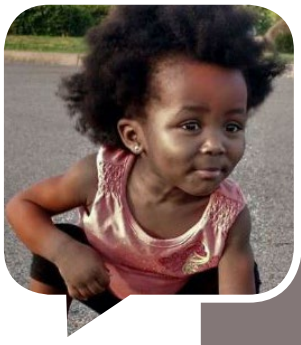

The Voices of LD

The Experiences of Individuals and Families living with Lipodystrophy.

Lipodystrophy United

Who We Are

Lipodystrophy United (LU) is an organization of committed individuals living strong with Lipodystrophy (LD). Our mission is to provide an interactive community, facilitating support and education for anyone affected by this rare disease. We serve as a resource and to increase awareness in the general population as well as the medical and insurance communities. We advocate and act as a catalyst for new patient diagnosis by assisting healthcare professionals in the understanding of Lipodystrophy trends, physical attributes and clinical symptoms in order to aid in the advancement of knowledge, treatment and future research.

When individuals with the same rare condition meet, either in person or on social media, something indescribably powerful happens. For individuals with lipodystrophy it means looking at yourself in the mirror. Seeing others that look like you, and hearing stories that sound like yours ends a lifetime of isolation. LU was founded to help others gain access to information and resources. We never anticipated hearing things like, "No exaggeration, LU saved my life!" and, "I once walked alone in this battle and felt like the only one, until I discovered LU. Now I walk in pride with my rare community, which has replenished my heart and soul. I now know I am never alone in the battle that is lipodystrophy."

We now know that receiving a diagnosis and gaining access to resources is only part of the picture. Providing a community that leads to a feeling of family and understanding is important to treating the whole individual.

Our Unmet Medical Needs

People with lipodystrophy experience an uncontrolled loss of fat tissue, especially fat under the skin. This causes a drop in an important hormone called Leptin. Without enough fat tissue or Leptin, the body's system for regulating energy use falls out of balance. This system is called the metabolic system. Each person's metabolic system requires a specific amount of fat and hormones to work properly. This serious imbalance causes fat to accumulate where it in the blood or organs—which can lead to life-threatening complications.

- Organ dysfunction and failure
- Severe Metabolic complications (high triglycerides, insulin resistance, diabetic)
- Hyperphasia
- Fatigue
- Pain
- Sleep and mood disorders
- PCOS
- Compromised Immunity

Causes

Congenital/Inherited (CGL, FPL):
Parents are carriers of alterations in one of their genes. In CGL, the affected person inherits mutations in their genes from both parents. In FPL, the affected person has a 50% chance of inheriting the mutation in their genes from one parent. The diagnosis of CGL is usually made at birth or soon after. Assuming that only 1 in 4 patients are reported in the published literature, the estimated worldwide prevalence is about 1 in 10 million.

Acquired (AGL, APL):

- Panniculitis variety (type 1):The patient presents with painful and inflamed subcutaneous nodules or maculopapular lesions. Upon healing, depressed scars remain but the overlying skin is normal. New nodules appear and there is progression of subcutaneous fat loss.
- Autoimmune disease variety (type 2): In this variety, the patients have past or present evidence of autoimmune diseases.
- Idiopathic variety (type 3): This is the most common variety. The cause of this variety is not known.

Symptoms:
The extent of fat loss may determine the severity of metabolic complications. Some patients may have only cosmetic problems while others may also have severe metabolic complications.

Physical changes
The exact locations of fat tissue loss varies from person to person. For example, some people with lipodystrophy may have areas on their body that look very thin (face and arms), while other areas might appear large (hips or buttocks). Other people with lipodystrophy might have very little fat tissue on the lower areas of the body (legs and buttocks) and excess fat tissue on the upper areas of the body (abdomen, chin, and neck). Still others might have very little visible fat tissue anywhere on their bodies and may appear extremely muscular.

