



WELCOME!

2021 Iowa Wellstone Dystroglycanopathy Patient and Family Conference

July 16-17
Iowa City, IA
(and on Whova)

Thank you to this community

- Your thoughtfulness was overwhelming and so appreciated
- I am grateful to know you

*Thank
you*



For those not
able to join in
person, Iowa
in July



Acknowledgements to key people

- Organizers

- Jaeda Harmon
- Carrie Stephan
- Shelley Mockler
- Katie Laubscher
- Chyan Decker



- Research team:

- Chandra Miller
- Corey McDaniel
- Evgenia Folts
- Bryn Miller
- Hanlong Fan



Thank you to our advisory council members!

- Dystroglycanopathy patient and family advisory council
 - Akemi Rogers
 - Tara Boone
 - Karen Cole
 - Vanessa Thorpe
 - Kelly Brazzo
 - Julie Savage
- Please let us know if you would be willing to serve on this board

Whole Wellstone Center Advisory Boards. Thank you!

- Internal (U of Iowa)

- Ted Abel
- Christopher Ahern
- Christopher Cooper
- Peggy Nopoulos
- George Richerson
- Michael Shy

- External

- Barry Byrne (Florida)
- Anne Connolly (Ohio)
- Jim Ervasti (Minnesota)
- Trisha Tully
(parent/patient representative)

Thank you to those who support this conference financially

- NIH
- CURELGMD2i Foundation (Kelly and Keith Brazzo)
- LGMD2i Fund (John Pierre Laurent)
- Private donors





Senator Paul D. Wellstone Muscular Dystrophy Specialized Research Center at the University of Iowa

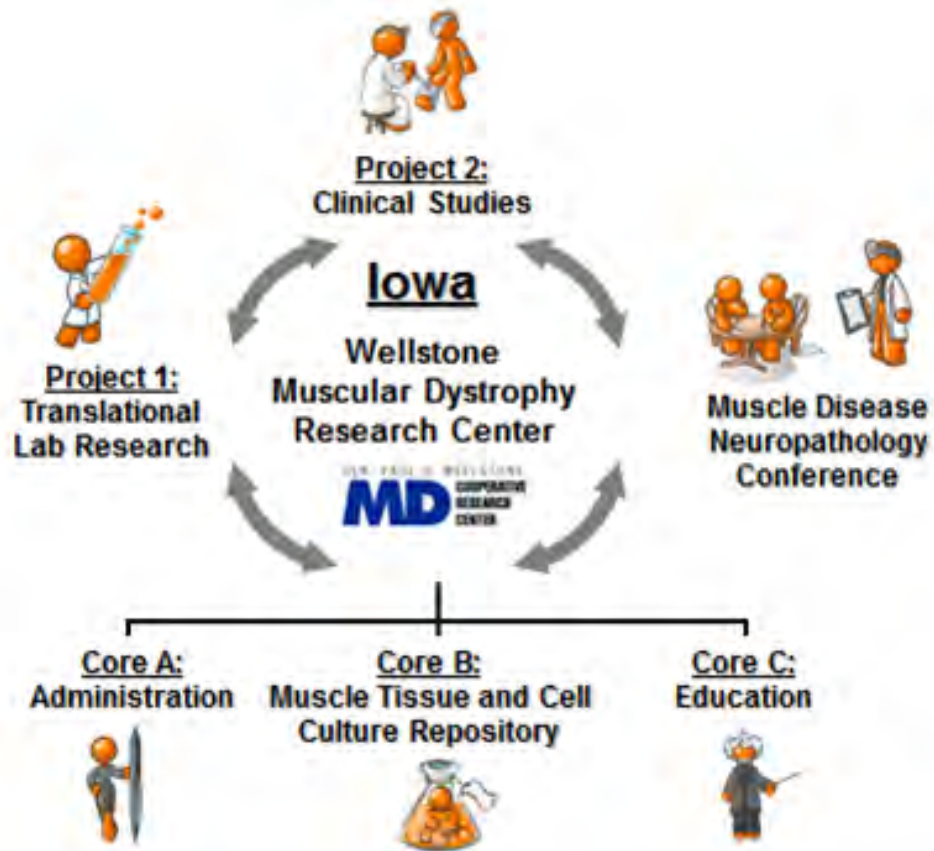


Kevin P. Campbell, Steven A. Moore, Katherine D. Mathews

Iowa Wellstone Muscular Dystrophy Center



National Institutes
of Health

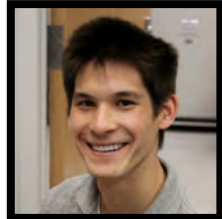


Overall goal: Improve treatment for patients now and in the future

Wellstone Medical Student Fellows



Jamie Eskuri (2010-2011)
Pediatric Neuromuscular
Physician
Gillette Children's



Steve McGaughey (2011-2012)
Assistant professor, Emergency
medicine
University of Oregon



