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Involuntary Painful Muscle Contractions in Satoyoshi Syndrome: A Surface Electromyographic Study

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Abstract: We report a child with Satoyoshi syndrome manifested by involuntary painful muscle contractions and alopecia. Although an autoimmune origin of Satoyoshi syndrome seems likely, its exact etiology remains as yet

unknown, as is the origin of the involuntary contractions. To gain a better understanding of the electrophysiological characteristics of the involuntary contractions, we performed a surface electromyographic (EMG) study. We investigated muscle contractions in the legs using two noninvasive techniques: high-density surface EMG (HD-sEMG) recordings on one muscle, and polymyographic surface EMG (sEMG) recordings on various muscles. During the involuntary contractions, HD-sEMG showed a fourfold increase in amplitude compared to maximal voluntary contractions. These high potentials were widely distributed across the whole muscle and showed a pronounced oscillatory behavior with a frequency around 45 Hz. Polymyographic sEMG revealed that the involuntary contractions often occur simultaneously in various muscles or showed a switch of activity from one muscle to another. These findings point to hyperactivity or a disinhibition at the alpha motor neuron level, originating probably at that level, although a central origin cannot be excluded. © 2006 Movement Disorder Society

Key words: Satoyoshi syndrome; high-density surface EMG; involuntary muscle contractions

Satoyoshi syndrome is a childhood-onset progressive multisystem disorder characterized by intermittent painful muscle contractions, malabsorption, diarrhea, endocrinopathy, alopecia, and secondary skeletal abnormalities.¹ Its etiology is unknown, although an autoimmune basis is likely through association with other autoimmune conditions, the presence of antibodies, and the improvement of symptoms with corticosteroid treatment and intravenous immunoglobulin.^{2,3} The painful contractions usually begin in the legs during childhood, and progress slowly with increasing severity and intensity, also involving limbs, trunk, neck, and masticatory muscles. Muscle contractions generally last for a few minutes and usually recur after an interval of less than a minute. These contractions are so intense that they induce abnormal posturing of affected limbs. Satoyoshi reported that the more severe the muscle contractions are and the earlier the disease onset in these patients is, the more severe the growth disturbance and the joint deformity are.⁵

The relation between the presumed autoimmune origin and the involuntary muscle contractions currently is still unclear. The answer to the question whether the painful muscle contractions in Satoyoshi syndrome have a peripheral (like muscle cramps) or central origin (like muscle spasms) would provide a clue to the pathophysiology and the cause of this rare syndrome.

Muscle cramp is a clinical diagnosis. It is characterized by a sudden, uncomfortable squeezing or contraction, lasting for seconds to minutes, often with a palpable hard knot in the affected muscle. Stretching the muscle

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or contraction of its antagonist muscle speeds relief of the cramp.⁶ Cramp is limited to a single muscle. The exact site of origin of muscle cramp is still a matter of debate. A surface electromyographic (EMG) technique, with multiple electrodes on one muscle, recently has been applied to gain better insights into its pathophysiology. It has been demonstrated that muscle cramps present themselves with a characteristic pattern of a slowly moving fraction of contracting muscle fibers in a single muscle, indicating that cramp spreads through neighboring groups of muscle fibers.⁷ A recently developed surface EMG technique, with multiple surface EMG electrodes in a single grid, named high-density surface EMG technique (HD-sEMG), allows simultaneous recordings from various locations across a muscle, providing both temporal and spatial information about motor unit activity.^{8,9}

Muscle spasms are ill-defined. They form part of a spastic syndrome that also includes exaggerated tendon reflexes and muscle tone.¹⁰ Zijdwind and Thomas showed that MU firing rates of the thenar muscles that have been weakened in patients with chronic spinal cord injury were low during both submaximal and maximal voluntary contractions.¹¹ During spasms in patients with chronic cervical spinal cord injury, motor unit firing rates either increased and then decreased with the spasm intensity or were relatively constant. Mean peak spasm firing rates were 18 ± 9 Hz (mean $\pm$ SD) for the rate-modulated units and 11 ± 10 Hz for units with little or no rate modulation.¹² Because muscle spasms are typically not limited to a single muscle, we also recorded polymyographic surface EMG (sEMG) patterns in various muscles simultaneously with conventional sEMG techniques.

