LGMD R1 Natural History Study

Status: RECRUITING

Eligibility Criteria

Age: 12 years to 50 years old

This study is NOT accepting healthy

Healthy Volunteers: volunteers

Inclusion Criteria:

1. Age between 12-50 at enrollment 2. Clinically affected (defined as weakness on bedside evaluation in a pattern consistent with LGMDR1) 3. Genetic confirmation of LGMDR1 (presence of homozygous or compound heterozygous pathogenic mutations in CAPN3). 4. 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 Criteria:

1. Have contraindications to MRI or MRS (e.g., non-MR compatible implanted medical devices or severe claustrophobia) 2. Non-ambulatory as defined by those who are not able to walk 10 meters without assistive devices (ankle foot orthotics excluded) 3. Positive pregnancy test at any timepoint during the trial 4. Have dominantly inherited CAPN3 mutations (LGMDD4) 5. 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.

Conditions & Interventions

Conditions:
Calpain-3 Deficiency Limb Girdle Muscular Dystrophy Type 2A, Limb Girdle Muscular Dystrophy, Limb Girdle Muscular Dystrophy Type R1, LGMD2A
Keywords:
LGMD, Limb Girdle Muscular Dystrophy, LGMD R1, LGMD2A, CAPN3

More Information

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