

Progressive Facial Hemiatrophy Revisited

A Role for Sympathetic Dysfunction

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Objective: To report a case of progressive facial hemiatrophy with unusual features of contralateral brain atrophy and transcranial Doppler ultrasound evidence of autonomic dysfunction.

Design: Case report.

Setting: A teaching hospital.

Patient: A 63-year-old man who presented with a 10-year history of progressive right-sided facial atrophy and recent facial pain.

Results: Brain magnetic resonance imaging revealed left frontoparietal atrophy. Transcranial Doppler ultrasound demonstrated evidence of autonomic dysfunction ipsilateral to brain atrophy.

Conclusion: This case expands the spectrum of findings in progressive facial hemiatrophy to include contralateral brain atrophy and suggests that sympathetic dysfunction might play a pathogenic role in progressive facial hemiatrophy.

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PROGRESSIVE FACIAL HEMIATROPHY (PFH), also known as Parry-Romberg syndrome, is a rare condition characterized by slow and progressive hemifacial atrophy of skin, subcutaneous tissue, muscle, and bone.^{1,2} The underlying pathogenesis is not well understood. Congenital mechanisms, disturbance of fat metabolism,^{3,4} and trophic malformation of the cervical sympathetic trunk^{5,6} have been implicated. We describe a case of PFH in which we detected the supine-standing cerebral blood flow velocity curve using transcranial Doppler ultrasound to assess the autonomic function and found evidence of sympathetic dysregulation.⁷

REPORT OF A CASE

A 63-year-old right-handed man presented with right-sided facial pain and atrophy of the right side of his face. His facial atrophy had been noted at least 10 years earlier. He reportedly did not seek medical attention because he had no discomfort except for the appearance. Two years before presentation, he developed a persistent mild right-sided facial pain with stabbing and burning features, aggravated by eating and by washing his face. The pain gradually progressed and in-

creased in severity, prompting him to refer to our hospital. He was otherwise healthy. On physical examination, blood pressure was 165/80 mm Hg in the supine position and 160/70 mm Hg in the standing position. The patient had facial asymmetry with marked atrophy of the right side of the face, right-sided deviation of the nose and lip, and enophthalmos of the right eye (**Figure, A**). Brain magnetic resonance imaging showed diffuse left frontoparietal atrophy (**Figure, B**). Dynamic transcranial Doppler ultrasound findings showed that the mean blood flow velocity of the middle cerebral artery of the apparently morphologically normal hemisphere (blue curve) decreased following standing up but quickly rebounded and reached the supine baseline level in 30 seconds, whereas the curve trend of the abnormal hemisphere (red curve) decreased following standing up and failed to return to the baseline level (**Figure, C**). Results of the nerve conduction studies of the facial and trigeminal nerves were normal. The patient was treated with carbamazepine with subsequent improvement of his trigeminal neuralgia.

