The Long-Chain Fatty Acid Oxidation Disorders (LC-FAOD) Resource Toolkit is a set of materials developed by Patient Advocacy at Ultragenyx Pharmaceutical for people and families living with LC-FAOD. Ultragenyx incorporated feedback from the LC-FAOD community and healthcare providers into the content and design of the Toolkit to help ensure the materials meet the needs of the LC-FAOD community. It consists of the below documents:

1. LC-FAOD Welcome Letter
2. LC-FAOD Building Your Support Network
3. LC-FAOD Communicating with Healthcare Providers
4. LC-FAOD Medical History Template
5. LC-FAOD Living Well for Caregivers
6. LC-FAOD Share Your Rare Journey
7. LC-FAOD Craft Your Story

This information is currently being shared with patients, families, patient advocacy groups and healthcare providers via print and digital channels. We welcome you to share this information with your patients. Resources 2-7 listed above can also be found at https://www.faodinfocus.com/resources/.
This toolkit is designed for people affected by rare conditions called long-chain fatty acid oxidation disorders (LC-FAOD, also called FAOD). The six resources included will support your journey whether you are living with or caring for someone with LC-FAOD. It includes information on how to:

- **Build Your Support Network.** Learn about organizations that can help you.
- **Communicate with Healthcare Providers.** Make the most of your appointments.
- **Prepare an LC-FAOD Medical History.** Gather your important information.
- **Live Well While Caring for Someone with LC-FAOD.** Prepare others to help and get the respite you need.
- **Share Your Rare Journey and Craft Your Story.** Learn to educate and advocate.

### ABOUT LC-FAOD

LC-FAOD are a group of rare, autosomal recessive genetic conditions. This means that the disorder is inherited when each parent is a carrier. In people with LC-FAOD, an enzyme that transports or breaks down dietary long-chain fat is either very low or does not work properly. The signs and symptoms seen in people with LC-FAOD occur because they are unable to properly break down long-chain fatty acids for energy. Therefore, they must find alternative energy sources.

### TYPES OF LC-FAOD

There are different types of LC-FAOD. Each type results from a different gene mutation.

Fats in our diet are an important source of energy production during times of metabolic stress or prolonged fasting. Long-chain fatty acids need to be transported into the mitochondrion (energy house of the cell) by three different enzymes (CPT I, CACT, CPT II), and then once in the mitochondrion, the fatty acids undergo further metabolism for energy (VLCAD, TFP, LCHAD).

#### CPT I (Carnitine Palmitoyltransferase I) Deficiency

A mutation in the CPTIA gene causes the CPT I enzyme to not function properly, which results in CPT I deficiency. This means that long-chain fatty acids cannot begin the first step in the carnitine shuttle to bring long-chain fatty acids into the mitochondrion.²

#### CACT (Carnitine-Acylcarnitine Translocase) Deficiency

A mutation in the SLC25A20 gene causes CACT deficiency. This means that the middle step of the carnitine shuttle, transporting the long-chain fatty acid into the mitochondrion, cannot be performed properly.³

#### CPT II (Carnitine Palmitoyltransferase II) Deficiency

A mutation in the CPTII gene causes CPT II deficiency. This means that the last step in the carnitine shuttle cannot be completed. Therefore, the long-chain fat cannot enter the mitochondrion.⁴

#### VLCAD (Very Long Chain Acyl-CoA Dehydrogenase) Deficiency

A mutation in the ACADVL gene causes VLCAD deficiency. This enzyme is part of the long-chain beta oxidation spiral. Once long-chain fats enter the mitochondria after transportation via the carnitine shuttle, they are processed by the long-chain beta oxidation spiral. If the VLCAD enzyme is not functioning properly, then long-chain fatty acids are not broken down properly, resulting in lower energy and damage from incomplete processing of fatty acids.⁵⁻⁶
**TFP (Trifunctional Protein) Deficiency**

TFP deficiency occurs when a person has a mutation in both the *HADHA* gene and *HADHB* gene. TFP is a three enzyme complex and performs the last three steps in the breakdown of long-chain fatty acids. One of these enzymes is LCHAD. If the TFP enzyme complex is not working properly long-chain fats cannot be broken down for energy and unused fatty acids can build up in the body and cause issues.⁷

