Hemifacial spasm and involuntary facial movements

N.-C. TAN, L.-L. CHAN¹ and E.-K. TAN²

From the SingHealth Polyclinics-Pasir Ris, and Departments of ¹Diagnostic Radiology and ²Neurology, Singapore General Hospital, Singapore

Received 13 December 2001 and in revised form 7 March 2002

Summary

Hemifacial spasm (HFS) is characterized by tonic and clonic contractions of the muscles innervated by the ipsilateral facial nerve. It is important to distinguish this from other causes of facial spasms, such as psychogenic facial spasm, facial tic, facial myokymia, blepharospasm, and tardive dyskinesia. Magnetic resonance imaging and angiography studies frequently demonstrate vascular compression of the root exit zone of the facial nerve. Importantly, an underlying space-occupying lesion needs to be excluded in patients with associated atypical features such as facial numbness and weakness. Botulinum toxin injection to the facial muscles is an effective treatment for HFS, with few disabling side-effects.

Introduction

Hemifacial spasm (HFS) is characterized by tonic and clonic contractions of the muscles innervated by the ipsilateral facial nerve.¹ It must be differentiated from other causes of involuntary facial movements (Table 1), all of which can potentially lead to social embarrassment and affect quality of life. As facial twitches are frequently attributed to stress and anxiety, the diagnosis of HFS may be missed. Early recognition is important, as it enables institution of appropriate therapy. We summarize the aetiology, clinical features and treatment options in HFS, and discuss the differential diagnosis of involuntary facial movements frequently encountered in clinical practice.

Epidemiology

There is generally a paucity of epidemiological data on HFS. The average age-adjusted annual incidence of HFS for all ages was 0.78/100 000 (0.81/100 000 in women and 0.74/100 000 in men) in a study in Olmstead County in Minnesota.² The prevalence was 14.5/100 000 in women and 7.4/100 000 in men, indicating that it affects predominantly women. The prevalence of HFS appears to be more common in some Asian populations than in Caucasians, but there have been no epidemiological studies in these populations to support this observation. Most HFS cases are sporadic, though occasional familial cases have been described,³–⁵ suggesting that some patients may be genetically predisposed to developing HFS.

Clinical features

Patients with HFS usually present at between 40 and 50 years of age.¹,⁶,⁷ They frequently complain of involuntary eye closure, which interferes with
Table 1  Differential diagnosis of involuntary facial movements

<table>
<thead>
<tr>
<th>Contraction</th>
<th>HFS</th>
<th>Facial myokymia</th>
<th>Psychogenic facial spasm</th>
<th>Blepharospasm and Meige syndrome</th>
<th>Tic</th>
<th>Tardive dyskinesia</th>
</tr>
</thead>
<tbody>
<tr>
<td>Nature of contraction</td>
<td>Intermittent clonic or tonic contraction of muscles supplied by facial nerve. Muscles are relaxed in between contractions.</td>
<td>Undulating movement of facial muscles</td>
<td>Intermittent or constant contraction of facial muscles</td>
<td>Dystonic movement of orbicularis oculi muscle only. Meige syndrome includes blepharospasm and dystonic movement of facial muscles. Often involves muscles not innervated by facial nerve</td>
<td>Rapid stereotyped movements that resemble normal coordinated movement. May involve muscles not supplied by the facial nerve</td>
<td>Choretic or dystonic movement of muscles. May involve muscles not supplied by facial nerve</td>
</tr>
<tr>
<td>Site of involvement</td>
<td>Usually unilateral</td>
<td>Commonly involves eyelids</td>
<td>Face and any body region</td>
<td>Usually bilateral</td>
<td>Commonly involves the face and limbs</td>
<td>Commonly involves the oro-facial region</td>
</tr>
<tr>
<td>Aggravating and relieving factors</td>
<td>Increased by voluntary facial movement, stress, fatigue, anxiety or change in head position. Persists in sleep</td>
<td>Increased by stress, anxiety, fatigue</td>
<td>Increased by stress, anxiety, fatigue, relieved by placebo treatment</td>
<td>Increased by stress, anxiety, fatigue, improved during sleep</td>
<td>Able to voluntarily reproduce and to transiently suppress the movement</td>
<td>Increased by stress, anxiety, fatigue</td>
</tr>
</tbody>
</table>

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vision and causes social embarrassment. In a large series of 158 HFS patients, the initial site of onset was the orbicularis oculi muscle in 90%, the cheek in 11% and the perioral region in <10% of cases. Similar findings have been reported in other series. Over months to years, the spasms spread gradually to other muscles innervated by the ipsilateral facial nerve in a synchronous manner. Bilateral HFS is occasionally reported, its prevalence in clinic-based series varying between 0.6 and 5%. When bilateral, the second side becomes involved after a long interval and the movements on each side are usually asynchronous.

