Progress Update
Key Achievements To Date

PROMOT
Performing a Rare Disease-Oriented
Master Observational Trial

1 Mapping the Data Landscape: An Inventory of Existing Data Sources

Understanding what data already exists – and how it can be used – is a vital first step in our mission. Over the past year, the PROMOT team has:

- Completed two comprehensive scoping reviews focused on Congenital Myasthenic Syndromes (CMS) and Congenital Myopathies (CM). These reviews helped identify the most common and relevant signs and symptoms in each disease area.
- Conducted targeted surveys and consultations with clinical experts in Oculopharyngeal Muscular Dystrophy (OPMD), CMS and CM to validate the findings and ensure clinical relevance.
- Developed the first version of a common data model, linking key clinical concepts to existing ontologies.

2 Deepening Stakeholder Engagement

PROMOT places strong emphasis on co-design and inclusivity, ensuring that those most affected by rare diseases have a voice in shaping research priorities.

- We have established a dedicated Patient Advisory Board, bringing together individuals with lived experience and representatives from patient organizations.
- Regular dialogue with clinicians, researchers, and advocacy groups has created a strong feedback loop, ensuring PROMOT remains grounded in real-world needs and priorities.

3 Strengthening Visibility and Advocacy

To build a movement, we must communicate clearly and effectively. Over the past year, we have:

- Launched the PROMOT website and activated our social media channels (LinkedIn and Facebook), creating vibrant platforms to highlight project milestones, showcase partner contributions, and promote public awareness of rare diseases.
- Published project news and milestones to reach new audiences, foster transparency, and encourage involvement from stakeholders across sectors.

Looking Ahead

- **Scaling our digital infrastructure and refining the LEAP FORWARD platform**
- **Launching pilot studies**
- **Hosting our second Consortium Meeting in 2026**

Stay connected with us for the latest updates on events, publications, and collaboration opportunities:

promotinitiative.com