Katie Lutz (2012-2013)
Assistant professor, Child
neurology
University of Iowa



Cameron Crockett (2013-2014)
Neuromuscular fellow
Washington University, St. Louis



Braden Jensen (2014-2015)
General surgery Resident
University of Iowa



Brianna Brun (2015-2016)
Child Neurology Resident
Ohio State University



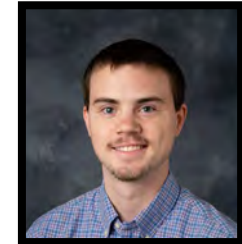
Courtney Carlson (2016-2017)
Orthopedic surgery Resident
Mayo Clinic, Rochester



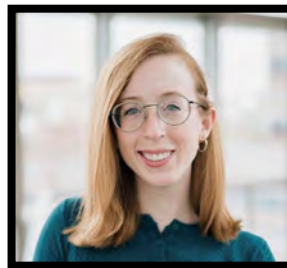
Angela Lee ((2017-2018)
Pediatrics/Genetics Resident
Washington University, St. Louis



Natalie Manhica (2019-2020)
Anesthesia resident
University of Iowa



Eric Libell (2019-2020)
Neurology resident
University of Cincinnati

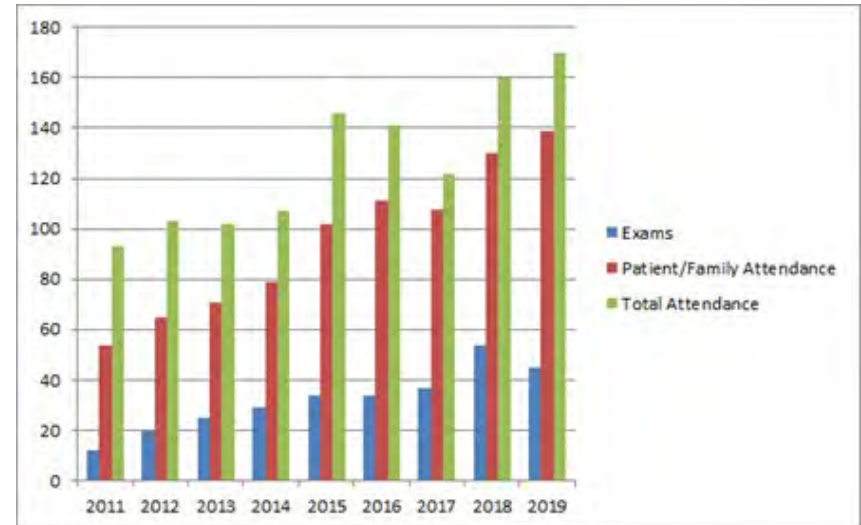


Lauren Coffey (2020-2021)
M4, University of Iowa

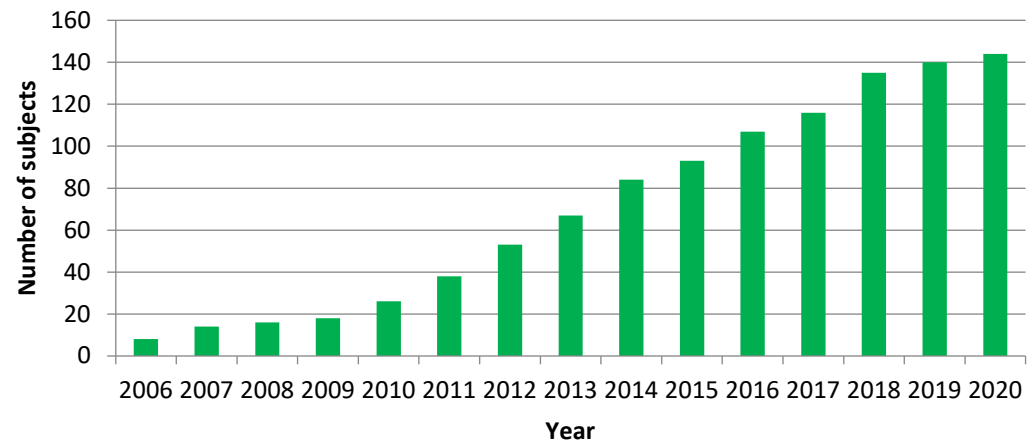
Enrollment was impacted by pandemic

- Covid19 had a major impact on clinical research
- 2020 conference via Zoom had 118 unique viewers from around the world
- New enrollment often associated with conference