**LCHAD (Long-chain 3-hydroxy-acyl-CoA Dehydrogenase) Deficiency**

LCHAD deficiency occurs when a person has a mutation in the *HADHA* gene. This is part of the TFP complex, but the mutation is just in the *HADHA* gene, causing LCHAD. If there is a deficiency in this step, long-chain fatty acids are not properly broken down so they cannot be used for energy.⁷

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**REFERENCES**


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Building Your Support Network

Living with a rare condition can feel isolating. Most people you encounter are not likely to have heard of long-chain fatty acid oxidation disorders (LC-FAOD), and many won’t understand what it’s like to manage a chronic or life-threatening condition. These are some reasons why it’s very important to find and connect with organizations and people who share similar experiences and challenges, and who can offer support.

Because LC-FAOD are a type of fatty acid oxidation disorder, some people may refer to them as FAOD for short.
How to Use this Resource

You can find support in many ways, such as through patient advocacy organizations, your physician or healthcare providers, academic institutions, online or in-person support groups and social media forums, and professional organizations.

**Patient advocacy organizations** are non-profit groups dedicated to helping patients and caregivers navigate life with a condition or diagnosis. To achieve their goals, advocacy organizations work in a variety of ways, such as:

- Hosting events and networking opportunities to bring patients and families together, both in person and online.
- Serving as a source of disease and treatment information.
- Promoting disease awareness, hosting support programs or sharing resources.
- Raising money to fund research for a disease or group of related diseases.
- Advancing public policy to meet the needs of patients with rare disease and their families.
- Supporting disease research by collecting medical history and health data from patients.

Organizations may focus on some or all of these areas, or even just one single area. They can be specific to LC-FAOD or one type of LC-FAOD, serve a related rare disease with similar symptoms or management needs, or support the common needs of people living with any rare disease. **The following support resources focus not only on FAOD, but on mitochondrial disorders, metabolic diseases, other rare conditions, and topics that may impact all rare families.**

When reading or using any resource, keep in mind that medical research moves quickly and information on the internet can become outdated. Remember to check the date of publication and always bring questions and new ideas to your healthcare team.
Metabolic Disorder Organizations

INTERNATIONAL NETWORK FOR FATTY ACID OXIDATION RESEARCH AND MANAGEMENT (INFORM) FAMILIES

Primary focus: FAOD disease-specific research and education
Website: informnetwork.org/inform-families

INFORM is a global scientific organization that focuses on FAOD and related metabolic disorders. Its main goals are to provide education and encourage or support research on these disorders. INFORM also hosts a FAOD conference each year for researchers.

INFORM Families is a dedicated part of the organization that provides families with information on newborn screening, diagnosis, and treatment of FAOD. They also include patient stories, information on clinical trials, and an “Ask the Experts” discussion forum.

MITOACTION

Primary focus: Mitochondrial disease education and awareness
Website: mitoaction.org

MitoAction's mission is to improve the quality of life for children, adults, and families living with mitochondrial disease, including FAOD, through support, education, outreach, advocacy, and clinical research initiatives.

- The organization helps patients and caregivers manage their day-to-day journey with mitochondrial disease, as well as helps clinicians and researchers better understand the burdens of living with the disease.
- MitoAction offers monthly educational webinars, one-on-one support through the Mito411 support line, weekly support teleconferences, financial assistance for families in need, college scholarships for students with mitochondrial disease, opportunities to attend summer camp, a physician locator, clinical protocols to help with disease management, and a state-of-the-art mobile platform.
Umbrella Rare Disease Organizations

EVERYLIFE FOUNDATION FOR RARE DISEASES

Primary focus: Rare disease public policy and legislative advocacy  
Website: everylifefoundation.org

EveryLife Foundation for Rare Diseases provides training, education, resources, and opportunities to guide patients to be advocates for their needs with the intent to affect and change public policy. Some programs include:

- **Rare Disease Legislative Advocates (RDLA)** is a collaborative organization that provides informational meetings, legislative resources, advocacy tools, and special events that support organizations and advocates working to promote rare disease legislation. RDLA offers travel stipends for a certain number of patients to attend Rare Disease Week on Capitol Hill.

