

## Reference List

Baker JJ, Burton BK. Diagnosis and clinical management of long-chain fatty-acid oxidation disorders: a review. *touchREV Endocrinol*. 2021;17(2):108-111. doi:10.17925/EE.2021.17.2.108

Brunengraber H, Roe CR. Anaplerotic molecules: current and future. *J Inherit Metab Dis*. 2006;29(2-3):327-331. doi:10.1007/s10545-006-0320-1

DeLany JP, Horgan A, Gregor A, Vockley J, Harding CO, Gillingham MB. Resting and total energy expenditure of patients with long-chain fatty acid oxidation disorders (LC-FAODs). *Mol Genet Metab*. 2023;138(3):107519. doi:10.1016/j.ymgme.2023.107519

Dojolvi (triheptanoin). Prescribing information. Ultragenyx Pharmaceutical Inc.; October 2023. Accessed November 8, 2024.

[https://www.accessdata.fda.gov/drugsatfda\\_docs/label/2020/213687s000lbl.pdf](https://www.accessdata.fda.gov/drugsatfda_docs/label/2020/213687s000lbl.pdf)

El-Gharbawy A, Vockley J. Inborn errors of metabolism with myopathy: defects of fatty acid oxidation and the carnitine shuttle system. *Pediatr Clin North Am*. 2018;65(2):317-335. doi:10.1016/j.pcl.2017.11.006

Gillingham MB, Elizondo G, Behrend A, et al. Higher dietary protein intake preserves lean body mass, lowers liver lipid deposition, and maintains metabolic control in participants with long-chain fatty acid oxidation disorders. *J Inherit Metab Dis*. 2019;42(5):857-869. doi:10.1002/jimd.12155

Gillingham MB, Heitner SB, Martin J, et al. Triheptanoin versus trioctanoin for long-chain fatty acid oxidation disorders: a double blinded, randomized controlled trial. *J Inherit Metab Dis*. 2017;40(6):831-843. doi:10.1007/s10545-017-0085-8

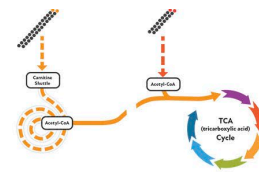
Houten SM, Wanders RJ. A general introduction to the biochemistry of mitochondrial fatty acid  $\beta$ -oxidation. *J Inherit Metab Dis*. 2010;33(5):469-477. doi:10.1007/s10545-010-9061-2

Janeiro P, Jotta R, Ramos R, et al. Follow-up of fatty acid  $\beta$ -oxidation disorders in expanded newborn screening era. *Eur J Pediatr*. 2019;178(3):387-394. doi:10.1007/s00431-018-03315-2

Karall D, Brunner-Krainz M, Kogelnig K, et al. Clinical outcome, biochemical and therapeutic follow-up in 14 Austrian patients with Long-Chain 3-Hydroxy Acyl CoA Dehydrogenase Deficiency (LCHADD). *Orphanet J Rare Dis*. 2015;10:21. doi:10.1186/s13023-015-0236-7

## EXPERT PERSPECTIVES:

# BRIDGING GAPS, OPTIMIZING TREATMENT IN ADULTS WITH LC-FAODS



Knottnerus SJG, Bleeker JC, Wüst RCI, et al. Disorders of mitochondrial long-chain fatty acid oxidation and the carnitine shuttle. *Rev Endocr Metab Disord*. 2018;19(1):93-106. doi:10.1007/s11154-018-9448-1

Longo N, Amat di San Filippo C, Pasquali M. Disorders of carnitine transport and the carnitine cycle. *Am J Med Genet C Semin Med Genet*. 2006;142C(2):77-85. doi:10.1002/ajmg.c.30087

Marsden D, Bedrosian CL, Vockley J. Impact of newborn screening on the reported incidence and clinical outcomes associated with medium- and long-chain fatty acid oxidation disorders. *Genet Med*. 2021;23(5):816-829. doi:10.1038/s41436-020-01070-0

Merritt JL 2nd, MacLeod E, Jurecka A, Hainline B. Clinical manifestations and management of fatty acid oxidation disorders. *Rev Endocr Metab Disord*. 2020;21(4):479-493. doi:10.1007/s11154-020-09568-3

Owen OE, Kalhan SC, Hanson RW. The key role of anaplerosis and cataplerosis for citric acid cycle function. *J Biol Chem*. 2002;277(34):30409-30412. doi:10.1074/jbc.R200006200

Peña-Quintana L, Correcher-Medina P. Nutritional management of patients with fatty acid oxidation disorders. *Nutrients*. 2024;16(16):2707. doi:10.3390/nu16162707

Roe CR, Brunengraber H. Anaplerotic treatment of long-chain fat oxidation disorders with triheptanoin: Review of 15 years experience. *Mol Genet Metab*. 2015;116(4):260-268. doi:10.1016/j.ymgme.2015.10.005

