

F S H D
WELLSTONE RESEARCH UPDATE
1.27.18
 friends of FSH research
 SEATTLE PAUL D. WELLSTONE MUSCULAR DYSTROPHY COOPERATIVE RESEARCH CENTER

UNIVERSITY of WASHINGTON
 Paul D. Wellstone Muscular Dystrophy Cooperative Research Center

Speakers today:

Amy Campbell, PhD
Joel Chamberlain, PhD
Dennis Shaw, MD
Stephen Tapscott, MD PhD
Leo H. Wang, MD PhD

UNIVERSITY of WASHINGTON
 Paul D. Wellstone Muscular Dystrophy Cooperative Research Center

Director:
Jeff Chamberlain, PhD

Co-director:
Stephen Tapscott, MD PhD

- Part of the MD-CARE Act of 2001
- Established in 2003
- Six NIH-sponsored centers
- Funded by the National Institute of Arthritis and Musculoskeletal and Skin Diseases
- UR + UW: 2014-2018
- Two projects: one basic science, one translational research project
- Our clinical project is focused on FSHD

Stephen Tapscott
Rabi Tawil

Chris Budech
Katy Eichinger
Seth Friedman
Nancy Gove
Sandy Poliachik
Dennis Shaw
Leo Wang

Wellstone Project 2

- 36 patients
- University of Rochester and University of Washington
- 23 males and 13 females
- At UW: Average 6 (range 4-9) D4Z4 units; all permissive haplotype
- Clinical Severity Score Median 5.5
- ~59 needle muscle biopsies



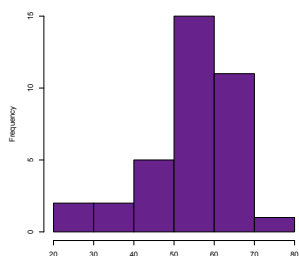
We have reached our goal of 36 patients across two sites.



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As of today 35/36 patients have returned for their year 2 visits.



We have reached our goal of 36 patients across two sites.
As of today 35/36 patients have returned for their year 2 visits.
We have obtained at least 62 muscle biopsies.



Age range = 20-75
Median = 57
Mean = 54

	Visit 1	Visit 2
	0	Month 12 (+/- 1 month)
Laboratory tests: PT, PTT	X	X
Vital signs (Height, weight, BP, RR, HR, temperature)	X	X
Neurological exam/ confrontational strength exam	X	X
Quantitative strength testing	X	X
CSS: Clinical Severity Score	X	X
MRI of lower extremities	X	X
Needle muscle biopsy	X	X

First hour

- Genetics and biology of FSHD: Stephen Tapscott, MD PhD
- Searching for DUX4 in FSHD muscle: Joel Chamberlain, PhD
- MRI biomarkers in FSHD: Dennis Shaw, MD

Break

- Around 2:16

Second hour

- Clinical and pathological correlations in the FSHD Wellstone cohort: Leo H. Wang, MD PhD
- Drug Development for FSHD: Amy Campbell, PhD

Introductions and informal discussion

- Introduction to the FSHD researchers

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Neuromuscular Testing Center

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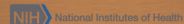


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Stephen Tapscott Rabi Tawil	Don Henderson Leann Lewis Bharati Shah	
Chris Budech Katy Eichinger Seth Friedman Sandy Gove Sandy Poliachik Dennis Shaw	Darren Bisset Joel Chamberlain John Hall Bobbi Miller Maja Zavaljevski	

NIH U01 Tawil/Statland study on Clinical Trial Readiness to Solve Barriers to Drug Development in FSHD

FSHD Immunosuppressive trial

- Open label tacrolimus + prednisone
- Short: Three months
- Follow blood markers and radiological markers
- MRI before, in the middle, and after treatment
- PT and strength measurements

