

# Living Your Best Life

Learn what to expect with long-chain fatty acid oxidation disorders (LC-FAOD) during **young adulthood and beyond**



Learn from others' experiences to help you on your journey as you become more and more independent in managing your LC-FAOD.



Ultragenyx has had the privilege of meeting with people and families living with LC-FAOD and gathering a collection of insights and tips directly from the community.

### **In these booklets, we aimed to:**

- Capture the collective voices and experiences of the LC-FAOD community
- Provide key information to help you feel more prepared and supported

# Your future awaits

Turning 18 is a big deal—a milestone into adulthood! At this age, you become a legal adult in many countries. You may now be able to vote, get married, buy a car, and—importantly—make your own medical decisions.



## YOU ARE IN CHARGE OF YOUR OWN LIFE NOW

This time marks a shift in taking responsibility for your health from your parents to you. You will learn how to rely more on yourself, take charge of your own LC-FAOD needs and do what is best for you. It might not happen overnight, but with time, you will understand what it means to be independent and make important decisions with the support of your healthcare team.

**You will also learn how to balance managing your condition while not letting it define you.**

As adults living with LC-FAOD, we know this can seem daunting at first. But know you will get through these changes and learn so much about yourself and what you are capable of.



## SHARING YOUR STORY

As you get older, you may find yourself interested in sharing your story, speaking up about life with LC-FAOD, or helping others navigate life with LC-FAOD. Telling your story is one of the most powerful ways to advocate for your needs and make an emotional connection with others who are unfamiliar with the disorder.



Share your story! See **Crafting Your Story** and **Share Your Rare Journey** included in this toolkit.



**This booklet will guide you along as you navigate through early adulthood.**

The goal is to **help prepare you for important issues that may arise, empower you to take action, and encourage you to work closely with your care team to make the best decisions** for yourself.

Remember, this is an exciting time filled with lots of new experiences, new relationships, and new freedoms.

Living with LC-FAOD has its challenges, but we can tell you from our own experiences that **your future looks bright** as long as you take the necessary steps to manage your health.



### First-hand tip

Managing your LC-FAOD can be challenging but it is important to find a way to stay motivated and work on your care goals to maintain the best health possible.

Here are some pointers to staying on track:

- **Build a support network** to help you through challenging times and offer encouragement
- **Keep a healthy perspective about your diet** and focus on how it is integral to your health
- **Find ways to make managing LC-FAOD work for you and your lifestyle** so you can engage in activities that you enjoy

# Taking charge of your health and care



## A PERSONAL EXPERIENCE

*I recall the day I realized I needed to step up my involvement in making my own doctor appointments and addressing my health needs.*

I was about to graduate high school and planning on moving out. My friend was over at my house and we overheard my mother calling to make an appointment with my metabolic team. My friend turned to me and asked me “How does your mom know your schedule? Shouldn’t she check with you to make sure you are available on that day?” It struck me—this was my life and I needed to take control of it. I wasn’t a child anymore and I was capable of handling many things on my own. I just needed to start.



See **Communicating with Healthcare Providers and Medical History** in this toolkit for help preparing for your doctor visit.



## ADVICE FROM YOUR PEERS

*Don’t take on all the responsibility for your health needs at once.*

That will likely lead you to feel overwhelmed by the many aspects that need attention. Instead, take them on gradually, one area at a time. Once you are confident with one area, you can add more. Here are some things you can start with:

- Make your own appointments
- Refill your own medications/supplements
- Follow up on your blood work
- If your clinic has a patient portal, gain access so you can view your laboratory results and communicate directly with your healthcare providers

As you know, LC-FAOD is a lifelong journey and managing it will require ongoing medical attention. It’s never too early or late to take more responsibility for your health.

