

Unilateral Limb Thinning –Thinking Out of the Box

Chandramouleeswaran Venkatraman, Shubha Subramanian, Daniel Sweetson Abraham, Kannan Vellaichamy
Institute of Neurology, Madras Medical College, Chennai, Tamil Nadu, India

Abstract

We report an unusual presentation in a 9-year-old girl with unilateral circumferential thinning of the entire right upper limb without any other neurological deficit, with normal nerve conduction and electromyography initially thought of as a neurodegenerative disorder based on clinical presentation. Magnetic resonance imaging of the upper limb showed partial lipoatrophy with normal glucose metabolism and lipid profile and negativity for HIV and autoimmune disease (panniculitis) with no family history of similar disorder. Remember to think out of box before labeling neurodegenerative disease.

Keywords: Lipoatrophy, magnetic resonance imaging muscle, neurological deficit, unilateral circumferential thinning

INTRODUCTION

This case of circumferential unilateral limb thinning is presented for its rarity. In general, clinicians make a diagnosis of monomelic atrophy, a rare neurodegenerative disorder in such cases. In our case, clinical, biochemical, and electrophysiological workup did not give any clue to the diagnosis. A diagnosis of focal lipodystrophy involving the right upper limb was made by doing muscle magnetic resonance imaging (MRI), and this was later confirmed by biopsy findings. This case highlights the fact that all wasting disorders need not have a neurodegenerative etiology, and a full workup including MRI muscle may give a completely different diagnosis.

CASE REPORT

A 9-year-old girl presented with a history of progressive thinning of her right upper limb that abruptly developed and stopped spontaneously in about 3 months' time [Figure 1]. This thinning was not associated with weakness, fasciculation, pain, cramps, or sensory disturbance. Her medical history was negative for fever, rashes, drug intake, infections. There was no

history to suggest trauma or compression of the neck before the onset of thinning. Her antenatal, perinatal, and postnatal history was uneventful. She had normal developmental milestones. No member in her family is having similar complaints.

On physical examination, her right upper limb appeared hypotrophic and thinned, mimicking monomelic amyotrophy. On closer view, the skin over her right upper arm was normal, and the skin was soft and pinchable, but prominent veins were visualized when compared to the left. The thinning is limited to the right arm alone; the shoulder girdle, face, and other parts of the body are normal. Neurological examination did not reveal any deficit in all axes. The power is normal in both proximal and distal group of muscles, with well-preserved reflexes and sensation.

Initially, she was evaluated in the line of monomelic

Address for correspondence: Prof. Chandramouleeswaran Venkatraman, Institute of Neurology, Madras Medical College, Government Rajiv Gandhi General Hospital, Chennai - 600 003, Tamil Nadu, India.
E-mail: drvcnmp@gmail.com

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Figure 1: Unilateral circumferential thinning of the entire right upper limb

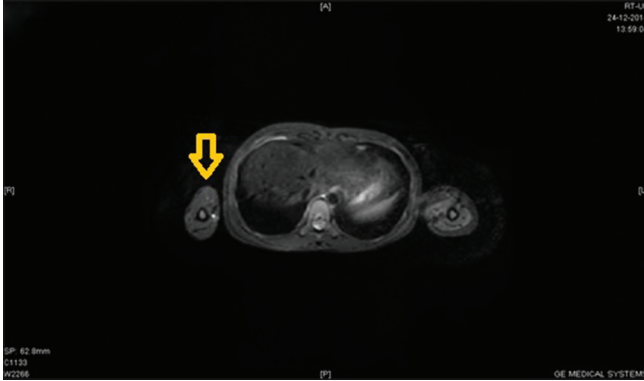


Figure 2: Magnetic resonance imaging muscle of the upper limb showed lipoatrophy of the entire upper limb

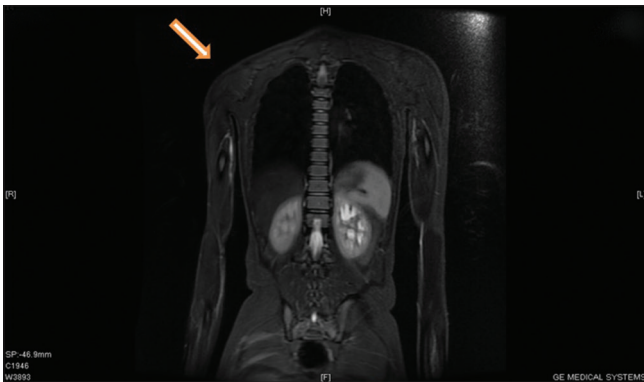


Figure 3: Magnetic resonance imaging muscle of the upper limb showed lipoatrophy of the thinning of the entire upper limb

amyotrophy. The electromyography done twice was normal. Both neutral and dynamic MRI of cervical spine were normal. Biopsy of the forearm muscle showed a neurogenic pattern of degeneration. Over 4 months, she was static and no worsening is noted in her clinical course. On re-evaluation, MRI muscle of the upper limb was done which showed lipoatrophy of the entire upper limb with preserved marrow fat [Figures 2 and 3].

