

Muscular Dystrophy Association (MDA) Studies

The MDA studies are supported by a gift from the Muscular Dystrophy Association to the Department of Pediatrics and include two projects led by Dr. Steven Lipshultz, Chair of the Pediatrics department and Dr. Tracie Miller, Chief of the division of Pediatric Clinical Research (DPCR). Muscular Dystrophy (MD) is a group of inherited neuromuscular disorders in which strength and muscle bulk gradually decline. It is typically characterized by progressive weakness of the skeletal muscles followed by weakness of the heart and other muscles.

The first project is a multi-site study involving youth with Duchenne (DMD) and Becker muscular dystrophy (BMD), two of the most common forms of muscular dystrophy. The study proposes to evaluate the genetic tests and their associations with clinical outcomes affecting skeletal and cardiac muscle of these patients. To evaluate cardiovascular outcomes of DMD/BMD patients from five sites across the US, it is important to appropriately measure, read, and interpret the echocardiograms of the patients. The University of Miami (UM) is one of the sites included in the study. In addition, UM will be the central coordinating site responsible for reading serial echocardiogram results of patients from other sites using the equipment and software that are housed in the UM Division of Pediatric Clinical Research (DPCR). Having a central reading site and having all echocardiogram measures read by a local cardiologist allows excellent quality control of study data.

The second project is a 12-week exercise intervention study involving youth with DMD/BMD who are being followed by a team of experts (neurologist, cardiologist, geneticist, physical therapist) at UM clinics. The primary objective of this study is to increase cardiorespiratory fitness, muscle strength, and endurance among DMD/BMD patients using the structured exercise intervention supervised by a trained exercise physiologist and monitored by a team of physicians caring for these patients.

This study also proposes to measure their body growth and nutrition, body composition, and the activities of daily living (ADL). The DPCR has equipment available to measure body composition (dual-energy x-ray absorptiometry or DEXA), cardiorespiratory fitness (treadmill, spirometry from a metabolic cart), and muscle strength (dynamometer). Additionally, the DPCR laboratory has exercise equipment appropriate for youth with neuromuscular diseases. Patients will be measured before and after 12-weeks of study participation. Although the initial study is proposed to be 12-weeks long, we plan to monitor these patients to assess the long-term effects of exercise on their health and neuromuscular conditions. The exercise laboratory and equipment at DPCR are available to the patients for their use even after completion of the study.