Here, we report a child with Satoyoshi syndrome in which HD-sEMG and polymyographic sEMG, were performed to gain a better understanding of the pathophysiology of the involuntary muscle contractions in Satoyoshi syndrome.

CASE REPORT

A 6-year-old girl of Turkish origin suffered from exercise-induced, progressive painful muscle contractions and thinning hair. Apart from abnormal muscle contractions and a short stature (body length below 3 SD), general and neurological examination revealed no abnormalities. Laboratory examination revealed a serum creatine kinase of 296 U/l (normal value < 200 U/L), the presence of anti-nuclear antibodies and a transient anemia. Endocrine investigation (thyroid gland, pituitary function, urine steroid profile, serum, and urine glucose) was unremarkable. Radiological investigations showed

no skeletal deformities. Quadriceps muscle biopsy was normal, as were immunohistochemistry (phosphorylase, phosphofructokinase) and mitochondrial respiratory chain enzyme activities. Hair root analysis showed a high percentage of catagen follicles. Standard nerve conduction and needle EMG (not during involuntary contractions) showed normal values. Satoyoshi syndrome was diagnosed, and therapy was started with prednisone, 1 mg/kg per day for 3 months. The painful muscle contractions completely disappeared during therapy. Hair growth was not restored.

Surface EMG Methods

EMG measurements were performed on the child's leg, where the involuntary muscle contractions were most pronounced. First, HD-sEMG recordings of the left vastus lateralis muscle were recorded during maximal voluntary contraction and during the painful muscle contractions. HD-sEMG allows multichannel recordings of a single muscle and is a noninvasive tool to record the distributions of muscle activation over a larger area (6×4.5 cm) of a muscle.^{8,9,13} The electrode grid used consisted of 130 gold-coated (13×10) electrodes with an interelectrode distance of 5 mm in both directions. The electrode grid was carefully placed over the vastus lateralis muscle with its columns of 13 electrodes parallel to the muscle fibers. All signals were amplified, band-pass filtered, and simultaneously AD converted (at 2,000 samples/sec) with a multichannel amplifier system.^{8,13} The signals were analyzed off-line and displayed in a bipolar montage for visual analysis.

In addition, muscle activity of different muscles were recorded simultaneously with bipolar polymyographic surface EMG recordings on different muscles of both legs, using Ag/AgCl electrode pairs at a 3-cm interelectrode distance. On both legs, the quadriceps muscles, the tibialis anterior muscles and the gastrocnemius muscles were investigated.

RESULTS

HD-sEMG recordings of the left vastus lateralis muscle during maximum voluntary contractions and during painful involuntary muscle contractions are shown in Figure 1A and B, respectively. During maximal voluntary contractions, the HD-sEMG recording showed a normal interference pattern with motor unit action potential amplitudes of approximately 50 μ V. During the involuntary contractions, bipolar HD-sEMG recordings showed a regular pattern, most probably of synchronized motor unit firings with a frequency of 40 to 50 Hz, resulting in much higher amplitudes (over 200 μ V) than in the maximal voluntary contractions. We observed that

FIG. 1. A–B: High-density surface electromyographic (EMG) recordings of the left vastus lateralis muscle show high-density EMG recordings of the Satoyoshi patient. One column of the grid (12 bipolar montages from 13 monopolar electrodes in a grid column positioned in fiber direction) with the clearest presentation of the phenomenon of muscle-wide synchronous oscillatory activity (Part B) is displayed. The x axes indicate time in seconds, and the y axes indicate 12 bipolar recordings derived from 13 electrodes. A: High-density surface electromyographic (HD-sEMG) during maximal voluntary contraction. The sEMG shows a normal motor unit interference pattern. B: Recording from the same area during painful contraction shows widely spread rhythmic EMG activity with a much higher amplitude (note the different gain settings) and an oscillatory frequency of approximately 45 Hz. C–D: Conventional bipolar polymyographic sEMG recordings of three different muscles of both legs: the first three traces of the right leg and the lower three traces of the left leg. C and D are recordings during the involuntary contractions of the Satoyoshi patient at different time points. The polymyographic recordings show simultaneous EMG activity in both right and left quadriceps muscles (C), and in different muscles (right quadriceps muscle and right tibialis anterior muscle) of the same leg (D).

