# 1. A CASE OF SUSPECTED LIPODYSTROPHY IN AN INSULIN-RESISTANT PATIENT

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**Introduction.** Lipodystrophy syndrome, a rare disease associated with leptin deficiency and characterized by disproportionate fat loss, is linked to metabolic disorders like dyslipidemia and severe insulin resistance. It is often accompanied by liver steatosis and can be familial or acquired. Diagnosis is built mainly on clinical findings and confirmed by genetic analysis, but it can be tricky sometimes as the extent of symptoms depends on the level of lipodystrophy.

**Case statement.** A 55-year-old female, known with type 2 diabetes mellitus (DM2), presents to the endocrinology department in November 2021 with complaints of extreme hunger, increased preprandial blood glucose of 12-15 mmol/l, postprandial glucose - 20-30 mmol/l, and high blood pressure of 140/70 mmHg. From anamnesis, she has been on insulin therapy since the onset of the disease in 2012 and has lost 20 kg of body weight mainly from subcutaneous fat in the upper and lower extremities. Besides DM2 she suffers from hepatitis B. Family history reveals that the patient's twin sister has the same constellation of hyperglycemia and hyperlipidemia, also her younger sister passed away at age 26 due to cirrhosis of unknown etiology. Her blood lipid profile shows abnormal triglyceride levels up to 86.39 mmol/l (N < 2.3 mmol/l). The ApoB gene mutation was suspected and the test was positive. Insulin doses were up-titrated, but despite administering 160 units of insulin, normal blood glucose levels were not achieved. During 2021-2023 she was hospitalized multiple times in the endocrinology department due to elevated blood sugars, hepatology – due to worsening liver function tests, and surgery department– due to episodes of acute pancreatitis. Glycemic control was finally achieved using a basal-bolus regimen, including U-300 insulin in combination with sodium-glucose cotransporter-2 inhibitors.

**Discussions.** Irregular distribution of body fat, family history, and extreme insulin resistance raised the suspicion of a lipodystrophy syndrome. Familial hyperlipidemia was another presumptive diagnosis, but positive gene mutation of ApoB does not correlate with the unusually increased triglyceride levels. Genetic analysis would confirm the suspected diagnosis based on the clinical picture.

**Conclusion.** Lipodystrophy is an uncommon syndrome, but this might be an underestimation, as many cases remain unrecognized due to the phenotypic variability of this syndrome. The affected storing capacity and endocrine function of adipocytes result in insulin resistance and ectopic fat accumulation with severe metabolic complications. Raised awareness amongst clinicians can lead to early suspicion and diagnosis of the syndrome and as a result – better management of comorbidities.

**Keywords**. Dyslipidemia, Insulin resistance, Lipodystrophy syndrome, triglycerides, Type 2 Diabetes mellitus.