RARE CASE OF ACQUIRED GENERALIZED LIPODYSTROPHY IN A 14-YEAR OLD PATIENT

Cristina Patricia Dumitrescu,[1] Monica Mailat[1]

[1] *C.I. Parhon National Institute of Endocrinology, Bucharest; Romania*

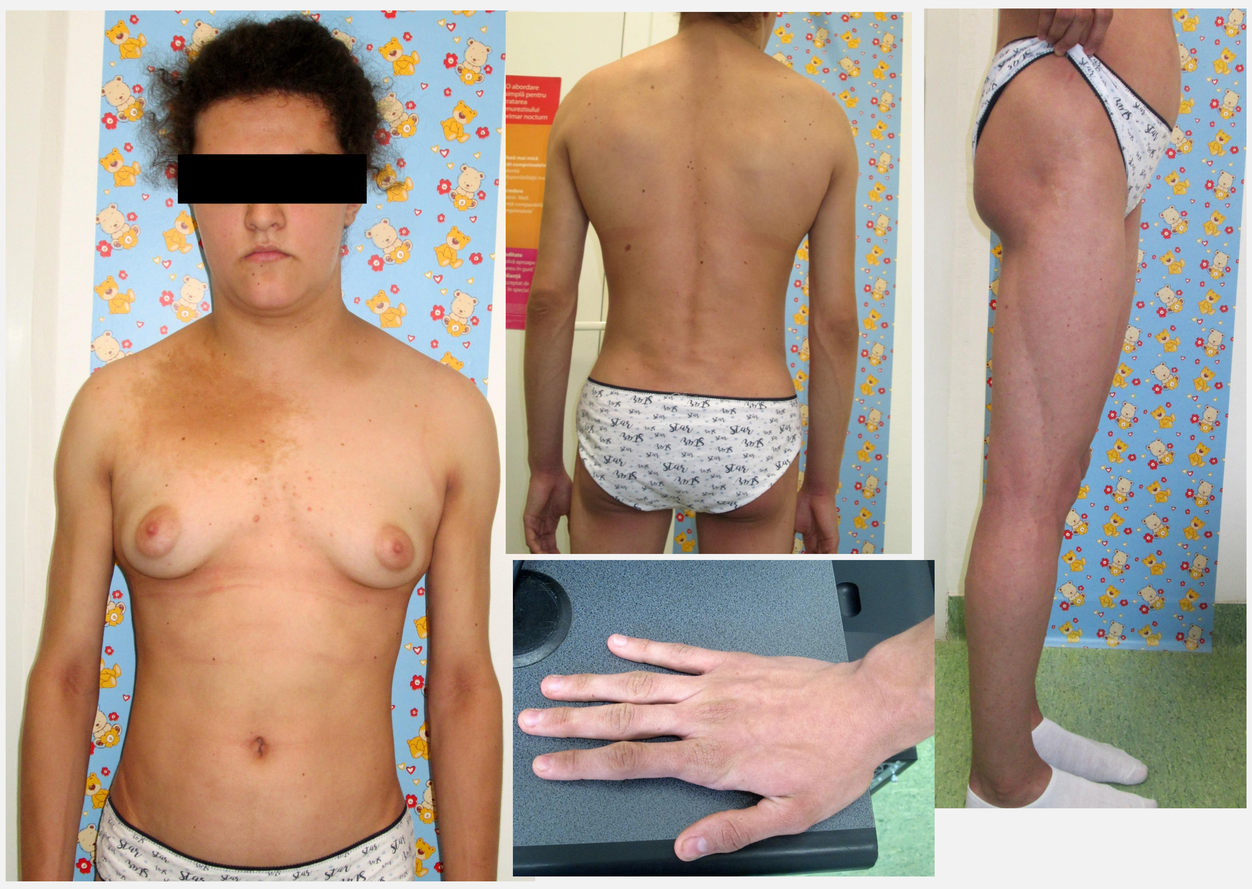
Background

Lipodystrophies are a group of rare disorders which are characterized by varying degrees of body fat loss. The metabolic abnormalities associated with lipodystrophy include insulin resistance, often leading to diabetes mellitus and its complications, hypertriglyceridemia that may be severe enough to cause acute pancreatitis and hepatic steatosis that may lead to cirrhosis.

They can be either genetic or acquired, generalized or partial. Acquired generalized lipodystrophy (AGL) is more common in females, usually appears before adolescence, is often associated with autoimmune diseases and is characterized by progressive loss of fat affecting the whole body but some fat accumulation can appear in the face, neck, axilla.

Case report

A 14-year old female presented with very low body-fat tissue, with increased muscle tissue at the trunk, upper and lower extremities. The adipose tissue is present in the face and the neck, causing a „buffalo-hump" appearance. She also presented acanthosis nigricans at bilateral axillary level and in the posterior region of the neck. The patient had normal fat distribution during childhood, followed by onset of progressive fat loss around late childhood.



Paraclinical Investigations

Blood Test	Value	Normal Range
Leptin	**2.8**	>12 ng/ml
Insulin	**65.3**	6-27 uUI/ml
TSH	**0.0002**	0.5-4.5 uUI/ml
FT4	**24**	9-19 pmol/l
TRab	**19.49**	0-1.75 UI/ml

Results

Given the clinical appearance of the patient, the very low level of leptin and the association with Graves Disease, the diagnosis of acquired generalized lipodystrophy is the most likely. For the hyperthyroidism the patient was started on antithyroid agent (30 mg Thyamasole) and Metformin was started for her progressing hyperinsulinism. Also, the patient has been instructed to begin the appropriate diet and exercise plan, recommended for all patients with this disease. Consequently to normalization of her thyroid function, cholesterol and triglyceride values started to increase. One year after starting antithyroid medication and 3 months after starting metformin she presented a transient episode of increased liver enzimes and was evaluated for autoimmune/infectios hepatitis and non-alcoholic fatty liver disease, but none was confirmed.
Also, given the very low level of leptin, the patient can be a good candidate for treatment with metreleptin.

Conclusion

This is the case of a young female with acquired generalized lipodystrophy with late childhood onset of disease, typical clinical presentation of fat loss, very low levels of leptin and insulin resistance from a young age, while also associating Graves' disease.

The association of Graves' hyperthyroidism and its therapy with AGL influences the evolution of AGL metabolic consequences and comorbidities.