LIPODYSTROPHY UNCOVERED

An ultra-rare, incurable condition that can reduce life expectancy

Lipodystrophy is a rare, serious and progressive disease impacting a person's ability to store fat in the body.

GENETIC OR ACQUIRED

People can be born with lipodystrophy or it can be acquired after an immune condition and occur in childhood or adolescence.[1]

ULTRA RARE

This little known condition affects approximately 1-4 people per million globally.[1] Many patients are diagnosed late in the course of their disease, when the physical symptoms may have become more severe and multi-organ damage may be irreversible.[1]

LIFE LIMITING

Lack of body fat results in a loss of leptin production, an important hormone that helps the body function properly. It can lead to severe, life-threatening metabolic problems, organ failure and premature death.[1,2,3,4] It is associated with other serious conditions, such as pancreatitis, liver disease, early onset severe diabetes, cardiovascular disease, and can cause severe organ damage if uncontrolled.[5]

INCURABLE

Lipodystrophy is incurable and can severely impair quality of life including emotional and social consequences.[2,3,4]

REFERENCES 1. Akinci, B., et al., Natural History of Congenital Generalized Lipodystrophy: A Nationwide Study From Turkey. J Clin Endocrinol Metab, 2016. 101(7): p. 2759-67. **2.** Dhankar, P., et al., Estimating Quality of Life of Patients with Lipodystrophy. Value in Health, 2015. 18(3): A292. **3.** Chiquette, E., et al. Estimating the prevalence of generalized and partial lipodystrophy: findings and challenges. Diabetes Metab Syndr omes, 2017. 10: p.375-383. **4.** Brown, R.J., et al., The Diagnosis and Management of Lipodystrophy Syndromes: A Multi-Society Practice Guideline. J Clin Endocrinol Metab, 2016. 101(12): p. 4500-4511. **5.** Kelesidis, T., et al., Narrative Review: The Role of Leptin in Human Physiology: Emerging Clinical Applications. Ann Intern Med, 2010. 152(2): p.93–100. **6.** Pope, E., et al., Childhood acquired lipodystrophy: A retrospective study. Journal of the American Academy of Dermatology, 2006. 55(6): p. 947-950 **7.** Gupta, N., et al., Clinical Features and Management of Non-HIV-Related Lipodystrophy in Children: A Systematic Review. J Clin Endocrinol Metab, 2017. 102(2): p. 363-374

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TYPES OF LIPODYSTOPHY

GENERALISED
Absence of subcutaneous fat *all over* the body.
Affects 1 in 1,000,000 people[2]

PARTIAL
Absence of subcutaneous body fat in *some parts*
Affects 3 in 1,000,000 people[2]

In both, fat is abnormally distributed with little or no subcutaneous fat in some areas and an accumulation of fat around internal organs and muscles.[4]

WHY DO WE NEED LEPTIN?

Leptin is a hormone produced by subcutaneous fat and is vital in helping control key metabolic processes in the body:

- Controls how fat is broken down or stored. If it is not used and stored properly, fat can collect in places it should not, for example, in organs, such as the liver and muscles[6]
- Helps the body respond to insulin. Without leptin, the body can develop insulin resistance, which may lead to early onset diabetes in people with lipodystrophy
- It lets the body know when you have eaten enough. People with lipodystrophy can live continuously with a severe hunger that cannot be satisfied

CHALLENGING TO LIVE WITH

Lipodystrophy is associated with an unusual body shape which can result in negative body image. Combined with significant fatigue and excessive hunger. this can cause more distress than the metabolic effects and leads to isolation, stigma and mental health issues.[7]

Lack of fat in areas such as the face, can lead to the patient looking prematurely aged.

Lipodystrophy can have devastating emotional and social consequences that restrict patients' everyday life, impacting their relationships, travel, socialising and career choices.[2,3,4]

LIPODYSTROPHY IS HARD TO TREAT

Uncontrolled lipodystrophy can cause severe organ damage.[5] Management options vary in effectiveness from person to person and include diet and exercise, medicinal treatment for the metabolic consequences of lipodystrophy such as anti-diabetics and lipid lowering agents as well as cosmetic options. Treatment options are somewhat limited and directed toward specific complications of the disease. There is currently only one treatment approved in the EU for the treatment of the complications of leptin deficiency: leptin replacement therapy.

REFERENCES 1. Akinci, B., et al., Natural History of Congenital Generalized Lipodystrophy: A Nationwide Study From Turkey. J Clin Endocrinol Metab, 2016. 101(7): p. 2759-67. **2.** Dhankar, P., et al., Estimating Quality of Life of Patients with Lipodystrophy. Value in Health, 2015. 18(3): A292. **3.** Chiquette, E., et al. Estimating the prevalence of generalized and partial lipodystrophy: findings and challenges. Diabetes Metab Syndr omes, 2017. 10: p.375-383. **4.** Brown, R.J., et al., The Diagnosis and Management of Lipodystrophy Syndromes: A Multi-Society Practice Guideline. J Clin Endocrinol Metab, 2016. 101(12): p. 4500-4511. **5.** Kelesidis, T., et al., Narrative Review: The Role of Leptin in Human Physiology: Emerging Clinical Applications. Ann Intern Med, 2010. 152(2): p.93–100. **6.** Pope, E., et al., Childhood acquired lipodystrophy: A retrospective study. Journal of the American Academy of Dermatology, 2006. 55(6): p. 947-950 **7.** Gupta, N., et al., Clinical Features and Management of Non-HIV-Related Lipodystrophy in Children: A Systematic Review. J Clin Endocrinol Metab, 2017. 102(2): p. 363-374

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