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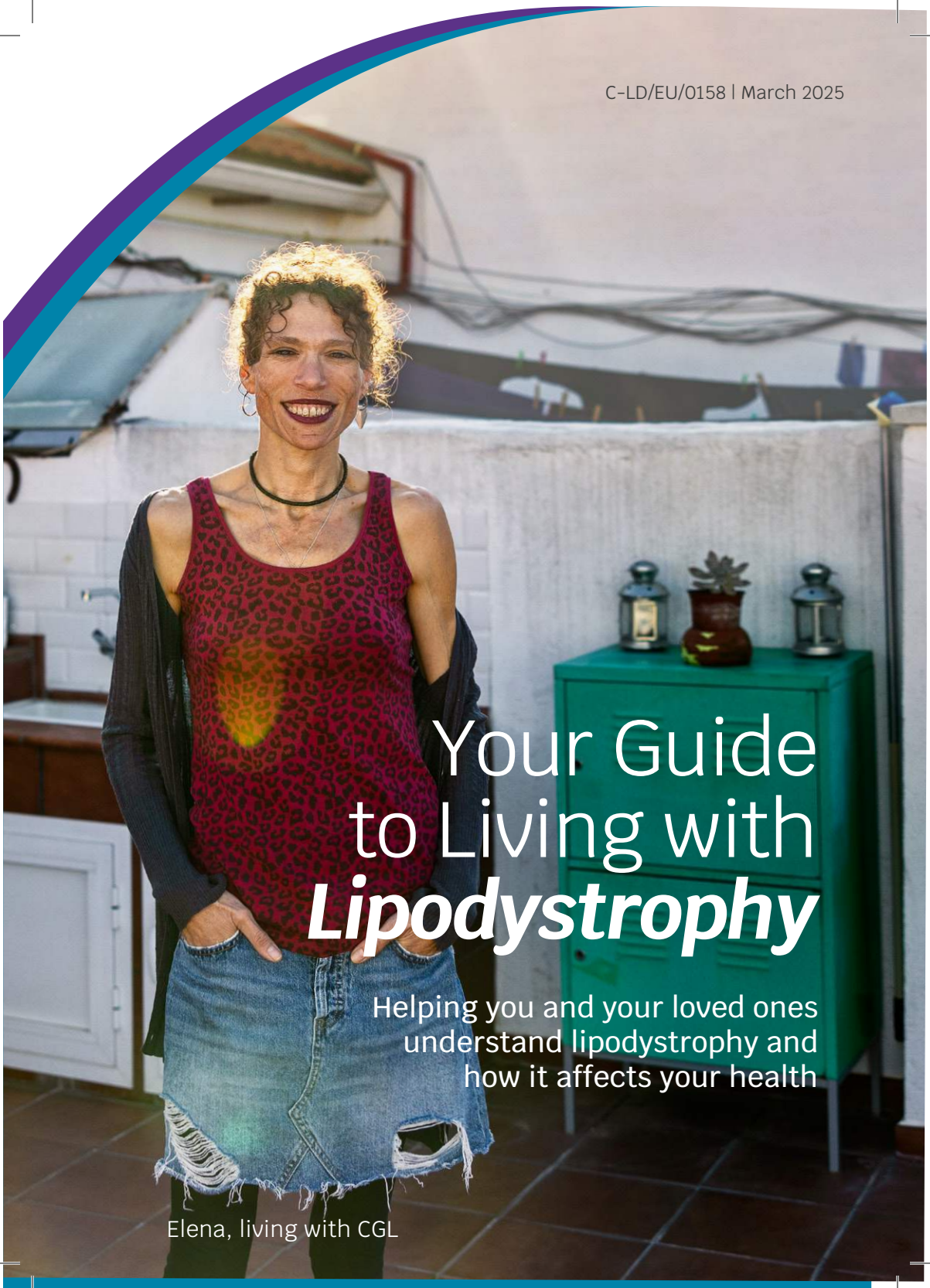

Your Guide to Living with *Lipodystrophy*

Helping you and your loved ones understand lipodystrophy and how it affects your health

Elena, living with CGL

Patricia, living with FPLD

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Your Guide to

Lipodystrophy

About This Booklet

- This booklet is here to help you and your family understand lipodystrophy and how it affects your health. It offers important information on how to manage the condition and navigate the challenges that may come with your diagnosis.
- The goal of this booklet is to give you the knowledge and resources you need to support your journey with lipodystrophy.
- Understanding lipodystrophy can help you and your family navigate living with this condition, managing health issues effectively and accessing the right care.
- Since lipodystrophy is rare and can be misunderstood, getting an accurate diagnosis is very important. Learning about treatment options will empower you to make informed choices about your health and well-being.
- You'll notice glossary terms are in ***bold*** and these terms are defined at the back of the booklet.

Your Doctor: _____

Your next appointments: _____

Your medication: _____

Notes: _____

A Note From Your Patient Group Leaders

Dear Reader,

We know that being diagnosed with lipodystrophy can feel like a lot to take in. It is okay to feel unsure or even a bit scared right now. We have been where you are, and we want you to know that you are not alone.

There is a whole community of people living with lipodystrophy who understand what you are going through. Many of us have felt the same way when we were first diagnosed. But with time, support, and the right information, things do get easier. A diagnosis is not the end, but rather the beginning of a new chapter in understanding your health and taking control of your journey.

Elena, living with CGL

This guide is here to help you with the first few weeks and months. Take your time going through it—there is no rush. It is filled with information that can help you understand more about lipodystrophy and what to expect.

Remember, this is just the beginning, and you have got a whole community ready to support you every step of the way.

Lipodystrophy UK
Hope · Awareness · Support

AFLIP
Association Française des Lipodystrophies

Lipodystrophy United - Israel

AILIP
ASSOCIAZIONE ITALIANA LIPODISTROFIE

Lipodystrophy United

AELIP
Asociación Internacional de Familiares y Afectados de Lipodistrofias
www.aelip.org

Lipodystrophy Canada

Introduction to Lipodystrophy

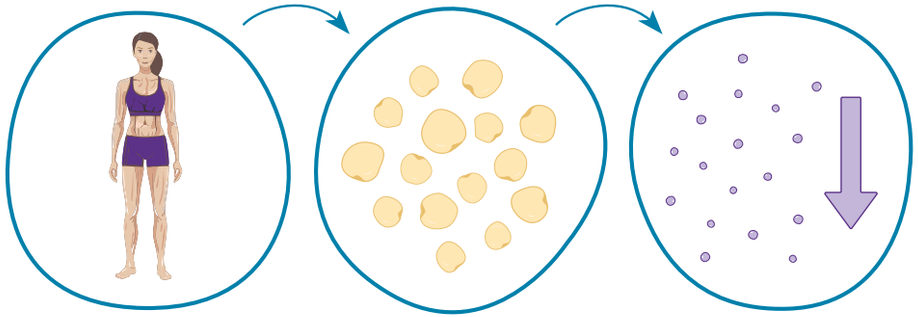
What is Lipodystrophy?

Lipodystrophy is a rare disease characterized by an abnormal distribution of fat throughout a person's body, generally leading to progressive metabolic disease.