The facial spasms are spontaneous and may persist during sleep. Symptoms are frequently aggravated by stress, fatigue, anxiety, and voluntary facial movements. Relaxation, alcohol intake, touching the affected areas, and exercise reportedly improve symptoms in some patients. Low-pitched tinnitus in the ipsilateral ear may occasionally be present, and is thought to be due to a stapedius muscle contraction which accompanies facial muscles movements. In one series, 13% of patients reported unilateral or bilateral hearing loss, which did not appear to correlate with the side or severity of HFS. Concomitant trigeminal neuralgia, though uncommon, has been reported in HFS. HFS is a chronic disease, with spontaneous resolution in <10% of patients. Clinically obvious facial weakness may be seen in long-standing cases.

Aetiology

The aetiology of HFS has puzzled investigators for many years. In 1947, Campbell and Keedy suggested that vascular abnormalities in the posterior fossa might be associated with HFS. This was supported by surgical posterior fossa explorations in HFS patients. With the advent of advanced imaging and improved surgical techniques, vascular compression of the facial nerve by an ectatic vessel has been demonstrated to be the most common underlying aetiology of HFS. The vascular abnormality is usually an atherosclerotic aberrant or ectatic intracranial artery, most commonly the anterior or posterior cerebellar artery or the vertebral artery. The introduction of magnetic resonance imaging (MRI) and magnetic resonance angiography (MRA) has improved the detection of neurovascular conflict (vascular contact and/or compression of the root exit zone of the facial
nerve).\textsuperscript{16–18} High-resolution MRI and MRA techniques highly sensitive for neurovascular conflict are frequently needed.\textsuperscript{17,18} However, neurovascular conflict can be identified in up to 25\% of controls, suggesting that neurovascular conflict alone is insufficient to produce HFS.\textsuperscript{16–18} In a study of 34 patients with HFS, MRI and MRA detected vascular abnormalities in 88\% of HFS patients.\textsuperscript{18} All vascular abnormalities were ipsilateral to the side of the HFS. Only 3/12 controls (25\%) had a vascular abnormality in both MRI and MRA studies. One of the nine HFS patients (11.1\%) who had MRI only had an ipsilateral vascular abnormality. It has been suggested that hypertension may be a risk factor for HFS, as elevated blood pressure may cause atherosclerosis and hence give rise to ectatic vessels and subsequent compression of the facial nerve. Alternatively, compression of the ventral-lateral medulla by an ectatic vessel may cause hypertension. However, the cause and effect of hypertension in HFS has not been clarified.\textsuperscript{19} Large-scale prospective studies are needed to address these issues. A review of the literature suggests that an underlying space occupying lesion such as tumour causing HFS is not common.\textsuperscript{15} Various types of intracranial tumours (e.g. epidermoid, meningioma, lipoma) have been associated with HFS.

There is electrophysiological evidence that compression of the nerve at the root exit zone is responsible for HFS. Gardner in 1968\textsuperscript{20} and later Nielsen\textsuperscript{21,22} in 1984 proposed the theory of ‘ectopic’ or ‘ephaptic’ transmission. Nerve compression and the resulting demyelination cause a ‘false’ synapse at which ectopic activity may be triggered by mechanical irritation or flow of extracellular current during passage of nerve impulses in adjacent nerves. Nielsen demonstrated that in HFS, stimulation of the zygomatic branch of the compressed facial nerve results in the expected response in the orbicularis oculi muscle but also a simultaneous response in the mentalis muscle.

\textbf{Figure 3.} Male, 53 years old, with left-sided HFS. Vascular loop identified as the left posterior inferior cerebellar artery (white arrowhead) on a coronal maximum intensity projection of MR angiography (B: basilar artery, V: left vertebral artery).

\textbf{Figure 4.} Male, 53 years old, with left-sided HFS. Solid line demonstrates plane of the left facial nerve at the cerebellopontine angle (MRI).

