Estab Biomarkers and Clinical Endpoints in Myotonic Dystrophy Type 1 (END-DM1)

Status: RECRUITING

Eligibility Criteria

Age: 18 years to 70 years old

This study is NOT accepting healthy

Healthy Volunteers: volunteers

Inclusion criteria: * Age 18 to 70 (inclusive) * Competent to provide informed consent * Clinical diagnosis of DM1 based on research criteria1 or positive genetic test * Comment: The clinical research criteria require myotonia, muscle weakness in a characteristic distribution, and history of similar findings in a first degree relative. Genetic testing confirmed the diagnosis of DM1 in \> 99% of individuals who satisfied these criteria.2 Exclusion criteria: * Symptomatic renal or liver disease, uncontrolled diabetes or thyroid disorder, or active malignancy other than skin cancer. * Current alcohol or substance abuse * Concurrent enrollment in clinical trial for DM1, or participation in trial within 6 months of entry. * Concurrent pregnancy or planned pregnancy during the course of the study. * Concurrent medical condition that would, in the opinion of the investigator or clinical evaluator, compromise performance on study measures. * Note: non-ambulatory participants are not excluded, but are limited to \<15% of enrollment. Inclusion criteria for participants in the muscle biopsy sub-study: • Of the 95 patients undergoing the tibialis anterior muscle biopsy, at least half will have at least moderate weakness of ankle dorsiflexion, defined as MRC score ≤ 4+. This is in order to obtain a muscle tissue sample in a person more severely affected with myotonic dystrophy. Approximately 10 patients at each site will undergo the muscle biopsy. Exclusion criteria for 95 participants in the muscle biopsy sub-study: * Known CTG repeat expansion size less than 100 repeats, unless there are clear cut signs of limb weakness and muscle wasting. This is in order to obtain a muscle tissue sample in a person more severely affected with myotonic dystrophy. * Use of anticoagulant such as warfarin or a direct oral anticoagulant (e.g. dabigatran) due to the increased risk of bleeding. * Use of aspirin or non-steroidal anti-inflammatory agents should be discontinued 3 days prior to the biopsy procedure, if possible. * Platelet count \<50,000 (if known) due to the increased risk of bleeding. * History of a bleeding disorder due to the increased risk of bleeding. * Advanced wasting of tibialis anterior (TA) muscle that precludes needle muscle biopsy in order to ensure that a sample taken would be of muscle and not just fat and fascia. * Previous muscle biopsy of either TA in order to provide muscle tissue samples of non-biopsied muscles.

Conditions & Interventions

Conditions:

Myotonic Dystrophy 1, DM1

Keywords:

Myotonic Dystrophy, END DM-1, Muscular Dystophy, DMCRN

More Information

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