- **Rare Disease Week** on Capitol Hill brings rare disease community members from across the country together to learn about federal legislative issues, network, and share their stories with legislators.

- **Rare on the Road**, a partnership with Global Genes, builds and activates the rare disease community at the local level by hosting regional one-day training sessions for rare disease patients, caregivers, and other advocates.

- **Young Adult Rare Disease Representatives** works to instill confidence in the next generation of rare disease advocates and provides support in the advocacy journey, especially during the transition from childhood to adulthood.

- **Rare Giving** financially supports individuals with rare diseases, as well as organizations that engage patients, caregivers, and others in the community in public policy.

GLOBAL GENES

Primary focus: Rare disease education and support  
Website: globalgenes.org

Global Genes is a global non-profit advocacy organization for individuals and families fighting rare, genetic diseases. The main purpose is to raise awareness and build community support of genetic disorders through social media, provide education to both the public and medical communities, and fund research focused on treating rare conditions.

- Its website includes **Global Genes Rare Toolkits and Rare University**, and features a portal where patients and caregivers can create a profile and connect with one other.

- Scholarships are also available for its **RARE Patient Advocacy Summit**, an annual event that brings together patients, caregivers, and other stakeholders to develop and take home actionable strategies to accelerate change.
NATIONAL ORGANIZATION FOR RARE DISORDERS

Primary focus: Rare disease public policy advocacy, education, and research
Website: rarediseases.org

NORD provides information, services, and support for patients and their families as well as patient advocacy organizations, medical professionals, and others seeking to develop new diagnostic tools and treatments for rare diseases.

- NORD provides a network of patient organizations, information, resources, frequently asked questions, and state-by-state advocacy information. It also offers patient and financial assistance programs to help support a variety of disease-related patient costs.
- Its network of ambassadors – volunteers in leadership positions – aim to build a strong community of rare disease advocates in their state to help increase awareness of rare diseases and the challenges patients and their families face.

RARE NEW ENGLAND

Primary focus: Rare disease support and education throughout New England
Website: rarenewengland.org

Rare New England (RNE) offers patients, families, and providers educational opportunities, resources, and support. The organization also provides information on genetic counseling, grief support, and regional and national events.

- Patients and families can watch episodes of RNE’s educational cable show series about rare and complex diseases.
- RNE sponsors an annual speaker series that connects patients and their families with rare disease experts to discuss the diagnostic journey. These presentations cover the experience of living with a rare disease, coping strategies, how to navigate the challenges in the healthcare and/or educational systems, among other topics.
- RNE also hosts an annual conference to provide resources, education, and networking opportunities for the rare disease community.
Caregiver Support

CAREGIVER ACTION NETWORK

Primary focus: Caregiver support
Website: rarecaregivers.org

The Caregiver Action Network (CAN) is an organization focused on supporting people who care for individuals with chronic conditions, disabilities, disease, or age-related issues. CAN also offers a rare disease caregiver-focused site that provides education, peer support, and resources to family caregivers across the country free of charge. CAN offers support and information for all caregivers, regardless of the condition.

NATIONAL ALLIANCE FOR CAREGIVING

Primary focus: Public policy advocacy regarding caregiver issues
Website: caregiving.org

The National Alliance for Caregiving (NAC) conducts research, analyzes public policy, develops national best-practice programs, and works to increase public awareness of family caregiving issues. Although not specific to LC-FAOD or rare genetic diseases, the NAC offers valuable resources and information to all types of caregivers. The NAC collaborated with Global Genes to publish Rare Disease Caregiving in America, a first-of-its-kind national research study capturing the experiences of caregivers of children and adults with a rare disease.
Organizations Outside the U.S.

There are also several organizations outside the U.S. that might have relevant information or be useful if you ever decide to visit, live or work in another country. In some cases, international organizations may collaborate with U.S.-based organizations on global issues and initiatives.

