Do you know this syndrome? *

Você conhece esta síndrome?

Mônica Santos [1]
Lisiane Nogueira [2]
Renata Rabelo [2]
Carolina Talhari [1]
Virgínia Vilasboas [2]
Sinésio Talhari [3]

CASE REPORT

IDENTIFICATION: female, 29 years old, single.

MEDICAL HISTORY: progressive reduction of body fat beginning at the age of eleven, after presenting symptoms of intestinal infection.

DERMATOLOGIC EXAMINATION: symmetrical loss of body fat in the face, trunk, upper and lower limbs, with an aspect of muscular hypertrophy (Figures 1 and 2). Yellowish and hard nodules, some painful, in the thighs, arms and buttocks were also present.

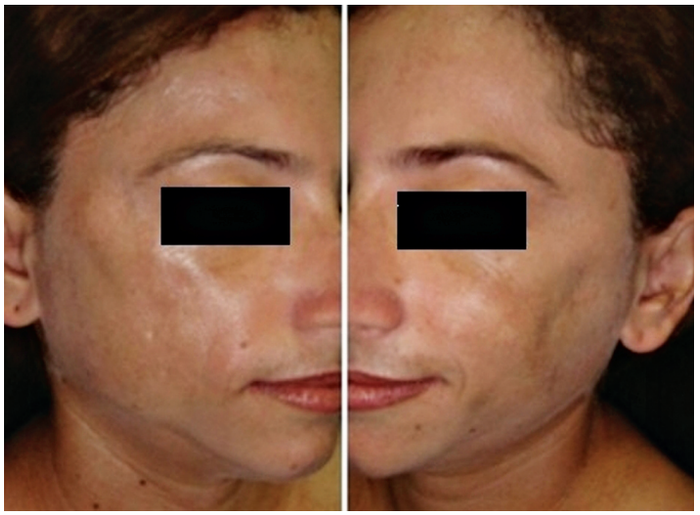

FIGURE 1: Lipoatrophy in zygomatic-temporal region and reduced Bichat's ball

COMPLEMENTARY TESTS: blood count, fasting glucose, electrolytes, normal liver and kidney function, normal glycemic levels (fasting = 71, 30min = 124; 60min = 124; 120min = 119mg/dL), absence of glycosuria on urinalysis, unaltered thyroid and abdominal ultrasound; anti-peroxidase antibody 9.3 U / l (normal up to 34U / l), nonreactive ANA (antinuclear factor); normal dosage of C3; HIV and hepatitis serology were negative. ❑

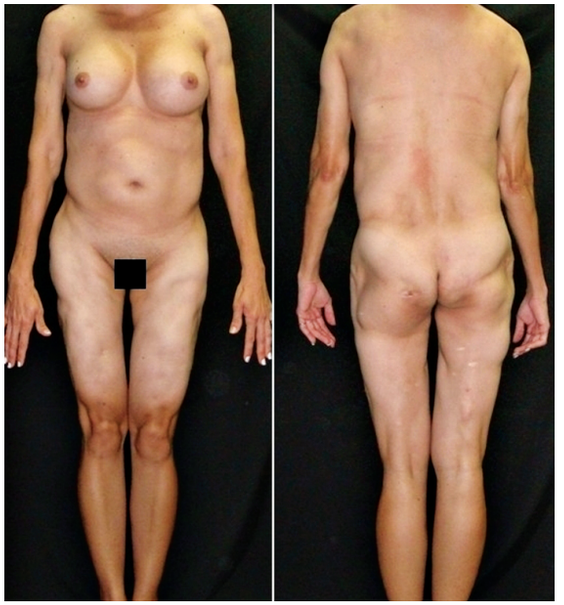

FIGURE 2: Symmetrical loss of body fat involving the trunk, buttocks, thighs and arms, giving an appearance of muscular hypertrophy

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[1] PhD in Infectious and Parasitic Diseases; Dermatologist at the Tropical Medicine Foundation of the Amazonas (FMTAM); Professor of Dermatology at the University of the State of Amazonas (UEA) – Amazonas (AM), Brazil.
[2] MD; Resident in Dermatology at the Tropical Medicine Foundation of the Amazonas (FMTAM).
[3] PhD in Dermatology; President/Director of the Tropical Medicine Foundation of the Amazonas (FMTAM) – Manaus (AM), Brazil.

