FOUNDATION
of the National Lipid Association

What is Lipodystrophy?

Lipodystrophies are a group of rare diseases involving the unexpected loss of body fat from various places of the body. They are frequently associated with conditions like high blood sugar, diabetes, high triglycerides in the blood, and an abnormal amount of fat in the liver. The fat loss can vary from very small areas on one part of the body to almost no fat in the entire body. Lipodystrophy can be inherited (genetic) or caused by other illnesses or drugs (acquired).

1. Genetic lipodystrophies: These are caused by gene changes and can show up soon after birth or later in life depending on type of gene change. Congenital generalized lipodystrophy and familial partial lipodystrophy are the two main types of inherited lipodystrophy; the other types are extremely rare.
2. Acquired lipodystrophies: These usually occur during childhood, adolescence or adulthood and can be caused by medications such as medications to treat HIV, from autoimmune disorders, or due to unknown reasons (this is called "idiopathic"). Common types of acquired lipodystrophy include acquired generalized lipodystrophy (Lawrence syndrome) and acquired partial lipodystrophy (Barraquer-Simons syndrome).

Criteria for Diagnosis of Lipodystrophy:

Diagnosis of lipodystrophy is based on a detailed medical history and a thorough clinical exam to evaluate body fat loss. Tools are used to help measure the loss of body fat. They include skin fold measurements, dual-energy X-ray absorptiometry (DEXA), and whole-body MRI. A variety of other tests including genetic testing can be helpful for the patient and family members who may be at risk for genetic lipodystrophies. In patients with acquired lipodystrophy, there are special lab tests that can be ordered, including serum complement levels and autoantibodies. These tests help can help with the diagnosis of four major types of lipodystrophies:

- Congenital generalized lipodystrophy
- Acquired generalized lipodystrophy
- Familial partial lipodystrophy
- Acquired partial lipodystrophy

What a Diagnosis of Lipodystrophy Means for My Child or Adolescent?

Patients with suspected lipodystrophy typically need specialized testing, and they should be referred to a lipid specialist. Some patients with lipodystrophies can develop health problems related to lipodystrophy. Some examples include diabetes requiring very high insulin doses, very high levels of triglycerides (extreme hypertriglyceridemia), abnormal fat storage in the liver, heart problems, and potentially life-threatening inflammation of the pancreas (acute pancreatitis). Once the diagnosis of lipodystrophy is made, clinicians should find out if the lipodystrophy is generalized, partial, or localized. In generalized forms, total or near-total loss of fat underneath the skin can be observed over the entire body. In partial forms, fat loss affects large areas, particularly the arms, and legs, but fatty tissue may build up in areas such as the abdomen, face, and neck. Localized forms of lipodystrophy are limited to small body areas.

A child or adult with lipodystrophy lacks fat under the skin either in some places or all over. People with lipodystrophy can have very muscular appearing arms and legs even if they are not strong or athletic. Sometimes there is fat in unusual places such as the neck. These features can lead to embarrassment and body shaming as well as to health problems.

Lifestyle Recommendations for Children with Lipodystrophy:

Current treatments focus on preventing the associated health problems from lipodystrophy. There is no treatment to reverse the loss of body fat. Most patients need a balanced diet with 50–60% of calories from carbohydrate, 20– 30% from fat, and about 20% from protein; however, patients with elevated triglycerides will need an extremely low-fat diet with < 15% of calories from dietary fat, an example of options for this diet are below.

Breakfast Ideas

- Nonfat fruit and yogurt parfait
  - 1 cup nonfat Greek yogurt (plain or vanilla with minimal added sugar or sweetened with non-nutritive sweetener)
  - ½ cup berries
  - ½ cup original Cheerios (or can use Multigrain cheerios)
- Scrambled egg (1 whole egg + 2 egg whites) with 1 slice whole wheat toast and 8oz glass nonfat milk
- Breakfast sandwich (egg whites + reduced fat slice of cheese + whole wheat English muffin)
- 1 cup original Cheerios + ¼- ½ cup blueberries + nonfat milk or unsweetened milk alternative
- Smoothie made with nonfat plain Greek yogurt, 1 cup fruit of choice, 1 tsp chia seeds or ground flaxseed

Lunch Ideas

- Sandwich (whole wheat bread, uncured deli turkey meat, reduced fat or fat-free cheese, lettuce/tomato), 1 small apple, carrot sticks with nonfat Ranch dressing
- Salad with grilled chicken, tomatoes, cucumbers, carrots, baked croutons, and reduced-fat salad dressing.
- ½ whole wheat pita filled with grilled chicken, lettuce, cucumbers, and 1-2 Tbsp hummus: pair with baked chips

