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CASE REPORT

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Barraquer-Simons syndrome in systemic lupus erythematosus: A case report

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Abstract

Barraquer-Simons syndrome is a rare entity characterized by progressive loss of subcutaneous tissue in the face and/or upper half of the body and can be associated with autoimmune conditions such as systemic lupus erythematosus. Close long-term follow-up is required to identify metabolic disturbances, potentially life-threatening renal problems, and other associated diseases.

KEYWORDS

Barraquer-Simons syndrome, lipodystrophy, partial lipodystrophy, systemic lupus erythematosus

1 | INTRODUCTION

Lipodystrophies are rare heterogeneous disorders in which there is a generalized and partial/localized absence of subcutaneous tissue, both of which can be inherited or acquired.[1]

Barraquer-Simons syndrome (BSS) is an acquired form of partial lipodystrophy, characterized by bilateral symmetrical loss of adipose tissue that begins in the face and can spread to other body parts such as the neck, shoulder, arms, and trunk, but variably without the involvement of the lower extremities.[2] The condition has female predisposition with a female to male ratio of 4:1.[3]

Acquired partial lipodystrophy (APL) is rare with approximately only 250 cases described to date.[4] Several reports have shown the association of BSS with autoimmune diseases in a minority of patients, in particular, systemic lupus erythematosus (SLE), and dermatomyositis.[5]

Herein, we report a case of a 26-year-old female patient who was diagnosed with SLE at an age of 18 years with poor compliance to medication and later developed partial lipodystrophy of the face.

2 | CASE REPORT

A 26-year-old female patient was diagnosed with systemic lupus erythematosus at the age of 18 when she presented to a health center with erythematous lesions over her face and joint pains. She was kept on maintenance medication of prednisolone 5 mg and Hydroxychloroquine 200 mg which she discontinued on her own after 5 years of medication and lost to follow-up. She presented to our

center this time with complaints of a similar facial rash for 3 months and fever for one month. She was managed with prednisolone 50 mg for 2 weeks which was tapered over the next 2 months to a maintenance dose of 5 mg along with hydroxychloroquine 300 mg daily and mycophenolate mofetil 2 gm daily.

She again presented to our center after 3 months of re-initiation of therapy with complaints of gradual and progressively shrunken eyeballs. (Figure 1) On physical assessment, she was diagnosed with partial lipodystrophy. There were no such lesions elsewhere in her body and examination of other systems was unremarkable. There was no history of similar lesions among any of the family members. Furthermore, there were no preceding skin symptoms or symptoms suggestive of viral illness prior to the symptom onset. Laboratory examination for thyroid function test, liver function test, renal function test, cortisol level, glucose, and lipid profile were within normal range. However, C3 and C4 levels were decreased. Table 1 shows these parameters along with the patient's autoimmune profile. Serology was negative for HIV, hepatitis B, and hepatitis C. Histopathological examination from the lipodystrophic area showed mild orthokeratosis, thinning of the epidermis with vacuolar degeneration of the basal layer, and decreased adipocytes lobules in the subcutaneous layer. (Figure 2).

The patient was counseled regarding the available treatment options. However, considering the financial strains, she opted not to undergo any form of treatment, is currently under medications for SLE, and is under regular follow-up. On the last follow-up, SLE is under remission without any change in the previous lipodystrophic area and no new similar lesions elsewhere.

3 | DISCUSSION

The lipodystrophy syndromes constitute a rare group of disorders characterized by a selective deficiency of adipose tissue in the absence of nutritional deprivation or catabolic state and without any evidence of inflammation.[6,7] It occurs among children and in young adults. BSS or APL usually begins between five to fifteen years of age. A study reporting on 35 patients with APL showed the median age of all patients at presentation was 25 years (range, 4–65 years); however, the onset is often earlier.[5] Our patient was 26 years old at the time of presentation and had not previously noticed any such skin lesion.