Internal changes
Because people with lipodystrophy are missing or have very low leptin, fat can be found in unusual places like the bloodstream, heart, kidneys, liver, and pancreas. Since fat is not meant to be in these places, it can lead to serious problems, such as insulin resistance, diabetes, high cholesterol, fatty liver disease, pancreatitis and heart disease.

PATH TO DIAGNOSIS

The path to diagnosis is regularly difficult no matter what type of lipodystrophy one has. Generalized lipodystrophy is typically diagnosed between infancy and adolescence. Parents may struggle to convince pediatricians that something is wrong and multiple specialists may be called upon prior to correct diagnosis. Individuals living with partial lipodystrophy do not typically display any symptoms until puberty and the progression from that point varies as does the health complications associated with the disease. Partial lipodystrophy it is commonly misdiagnosed as Cushing's Syndrome or mistaken as problems only associated with Polycystic Ovarian Syndrome (PCOS) or Metabolic Syndrome while the rest of the health problems are ignored. Among all types, patient and caregiver reports indicate significant challenges in being taken seriously.

Our Stories

Andra's story:

I am a mother, a patient and the co-founder and President of Lipodystrophy United. My story is only one of hundreds living with Lipodystrophy (LD), each varying by subtype or severity. My specific type, FPL, is a genetic condition with onset at puberty. Although I was not diagnosed until I was 38, I'd always known my body was different and even assumed that some of my health problems were associated with my lack of visible fat. One day, after a trail of questions not related to my condition, I walked into the office of a knowledgeable, old-school endocrinologist and he simple said, "I think you have something called lipodystrophy." This explained my years of high cholesterol, failed normative tests, high blood pressure, diabetes, near-death pregnancies, exhaustion and insatiable hunger. You mean not everyone is this hungry? I thought I was just a wimp. I suffered in silence because I thought everyone felt like this. Yes, it seemed strange that I could eat an ENTIRE pizza and promptly start thinking about my next meal while my body was thin and getting thinner (Except that darn face that keeps getting rounder and rounder). Without the subcutaneous fat to cover the muscle, I look like I spend my days at the gym. No, I am not a body builder, a swimmer, a runner, and no, I don't take steroids, really, doctor, I don't. I am watched and stared at everywhere I go. Often people will say, "Look at that woman, gross!", as though I cannot hear them from 5 feet away. Others think it's cool, but either way, I draw attention. I am told "You are sooo lucky!" on a regular basis because I don't have cellulite....that adipose tissue that creates the hormone, leptin, which helps your body regulate energy, metabolism, satiation, and so much more. I'll gladly trade some fat for my daily 20+ pills and restricted diet, for the swollen legs, the apnea, the exhaustion, the headaches, the nerve damage, chronic angina and that stroke or heart attack that is hiding behind every corner for some of that cellulite.

Lipodystrophy is progressive. I lived many years mostly healthy and still feel lucky to live an active and productive life. But I'm increasingly tired and "sick". I would say the fact that we look healthy hinders the ability for people to take our illness seriously. I find it difficult to express that to friends and family in such a way that I am taken seriously yet don't sound like a complainer.

I lived many years thinking something was wrong with me by not having a diagnosis. The label "lipodystrophy" has allowed me to seek information, demand attention and learn to advocate for myself.

There is still limited information, no approved treatment, and certainly no cure in sight. I now work with some of the most inspiring patients, doctors and researchers around the world. I am able to answer questions and provide information with the hope that I can help make the path for every young person with LD a bit smoother. All in all, I think I'm pretty lucky too."

~ Andra Stratton, President & Co-Founder of Lipodystrophy United

Cynthia's Story:

This all started for me when I was 16, 5'5 tall and 110 pounds. At this time I was told that my triglycerides were over 550. I didn't know what triglycerides were at the time and nothing was done. When I was 23 I woke up at 5am in my apartment with the worst stomach & back pain I had ever felt in my life. I drove to my mother's house and had her take me to the hospital. I was admitted to the ICU for 2 weeks with triglycerides over 14,000. I had a fever they couldn't control, pseudo cyst and an NG tube to drain all the fluid. My small home town hospital couldn't handle how sick I was or control my fever. They transported me via ambulance to The University of Maryland Hospital in Baltimore. I spent another 2 weeks in that hospital where I was told I was the youngest patient they had ever seen with acute pancreatitis. I was told I couldn't work for 3-4 months. It was a painful recovery, but I had almost died from this, so I was thankful to be alive. I was started on medicines to help lower triglycerides.

