Cerebrospinal Fluid Biomarkers of Myotonic Dystrophy

NCT06075693

Status	RECRUITING
Sponsor	Massachusetts General Hospital
Enrollment	88 participants

Key Eligibility Criteria

Inclusion (4)

- Subjects with DM1 based on genetic testing and/or clinical criteria (some subjects who have positive genetic testing may be asymptomatic, while other subjects who show characteristic clinical features may have declined to have genetic testing done).
- Unaffected subjects are unknown to have myotonic dystropphy or any other muscular dystrophy by history and may have had no genetic testing.
- Clinical indicators of current status, as measured within 30 days of study start: Able to provide informed consent or assent for participation in the study.
- Demographic characteristics (e.g., biologic sex, age): Males and females age 18 years and older.

Exclusion (5)

- Medical history of any of the following. State of immunosuppression; pre-existing liver or kidney disease; documented HIV positive; documented hepatitis B and/or C positive.
- Medications and other drugs. Use of anticoagulants within 60 days prior to lumbar puncture and/or blood draw. Use of anti-platelet drugs within 7 days prior to blood draw.
- Contraindications to MRI. The presence of any metal within the body, which would include any medical device containing metal, such as a pacemaker, defibrillator, some heart valves or stents, artificial joint, aneurysm clip, or inner ear device, a history of working with sheet metal, or an injury with metal shrapnel; pregnancy, due to effects of MRI on unborn children.
- Contraindications to Lumbar Puncture. Evidence of increased intracranial pressure or active infection on exam; platelets less than 50,000.
- Other. Inability or unwillingness of the subject to give written informed consent.

Locations (1 total)

Massachusetts General Hospital, Boston, Massachusetts, United States