



My Personal Journey and Career With FSHD

Presenter
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My Childhood

Sports, year round

- Traveling Soccer Team for the City of Livermore, CA
- Basketball and Baseball

My Adolescence

Family moved to Illinois where my father had received a position at the University

Played Varsity Soccer and Basketball in High School

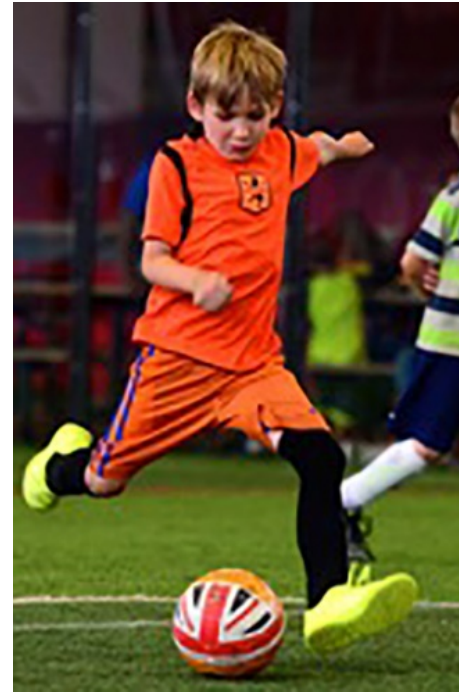
Was accepted as a student of electrical engineering at the University of Illinois

- Weight Lifting
- Scapular weakness



I was diagnosed with FacioScapuloHumeral Muscular Dystrophy (FSHD) at the age of 20.

- Relatively unknown disease
- Looking Back at the signs



The Signs that I had FSHD

- Congenital absence of the Pectoralis major
- Inability to complete a sit-up at the age of 13
- Loss of speed and jumping height beginning at age 15
- Extreme hamstring weakness at age 18
- Massive scapular winging during weight lifting at age 19
- Difficulty walking up stairs at age 27
- Difficulty getting up from seats at age 29
- Difficulty walking up slopes at age 32
- Difficulty walking period at age 36

Facioscapulohumeral Muscular Dystrophy

FSH SOCIETY
FACIOSCAPULOHUMERAL
MUSCULAR DYSTROPHY

FACT:
FSHD is among the most prevalent forms of muscular dystrophy.

FSH SOCIETY
FACIOSCAPULOHUMERAL
MUSCULAR DYSTROPHY

FACT:
95% of people with FSHD develop noticeable muscle weakness by the age of 20.

FSH SOCIETY
FACIOSCAPULOHUMERAL
MUSCULAR DYSTROPHY

FACT:
FSHD occurs with equal frequency in both males and females.

FSH SOCIETY
FACIOSCAPULOHUMERAL
MUSCULAR DYSTROPHY

FACT:
An early sign of FSHD is the inability to whistle or sip through a straw.

FSH SOCIETY
FACIOSCAPULOHUMERAL
MUSCULAR DYSTROPHY

FACT:
Typically, FSHD muscle weakness is first noticed in the face, shoulders, upper arms and by foot drop.

FSH SOCIETY
FACIOSCAPULOHUMERAL
MUSCULAR DYSTROPHY

FACT:
FSHD can cause progressive loss of skeletal muscle and make even the simplest daily tasks impossible.

FSH SOCIETY
FACIOSCAPULOHUMERAL
MUSCULAR DYSTROPHY

FACT:
Around 20% of people with FSHD will become dependent on a wheelchair or scooter.

FSH SOCIETY
FACIOSCAPULOHUMERAL
MUSCULAR DYSTROPHY

FACT:
FSHD can cause significant hearing loss and abnormalities in the eye.

FSH SOCIETY
FACIOSCAPULOHUMERAL
MUSCULAR DYSTROPHY

FACT:
The age of onset for FSHD and the extent of muscle loss varies from person to person.

