

# SMA Research Update – Providing Hope!

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## Currently, there are (7) active studies within Columbia University Medical Center:

### Oxidative Capacity and Exercise Tolerance in Ambulatory SMA

**\*\*Active and recruiting\*\***

Spinal Muscular Atrophy (SMA) is a progressive, recessively-inherited neuromuscular disease characterized by weakness and muscle atrophy due to the loss of spinal cord motor neurons. The results from this study would provide preliminary data, using non-invasive methods, on oxidative capacity in ambulatory SMA patients and disease controls to aid in the design of exercise intervention studies. Furthermore, this information would link previous laboratory and preclinical findings of mitochondrial depletion in SMA to the clinical condition and provide important information for future studies designed to improve oxidative capacity and fitness in SMA patients.

#### Eligibility

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<b>Ages Eligible for Study:</b>	8 Years to 55 Years (Child, Adult)
<b>Sexes Eligible for Study:</b>	All
<b>Accepts Healthy Volunteers:</b>	Yes
<b>Sampling Method:</b>	Non-Probability Sample

#### Study Population

The study sample will include 14 ambulatory SMA patients, 14 ambulatory mitochondrial myopathy patients, and 14 healthy controls.

#### Criteria

Inclusion Criteria:

1. One of the following categories:
  - o Genetic confirmation of SMA with laboratory documentation of homozygous deletion of SMN1 exon 7;
  - o Genetic confirmation of mitochondrial myopathy or evidence from muscle biopsy confirming the diagnosis; or
  - o Healthy individuals.
2. Able to walk independently at least 25 meters, and able to tread a stationary cycle ergometer.

Exclusion Criteria:

1. Unable to walk 25 meters independently.

2. Use of investigational medications intended for the treatment of SMA within 30 days prior to study entry.
3. The presence of any contraindication to exercise according the ACSM criteria.

**Primary Outcome Measures:**

- Change in NIRS derived index of muscle oxygen extraction [ Time Frame: baseline, 6 months ]  
Near Infrared Spectroscopy (NIRS) is a simple, non-invasive method to measure oxygen in muscle and other tissues in vivo.

**Secondary Outcome Measures:**

- Change in Peak oxygen uptake (VO<sub>2</sub> max) [ Time Frame: baseline, 6 months ]  
Participants will undergo an exercise stress test performed by a clinical exercise physiologist using an electronically-braked recumbent cycle ergometer to determine peak oxygen uptake (VO<sub>2</sub> max).
- Change in Distance walked during the Six Minute Walk Test (6MWT) [ Time Frame: baseline, 6 months ]  
6MWT is an objective evaluation of functional exercise capacity, measures the maximum distance a person can walk in six minutes over a 25-meter linear course.
- Change in Lean body mass assessed with Dual Energy X-ray Absorptiometry (DEXA) [ Time Frame: baseline, 6 months ]  
Dual-Energy X-ray Absorptiometry (DEXA) is a method of estimating bone and lean body mass by comparing the absorption of two distinct energy level beams at 46.8 keV and 80 keV, which are effective at differentiating soft tissue and bone. A standard DEXA scan will be performed in supine.

Estimated Enrollment: 42

Study Start Date: November  
2016

Estimated Study Completion Date: May 2020

**If you are interested in participating, please contact Jackie Montes at 212-342-5767 or [jm598@columbia.edu](mailto:jm598@columbia.edu).**

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