Novel therapy for neuromuscular disease caused by mutations in myotubularins

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SUMMARY

Mutations in myotubularin1 affect 1:50,000 newborn males and cause a severe muscle disorder (XLMTM) which is characterized by severe generalized muscle weakness with ventilator, wheelchair and feeding tube dependence in addition to dramatically reducing survival.
Despite it's severity, there is no treatment yet. This project aims at utilizing a novel target for the treatment of this disease.
By screening an in-house library and optimizing hits via medicinal chemistry, the team aims at finding and developing the first small molecule inhibitor for the treatment of this fatal disease.

PROJECT GOALS

- Identification and generation of lead compounds
- *In vitro* and *in vivo* proof-of-concept studies
- Hit-to-lead optimization

LONG-TERM GOALS

- Develop first small molecule inhibitor as treatment for XLMTM
- Licensing to Pharma company