



An Engaged and Empowered Community  
June Kinoshita, Chief Strategic Programs Officer  
FSHD Family Day Conference  
February 23, Miami



Year 2025

Community  
Engagement

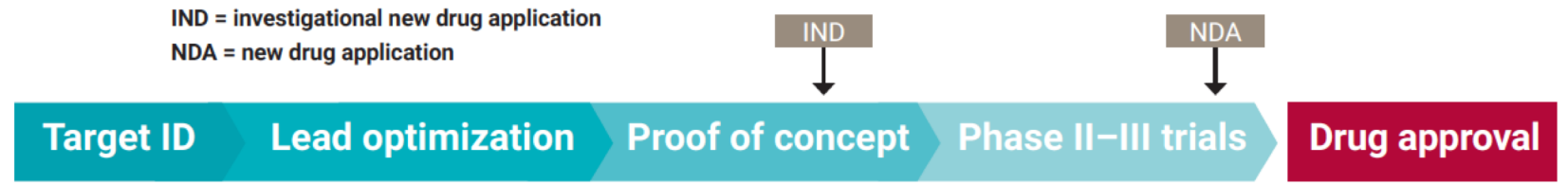
Accelerate  
Research







# The FSHD drug development pipeline



### Therapies to promote muscle growth & regeneration

- Acceleron—ACE-083 (myostatin inhibitor)
- Harper lab—follistatin gene therapy
- Strykagen—small molecule drug
- Perlingeiro lab—stem cell therapy

### Therapies to block DUX4

- Fulcrum Therapeutics—small molecule
- Novartis—small molecule
- GBC0905—small molecule
- Harper lab—anti DUX4 gene therapy
- Facio Therapies—small molecule
- Chen lab—antisense oligomer
- Dumonceaux lab—antisense oligomer
- Lek lab—small molecule
- Ultragenyx—small molecule
- Pfizer—gene therapy
- ...and many more

**Promising  
Treatments**



Patient registry  
& natural history

Biomarkers

Imaging  
markers

Clinical  
outcomes

**Patient engagement**

**FDA Approval**

**Our Families**



## What Do We Mean by Patient Engagement?





## Community Engagement

YOU—THE PATIENTS, FAMILIES AND CAREGIVERS—CAN  
DRIVE US FURTHER AND FASTER TOWARD OUR GOAL

- The more you learn about FSHD, the better able you will be to advocate and care for yourself and your family.
- Sharing your experiences adds to knowledge and improves medical care and clinical trial design.
- You are essential as collaborators and activists.



## Factors affecting quality of life

- Patients' quality of life was affected "Moderately" to "Very much" by weakness in arm/shoulder (88%), foot/leg (80%), and core/abdominal muscles (81%).
- Fatigue and pain also frequently impacted patients' quality of life.

**Table 2: Factors Affecting Patient Quality of Life (n=388)**

	Very much	Moderately	Slightly	Not at all
Foot/leg weakness	58%	22%	14%	7%
Arm/shoulder weakness	53%	35%	11%	1%
Core/abdominal weakness	51%	30%	13%	6%
Fatigue	35%	39%	21%	4%
Pain	19%	34%	33%	15%
People's lack of understanding	19%	25%	33%	24%
Having to keep my FSHD secret	10%	8%	16%	67%
Loss of facial expression	9%	20%	35%	38%
Breathing issues	6%	16%	28%	50%
Hearing loss	5%	11%	25%	59%
Speech impairment	2%	6%	28%	64%

**Table 3: Motivators for Clinical Trial Participation**  
(n=388)

Even if it might not help me, it will help others some day	69%
The feeling that I'm doing everything I can do to help	57%
The idea of advancing science	57%
The chance to be first in line to try a treatment	42%
I'll only try something that's proven, so I wouldn't volunteer	5%
Other	4%
I'm not seeking a drug to cure FSHD, so I wouldn't volunteer	2%

Note: Patients could select more than one motivator for clinical trial participation.



