Progressive right lower extremity enlargement

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Received: 3 June 2011 / Revised: 29 August 2011 / Accepted: 5 September 2011 / Published online: 10 November 2011
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Diagnosis

Acquired partial lipodystrophy of the left lower extremity.
Normal right lower extremity.

Discussion

The patient and clinicians were concerned about the right lower extremity when in fact MRI showed the diseased extremity to be the left. The MR images (Figs. 1 and 2) show loss of subcutaneous fat throughout the left lower extremity with relative sparing of the muscle mass and medial thigh subcutaneous fat, accounting for the asymmetric size of the lower extremities and the misleading clinical appearance of an abnormally enlarged right lower extremity. The MRI distribution of subcutaneous fat loss of the left extremity was felt to be consistent with the diagnosis of lipodystrophy and in this patient attributed to adult onset dermatomyositis, which has been previously associated with lipodystrophy [1–3]. In addition, increased T2 signal on the STIR sequence within the subcutaneous tissues, particularly overlying the lateral left leg are indicative of active disease.

Lipodystrophies are a group of disorders characterized by the loss of adipose tissue from various parts of the body, and have been categorized as being congenital or acquired and further subcategorized as generalized, partial, or

localized [4]. The generalized varieties include congenital generalized lipodystrophy (Berardinelli–Seip syndrome) and acquired generalized lipodystrophy. Berardinelli–Seip syndrome is a rare autosomal recessive disorder in which there is a complete absence of adipose tissue at birth, while acquired generalized lipodystrophy manifests in late childhood and adolescence. Partial lipodystrophies include familial partial lipodystrophy (Dunnigan variety), which presents at onset of puberty, and acquired forms, which have been linked to HIV, drug use, and various autoimmune diseases. Localized forms of lipodystrophy are most often related to trauma, drug use, and autoimmune causes [4].

Patients affected by lipodystrophy are predisposed to insulin resistance. The central cause of morbidity and mortality in patients with lipodystrophy are diabetes mellitus and its long-term complications, including recurrent pancreatitis related to hypertriglyceridemia, chronic liver disease related to hepatic steatosis, and atherosclerotic disease [5].

Acquired lipodystrophies are the most common subtype, with HIV lipodystrophy accounting for over 100,000 cases of lipodystrophy in the United States alone [5]. The typical pattern of HIV lipodystrophy is loss of subcutaneous fat in the face and extremities, with compensatory circumferential adipose deposition in the submental (double chin), dorso-cervical spine (buffalo hump), chest, and intra-abdominal regions [6]. This is in contradistinction to acquired partial lipodystrophy (Barraquer–Simons syndrome) and acquired generalized partial lipodystrophy. Acquired partial lipodystrophy is very rare (250 cases in the English literature), and characterized by subcutaneous adipose loss involving the face, neck, arms, chest, and abdomen, with compensatory circumferential adipose deposition in the thighs and lower legs [5]. Acquired generalized lipodystrophy (80 reported cases in the English literature) is characterized by circum-

The case question and presentation can be found at doi:10.1007/s00256-011-1271-7.

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ferential fat loss involving the face, chest, abdomen, and extremities [5].

Lipodystrophy has been linked to patients with various autoimmune conditions, and is particularly well established in juvenile dermatomyositis [7–10]. A study by Singh and Bansal [8] retrospectively reviewed 33 cases of juvenile dermatomyositis and found 10 cases of lipodystrophy, and a review performed by Pope et al. [7] found that of 23 patients identified with lipodystrophy, 78% of them had an underlying diagnosis of juvenile dermatomyositis. More recently, Bingham et al. [1] identified 28 cases of lipodystrophy in a cohort of 353 patients (7.9%) with juvenile dermatomyositis and an additional case of lipodystrophy in a patient with adult dermatomyositis. Generalized, partial, and localized lipodystrophy have all been described with juvenile dermatomyositis [1, 7–9]. Patients with generalized lipodystrophy have fat loss from all extremities, face, chest, and trunk; partial lipodystrophy typically involves fat loss in the arms, face, and usually the lower extremities; and focal disease typically involves the buttock or thigh [1]. Fat redistribution has been reported in dermatomyositis patients with generalized (38%) and partial (69%) lipodystrophy [1]. A particularly unique pattern of fat loss demonstrated in dermatomyositis patients with lipodystrophy is a relative sparing of the medial thigh subcutaneous fat compared with the lateral thigh (56% of patients with lipodystrophy compared with 6% of age- and sex-matched dermatomyositis patients without lipodystrophy) [1]. There have only been 3 cases of lipodystrophy affecting patients with adult onset dermatomyositis reported in the English medical literature [1–3]. Of these, one patient was reported to have subcutaneous fat loss in the anterior thigh, and another with severe lipoatrophy of the face, neck, arms, upper chest, and focally over the abdomen and calf. The exact pathogenetic basis of lipodystrophy in patients with dermatomyositis is unknown.

The patient in this case had a biopsy-proven history of dermatomyositis initially diagnosed in 2002 at the age of 64, when she presented with neck, chest, and eyelid rash, extremity weakness, and elevated aldolase and CPK. The biopsy was performed at another institution by her dermatologist, and the exact type of biopsy (skin, muscle, or subcutaneous) is unknown. She was treated initially with steroids, followed by regimens of methotrexate, mycophenolate, and hydroxychloroquine. Her last medication for dermatomyositis was prednisone, which she had been weaned from 2 years earlier as her symptoms had abated. She now presented with vague bilateral lower extremity

weakness and right leg swelling; she denied rash or upper extremity weakness. At her current presentation, she had normal aldolase and CPK. She had no apparent weakness on clinical examination. Given her normal aldolase and CPK values, the diagnosis of myositis as a cause of her weakness was not favored, and currently other causes of weakness are being explored, particularly peripheral vascular disease. Lipodystrophy is not associated directly with muscle weakness as the musculature is preserved.

This is a rare case of a patient with biopsy-proven adult onset dermatomyositis presenting with lipodystrophy. The diagnosis of lipodystrophy rendered by imaging turned attention from the right thigh and prompted the evaluation for associated metabolic disturbances. The patient was found to have elevated triglycerides (436 mg/dL) and hemoglobin A1c (15.9%). Currently, the patient is being treated for her hypertriglyceridemia and uncontrolled diabetes mellitus, while the cause of her weakness is undergoing further investigation.

Conflict of interest The authors declare that they have no conflict of interest.

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