Though highly active antiretroviral therapy-induced lipodystrophy has become the most common form of acquired partial lipodystrophy, other forms of partial lipodystrophy syndromes are uncommon and the prevalence is currently estimated to be only 1.67 per million cases.[4,8] Although the exact mechanism of fat loss in patients with APL is largely unknown, alternate complement pathway activation, and C3 hypocomplementemia with lysis of adipocytes induced by C3NeF have been implicated.[9] As C3 hypocomplementemia has been reported in 70%–80% of patients with BSS, this feature is considered a critical marker for the differential diagnosis of this type of lipodystrophy.[3,6] C3 and C4 levels were below the normal in our patient too. Moreover, the immunological disorder might be related to APL as there have been several cases of lipodystrophy with SLE and systemic sclerosis.[10,11] In addition, though less frequent, other autoimmune diseases that are associated with APL are discoid lupus erythematosus, rheumatoid arthritis, autoimmune thyroiditis, autoimmune hepatitis among many others.[5] Our patient was diagnosed case of SLE and was under remission therapy; however, she had left the medications a few years back on her own.

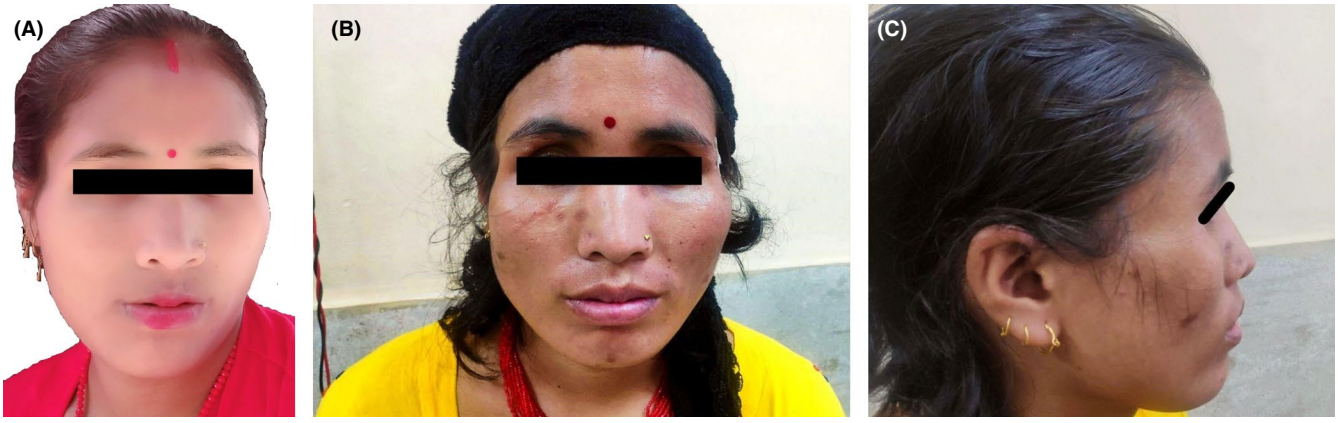

FIGURE 1 Patient facial appearances before the onset of partial lipodystrophy (A) and anterior and lateral facial appearance of the patient at the time of hospital presentation after the onset of partial lipodystrophy (B and C)

TABLE 1 Various laboratory parameters of the patient

Parameter	Reference range
Random Blood Glucose = 4.0	3.5–6.1 mmol/L
Total Cholesterol = 4.7	3.5–5.1 mmol/L
HDL Cholesterol = 1.0	0.8–1.6 mmol/L
LDL Cholesterol = 3.2	Upto 4.0 mmol/L
Triacylglycerol = 1.0	0.5–1.8 mmol/L
Thyroid function test	
Thyroid stimulating hormone (TSH) = 4.0	0.46–4.68 microIU/L
Free T3 = 7.45	4.26–8.1 pmol/L
Free T4 = 15.6	10.2–28.2 pmol/L
8:00 am cortisol level- Normal	
Complement Level	
C3 level = 0.34	0.9–1.8
C4 level = 0.03	0.1–0.4
Renal Function Test	
Serum Urea = 3.5	1.6–7.0 mmol/L
Serum Creatinine = 65	40-110 micromol/L
Serum Sodium = 142	135-146 mEq/L
Serum Potassium = 3.0	3.5–5.2 mEq/L
Autoimmune Profile	
ANA +ve	
Anti-Smith antibody +ve	
P-ANCA +ve	
Anti-histone +ve	
Anti-dsDNA +ve	