A year later I started with the same pain and again was admitted to the hospital. This time it was a combo of a gallstone stuck in the duct to my pancreas. I had emergency surgery to remove my gallbladder. I was again in the hospital for 2 weeks.

I then started developing other health issues; diabetes, darkened skin in areas on my body, weight gain only in my stomach, kidney disease, nonalcoholic fatty liver disease, anxiety and depression, PCOS, infertility, herniated disks in my neck, joint pain, insulin resistance, severe pain in my shoulders due to calcium deposits, fatigue, hair loss, psoriasis, uncontrolled triglycerides, etc. I could not lose weight even with a healthy diet and exercise. My doctors didn't understand what was going on and I was treated individually for each health problem. I was tested for Cushing's and lupus and finally one of my doctors suggested I have bariatric surgery. All my doctors blamed all my conditions on my weight. No one believed that I ate healthy and exercised because of my body shape. It was really frustrating!

In 2008 I had a gastric sleeve and only lost 18 pounds and only during the liquid phase of the diet. I was devastated, 18 pounds after surgery following the post op diet and exercise! The surgeon couldn't explain it and gave me diet pills. I still to this day never lost any more than the 18 pounds. I kept it off, so I guess that's something!

I had my next bouts of pancreatitis in Oct of 2014 and again in June & July of 2015. It was at this time my kidney Doctor contacted the NIH and got me into the clinical trial.
I started in May of 2016 at the NIH and was diagnosed with Partial Lipodystrophy. It all finally made sense! Every single one of my medical conditions were from the Lipodystrophy, even my body shape!

I started on the Isis trial injection once a week. In one month my triglycerides went from over 2,000 to under 200! Today my triglycerides are 70! My triglycerides are lower now at 225 pounds then when I was 110 pounds. That's because this disease has nothing to do with weight. It has to do with weight distribution and for me that looks like all my weight in my middle. I have no fat on my legs, buttocks, feet, forearm and hands.

My trial ends in Nov of 2017, but I will still need this medicine! For me this medicine has done what no other medicine has been able to do and that is to lower my triglycerides. This medicine has to be approved for people like me! We need more research into this horrible disease and we need your help!

Thank you,
Cynthia Wooters

Julian's story:

Julian was born in July of 2015. He was tiny, weighing in at 6 pounds, with no subcutaneous fat and gorgeous little muscles covering his body. He had a collapsed lung and fluid in his lungs, so spent a few days in the NICU before coming home with a clean bill of health. We had questioned the lack of subcutaneous fat, but were told he was a few weeks early, so he would put on weight in a few days. In other words, it was nothing to worry about. At our first weight-check at our pediatrician's office, she kept telling us how shocked she was that he had zero subcutaneous fat. She had never seen that before. I felt so much pressure, as I was breastfeeding, to try and make my milk more fatty in hopes to put some fat on him. But 2 weeks later, he still had no fat. We learned that one of his protein levels was elevated, from his newborn screening, so we saw a geneticist about it. She had no explanation for the lack of fat, and the high protein, as he wasn't showing any symptoms related to that. He was a happy baby, calm and so alert.

