OLGU SUNUMU CASE REPORT

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Barraquer-Simons Syndrome Occurred After Pregnancy: A Rare Case Report

Gebelik Sonrası Meydana Gelen Barraquer-Simons Sendromu: Nadir Görülen Bir Olgu

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ABSTRACT Lipodystrophies are a widespread group of genetic or acquired metabolic disorders that are characterized by varying degrees of body fat loss. The Barraquer-Simons syndrome also called Acquired Partial Lipodistrophy is a form of partial lipodistrophy of unknown etiology, characterised by the loss of subcutaneous adipose tissue, limited to upper part of the body. Also it can be associated with hypocomplementemia, diabetes, and hypertriglyceridemia. In this paper a 34-year-old woman with progresive loss of subcutaneous fat limited to upper arm, which was developed after pregnancy, is reported and literature was reviewed.

Keywords: Acquired partial lipodistrophy; Barraquer-Simons syndrome; upper arm lipodistrophy

ÖZET Lipodistrofiler genetik veya sonradan kazanılan vücutta değişik derecelerde yağ doku kaybı ile seyreden bir grup metabolik bozukluklardır. Barraquer-Simons sendromu diğer ismi ile "Kazanılmış Parsiyel Lökodistrofi" parsiyel lökodistrofilerin bir formudur. Etiyolojisi bilinmemektedir ve genellikle üst ekstremitede yağ doku kaybı ile seyretmektedir. Bunun dışında hipokomplemantemi, diyabet ve hipertrigliseridemi ile ilişkili olabilmektedir. Bu yazıda gebelik sonrasında üst kolda ilerleyici subkutan yağ doku kaybı olan Barraquer-Simons sendromlu 34 yaşındaki kadın olgu sunulmuştur ve bu olgu ile ilgili literatür bilgileri gözden geçirilmiştir.

Anahtar Kelimeler: Kazanılmış parsiyel lökodistrofi; Barraquer Simons sendromu; üst kolda lipodistrofi

The lipodystrophies are rare disorders characterized by selective but variable loss of adipose tissue. They are a group of acquired or genetic disorders which are characterised by selective fat loss, ranging from partial to generated.[1,2] Metabolic complications, such as insulin resistance, diabetes mellitus, hypertriglyceridemia, and fatty liver, increase in severity with the extent of fat loss.[3]

Barraquer-Simons syndrome -now called Acquired Partial Lipodystrophy (APL)- was first defined as lipodystrophic disorder about a century ago. Only 250 cases had been reported in the literature since then. It is characterized by the loss of subcutaneous tissue, limited to upper part of the body, with the face, neck, arms, thorax, and upper abdomen.[4] Women are more often affected than men.[5] Different from other types of lipodystrophies, insulin resistance and hipertriglyceridemia are less severe. Although patients

may have decreased serum complement-component 3 (C3) levels which is associated with the presence of renal involvement such as membranoproliferative glomerulonephritis.[6]

CASE REPORT

A 34-year-old female patient admitted to our clinic with sudden-onset complaints of local atrophy occurred in the distal part of the left upper arm after pregnancy (Figure 1). Laboratory examinations of biochemistry, including urea, creatinine, creatine kinase, thyroid function test, complete blood count, erythrocyte sedimentation rate, C-reactive protein, C3 level, and rheumatoid factor levels were normal. Her fasting blood glucose (87mg/dl) and insülin (6,2 mU/mL) levels were in the normal range. There was a mild elevation of LDL-cholesterol (140 mg/dl), with normal triglycerides level.

There was no evidence of myopathy in electromyographic study. Humerus magnetic resonance imaging showed that loss of subcutanous fat in the posterolateral region of distal arm (Figure 2). The patient presented no renal disease, and/or more severe metabolic and other systemic disease. A family history of lipodisthrophia was absent. The overall clinical and biochemical characteristics of our patient led us to think about Barraquer-Simons syndrome as being the main diagnosis. A written informed consent was obtained from the patient to publish her condition as a case report.