FSH SOCIETY
FACIOSCAPULOHUMERAL
MUSCULAR DYSTROPHY

FACT:
FSHD is estimated to affect 1 in 8,333 people around the world.

FSH SOCIETY
FACIOSCAPULOHUMERAL
MUSCULAR DYSTROPHY

FACT:
The frequency of FSHD may be 3x higher than reported due to undiagnosed cases.

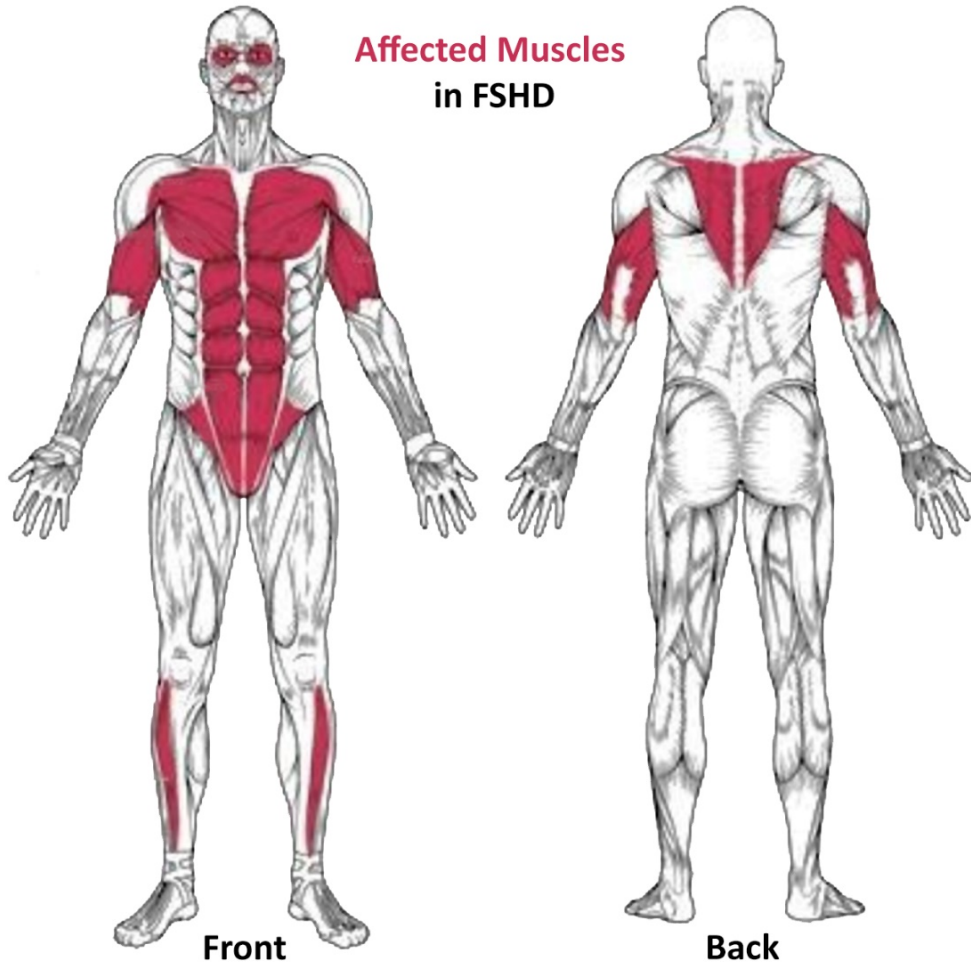
FSH SOCIETY
FACIOSCAPULOHUMERAL
MUSCULAR DYSTROPHY

FACT:
Currently, there is no cure for FSHD.