**METABOLIC SUPPORT UK**

**Primary focus:** Metabolic disease patient and caregiver support  
**Website:** metabolicsupportuk.org

Metabolic Support UK is an organization supporting patients and families with inherited metabolic disorders across the United Kingdom. It offers a **Family Advice Service and Metabolic Connect** (a peer support matching service). Metabolic Support UK also sponsors conferences, workshops, and education days, among other programs.

**MITOCANADA**

**Primary focus:** Awareness, education, support, and funding research for mitochondrial disease in Canada  
**Website:** mitocanada.org

MitoCanada is a Canadian-based organization offering support to **patients with mitochondrial disease**, their families, and caregivers. It also strives to increase public awareness of mitochondrial disease, ultimately advancing research towards disease prevention and cure.
Newborn Screening

Many people with LC-FAOD may have been or will be identified with a positive newborn screening test. A positive result means that the baby may have a higher risk of having one or more of the conditions included on the newborn screening panel. Confirmation of LC-FAOD diagnosis occurs with an additional blood, urine, or genetic test.

There are advocacy organizations that provide more information about the newborn screening process and mobilize patient advocates to influence newborn screening regulations and policies.

**BABY’S FIRST TEST**

*Primary focus:* Newborn screening information and education  
*Website:* babysfirsttest.org

Baby’s First Test provides materials and resources about newborn screening at the local, state, and national levels. The organization’s website features facts about the conditions screened and what to expect from the newborn screening process. Some topics include:

- Details on the newborn screening **tests offered in every state**
- **Tools for parents** including what to do after you receive newborn screening results
- **Condition-specific information** including causes, treatment, and family experiences

**SAVE BABIES THROUGH SCREENING FOUNDATION**

*Primary focus:* Newborn screening education and advocacy  
*Website:* savebabies.org

The Save Babies Through Screening Foundation educates parents, pediatric healthcare providers, and policy makers about the complexity of newborn screening, the importance of timely and early screening, and the importance of prompt testing to confirm a diagnosis. This confirmatory testing is a critical step and helps to allow for early treatment and management of rare disorders when indicated.

In the U.S., adoption of newborn screening for LC-FAOD occurred in a few states in 1999, while most states adopted the screening between 2002 and 2008.¹

**REFERENCES**

Living with or caring for someone with one of the long-chain fatty acid oxidation disorders (LC-FAOD) can be an overwhelming responsibility. It will require navigating the healthcare system, which can be complex and confusing.

LC-FAOD can affect multiple organs and you may need to see many healthcare providers including, but not limited to:

• Metabolic dietitian
• Geneticist
• Cardiologist
• Neurologist
• Ophthalmologist
• Gastroenterologist

As a result, you will likely need to learn how to serve as a coordinator and self-advocate. It's important to provide each member of your healthcare team with consistent, detailed information so they can get a complete picture.

It may not always be easy to talk to healthcare providers, but creating a partnership with them based on mutual trust and respect can lead to better care. It can also potentially reduce some challenges and frustrations.

Your conversations may change over time based on your understanding of and experience with the disease, and can vary depending on the type of provider and their experience with LC-FAOD. Here we provide some tips that may help you feel empowered so that you can successfully communicate and collaborate with providers.

## TAKE TIME TO PREPARE

- **Note questions or discussion points.** Write down any questions you may have and think about what information you want to learn at this visit. Bring this list with you and write down the answers, taking time to repeat them back to ensure you’ve captured everything.
  - What symptoms should I expect? Could these change over time?
  - Am I at risk for developing other health conditions because I’m diagnosed with LC-FAOD?
  - What lab tests or procedures should I expect to regularly undergo?
  - How do you prefer to communicate with your patients and families in between visits?
  - What should I do in the case of an emergency, both when in my local area and while traveling?

- **Do your homework.** Learn as much as possible about the disease and current research. Your healthcare team, government agencies, patient advocacy organizations, peer-reviewed journals that offer free, open access to scientific articles, and professional organizations are good resources for reliable information.

