Facial Myokymia in Multiple System Atrophy

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Summary: Two patients are described with clinical and neuroimaging features consistent with a diagnosis of multiple system atrophy (MSA). The patients are unusual in that facial myokymia became apparent clinically at some stage in their illness. In each patient, the nature and severity of the involuntary facial movements evolved over the course of the illness. Electrophysiologically the movement pattern was consistent with myokymia, and studies of blink-reflex responses suggested that the myokymic discharges were of brainstem origin. Involuntary facial movements described as facial action myoclonus with electrical characteristics consistent with myoclonus have been described previously in hereditary olivopontocerebellar atrophy (OPCA). Our report describes electrical and clinical features of facial myokymia in MSA with electrical features suggesting hyperexcitability of the facial motoneurons in the brainstem. Such myokymic movements may occur more frequently in MSA than previously recognized but may be missed clinically because of their evolving nature. Key Words: MSA—Myokymia—Blink reflex.

Olivopontocerebellar atrophy (OPCA) was the description used by Dejerine and Thomas (1900) (1,2) to denote a disorder seen in two middle-aged patients with an apparently sporadic progressive disorder of the cerebellum. The chief clinical features were broad-based, short-stepped gait, slowness and hesitation of body movements, clumsiness of the hands, action tremor, and slow-scanning speech. This sporadic condition was distinguished from a group of autosomal dominant cases reported by Menzel [see (3)], who had an earlier age of onset and chorea in the early stages of the illness.

Classification of OPCA conditions has proved difficult because of heterogeneity not only in apparently sporadic cases, but also between individuals of the same kindred in familial cases. Furthermore, the familial OPCA syndrome may encompass a number of different disorders including autosomal dominant or recessive forms, late onset cerebellar ataxia (4), and some examples of Joseph disease (5). Computed tomographic (CT) brain appearances may be helpful in making the diagnosis, but a recent study showed little correlation with the clinical or pathologic picture (6).

Sporadic OPCA is today included as one type of multiple system atrophy (MSA) (7). Additional clinical features have since been added to those classically accepted for the syndromes of familial or sporadic OPCA, as described previously. These include spasticity, dementia, chorea, parkinsonism, myoclonic jerks, retinal degeneration, optic atrophy, dysautonomia, and amyotrophy (8). Recently electrophysiologic features of facial myokymia (without concurrent involuntary facial movement) have been reported in familial OPCA, together with the appearance of involuntary facial movements, which have been described as "facial action myoclonus" (9,10).

We describe two patients with MSA who displayed clinically evident facial myokymia at some stage during their illness. Electromyogram (EMG) studies confirmed characteristic myokymic discharges both at rest and on voluntary movements of the facial musculature. The blink-reflex recovery cycle was enhanced, suggesting that intrinsic brainstem hyperexcitability of the facial nucleus motoneurons may be responsible for these involuntary facial muscle contractions.

CASE 1

A man of Afro-Caribbean origin was first seen in an ENT department in 1992 at the age of 53, with a 2-year history of imbalance and vertigo and a tendency to stagger to one side. He had had a vague feeling of weakness for 36 months. No otologic cause for his symptoms was found, and he was referred to the neurology department. We obtained a 2-year history of a feeling of being in a "semidrunken state." He had had an increasing tendency to fall. He was no longer able to run and felt dizzy when rising out of bed. He complained of unusual occipital headaches. His legs felt weak, and he had a periodic tremor of the right hand. He had noticed a deterioration in his writing. He had become impotent without loss of libido and had developed constipation and increased frequency of micturition. There was no family history of neurologic disease.
Initial examination revealed a postural decrease in blood pressure (BP) from 155/100 to 13/95 mm Hg. He walked with a wide-based gait, with great difficulty turning, and was unable to heel-toe walk. He had a strongly positive glabellar tap, present only on the right. He had an impassive facies with increased nuchal tone. The optic discs were pale, and he had broken pursuit eye movements. The right arm showed cogwheel rigidity but no bradykinesia or incoordination. There was an action tremor of the right arm. Tone was mildly increased in the legs, and there was marked heel-shin ataxia. Reflexes were brisk with flexor plantars. Over the subsequent months, his symptoms and signs progressed. Nystagmus developed on right lateral gaze. His postural hypotension became more marked, and he was given elastic stockings.

**Myokymic Movements**

Over the first year of observation in the neurology clinic, he was noted to develop spontaneous fine myokymic movements initially above and below the left side of the mouth. These had not been apparent on his earlier visits. He was noted to have tremulous facial muscles when he talked and on jaw opening. Four months after these movements first appeared, further spontaneous myokymic movements were seen below the right and later the left eye. These movements varied in intensity and distribution between visits and, over recent months, have tended to become less evident. There is no evidence that facial movements have become weaker. On his most recent visit to the clinic, ~3 years after first presentation, the myokymic movements were barely visible around the mouth, and only just visible below the right eye (video). A magnetic resonance imaging (MRI) scan of the brain performed 6 months after presentation showed atrophy of the cerebellar vermis.

