Funded by
the European Union

European
Reference
Network
for rare or low prevalence
complex diseases

Network
Neuromuscular
Diseases (ERN EURO-NMD)

8th ERN EURO-NMD
Annual Meeting

FSHD Europe & FSHD European Trial Network (ETN)

5th – 7th March 2025

Nicol Voermans & Ria de Haas

Radboudumc, Nijmegen, The Netherlands

The ERN EURO-NMD is funded by the European Commission under the EU4Health programme (EURO-NMD 23-27 — 101156434 — EU4H-2023-ERN2-IBA)

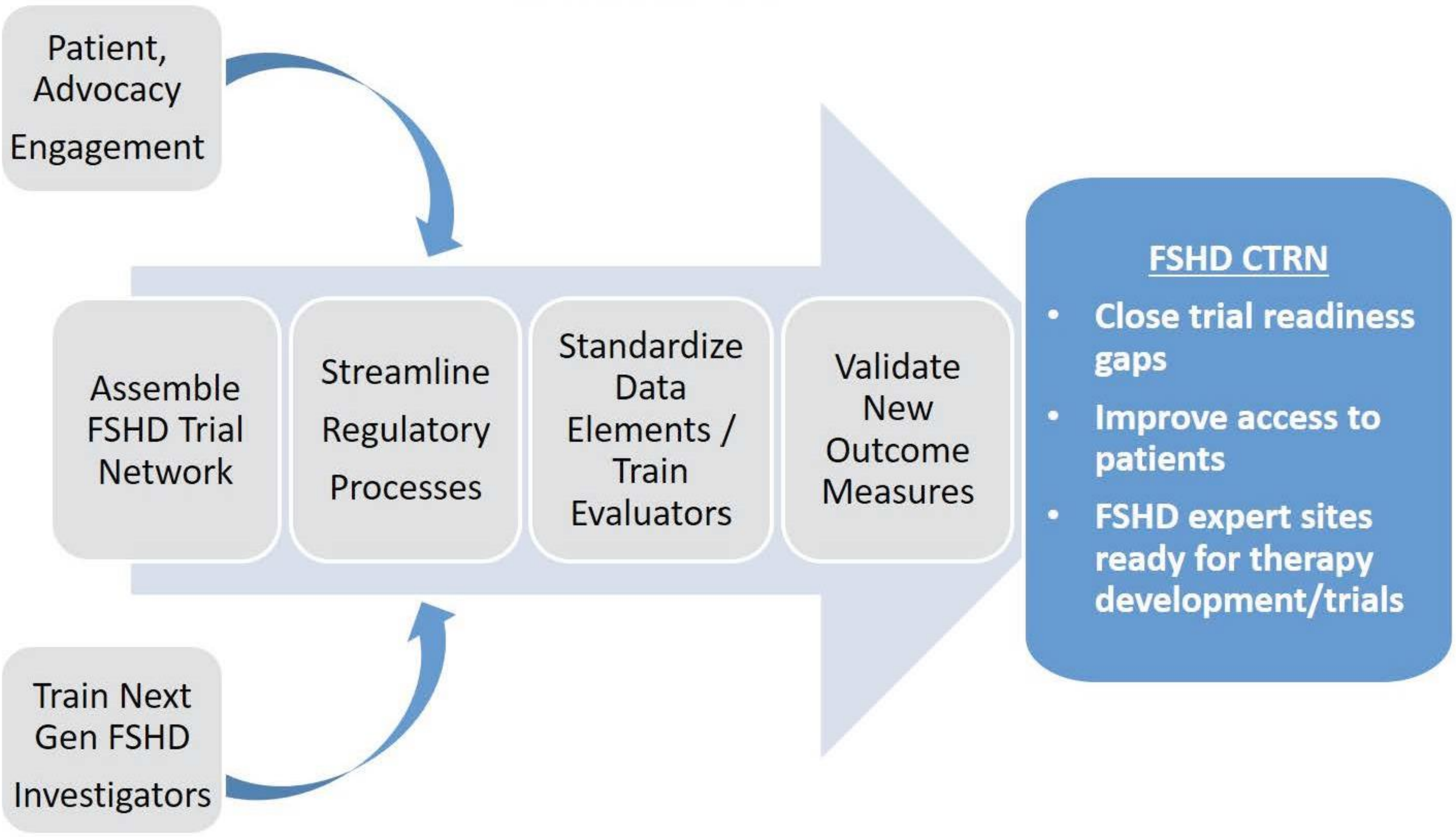
FSHD Europe started

2010

Launch
7 organisations

‘A united, consistent patient voice
for FSHD in Europe’

