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

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 years (inclusive) * Written, voluntary informed consent must be obtained prior to any study procedures. In cases where a Legally Authorized Representative (LAR) provides consent, verbal assent will be obtained from the subject, as determined by the investigator and documented directly on the consent form. Capacity to consent will be determined by the neurologist at the Baseline visit and will be signed off on the Inclusion/Exclusion checklist. * Clinical diagnosis of DM1 based on research criteria or positive genetic test. The research criteria for clinical diagnosis of DM1 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. OR A diagnosis of CDM, which is defined as children having symptoms of myotonic dystrophy in the newborn period (\<30 days), such as hypotonia, feeding or respiratory difficulty, requiring hospitalization to a ward or to the neonatal intensive care unit for 3 days or more; and a genetic test suspicious of an expanded trinucleotide (CTG) repeat in the DMPK gene in the child or first degree relative. An expanded CTG repeat size in the child is considered greater than 200 repeats or E1-E4 classification (E1= 200-500, E2=500-1,000, E3=1,000-1,500, E4\>1,500)

Exclusion Criteria:

* Symptomatic renal or liver disease, uncontrolled diabetes or thyroid disorder, or active malignancy other than in situ skin cancer. * Current alcohol or substance use disorder. * 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. * Use of mexiletine or other anti-myotonia agents within 72 hours of any study visit.

Conditions & Interventions

Conditions:
DM1, Myotonic Dystrophy, Myotonic Dystrophy 1, Myotonic Dystrophy Type 1, Myotonic Dystrophy Type-1, Myotonic Dystrophy, Type 1 (DM1), Myotonic Muscular Dystrophy

Keywords:
DM1, END-EXT, END-DM1 Extension, Myotonic Dystrophy Type 1, Steinert's Disease, Muscular Dystrophy, Neuromuscular Disease, DMPK, Natural History, Myotonia, DMCRN, Myotonic Dystrophy Clinical Research Network

More Information

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