The word can be broken down as below:

Lipo means fat | ***Dys means bad*** | ***Trophy means growth***

Lipo-dys-trophy means bad growth of fat



Lipodystrophy | **Reduced Fat Tissue** | **Decreased Leptin Production**

Body fat, known as adipose tissue, is essential for energy storage, hormone production, and providing insulation to the body. It produces ***leptin***, a hormone that helps to regulate appetite and metabolism. People living with lipodystrophy often have either too little or no fat in certain parts of their bodies. This fat loss may be ***generalized*** (affecting the whole body) or ***partial*** (affecting specific areas), which can lead to excessive fat storage in other areas. Due to the loss of fat, ***leptin*** levels can be low, leading to complications of ***leptin*** deficiency. You can read more about health complications on page ***16-18***.

Types of Lipodystrophy

Lipodystrophy is a complex and variable disease. While the disease can vary from person to person, there are two main forms of lipodystrophy:

Generalized Lipodystrophy (GL)

In ***generalized lipodystrophy***, there is a near total absence of ***adipose tissue*** from the entire body.

GL occurs in around 1 per million people globally.

Partial Lipodystrophy (PL)

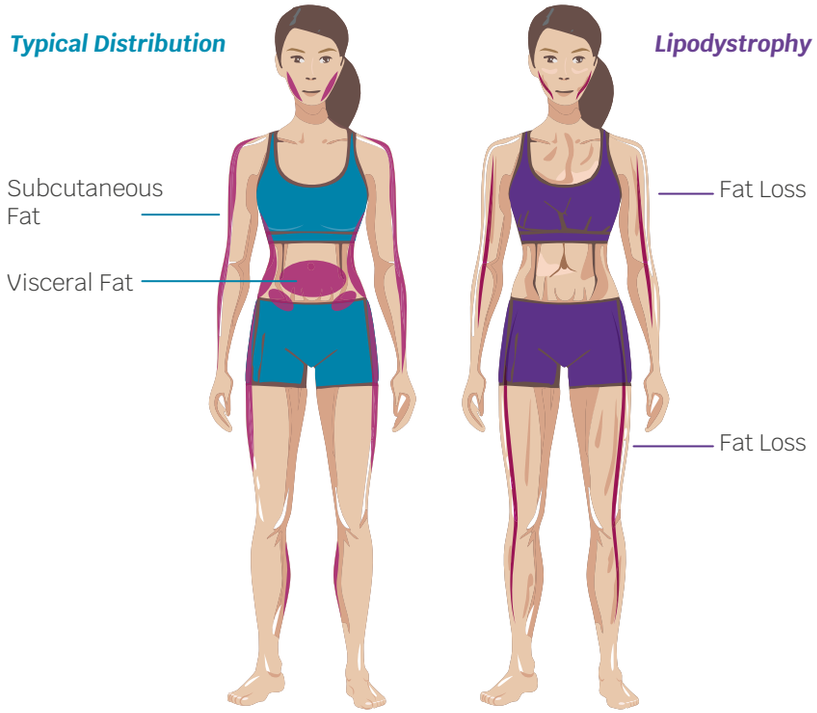
In ***partial lipodystrophy***, specific areas of the body are affected by fat loss, while others might retain fat or even have excess fat. This phenotype may not become apparent until after puberty.

PL is slightly more common at around 3 per million people globally, although it is notable that PL is significantly under diagnosed and actual prevalence rates are deemed to be much higher.

Learn About Lipodystrophy:
Take time to understand the condition, its types, and potential complications. Ask your healthcare team for reliable resources to learn more.

Usually, people lose or gain weight due to changes in the amount of fat stored within ***adipose tissue***. However, in ***lipodystrophy***, the body's capacity to store fat is reduced or lost. This means the body tends to store fat in areas where it would not typically be stored, like the liver or muscles. This is known as ***ectopic fat***.

Typical Fat Distribution vs Fat Distribution in Lipodystrophy Patients

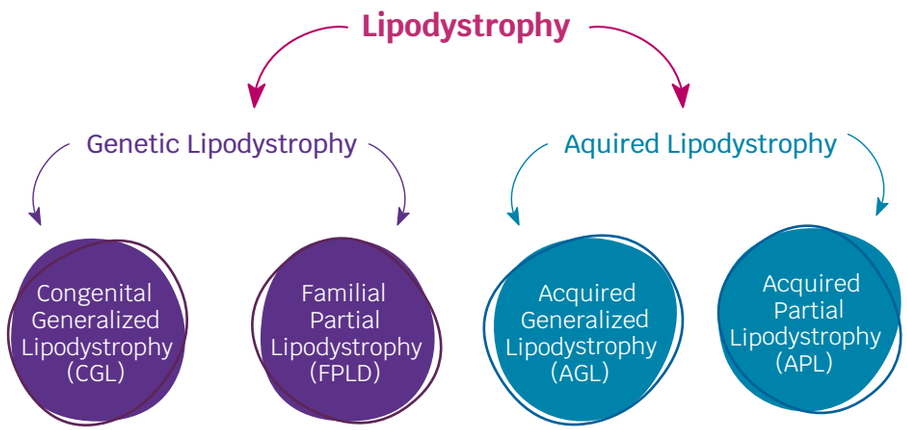


This abnormal fat storage affects physical appearance and can lead to significant health problems. The extent of fat loss often correlates with the severity of these health complications; however, your healthcare team will be able to provide more information on how lipodystrophy affects you and your health.

What Causes Lipodystrophy?

Both ***generalized lipodystrophy*** (GL) and ***partial lipodystrophy*** (PL) can be either genetic or acquired. This results in the four main subtypes of lipodystrophy, which are explained in more detail below.

Different Types of Lipodystrophy



Ashley, living with FPLD

Genetic Lipodystrophy:

Genetic types of lipodystrophy are caused by pathological changes (mutations) in our DNA, which can be passed down through families. These types are often called ***inherited, familial,*** or ***congenital.***

More than 20 genes have been linked to different forms of genetic lipodystrophy. Many have been discovered recently, so it is likely that there are additional genes yet to be discovered. Most of these genes appear to play a role in the development, function or survival of ***adipose tissue***, leading to either partial or generalized forms of lipodystrophy (e.g. LMNA).

Speak to your doctor about your subtype and what this could mean for your family.

1. ***Congenital Generalized Lipodystrophy (CGL)***: Individuals with ***CGL*** lack almost all body fat from birth. While the absence of fat is present early, the diagnosis is often made in childhood or even adulthood when metabolic complications become apparent. Those with ***CGL*** may have prominent muscles, and severe metabolic complications, including insulin resistance, hypertriglyceridaemia and fatty liver.

2. ***Familial Partial Lipodystrophy (FPLD)***: Physical changes in fat distribution in ***FPLD*** can present as fat loss, mainly in the limbs and buttocks, with fat remaining or increasing around the neck and face. These features often become more noticeable around puberty, though metabolic changes may develop at different times. It is often associated with insulin resistance, high ***triglycerides***, and lipoatrophic diabetes (sometimes misdiagnosed as type 2 diabetes).

Acquired Lipodystrophy:

Acquired lipodystrophy means it is not ***inherited*** and may be caused by other factors. Autoimmunity, in which the body mistakenly attacks its own cells, is a common finding in patients with acquired lipodystrophy. However, in some cases, the cause of acquired lipodystrophy is unclear.