FSHD Society’s steps forward



Patient, Advocacy Engagement

Assemble FSHD Trial Network

Streamline Regulatory Processes

Standardize Data Elements / Train Evaluators

Validate New Outcome Measures

Train Next Gen FSHD Investigators

FSHD CTRN

- Close trial readiness gaps
- Improve access to patients
- FSHD expert sites ready for therapy development/trials

FSHD SOCIETY

FSHD CTRN
FSHD CLINICAL TRIAL RESEARCH NETWORK

World FSHD Alliance
STRONGER TOGETHER

FSHD europe

Europe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- 748 million inhabitants
  (11 % world populations)
- FSHD prevalence rate 12/100,000 (Nl)
  → 90,000 European FSHD patients
- 44 Countries
- 27 EU partners
- 200 Languages (24 official)

Europe

Differences Europe - USA:

- Guidelines for clinical trials
- Pharma regulation and participation
- Health care provisions
- Supporting organizations

EUROPEAN NEURO MUSCULAR CENTRE

European Reference Network | Neuromuscular Diseases (ERN EURO-NMD)

EURORDIS RARE DISEASES EUROPE

FSHD europe

FSHD Europe developed

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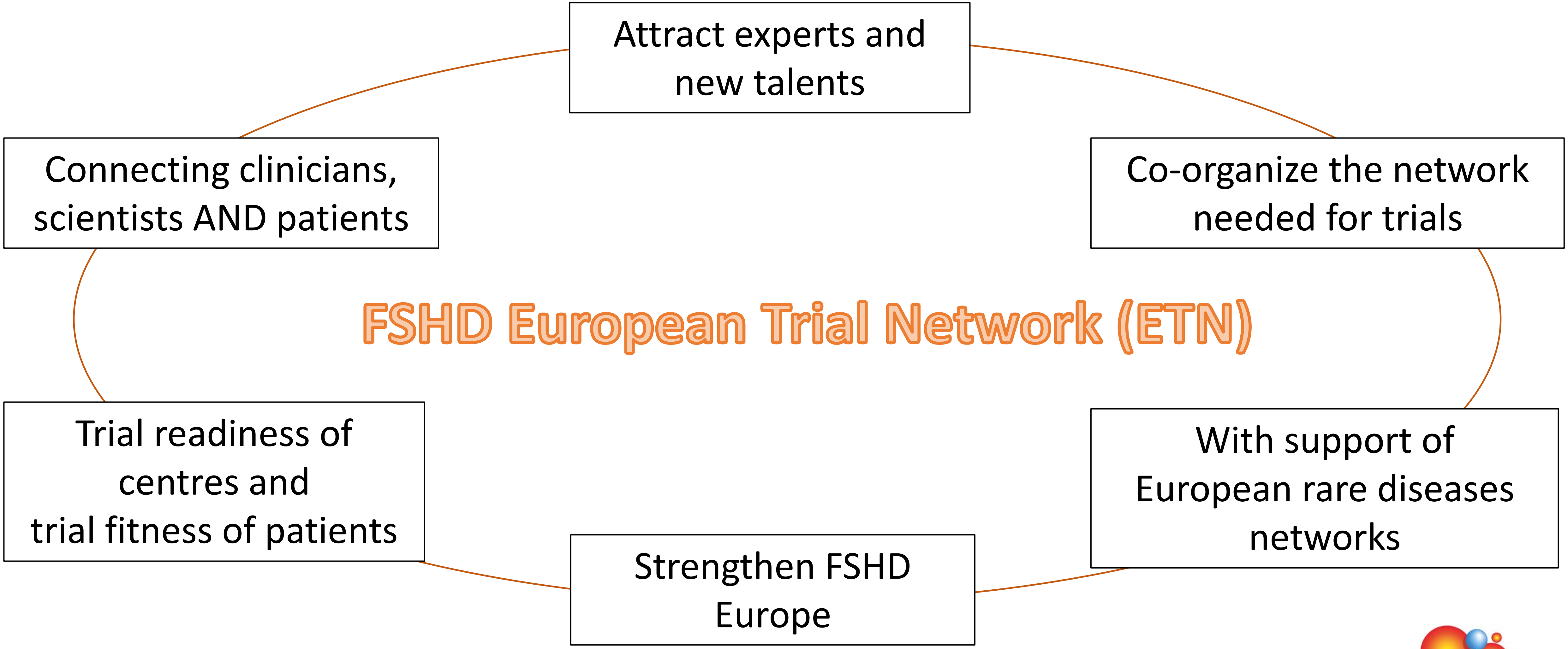
2021