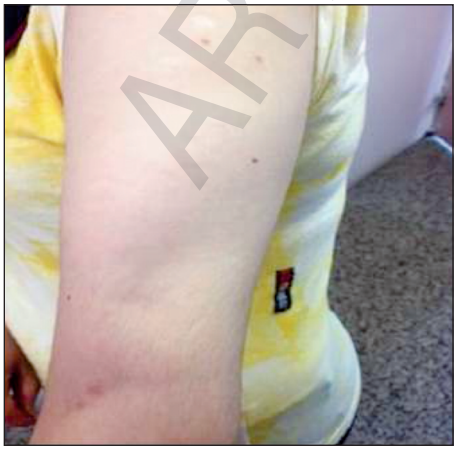

FIGURE 1: Progressive fat tissue loss on the distal arm.

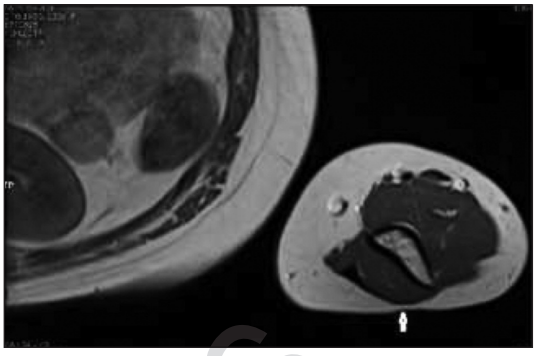

FIGURE 2: Axial non-contrast T1-weighted magnetic resonance image showing fat tissue thinning on the distal arm (white arrow).

DISCUSSION

The Barraquer-Simons syndrome is a form of partial lipodystrophy with an unknown etiology, featured by the loss of subcutaneous adipose tissue and limited to upper part of the body. This 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.[7] We classified our patients as Barraquer-Simons syndrome subtype III hence she did not have any other comorbidities.

Mutations in several genes have been found in patients with inherited lipodystrophies, including mutations in *LMNA*, *PPARG, AKT2,* and *ZMPSTE24* in partial lipodystrophy, and mutations in *AGPAT2, BSCL2, CAV1,* and *PTRF* in congenital total lipodystrophy.[8-11] However, the molecular pathogenesis of APL has not been clearly established. In 2006, Hegele proposed that *LMNB2* could be a mutation responsible for APL. In four out of nine patients he found three new rare *LMNB2* mutations, by using candidate gene sequencing.[12]

The abnormal fat repartition was in conformity with the essential criterion proposed by Misra et al. with subcutaneous fat loss from the face, neck, upper extremities, thorax, and abdomen, sparing

the lower extremities.[13] Some supportive criteria were also onset during adolescence, the absence of a family history of lipodystrophy, and low serum levels of C3. The C3-nephritic factor induces lysis of adipocytes expressing factor D (adipsin) which is a serine protease enzyme leads to loss in the fatty tissue when it is overexpressed.[14,15]

Acquired partial lipodystrophy is the major extrarenal manifestation of C3 glomerulopathy, and presented with complement alternative pathway disorders, associated frequently to the presence of C3 nephritic factor.[16] In a recent study, a 26-year-old female patient have been presented with low C3 levels and crescentic glomerulonephritis associated with acquired partial lipodystrophy.[17]

Barraquer-Simons syndrome may be presented without renal disorders. For instance, Heidemann et al described a female patient with symmetrical loss of adipose tissue from face, neck, upper extremities, and the trunk with onset in early childhood.[18] Similar to our case, this patient had no renal impairment and sistemic other disorders that may be associated with this syndrome.

Treatment of lipodystrophy in these patients is limited to cosmetic restoration, and autologous fat grafting has been shown sustained positive effects with no or very little loss of volume at follow-ups.[19]

CONCLUSION

In this paper a case of Barraquer-Simons syndrome with sudden onset after pregnancy was reported. The patient presented loss of fat on the distal upper arm, and she had no complaints of pain. There was no systemic disease which was related to lipodystrophia. Cosmetic restoration was suggested as a treatment option. It is crucial that the diagnosis of this very rare syndrome should be kept in mind by physicians in local atrophies at the extremities.

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