Acquired lipodystrophies can develop at any stage of life and, like genetic types, can cause a complete or partial loss of fat tissue.

1. ***Acquired Generalised Lipodystrophy (AGL)***: ***AGL*** can develop at any age and is often associated with autoimmunity. It causes widespread fat loss, which may lead to high blood sugar, raised ***triglycerides***, and fatty liver.

2. ***Acquired Partial Lipodystrophy (APL)***: ***APL*** is often triggered by autoimmunity and mainly causes fat loss in the upper body, including the face, arms, and trunk, while the lower body retains or accumulates fat.

In addition to these categories, there are other rare types of lipodystrophy, for example progeroid syndromes and complex genetic syndromes. Please note, localized lipodystrophy and HIV-associated are not covered within this booklet. For more information on these forms, please speak with your healthcare team.

Your healthcare team will be able to provide more information regarding your condition. For more information on the symptoms of these different subtypes of lipodystrophy, please refer to page ***14*** of this booklet.

Symptoms of Lipodystrophy

People with lipodystrophy are affected in a variety of ways. This section offers an overview of:

- **Some of the most common symptoms**
- **A breakdown of how symptoms vary based on specific subtypes of lipodystrophy**
- **Real-life experiences from other patients and families who have navigated similar journeys**

Track Your Symptoms
Keep a journal of your symptoms, treatments, and lifestyle changes to help your healthcare team adjust your plan as needed.

Prepare for Symptom Changes
Symptoms can change over time. Work with your healthcare team to adapt your management plan as needed.

Common Symptoms of Lipodystrophy

Changes to Your Physical Appearance:

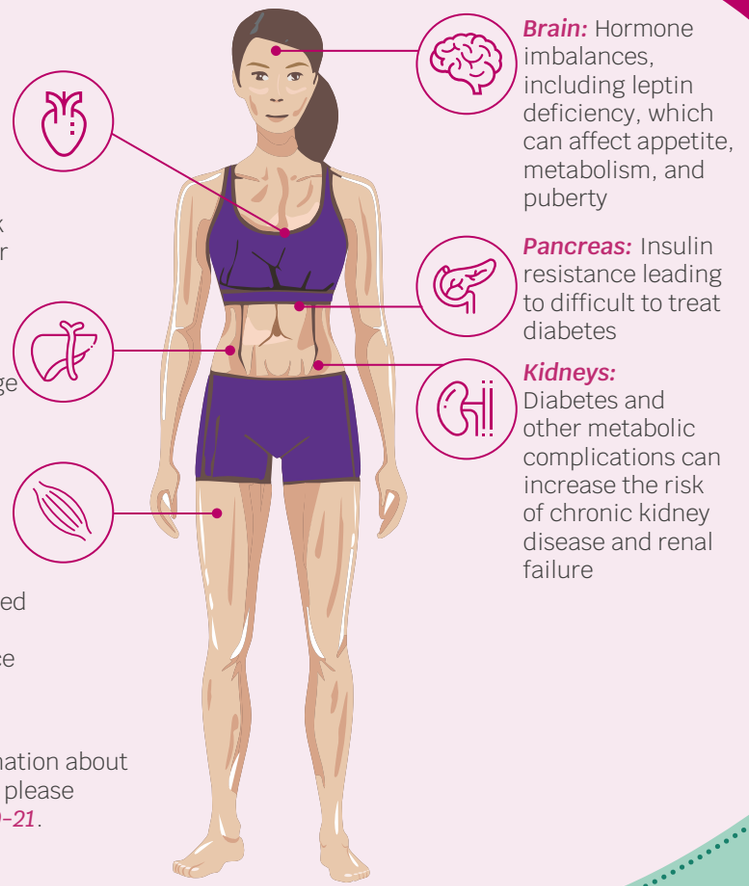
People with lipodystrophy often notice significant changes in their physical appearance due to differences in how their bodies store or lack fat. The condition can affect multiple organs, and while symptoms vary depending on the individual and subtype, some of the most common physical features include:

- **Reduced fat** ***(lipoatrophy)***: Many people with lipodystrophy lack fat in certain areas like the face, arms, or legs. In generalized types, fat is absent throughout the body, leading to a distinct, leaner look.
- **Increased fat**: In certain forms of ***partial lipodystrophy (PL)***, there can be an increase in ***adipose tissue*** in certain parts of the body, typically the face (double chin), neck ('Cushingoid appearance' of facial puffiness), and between the shoulder blades (buffalo hump). Some women also experience increased ***adipose tissue*** in the labia and some men can experience an increase in pubic fat.
- ***Muscular hypertrophy***: People with lipodystrophy quite often have muscles that appear more defined and prominent, creating a naturally muscular and athletic appearance even without exercise, especially in the arms and legs.
- ***Acanthosis Nigricans***: Dark, thickened skin patches often appear in body folds (neck, armpits, groin) due to insulin resistance, which is common in lipodystrophy.
- **Xanthoma**: Xanthomas are fatty deposits in the skin due to elevated blood lipid levels.

Metabolic Complications

Adipose (fat) tissue plays a vital role in the body by storing energy, regulating metabolism, and providing insulation to the body. In lipodystrophy, the loss of ***adipose tissue*** disrupts these essential functions, resulting in a range of complications. Below is an overview of some of the most common metabolic complications and their potential impact on health:

Potential Metabolic Complications of Lipodystrophy



Heart & Blood Vessels: High triglycerides and other lipid abnormalities increase the risk of cardiovascular disease

Liver: Fatty liver disease due to excess fat storage and lipid uptake

Muscles: Fat accumulation and increased glucose uptake leading to reduced function and insulin resistance

Brain: Hormone imbalances, including leptin deficiency, which can affect appetite, metabolism, and puberty

Pancreas: Insulin resistance leading to difficult to treat diabetes

Kidneys: Diabetes and other metabolic complications can increase the risk of chronic kidney disease and renal failure

For more information about co-morbidities, please refer to page *20–21*.

- **Hypertriglyceridaemia**: People with lipodystrophy often have high levels of ***triglycerides***, a type of fat in the blood. Severely elevated ***triglycerides*** can trigger an episode of acute pancreatitis. Elevated triglycerides in lipodystrophy may contribute to increased risk of developing cardiovascular issues, like heart disease.

- **Insulin resistance and diabetes mellitus**: ***Adipose tissue*** plays a key role in regulating blood sugar by storing energy in the form of fat. When ***adipose tissue*** is absent or reduced, the body becomes less responsive to insulin—the hormone responsible for regulating blood sugar. This can result in insulin resistance, making it harder for the body to lower blood sugar levels, and may eventually lead to diabetes mellitus.

- **Leptin deficiency *(hypoleptinemia)***: ***Leptin*** is a hormone produced by fat cells that helps regulate appetite and energy balance by signalling the brain when the body has enough stored energy. In lipodystrophy, the loss of fat cells reduces leptin production, leading to absolute or relative deficiency. This results in increased appetite and ***hyperphagia***, even when the body has recently eaten or doesn't need more energy. This can result in overeating and the worsening of metabolic complications.
  ~ *Note, **leptin** values are not a diagnostic tool.*

Speak to Your Healthcare Team

Patients with Lipodystrophy may develop Lipoatrophic Diabetes, which can often explain why the body might not respond to "normal" doses of insulin.

