

World FSHD Alliance annual leadership summit

Working to advance clinical trial readiness and access to treatments for FSHD

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Ricardo Gerpe from FSHD Spain addresses colleagues attending the annual World FSHD Alliance leadership summit.

The World FSHD Alliance, a network of two dozen national patient groups and advocacy organizations for facioscapulohumeral muscular dystrophy (FSHD), held its fifth annual leadership summit on June 17-18, 2023, in Milan, Italy. Forty in-person and nine virtual delegates from 19 organizations attended the meeting. They share a mission to support patients and speed up the development of treatments for FSHD by:

- educating, supporting, and activating people with FSHD;
- finding and engaging clinicians who are interested in providing the best care for patients;
- working with national registries and supporting research;
- advocating for patients with their governments, healthcare agencies, insurance, and policymakers.

As drug development has advanced rapidly in the past several years, Alliance members also recognized they need a global strategy of coordination and collaboration to ensure that there are enough “trial-ready” patients and clinical trial sites. Some members are actively working to develop trial infrastructure, genetic testing, and patient registries in countries such as Japan, Brazil, and Israel, that might not other-




wise be on the radar for clinical trial sponsors.

With the prospect of new treatments coming on the market in the US and EU over the next few years, the Alliance is also discussing country-by-country strategies to advocate for patients everywhere to have access to treatments.

In response to these emerging challenges, several World FSHD Alliance members proposed a new global initiative called Project Mercury (see story on page 4) to accelerate trial readiness and ensure widespread access to treatments. This was, appropriately, a focus of this year’s Alliance leadership summit.

“In a rare disease like FSHD, patients, expertise, and resources are scarce and precious,” said Mark Stone, CEO of the FSHD Society, who serves as chair of the Project Mercury Global Task Force. “To face the urgent challenges in clinical trials and treatment access, we must bring all stakeholders together in an effective, collaborative, and transparent network. Solutions to the challenges we face require global collaboration, led by the patient advocacy leaders of the World FSHD Alliance.”

For more information about the World FSHD Alliance, visit fsbdsociety.org/connect/worldfsbdalliance/. 

World FSHD Alliance 2023 updates

The World FSHD Alliance officially launched in 2019 in Marseille, France. Since then, members have met regularly to discuss and implement strategies to build and strengthen stakeholder networks and work toward clinical trial readiness. Here is a snapshot of just a few of the members. For more information, visit fshdsociety.org/connect/worldfshdalliance/.

FSHD UK

- Guidance team of 30 stakeholders
- Six clinical sites, reach of approximately 800 patients (2023)
- ReSOLVE and MOVE studies underway at two sites
- Two clinical trials starting at two sites
- Started standardized in-clinic assessments across the network
- Health Economics study ready to launch
- Ongoing engagement with FSHD pharmaceuticals
- Initiated engagement with UK regulatory groups
- Socializing via FSHD UK-European Patient Survey and other FSHD initiatives

FSHD Israel

- Running in-person and Zoom meetings
- Growing a network of people with FSHD
- Engaged a clinician who has started Israel's first FSHD center
- Launched website
- Discussing a registry for Israel

ABRAFEU (Brazil)

- First international conference on FSHD in Brazil (on Zoom)
- Clear strategy for trial readiness
- Country-wide network for genetic testing
- Affordable genetic test
- Patient registry mobile app
- A total of 700 patients registered, more than 150 genetically tested
- Collaboration of physiotherapists with Radboud University, the Netherlands
- A scientific research team in the largest hospital with the highest concentration of people in Brazil
- Memorandum signed to join MOVE study
- Identified MRI facility for studies and trials

Aims for 2024-2025:

- Activate MOVE study
- Join FSHD Clinical Trial Research Network
- Engage with pharmaceutical companies about clinical trial sites in Brazil
- Engage with Project Mercury
- Develop funding sources



UILDM-FSHD (Italy)

- Established a group focused on FSHD within UILDM
- Collaborated on EU-funded EPIthe4FSHD scientific research and programs advocating for more communication about FSHD in Italy and internationally
- Entered into mutual support and communication agreement with FSHD Society
- Launched the first Italian-English FSHD website (epithe4fshd.org)
- UILDM-FSHD patients collaborated with newly announced FSHD neurologist group led by Dr. Massimiliano Filosto
- Conducted two webinars with more than 130 patients attending on FSHD diagnosis and care, research at NeMo centers, upcoming Fulcrum trial, future research landscape
- Published the first UILDM-FSHD Society newsletter for Italian FSHD patients
- Encouraged Italian patients to register on the FSHD Society website

Aims for 2024-2025:

- Formalize our organization as it matures
- Continue to seek funding for MOVE for other sites
- Advance in-clinic assessments internationally
- Commence fundraising to support infrastructure, IT, and network projects
- Launch our research strategy/collaboration group
- Continue with Patients@Site
- Continue engaging with pharmaceuticals and regulatory authorities
- Get our website up and running
- As part of Project Mercury, become fully clinical trial ready with patients, clinicians, sites, processes, infrastructure, and more
- Patients@Site Engagement initiative

FSHD Japan

- Encouraged more core members to better organize the community
- Held online meetings with key clinicians and strengthened relationships
- Set regular online events targeting patients and families to support education and build a stronger community

Aims for 2024-2025:

- Communicate with pharmaceutical companies about clinical trials
- Avoid delays in drug access by collaborating with other neuromuscular disease groups
- Promote development of domestic drug candidates

FSHD China

- Organized first-ever meetings for FSHD community
- Online courses for FSHD community
- Voice of the Patient report on Chinese patients
- Facilitating development of new genetic test
- Building story platform for rare disease patients
- Outreach to companies for future trials in China

FSHD India

- Growing a network of patients in India on WhatsApp
- Identifying and addressing patients' needs
- Engaging with genetic testing labs
- Engaging with clinicians who treat FSHD patients
- Building a website

- Publicized FSHD IRC and World FSHD Alliance meetings widely throughout Italy, with EPIthe4FSHD facilitating transportation of international guests and Italian patients

Aims for 2024-2025:

- Integrate FSHD individuals across UILDM's political and social activities
- Communicate about upcoming trials and future access to treatments
- Encourage FSHD patients and caregivers to participate in scientific meetings (starting from the FSHD IRC in Milan)
- Extend network of FSHD patients in Italy
- Continue to develop webinars, meetings, media communications, and scientific publications for Italian patients, researchers, and clinicians