One example is the [International Network for Fatty Acid Oxidation Research and Management](https://informnetwork.org/inform-families/) (INFORM). It's INFORM Families section has information about all of the LC-FAOD types and stories of people living with LC-FAOD. (https://informnetwork.org/inform-families/)

See the [Building Your Support Network](#) section of this toolkit for more detailed information about resources from advocacy organizations.
• **Create a complete medical history.** Some ideas may include:
  - A list of your current providers and their contact information.
  - Details about all medications, exact doses, when you take them, and who prescribed them.
  - A simple reference or description of your condition to share with new providers.
  - Organizing your information in one place such as a notebook, binder, or online file. Be sure it is easy to access and make extra copies to share with your healthcare providers.

**GET THE MOST FROM YOUR VISIT**

• **Be thorough, honest, and factual.** You may be tempted to downplay or perhaps exaggerate symptoms; instead, be realistic about your experiences.
  - Tell healthcare providers your specific symptoms, how often you experience them, how those symptoms interfere with your daily activities, and whether they impact your emotional or mental health.
  - Use numbers to explain symptoms like fatigue, energy level or pain. For example, many healthcare providers use scales or pictures to understand if your symptoms are not noticeable, mild, or at their absolute worst.

• **It's okay to repeat yourself, speak up, ask questions, and take notes.** If there is something you want to make sure your healthcare providers know, repeat it. It is also important that you understand the information the healthcare providers share with you, including lab and test results. Ask your providers clarifying questions, to repeat what they said or for a print out of the information discussed. Be mindful of your words and use statements like “I don't understand” or “I would like to discuss this further.”

• **Let your child speak for him/herself when possible.** This can ease a child into the adult transition down the road. It is important for a child to learn about how the disease affects them and how to advocate for themself.

**CREATE YOUR BEST TEAM**

• **Build mutual respect.** Trusting and respecting your team’s medical expertise helps to form the foundation for a strong partnership. Carefully consider your team’s medical expertise while making sure to state what you disagree with or would like to further discuss.

• **Feel comfortable with your healthcare team.** You should feel confident in how a healthcare provider is managing your child’s or your care. It may take some time to establish a connection with your team. Maximizing your time at the office by preparing questions in advance and ensuring questions are answered will help you gain confidence in your team.

• **It’s okay to ask for a second opinion and explore your options.** If you’re happy with your healthcare team but unsure about their advice or recommendations, you may want to seek a second opinion.
  - Be upfront about your desire to gain a few perspectives to make an informed decision.
  - Be sure to check with your insurance company first to understand if you have coverage for a second opinion.

Knowing the information that is important to your healthcare team and what to include in your medical history can be a challenge. This toolkit includes a [Medical History Template](https://vimeo.com/178253392) you can use to get started.

For more information, check out the “Keys to Effectively Communicate with Healthcare Providers” webinar from Global Genes. ([https://vimeo.com/178253392](https://vimeo.com/178253392))

MRCM-UX007-00070 January 2020

FAODinfocus.com
This is a sample medical history template that you can use to prepare for appointments with existing or new healthcare providers. Your medical history, goals, and questions will change over time. Keeping an up-to-date medical history and providing copies to your healthcare team can help you make the most of your office visits. List any ideas, questions or concerns you want to discuss at your next appointment, or support you need for navigating the healthcare system.

**DISEASE AND JOURNEY OVERVIEW**

In this section, include information such as your primary diagnosis and important details about it, when and how it happened, any misdiagnoses, and key medical events since your diagnosis. You can also include your current health goals and anything else that might be helpful for your healthcare team to know about your disease and experiences.

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<th>Symptom</th>
<th>Description</th>
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**OTHER CONDITIONS (INCLUDE DATE OF DIAGNOSIS, IF KNOWN):**

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2. 
3. 
4. 

