Calf-Head Sign in Miyoshi Myopathy

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Objective: To observe whether patients with Miyoshi-type dysferlinopathy demonstrate any distinct appearance in the back of the shoulders and upper back in a specific posture.

Design: Case series.

Setting: Neurology outpatient clinic of a north Indian tertiary care medical institute.

Patients: Fifteen patients from 9 families (10 males and 5 females; age range, 16-42 years) who had Miyoshi myopathy with onset of calf and shin muscle wasting between the ages of 9 and 28 years and a myopathic pattern at electromyography, moderately high serum creatine kinase levels, and absence of dysferlin at immunohistochemical staining.

Interventions: Patients were asked to raise their arms with shoulders abducted and elbows flexed to 90°. All were observed from behind for the morphological change in the bulk of different muscles of the upper back and shoulders. T1-weighted magnetic resonance imaging of the upper thorax and shoulder was performed in an oblique sagittal plane to confirm the clinical findings.

Main Outcome Measure: Consistent pattern of muscle enlargement or wasting seen clinically and at magnetic resonance imaging that produces a particular diagnostic appearance.

Results: A characteristic appearance was observed on the upper back and shoulders that could be described as calf heads on a trophy sign. The sign was clearly visible in 6 patients, whereas it was present in a subtle manner in another 4. Some but not all components of the sign were visible in the rest of the patients.

Conclusion: The calf heads on a trophy sign may be useful in identifying most patients with Miyoshi-type dysferlinopathy.

MIYOSHI MYOPATHY (MM) is an autosomal recessive disease that was first described in Japan1,2 and subsequently other countries. In this disease, muscular wasting and weakness start from distal parts of the lower extremities, particularly the calves. Serum creatine kinase levels may be moderately high. Results of recent studies suggest that MM and limb-girdle muscular dystrophy type 2B are allelic disorders; both diseases are linked to the same locus on chromosome 2p13-35 that includes the DYSF gene encoding the protein dysferlin.6,7 Dysferlin is a sarcolemma-bound protein8,9 involved in membrane repair.10

Clinical diagnosis of MM is difficult in the early stage despite advancement in definitive diagnostic methods such as molecular biology and muscle immunohistochemistry. The reasons for the delay in diagnosis could be the rarity of the disease and the onset from distal limb muscles. Muscular dystrophies are known for the selective involvement of skeletal muscles. This selectivity is not restricted only to weakness but extends also to wasting and enlargement. The regional topography of involved muscles has played a major role in early diagnosis, and computed tomography and magnetic resonance imaging studies have been performed in distal limb muscles to show that imaging abnormalities may precede clinical symptoms.11,12 Sometimes, one part of a muscle is enlarged and another part wasted.13 Subtle enlargement can be appreciated better when the muscle is under mild contraction as happens when a particular posture is adopted.13 Making use of these phenomena, researchers have described particular appearances in different muscular dystrophies, such as valley sign in Duchenne-Becker muscular dystrophy,14,15 poly-hill sign in facioscapulohumeral dystrophy,16 and shank sign in myotonic dystrophy.17 These signs make use of the fact that some muscles show relative enlargement or preserved bulk, whereas others show wasting in the same region. In this context, a particular appearance in the upper back and the back of the shoulders has been observed in MM-type dysferlinopathy.

REPORT OF CASES

Fifteen patients from 9 families (10 males and 5 females; age range, 16-42 years) with MM-type dysferlinopathy who had onset
of symptoms in the lower limbs between the ages of 9 and 28 years were selected for this study. The patients were selected from a neurology outpatient clinic of a north Indian tertiary care medical institute (Sanjay Gandhi Postgraduate Institute of Medical Sciences). All patients had wasting of the calf muscles; 8 had remarkable wasting of the anterior leg (shin) muscles as well. They were examined 3 to 11 years after the onset of symptoms, at a stage when they were independently walking but with difficulty in clearing the steps from the ground or getting up from a squatting position. There were no symptoms in the upper limbs in 7 patients, whereas the other 8 had mild generalized weakness in the arms, with sparing of the small muscles of the hand. At examination, 10 patients had mild wasting in the forearm muscles. The diagnosis of MM was based on gradual wasting of the leg (calf/shin) muscles, moderately high serum creatine kinase levels, myopathic pattern at electromyography, and absence of dysferlin at immunostaining of muscle biopsy specimens.

All patients were asked to raise their arms with shoulders abducted and elbows flexed to 90° as shown in Figure 1A. In this position, all patients were observed from behind for any specific appearance of the upper back and shoulders. This sign, which was described as calf heads on a trophy sign, diagrammatically shown in Figure 1B, was considered positive when 6 specific features were present. First is a prominent deltoid muscle looking like a calf's head with the mouth at the muscle's insertion and one of the several linear depressions appearing like a half-closed eye; the prominence of the calf's head was further enhanced at its lower border by slight wasting of the long head of the triceps muscle laterally and the lateral part of the infraspinatus muscle medially (the prominent posterior lower border of the deltoid muscle sometimes created a double posterior axillary fold). Second is a cordlike upper border of the trapezius muscle appearing like a horn on the calf's head; the horn stood further prominent because of wasting of the supraspinatus muscle and the overlying trapezius muscle below it. Third is a prominent upper medial part of the infraspinatus muscle with sharply demarcated wasting of the lower and lateral part, looking like a downward directed ear of the calf. Fourth is the spinous process of the scapula, which becomes prominent because of the wasted supraspinatus above it appearing like the upper border of the calf's neck with its continuation medially as a submucosal dorsal hump caused by the levator scapulae and rhomboideus minor muscles, which show a lump-like prominence at their insertion over the upper medial border of the scapula. Fifth is the ventral frill of the calf's neck formed by enlarged latissimus dorsi and teres major muscles. Sixth is the overall appearance of the upper back as that of a trophy with superolateral concave arches formed by calf horns, upper lateral projections formed by calf heads, and lower lateral convex arches formed by the ventral frill of the calf's neck. The inferior border of the trophy is formed laterally by the prominent upper borders of the latissimus dorsi muscles on either side and medially by the prominent lower borders of the trapezius muscles, which meet each other medially. These calf heads appeared prominent over the upper back because of the hollowing of the interscapular region owing to wasting of the rhomboideus major and overlying trapezius muscles.

