

X-linked myotubular myopathy (XLMTM, or commonly referred to as MTM) is a rare genetic disorder that primarily affects muscle strength and function and can cause muscle weakness, difficulties in breathing, and developmental delays. Recent evidence from natural history and gene therapy studies shows that there are problems with the liver in some people with MTM. However, there is limited understanding of these liver-related health issues.

The Epidemiologic Study of XLMTM and Clinical Expression in the Liver (EXCEL) study (NCT06581146), which is sponsored by Astellas Gene Therapies, is designed to gain a better understanding of the effect of MTM on the liver.

### Overview of EXCEL:

- Observational, prospective study (participants and certain outcomes will be observed for period of time) using liver-focused assessments
- No specific therapeutic treatment or medicine will be given
- Study duration is about 1 year
- Approximately 50 male participants with XLMTM will be enrolled at specialist sites in the United States, Canada, and the United Kingdom



Credit: Zak Hughes, 2024

### Inclusion Criteria:

- Participant has a diagnosis of XLMTM resulting from a genetically confirmed mutation in the *MTM1* gene
- Participant is male
- Participant is <18 years of age at enrollment
- Participant requires some mechanical ventilatory support (e.g., ranging from 24 hours per day full-time to non-invasive support, such as CPAP or BiPAP during sleeping hours)
- Participant or guardian is willing to provide written informed consent
- Participant or guardian is willing to comply with the recommended schedule of assessments

### Exclusion Criteria:

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

The study is designed in a way to make it easier for patients to participate. The health of their liver and gallbladder will be checked about every 6 weeks. Some of these tests and data can be collected at home. Data from EXCEL will increase our understanding of liver health in those with MTM and can help to improve current treatment practices and future potential clinical study designs for investigational treatments.

Enrollment for EXCEL will be for patients with XLMTM meeting the inclusion criteria based in the United States, United Kingdom or Canada. For further queries, please email [medinfo.us@astellas.com](mailto:medinfo.us@astellas.com) in the United States or [medinfo.est-m@astellas.com](mailto:medinfo.est-m@astellas.com) in the United Kingdom or Canada.