

# 2021 Annual Summit Member action plans 2021-2022



**World FSHD Alliance**

annual meeting 2021

# FSHD Europe



## **Done in 2020/2021 :**

- First 2 meetings of European FSHD trial readiness
- General Assembly scheduled in France in april 2020 cancelled because of Covid
- Welcomed new member : FSHD Italia

## **Will do in 2021/2022 :**

- Support activity of FSHD European trial network with Dr Nicol Voermans, participate to working groups
- Realize a patient survey on their expectations on clinical trails
- Revise statuses and write Rules of Governance to comply to new dutch law on associations
- Look for new members (associations) and new volunteers (persons)
- Develop presence on the web (website and social network)



# 2021 Annual Summit



FSHD Society Action Plan 2021-2022

Revenue goal \$3.9 million

## Patient Education and Engagement

- Continue to grow international chapter program
  - Now 33, including Early-onset Chapter
- Volunteer-led fundraisers 2021 goal: \$800,000
- Increase the number of confirmed FSHD patients in our database
- 6 FSHD 360 conferences/year
- Monthly FSHD University webinars and FSHD Radio shows
- International research contact registry

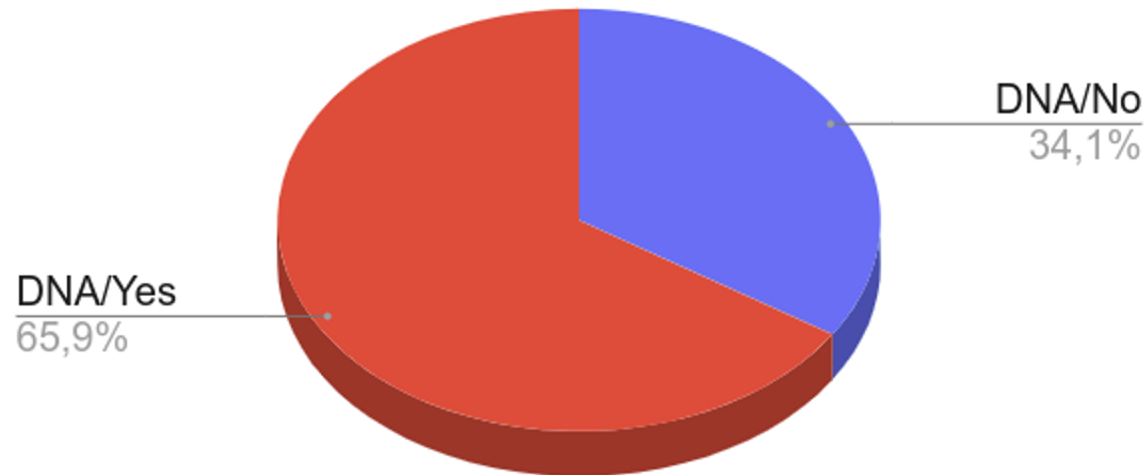
## Research & Therapeutic Accelerator

- Expand Clinical Trial Research Network (CTRN) with 3 new EU sites
- Industry collaborative workshops
- Biomarker and imaging marker initiatives
- Genetic testing initiative
- FSHD Masterclass for medical professionals
- International care considerations guide
- Health economics study of FSHD

## FSHD Argentina Action Plan 2021-2022

- FSHD webinar for World FSHD Day organized in conjunction among **Argentina, Chile and Spain.**
- Following **Brazil's** initiative we'll start trying a cheap and fast saliva DNA test.

FSHD Argentina - Total Members 89





# 2021 Annual Summit



## FSHD Global Research Foundation Action Plan 2021-2022

- **Medical Research**

- ✓ While we continue to fund global medical research, we invest into FSHD biotech, and support scientists that wish to commercialize their FSHD technology.
- ✓ We have advanced our focus towards areas of muscle wellness and muscle technology such as 3D muscle printing.
- ✓ Establishing Australia's first FSHD fully characterized tissue cell bank which will complement data registry and provide a library of cell tissue which can be shared globally for medical research
- ✓ Launched the FSHD Medical Education Portal ([www.fshdmedicalportal.org](http://www.fshdmedicalportal.org))
  - Offers a complimentary saliva based DNA methylation test for Australians (collaboration with Peter Jones). Australians can donate their results to the HIPPA compliant Portal (FSHD Global Registry). The Portal also offers a directory of services e.g.: allied health, medical (GPs, Neurologists, Surgeons), accommodation, mental health who have experience with FSHD patients. Connect communities to avenues of support

