

Characterization of Clinical Skeletal and Cardiac Impairment in Carriers of DMD and BMD

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Study Team

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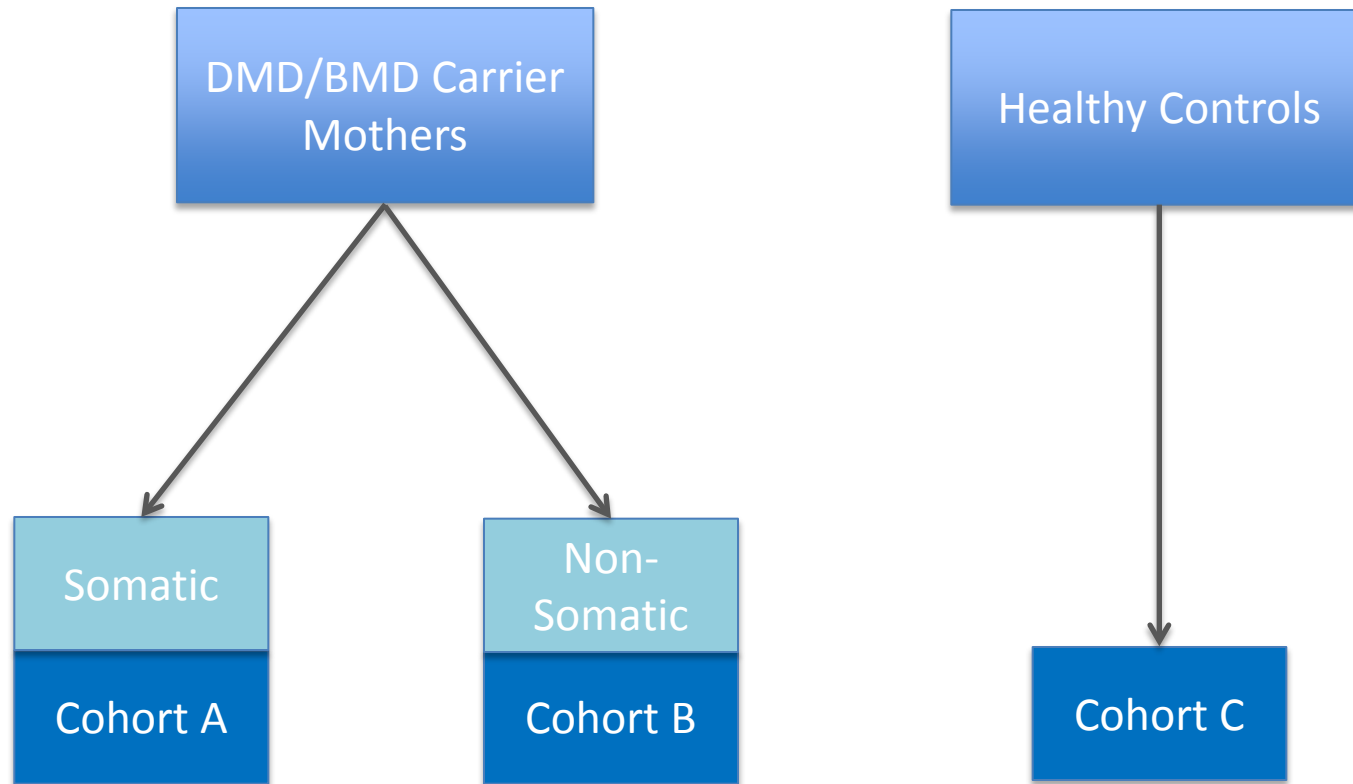
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Study Population

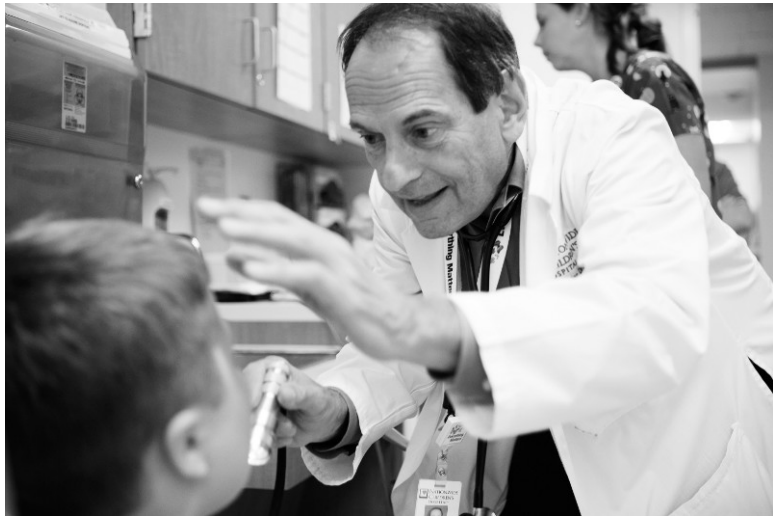


Study Design

- Longitudinal observational study
 - Baseline, 12-month, 24-month visits
- Cardiology
 - Determine prevalence and progression of cardiac involvement in somatic carriers
- Neuromuscular
 - Assess for evidence of skeletal muscle involvement
- Biobehavioral
 - Elucidate consequences of stress for genetic carriers