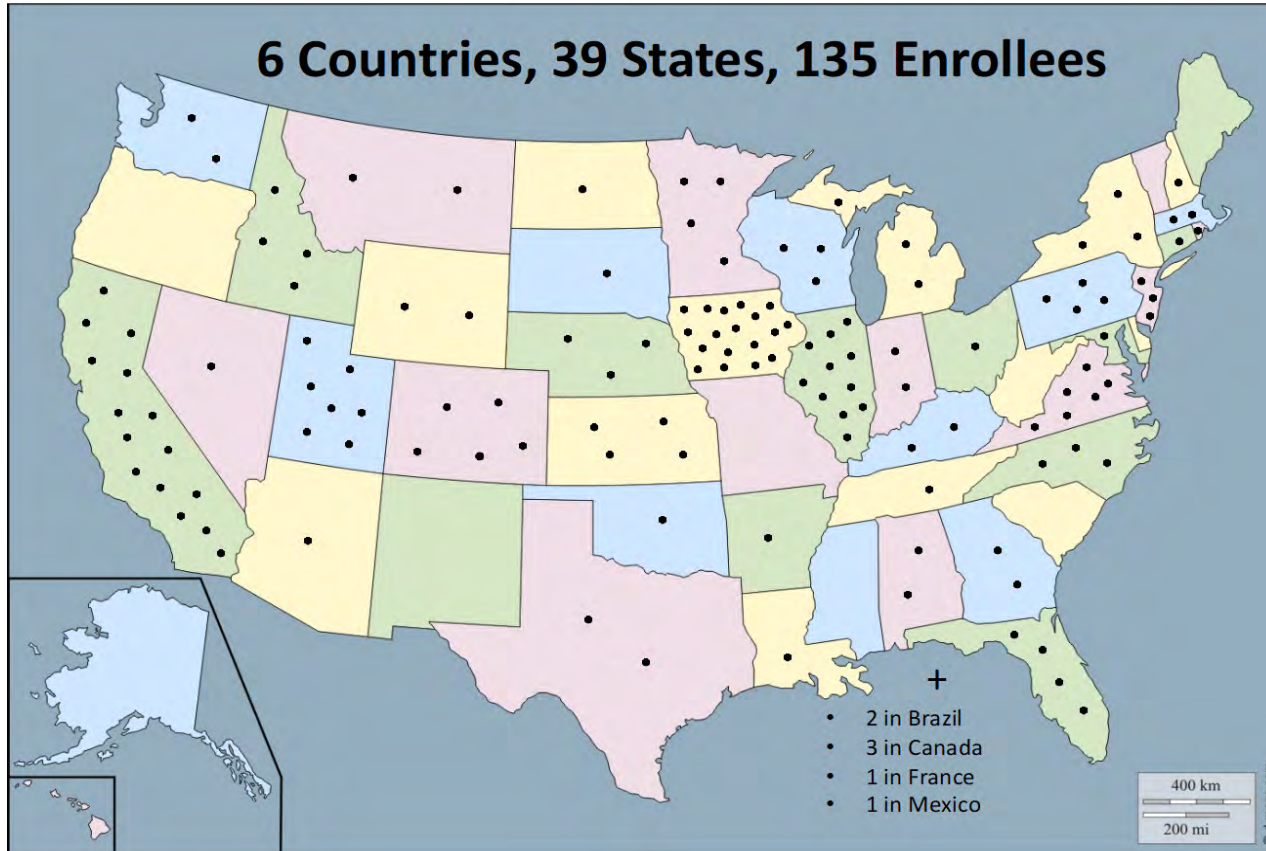
Conference attendance through 2019



Cumulative Enrollment 2006-2020



Enrollment is geographically diverse



Previous conferences

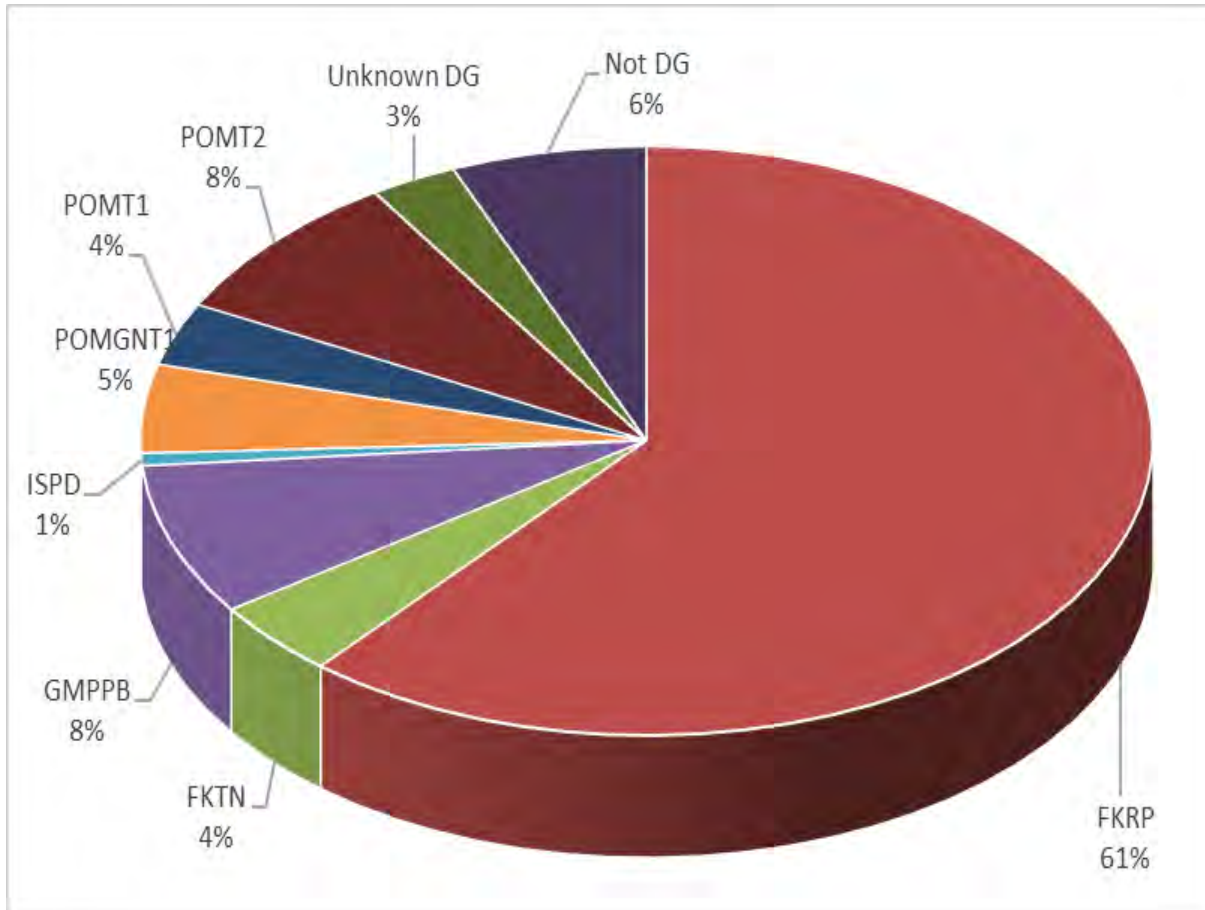


Community is an important part of the conferences

Virtual attendees--you can participate if you are able to travel

- We can schedule research visits throughout the year Monday-Friday
- Weekend exam date: October 23, 2021
- Email Carrie Stephan if interested in arranging a research visit
 - Carrie-Stephan@uiowa.edu

Distribution of Wellstone enrollees consented by genotype



Gene	
FKRP	88
FKTN	6
GMPPB	12
ISPD	1
POMGNT1	7
POMT1	5
POMT2	12
Unknown DG	4
Not DG	9

Publications this year
from your information

Motor outcome measures in patients with *FKRP* mutations

Cumulative % who are not able to do the test by age

AGE IN YEARS	10 M WALK	4 STAIR CLIMB	STAND FROM LYING	6 MINUTE WALK
20	23	25	58	24
30	23	34	78	24
40	33	45	78	33
50	40	54	87	56

Adapted from
Gedlinski,A. et al. Neurology October 13, 2020

Change in motor outcome measures after 6 years in patients with homozygous c.826C>A *FKRP* mutations compared to European cohort

Summary: they are remarkably similar

Table 6 Disease progression over 6 years in the present cohort compared with published results: adults homozygous for c.826C>A mutation