\textbf{Figure 5.} Male, 53 years old, with left-sided HFS. Oblique coronal reconstruction along the plane of the left facial nerve (see Figure 4), showing gross indentation of the inferior-lateral surface of the left pons by the vascular loop (arrow).
which is supplied by the mandibular branch. This phenomenon is absent in controls, and is resolved after surgical decompression of the facial nerve.\textsuperscript{22} Other investigators examined intraoperative intra-cranial recordings of the facial muscles and the facial nerve near its entrance into the brain stem in HFS patients.\textsuperscript{23} Their findings suggest that HFS caused by injury of the facial nerve may be a result of reverberant activity in the facial motonucleus, possibly caused by mechanisms that are similar to kindling.\textsuperscript{23}

**Differential diagnosis**

Involuntary facial movements are not uncommonly encountered in the general population. As facial twitches are frequently attributed to stress and anxiety, the diagnosis of HFS may be missed. Other aetiological causes of involuntary facial movements such as tardive dyskinesias, myokymia, tics, cranial dystonia, and psychogenic facial spasm must be differentiated from HFS, as early diagnosis allows institution of appropriate treatment (Table 1).

Facial myokymia is manifested clinically by involuntary undulating movements of the facial muscles. The eyelids are frequently involved. Facial tics may affect the facial muscles but other body regions are commonly involved. These movements may be preceded by premonitory symptoms, and are quick and stereotypical, but frequently vary in intensity, and alternate between left and right sides. Tics may be partially suppressible. Patients suffering from Tourette’s syndrome have both motor and vocal tics associated with behavioural symptoms. In blepharospasm, there is bilateral, frequently symmetrical and synchronous contractions of the orbicularis oculi. The frontalis and corrugator muscles as well as other facial muscles may also be affected. Blepharospasm may be preceded by frequent blinking. Choreiform movements in the face are usually random and non repetitive. Careful examination may reveal presence of choreoathetosis in other body regions. For instance, patients with Huntington’s disease present with cognitive impairment and generalized choreoathetosis. Choreiform movements may be observed in the frontalis and other facial muscles in these patients.

Oromandibular dystonia refers to sustained and repetitive muscle contractions affecting the lower face, jaw, tongue, pharynx and mouth. Jaw-closing dystonia is the most common, and this may be associated with bruxism.\textsuperscript{24} Unlike HFS, the eyelids are not involved. Patients who have been exposed to neuroleptic agents (such as haloperidol) may develop stereotypical movements of the face, neck, trunk and limbs called tardive dyskinesia.\textsuperscript{25} Actions such as ‘marching in space’, ‘truncal rocking’, ‘facial grimacing’ and ‘tongue protrusion’ are characteristic features. Because the orofacial-lingual regions are frequently affected, tardive dyskinesias have to be distinguished from HFS. Focal seizures involving one side of the facial muscles frequently progress to other body regions such as the neck and limbs. Electroencephalography may be useful in supporting the diagnosis.

Psychogenic causes of facial movements can mimic HFS, and may lead to unnecessary treatment. However, these appear to be uncommon in the Caucasian population. In one study, psychogenic facial spasm constituted only 2.4\% of 210 consecutive patients evaluated for HFS in a large American movement disorder clinic.\textsuperscript{6} Psychogenic HFS is characterized by non-patterned facial movements that frequently vary in intensity and frequency, and are distractible.\textsuperscript{6} However, psychological components superimposing on an underlying organic HFS may create diagnostic dilemma in some instances. In a study of patients with various forms of facial spasms, more than half of them considered themselves to have psychological problems, which they attributed to be secondary to their symptoms.\textsuperscript{26} Other rare differentials for HFS include hemimasticatory spasm (consists of a unilateral contraction of muscles innervated by the motor trigeminal nerve resulting in a painful jaw-closing masseter and temporalis muscle spasm) and aberrant regeneration with synkinesis, especially after Bell’s palsy. Post-paralytic facial synkinesis as a result of Bell’s palsy is due to transmission by aberrantly regenerating facial nerve fibres. Involuntary eye closure may occur with voluntary mouth opening in these patients.\textsuperscript{1}

**Investigations**

In patients with atypical features such as facial numbness and weakness, MRI and MRA are recommended if facilities are available. However, since HFS is rarely associated with tumours,\textsuperscript{27} imaging in patients with typical HFS may not be cost-effective. Nevertheless, it is important to perform a careful neurological examination in all HFS patients for evidence of focal neurological deficits. Imaging is also suggested for patients who are amenable to surgery after failed conservative treatment. Advanced MR imaging techniques (such as constructive interference in steady state) are highly sensitive for neurovascular contact, and may be useful as a pre-operative investigation before microvascular decompression surgery.\textsuperscript{17}
Treatment