Study Design - Neuromuscular

- Physical exam
 - Patient medical history
 - Motor strength testing
- Physical Therapy
 - 6 Minute Walk Test
 - Quantitative Myometry
 - Exploratory measures
 - “Time-to-Rise”
 - ACTIVE-seated



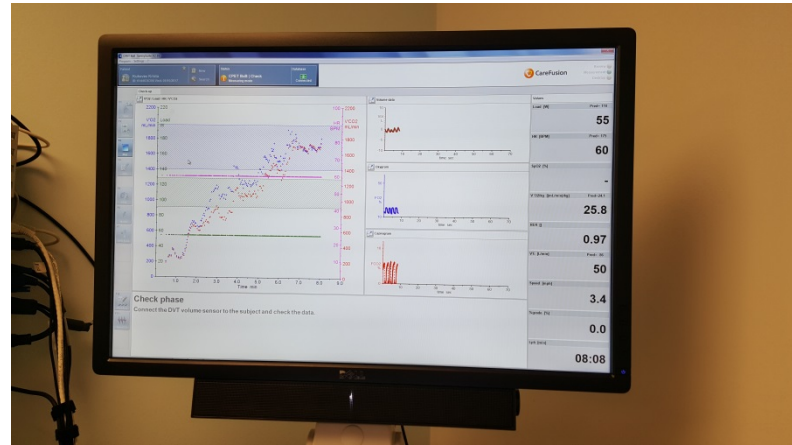
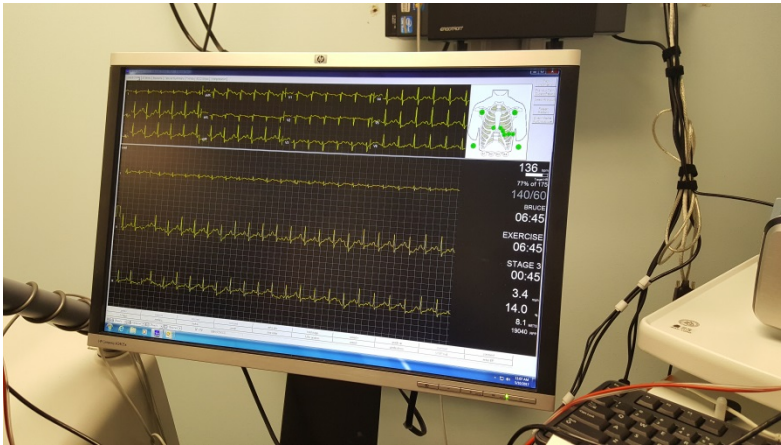
Study Design - Cardiology

- Treadmill Stress testing with Electrocardiogram
 - Measure effectiveness of oxygen use of the body
 - EKG continuously recorded with BP measured every 2 minutes
- Cardiac Magnetic Resonance Imaging
 - 30 minutes of cardiac imaging without dye
 - 15 minutes of imaging with gadolinium contrast

Interim Results - Cardiology



1. Set-up: EKG, Face Mask
2. Obligatory Selfie/Facebook Post
3. Exercise to Exhaustion
4. Data Collection



Courtesy May Ling Mah, MD

Study Design - Biobehavioral

- Collection of biomarkers for disease and stress burden
 - Hair cortisol, interleukin 6, C-reactive protein
 - Online survey to identify patients perceived stress
- Qualitative interview with mothers identifying themes in their caregiving experiences
- Cognitive evaluation to determine understanding, awareness and comprehension

What your typical visit looks like...

- Day 1

10:30am – Biobehavioral

- Qualitative Interview
- Hair Cortisol Collection
- Cognitive Evaluation

1:00pm – Neuromuscular

- Medical History
- Manual Muscle Testing

2:00pm – Physical Therapy

- Quantitative Myometry
- 6MWT
- ‘Time to Rise’ and Active-seated

- Day 2

6:30am – Cardiac
MRI

8:00am – Stress
Test with EKG

If interested in participating contact:

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Thank You

Neuromuscular

- Samiah Al-Zaidy
- Jerry Mendell
- Linda Lowes
- Kelly Lehman
- Lindsay Alfano
- Natalie Miller
- Megan Iammarino
- Kathleen Church
- Stephanie Diemer

Cardiology

- May Ling Mah
- Linda Cripe
- Kan Hor
- Houchun Hu
- Ramkumar Krishnamurthy
- Michelle Slawinski

Biobehavioral

- Jamie Jackson
- Kathryn Vannatta
- Kathryn Balistreri
- Jennifer Cotto
- Mary Hagan
- Carine Leslie

Parent Project Muscular Dystrophy **JOIN THE FIGHT. END DUCHENNE.**