With no answers, we went home with directions to repeat labs in 2 weeks. We didn't make it that long. At 3 weeks old, we found a red spot in his diaper: he was dehydrated. At our pediatrician's office, they checked his urine for blood, found none, but did find that his sugar levels were 500. When checking his blood sugars, they were 318. We went to the emergency room, but were sent home later that night because he had regulated his own sugars. We had so many questions, and still no answers. Was he diabetic? Why did the sugars regulate on their own? Still, why does he have no fat and so many muscles? The following week, we had his sugar levels checked again, and they were elevated at over 300, so we were admitted to the hospital. Many labs were done to try and find what was going on in his body. We had a wonderful endocrinologist, who had sat us down and told us she thought Julian had one of 3 conditions: Diabetes, Lipodystrophy, and some other one that I can't remember now. I had never heard of lipodystrophy, so didn't think much about it at first. But after testing his triglycerides, which were 3,428, and insulin which was 56, (amongst other various blood tests,) and an abdominal ultrasound with results that showed his liver was enlarged, our endocrinologist and her wonderful team, came to us with the diagnosis of Congenital Generalized Lipodystrophy. Our team had reached out to Dr. Garg at the University of Texas Southwest, to try and confirm the diagnosis via pictures and lab results. He agreed.

Having never have heard about this before, we were handed a 2-page general explanation. In short, our brand-new baby couldn't process fat like the typical body did. Instead of storing it under his skin, it was stored in his blood and liver. In short, my breast milk was literally killing him. I was devastated. We were sent home from the hospital with a prescription formula, made from MCT oil.

The next few months went by as a blur. We started seeing multiple specialists to monitor his kidneys, liver, and endocrine system. What we quickly learned was that being 1 in 10million people to have this disease was not easy. Our doctors had never seen anyone like Julian, and quickly learned they had a lot of research to do. I found myself doing a lot of research online (mainly at lipodystrophyunited.org), talking to Dr. Garg and a nurse at the NIH, and joining the various Facebook groups so that I could learn as much about this rare disorder as a I could. I wanted to be aware so that I could advocate for Julian at all of his appointments, and have the knowledge that we and his doctors needed, when they didn't have it originally.

At 3 months old, Julian was diagnosed with hypertrophic cardiomyopathy. When he was initially diagnosed, our cardiologist looked at us and told us "Your son's heart walls are very thick. He has something called hypertrophic cardiomyopathy. He can die at any time from cardiac arrest, and there's nothing we can do about it." We were devastated. But I refused to accept it. I called Boston Children's Hospital and got an appointment with them for the following week. It was there that we learned that He did have cardiomyopathy, but by looking at his heart, it didn't look like one that would experience sudden cardiac arrest. We learned that having developed cardiomyopathy so young was even more rare than the original 1 in 10 million diagnosis of CGL. Julian is the only person in the U.S. to have developed both conditions at such a young age (as far as we are currently aware.)

It has been a difficult road so far, with many ups and downs. We have finally formed a team of 11 specialists, all of whom we love. But it took time and negative experiences to be happy with our current team. Julian has a hepatologist, nephrologist, 2 dieticians, 3 cardiologists, 2 metabolism specialists, a genetetecist, an endocrinologist. He has routine bloodwork done every 3 months to monitor his essential fatty acid levels, lipid levels, liver and kidney function, and other various things.

Julian is now a very busy and energetic 17 month old. He is very tall, with advanced muscles covering every inch of his body. His belly is distended because of his enlarged liver, kidneys and pancreas (due to high insulin levels and fat in his liver.) His speech is delayed, which may be related to the CGL, but he uses sign language to communicate with us. His arms and legs are "skinny." They have a lot of beautiful muscles, but are small in comparison to his belly. It is very difficult to find clothes that fit him properly. He wears 3T-4T shirts to cover his belly. He needs 6-12 month pants to stay up on his tiny hiney, but his legs are so long, we have to get 2T pants and roll them over to try and get them to stay up. We are constantly puling his pants up throughout the day because they fall down so often!

We maintain healthy TG levels, sugar levels, etc. by way of diet. He is on an extremely strict low fat, no sugar, high protein diet. His carbohydrates consist only of insulin and sugar friendly foods. His fat intake is monitored closely, with most of his intake consisting of medium chain triglycerides.