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Santos M, Rabelo R, Vilasboas V, Nogueira L, Talbari C, Talbari S

DISCUSSION

The Barraquer-Simons syndrome, also called progressive partial lipodystrophy or cephalothoracic lipodystrophy, was described by Barraquer in 1906 and Simons in 1911.[1] It is characterized by progressive and symmetrical loss of subcutaneous tissue in craniocaudal direction, starting on the head and progressively involving the trunk, arms and legs up to the thighs. However, arms and legs are rarely affected. The onset of the disease a often occurs in the first and second decade of life, predominantly affecting females.[2]

The etiology of this syndrome remains unknown. Although considered an acquired form of lipodystrophy often associated with viral infections, the disease has also been recently linked to mutations in the gene encoding the nuclear lamina protein (lamin B2- LMNB2).[3] In 2006 Hegele et al. analyzed eight patients with the Barraquer-Simons syndrome, finding in four of them a mutation in this gene.[4] Other studies show that some patients with this syndrome have altered levels of C3, consistent with the presence of a nephritic factor, which increases the consumption of C3 and reduces its synthesis. *In vitro* studies demonstrate that the nephritic factor also has a lipolytic effect, what explains the lipodystrophy in these patients.[5]

The major differential diagnoses of the syndrome are other forms of lipodystrophy, particularly congenital partial lipodystrophy and lipodystrophy in AIDS patients undergoing antiretroviral therapy.[6] However, metabolic disorders such as glucose intolerance, dyslipidemia and diabetes are frequent in these diseases, while in the Barraquer-Simons syndrome they are uncommon.[7]

The Barraquer-Simons syndrome is classified into three subtypes: subtype I, associated with panniculitis; subtype II, associated with systemic diseases, especially hypothyroidism, dermatomyositis, dermatitis herpetiformis, systemic lupus erythematosus, leukocytoclastic vasculitis, mesangiocapillary glomerulonephritis; subtype III or idiopathic, which represents more than 50% of the cases and is not associated with systemic diseases.[8] The patient described did not show associated co-morbidity, being classified as Barraquer-Simons syndrome subtype III.

Treatment options are mostly indicated to correct facial lipoatrophy; facial reconstruction techniques have been used to restore lost facial contours, such as fat grafting or filling with polylactic acid or polymethylmethacrylate.[9] In more severe cases of lipodystrophy, especially those with metabolic alterations, recombinant leptin has been used, a hormone secreted by fat cells responsible for regulating glucose metabolism and storing fat cells.[10]

In this paper we report a case of Barraquer-Simons syndrome with onset in puberty and which is not associated with systemic disorders. Apart from lipodystrophy, the patient presented with hardened areas on the arms, thighs and buttocks; histological examination confirmed the diagnosis of calcinosis, an uncommon finding in these patients. ❑

Abstract: Barraquer-Simons syndrome, also called acquired partial lipodystrophy or cephalothoracic lipodystrophy, is a rare form of progressive lipodystrophy, characterized by symmetrical lipoatrophy of subcutaneous adipose tissue starting in the head and spreading to the thorax, upper and lower limbs and thighs. In this work, we report the case of a patient with Barraquer-Simons syndrome without systemic complications.
Keywords: Diagnosis; Lipodystrophy; Physical examination

Resumo: A síndrome de Barraquer-Simons, também denominada lipodistrofia parcial progressiva ou lipodistrofia céfalo-torácica, caracteriza-se por perda progressiva do tecido celular subcutâneo, em direção crânio-caudal, de modo simétrico, iniciando na face e envolvendo progressivamente o tronco, membros superiores e inferiores, até as coxas. Nesse trabalho relata-se caso de paciente, com síndrome de Barraquer-Simons, sem associação com co-morbidades sistêmicas.
Palavras-chave: Diagnóstico; Exame físico; Lipodistrofia

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Endereço para correspondência / Mailing address:
Mônica Nunes de Souza Santos
Av. Djalma Batista, 1061, sala 610, chapada.
69050-010. Manaus-Am
E-mail: m.n.souza.santos@gmail.com

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