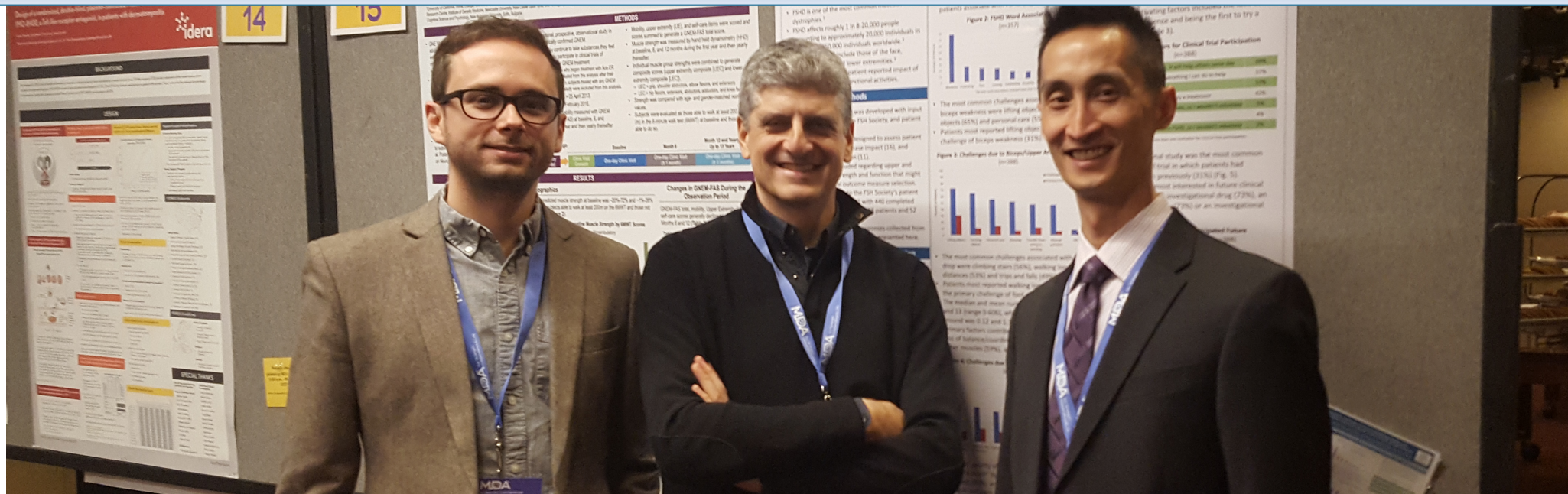
# Your engagement informs future clinical trial design

## Manifestations and Impact of Facioscapulohumeral Muscular Dystrophy (FSHD): Preliminary Results from a Survey of FSHD Patients

Thienhuu Q. Nguyen<sup>1</sup>, June Kinoshita<sup>2</sup>, Barry Miller<sup>1</sup>, Chad Glasser<sup>1</sup>, Kenneth M. Attie<sup>1</sup>, Jeffrey M. Statland<sup>3</sup>  
<sup>1</sup>Acceleron Pharma, Cambridge MA; <sup>2</sup>FSH Society, Lexington, MA; <sup>3</sup>University of Kansas Medical Center, Kansas City, KS

