

Clinical Manifestations and Biomarkers in Amyotrophic Lateral Sclerosis Type 4 and Other Inherited Neurological Disorders of RNA Processing

NCT04394871

Status	RECRUITING
Sponsor	National Institute of Neurological Disorders and Stroke (NINDS)
Enrollment	330 participants

Key Eligibility Criteria

Inclusion (19)

- Age 5 or above
 - Genetic diagnosis of ALS4 (heterozygous mutation in SETX)
 - Able to communicate well with the investigator, to understand and comply with the requirements of the study
 - Capacity to consent (adults) or assent (pediatric subjects) to the study
 - Age 5 or above
- ... and 14 more (see full listing online)

Exclusion (29)

- Patients with known claustrophobia, presence of pacemaker, ferromagnetic material in their body, or any other condition that would preclude MRI assessments
 - Pregnancy
 - Note: An Adult RNA metabolism ALS4 Patient who meets any of the following criteria will be excluded from the lumbar puncture procedure:
 - PT/PTT values that are prolonged greater than or equal to 3 seconds from the upper limit of normal (including treatment with oral and parenteral anticoagulants)
 - INR greater than or equal to 1.5, thrombocytopenia ($<70,000$), or abnormal bleeding time or platelet dysfunction
- ... and 24 more (see full listing online)

Locations (1 total)

National Institutes of Health Clinical Center, Bethesda, Maryland, United States