- **Acute pancreatitis:** Severe ***hypertriglyceridemia*** can sometimes lead to acute pancreatitis, a painful inflammation of the pancreas. Acute pancreatitis is a medical emergency that causes intense abdominal pain, nausea, and vomiting, and requires prompt treatment to avoid complications.

These metabolic complications can also increase the risk of further health issues:

- **Heart disease**: High ***triglyceride*** levels, insulin resistance, and diabetes all increase the risk of heart disease, which can include heart attacks and other cardiovascular issues. Monitoring and managing these factors is essential to help reduce cardiovascular risks.

- ***Cardiomyopathy***: Some people with lipodystrophy may develop ***cardiomyopathy***, a disease that affects the heart muscle. If cardiomyopathy leads to heart failure, it can result in people feeling fatigue, shortness of breath, and reduced energy levels.

- **Fatty liver disease** ***(hepatic steatosis)***: Excess fat can build up in the liver due to the metabolic changes associated with lipodystrophy. This buildup, known as ***hepatic steatosis***, can lead to liver inflammation and, over time, increase the risk of liver damage or liver failure.

- **Chronic kidney disease**: People with diabetes and other metabolic complications related to lipodystrophy have a higher risk of developing chronic kidney disease, where the kidneys gradually lose function. Chronic kidney disease may require close monitoring and, in some cases, additional medications to protect kidney health.

- **Organ damage**: Organ damage occurs due to poor metabolic control, and sometimes due to underlying etiologies such as molecular factors or autoimmunity, depending on the type of lipodystrophy.

Speak to your healthcare team for further information about symptoms and metabolic complications. To help track your symptoms, please refer to page ***50–51*** of this booklet.

My lipodystrophy diagnosis explained my lifetime of health challenges and provided me the opportunity to understand better what was happening with my health. Learning more about the disease helps me to manage, slow, and in some cases prevent further serious complications associated with the disease

Andra, living with FPLD

Quality of Life Impacts

Lipodystrophy affects more than just metabolism—it can impact physical health, emotional well-being, and daily life in ways that may challenge overall quality of life. These include:

Intense hunger *(hyperphagia)*:
Many patients describe a constant and sometimes overwhelming sense of hunger, which is difficult to manage even with regular meals.

Muscle pain and joint pain:
Chronic pain can occur and impact daily mobility and overall quality of life.

Fatigue:
Persistent tiredness, even after a full night's rest, can be a common struggle for many patients.

Social anxiety and limitations:
Physical symptoms may lead to self-consciousness and discomfort in social situations.

Sex hormones and fertility:
Insulin resistance can disrupt ovarian hormone balance, leading to irregular periods, excessive hair growth, and acne in females with Lipodystrophy. This can cause symptoms similar to a condition called polycystic ovary syndrome (PCOS).

The lack of adipose tissue in lipodystrophy can result in very low or undetectable leptin levels, which contributes to reproductive problems and the onset of puberty.

Other Symptoms

In some cases, people with lipodystrophy may also experience:

- **Bone cysts**: Fluid-filled or hollow spaces that can develop in bones in some types of lipodystrophy.
- **Scoliosis**: Curvature of the spine, which is seen in some patients.
- **Concern with appearance**: Fat loss may cause self-consciousness, impacting confidence and mental well-being.
- **Non-alcoholic fatty liver disease (NASH)**: A condition in which the liver builds up excessive fat deposits.
- **Metabolic dysfunction-associated steatotic liver disease (MASLD)**: A condition where excess fat accumulates in the liver due to metabolic dysfunction, not caused by alcohol.
- **Metabolic dysfunction-associated steatohepatitis (MASH)**: A more severe form of MASLD, involving liver inflammation and damage due to fat buildup.
- **Sleep apnea**: A sleep disorder in which breathing repeatedly stops and starts. This can be experienced by ***FPLD*** patients.

These symptoms may not affect everyone, but are worth discussing with your healthcare team if they occur.

Lipodystrophy is a rare disease, and several symptoms and ***comorbidities*** can be linked to your condition. Speak to your healthcare team about managing your symptoms, ***comorbidities*** and the impact of lipodystrophy on your quality of life. Local patient organizations may also be able to guide you to appropriate clothing, footwear and mobility accessories to help ease aches and pains.

I can finish a full meal and think I'm full but then go for round 2 in a blink of an eye

Amanda, living with Familial Partial Lipodystrophy

Managing Your Mental Health and Lipodystrophy

Living with a chronic condition is undeniably challenging. It affects not only your physical well-being but also your mental health. As the disease progresses and changes over time, so do the obstacles you face. Many patients experience mental health issues such as social anxiety, general anxiety, and depression.

The 2024 QuaLip study showed over 25% of 65 adult patients* were diagnosed with a psychiatric disorder (e.g., depressive episodes, mixed anxiety and depressive disorder, anxiety disorder, adjustment disorder). Lipodystrophy disease and Quality of Life (QoL) questionnaires revealed a significant disease burden over the study period.

- More than one-third of patients reported depression symptoms on the Beck Depression Inventory.
- Patients reported physical appearance, fatigue, and pain contributed to their overall disease burden and QoL scores were lower in patients with psychiatric disease and in those with poor metabolic control.
- The study showed a significant portion of patients experienced psychiatric disorders and without the right interventions and support, they can remain under diagnosed.

If you notice changes in your mental state or find yourself struggling, seek support from a healthcare professional, in addition to a trusted family member, friend or patient organization. Remember, you are not alone. Your healthcare team and local patient organizations are there to support you on your journey – they can guide you to helpful resources and patient groups. Support is available for both you and your loved ones, helping you navigate these challenges together.

*Two subjects refused psychiatric assessment.

It can be really hard to look different, but there's always someone like you in our community
Julie living with FPLD type 3

It was so comforting to hear that so many others have the same health and mental struggles that I have
Person living with Lipodystrophy

Elena, living with CGL

Symptom Variation by Lipodystrophy Type

Congenital Generalized Lipodystrophy (CGL)

CGL is inherited and presents from birth or shortly after. Symptoms include near-total loss of fat throughout the body, severe insulin resistance, prominent muscles, and extreme hunger.

Familial Partial Lipodystrophy (FPLD)

FPLD is inherited but often does not appear until adolescence. Fat loss tends to be limited to certain areas, such as the arms, legs, and trunk, while fat may accumulate around the neck, face, and abdomen. Patients with ***FPLD*** often face insulin resistance, diabetes, and lipid abnormalities as primary symptoms, with varying degrees of hunger and fatigue.

Acquired Generalized Lipodystrophy (AGL)

AGL can develop at any time point throughout your life and usually develops suddenly, often associated with autoimmunity. Symptoms closely resemble those of ***CGL***, with a total or near-total fat loss, severe metabolic disturbances, and high levels of hunger.

Acquired Partial Lipodystrophy (APL)

In this form, fat loss typically begins in the face and gradually affects the upper body. Though less severe than other forms, people with ***APL*** may still experience metabolic issues like diabetes, high ***triglycerides***, and moderate to severe hunger.

