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

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Study Team

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Study Population

DMD/BMD Carrier Mothers

- Somatic Cohort A
- Non-Somatic Cohort B

Healthy Controls

Cohort C
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

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