Brief Communication

Recurrent stroke as a presenting feature of acquired partial lipodystrophy

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ABSTRACT

Acquired partial lipodystrophy (PL) (Barraquer–Simons syndrome) is a rare condition with onset in childhood, and it is characterized by progressive loss of subcutaneous fat in a cephalocaudal fashion. This report describes a case of acquired PL in a 16-year-old girl, who had progressive loss of facial fat since 3 years. Systemic lupus erythematosus (SLE), anticardiolipin antibody, primary hypothyroidism, diabetes, and dyslipidemia may antedate the development of complications such as cerebrovascular stroke and cardiovascular disease. The girl had developed recurrent left hemiparesis, and withdrawn from school due to poor performance.

Key words: Recurrent stroke, acquired partial lipodystrophy, Barraquer-simons syndrome

Introduction

Lipodystrophy is a rare and heterogenous disorder characterized by loss of adipose tissue from different parts of the body. Classification of the different lipodystrophy syndromes is based mainly on whether the disease is congenital/familial or acquired, and whether the fat loss is generalized or partial.[1] Partial lipodystrophy (PL) usually begins during the first decade of life and occurs more frequently in females. There is a gradual symmetric loss of subcutaneous tissue. The sites of loss are heterogenous. The most common variant involves loss of fat from the face and the upper half the body, resulting in a cadaverous faces and marked disproportion between the upper and lower halves of the body (Weir-Mitchell type).[2]

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Several autoimmune diseases, in particular systemic lupus erythematosus (SLE) and dermatomyositis, were associated with APL. The prevalence rates of diabetes mellitus and impaired glucose tolerance were 6.7% and 8.9%, respectively.[3] Approximately, 83% of acquired partial lipodystrophy (APL) patients had low complement (C3) levels and the presence of polyclonal immunoglobulin C3 nephritic factor (C3NeF). Twenty-two percent of patients developed membranoproliferative glomerulonephritis (MPGN) after a median of approximately 8 years following the onset of lipodystrophy.[3]

This report describes a case of acquired PL in a 16-year female patient with facial fat atrophy with SLE, anticardiolipin antibody, primary hypothyroidism, and diabetes.

Case Report

A 16-year-old girl presented with a history of weakness of left upper limb and lower limb to emergency department. She had similar history of left-sided weakness 6 months back. She is a product of consanguineous marriage of healthy parents, born at term with a birth weight of 2.5 kg. There was no history of birth asphyxia or gestational diabetes in the mother. She has one younger brother aged

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6 years and was reported to be healthy. There was no history of similar cases in the family. There was progressive loss of facial fat for the last 3 years [Figure 1].

The child was diagnosed primary hypothyroidism 10 months back on thyroxine (100 μg/day) replacement. She has been diagnosed as type 2 diabetes mellitus 3 months back on oral hypoglycemic agents (glimepiride, 2 mg; metformin, 1,000 mg; and pioglitazone, 30 mg) with moderate glycemic control. There was history of lethargy and daytime sleepiness. There was history of operation for bilateral cataract at the age of 8 years. She had attained menarche at the age of 13 years and the periods are regular.

Physical examination showed marked symmetrical atrophy of fat over face, neck, shoulders, arms, and forearms and that extends to the thoracic region and upper abdomen. Muscular prominence is noticed in the above areas. Lower abdomen, pelvic girdle, and lower limbs were spared. Acanthosis nigricans and diffuse thyromegaly were noted. There was weakness of left upper and lower limbs. There was no facial muscle weakness.

Her weight was 54 kg (25-50th percentile) and height was 159 cm (50-75th percentile). The clinical assessment of cardiovascular and respiratory system was normal. There was no hepatosplenomegaly. The biochemical and laboratory details are presented in Table 1.