Ashley, living with FPLD

The Importance of Early Diagnosis and Management

Lipodystrophy can be progressive, which means that the condition can worsen over time. Therefore, early diagnosis and management are crucial. Early diagnosis and consistent management can help slow disease progression and reduce metabolic complications, which may otherwise damage vital organs.

Early diagnosis allows your healthcare team to take prompt action, helping you manage your condition more effectively. This proactive approach can improve your overall health, reduce the risk of complications, and support a healthier lifestyle.

If you suspect you have lipodystrophy or have already been diagnosed, it's important to work closely with your healthcare team to effectively monitor and manage the condition.

Once I got my diagnosis I could begin to accept my body changes

Laura living with partial lipodystrophy type 1

It felt like I was actually being heard and believed and that my symptoms finally had a name

Krista living with acquired partial lipodystrophy

Ryan, living with CGL

Receiving a Diagnosis

As a rare disease, awareness of lipodystrophy is generally low – even among medical professionals – and can take years to diagnose. When symptoms are spotted, they can also be mistaken for other conditions before a diagnosis of lipodystrophy is eventually made.

In ***generalized lipodystrophy (GL)***, there is often a complete lack of body fat from an early age. Parents and healthcare professionals are often able to spot this easier.

In ***partial lipodystrophy (PL)***, the path to diagnosis can be more complicated. The pattern of body fat loss is often less predictable and less pronounced, so this symptom alone may not lead directly to a diagnosis, as it does in GL. Other symptoms vary greatly, presenting in different ways and at different times for each person. This can lead to people being incorrectly diagnosed with other conditions that share similar symptoms. Some of the **common misdiagnoses** are listed below.

- Cushing's Syndrome
- Polycystic Ovary Syndrome (PCOS)
- Metabolic Syndrome
- Type 2 Diabetes
- Other disorders that present ***lipid*** abnormalities
- Truncal obesity

In 2024, a panel of lipodystrophy experts outlined key steps for diagnosing suspected lipodystrophy. These include:

Clinical assessment: A healthcare professional will take a detailed medical history, including family history, to look for signs of lipodystrophy and assess fat distribution.

Physical examination: A thorough exam helps identify changes in body composition, such as areas of fat loss or abnormal fat accumulation.

Blood tests: These assess the body's response to insulin, measure lipid levels (such as ***cholesterol*** and ***triglycerides***), and check liver function. ***Leptin*** levels may also be measured, as low ***leptin*** can be associated with lipodystrophy, though it is not a diagnostic marker on its own.

Imaging tests: A doctor may order imaging studies, such as a DXA scan, MRI, or ultrasound, to assess body fat distribution and detect any liver abnormalities.

Heart evaluation: Since lipodystrophy can affect the heart, tests like an ***electrocardiogram (ECG)*** or echocardiogram may be used to check for abnormalities.

Genetic testing: If a hereditary form of lipodystrophy is suspected, a genetic test from a blood sample may be recommended to confirm the diagnosis.

Read Up!

There are many diseases associated with fat disorders. Please ensure you are reading about the correct condition to avoid any confusion or misunderstanding.

Treatment Options for Lipodystrophy

People with lipodystrophy have either a near total or partial deficiency of *adipose tissue*. This tissue produces a hormone called *leptin*, which helps regulate many important body functions.

Your healthcare team will carefully review all your symptoms and the available treatment options to create the best plan for you. This plan might include:

- A controlled diet to improve your metabolic health
- Physical activity
- Anti-diabetic medication to manage blood sugar levels
- Medication to lower ***triglycerides*** and/or ***cholesterol***

Even with a diagnosis of lipodystrophy, you still have control over many areas of your health. Ask your healthcare team about how you can live a healthy lifestyle, such as a diet and exercise program.

Depending on progression and severity of disease, the response to the treatments listed above may vary. For example, anti-diabetic medication often requires very high doses to treat severe insulin resistance. In some countries, there is a treatment option called ***leptin-replacement therapy***, which mimics the natural function of ***leptin*** in the body. This therapy

works across multiple body systems that rely on ***leptin***, addressing several symptoms at once. This makes it different from other approaches that target each symptom individually.

It's important to know that each country carefully decides which medicines can be used there. As a result, some medicines may be approved or undergoing clinical trials in one country but not in another. Remember, a healthy lifestyle is unique to each person. Work with your healthcare team to find the approach that best supports your individual needs and goals.

Understanding Leptin Replacement Therapy

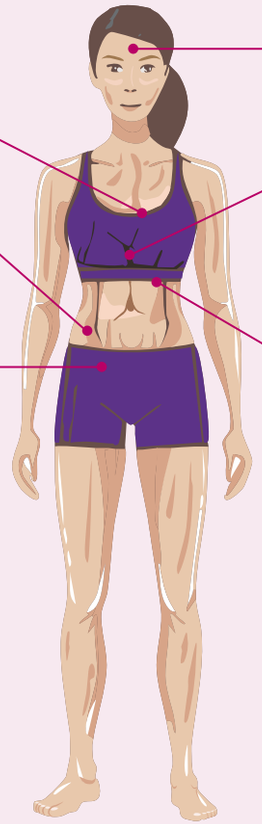
Heart: Improved cardiovascular health and lipid profile

Liver: Supports liver function and reduces ectopic fat

Reproductive: Improved female reproductive function

Immune function: Improved immune function and responsiveness

Quality of life and survival: Improved quality of life and reduced mortality risk



Physical Appearance: Improvement in self-perception

Metabolic effects: Improved metabolic regulation, including better glucose control, lower triglycerides, and reduced hunger

Pancreas: Improvement in pancreatitis

Key Takeaway: Leptin therapy improves whole-body metabolism, preventing fat accumulation in harmful places and reducing disease risks.

Ongoing Management

Understanding Your Healthcare Team

During your journey with lipodystrophy, you may work with various healthcare professionals and specialists who can help manage different aspects of your condition. As lipodystrophy is a progressive condition, it is important to regularly check in with your healthcare team as your comorbidities and symptoms may change over time.

Elena, living with CGL

Here's a guide to who they are and how they can support you. Depending on your age, you may need to see adult or paediatric versions of the specialists listed below:

- **Endocrinologists**: These doctors are the main specialists who manage lipodystrophy. They help with many aspects of the condition, including hormone problems like insulin resistance and complications from low ***leptin*** levels. In most countries, endocrinologists oversee overall care for lipodystrophy.

- **Geneticists**: Since lipodystrophy can have a genetic cause, geneticists can help confirm a diagnosis, provide genetic counseling, and guide families on inherited aspects of the condition.

- **Cardiologists**: They help protect your heart health and manage issues like high cholesterol or blood pressure.

- **Dietitian**: A dietitian can help you make a healthy eating plan that's right for your body. They will guide you on food choices to help manage your lipodystrophy.

