A Study to Check Liver Health in Boys With XLMTM, a Serious Genetic Muscle Condition

Status: RECRUITING

Eligibility Criteria

Sex: MALE
Age: Up to 17 years old
Healthy Volunteers: This study is NOT accepting healthy volunteers
Inclusion Criteria:

- Participant has a diagnosis of XLMTM resulting from a genetically confirmed mutation in the MTM1 gene based on genetic test reports.
- Participant requires some mechanical ventilatory support (e.g., ranging from 24 hours per day full-time mechanical ventilation, to non-invasive support such as continuous positive airway pressure (CPAP) or bilevel positive airway pressure (BiPAP) during sleeping hours)
- Participant (as applicable) and/or parent(s)/carer is willing to comply with the recommended schedule of assessments.

Exclusion Criteria:

- Participant is currently enrolled in an interventional study designed to treat XLMTM.

Conditions & Interventions

Interventions:
OTHER: No Intervention
Conditions:
X-Linked Myotubular Myopathy
Keywords:
XLMTM, Hepatobiliary

More Information

Description:
XLMTM (X-linked myotubular myopathy) is a serious genetic muscle condition. It is caused by changes in the MTM1 gene which stops or slows down normal muscle development, causing severe muscle weakness. There is currently no cure for XLMTM. Ongoing care is needed to manage symptoms and prevent further medical problems from this condition. Recent research shows that individuals with XLMTM often have reduced bile flow which can affect liver and gallbladder health. Bile is a liquid made in the liver that helps digest fat. Ongoing liver health checks may help with the routine care of people with XLMTM. There is a need to understand liver problems that develop in individuals with XLMTM over time. The main aim of the study is to learn how many boys with XLMTM have new cases of liver problems during the study. This study is about collecting information only. This is known as an observational study. The individual's doctor decides on treatment, not the study sponsor (Astellas). In this study, boys under 18 diagnosed with XLMTM will be followed for about 1 year. The health of their liver and gallbladder will be checked about every 6 weeks. This can be done at home, if preferred. A scan called a Fibroscan (also known as transient elastography) will check for signs of scarring in the liver (fibrosis) and the build-up of lipids. It is suggested that each boy will have a Fibroscan when they start the study and another scan when they complete the study. This study will help understand liver, gallbladder, and bile duct issues in individuals with XLMTM over time. The goal is to improve their care and provide information to use in future clinical studies.

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