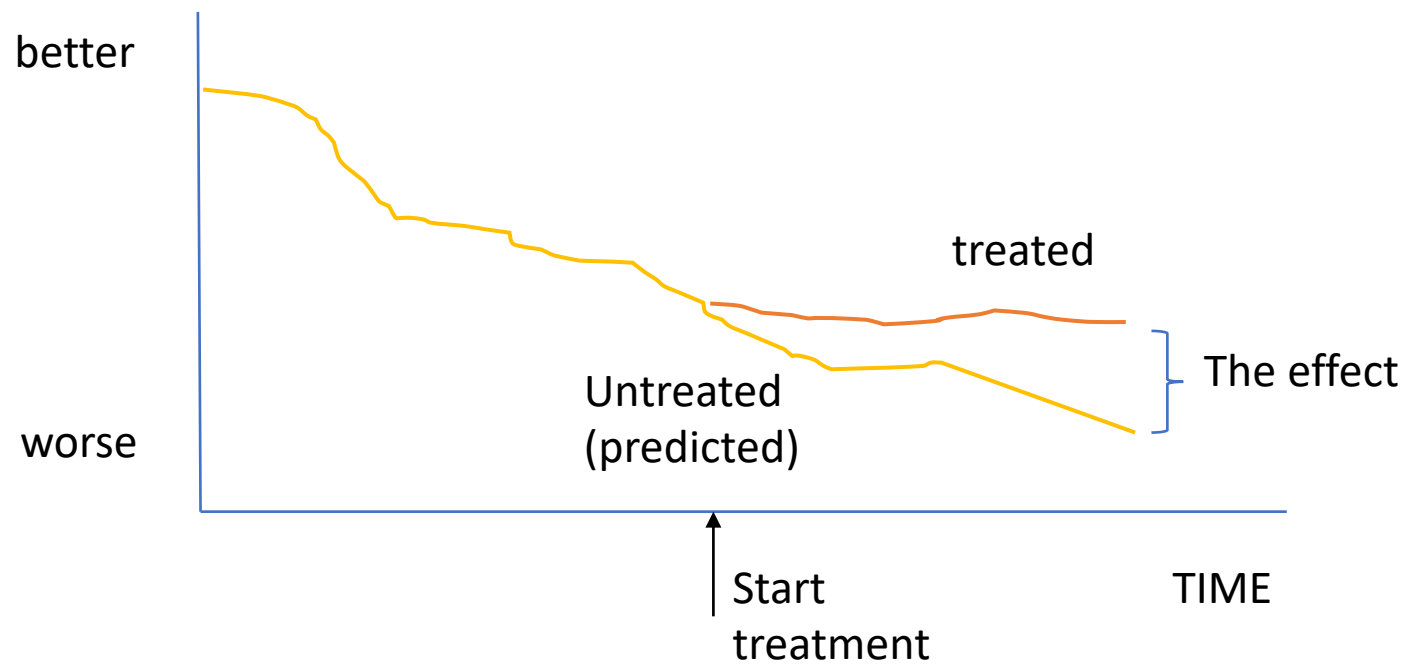
2017 MDA  
Scientific  
Conference

Arlington, VA





\$64 MILLION QUESTION  
DOES A TREATMENT WORK?





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**VOLUNTEER FOR RESEARCH**

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- [Donate Tissue](#)
- [Join an FSHD Registry](#)
- [What Is a Clinical Trial?](#)
- [Why Volunteer?](#)

**CONNECT LOCALLY**

- [FSH Society Chapters](#)
- [Start a Chapter](#)
- [Walk & Roll to Cure FSHD](#)
- [Support Groups Online](#)
- [Communities](#)

**EVENTS**

- [Events Calendar](#)
- [FSHD Connect Conference](#)
- [International Research Congress](#)



## Studies recruiting research subjects

Clinical Trial Readiness to Solve Barriers to Drug Development in FSHD. PIs Jeffrey Statland, MD, and Rabi Tawil, MD.	▼
FSHD Biomarker study. Wellstone Center for FSHD Research, Univ. of Massachusetts Medical School. Robert Brown, Jr., MD, and Larry Hayward, MD PhD.	▼
Research study for FSHD (Facioscapulohumeral Muscular Dystrophy) conducted by Dr. Michael Kyba and Dr. Peter Karachunski, University of Minnesota.	▼
Research study to understand if the resting metabolic rate and cardiovascular response to exercise are affected by the genetic mutation that causes for FSHD. By Manda L Keller-Ross, PhD, DPT, PT, University of Minnesota.	▼
Volunteers who are 18 to 55 years of age with FSHD needed for a study to evaluate functional mobility. Principal investigator, Jay Han, MD, UC Irvine.	▼
A Phase 2 Randomized, Double-Blind, Placebo-Controlled Study of ACE-083 in Patients With Facioscapulohumeral Muscular Dystrophy. Acceleron Pharma. Multiple sites in U.S.	▼
FSHD Blood Draw Study at Johns Hopkins Hospital, Baltimore, MD (Non-local patients encouraged to apply)	▼
Facioscapulohumeral Muscular Dystrophy (FSHD) Imaging Study at Kennedy Krieger Institute, Baltimore, MD.	▼

## A sobering fact

Across all research, 85 percent of trials face delays and 30 percent never get off the ground due to a lack of volunteers.

**We can't let this happen in FSHD. Our families are waiting.**





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## FSH Society Chapter Program

- Fundraising
- Community Building
- Education & Empowerment



## The Power of a Chapter

- Think globally, act locally
- Volunteer led, staff-supported
- Leadership training
- Serve families where they live
- Strengthen relationships with research centers
- Empowerment





