Observation Study in Patients Age 0-5 Years With LAMA2-related Congenital Muscular Dystrophy

Status: RECRUITING

Eligibility Criteria

Sex: ALL
Age: Up to 5 years old
Healthy Volunteers: This study is NOT accepting healthy volunteers
Inclusion Criteria:

- Signed informed consent by the subject, parent(s) or legally authorized representative (LAR) and/or assent by the subject (when applicable).
- Subject must be aged birth to less than 5.0 years of age at time of consent.
- A confirmed diagnosis of LAMA2-RD confirmed via:

a: Two pathogenic variants in the LAMA2 gene (via a CLIA-approved laboratory) or: b. muscle biopsy with absence of merosin (laminin-211) and at least one pathogenic variant in the LAMA2 gene
* Absence of another confirmed genetic disease.
* Willingness to maintain current exercise and/or physical therapy regimen for the duration of the clinical study.
* Willingness to comply with the study protocol, including but not limited to, all study procedures and visits.

Exclusion Criteria:

- Acute medical illness or hospitalization within 30 days prior to informed consent.
- Participation in a previous trial of any investigational agent for LAMA2-RD within 1 month prior to informed consent, or use of any other investigational therapy (including off-label use of Losartan) within 30 days prior to informed consent, or participation in other clinical studies, within 30 days (or 3 half-lives, whichever is longer) prior to informed consent, which in the opinion of the PI, may potentially confound results from this study.
- Other significant medical condition, which in the opinion of the site Principal Investigator may confound interpretation of the clinical course of LAMA2- RD.

Conditions & Interventions

Conditions:
LAMA2-MD \(Merosin Deficient Congenital Muscular Dystrophy, MDC1A\)
Keywords:
Early Phase 1, Observational, Natural History, Neuromuscular, Functional Assessments

More Information

Description:
The goal of this observational study is to understand how young children with LAMA2-related dystrophy move and change over time. We will also learn about how this condition impacts other body systems. Participants will undergo: * Neuromuscular assessments * Blood collections * Swallowing and breathing assessments * Questionnaires

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