It’s important that you work with your care team to know everything that you need to know about your specific situation. Here are important things to know about this chronic condition over the coming years:

- **Symptoms can be brought on or made worse** by fasting, illness, sustained exercise, and physiologic stress
- **The organs that are primarily affected** are the heart, skeletal muscle, and liver, which means LC-FAOD can cause a breakdown of muscle tissue (rhabdomyolysis), disease of the heart muscle (cardiomyopathy), and low blood sugar/low blood ketones (hypoketotic hypoglycemia)
- **Chronic symptoms** may lead to decreased muscle tone and weakness (hypotonia), damage to the retina of the eyes (retinopathy),\* damage to the nerves outside the brain and spinal cord (peripheral neuropathy)\*
- LC-FAOD impacts nearly all parts of life, including your **physical, mental, emotional and social well-being**. Some days may feel harder than others, and that’s OK. It’s really important to honor where your body is one day at a time

**If you are feeling anxious, fearful, or depressed about your LC-FAOD, the continual avoidance of potential triggers, or the long-term impact, reach out to your care team to let them know.** There may be things they can do to help. Just as your physical health is important, so is your emotional well-being.



### First-hand tip

Over time, it may be difficult to stay motivated to manage your health and go for follow-up visits when life gets busy or when you’re enjoying new experiences, but it’s so important that you do.

**Consider including a trusted friend or relative in your health plan so they can encourage you when things get hard.**

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

# Considerations for college



## A PERSONAL EXPERIENCE

*I was thrilled when I got accepted to my first-choice college in another state. I chose this college because while it was far from my home, it was still driving distance so I could travel back and forth easily if needed.*

Then it hit me—I'd still be away from my family and have to take care of my own meals, doctor appointments, and other medical needs. While the thought overwhelmed me initially, I then realized that my parents had been preparing me for this moment for years. With my friends and family to support me—even from a distance—I knew I could do it.



## ADVICE FROM YOUR PEERS

*Choosing to go away to college is a big commitment, and it may not be the right choice for everyone.*

For those who are interested and able to pursue a college away from home, this may be the first time you live apart from your family for long stretches. On the one hand, the independence and all that comes with it can be thrilling. On the other hand, it can be overwhelming: figuring out how to juggle schoolwork, meet new friends and manage doctors' appointments, your diet, and your other health needs—in a new environment no less!

**It might take some time to find a healthy balance for all these areas, but don't give up—it's attainable.** Be sure to speak to people in your support network. You can also reach out for guidance on campus; many colleges have advisors who can assist with helping navigate this new world. You are never alone, but you will need to seek out support when you need it.

If you do decide that attending a college away from home is right for you, here are some suggestions to help with a safe and smooth transition:



**Identify the nearest hospital for emergencies**



**Have your clinic contact information** available to you at all times. Many clinics now offer telemedicine visits allowing you to stay in touch with your team



**Notify** your pharmacy that your address has changed



**Set reminders** for important tasks, such as making doctor appointments and ensuring that your disease management and treatment plan remains up to date



**Discuss** with your care team the effects of drinking alcohol. Excessive drinking should be avoided, as this adds metabolic stress and could make people with LC-FAOD extremely ill



**Tell your roommates and close friends about your condition.** They can be your new support system, help you identify symptoms, and get you medical attention if needed



If you play a sport, **talk to your coach and teammates** so they are aware of your medical needs (eg, breaks, hydration, etc)



See the **next page** for important considerations when choosing a college

If you decide to commute or to live at home, you can still follow many of these suggestions to help you make the transition to independently caring for your own health.

# Questions for college

As you prepare for your college years, ask yourself the following questions to make sure you are set up for a successful college experience. Use the lines below each section to jot down your own thoughts and discuss with your healthcare team.



## Lifestyle/dorm living

- What foods can I eat from the dining halls?
- What accommodations will the school dining hall or dormitory be willing to make?
- Will they have nutrition information available to me?
- Will they allow me to prepare my formula/medical food?
- Will I have a fridge in my dorm room?
- Do I have access to food/beverages, if needed, at all times?
- Can I prepare my formula/medical food or other supplements in my dorm?

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## Friends and social circle

- When and how should I share information about LC-FAOD and the impact it has on my life with new friends and classmates?

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## Career planning/hobbies

- How and to what extent (if any) will LC-FAOD play a role in my career choice and hobbies?
- What activities can I participate in that would be safe for me?
- If I decide to play a sport or choose an activity that requires extra energy, what accommodations do I need to make?