- **Government Support**

- ✓ Australia considers the Southern Blot as the gold standard diagnostic test for FSHD. FSHD Global is in conversations with Federal Government and Australia's diagnostic clinic to replace with new diagnostic technologies such as DNA Molecular Combing, Nanopore Long Sequencing, Bionano Optical Mapping etc.
- ✓ On 2<sup>nd</sup> July Prime Minister of Australia and Health cabinet will meet with Bill and Natalie Moss to form a plan on how Australia can improve its diagnostics and become a site for clinical trials. **What is this forum's opinion of what type of diagnostic test/technology we should encourage Australia to embrace? Is there formal International gold standard test to replace Southern Blot?**

- **Community**

- ✓ We have encouraged all Australians living with FSHD to write to their local government representative to put pressure on the Federal Government to take notice of the community and action support, services, education, awareness, funding for FSHD. This has been successful. This has helped open conversations with Federal Government around Australia formally embracing World FSHD Day.

## Brazilian Association of Facioscapulohumeral Muscular Dystrophy

2021



**DNA**



SERVING PATIENTS

MyFSHD

Introduce a new saliva test the Peter and Takako Jones Lab at an affordable cost will permit us to give direction and initiate our task to create data for all patients here in Brazil.



We are promoting a project to create a patient and family history database. Together with 19 Universities and Medical Centers with MyFSHD in Nevada:

- 1- Unifesp - SP - With the team of Dr. Acary B de Oliveira
- 2- Universities and Medical Centers - 17 distributed in Brazil
- 3- Genome of the University of São Paulo - With the team of Dr. Mayana Zatz

genoma

2022



**Register**



RESEARCH

To qualify for DNA testing by Abrafeu, we will have an international standard questionnaire to create the family registries.

We plan to create patient registries on a large scale to get the Natural history of the region. We will develop donation campaigns to test people who cannot afford the test.

2023

**Clinical Trial**

By the end of 2022, we believe we will have a patient registry to study the possibility of requesting a Clinical Trial to start the certification process of the treatment in Brazil.

Project to disseminate a standard protocol for treating FSHD to physical and occupational therapists throughout Brazil.



**Standardized Protocol**



SERVING PATIENTS

INSTITUTO  
VITA  
pesquisa e performance

Develop a physiotherapy treatment for FSHD based on international protocols to training all physiotherapists interested in Brazil. Introduce metrics standards protocols, using technology for distance assessment to better measure the evolution. The main goal is to bring the standardization treatment protocols for FSHD to treat all patients anywhere in Brazil.

Radboudumc

Together with Vita, one largest physical therapy institute in Brazil is evaluating if there might be a possibility to establish a research collaboration with an existing PT research program with Radboud university. The main goal are to use the same protocol and training to run a cohort in Brazil of FSHD patients to be part of a more extensive study. (Dra. Nicole Voet)



# World FSHD Alliance 2021 Annual Summit

## FSHD-China Action Plan 2021-2022



- Action 1: Improve patient registration and encourage patients to complete patient real-name registration.
- Action 2: Increasing the number of genetic tests, plans to cooperate with genetic testing companies and hospitals, so that patients can afford the cost of testing.
- Action 3: In terms of helping patients to solve their fertility needs, we will further promote the necessity of prenatal testing and PGT, and cooperate with hospitals to reduce or waive related expenses.
- Action 4: A documentary is planned to introduce the FSHD disease and the stories of FSHD patients.
- Action 5: It is planned to launch a long-term online class {20 sessions}, inviting neurology, rehabilitation, and psychologists to explain professional knowledge to help patients with FSHD, so that patients can better understand the disease and face the disease correctly.



**FSHD-CHINA**  
PATIENT ADVOCACY AND SUPPORT ORGANIZATION  
你并不孤单 FSHD 患者关爱组织





**World FSHD Alliance**

annual meeting 2021

# AFM-Téléthon France FSHD group



## **Done in 2020/2021 :**

- 4 family information in video-conference
- 1 newsletter
- New blog / website
- We lost our friend and colleague Marie-Martine on June 2

## **Will do in 2021/2022 :**

- More family information in video-conference and in presence
- Try and "hire" more volunteers
- Continue "routine" work (answer patient's questions, participate to french registry, etc.)