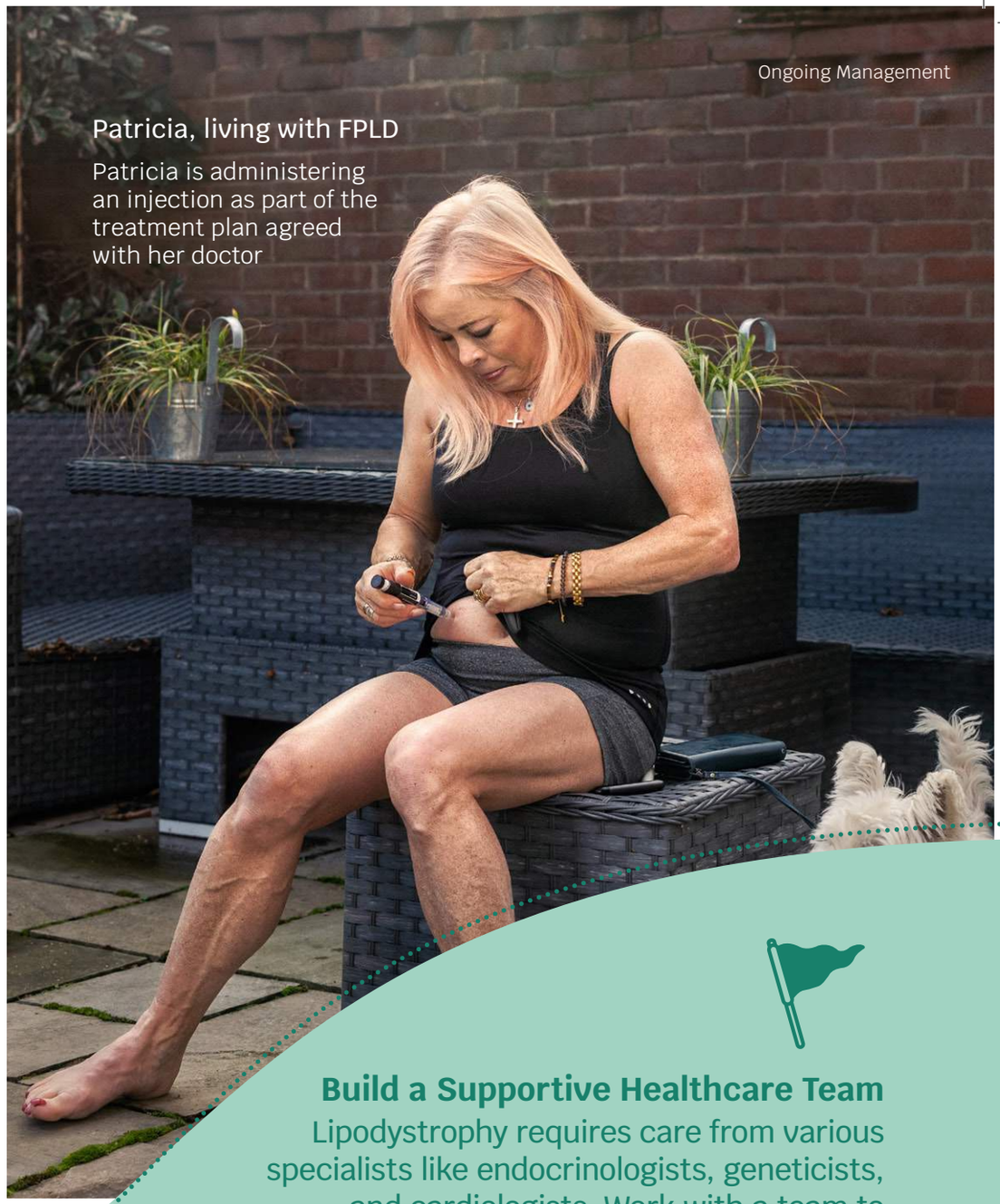
- **Dermatologists**: If you experience skin changes related to lipodystrophy, a dermatologist can offer support to help improve your skin's condition.

- **Diabetologists**: If you have diabetes or are at risk, a diabetologist can help you manage your blood sugar levels and prevent complications.

- **Reproductive Gynaecologists**: A reproductive gynaecologist can help with any reproductive health concerns or questions you may have.

- **Hepatologists**: These doctors look after your liver health, especially if conditions like fatty liver disease occur.
- **Pediatricians**: If lipodystrophy affects children or teenagers, paediatricians provide care tailored to their specific needs.
- **Lipid Experts**: These specialists focus on managing high cholesterol and triglyceride levels, which are common in lipodystrophy.
- **Nutritionists**: A nutritionist is an expert who provides advice and support on food and your diet.
- **Nephrologists**: Nephrologists are kidney specialists who monitor and treat kidney-related complications, which can occur in people living with lipodystrophy.
- **Psychiatrists & Psychologists**: Psychiatrists and psychologists provide mental health support and can help with emotional well-being.

These healthcare professionals often work together as a team, and may refer you to additional specialties based on their findings. You may meet some of these specialists directly during your care, while others may support your treatment behind the scenes. Your doctor will coordinate with them as needed and keep you informed about your treatment plan.



Patricia, living with FPLD

Patricia is administering an injection as part of the treatment plan agreed with her doctor

Build a Supportive Healthcare Team

Lipodystrophy requires care from various specialists like endocrinologists, geneticists, and cardiologists. Work with a team to manage the condition holistically.

What Should I do Next?

- **Document your symptoms**: Create a list of your symptoms, including any new ones that have not been discussed with your doctor yet. Do not worry if they seem insignificant right now. See page ***48*** for more information.

- **Collect photos**: Pictures from the past can help healthcare providers see how fat loss has changed over time. If possible, collecting photos of family members can also be useful in identifying familial forms of the condition.

- **Prepare your questions**: On page ***38***, you will find some commonly asked questions by newly diagnosed patients. You may also have additional questions about your lipodystrophy diagnosis. It is important to note down any questions you want to ask your doctor – this will help you feel prepared for your appointment and make sure you do not forget anything.

- **Anticipate questions**: Think about the types of questions your doctor might ask you regarding your health. For example, they may ask you about your diet, alcohol intake, sleep or any changes in your symptoms. It is important to be honest – your healthcare team is here to help you.

- **Bring a support person**: Consider taking a friend or family member with you for moral support. They can help by asking questions on your behalf or taking notes for you to refer to later.

- **Discuss family history**: Some forms of lipodystrophy, such as Familial Partial Lipodystrophy (FPLD), can be ***inherited***. Speak with your doctor about genetic testing for your family, as early diagnosis can make a big difference. If you already know of family members living with lipodystrophy, consider reaching out to them as their experiences can provide valuable insights.

- **Expand your support network**: Look into connecting with others who share your condition through a patient organization, as this can offer extra support and resources. A list of patient organizations can be found at the end of this booklet on page ***46***.

- **Explore additional resources**: Review other available information that might inspire further topics to discuss with your doctor. Please refer to page ***45*** for additional resources.

Wear the clothes you feel most comfortable in. Don't worry if it shows too much muscle. People are going to comment on your body out of fascination. Say 'Thank You' for asking or noticing. Take the opportunity to educate with kindness

Cynthia, living with FPLD

Be Patient with Yourself

Adjusting to a new diagnosis takes time. Be kind to yourself as you navigate this journey and seek support when needed.

