



Our Mission & Vision: Together with key stakeholders and active players in FSHD, accelerate therapy development and ensure speedy access to treatments. Become the coordinating group for driving FSHD strategy in the UK.

Background and Development

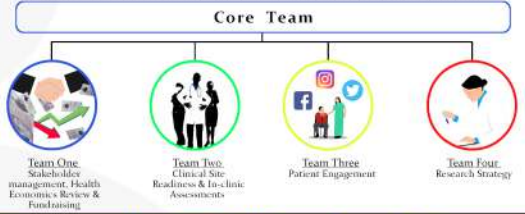
The UK has an excellent reputation for high quality FSHD clinical services, research teams and an established Patient Registry. However, these accomplished UK centres were mainly working in their own areas without an overarching strategy or central co-ordination.

In July 2021 FSHD UK was formed with representation from key stakeholder groups; clinicians, clinician support teams, patients, UK FSHD Registry, research scientists, MDUK and the FSHD Society. Our mission is to ready the UK for clinical trials and to become the recognised coordinating group for FSHD in the UK.

Our Team Set-Up

Our team now with over 30 members span the key stakeholder groups working together to drive FSHD priorities in the UK. The core team members form the key decision-making group with an extended support team. There is a dotted connection to external stakeholders including industry and UK regulatory authorities who play an instrumental role in our success.

We have four teams with an overarching Core Team. Our plans are driven by the priorities/gaps and opportunities identified by the members. Since July 2021 our key achievements have been a direct reflection of our team purpose and more importantly each teams' own areas of focus. This model has been a key to our success!



FSHD UK - Our Key Achievements and Plans

- July 2021 FSHD UK Launched
- 3 Sites, Team of 14, connection to 368 Patients
- August - Site Surveys for gaps/opportunities started
- September - Established regular Stakeholder Engagement with Pharmaceuticals: Fulcrum Therapeutics, Dyne Therapeutics, Avidity Biosciences, Myocera Inc, Fascio Therapies, Healex
- FSHD UK joins World FSHD Alliance
- November - In-Clinic Assessments Designed
- December - Engagement with Clinical Trial Research Network
- December - FSHD UK Grows to 5 Sites, Team of 27 connection to c700
- In-Clinic Assessment launched with training for 5 Sites
- First Meeting of Research Strategy Group – CDA started
- Commence on-going engagement with Regulatory Bodies in UK –Medicines and Healthcare products Regulatory Agency (MHRA) and an introductory meeting with National Institute for Health and Care Excellence (NICE)
- Spring - Logo and Social media platform launch
- Socialized European Patient Pharma Project - 30% UK responses
- November - Patients@Site Engagement initiative (at St Georges)
- Site 6 joins FSHD UK – total connection to c800 patients
- Health Economics Study (cost/benefit) started
- ReSOLVE and MOVE underway at 2 sites
- Novel therapies at 2 Sites
- International collaboration on In-clinic assessments
- FSHD UK Network Database initiated
- FSHD UK joins Project Mercury
- Launch HEOR (Cost/Benefit) study
- Continue with Patients@Site Engagement
- Formalize FSHD UK and commence Fundraising for clinical and IT infrastructure projects
- Launch FSHD UK website / Start on FSHD UK Patients, Clinicians & Research Information Platform
- Launch our Research Strategy & Collaboration Group