FSHD European Trial Network

‘Connecting the clinicians and researchers in Europe’

FSHD European Trial Network



FSHD European Trial Network (ETN)

Attract experts and new talents

Co-organize the network needed for trials

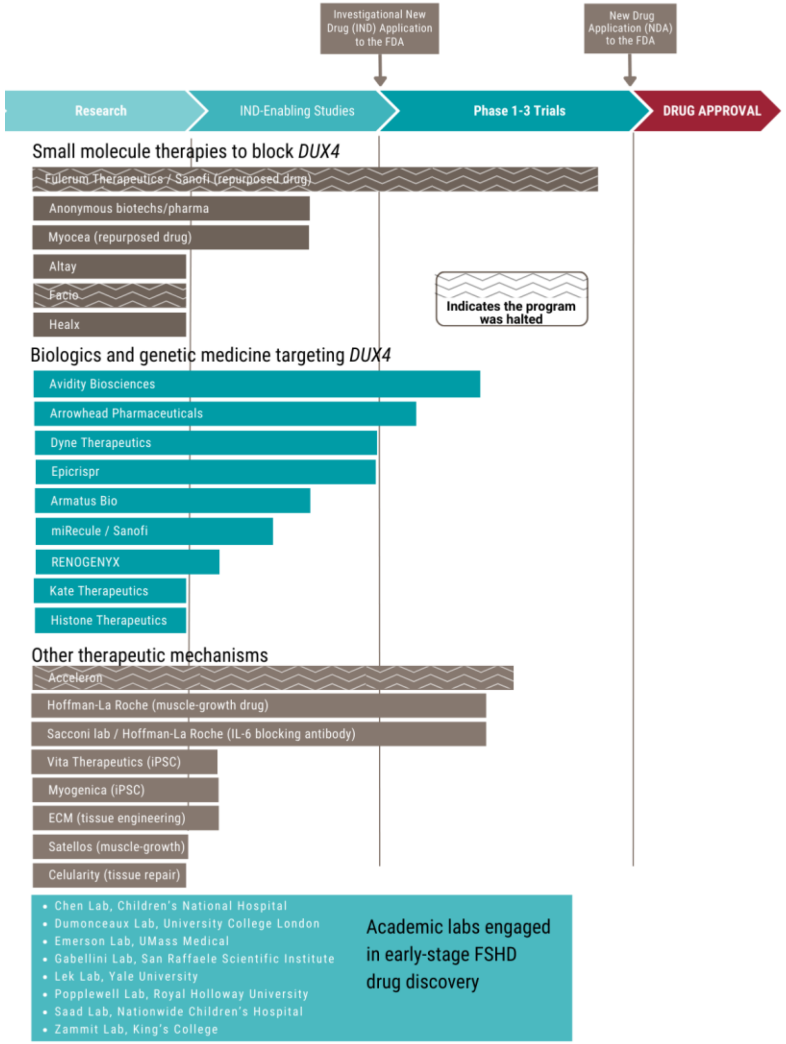
With support of European rare diseases networks

Strengthen FSHD Europe

Trial readiness of centres and trial fitness of patients

Connecting clinicians, scientists AND patients

Drug development pipeline



Drug Development Pipeline | FSHD Society

- Currently no treatment for FSHD
- Growing numbers of companies and academic laboratories are pressing forward with early-stage drug development efforts.
- Clinical trials ongoing