these high-amplitude EMG signals were present with rather constant amplitudes over a substantial part of the vastus lateralis muscle. It is important to note that there were no signs of slow spread of local EMG activity in the muscle as found with multichannel EMG in muscle cramps.⁷

Conventional polymyographic sEMG recordings showed simultaneous activity both of various muscles of the same leg, and of muscles of both legs during the involuntary contractions (Fig. 1C and D, respectively). In the course of the involuntary contractions, there was a switch over of EMG activity from one muscle to another, with a “waxing and waning” character (Fig. 1C).

DISCUSSION

The EMG findings of this study strongly support the concept that the involuntary muscle contractions in Satoyoshi syndrome do not fit into the dichotomy between

muscle cramps and spasms in a “classic sense.” We will give reasons why another mechanism seems likely.

There are several arguments against the mass activity found in this patient being a muscle cramp. First, our HD-sEMG recording during the involuntary contractions showed an over fourfold increase in amplitudes of the EMG activity compared to the amplitudes recorded during maximal voluntary contraction and a regular firing frequency of 40 to 50 Hz. The synchronous regular firings and the high amplitude during the involuntary contractions suggest strictly synchronous repetitive active motor unit discharges, which have not been found in cramps.^{7,14} Second, the motor unit action potentials in the vastus lateralis muscle during the involuntary contractions were present in a large part of the muscle, which is in contrast to what has been found in muscle cramps, in which only a (slowly shifting) fraction of the

muscle is involved.⁷ Third, the discharge frequency of the EMG activity during the involuntary contractions was 40 to 50 Hz. Discharge rates in cramps are typically around 150 Hz.⁶ Last but not least, our polymyographic recordings showed that massive electric muscle activity was not limited to one muscle. It extended over different leg muscles, even more or less simultaneously in both legs. The latter directed us to consider this mass activity as muscle spasms.

As already stated, spasms are ill defined.¹⁰ That there were no clinical signs of pyramidal tract lesion provides an important argument that the involuntary painful contractions in this patient are not "conventional" spasms. In healthy subjects, motor units usually fire at 6 to 10 Hz when first recruited and at 15 to 60 Hz during maximal voluntary contractions.¹⁵ Thomas and Ross showed with needle EMG studies of motor unit activity in patients after spinal cord injury that, during spasms, the motor unit firing frequencies show mean peak firing rates of 18 ± 9 Hz.¹² Rate coding for many motor units appears to be similar whether descending motor input is intact or whether it has been reduced severely by spinal cord injury.

In 1978, Satoyoshi already described that, during the involuntary contractions in his patients, needle EMG recordings revealed synchronized motor unit discharges of 40 to 50 Hz and of 4 to 10 mV amplitude. He suggested that abnormal discharge of anterior horn cells is responsible for the mass activity.¹ The HD-sEMG recordings showed a stable spatial extension of such synchronized activity throughout the vastus lateralis muscle during the contractions. This finding supports and provides additional arguments for this hypothesis of massive hyperactivity or disinhibition at the alpha motor neuron level. In conclusion, although a more proximal origin cannot be excluded, the surface EMG findings strongly suggest a deregulation at the alpha motor neuron level leading to involuntary muscle contractions in Satoyoshi syndrome.

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Alzheimer's Disease Presenting as Corticobasal Syndrome

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Video

Abstract: A 60-year-old man presented with slowly progressive left hemi-Parkinsonism, left hand apraxia, myoclonus, dystonia, visuospatial disturbances, and alien limb phenomenon, resembling corticobasal syndrome. Eight years later,

This article includes Supplementary Video, available online at <http://www.interscience.wiley.com/jpages/0885-3185/suppmat>

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