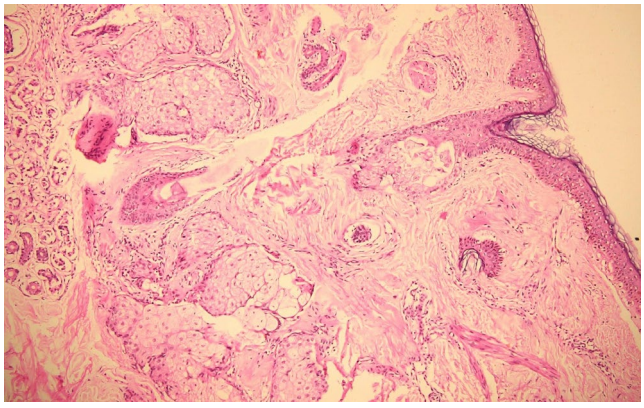

FIGURE 2 Sections from the skin tissue show orthokeratosis with thinned epidermis and decreased adipocytic lobules in the subcutaneous layer

Partial lipodystrophy causes a slow and symmetrical progressive loss of fat in the face and can also involve the upper half of the body in about 10% of the cases.[12] The condition can also be associated with metabolic syndrome, acanthosis nigricans, hirsutism, and liver cirrhosis.[11] The patient had a bilateral fat loss in her orbital region without any other systemic abnormalities. Owing to the rarity of the disease condition, lipodystrophy syndromes are frequently challenging, unrecognized, or misdiagnosed, especially in men as the fat loss is very gradual. Also, as no diagnostic criteria have been established based on skinfold measurements or imaging procedures, lipodystrophy should be suspected in patients with regional or generalized lack of adipose tissue outside of the normal range by physical examination, the findings can be aided by various investigations including anthropometry, dual-energy x-ray absorptiometry, and whole-body magnetic resonance imaging. Along with fat loss and the presence of autoimmune disease, the suspicion of acquired lipodystrophy grows stronger.[6] The presence of Systemic lupus erythematosus, not under antiretroviral therapy and exclusion of other causes that can potentially explain the cause of fat loss led to the diagnosis in our case. The histopathological findings in our case showed decreased adipocytic lobules in the subcutaneous layer with thinning of the epidermis without infiltration with lymphocytes, mononuclear cells, or giant cells which is in accordance with reported histopathological findings.[5]

Because there is no cure for lipodystrophy and no treatment that can regrow adipose tissue, the management of subcutaneous fat loss is challenging and often frustrating for physicians and surgeons.[5,6] Plastic surgery may improve appearance in some people and the possible interventions include autologous fat transfer, dermal fillers, or muscle grafts to treat facial lipoatrophy, although data are limited. Though facial lipofilling can give excellent results, reinjected fat disappears after a few years, and thus, the effect is transient.[3] Walker et al.[13] demonstrated a good cosmetic outcome of rosiglitazone treatment in a young woman with APL. Metreleptin is another drug that has been used in the treatment of generalized lipodystrophy often when there are associated metabolic or endocrine abnormalities. However, the response to metreleptin in partial lipodystrophy is less robust.[6] Moreover, changes in physical appearance from lipodystrophy can cause physical and psychological distress which is to be taken care of as a part of treatment.

Membranoproliferative glomerulonephritis (MPGN) is reported in approximately 20% of patients with APL and also, as the deficient adipose mass in lipodystrophy can result in a collection of metabolic complications such as diabetes, dyslipidemia, non-alcoholic fatty liver disease, and cardiovascular and reproductive dysfunction, the patients should be screened regularly.[5] However, the risk of metabolic complications is low, and the clinical judgment should guide follow-up screening.[6] Our patient did not opt for any of the treatment, has no metabolic or endocrine abnormalities, and is under regular follow-up.

4 | CONCLUSION

Barraquer-Simons syndrome is an extremely rare disorder with important clinical consequences and psychosocial effects. For diagnosis, clinical recognition and physical examination are critical. Close long-term follow-up is required to identify metabolic disturbances, potentially life-threatening renal problems, and other associated diseases.

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CONFLICT OF INTEREST

None.

AUTHOR CONTRIBUTIONS

SA and SJ were involved in the management of the patient. SBT and AB examined and interpreted the pathology. SA, SK, and SS reviewed the literature, collected all the required case information, images, slides, reports, and contributed to writing manuscripts. SJ, SSP, SBT, and AB reviewed and edited the manuscript. All authors read and approved the final manuscript.

CONSENT

Written informed consent was obtained from the patient before the submission of the report. The signed Institutional Consent Form is on file.

DATA AVAILABILITY STATEMENT

The data that support the findings of this study are available from the corresponding author upon reasonable request.

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