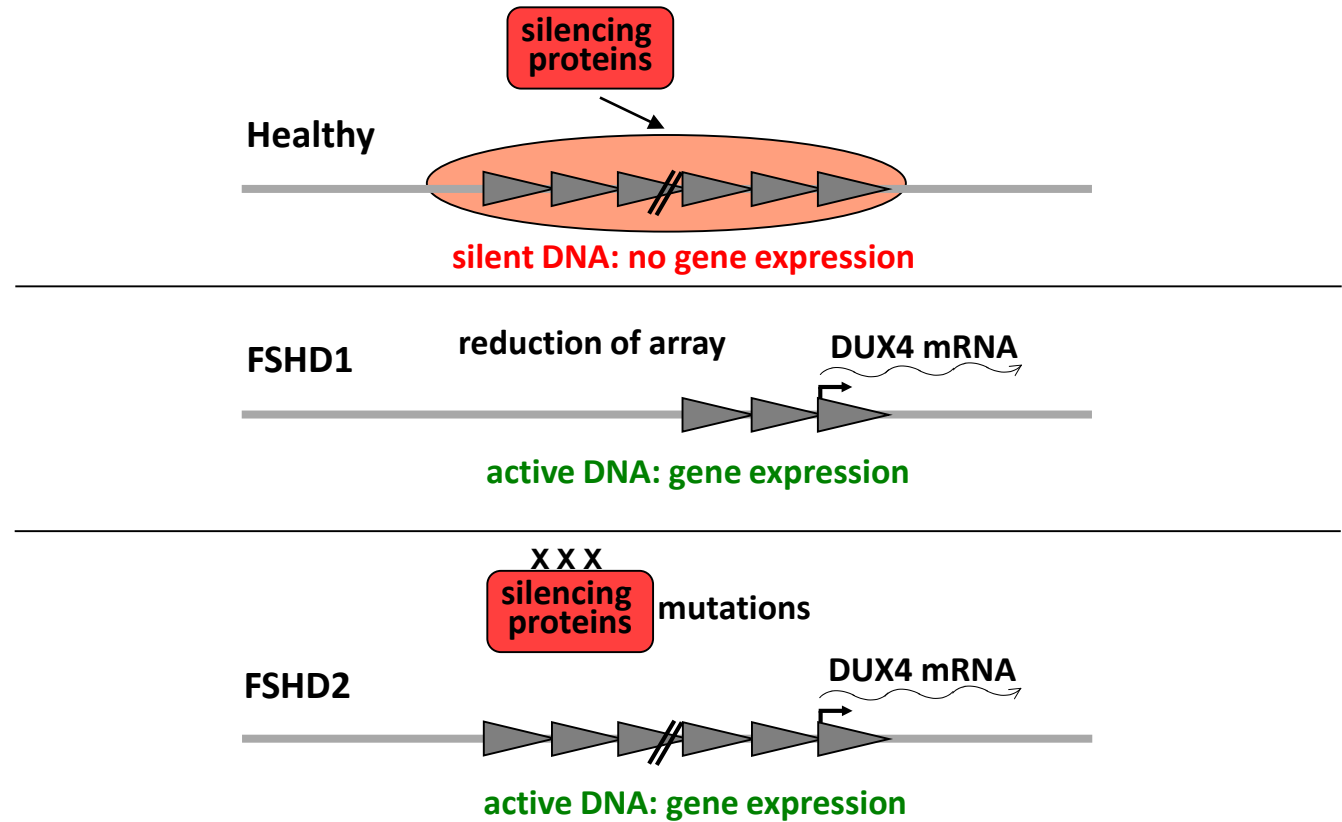
#CureFSHD

FacioScapuloHumeral Dystrophy (FSHD)

FSHD is caused by misexpression of the DUX4 gene in skeletal muscle



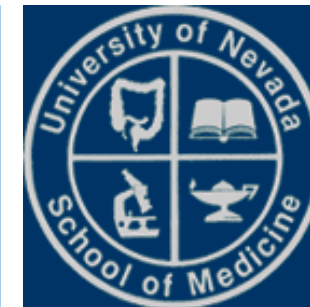
D4Z4 repeat array at chromosome 4



DUX4 Expression is Highly Toxic to Most Human Cells!