A Study to Evaluate RO7204239 in Participants with Facioscapulohumeral Muscular Dystrophy

A Phase 1/2 Trial of ARO-DUX4

Motor Outcomes to Validate Evaluations in FSHD (MOVE FSHD)

FORTITUDE

Study of safety and efficacy of RNA therapy to block DUX4

MANOEUVRE

Phase 2 study of safety and to test if drug can help muscles grow

Arrowhead

Study of safety and dosage of RNA therapeutic to block DUX4

MOVE & MOVE+

Natural history study to help improve future clinical trials

Current Trials and Studies | FSHD Society

Challenges ahead

- Clinical Trial Readiness
- Clinical Trial Infrastructure
- Patient involvement, engagement, participation and availability
- Equal access to approved therapies
- Treatment Readiness

FSHD Europe

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2021

FSHD European Trial Network

‘Connecting the clinicians and researchers in Europe’

2024

FSHD Europe in Project Mercury

‘Collaborating in global collaboration PROJECT MERCURY’

Project Mercury

A **collaboration program led by FSHD patient advocacy organizations** in partnership with experts, biopharma companies and researchers – all networked together.

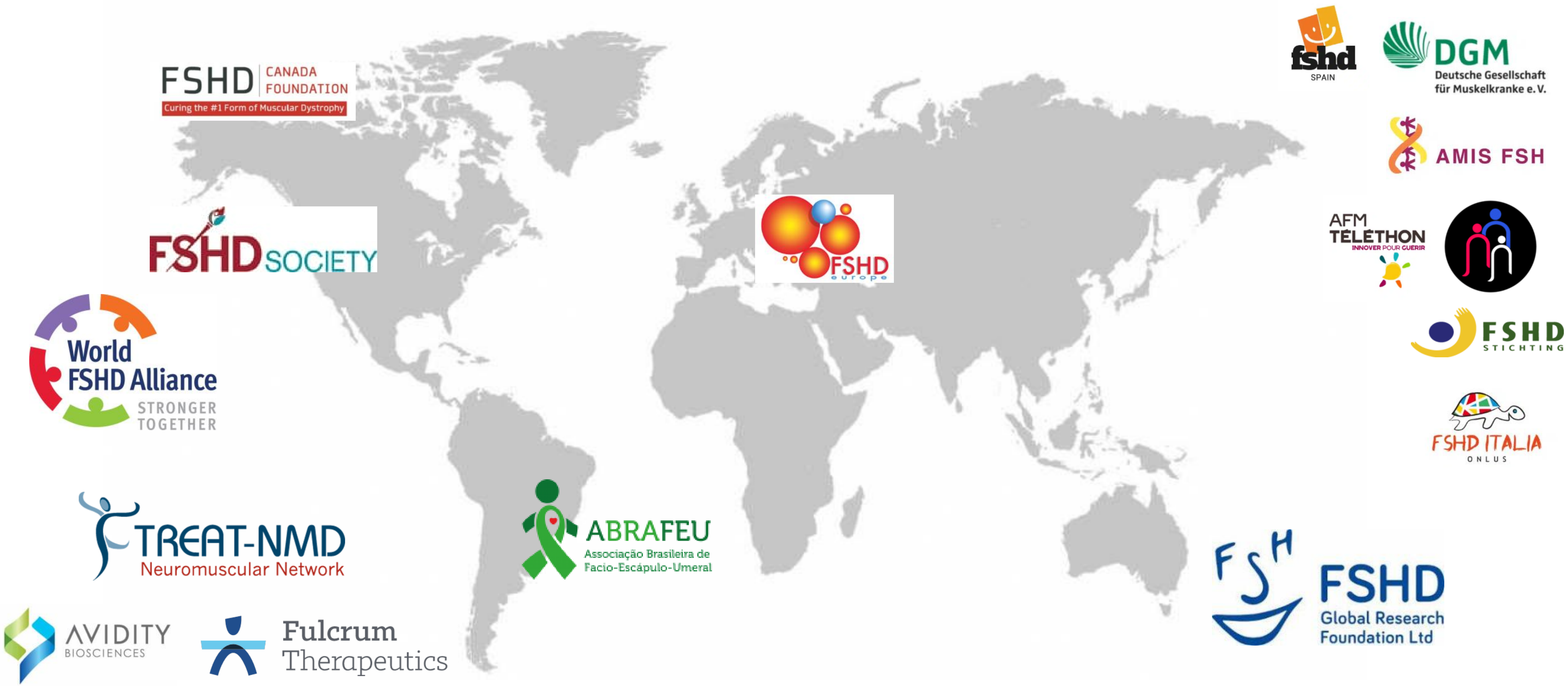
The initiative is focused on **solving specific challenges that slow or stop the development and delivery of therapies** to people affected by FSHD around the world.