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## Traveling

- What items do I need to carry on a plane or other long-distance travel?
- Do I need to get a letter of medical necessity from my doctor for traveling with medication?
- Do I need a refrigerator (or anything else) in my hotel room when traveling?

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# Entering the workforce



## A PERSONAL EXPERIENCE

*As I was preparing to look for jobs, I began to think about the interviewing process for full-time employment.*

I had many questions—what should I share about my LC-FAOD with prospective employers? What should I not share? Is it appropriate to discuss in an interview, and if so, when and how? It took a few interviews, and with some trial and error, I eventually figured out what I felt comfortable sharing and what was critical for a future employer to know. You will figure it out too!



## ADVICE FROM YOUR PEERS

*It's your choice what information about LC-FAOD you want to share or don't want to share.*

You are not legally obligated to disclose any information about your condition to your employer. Some people might not be comfortable sharing any information because they are very private by nature, they worry that it could jeopardize a job opportunity, or it feels like they are constantly repeating themselves; others might feel that transparency may help their employer better able to make accommodations, if needed.

You will also need to prepare yourself for balancing full-time work with managing your health. You might want to consider:



**How you will take time off for appointments.** Can you use sick days for appointments? Can you work from home or make up the hours you missed?



**Traveling for work.** How long will you be away and what, if any, are the implications for your health plan? Do you need to make special arrangements for the plane or hotel?



**Dealing with sick leave.** How many sick days are you allowed? Can you tap into vacation days if need be?



**Short-term/long-term disability.** What are your company policies regarding disability leave?

NOTE: The Family Medical Leave Act (FMLA) is a federal law which allows an employee to take up to 12 *unpaid* weeks off per year for specified medical reasons. (Applies to the United States only.)



### First-hand tip

As someone living with a chronic and potentially disabling condition, it's important to know that there are laws in place to protect you and guarantee certain rights in the workplace.

This can include your employer making reasonable accommodations for you to do your job. Familiarizing yourself with your rights as you navigate your career can help you succeed. **You can learn about The Americans with Disabilities Act by visiting ADA.gov.** (NOTE: this applies to people living in the United States only.)



# Navigating the world of health insurance



## A PERSONAL EXPERIENCE

*I never realized how much there was to know about health insurance before I started looking into my options.*

For me, words like *deductibles*, *copays*, *premiums*, and *co-insurance* were so unfamiliar, it felt like I was learning a new language. I asked a lot of questions and wasn't satisfied until I really understood the answers. It helps to have all the facts first so you can make a decision about what will work best for you.



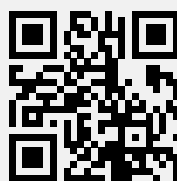
## ADVICE FROM YOUR PEERS

*Do your research! It's important to understand the options that you have and what will work best for your needs.*

Plans and coverage can vary significantly depending on the employer and the plan you choose, so it is important to check on the specific details of the plan before signing up. There's a lot of information to take in: write it down and create a pro/con list for each option that is available to you.

It's also important to know that when you turn 26, you may no longer be eligible for insurance coverage under your parent's health plan. Be sure to plan ahead to avoid a lapse in coverage.

Your care team can support you through this process. Reach out to them with your questions. **You can also learn more about health insurance at [ultrarareadvocacy.com](https://ultrarareadvocacy.com).**



Definitions for common health insurance terms that you will need to understand:

**Deductible**—The amount you pay annually for health services before your insurance company pays its share. (For example, if you have a deductible of \$1,000, your insurance plan might not start covering its share of your bills until you've paid \$1,000 for healthcare in a given year.)

**Copay (or copayment)**—The amount you pay each time you visit your healthcare provider. Copays may also apply to prescriptions, blood draws, or other medical procedures.

**Premium**—The amount you pay to the health insurance company each month to maintain your coverage.

**Co-insurance**—The percentage of covered services that you're required to pay after your deductible is met. (For example, your health insurance may cover 70% of the charges for a covered hospitalization, leaving you responsible for 30%.)