## Organization AMIS FSH Action Plan 2021-2022

- With a young company named KONDREE we are working on the development and distribution of software based on the work of Professor Dalila Laoudj-Chenivresse from the Montpellier University Hospital (France) to fight against oxidative-stress in our muscular cells. With this software, the aim is to allow doctors to prescribe supplementation (Vitamin C and E, Zinc, Selenium and copper) personalized from a biological blood test.
- Continue to fund seed money on innovative research and help to develop the consideration of FSHD in countries where it is unknown



# 2021 Annual Summit

## German FSHD Diagnosis Group within the DGM e. V.



### Aims and Activities 2021/22:

- expanding the German FSHD patient registry (currently 640 registered patients)
- start of two virtual discussion groups in November 2020 (FSHDers, relatives), which take place regularly once a month
- joint organisation of a 3-day patient day for two diagnostic groups (FSHD and Myositis) in September with many lectures and workshops on the topics of diagnostics, sports/physiotherapy, ventilation, swallowing difficulties, palliative medicine
- in May 2022, a large scientific symposium with international researchers and clinicians working in the field of FSHD from the USA, France, the Netherlands and Germany (originally planned for 2020, but postponed to 2021 and now 2022 due to the global pandemic)
- continue to inform, educate and raise awareness in society about FSHD
- continue with our regularly podcast ("FSHD-LIFE")

# FSHD India

## Background:

There are many organisations working on DMD, LGMD and Muscular Dystrophy in general. A need was felt to create a community group specific to people with FSHD.

‘FSHD India’ was then conceptualised with the help and motivation from June Kinoshita. It is managed by four members who are part of the FSHD community. Main aim of the organisation is to get the benefit of synergies from various organisations working globally and make lives of people with FSHD better.

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### Objectives of the organization:

FSHD India is determined to provide end to end support to FSHD community members and plan to do so in two stages.

#### Stage 1:

Non financial. Only information is researched and circulated to the community once verified. This will include various details like list of doctors available, results of clinical trials, etc

#### Stage 2:

**Financial** – Organisation will get involved in activities where money needs to be invested.

### Targets for Next one Year

- Register the organisation
- Add minimum 100 members
- Build a support group of experts (doctors, counsellors, physiotherapist, etc)
- Implement a robust social media strategy.



# 2021 Annual Summit

## FSHD Israel Action Plan 2021-2022

- Continue to grow our group (currently 50 members)
- Engage with medical community members (e.g. invite to speak at virtual meetings)
- Hold an in-person support meeting
- Develop a list of healthcare resources



PRESIDENT MARCO RASCONI



COMMUNITY

HEALTHCARE

RESEARCH

COMMUNICATION

Uildm-FSHD

**SOCIAL INCLUSION: SPORTS, SCHOOL, WORK ACCESS**  
**INDEPENDENT LIFE PROGRAMS**  
**ONLINE HANGOUTS**  
**IN-PERSON SOCIAL EVENTS**



**ROUTINE MEDICAL CHECKS @NEMO**  
**MULTI-DISCIPLINARY CENTERS**  
**PHYSIOTHERAPY @Uildm SECTIONS NATIONWIDE**  
**VACCINATION PROGRAMS**



**A.I. NEMOLABS IMPLEMENTATION WITH**  
**9 NEMO CENTERS NATIONWIDE**  
**TO IMPROVE QUALITY OF LIFE**  
**FSHD-RESEARCH STUDIES**  
**TELETHON NMD STUDIES**  
**INTERNATIONAL ENGAGEMENT**



**WEBSITE**  
**NEWSLETTERS**  
**SOCIAL MEDIA**  
**FSHD-SPECIFIC WEBINARS**

## FSHD Italia Onlus' action Plan 2021-2022

- **Monitoring of FSHD progression and severity through blood draws combined with data coming from MRI**
  - Analysis of variation in time of Cell-free DNA (cfDNA);
  - Possibility to repeat the test multiple times;
  - Research conducted by Policlinico Agostino Gemelli (Rome);
- **Microdialysis tests**
  - Placement of microcatheter in the muscle and observation of the patient for five consecutive days;
  - Identification of biomarkers responsible for the inflammation of the muscle;
  - Development of innovative therapeutic strategies;
- **Psychological support**
  - Mindfulness classes;
  - Psychology sessions;

# 2021 Annual Summit



## FSHD Japan communities Action Plan 2021-2022

- To promote the cases with genetically confirmed diagnosis based on the current diagnosis standards (including 4qA test) and increase the number of patient registration.
- To increase the chances for patients and families to know each other.
- To increase younger participants to our community and foster members with potential of global communication.
- To support domestic researchers activities focusing on FSHD via online tools.

