

## **Development of a Patient-Reported Outcome Questionnaire for Patients with Fatty Acid Oxidation Disorders**

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**Background:** Individuals with fatty acid oxidation disorders (FAOD) experience muscle symptoms due to their disease that can vary in type and frequency and can impact physical function and health-related quality of life (HRQoL). Patient-reported outcome questionnaires (PROs) can be useful to assess symptoms, and associated impact and treatment response. Available validated PROs for pain, muscle fatigue, and HRQoL may not adequately capture all the relevant muscular symptoms in patients with FAOD.

**Objective:** To understand from the patient perspective the important FAOD symptoms and their impact on daily activity and HRQoL and determine the suitability of existing PROs or need for a novel PRO.

**Methods:** An interview guide was developed to assess symptoms and impacts associated with FAOD. Individuals with established FAOD were invited through a support group to participate in an in person or telephone interview. Saturation grids identified the point at which no new concepts were reported. Elicited concepts were mapped to items in existing PROs. The study received IRB approval.

**Results:** 11 patients ages 15-43 with LCHAD, CPTII or VLCAD were enrolled. All experienced muscle weakness, muscle fatigue, longer recovery time from illness and vision problems. Other symptoms included muscle aches/pains, extreme fatigue (91%), and difficulty or inability to exercise (82%). 90% said their symptoms impacted others; 67% said their work was impacted by symptoms. Saturation was achieved by the 10<sup>th</sup> interview as no new symptoms were mentioned, providing confidence that all relevant symptoms and impacts were identified. Gaps were identified in currently available PROs, particularly for muscle pain, aches and fatigue.

**Discussion:** Results from this qualitative study are being used to draft a novel PRO with a focus on muscle-related symptoms. The PRO will then be evaluated with cognitive debriefing interviews in FAOD patients for content, clarity and relevance before it is finalized for use in upcoming clinical studies.