The calf heads on a trophy sign was clearly visible (Figure 1A and Figure 2) in 6 patients, whereas it was present in a subtle manner in another 4, which included patients in both late and early stages of the disease. In the rest of the patients, some but not all components of the sign were visible, and the overall appearance did not look like the calf's head. The status of the individual components of this clinical sign is presented in the Table.

Magnetic resonance imaging of the upper thorax and left shoulder was performed with a 1.5-T superconducting magnetic resonance imaging machine (LX Echo Speed Plus; General Electric Medical Systems, Milwaukee, Wis) and a body coil. T1-weighted sagittal oblique imaging was performed by using a spin-echo sequence with the following experimental parameters: relaxation time, 500 milliseconds; echo time, 15 milliseconds; number of excitations, 4; and field of view, 340 mm. Section thickness was 5 mm with no intersection gap and a 36 × 12 matrix. The oblique sections were obtained at about a 40° angle, starting from 5 cm medial to the medial border of the scapula and ending at the lateral border of the shoulder. Four sections aptly described the status of the muscles that were involved in the clinical sign; these passed through the oblique sagittal planes 3 cm medial and lateral to the medial angle of the scapula and 3 cm medial and lateral to the acromioclavicular joint (Figure 2). Skin over the bulky deltoid muscle showed depression at those
places where fascia separating the muscle fiber bundles reached up to the periphery. Muscle fibers at the superior-most level of the trapezius muscle showed tubular enlargement that appeared circular in cross section; immediately below, the trapezius muscle showed wasting over the wasted and fibrosed supraspinatus muscle. The lower border of the trapezius muscle also showed slight enlargement, but the rhomboideus major muscle showed wasting. The bulk of the levator scapulae and infraspinatus muscles was preserved, but the teres major and latissimus dorsi muscles showed slight enlargement. These magnetic resonance imaging findings were consistent with the clinical findings.

**COMMENT**

In MM-type dysferlinopathy, personal observations have revealed thinning of the whole trapezius muscle so that the wasting or enlargement of the underlying muscles is prominent. The upper and lower borders of the trapezius muscle, however, show sparing or enlargement of muscle fibers with possible fibrosis and appear like a cord running from the occipital protuberance to the lateral part of the clavicle/acromion process and lower thoracic spine to the base of the spinous process of the scapula. The upper border of the latissimus dorsi muscle also shows prominence that runs as a cord from the midthoracic spinous process to the inferior angle of the scapula and as a bulge in the inferomedial part of the posterior axillary fold (simulating ventral skin fold of the calf's neck). These magnetic resonance imaging findings were consistent with the clinical findings.
Humeral dystrophy with high sensitivity. The shank sign the wasted biceps muscle on its lateral side, and (4) mildly distal three fourths of the deltoid muscle’s bulk relative to on its lateral side, (3) enlargement or preservation of the zontal placement and wasting of the trapezius muscle on medially and backward enlargement of the infraspinatus, deltoid, and latissimus dorsi, are wasted. The appearance at the back of the shoulder is like a valley (posterior axillary fold) between the 2 mounts (the deltoid muscle supero laterally and the infraspinatus muscle inferomedially). The valley sign is seen convincingly in about 90% of patients with Duchenne muscular dystrophy, including those showing no calf enlargement. The valley sign is also visible in adult counterpart Becker muscular dystrophy. In facioscapulohumeral dystrophy, the posture similar to the one adopted in the present study demonstrates the poly hill sign, in which 2 projections are seen over the shoulder blade (the inferior angle of the scapula projecting medially and backward enlargement of the infraspinatus muscle). Four projections are seen over the shoulder and arm: (1) the superior angle of the scapula projecting upward under the wasted trapezius muscle, (2) the acromio clavicular joint projecting laterally owing to its more horizontal placement and wasting of the trapezius muscle on its medial and proximal one fourth of the deltoid muscle on its lateral side, (3) enlargement or preservation of the distal three fourths of the deltoid muscle’s bulk relative to the wasted biceps muscle on its lateral side, and (4) mildly enlarged brachioradialis/extensor digitorum communis muscles. The poly-hill sign is specific to facioscapulohumeral dystrophy with high sensitivity. The shank sign in myotonic dystrophy type 1 shows well-preserved deltoid muscle bulk with slight enlargement of its fibers originating from the lateral part of the clavicle; slight wasting of the trapezius and supraspinatus muscles on the medial side; and the relatively preserved or slightly enlarged inferior border of the shoulder because of the preserved bulk of muscles constituting the posterior axillary fold such as the infraspinatus, teres major, teres minor, and latissimus dorsi. This characteristic bulky appearance of the shoulder is followed laterally by sudden tapering of the upper extremity near the middle of the upper arm because of wasting of all the distal muscles, which looks like the shank of an animal. The calf heads on a trophy sign was not visible in any patient with muscle disease other than MM and thus appears to be highly specific. However, its sensitivity needs to be evaluated in a larger number of patients. As the final diagnosis nowadays is made with immunohistochemical staining of muscle biopsy specimens and genetic studies, the calf heads on a trophy sign may be useful in directing patients to appropriate tests, thus minimizing the time and cost of diagnosis.

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REFERENCES