FAQ's

Here are some of the most common questions that newly diagnosed patients ask:

1. **How common is lipodystrophy? Are there other people like me?**
   Lipodystrophy is a rare condition, affecting approximately 3 in every 1 million people globally. While it is not commonly seen, there are others who also live with lipodystrophy. The condition can vary in its forms, and each person's experience is unique. You are not alone – there are communities and support networks where you can connect with others facing similar challenges. For more information on the different types and how common each one is, please see page ***9***.
2. **How will lipodystrophy affect my overall health and daily life?**
   Lipodystrophy can increase the risk of other health conditions, such as diabetes, high triglycerides, and liver disease. It can also cause physical symptoms like extreme hunger, pain and fatigue. People with lipodystrophy may also experience emotional challenges and body image concerns. However, with the right treatment, lifestyle changes, and support, these effects can be managed, helping to improve your overall wellbeing.

Patricia, living with FPLD

Lean on the support groups, they are your equals, they know how you feel

Laura, living with partial lipodystrophy type 1

Gender Distribution:
Lipodystrophy can affect both genders but is generally more common in women than in men.

3. **What tests are used to diagnose lipodystrophy and how can I access them?**
   Lipodystrophy is a clinical diagnosis, and medical history and physical examination are the most important steps in identifying the condition. However, the diagnosis of lipodystrophy, regardless of its type, will involve different types of tests. This may include blood tests (such as HbA1c, blood glucose, insulin resistance and triglycerides) and imaging scans (including DEXA and MRI). In cases where a genetic cause is suspected, specific genetic tests may also be performed. For more information about the tests available and how to access them, please talk to your healthcare team.

4. **Are my current symptoms related to lipodystrophy?**
   It is important to discuss all your symptoms with your healthcare team to understand how they may be related to lipodystrophy. Your healthcare team can also suggest ways to manage these symptoms.

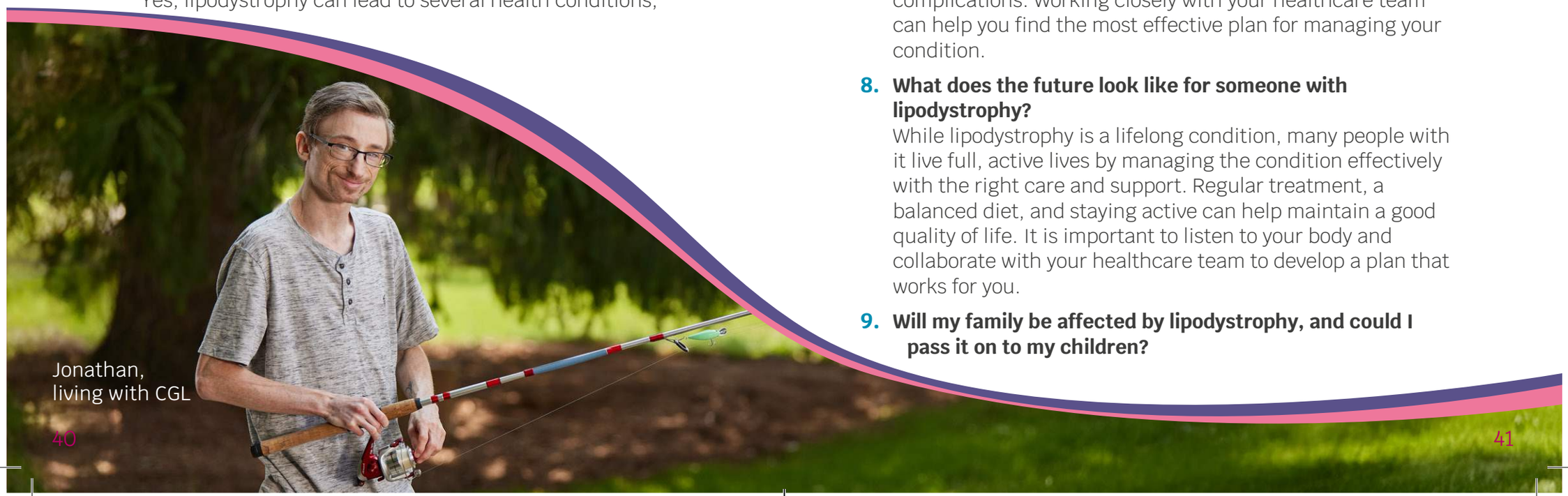
5. **Is lipodystrophy linked to other health problems?**
   Yes, lipodystrophy can lead to several health conditions, including diabetes, heart disease, high triglycerides, liver and kidney disease and complications in female reproductive systems. The lack of normal fat storage can disrupt your body's metabolism, increasing your risk of these issues. Regular visits to your healthcare team are essential to monitor your health and manage these risks effectively.

6. **What should I do if my symptoms change or get worse?**
   If you notice any changes or worsening of your symptoms, it is important to contact your doctor. Your healthcare team can reassess your condition and adjust your treatment plan as needed to better manage any new challenges. Don't hesitate to reach out if something feels different or more concerning than usual.

7. **Can lipodystrophy be cured?**
   Currently, there is no cure for lipodystrophy, but it can be managed with the right treatments and lifestyle changes. Medications and lifestyle adjustments, including diet and exercise, can help control symptoms and reduce the risk of complications. Working closely with your healthcare team can help you find the most effective plan for managing your condition.

8. **What does the future look like for someone with lipodystrophy?**
   While lipodystrophy is a lifelong condition, many people with it live full, active lives by managing the condition effectively with the right care and support. Regular treatment, a balanced diet, and staying active can help maintain a good quality of life. It is important to listen to your body and collaborate with your healthcare team to develop a plan that works for you.

9. **Will my family be affected by lipodystrophy, and could I pass it on to my children?**



Jonathan,
living with CGL

There's always someone with knowledge about anything. Don't be afraid to reach out and ask questions, we were all there once!

Sharon, living with FPLD

There's a community built ready to welcome you, you're not alone

Miranda living with FPLD type 2

Elena, living with CGL

Lipodystrophy can be inherited, so there is a chance that other family members may be affected, especially if it runs in your family. If you have a family history of lipodystrophy, similar symptoms or if you suffer from a familial or congenital subtype of lipodystrophy, it is important to discuss genetic testing with your doctor. They can help you understand the likelihood of passing lipodystrophy on to your children, based on your family's genetic background and the type of lipodystrophy involved.

10. **How can I explain lipodystrophy to my family and friends?**
It can be helpful to explain lipodystrophy as a rare condition that causes near total or partial loss of fat. Let your family and friends know that, while there is no cure, lipodystrophy can be managed. Encouraging them to ask questions and learn more will help them understand your experience and be better equipped to support you emotionally and practically. For example, sharing that managing the condition often involves specific dietary needs and meal planning can help reduce misunderstanding.

11. **Are there any support groups or communities for people with lipodystrophy?**
Yes, there are many support groups and online communities where people with lipodystrophy share experiences, advice, and emotional support. Connecting with others who understand what you are going through can be incredibly valuable. For more information, please refer to page **46**.

Jonathan, living with CGL

Here are some questions you may want to ask your doctor or healthcare professional:

- What's the best way for me to manage lipodystrophy every day?
- Which doctors or specialists should I see for lipodystrophy?
- Are there lifestyle changes that can help with my symptoms?
- What treatments are available for lipodystrophy?
- How can I manage symptoms related to lipodystrophy?
- Can you suggest any resources for dealing with body image issues or treatments for it?
- How can I cope with the emotional and psychological effects of lipodystrophy?
- What can I do to manage fatigue from lipodystrophy?
- Are there any new treatments or clinical trials I should know about?
- What are the key aspects of managing the metabolic issues associated with lipodystrophy?
- Are there specific steps I can take to improve my general health?
- Based on my type of lipodystrophy, should any of my family members be tested?