He is a very happy boy, unless he is hungry. Because of his low Leptin levels, he feels hungry often. We spend each day keeping Julian busy so that he doesn't think of food, because if he gets bored, all he wants to do is eat. There are times when he tells us he is hungry, and we can distract him. There are other times when no distraction in the world works to get his mind off of food. This can happen at any time, sometimes even after he just ate. These "food frenzies" (as I call them) can be awful, with a lot of screaming, crying and "tantrumming." Nothing stops them, but food. He still wakes up at least twice at night to eat as well.

We have learned our future will not be what we have always planned when thinking about having children.

Julian won't be able to play in most sports. He will never know what "fast food" tastes like, parties and functions will not be noted for their yummy and different foods that are usually the highlight of the events, he will have more bloodwork done and see more doctors than most families combined do in a lifetime. But our love, faith and hope will get us through each day with smiles.

It is so easy to feel alone in this journey, with no one that you know personally to relate to anything you are going through. My family is so thankful to this lipodystrophy family who has welcomed us with open arms. Lipodystriophy United is not only full of life-saving information, but has wonderful people behind the scenes who welcome all questions and are there to lend as an open ear in times of need.

Alani's Story:

Our Lipodystrophy: "I watch my daughter's belly extend, her legs so frail and thin. Her arms so tiny and her hands barely able to hold anything for any length of time. Her face no longer the cute chubby cheeks and pudgy nose. She tries so hard to sit and stand but succumbs to the aches and pain of her joints and spends most of her day laying in my lap. Her carseat is riddled with cushions just to make it to the doctor. Alani has 24hr wake periods and 12hr binges of sleep. Why? We don't know...Her episodes reacting to extreme hunger can occur anywhere at anytime and are extreme mental breakdowns for all who have to witness and comfort her through it. Leptin helps her (A LOT) and, without it, I don't believe I'd be able to make it through her rage that can last for hours and include destruction of anything she puts her hands on all in the name of food. How do you tell a baby that she just ate? She has ripped out her earrings from her ears and a second occurrence forced me to remove them indefinitely. She has pulled out her hair and scratched her scalp until a wound required urgent attention.

She is so, so cold all the time lacking total body fat that at our temps here in Oklahoma peaking over 100's, we still have to run space heaters in the rooms for her.

At nearly 3 years old and only 24lbs she still wears clothing fit for a 6 month old. She is non-verbal and wears diapers fit for a newborn. She can wear doll clothing. Holding her is an art, as I have to be careful not to squeeze her too tightly as I am gripping only skin and bones.

She has Congenital Generalized Lipodystrophy (CGL) with muscular hypertrophy, hypertriglyceridemia, hepatic steatosis, hepatomegaly, etc. I have to (with assistance) pin her down every single day to administer her medication as she fights and screams unlike anything you could imagine or ever heard.

This disease is not to be taken lightly. It is progressive and aggressive and death has no age limit. It has no face and is no age of onset. One minute you're ok and the next you're not. This is just our story, our lives, everyday, our love to you. Please fight and know you can always depend on me to fight too.

- Rach and Alani

Our Hope

There is no cure for lipodystrophy and the effects of treatment vary from person to person. There is only one FDA approved treatment for Generalized Lipodystrophy (Myalept) and there are no approved treatments for Partial Lipodystrophy. There is no cure for lipodystrophy and the effects of treatment vary from person to person. There is only one FDA approved treatment for Generalized Lipodystrophy (Myalept) and there are no approved treatments for Partial Lipodystrophy. Other tools that vary in efficacy include traditional therapies for metabolic syndrome such oral and injectable diabetes treatment, hormone replacement therapy (approved for generalized only), statin therapy, diet and exercise and cosmetic options. Typically, traditional therapies are not effective and individuals need preventative treatment for vital organ protection close monitoring. New research and treatment options are extremely necessary. New research and treatment options are extremely necessary.

Website:
lipodystrophyunited.org

Facebook:
facebook.com/LipodystrophyUnited

Twitter:
@LipodystrophyLU


Lipodystrophy
United