LIVED EXPERIENCE: INSIGHTS INTO LONG CHAIN FATTY ACID OXIDATION DISORDERS

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Introduction:

• Long-Chain Fatty Acid Oxidation Disorders (LC-FAODs) are a class of six rare inherited metabolic disorders: Carnitine palmitoyltransferase I (CPTI) deficiency, Carnitine palmitoyltransferase II (CPTII) deficiency, Carnitine-acylcarnitine translocase (CACT) deficiency, Long-chain 3-hydroxy-acyl-CoA dehydrogenase (LCHAD) deficiency, Trifunctional protein (TFP) deficiency, and Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency.
• Together, they affect roughly 1 in 50,000 births (Lindner et al., 2010). However, some are significantly more prevalent than others with VLCAD being the most common affecting roughly 1:85,000 births and CPT I, CPT II, and CACT being the least common affecting approximately 1:750,000-2,000,000 births (Merrit et al., 2020).
• These disorders impair the body's ability to metabolise long-chain fatty acids for energy, leading to acute energy crises (Vockley 2020).
• Severity, age of onset, and symptoms vary between disorders, but muscle weakness and rhabdomyolysis (muscle wastage) are common (Vockley 2020).
• Treatment has typically focused on a fat-restricted diet and medium-chain fatty acid supplements as alternative energy sources. A new treatment option, Triheptanoin (C7), is currently being investigated and has become available to people with LC-FAOD in the UK through the Early Access to Medicines Scheme.

Aim: To gain insight into the lived experience of people with Long Chain Fatty Acid Oxidation Disorders

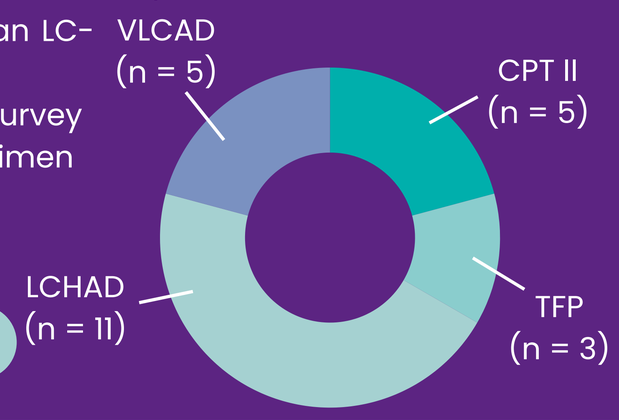
Methods:

• A digital community-facing survey was conducted by Metabolic Support UK, which ran between 10/01/2025 and 12/05/2025.
• Questions were designed based on the NICE patient organisation submission template and in-house LC-FAOD knowledge.
• The survey was disseminated via social media, newsletters and targeted outreach.
• Data were analysed using a mixed methods approach using both descriptive statistics and thematic content analysis.

Family with a child with a long chain fatty acid oxidation disorder

Results:

• 24 responses were received and included people living with an LC-FAOD (n = 6), parents/carers of someone living with an LC-FAOD (n = 16) and bereaved parents/carers (n = 2).
• 4 out of the 6 types of LC-FAOD are represented in these data, people living with CPT I and CACT are not represented in this survey
• 11 respondents had had prior experience with triheptanoin, and 9 respondents listed it as part of their current medication regimen
• Respondents were from Australia, Austria, Brazil, Canada, Croatia, Hong Kong, Malta, the UK and the USA
• The average patient age was 17 years (range: 0–59).



The current day-day burden of living with an LC-FAOD is high

The medication and food regimen burden is high

On average people with an LC-FAOD take **2.7 medications or dietary supplements** (range 1 to 6) in addition to monitoring their diet.

The most common medications people are currently taking are **MCT oil (50%), Carnitine Supplements (40%), Triheptanoin (40%) and Vitamin Supplements (36%)**

"I think it's very difficult to follow a diet, because life goes by very fast, the day flies by, I don't always have time to eat 5-6 meals a day and pay attention to nutritional values." – Person living with CPT II

The hospital visit burden is high

On average, people with an LC-FAOD had **12 planned hospital visits per year** (range 0-45); where they see on average **7 specialists** (range 3-16) and the average yearly **cost to attend appointments is £502** (range £20-£1484)

Many respondents say that **hospitalisations used to be common** but have decreased in the last few years.

"He has not been admitted for 5 years [since beginning to take triheptanoin]" – Person caring for someone living with LCHAD

The burden of providing care from home is high

33% of respondents had to make **home adaptations** to allow for care at home (N=18)

22% of respondents had to have **specialist equipment installed** to allow for care at home (N=18)

80% of respondents had **additional out-of-pocket expenses** to allow for care at home (N=20)

"The special diet is expensive. Also have had to purchase some equipment (travel bottles; dosing syringes) for the triheptanoin" - Person caring for someone living with CPTII

Living with an LC-FAOD significantly affects quality of life

85% of respondents stated that the LC-FAOD's symptoms impacted their life significantly (N=21)

'There are days when I am very tired and I have to take it easy because I am on the verge of rhabdomyolysis, my mental health is very unstable, I have days when I am strong and some days it is too much for me, too much for myself, too much for my family, etc.' -Person living with CPT II

Average **EQ Index Score 0.48**
Range -0.484 to 1, Median: 0.725

Many respondents felt that they or the person they cared for experienced some **some level of pain and could not undertake usual activities for their age group**

Triheptanoin (C7) as a novel treatment is generally well received

50% of survey respondents had **experience using triheptanoin** (N=22)

40% who had experience using triheptanoin said there were **no disadvantages** (N=10), with the only common **disadvantage** described to triheptanoin was causing **upset stomachs (36%)**

Common **advantages** to taking triheptanoin included **more energy (63%), better heart condition (36%),** and **less rhabdomyolysis (36%)**

"Triheptanoin (C7) is great, just need to make sure to take it regularly. Little to no side effects"– Parent of a child with LCHAD

Conclusions:

• This survey captures perspectives from a wide range of global respondents living with or caring for someone with an LC-FAOD. It covers the majority of the different LC-FAODs, offering valuable insights into life with this class of rare genetic conditions.
• The findings highlight the significant burden, from multiple angles, of living with or caring for someone with an LC-FAOD, which significantly impacts quality of life.
• The data also shows that triheptanoin is well received with a minimal side effects and has meaningful benefits to the LC-FAOD community

Acknowledgments:

Metabolic Support UK would like to thank the members of the LC-FAOD community for their participation in the survey

References:

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