On further evaluation for lipodystrophy and related conditions, her glucose metabolism, lipid profile, serum complement C3, and lupus workup were normal. HIV status was negative. Chest X-ray and MRI lung showed no restrictive lung disease. Morphea was ruled out. The clinical presentation, the laboratory investigations, and the natural history in our patient do not match the diagnostic criteria for any of the established lipodystrophy subsets.

DISCUSSION

Lipodystrophy or lipoatrophy is a rare primary idiopathic

atrophy of adipose tissue of unknown etiology.^[1] Lipodystrophy can be total, partial, or localized. Total lipodystrophy can be congenital or acquired, characterized by complete loss of adipose tissue. Usually, lipodystrophy is associated with insulin resistance, altered lipid profile, hyperglycemia, and hepatomegaly.^[2]

Acquired partial lipodystrophy (APL) is characterized by loss of facial fat with or without involvement of the arm and upper trunk. APL is a rare disease, and so far 250 cases are reported in literature.^[3] It is common among females, with the male to female ratio being 4:1, and the median age of onset is 7 years.^[3] These patients are more prone to develop autoimmune disease, especially SLE, membranoproliferative glomerulonephritis, dermatomyositis, pernicious anemia, celiac disease, and rheumatoid arthritis.^[4]

The pathophysiology of APL is unknown. The proposed mechanism of activation of alternate complement pathway leading to c3Nef-induced adipolysis, resulting in hypocomplementenemia and lipodystrophy has been postulated.^[5] Other hypothesis states autoimmune process triggered by viral infection and genetic association. Classic clinical presentation is slowly progressive fat loss spreading in a cephalocaudal distribution from the face, neck, shoulder arm, and forearm and extending to the trunk and upper abdomen. Other presentation includes isolated involvement of the limb, trunk, and upper abdomen. The hip and legs may start accumulating fat disproportionately after puberty.^[6] The onset may be precipitated by febrile illness or surgical procedure or psychological stress. Thinning is not associated with pain, but muscle weakness can accompany occasionally. It is important to look for metabolic and endocrinal abnormalities inquiring about menstrual irregularities, hirsutism, and altered glucose metabolism.^[7] Twenty percent of patients with APL are associated with renal involvement. The presence of hepatomegaly is often associated with autoimmune diseases.^[8] Physical growth and secondary sexual characteristic are usually normal. The usual site of fat loss is around the cheeks or temple, resulting in wasted appearance of the cheek and temple region. Breast may lose fat and consist of firm glandular tissue. Extremities showing prominent veins with a muscular appearance are very characteristic of APL. After puberty, the patient may show fat deposition in the abdomen mimicking Cushing's syndrome, but classic Cushingoid facies is not seen.

Diagnosis of this condition is clinical, though in rare cases, MRI of the affected limb is needed. The classic MRI finding in these patients includes loss of fat in T1 sequence, with preserved marrow fat and muscle fat. In some patients, abnormal lipid deposition occurs in the liver, hip, and thighs.^[9] Associated metabolic, autoimmune, and renal disease and HIV infections have to be ruled out.

APL has been reported to occur along with myopathy.^[9] However, our patient had neurogenic changes in muscle biopsy, and so far such neurogenic pattern has not been reported in APL. Biopsy findings (light microscopy) usually show loss of

subcutaneous fat and reduction in adipocyte volume in these types of patients. Lipocyte appears atrophic with reduced count. Atrophic area is not infiltrated by lymphocytes.^[10]

The treatment for APL is limited to cosmetic and dietary management. The medical treatment is limited to the management of associated metabolic abnormality. No effective treatment exists to halt the progression of lipodystrophy.^[11]

CONCLUSION

The cause for unilateral thinning of the limb in a young individual need not be always neurological. Lipodystrophy can be diagnosed clinically if strongly suspected, though MRI muscle is the gold standard to diagnose lipodystrophy. Hence, lipodystrophy should be actively looked for before labeling a patient as having neurodegenerative disease.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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