Trial Readiness and Endpoint Assessment in Pediatric Myotonic Dystrophy Extension

Status: RECRUITING

Eligibility Criteria

Age: 3 years to 17 years old

This study is NOT accepting healthy

Healthy Volunteers: volunteers

Inclusion Criteria (Congenital Myotonic Dystrophy Group): * Age 5-17 years, 11 months at enrollment. Lower age limit not applicable for participants who have completed ASPIRE-DM1 protocol. Upper age limit not applicable for participants who previously participated in TREAT-01-001 (TREAT-CDM) study * A diagnosis of CDM, 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 more than 72 hours; and a genetic test confirming an expanded trinucleotide (CTG) repeat in the DMPK gene in the child or mother. 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). * Written, voluntary informed consent must be obtained before any study related procedures are conducted. Inclusion Criteria (Childhood Myotonic Dystrophy Group): * Age 3-17 years, 11 months at enrollment. Upper age limit not applicable for participants who previously participated in TREAT-01-001 (TREAT-CDM) study. * A diagnosis of ChDM, defined as: children having cognitive deficits, muscle weakness, myotonia that developed after age 1 and prior to age 10 and a genetic test confirming an expanded trinucleotide (CTG) repeat in the DMPK gene in the child or mother. 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). * Written, voluntary informed consent must be obtained before any study related procedures are conducted.

Exclusion Criteria:

* Any other non-DM1 illness that would interfere with the ability to undergo safe testing or would affect the interpretation of the results, in the opinion of the site investigator * Significant trauma within the past month * Internal metal or devices (exclusion for DEXA component) * Use of anticoagulants, such as warfarin or a direct oral anticoagulant (e.g., dabigatran) due to the increased risk of bleeding with biopsy * Platelet count \<50,000 * History of a bleeding disorder * Participation in a clinical trial involving an investigational product * History of adverse reaction to lidocaine (if participating in muscle biopsy)

Conditions & Interventions

Conditions:

Congenital Myotonic Dystrophy, Childhood Myotonic Dystrophy, Myotonic Dystrophy

Keywords:

DM1, Childhood Myotonic Dystrophy, Congenital Myotonic Dystrophy, Myotonic Dystrophy, Myotonia, Dystrophy Myotonic, DMCRN, TREAT-EXT, TREAT CDM

More Information

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Phase:

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