My Career Choices After the FSHD Diagnosis

- I switched career trajectories towards Chemistry/Biology and away from Electrical Engineering.
- In Graduate School, I approached a newly arrived Professor from NIH by the name of Dr. Peter Jones (now UNR Mick Hitchcock, PhD Endowed Chair in Medical Biochemistry) about working in his lab and studying FSHD.
- As a post-doctoral fellow, I transitioned to Dr. Dean Burkin's lab in the Pharmacology department of the UNR school of medicine to learn about the drug development process and develop treatments for the muscular dystrophies.
- In 2013, Dr. Burkin and I began a company called StrykaGen in order to help transition the Integrin enhancing therapeutics we have developed towards the clinic.



2018 UNR Synapse Article
details part of this story

FSHD affect on my role as a UNR Research Associate Professor of Pharmacology

Research (affected)

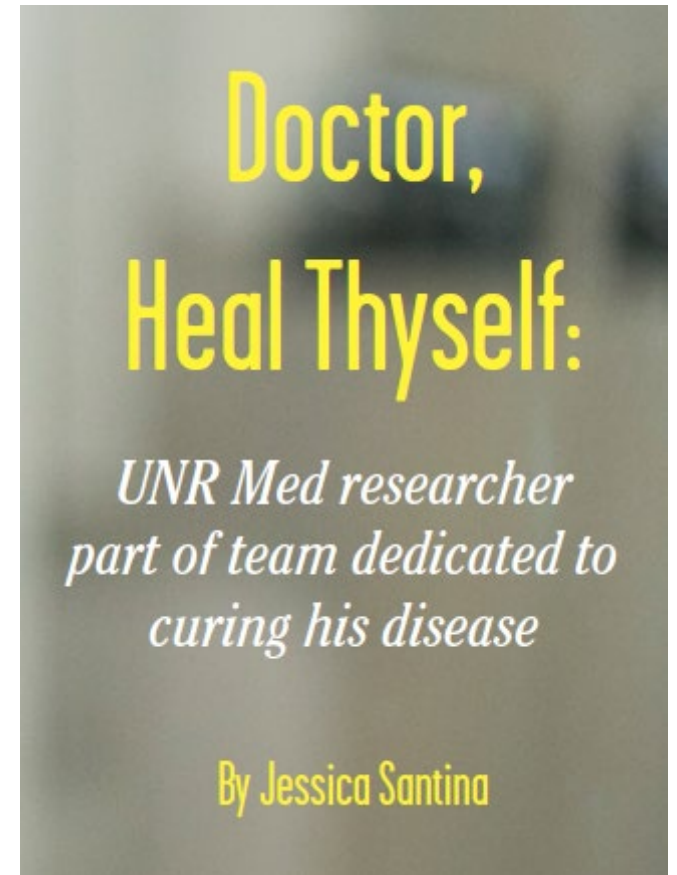
- My physical abilities are declining and thus I am able to do less direct research than I previously could.
- I now have an expanded role in experimental planning and design, data analysis, certain types of data collection.

Grants (Unaffected)

- Like many researchers, I spend a lot of time trying to procure funding.

Publications (Unaffected)

- 4 publications (1 first author) this year.



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I am currently finishing a study assessing one of our muscle regenerating therapeutics in a new FSHD mouse model developed by Dr. Peter Jones.

My Perspective on FSHD Patients and Career Opportunities

~46% of FSHD patients between 18-65 are working

Careers requiring any physical activities should be avoided

- This could even include low exertion activities

Competition with non-FSHD patients

Most FSHD patients I have met are in White collar jobs (MD, Lawyers, Accountants, Professors)

My Future Career Goal

To become a Tenured Professor studying cures for muscular dystrophies.

FSHD may make this more difficult than normal.

- High amount of research is expected of junior PI
- Funding is dependent on productivity