**Health Maintenance Organization (HMO)**—An insurance plan in which coverage restricts patients to a particular group of physicians (called a network). It generally won't cover out-of-network care except in an emergency.

**Preferred Provider Organization (PPO)**—An insurance plan that allows patients to choose any physician they wish, either inside or outside of their network. You pay less if you use providers that belong to the plan's network. You can use doctors, hospitals, and providers outside of the network for an additional cost.



### First-hand tip

If your health insurance refuses to pay a claim for a necessary treatment or resource for LC-FAOD, do not take "no" for an answer!

You have the right to appeal the decision. **Ask for the exact reason for the denial, collect all documentation that explains the need for treatment, and consult your specific policy for the proper appeals process.** Often, care providers are able to assist with the process, so reach out to your team for help.

# Family planning



## A PERSONAL EXPERIENCE

*My partner and I were getting serious in our relationship, and we started thinking about our future together, including raising a family one day.*

In all the years of care for my LC-FAOD, I was so focused on me; I never put much thought into how it might affect my decision to have a family one day. We decided to seek answers, and that's when we started talking to a genetic counselor at the metabolic clinic. It was so helpful to speak to someone who really understood how LC-FAOD can be passed down and could help us explore our options.



## ADVICE FROM YOUR PEERS

*As you begin to think about starting a family, it's important to understand the chances of having a child with LC-FAOD.*

As someone diagnosed with LC-FAOD, your children would be at increased risk if your partner has, or is a carrier of the same LC-FAOD condition.

It may help to start by learning how you inherited this metabolic disorder and the way genes are passed down.

It may seem scary or overwhelming to think about passing along the disease, but know that your care team and family are there to support you. Keep on reading to learn more about how you can prepare for this stage of your life.



Below are important details to know about genetic testing and the risks of your future children also having LC-FAOD:



Your partner can get **genetic testing** to find out if they are a carrier of the same type of LC-FAOD. Screening for LC-FAOD can help empower you to make informed health decisions for your future. Genetic testing may also:

- Increase your understanding of the science of the disorder
- Lead to management options that you may have not known were available
- Allow families or individuals with the same genetic condition to connect



After your partner receives their genetic test results, you may find it helpful and supportive to meet with a **genetic counselor**. Together, they can help you understand different scenarios such as:

- What are the outcomes if my partner is also affected by LC-FAOD?
- What are the outcomes if my partner is a carrier for LC-FAOD?
- What are the outcomes if my partner is unaffected and is not a carrier of LC-FAOD?
- What genetic testing options are available before, during or following pregnancy to see if the child is affected?



### First-hand tip for females

#### Planning for pregnancy

LC-FAOD may affect the management of your pregnancy. Contact your metabolic team if you are thinking about becoming pregnant. They will work together with your obstetrician to develop a plan and provide guidance on your health before and during pregnancy, and after delivery. There may also be special considerations for testing and management of your newborn baby.



# Goals to consider

## as you transition to adulthood

You want to make sure that you are set up for success as an adult living with LC-FAOD. Here are some suggestions to ensure that you are properly prepared for and ready for this next stage in your life.

- I know what foods are healthy for me*
- I plan my meals independently*
- I keep track of my nutrition independently*
- I keep track of my medications and supplements independently*
- I prepare and administer my own medications and supplements*
- I know how to order/refill my medications and supplements*
- I schedule and attend medical appointments with my LC-FAOD team regularly*
- I understand how to manage and treat my condition based on my care team's medical guidance*
- I can freely discuss with my care team any symptoms or challenges I may be still experiencing*
- I manage my insurance independently*
- I ask questions to my care team and get the answers I need to help me stay healthy*
- I am aware of the importance of managing LC-FAOD*
- I understand that LC-FAOD can impact my health both today/immediately and over the long-term*
- I know to be prepared for an unexpected crisis at an inconvenient time*
- I can recognize the signs of a crisis and make it a priority to address*
- I feel comfortable seeking advice and support from my care team*
- I have an emergency protocol letter; it identifies my disorder, how I must be treated and what agents must be avoided*
- I know where to find details about my health insurance plan*
- I am aware of the disease state, management, and financial resources available to me*