Ask For Specialist Advice

If your clinician is unable to provide answers or guidance about lipodystrophy, don't hesitate to ask for a referral to a specialist who has experience with the condition. Building a healthcare team knowledgeable about lipodystrophy can be challenging, but it's important to seek the right support to manage your care effectively

Sources of Support

As you navigate your journey with lipodystrophy, it is important to remember that there is plenty of support available – both for people impacted by lipodystrophy and their loved ones. While lipodystrophy presents unique challenges, you are not alone in facing them.

The following organizations provide information, support and advocacy to help you understand and manage your condition. If you can not find a suitable resource in this booklet, you can also speak to your healthcare team about other options that may be available.

- **Lipodystrophy UK** – https://lipodystrophyuk.org/
- **Lipodystrophy United** – https://lipodystrophyunited.org/
- **AFLIP** – https://aflip.org/
- **AELIP** – http://www.aelip.org/
- **AiLIP** – https://ailip.it/
- **Lipodystrophy Canada** – https://lipodystrophycanada.org/
- **Lipodystrophy Israel** – https://www.lipodystrophyisrael.org/
- **Lipodystrophy Germany** – https://netlip.org/
- **Acopel** – https://www.facebook.com/acopelcomunica

Resources:

Living with lipodystrophy can come with unique challenges, and finding the right information and support can make a big difference. The following resources are designed to help you and your loved ones better understand the condition, manage symptoms, and connect with others in the community.

	Lipodystrophy UK Hope Awareness Support UK	***Watch 6 short films from individuals who are navigating life with lipodystrophy and listen to their personal stories.***
	Lipodystrophy United US	***Access a storytelling workbook to help guide you in writing your unique lipodystrophy story.***
	ECLIP EU	***Access a catalogue of support groups across EU and SWANA countries.***
	Lipodystrophy United - Israel Israel	***Access the registration page for the Lipodystrophy patient group based in Israel***
	AELIP Spain	***Click here to access the resources offered by the AELIP association. Their website appears in Spanish, Portuguese, English, French, Italian and German; and their services are provided to any person or family living with lipodystrophy anywhere in the world.***

Notes

This part of the booklet can be used to note down any questions you may want to discuss with your healthcare team. Your notes from this booklet and your appointments can help you feel more prepared and ensure that you cover all the points you wish to discuss.

Notes:

Tracking Your Symptoms

Keeping track of your symptoms using the table below can help you and your healthcare team better understand your condition and manage it effectively.

Date	Symptoms	Severity	Duration	Potential Triggers	Notes

Glossary

This glossary is designed to help you navigate through this new vocabulary associated with your diagnosis. It contains definitions of key terms and phrases that you are likely to encounter on your journey.

- ***Lipodystrophy*** – A rare disease characterised by an abnormal distribution of fat throughout a person's body.
- ***Generalized lipodystrophy (GL)*** – In generalized lipodystrophy, there is a total or almost total absence of adipose tissue from the entire body.
- ***Partial lipodystrophy (PL)*** – In PL, specific regions of the body are affected by a loss of fat.
- ***Congenital Generalised Lipodystrophy (CGL)*** – CGL is a rare genetic disorder characterized by the near total loss of body fat.
- ***Familial Partial Lipodystrophy (FPLD)*** – A rare genetic disorder characterized by a partial loss of fat predominantly affecting the limbs.
- ***Acquired Generalised Lipodystrophy (AGL)*** – A rare genetic disorder characterized by a progressive loss of fat leading to near-complete lack of adipose tissue.
- ***Acquired Partial Lipodystrophy (APL)*** – A rare genetic disorder characterized by the gradual loss of tissue from the head downwards.
- ***Acanthosis Nigricans*** – Thick, dry and dark patches found on the skin. They can frequently be found in skin folds including on armpits, neck or groin and can be associated with insulin resistance.
- ***Adipose tissue*** – A technical term for body fat. This type of tissue can be found throughout the body and under the skin.
- ***Cardiomyopathy*** – A general term for disorders which affect heart muscle.
- ***Cholesterol*** – Waxy, fat-like substance produced by the liver and found in all cells of the body, essential for various bodily functions including hormone production and cell membrane structure.

- ***Comorbidities*** – Separate health conditions which are present in a person at the same time. They can be related to a specific health condition and interact with each other or can be unrelated.
- ***Echocardiogram (Echo)*** – The use of ultrasound to look at the valves and chambers in the heart and surrounding blood vessels.
- ***Ectopic fat*** – Fat which accumulates in tissues or organs where fat is not normally stored, this can include the liver, muscles, heart and pancreas.
- ***Electrocardiogram (ECG)*** - A test used to visualize the activity of the heart including heart rate and rhythm.
- ***Glucose*** – A simple sugar which is an important source of energy in the body and a major component of many carbohydrates.
- ***Hepatic Steatosis*** - A technical term for fatty liver disease. This occurs when there is excess fat in the liver. If diagnosed and managed at an early stage it is possible to slow the progression and prevent serious liver damage.
- ***Hyperphagia*** – A feeling of intense, insatiable hunger which often results in overeating due to a lack of feeling full.
- ***Hypertriglyceridemia*** – When triglyceride levels in the blood are higher than normal levels.
- ***Leptin*** – A hormone produced by fat cells. It plays an important role in appetite control and often produced at a lower level in lipodystrophy patients.
- ***Leptin replacement therapy*** – A medical treatment available in certain countries to treat complications of leptin deficiency in lipodystrophy. Leptin replacement therapy treats complications of leptin deficiency in people with lipodystrophy.
- ***Lifestyle changes*** – Non-medical treatments including diet changes and exercise that can improve disease management and improve overall wellbeing.
- ***Lipoatrophy*** – Loss/lack of adipose tissue.
- ***Lipoatrophic diabetes*** - A condition characterized by severe insulin resistance and metabolic disturbances caused by a lack of fat in the body.
- ***Muscular hypertrophy*** – An increase in muscle mass and volume.
- ***Triglyceride*** – The most common type of fat in the body. They serve as a primary source of energy, circulate in the bloodstream, and are stored in fat cells.

Your Guide to Living with
Lipodystrophy

References for further information:

Fourman LT, Lima JG, Simha V, et al. A rapid action plan to improve diagnosis and management of lipodystrophy syndromes. Front Endocrinol (Lausanne). 2024;15:1383318.

Santos V, Almeida G, Costa C, et al. Lipodystrophy disease and quality of life questionnaires: A longitudinal assessment of hunger and well-being throughout the study period. Orphanet J Rare Dis. 2023;18(1):144.

Mosbah H, Vatier C, Andriss B, et al. Patients' perspective on the medical pathway from first symptoms to diagnosis in genetic lipodystrophy. Eur J Endocrinol. 2024;190(1):23-33.

Araújo-Vilar D, Santini F. Diagnosis and treatment of lipodystrophy: a step-by-step approach. J Endocrinol Invest. 2018;42(1):61-73.