

Research participation opportunity

Research study title:

Extracellular RNA biomarkers of myotonic dystrophy

Participating investigators and research facilities:

Thurman Wheeler, MD, Massachusetts General Hospital (MGH) in Boston, MA

Anthony Amato, MD, Brigham and Women's Hospital in Boston, MA

Seward Rutkove, MD, Beth Israel Deaconess Medical Center in Boston, MA

Courtney McIlduff, MD, Beth Israel Deaconess Medical Center in Boston, MA

Purpose of the research:

Current methods of measuring the response to new treatments for muscular dystrophies involve the examination of small pieces of muscle tissue called biopsies. We are interested in finding less invasive methods that reduce the need for muscle biopsies. The purpose of this research is to learn about the possibility of detecting and measuring the activity and severity of muscular dystrophies by examining a urine sample and a blood sample.

Eligibility criteria:

Males and females ages 5 years and older with myotonic dystrophy type 1 (DM1) or myotonic dystrophy type 2 (DM2) confirmed by genetic testing or by clinical history and examination are invited to participate.

In addition, male and female healthy volunteers ages 18 and older also are invited to participate.

Research study activities:

This study consists of three separate Groups. Eligible volunteers may choose to participate in one, two, or all three Groups of this study.

Group 1: we will ask eligible volunteers to provide a single urine sample and undergo a single blood draw.

Group 2: we will ask eligible volunteers to provide a urine sample, undergo a blood draw, and undergo standard muscle function tests once every six months over a two-year period.

Group 3: we will ask eligible volunteers to provide a urine sample and undergo a muscle biopsy.

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Duration of participation in the research study:

Group 1: We will ask you to make one visit to MGH, BWH, or BIDMC where it will take about 10 - 15 minutes for you to complete the study.

Group 2: It will take about two years to complete the study. During this time we will ask you to make five visits to MGH or BWH. Each visit will take about four hours to complete.

Group 3: It will take about one month to complete the study. During this time we will ask you to make three study visits to MGH. The first visit will take about one hour, the second visit will take about four hours, and the third visit 30 minutes or less.

Benefits of participation:

You will have no direct benefit from participation in this research. However, others that have myotonic dystrophy may benefit in the future from what we learn in this research study.

Payment for participation:

Group 1: There is no payment for participation.

Group 2: Individuals will be paid \$75 for each visit, or \$375 for completion of all five visits.

Group 3: Individuals will be paid \$175 for participation.

Contact persons for more information:

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