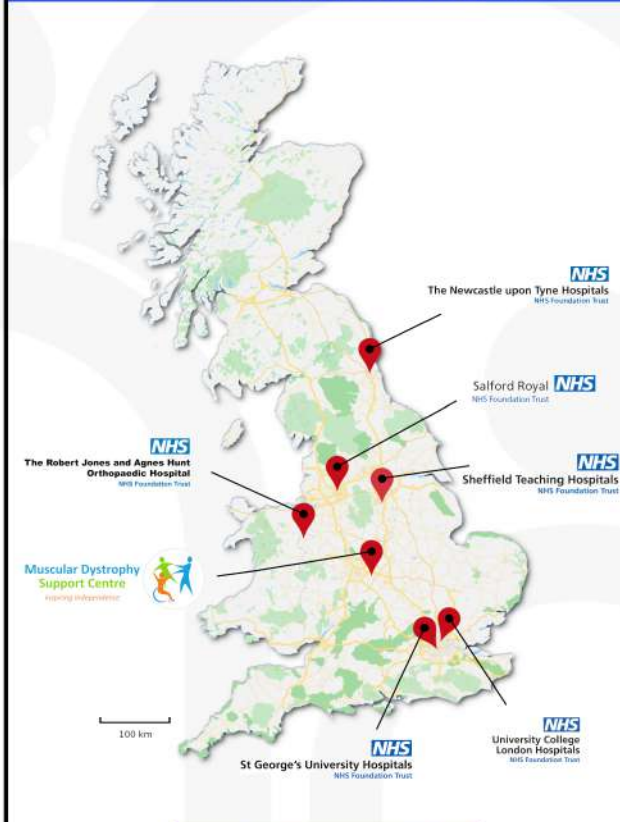
FSHD UK and Social Media

In combination with the UK FSHD Registry FSHD socialized the European Patient Survey in Spring 2022. The UK Response was c.30% of total responses.

Our key aim is engage the UK FSHD patients, carers and families to become more aware of FSHD clinical and research activities.



FSHD UK Sites & Connection to Patients



Location	Adult
Newcastle	200
UCL London	200
St Georges (London South)	150
Salford	76
Sheffield	70
Oswestry	50
Total @ Sites	746
+ MDSC Coventry	82
Total Connections	828

FSHD UK Core Team Collaborators

- Dr Enrico Bugiardini, PhD**
Consultant Neurologist & Clinical Research Associate
National Hospital for Neurology and Neurosurgery - UCL Queen Square Institute of Neurology, London & Southern England
- Professor. Tracey A Willis**
Consultant Paediatric Neurologist
Visiting Professor Chester University
The Robert Jones and Agnes Hunt Orthopaedic Hospital, Oswestry
- Dr James B Lilleker**
Consultant Neurologist and Honorary Senior Lecturer
Manchester Centre for Clinical Neurosciences, Northern Care Alliance NHS Foundation Trust.
- Professor. Giorgio Tasca**
Clinical Professor of Neuromuscular Science
John Walton Muscular Dystrophy Research Centre, Newcastle, North England
- Dr Emma Matthews, PhD, FRCP.**
Reader of Neurology and Honorary Consultant Neurologist, St Georges University of London – London and Southern England
- Dr Channa Hewamadduma**
Consultant neurologist and honorary Senior lecturer
Sheffield Institute for Translational Neurosciences (SITRAN), University of Sheffield, Sheffield.
- Ruth Hereford**
Chair of Trustees,
Muscular Dystrophy Support Centre
Coventry, Midlands
- Professor. Julie Dumonceaux**
Professorial Research Fellow
Developmental Neurosciences Dept.
GOS Institute of Child Health, UCL, London
- Professor. Peter. S. Zammit**
Randall Centre for Cell and Molecular Biophysics,
King's College London.
- Helen Walker**
UK FSHD Patient Registry Manager and Curator
The John Walton Muscular Dystrophy Research Centre, Translational and Clinical Research Institute, Newcastle University.

FSHD UK Standardised In-Clinic Assessment

In January 2022, Dr Meredith James and Dr Robert Muni-Lofra launched standardised In-Clinic Assessment involving Physio/Occupational Therapists at our 6 sites.

Further collaboration with Europe and USA (FSHD Society) is planned with a view to creating standardised In-clinic assessments across other areas.

FSHD UK Patient Engagement

Special Invitation for FSHD Patients @ St George's!

Take a leaf from the FSHD Society Chapter idea, we have set up Patients Engagement@Site, starting with St George's Hospital, London.

The aim is to roll this out to other sites -another session is being planned for the Autumn 2023 at a different site.

FSHD UK Information Platform - Concept/Vision

A proposal for an FSHD UK Network database has requested funding. This forms part of a longer-term vision to help move from a fragmented to a consolidated view of FSHD patient, clinical and research information needs to support an anonymised, aggregated view of FSHD data for patient/clinical care and research. Site services and facilities information could be managed here for clinical trial readiness.

FSHD CARIS
FSHD - Clinical and Research/Registry Information System (Patient Registry), In-clinic Assessments, Research Data)
Aggregated, Anonymous, GDPR, ETHICed