Snack Ideas

- 3 cups air-popped popcorn
- 1 cup fruit
- ½ cup fruit with 1 reduced-fat cheese stick

Dinner Ideas

- Homemade air-fried fish sticks (made with white fish), roasted broccoli and cauliflower, corn.
- Turkey tacos (ground turkey for protein, reduced fat cheese, lettuce, tomato, salsa) with 1-2 whole wheat tortillas.
- Spaghetti made with ground turkey breast, marinara sauce, and chickpea pasta or whole wheat pasta. Serve with a side salad.
- Grilled or baked chicken with roasted sweet potato and carrots

Physical activity and exercise can help; however, strenuous activity should be avoided in patients who have heart problems.

Treatment Options for Children with Lipodystrophy:

Metreleptin therapy with a diet described above should be considered for generalized lipodystrophy. Metformin and insulin therapy are usually needed for patients with diabetes. Triglyceride-lowering therapies like fibrates and fish oil are needed for patients with hypertriglyceridemia.

Metreleptin: Metreleptin is an injected medication that acts like a naturally occurring substance called leptin. Leptin is a protein that helps to regulate fat stores in the body and also how the brain perceives satiety the feeling of being full after eating). It is used by patients with lipodystrophy who may not be producing fat tissue appropriately. This medication should be diluted in sterile water and injected into a fatty area of the skin such as the abdomen once daily around the same time each day. The specific dose of the medication depends on the weight of the child and should be decided by the Healthcare Team. It can be taken with or without eating. There can be risks with this medication, including changes in blood sugar and a risk of some cancers; however, it can still play a crucial role in patients' management of this condition in collaboration with the child's Healthcare Team.

Fish Oils: Fish oils are medications that are widely available over the counter, meaning without a prescription, or with a prescription from a member of your Healthcare Team. Fish oils are contain two omega-3 fatty acids, known as EPA and DHA, that help to lower triglycerides, Fish oils that are available without a prescription contain differing amounts of EPA and DHA and may not work well. Prescription omega-3 fish oil should be used for treating hypertriglyceridemia in children. Prescription fish oils are gel-like capsules that contain liquid omega-3 fish oil and can be given once or twice daily, depending on the prescribed dose. They should be taken with a meal and should not be broken or crushed before swallowing. These medications are usually well tolerated but can lead to some upset stomach and fishy-smelling burping. Very rare side effects include variable heart rates or increased risk of bleeding. Fish oil medications should not be used in patients with a seafood allergy, and a child's Healthcare Team should be consulted if there is a concern for increased bleeding, such as dark stools or easy bruising.

Fibrates: Fibrates are a class of medication that reduces triglycerides by reducing the creation of triglycerides and the breakdown triglycerides in the body. Examples of these medications include fenofibrate or gemfibrozil. Fenofibrate is the most used medication in this class and can be given at variable doses, mostly once daily, with or without food, in a capsule or tablet formulation. These medications may cause a risk of muscle aches when used with statin medications. Monitor for these muscle aches when the cause is not known and discuss with the Healthcare Team if this occurs. Patients with a history of gallbladder issues or kidney disease may need to avoid these medications; however, for individuals without these conditions, they are typically well tolerated.

For all of these medications, it is important to talk to your Healthcare Team prior to starting any new medications or supplements to ensure that they are safe to take with prescribed medications. Following the start of new medications, dose adjustments, or discontinuation of certain cholesterol-lowering medications, your Healthcare Team will request repeat laboratory tests to check the impact of the medication on cholesterol levels, which can serve as a marker for risk of heart disease. Additional monitoring tests, such as liver tests, may be needed on a less frequent basis. As always, if you have any concerns with medications, please discuss them with members of your Healthcare Team.

Finding a Pediatric Lipid Specialist in Your Area:

Pediatric lipidology, or the treatment of high cholesterol in children, is a small but growing field in medicine. Some pediatric lipid specialists are pediatric cardiologists, some are pediatric endocrinologists, some are pediatricians, and some are adult lipidologists with an interest in caring for children with high cholesterol.

It can be challenging to find a pediatrician that specializes in lipodystrophy. Your child's primary care provider may know the pediatric lipid specialists in your area. On the website for your health care system, review providers in pediatric cardiology and pediatric endocrinology and look for key interests like "lipid clinic", "cholesterol", or "preventive cardiology", or search those terms under medical conditions the system treats.

Lipodystrophies are rare disorders. So many people with lipodystrophy do not know anyone else who has this problem. Patient groups like Lipodystrophy United can connect patients with lipodystrophy with other people with the same disorder.