PROJECT TITLE: **Improving skeletal muscle function in distal arthrogryposis**

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**Project Description**

1. Area of the research  
Distal arthrogryposis is an autosomal dominant congenital anomaly characterized by contractures of the distal regions of the hands and feet with no facial involvement or any additional anomalies prior to birth. Distal arthrogryposis type 1 affects an estimated 1 in 10,000 people worldwide. Distal arthrogryposis is related to decreased fetal movement during development which can be caused by environmental factors, single gene changes, chromosomal abnormalities and various syndromes. Treatment depends on the signs and symptoms found in each person, but it may include physical therapy, removable splints, exercise, and/or surgery. Research studies involve characterizing skeletal muscle function by determining muscle-specific gene and protein expression and measuring skeletal muscle function in vitro.

2. Research tasks the student will be performing  
Undergraduate students will perform DNA and RNA extraction, real-time polymerase chain reaction, agarose gel electrophoresis, protein isolation, quantitation, standard graph creation and Western blot analysis. Furthermore, students will use skinned muscle strips from mouse model of distal arthrogryposis to measure skeletal muscle function in vitro using the Aurora
Scientific 1300A 3-in-1 System. Briefly, the muscle of interest is surgically removed and placed in an oxygenated and temperature-controlled bath to maintain viability. One tendon is anchored to a fixation clamp, while the other tendon is sutured onto a dual-mode lever. The muscle is then stimulated by field electrodes fixed inside the bath to measure muscle contractility. This technique requires two weeks of training prior to performing the experiments.

3. Training that the mentor will provide to the WISE student
The objective of the twelve-week Women in Science and Engineering (WISE) Program is to expose undergraduate women to skeletal muscle research. The skeletal muscle system is responsible for movement of the human body, posture, movement of substances inside the body and generation of body heat. Therefore, studying skeletal muscle physiology in health and disease is essential for those contemplating careers in skeletal muscle biology. We hypothesize that undergraduate students fill these positions, as the upcoming innovators who will bring new ideas and technologies to improve skeletal muscle function in distal arthrogryposis. As proposed in this study, students will be exposed to systematic and critical thinking, planning, teamwork, experimental design and documentation thereof. They will learn how to review the pertinent literature, ask relevant questions, and develop an appropriate hypothesis. They will learn how to perform experiments in a rigorous and reproducible manner. In addition, students will be asked to attend research seminars and present the results of their studies at lab meetings. Overall, the WISE training plans will include orientation, hands-on research experience, weekly discussions of projects, weekly journal club, workshops on pertinent research tools, and poster presentations at the one-day local College of Medicine symposium.

4. Specific requirements, if any, that the mentor expects the student to meet
We prefer students who are highly motivated and interested in skeletal muscle research which is another focus of our research group.