Drugs

The efficacy of oral medications is often transient. Carbamazepine, anticholinergics, baclofen, clonazepam, haloperidol have all been studied in HFS. However, these trials involved small numbers of patients and cautious interpretations of their results are needed. Sedation is a common adverse effect of all these medications, particularly when high doses are used. Recently, the efficacy of gabapentin in HFS has been studied in a number of open-label trials. In one study, 23 patients with hemifacial spasm not suitable for surgery or therapy with botulinum toxin were treated with gabapentin. A clinically significant reduction of facial spasms was obtained in 16 patients (69.6%). Future placebo-controlled trials using a validated HFS severity scale will be needed.

Botulinum toxin injection

Botulinum toxin (BTX) is one of the most potent biological toxins known. The toxin is a zinc endopeptidase that acts on one or more of the neurosecretory proteins in the presynaptic nerve terminal. It inhibits the calcium-mediated release of the acetylcholine into synaptic junction resulting in local chemical denervation and loss of neuronal activity in the targeted organ. There are seven immunologically distinct serotypes of BTX (A-G). Because different serotypes act on different neurosecretory proteins at different sites, there are differences in relative potency and duration of action. For instance, type B has shorter action than type A. Type A is the only commercially available BTX in most countries, as the preparations Dysport and Botox. One unit of Botox is equivalent to 3 to 4 units of Dysport. The muscular weakness produced is reversible and last 3–6 months. BTX-A is injected into the subcutaneous tissue overlying the orbicularis oculi muscle and lower facial muscles. A review of the literature of BTX treatment in HFS showed that there have been numerous open-label studies and a few double-blind placebo-controlled studies, involving more than 2000 HFS patients. Despite the variation in the techniques of BTX injection and the lack of a validated scale to assess treatment response, good to excellent improvement was reported in 75% to 100% of these patients. The mean duration of action was about a few months. Adverse effects included dry eyes, ptosis, eyelid and facial weakness, ptosis, diplopia, and excessive tearing. However, these effects were transient, and no serious systemic effects have been reported. In experienced hands, adjustment of dosage, and site of injection may reduce some of these adverse effects in subsequent treatment. Repeated injections are generally well tolerated, and benefit is maintained over the years of therapy. Immuno-resistance to BTX is rare in HFS, due to the low dosage of BTX used. Many neurologists currently regard botulinum toxin as the treatment of choice for HFS. The major consideration for this treatment is its high cost.

Surgery

Microvascular decompression (MVP) of the facial nerve at the cerebellopontine angle, the most common surgical procedure carried out today, results in markedly improved HFS in the majority of patients, with success rates of >90% in some series. However, a recurrence rate of up to 20% has been reported. With the advent of BTX treatment, which has been shown to be safe and effective, potential complications associated with MVP may be unacceptable to some patients. Common complications of MVP include temporary or permanent dysfunction of facial or auditory nerve, with 7–26% suffering from hearing loss in some reports. Other complications, such as lower cranial nerve dysfunction and intracranial infections, are less common. In a recent review of 4415 MVP operations, in the 2420 operations performed before 1990 for HFS, trigeminal neuralgia, and glossopharyngeal neuralgia, cerebellar injury was reported in 21 cases (0.87%), hearing loss in 48 (1.98%), and cerebrospinal fluid (CSF) leakage in 59 cases (2.44%). In the 1995 operations performed since 1990, cerebellar injuries dropped to nine cases (0.45%), hearing loss to 16 (0.8%), and CSF leakage to 37 (1.85%). These results suggest that improved surgical techniques and experience have led to decreasing complication rates in recent years. However, in unsuccessful operative cases, repeat surgery may run a greater risk of complications.

Extracranial neurosurgical operations involving sectioning the peripheral nerve trunk or its branches, unilateral removal of the orbicularis oculi and corrugator superciliaris muscles, injection of alcohol or phenol to injure the facial nerve, and percutaneous puncture of the facial nerve at the stylomastoid foramen have all been used with varying success in the past and seldom performed nowadays.
Conclusions

Involuntary facial movements such as facial twichings and grimacing are frequently accepted as a ‘normal’ response to stress and anxiety. However, one needs to be vigilant for organic causes such as HFS, a condition which can be effectively treated. Early diagnosis of HFS or other organic involuntary facial movements allows institution of appropriate therapy and improves quality of life.

References