Adolescent onset progressive loss of subcutaneous fat over the face and associated autoimmune diseases such as SLE, anticardiolipin antibody, primary hypothyroidism, and diabetes complicating cerebrovascular accident with recurrent left hemiparesis suggests the diagnosis of acquired PL. The girl was started on antiplatelet agents, statins, 2 mg coumarin, 100 μg thyroxine, 1,000 mg metformin, and 15 mg pioglitazone

Figure 1: Lipoatrophy of face (characteristic feature of acquired partial lipodystrophy)

Discussion

Acquired PL, also known as Barraquer–Simons syndrome or cephalothoracic lipodystrophy, is a very rare condition with onset in childhood or adolescence.[3] PL is characterized by progressive loss of subcutaneous fat of the face, neck, trunk, and upper extremities in a cephalocaudal fashion sparing the lower extremities. Majority of patients with PL appear normal, whereas there is evidence supporting an association with renal disorders, autoimmune function, hepatomegaly, hyperlipidemia, and glucose intolerance.[4] Women are affected approximately three times more often than men. The diagnosis of the disease is mainly clinical. The laboratory workup is needed mainly to investigate for the presence of associated disorders, which are metabolic, autoimmune, and renal diseases.

Lipodystrophy progresses slowly and occurs over a period of a few months to 2 years. Seventy-five percent of patients had significant fat loss when younger than 13 years of age. Acquired PL is associated with autoimmune disorders such as SLE, dermatomyositis, hypothyroidism, pernicious anemia, celiac disease, dermatitis herpetiformis, rheumatoid arthritis, temporal arteritis, and leukocytoclastic vasculitis.[5]

Table 1: Laboratory details of the patient

Analyte	Patient value	Reference range
Hemoglobin (gm/dl)	8.6	12-15
Packed cell volume (hematocrit)	29	38-47
Total WBC count (cells/cumm)	8,300	4,000-11,000
Platelet count (per cumm)	195,000	150,000-400,000
ESR (mm 1st h)	42	20
Urine microalbumin (mg/L)	86	5-20
C 3 complement	Normal	
C 4 complement	Normal	
Creatinine (mg/dl)	0.89	0.6-1.3
Albumin (gm/dl)	3.9	3.5-4.5
Sodium (meq/L)	135	130-145
Potassium (meq/L)	4.8	3.5-5.0
Thyroid stimulating hormone (TSH) (mIU/ml)	8.1	0.35-5.6
Fasting blood glucose (mg/dl)	134	70-100
Post-glucose blood sugar (2 h) (mg/dl)	245	<140
Total cholesterol (mg/dl)	216	<200
Triglycerides (mg/dl)	201	<150
High density cholesterol (mg/dl)	25	>50
Low density cholesterol (mg/dl)	151	<130
Calcium (mg/dl)	8.6	8.5-10.5
Phosphorous (mg/dl)	3.8	2.5-4.8
Alkaline phosphatase (IU/L)	242	164-324
Iron (μg/dl)	67	75-165
Total iron binding capacity	310	200-400
Antinuclear antibody-IgM	Positive	Negative
Anticardiolipid antibody	Moderate positive	Negative
dsDNA antibody-IgM	Positive	Negative
Rheumatoid factor	Negative	Negative

ESR: Erythrocyte sedimentation rate

Nephropathy, in the form of MPGN, occurs in approximately 20% of the patients.[3] Usually, patients do not have clinically evident renal disease or abnormalities in renal function until they have had the disease for 8 or more years.[6] In our patient, there was no renal involvement, but we will follow the renal function tests of the patient in outpatient clinic visits.

The precise pathophysiology of fat loss is unclear. Activation of an alternate complement pathway, C3 hypocomplementemia with lysis of adipocytes induced by C3NeF, has been implicated.[7] C3 hypocomplementemia likely contributes to the association of this syndrome with autoimmune diseases and with a propensity for patients to acquire bacterial infections.[3] Other proposed mechanisms include an autoimmune process and genetic associations.[8]

The exact frequency of stroke in SLE is not known, but the risk of recurrence may be 50% or higher, and the occurrence of stroke increases the risk of seizures.[9] The most frequent etiology was a cardiogenic embolus or an antibody-mediated hypercoagulable state, with cerebral vasculitis occurring in association with infection. Patients who have had a stroke are at high (64%) risk for a recurrent stroke.[10] Because of the decreased fibrinolysis seen in patients with SLE, anticoagulant therapy may be the most effective preventive treatment currently available.

In conclusion, progressive PL in association with autoimmune disorders should be regularly observed for the development of complications. These children and parents often require considerable support to prevent metabolic complications and consequences of distressing physical abnormalities and appearance.

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