



Understanding LC-FAOD

Information for individuals—and their caregivers,
families, and loved ones—living with long-chain
fatty acid oxidation disorders (LC-FAOD)



What causes long-chain fatty acid oxidation disorders (LC-FAOD)?

METABOLISM IS ESSENTIAL TO LIFE

- Metabolism is how the body breaks down and converts food (carbohydrates, fats, and proteins) into energy
- Metabolism relies on enzymes to drive chemical reactions in your body

SPECIFIC ENZYMES HELP BREAK DOWN FAT AND CONVERT IT INTO ENERGY

FATS ARE ESSENTIAL TO ENERGY

Most dietary fats begin as long-chain fatty acids (LCFAs), which are broken down and used by the body as an important source of energy during times of:



Fasting (including sleep)



Exercise



Illness



Stress



UNAFFECTED METABOLISM IS DISRUPTED IN LC-FAOD

LC-FAOD are a group of rare, genetic metabolic disorders that prevent the body from breaking down LCFAs into energy during metabolism.

LC-FAOD are caused by a lack of, or defect in, one of the enzymes needed to break down fats. The following are specific types of enzyme deficiencies associated with LC-FAOD.

CPT I

CACT

CPT II

VLCAD

TFP

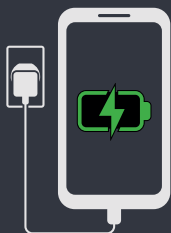
LCHAD

How does LC-FAOD impact the body?

IN UNAFFECTED METABOLISM

the body has access to 2 sources of energy, similar to a smartphone

IMMEDIATE ENERGY (EATING)



Immediate energy functions like electricity from a wall outlet that:

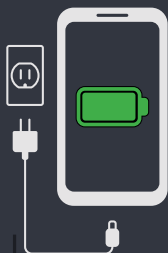
- Directly powers a smartphone
- Charges its battery



Eating proteins, carbohydrates, and fats:

- Provides a direct source of immediate energy
- “Charges” the body’s battery by converting these nutrients, including LCFAs, into reserve energy

RESERVE ENERGY (FASTING)



Reserve energy functions like an internal battery when the separate direct power source is no longer available



Reserve energy helps power the body during periods of fasting (including sleep) or physical stress (illness or exercise)

In unaffected metabolism, the body can exert energy and/or fast for a prolonged period by drawing on reserve energy to keep the body powered.

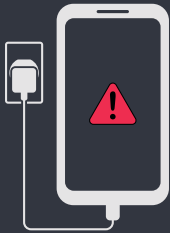
Watch how LC-FAOD impacts the body



IN LC-FAOD

the body has a faulty battery because it cannot rely on LCFAs to help it stay “charged”

IMMEDIATE ENERGY (EATING)



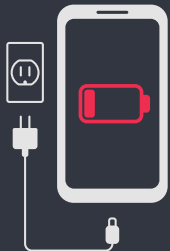
A faulty smartphone battery drains faster, requiring you to plug into the wall outlet more frequently



Eating proteins, carbohydrates, and fats:

- Provides a direct source of immediate energy, but not being able to use LCFAs leads to lower energy levels
- Can “charge” the body’s battery (reserve energy), but the body can’t use the stored LCFAs for energy

RESERVE ENERGY (FASTING)



Without a fully charged battery, a smartphone will begin powering down faster than normal when it’s unplugged from a direct power source



Since the body’s battery (reserve energy) can’t use LCFAs to stay fully charged, the body may not have enough energy during prolonged periods of fasting or physical stress

In LC-FAOD, when the body can’t supply enough energy to meet its needs, people may develop chronic symptoms or experience acute episodes.

Recognizing the signs and symptoms of LC-FAOD

Symptoms can vary between different types of LC-FAOD and can impact different body organs. No matter what enzyme deficiency you're living with, it's important to understand how to:



Recognize these signs and symptoms



Develop a plan for emergency situations with your LC-FAOD healthcare team

CHRONIC SYMPTOMS (PERSISTENT OR RECURRING)

Fatigue, muscle pain, muscle cramps, muscle weakness, and foggy thinking can be brought on or made worse by fasting, illness, sustained exercise, and physiologic stress, with chronic lack of energy potentially leading to:

- Decreased muscle tone and weakness (hypotonia)
- Damage to the retina of the eyes (retinopathy)*
- Damage to small nerves in the legs and feet (peripheral neuropathy)*

*Applies to certain LC-FAOD types such as LCHAD and TFP. Consult your healthcare team for more information.

ACUTE EPISODES (IMMEDIATE AND SEVERE)

These episodes can be triggered by illness or fasting, but can also occur spontaneously and unpredictably. Individuals with LC-FAOD may experience acute metabolic crises that include these serious conditions:

- Heart muscle damage (cardiomyopathy)[†]
- Muscle breakdown (rhabdomyolysis)
- Low glucose, also called low blood sugar (hypoglycemia)

[†]May have a chronic impact.

THE IMPACT OF LC-FAOD

Acute episodes can sometimes lead to hospitalization, emergency room visits, and emergency interventions, with the potential for sudden death.

More research is needed on the life expectancy and long-term impact of LC-FAOD.

Living with LC-FAOD

Living with LC-FAOD comes with unique challenges. By working with your healthcare team, you can learn ways to optimally manage your condition, including:

DIETARY

- Maintaining a mealtime routine (avoiding fasting)
- Managing diet (eating certain foods and avoiding, or limiting, others)

LIFESTYLE

- Learning how to modify physical activities or make adjustments in certain settings or environments
- Knowing how long to walk or play
- Recognizing symptoms and early signs of fatigue
- Understanding your body's early warning signs and what you need to do to stay safe

LC-FAOD IS RARE, BUT YOU ARE NOT ALONE

Talk to your LC-FAOD healthcare team about any signs or symptoms you may be experiencing or have questions about.

You can find a discussion guide with helpful tips on working with your healthcare team, download other patient and caregiver resources, and gain support from others at

FAODinFocus.ca