**EMG Studies Case 1**

**Right Orbicularis Oculi**

Needle electrode studies of right orbicularis oculi revealed single motor unit activity firing at ~10 Hz at rest. During movement, a full interference pattern was seen and some discrete EMG bursts of higher amplitude noted. When the patient was asked to stop movement, discrete bursts of EMG activity composed of multiple motor units (multiplets) were seen. The frequency within bursts exceeded 100 Hz. An example of the myokymic pattern of discharge seen in this patient is shown in Fig. 1.

**Right Orbicularis Oris**

Needle electrode studies of the right orbicularis oris showed no activity at rest. Motor unit activity during movement was normal with normal recruitment and full interference pattern. When movement stopped, activity vanished but not completely: a single motor unit activity was seen for a few seconds.

**Blink Reflexes**

With single stimuli, the latencies and amplitudes of R1, and ipsilateral and contralateral R2 components were normal. The recovery cycle of the blink reflex was enhanced (Table 1). The excitability of the blink reflex was assessed by using paired stimuli at interstimulus intervals of 100, 200, 300, and 500 ms to study the recovery cycle of the R2 component. Normally, marked suppression of R2 component is seen at these intervals. In this patient, the amplitude of the R2 component was not suppressed, indicating an enhanced reflex recovery cycle.

**CASE 2**

A 52-year-old man was seen with an 18-month history of unsteadiness and a feeling that his legs "had a mind of their own." He had difficulty going downstairs and changing direction when walking. His legs tended to jerk and shake uncontrollably when he attempted to put on socks or wash his feet. He had occasional falls with a tendency to fall backward. He had generally slowed down. There was no family history of neurologic disease, and he was of English descent.

Examination revealed a generally tremulous-looking man. Nuchal tone was increased, and he walked with a wide-based, unsteady gait. Postural reflexes and arm swing were normal. His forehead was furrowed. The glabellar tap was strongly positive. The left pupil was larger than the right, but both reacted normally. Pursuit eye movements were broken, but there was a full range of eye movements. There was cogwheel rigidity in both arms, no tremor, full power, and normal sensation. There was no wasting. Fine finger movements were impaired. Tone was increased in the legs, but power was full. However, attempted movements of the legs was accompanied by marked violent jerking of the legs with variable amplitude; this shaking also could be induced if the patient held his legs flexed in certain positions. There was a stocking distribution loss to pin prick. Reflexes were normal. There was marked truncal and heel-shin ataxia and mild finger–nose intention tremor.

**FIG. 1.** Example of myokymic discharges from patient 1. The discharges consist of singlets and multiplets from orbicularis oculi, induced by movement. The frequency between bursts is ~10 Hz. Calibration: 100 ms, 100 μV.
TABLE 1. Blink reflexes showing latencies and amplitudes of R1 and ipsilateral and contralateral R2 components

<table>
<thead>
<tr>
<th>Case</th>
<th>R1 lat. amp.</th>
<th>Ips. R2 lat. amp.</th>
<th>Contra. R2 lat. amp.</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Stimulation</td>
<td>R supraorb. nerve</td>
<td>10 ms/50 ( \mu \text{V} )</td>
</tr>
<tr>
<td>2</td>
<td>Stimulation</td>
<td>R supraorb. nerve</td>
<td>10 ms/65 ( \mu \text{V} )</td>
</tr>
</tbody>
</table>

Lat., latency; amp., amplitude; Ips., ipsilateral; Contra., contralateral; supra-orb., supraorbital.

**Myokymic Movements**

Examination of the face at rest on his first visit revealed fine rippling movements of the facial muscles around both eyes (left more than right) and both sides of the mouth, with the appearance of myokymia. These flickering movements varied greatly in frequency and severity, and the patient was not aware of the finer movements. With the passage of time, the myokymic movements in the upper part of the face (below the eyes) became less obvious, whereas twitching movements of a much coarser nature began to appear in the lower part of the face bilaterally. When the patient spoke or smiled, the facial contractions became much more obvious and sustained (see video). CT scan of the brain showed marked atrophy of the pons and cerebellum.

**EMG Studies Case 2**

**Right Orbicularis Oculi**

Needle electrode examination revealed EMG bursts firing several times per second (~5 Hz). Each burst consisted of multiletts, and the frequency within bursts exceeded 120 Hz. These were present at rest and enhanced by movement.

