Partners Human Research APPROVAL EFFECTIVE DATE 7/29/2020

Research participation opportunity

Research study title:

Biomarker development for muscular dystrophies

Participating investigators and research facilities:

Thurman Wheeler, MD, Massachusetts General Hospital (MGH) in Boston, MA Paloma González-Pérez, Massachusetts General Hospital (MGH) 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 muscle diseases by examining a urine sample, a blood sample, and some muscles in your arms and legs using tests called ultrasound and electrical impedance myography; both tests are painless and non-invasive. The information that we gather from this study may help us evaluate, prevent, diagnose, treat, and improve our understanding of human muscle diseases.

Eligibility criteria:

Males and females ages 14 years and older with myotonic dystrophy type 1 (DM1), myotonic dystrophy type 2 (DM2), Duchenne muscular dystrophy (DMD), Becker muscular dystrophy (BMD), or facioscapulohumeral muscular dystrophy (FSHD) confirmed by genetic testing or clinical history and examination are invited to participate.

In addition, male and female healthy volunteers ages 18 and older who are family members of an individual with muscular dystrophy also are invited to participate.

Research study activities:

We will ask eligible volunteers to provide a single urine sample and undergo a single blood draw. We will clean the skin over the muscles that we want to study by ultrasound and electrical impedance myography. Each muscle will take about four or five minutes to study.

Duration of participation in the research study:

We will ask you to come in for one visit to MGH where it will take about 60 - 90 minutes for you to complete the study.

Benefits of participation:

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

Payment for participation:

After the visit, we will pay you \$50 as reimbursement for time, inconvenience, parking, and meals.

Contact persons for more information:

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