Rohr F. Nutrition management of fatty acid oxidation disorders. In: Bernstein LE, Rohr F, van Calcar SC, eds. *Nutrition Management of Inherited Metabolic Diseases*. 3rd ed. Cham, Switzerland: Springer; 2022:417-432. doi:10.1007/978-3-030-94510-7\_23

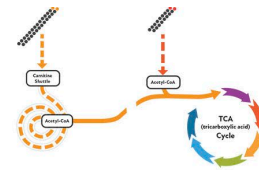
Saudubray JM, Martin D, de Lonlay P, et al. Recognition and management of fatty acid oxidation defects: a series of 107 patients. *J Inherit Metab Dis*. 1999;22(4):488-502. doi:10.1023/a:1005556207210

Shekhawat PS, Matern D, Strauss AW. Fetal fatty acid oxidation disorders, their effect on maternal health and neonatal outcome: impact of expanded newborn screening on their diagnosis and management. *Pediatr Res*. 2005;57(5 Pt 2):78R-86R. doi:10.1203/01.PDR.0000159631.63843.3E

Spiekerkoetter U. Mitochondrial fatty acid oxidation disorders: clinical presentation of long-chain fatty acid oxidation defects before and after newborn screening. *J Inherit Metab Dis*. 2010;33(5):527-532. doi:10.1007/s10545-010-9090-x

## EXPERT PERSPECTIVES:

# BRIDGING GAPS, OPTIMIZING TREATMENT IN ADULTS WITH LC-FAODS



Tamas C, Tamas F, Kovacs A, Cehan A, Balasa A. Metabolic contrasts: fatty acid oxidation and ketone bodies in healthy brains vs. glioblastoma multiforme. *Int J Mol Sci.* 2024;25(10):5482. doi:10.3390/ijms25105482

Vockley J, Burton BK, Berry G, et al. Triheptanoin for the treatment of long-chain fatty acid oxidation disorders: Final results of an open-label, long-term extension study. *J Inherit Metab Dis.* 2023;46(5):943-955. doi:10.1002/jimd.12640

Vockley J, Burton B, Berry G, et al. Effects of triheptanoin (UX007) in patients with long-chain fatty acid oxidation disorders: Results from an open-label, long-term extension study. *J Inherit Metab Dis.* 2021;44(1):253-263. doi:10.1002/jimd.12313

Vockley J, Longo N, Madden M, et al. Dietary management and major clinical events in patients with long-chain fatty acid oxidation disorders enrolled in a phase 2 triheptanoin study. *Clin Nutr ESPEN.* 2021;41:293-298. doi:10.1016/j.clnesp.2020.11.018

Vockley J. Long-chain fatty acid oxidation disorders and current management strategies. *Am J Manag Care.* 2020;26(7 Suppl):S147-S154. doi:10.37765/ajmc.2020.88480

Vockley J, Burton B, Berry GT, et al. UX007 for the treatment of long chain-fatty acid oxidation disorders: safety and efficacy in children and adults following 24weeks of treatment. *Mol Genet Metab.* 2017;120(4):370-377. doi:10.1016/j.ymgme.2017.02.005

Vockley J, Longo N, Andresen BS, Bennett MJ. Mitochondrial fatty acid oxidation defects. In: Sarafoglou K, Hoffmann GF, Roth KS. eds. *Pediatric Endocrinology and Inborn Errors of Metabolism, 2e.* McGraw-Hill Education; 2017. Accessed November 10, 2024. <https://accesspediatrics.mhmedical.com/content.aspx?bookid=2042&sectionid=154109616>

Vockley J, Marsden D, McCracken E, et al. Long-term major clinical outcomes in patients with long chain fatty acid oxidation disorders before and after transition to triheptanoin treatment-- A retrospective chart review [published correction appears in Mol Genet Metab. 2015 Nov;116(3):221]. *Mol Genet Metab.* 2015;116(1-2):53-60. doi:10.1016/j.ymgme.2015.06.006

Wajner M, Amaral AU. Mitochondrial dysfunction in fatty acid oxidation disorders: insights from human and animal studies. *Biosci Rep.* 2015;36(1):e00281. doi:10.1042/BSR20150240

Wanders RJA, Ruiten JPN, IJlst L, Waterham HR, Houten SM. The enzymology of mitochondrial fatty acid beta-oxidation and its application to follow-up analysis of positive neonatal screening results. *J Inherit Metab Dis.* 2010;33(5):479-494. doi:10.1007/s10545-010-9104-8

Wilcken B. Fatty acid oxidation disorders: outcome and long-term prognosis. *J Inherit Metab Dis.* 2010;33(5):501-506. doi:10.1007/s10545-009-9001-1