**ALLERGIES OR FOOD SENSITIVITIES OR PREFERENCES:**
**CURRENT AND PAST MEDICATIONS:**

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<th>Medication/Supplement</th>
<th>Dose</th>
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**MEDICATIONS YOU CANNOT BE TREATED WITH:**

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**FAMILY HISTORY:**

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<th>Medication/Treatment</th>
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**HEALTHCARE PROVIDER CONTACT INFORMATION:**

Primary LC-FAOD Healthcare Provider(s) (the person responsible for your day-to-day LC-FAOD management)

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<th>Institution</th>
<th>Phone</th>
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Additional Healthcare Providers

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**ADDITIONAL NOTES OR IMPORTANT TOPICS:**

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Living Well While Caring for Someone with LC-FAOD

Being a parent or caregiver to a child or adult with a long-chain fatty acid oxidation disorder (LC-FAOD) requires around-the-clock commitment, which can include:

- Carefully managing diet and nutrition
- Monitoring energy levels
- Watching for hard-to-predict and frequently changing symptoms

This can make it extremely hard to entrust your child or loved one's care to another person. It can feel overwhelming, stressful, and unsettling. It's normal to worry. But it's important to remember that you can better care for your child if you also take care of yourself.

Inside find information on how to **prepare** and **educate** others to care for your child and **help you** get the breather you need.

“LC-FAOD is complex but manageable and these children can have a good life with the right care.”

-Michelle, mom to Jake, who is living with LCHAD deficiency

**A first step may be identifying who can help.** For example, are there certain people who regularly visit with your child or loved one? Do you have extended family or close friends who live nearby? Are there certain people with whom you have built a close, trusted relationship?
Prepare Others

EXPLAIN LC-FAOD

- Help your designated caregivers understand what LC-FAOD is and how it affects the body. Provide disease information (find some at informnetwork.org/inform-families) in advance; give caregivers enough time to digest the information and to ask questions.
- Describe how LC-FAOD affects your child. Explain the specific signs and symptoms that your child experiences, what actions or circumstances might trigger a crisis, and what may indicate that something could be wrong.
- Invite potential caregivers over to prepare meals or medical formulas together. After you’ve done this a few times, have the caregiver do most of the work while you’re there.
- Carefully explain disease management needs. If you prepare food, medical formula, and/or dietary supplements in advance, mark each item with the time your child should take it, the amount they should take, and place the items in a designated area.

CREATE “CHEAT SHEETS”

Laminated cheat sheets or files on your phone that are easily shareable can be helpful tools for quick reference. Separate out the most important information so it is easy to spot and find. Arming your caregivers with this information will give them (and you) more confidence.

Be sure to include:

- A meal/feeding schedule: While most children snack during the day, you know it’s even more important for kids with LC-FAOD to eat at regular intervals. Specify the times at which your child will need to eat something and indicate whether it’s a full meal or snack.
- A list of dietary supplements or medical formulas, including doses, and when and how they should be prepared and/or taken
- A list that explains:
  - Foods they cannot have
  - Foods they can eat in limited amounts
  - Foods they can eat as much of as desired
  - Food preferences and favorites
- A list that explains limitations your child may have regarding certain activities, for example:
  - How far or long they are able to walk before they may experience pain or become unable to return without assistance
  - Specific activities or exercises they can enjoy in moderation
- Warning signs/symptoms to watch out for and what to do when they occur
- Key phone numbers (family, doctors, nearby hospitals)
TAKE THESE HELPFUL STEPS

There are additional ways to make sure others know how best to care for your child:

- **Make sure they wear a medical alert tag or bracelet.** This is a great way to inform others of their condition, which could require immediate attention in an emergency.

- **Have an emergency protocol letter.** If your child needs to visit the emergency room, an emergency letter provides the healthcare team with relevant medical and contact information. Your physician may already have a template they use. If not, MitoAction, a patient support organization, provides details about all of the information you might need in an emergency (https://www.mitoaction.org/planning-for-emergencies/). Work with your healthcare team to make sure you have all of the information you need.

- **Request a Section 504 plan for school-aged kids.** Children who have a specific medical diagnosis that affects their physical abilities can qualify for a 504 plan. This plan is a formal, written document that outlines specific accommodations the child will need at school. Specific accommodations may include allowing the child to have fluids or snacks during the day, limiting strenuous exercise in physical education class, providing excused absences for medical appointments, or allowing extra time to complete assignments or exams. Ask your school counselor about how to request a 504 plan.

- **Inform care providers about the emotional impact of LC-FAOD.** Following a strict diet or eating regimen can be difficult and isolating, especially for a child. Ask that caregivers be sensitive and try to minimize calling unneeded attention to the situation.