PROJECT MERCURY

The Global Initiative to Speed the Delivery of Therapies for FSHD

FSHD europe

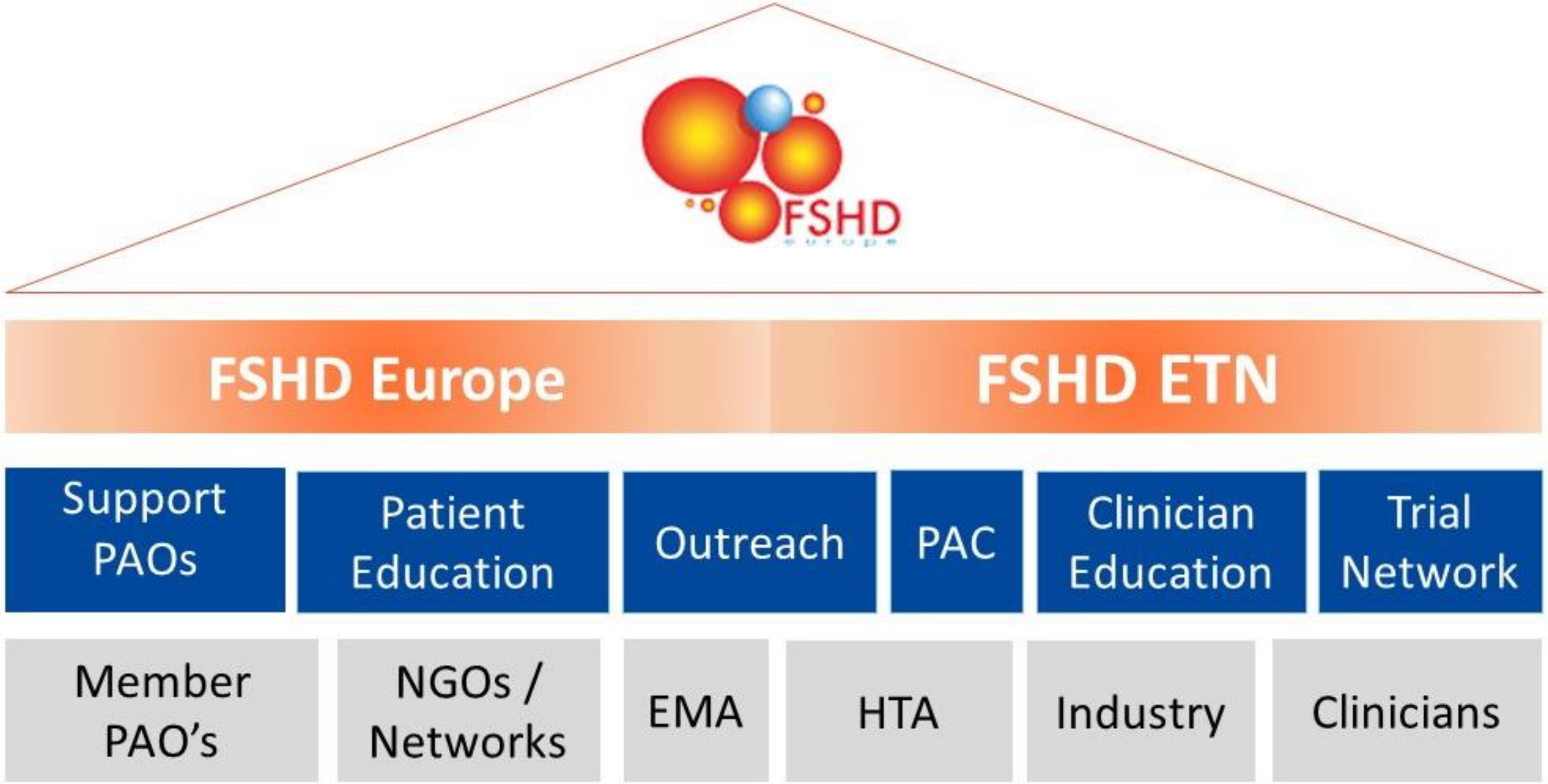
Project Mercury



FSHD Europe

To improve the quality of life for all patients and their families,
and facilitate diagnosis of FSHD,
and enable all patients have access to the best treatment.

Organizational structure



FSHD Europe

FSHD ETN

Programs

- Support PAOs
- Patient Education
- Outreach
- PAC
- Clinician Education
- Trial Network

Stakeholders

- Member PAO's
- NGOs / Networks
- EMA
- HTA
- Industry
- Clinicians

FSHD European Trial Network (ETN)

FSHD Europe established the FSHD European Trial Network (ETN) to **support collaboration to reach trial readiness**

WG1: **Clinical and genetic diagnosis**
(Richard Lemmers & Emiliano Giardina)

WG2: **Clinical outcome measures**
(Enrico Bugiardini & Elena Carraro)

WG3: **Biomarkers**
(Julie Dumonceaux & Yann Pereon)

WG4: **Imaging outcome measures**
(Giorgio Tasca & Mauro Monforte)

WG5: **Pediatrics**
(Katy de Valle & Tracey Willis)

FSHD CTRN
FSHD CLINICAL TRIAL RESEARCH NETWORK

EUROPEAN
NEURO
MUSCULAR
CENTRE

FSHD
europe

Program overview

- Support PAOs
- Education
- Patient Registries
- Care/ QoL
- Clinical Trial Network
- Patient Advisory Council (PAC)
- Governance
- Sustainability
- Outreach

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Patient registries

Increase number of patients in registries

- Patient surveys to identify facilitators and barriers to participation
- Inform and educate patients
- Support development of new patient registries

Clinical Trial Network

Enhance Your Site's Visibility: Register with the Clinical Trial Site Registry (CTSR)