**Right Orbicularis Oris**

Singlet discharges were seen at rest, firing at variable frequency. Some EMG bursts appeared after voluntary movements had ceased.

**Blink Reflexes**

The latencies of R1 and ipsilateral and contralateral R2 components were normal. The amplitude of R2 component was increased. With paired stimuli, the recovery cycle of R2 component of the blink reflex was enhanced. An example of the enhanced blink-recovery cycle of R2 component from case 2 is shown in Fig. 2.

**Summary of EMG Findings for Both Cases**

The abnormal electrical discharges have the characteristics of myokymia. These discharges were seen at rest (spontaneous), both together with and in the absence of clinically evident facial myokymia. Voluntary facial movements triggered enhanced myokymic movements, which were associated with enhanced myokymic discharges electrically. The increased amplitude and enhanced blink reflex recovery of the R2 component suggest hyperexcitability of the intrinsic brainstem neuronal network mediating the R2 component. Intrinsic brainstem hyperexcitability may be responsible for the facial myokymic discharges.

**DISCUSSION**

We describe two patients who fulfilled the clinical criteria of MSA, but who had the additional feature of facial myokymia at rest, with more obvious facial muscle contractions or "tremu-
lousness” precipitated by voluntary facial movements. In both patients, EMG features of myokymia were seen, with enhanced blink-reflex recovery cycles, suggestive of hyperexcitability of the intrinsic brainstem neuronal network mediating this reflex (10,11).

The presence of clinically detectable facial myokymia in MSA has not previously been described as such, although it may be that the recent description given by Lou et al. (9), which was termed “facial action myoklonus,” may be the same as the movements we have seen in our patients. Schut (12) described one patient with “hereditary cerebellar ataxia” who had a “constant coarse fasciculation of the facial musculature, the amplitude of which was increased on grimacing so as almost to constitute a tremor.” Later review of that patient showed that the facial movements had vanished, but fasciculations were still seen in the tongue. Schut described a second patient who had fine fasciculations at the angle of the mouth with an associated facial paresis, together with fasciculations of the tongue. Facial fasciculations at rest have also been described in Azorean disease of the nervous system (13), a progressive inherited neurologic disease characterised by gait ataxia, parkinsonism, impaired eye movements, widespread fasciculations, loss of limb reflexes, cerebellar tremor, and extensor plantars. Two of the patients described by Romanul et al. (13) showed an action-induced “tremor” of the perioral muscles, comparable to that seen in our case 2 on jaw opening. Other families of non-Azorean descent have been described who had the additional finding of facial and lingual fasciculations (14,15).

Based on the two patients presented here, we suggest that clinically obvious facial myokymia may be a feature of MSA. The EMG features seen in both patients are typical of myokymia, occurring spontaneously both with and without corresponding facial movements. Voluntary facial movements were associated with an exaggerated myokymia or tremulousness of facial musculature, which was associated with enhanced myokymic discharges electrically. The enhanced blink-reflex responses point to hyperexcitability of facial nucleus motor neurones in the brainstem as the cause of the movements, as in the familial cases described (9,10). Previous studies have suggested that facial myokymia is due to interruption of the supranuclear inputs to the facial nucleus (16,17). In both patients, the facial movements were bilateral (but not symmetrical), distinguishable clinically from hemifacial spasm; they occurred at rest (distinguishing them from action myoklonus) (9) but were increased by movement. From electrical and clinical evidence, we believe the movements seen in these two patients can most accurately be described as myokymic.

The facial muscle contractions in both of these patients, and seemingly also in others with hereditary conditions (10), changed both in distribution and severity with time. This is further evidence that these movements reflect a progressive brainstem pathologic condition. Indeed, in our case 2, by the time of preparation of the video for this article, the facial myokymia had decreased considerably compared with his examination 1 year earlier. We suggest that, perhaps, due to its evolving nature, facial myokymia may be missed in patients with MSA, and may be more common than has previously been recognised.

**LEGENDS TO VIDEOTAPE**

**Case 1.** This video was taken ~3 years after the patient's initial presentation to us. The facial myokymia—which was at one period a prominent feature of his condition—has now all but disappeared. Faint myokymic movements are just visible below the right eye and seem to be triggered by eye closure. No myokymia was present around the mouth on the day of recording.

**Case 2.** In contrast to the first patient, this patient of whom the video was made ~18 months into his illness, continues to show a number of abnormal involuntary movements. First, the whole body can be seen to be jolting irregularly. This was due to large-amplitude irregular jerking of both legs. Second, he shows frequent blinking of both eyes. Finally, around the mouth there are coarse myokymic and more sustained twitching movements, particularly noticeable below the right and left angles of the mouth and over the chin. These movements occur particularly after voluntary movement of the mouth or face and can continue for several seconds.

**REFERENCES**