We have two FSHD communities in Japan. We are collaborating with each other to hold events and share information.





## Annual plan of the Dutch FSHD diagnosis group:

- Participate in the Dutch Trial Network
- Participate in a 'set up' Care Network
- Combine the research agenda from patient perspective with the professional one
- Raise patients awareness of the importance of the registration database
- Help to organize annual webconference in November meant for patients
- Organize regular meetings for patients (online and offline)



## **FSHD patients group the Netherlands**

- 7 persons , of which 6 are volunteers
- Muscular diseases association the Netherlands
  - 120 diagnoses
  - 9000 members
  - Goals
  - Cooperation
- Members roles and responsibilities





# 2021 Annual Summit



FSHD Stichting (NL). Established 1997, funds research and aims to help find a cure for FSHD. Largely new board since end 2020; redefining strategy

The next few years will be critical with a number of potential drugs entering clinical trial phase... and beyond! Focus now is on enabling this.

- Registry of FSHD patients
- Biomarker study

## Goals for 2021/2022

- New marketing campaign to make disease more known in NL; link with stand-up comedians to generate more income to ensure ability to continue to fund significant research
- Close alignment with VSN, PBS, other muscular disease organisations in NL and internationally to ensure efficiencies, more effective patient advocacy and activation
- Enable registration of more FSHD patients



## **Russian FSHD Patients Organization Action Plan 2021-2022**

**Speaker: Anastasia Krainova**

representative of Russian FSHD community

**Contact person: Kirill Shakirov**

shakirov\_k@yahoo.com



## 2021 Annual Summit

### **Patients Organization Action Plan 2021-2022**

- Registration of official organization
- Creating web-site of organization with information about disease
- Holding online meetings and consultations between patients and doctors
- Obtaining orphan disease status for FSHD
- Attract funding from the government
- Communications with other Russian patients organizations of rare diseases and participations in all events of World FSHD Alliance



# 2021 Annual Summit



## Muscular Dystrophy Foundation of South Africa Action Plan 2021-2022

- Awareness
- Patient registry
- Support services
- Support from medical professionals

## FSHD Spain Plan 2021-2022

### 1. Consolidate the Association

Grow number of members, Annual summit, Patient registry, Education for the board, Increase income –book–, Get government certification as “NGO public benefit”

### 2. Improve internal processes

Four newsletters/year for members and scientists, Generate communications plan for all channels, Transparency, Reorganize web site, Expand WhatsApp

### 3. Grow awareness

Presence in local/national media and social networks, Organize 5 events and webinars/year

### 4. Develop institutional relationships

Politicians, Public hospitals, Researchers, Neurologists, Physiotherapists and Rehabilitation centers, attend to other associations’ 10 events/year

	2020	2021	2022
<b>Consolidate the association</b>			
Grow number of members	✓	✓	✗
Celebrate June 20	✓	✓	✗
Annual summit	✗	✗	✗
Patient registry	✗	✗	
Training	✓	✓	✗
Increase income	✓	✓	✗
Government certification	✓	✗	
<b>Improve internal processes</b>			
Better website	✗	✗	
WhatsApp for members	✓		
Newsletters	✓	✓	✗
Communications plan	✗	✓	
Fund research activities	✓	✗	✗
Transparency	✓	✓	✗
<b>Grow awareness</b>			
Media	✓	✓	✗
Social networks	✓	✓	✗
Events	✗	✗	✗
<b>Institutional relationships</b>			
Meet politicians	✗	✗	✗
Meet researchers	✗	✗	✗
Meet neurologists	✗	✗	✗
Other associations	✓	✓	✗
Develop clinical guide		✗	✗
European Trail Network		✓	✗



## Muscular Dystrophy UK Action Plan 2021-2022

- FSHD webinar 9th July
- FSHD in 2021 research grant round
- participating in the European Patient Pharma project