CTSR provides critical site-level information:
facilities, equipment, personnel, trial experience, and aggregated patient data

Benefits of Registration

✓ For Healthcare Providers:
- Increased visibility
- Participation in clinical trials and research collaborations
- Join international network of specialized neuromuscular disease centers

✓ For Industry & Researchers:
- A single point of access
- Access to a trusted network of specialized trial sites
- Improved recruitment

CTSR
Care and Trial Site Registry

Clinician Education

- EURO-NMD Webinar series on FSHD

When	Topic	Presenter
Thursday May 8th 4pm-5pm CET	Genetic Diagnosis	Emiliano Giardina
Thursday May 15th 4pm-5pm CET	International Clinical Care Guidelines	Teresinha Evangelista
Thursday May 22nd 4pm-5pm CET	Pediatric FSHD	Tracey Willis
Thursday May 28th 4pm-5pm CET	Clinical trial readiness	Nicol Voermans

- Masterclass (in person) 2026, in collaboration with TREAT-NMD, TBD

Clinician and Patient Education

[International Research Congress | FSHD Society]

WORLD
FSHD
DAY
Unite to find a cure

JUNE 20th

2025
FSHD
CONNECT
EUROPE

JUNE 13-15, 2025, Amsterdam

[FSHD Europe - The European Voice for People with FSHD (fshd-europe.info)]

Take home messages

- **Support collaboration:** FSHD Europe newsletter www.fshd-europe.info
- **Enhance Your Site's Visibility**: Register with the Clinical Trial Site Registry (CTSR)
- **Increase patient engagement**: Inform patients about registries and trial participation
- **Strengthening FSHD Europe**: Increase number of member organizations
- **Clinician and Patient Education**: EURO-NMD Webinar series and IRC and FSHD Connect

Questions?

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