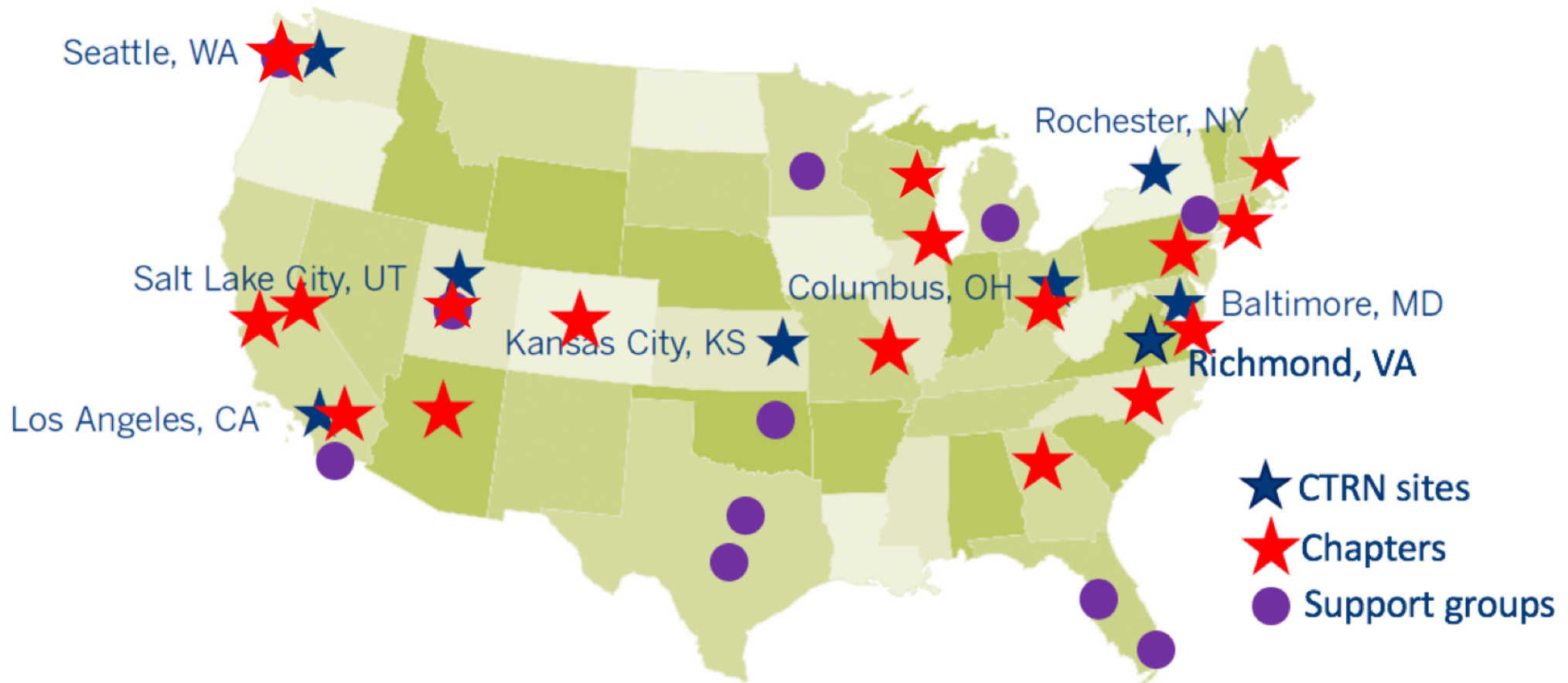
## The Power of a Walk & Roll

- Build programs
- Build community
- Empower patients/families
- Tells a story
- Limitless fundraising potential
- In 2018, 5 walks raised \$230K!





## Location of chapters, support groups, and CTRN sites



## What about Miami (and elsewhere in Florida)?





## Thank you to

- Dr. Mario Saporta and the University of Miami Miller School of Medicine team
- Drs. Ora and Isaac Prilleltensky
- Our guest speakers
- Robyn O'Leary, FSH Society

# Your gateway to all things FSHD

[www.fshsociety.org](http://www.fshsociety.org)

The screenshot shows the FSH Society website homepage. At the top left is the logo: "FSH SOCIETY FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY". The navigation menu includes: Financials, Library, News Center, Contact Us, a search bar, and buttons for "Donate" and "Join Us". Below the menu is a hero banner with a photo of two women and the text: "With your support, we are leading the way to a better future for all who live with FSH muscular dystrophy." A "JOIN FSH SOCIETY" button is in the bottom right of the banner. Below the banner are two main sections: "Get Involved in the Fight to Cure FSHD" with icons for "Join Us", "Volunteer", and "Donate"; and "Newly Diagnosed?" with an icon for "We Are Here To Help".





Year 2025

**The Cure is out there.  
It's up to all of us make it happen.**