

LGMD R1 Natural History Study

NCT05618080

Status	RECRUITING
Sponsor	Virginia Commonwealth University
Enrollment	100 participants

Key Eligibility Criteria

Inclusion (4)

- Age between 12-50 at enrollment
- Clinically affected (defined as weakness on bedside evaluation in a pattern consistent with LGMDR1)
- Genetic confirmation of LGMDR1 (presence of homozygous or compound heterozygous pathogenic mutations in CAPN3).
- Must be able to provide written informed consent and be willing and able to comply with all study requirements. Note: Adult participants must be able to provide consent themselves. Legally authorized representatives are not permitted to consent on behalf of adult participants.

Exclusion (5)

- Have contraindications to MRI or MRS (e.g., non-MR compatible implanted medical devices or severe claustrophobia)
- Non-ambulatory as defined by those who are not able to walk 10 meters without assistive devices (ankle foot orthotics excluded)
- Positive pregnancy test at any timepoint during the trial
- Have dominantly inherited CAPN3 mutations (LGMD4)
- Any other illness that would interfere with the ability to undergo safe testing or would interfere with interpretation of the results in the opinion of the site investigator.

Locations (12 total)

University of California, Irvine, Orange, California, United States
University of Colorado Anschutz Medical Campus, Aurora, Colorado, United States
University of Florida, Gainesville, Florida, United States
... and 9 more locations