	Published cohort ²⁹ (n = 23)		Difference	Current cohort (n = 32)	
	Baseline median	Year 6 median		Mean annual change (95% CI)	Estimated 6-year change (95% CI)
FVC, sitting, %	77	64	-13	-1.97 (-2.4 to -1.6)	-11.8 (-14.1 to -9.5)
6MWT, m	391	286	-105	-15.23 (-22.6 to -7.8)	-91.4 (-135.7 to -47.0)
10MW/R, m/s	1.2	0.8	-0.4	-0.06 (-0.1 to -0.04)	-0.39 (-0.5 to -0.3)
4SC, stairs/s	0.7	0.4	-0.3	-0.05 (-0.1 to -0.03)	-0.3 (-0.5 to -0.2)
Knee extension, pounds	24.5	26.5	+2.0	-0.15 (-0.9 to 0.6)	-0.9 (-5.6 to 3.7)
Knee flexion, pounds	18.5	10.6	-7.9	-0.60 (-1.0 to -0.2)	-3.6 (-6.2 to 1.1)

Abbreviations: 4SC = time to climb 4 steps; 6MWT = 6-minute walk test; 10MW/R = 10-meter walk/run; CI = confidence interval; FVC = forced vital capacity. Values for knee extension and knee flexion were converted from kilograms to pounds.

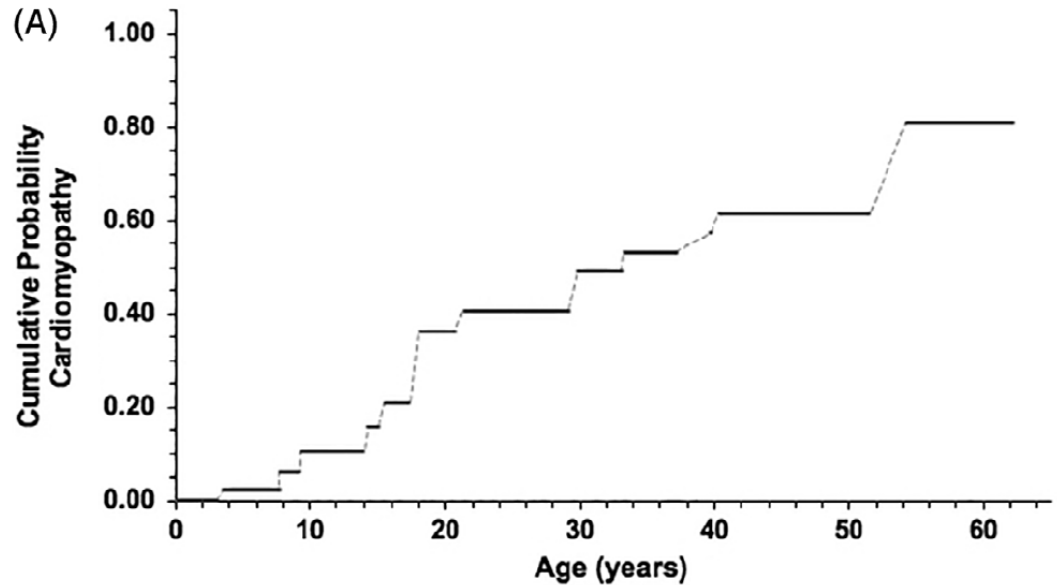
- Gedlinski, A. et al. Neurology 2020
- Murphy AP, et al. Ann Clin Transl Neurol 2019

Pregnancy outcomes in LGMDR9

- More assisted deliveries (ex. vacuum extraction) but fewer c-sections than national average
 - Babies were healthy
 - About half of pregnancies resulted in progression of weakness that did not return to baseline
 - No clear predictors identified
- Libell, E, et al. Muscle and Nerve 2020

Cardiomyopathy in limb girdle muscular dystrophy R9, FKRP related

Libell, E. et al. Muscle and Nerve, 2021



Genotype	Number of participants	Number of echocardiograms reviewed	With abnormal echocardiogram count (%)	Age at first abnormal echocardiogram, y Median (95% CI)
c.826C>A, c.826C>A	29	86	9 (31)	54.2 (39.7-54.2)*
Other <i>FKRP</i> genotypes	27	71	16 (59)	18.1 (14.3-18.1)*
Total	56	157	25 (45)	33.3 (18.0-54.2)

This year's Wellstone fellow projects

- Diagnostic odyssey for patients with LGMD 2I/R9
 - See Lauren Coffey's video presentation
 - Spoiler: diagnosis is often delayed, delay differs by presenting symptom, but the delay is getting shorter
- Predicting time to loss of ambulation for patients with LGMD2I/R9
 - Lauren Coffey presentation at Ask the Doctor, FKRPP related

Questions?

(email, use Whova or ask any time during the weekend!)

- Katherine-mathews@uiowa.edu
- Please complete conference evaluations through the Whova app
 - Paper copies available for Iowa City attendees if preferred

