the Netherlands on June 24th and 25th 2021. The registry plans to present a poster at this meeting.

The FSHD Society also runs its **FSHD University** webinars on the third Thursday of every month at 18:00pm (GMT). These are online educational webinars covering a variety of topics around physical health, wellness and research.

5. New and future registry developments

In the past 12 months, the registry has made some developments to provide patients with more communications and potentially increased research involvement.


Publications

The registry has been involved in supporting a variety of clinical trials and research studies since 2013. Please find below publications involving the registry, and projects that the registry has supported (updated as of November 2020):

Kulshrestha, R., Emery, N., Faux-Nightingale, A., Willis, T. and Philp, F., (2020). '**Upper limb rehabilitation in facioscapulohumeral dystrophy (FSHD): a patients perspective**'. *medRxiv*.

Banerji, C.R., Cammish, P., Evangelista, T., Zammit, P.S., Straub, V. and Marini-Bettolo, C., (2020). '**Facioscapulohumeral muscular dystrophy 1 patients participating in the UK FSHD registry can be subdivided into 4 patterns of self-reported symptoms**'. *Neuromuscular Disorders*, *30*(4), pp.315-328.

Porter B, Cammish P, Orrell R, Heslop E, Marini-Bettolo C. '**The UK FSHD Patient Registry: A Key Tool in the Facilitation of Clinical Research**'. In: 12th UK Neuromuscular Translational Research Conference. 2019; Newcastle.

The registry website now has a new **Publications** page which includes all of the research publications that have involved the UK FSHD Patient Registry. To date there have been **12** publications.

New developments coming to the registry and registry website:

- List of >25 registry enquiries that the registry has supported since 2013.
- Linkage to the **Newcastle Research Biobank for Rare and Neuromuscular Diseases**. This is a repository that creates a 'bank' of samples including: DNA and cells obtained from blood, urine, saliva, skin, muscle and nerve. Being able to link the registry data anonymously to the biobank could help support future research projects and help us understand more about FSHD.

The registry is also always looking for feedback on its communications, and on what type of information participants on the registry may like to see from the registry in the future.

Top registry tips:

- Remind your NM doctor/neurologist that you are on the registry if they have not entered your genetic information on there.
- Ask your doctor to contact the registry if they are not listed on there.
- Remember to log in every 12 months in case any of your details have changed (this includes your contact details too).

Please feel free to share information about the registry with anyone you may know who has FSHD. Thank you very much for being a part of the UK FSHD Patient Registry and please take care during these difficult times.

E-mail: fshdregistry@treat-nmd.org

Telephone: 0191 241 8640 (we are still working from home so it would be preferable to email us)

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