“Being a caregiver for a person with LC-FAOD is about learning how to step outside your comfort zone and continuously put the needs of another person above your own. Take each day one hour at a time, one feeding at a time. Remember to take care of yourself and your other relationships. Learn how to navigate the healthcare system and advocate for your family.”

- Jennifer, mom to Ava, living with VLCAD deficiency
Focus on Your Health

Forty percent (40%) of rare disease caregivers report having fair or poor emotional or mental health.1 Although you likely realize how important it is to take care of yourself, it may not always feel like something you can do. But taking care of yourself is just as important as taking care of your child.

Taking some time for yourself can be an important way to recharge and de-stress, ultimately helping ensure that you have the energy and focus to care for your child.

It may help to:

- **Talk to a healthcare provider or mental health professional,** such as a counselor, psychologist, or psychiatrist if you're experiencing symptoms of depression. These symptoms may include a persistent sad, anxious, or “empty” mood, loss of interest in or pleasure from activities you enjoy, feeling hopeless or helpless, difficulty concentrating or making decisions, or difficulty sleeping, among others.2
- **Connect with other caregivers.** Talking to other people who share similar experiences can be a powerful way to cope with stress and anxiety.
- **Find a stress-reducing activity** you like and try to make time for it or incorporate it into your routine; exercising, meditating, going for a walk, making art – or whatever works best for you.
- **Do what you can to stay well,** even if it’s just one small activity a day until it becomes a habit - eat a balanced meal, go to bed early to get enough sleep, or take a few deep calming breaths in a quiet place.
- **Get annual medical check-ups for yourself.** It can be easy to focus on your loved one’s medical appointments, but make sure that your calendar also includes visits with your own healthcare team.
- **Find a respite care program.** This can provide a temporary break for primary caregivers, whether in the home or at another facility.

Resources

- “The Respite Care Notebook” from the Child Neurology Foundation (childneurologyfoundation.org)
- “Rare Disease Caregiving in America” report from National Alliance for Caregiving (caregiving.org)
- “10 Tips for Family Caregivers” from The Caregiver Action Network (rarecaregivers.org)
- “Parenting a Child with a Life-Limiting Illness” toolkit from Global Genes (globalgenes.org)

REFERENCES

You’ve just received a diagnosis. Now what? A common next step is to learn as much as you can about your type of LC-FAOD, such as how to identify and manage symptoms, approaches to disease management and possible health complications that may occur in the future. You may also start to think about what the diagnosis means for you and your family and how it will impact daily life.

An important part of navigating life with LC-FAOD is identifying who needs to know about it and deciding how and when to explain it.

Who’s Listening?

A first step is to think about the people with whom you interact frequently. What do they need to know? Do they need to know every detail, or would a broad explanation be enough? How might the emphasis of your story differ from one person to the next? Are there certain factors to consider such as your age, disease management needs, or other special circumstances?

FAMILY, FRIENDS, AND LOVED ONES

Keep in mind that you may need to have these conversations more than once before loved ones understand what LC-FAOD is and your needs. Learn to accept that your experiences are your own. While others may never be able to completely understand what your diagnosis is like, have patience with loved ones who make an effort to support you. Be patient and prepared for a potentially emotional conversation.

- **Put it in context.** Compare your experience to something they might already understand.
- **Find time to have a one-on-one conversation** in a casual setting, not at a large family or group gathering. This will give them, and you, the appropriate focus.
- **Talk before a potential emergency** when it will be harder to process and understand.
- **Give them time to digest,** knowing that some may not be sure what to say or how to help.
- **Be open to discussing emotional and physical needs** of other family members, particularly siblings of a diagnosed child.
- **Leave the conversation open-ended** and ask them if they have any questions.

**DOCTORS AND HEALTHCARE PROVIDERS**

Because it’s rare, you will need to continually educate healthcare providers who aren’t familiar with LC-FAOD. To make this easier, build and maintain a care notebook/binder to keep all important information in one place. You may also choose to save some information on your phone or tablet for easy reference.

You’ll most likely have regular visits with a variety of healthcare providers, each one playing a different role in the care of LC-FAOD. You may also interact with emergency care personnel who will need to quickly access certain medical information. You can help facilitate better care by making sure they have complete information. See Communicating with Healthcare Providers in this toolkit for additional tips.
EMPLOYERS OR COWORKERS

You aren’t legally required to tell your employer or coworkers. But, if you are in a supportive work environment, discussing your needs can be beneficial so you know about policies or accommodations that may be available to help you with your caregiving or disease management responsibilities. The Family Medical Leave Act (FMLA) is a federal law which allows an employee to take up to 12 weeks off per year for medical emergencies (this is unpaid time). Learn more about FMLA here: https://www.dol.gov/agencies/whd/fmla.

TEACHERS, SCHOOL STAFF, AND NURSES

Have your go-to resource (an article, pamphlet, toolkit, etc.) ready to help explain LC-FAOD and any specific needs resulting from the condition. There are accommodations required by law for people with medical needs. Be prepared to work with the school to develop a plan. It may take several conversations, but advocacy organizations have tools to help. See the Global Genes School Advocacy Toolkit for tips on navigating school systems with a rare condition here: https://globalgenes.org/wp-content/uploads/2018/10/School-Advocacy_ToolKit_spread_DIGITAL.pdf

Advocate to a Broader Audience

Once you’ve mastered being an advocate in your everyday life, there are other ways you can make a broader impact – for yourself and for others living with LC-FAOD, or other rare diseases.

LEGISLATIVE OUTREACH

Reaching out to government representatives about your needs can influence and inform changes to healthcare policy. Reach out to EveryLife Foundation for Rare Diseases or the National Organization for Rare Disorders to get started.

AWARENESS BUILDING

Sharing information about LC-FAOD with reporters and community members in person and on social media can increase awareness and help raise funds for research. Some patient advocacy organizations have resources to kick off your efforts. For example, EURORDIS sponsors Rare Disease Day every year on the last day of February and provides many ideas for spreading the word.¹

RESEARCH PARTICIPATION

Clinical trials are one form of research, but there are many others such as registries, surveys, and advisory boards. To learn more about clinical trials visit ClinicalTrials.Gov or the Center for Information and Study on Clinical Research Participation (CISCRP) at ciscrp.org. Ask your healthcare teams or patient advocacy organizations for other types of research opportunities.

REFERENCES

Crafting Your Story

Below are some things to think about when crafting and sharing your story across audiences. The first three are always important, and the rest you will refine as you practice and gain experience. It’s okay if you feel nervous or uncomfortable. The most important step is the first – getting started! Telling your story will become easier over time.

Join the conversation.
Many people are helping to advance medicine and science. In order for the rare disease puzzle to be complete, it needs to include all voices – including yours!

Outline your goals.
Decide why you’re telling your story – is it to help someone understand your needs, raise money, or change laws or the direction of research? Pinpoint what you want to accomplish so that you can effectively inspire and motivate your listeners.

Understand your audience.
Before preparing your story, it helps to know your audience. Think about their perspective, what questions they might have, and what is most important for them to know.

Remember your key messages.
Before you start, it’s helpful to write down key points to deliver. Aim for 3 to 5 specific messages, with no more than 1 to 3 sentences each.

Nail down your elevator speech.
An elevator speech should be as short as the time it takes to ride in an elevator (about 60 to 90 seconds). Introduce yourself, describe your condition and its impact, and preview what you want your audience to do.

Elevate your confidence.
Practice your elevator speech with a friend or family member, or use your phone to record yourself. Ask the following questions:
• How did it sound? Interesting? Boring?
• Did you keep it short and clear, without extra words (you know, um, like)? Did you include a call to action?

Note what to change and go for it again! Think about how you’ll answer easy and hard questions.

You are the expert!
Your story belongs to YOU. While opinions and priorities may differ for each audience, no one can question your experience. You get to decide what to share and when.

Do these things for an impactful story
1. Offer background information, external resources
2. Get your key messages in early
3. Show your audience how your story applies to them
4. Be concise – less is more!
5. Be authentic – don’t memorize your answer
6. Balance fact and emotion