COMMENT

Progressive facial hemiatrophy is a craniofacial disorder characterized by pro-

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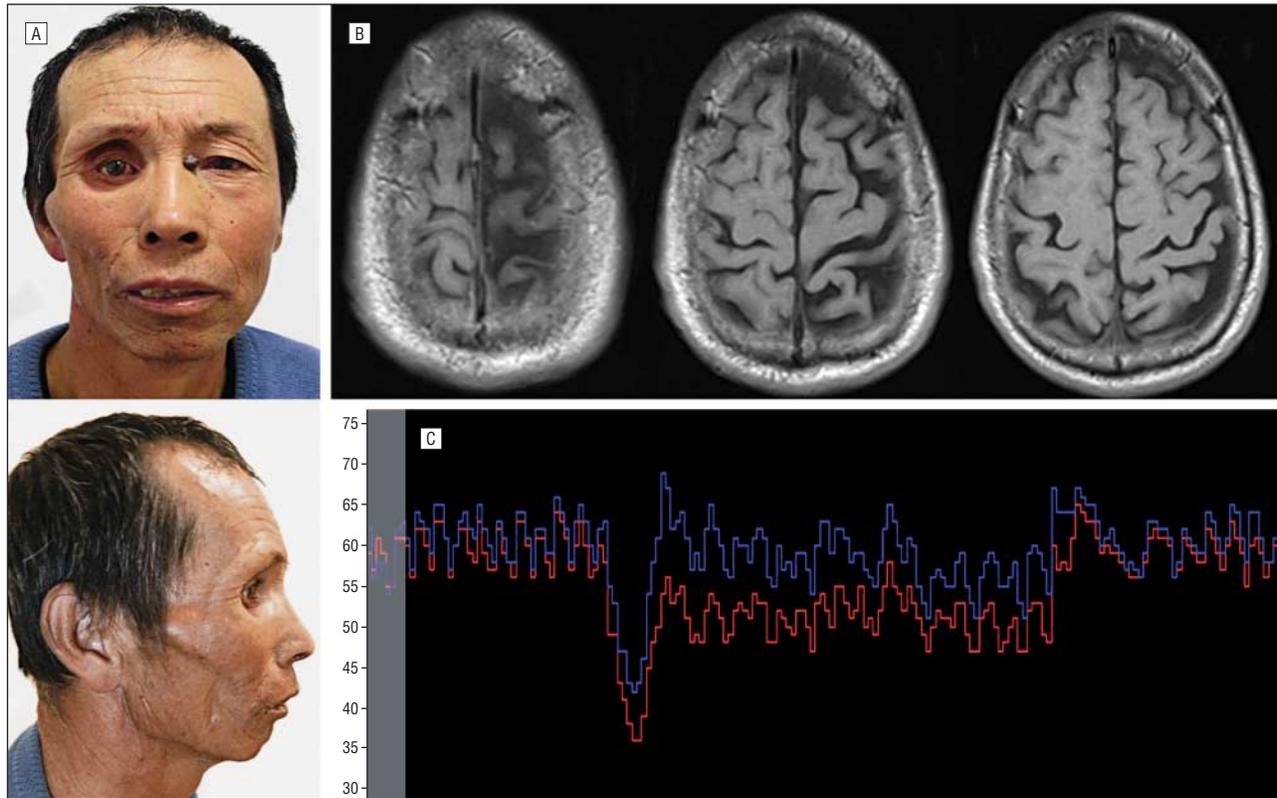


Figure. Morphologic, radiologic, and sonographic features of progressive facial hemiatrophy. A, Facial asymmetry with marked atrophy of the right side of the face, right-sided deviation of the nose and lip, and enophthalmos of the right eye. B, T2-weighted magnetic resonance imaging (1.5 T) shows left frontoparietal hemiatrophy. C, Transcranial Doppler ultrasound shows that the mean blood flow velocity of the left middle cerebral artery (red curve) drops following standing up and does not return to the baseline level in 2 minutes. The curve trend of the right middle cerebral artery (blue curve) is normal.

gressive shrinking and deformation of one side of the face with atrophy of the subcutaneous connective and fatty tissues.⁸ The atrophy often progresses slowly for many years and then stabilizes. The main morphologic features are unilateral facial atrophy and ipsilateral enophthalmos and deviation of the mouth and nose toward the affected side.^{9,10} Neurologic abnormalities occur in 15% of patients and include focal seizures, visual loss due to involvement of the retina and optic nerve, facial pain due to trigeminal neuralgia, and migraine.¹¹ Neuroradiologic features include ipsilateral cerebral hemiatrophy and white matter hyperintensity on magnetic resonance imaging and intracortical calcifications.¹¹ Our patient exhibited several of these morphologic and neurologic features. The stabbing and burning pain on the right-sided face of the patient presented almost all the time and was aggravated by eating and by washing his face. We attributed his facial pain to trigeminal neuralgia, but a possible neuropathic pain cannot be fully excluded. The pathologic etiology of PFH is poorly understood; trauma, infection with virus, immunologic abnormality, cranial vascular malformation, connective tissue disorder, and disturbance of fat metabolism have been proposed.¹² In addition, PFH may be congenital in nature.⁸ Because the cells participating in the formation of the frontonasal bud and the cells that give rise to the cerebral hemisphere have common progenitors when one side of the rostral neural tube is affected, it can cause ipsilateral cerebral and facial lesions later in life.^{3,12} Interestingly, our patient had

cerebral hemiatrophy, contralateral to the side of facial atrophy. To our knowledge, this is the first case presenting with a contralateral brain hemiatrophy. This finding expands the current spectrum of PFH, indicating that it may also be a bilateral disorder.

Trophic malformation of the cervical sympathetic trunk leading to sympathetic dysfunction has been implicated in the pathogenesis of PFH.^{5,6} We included a new transcranial Doppler ultrasound–based technique, developed by Xu et al,⁷ in our diagnostic evaluation to assess the autonomic function. In healthy individuals, the cerebral blood flow velocity decreases following standing up but quickly rebounds and returns to the supine baseline level or higher in 30 seconds and then is maintained at the baseline level. In our patient, the blood flow velocity curve of the left middle cerebral artery dropped following standing up and did not return to the baseline level in 2 minutes, but the curve trend of the right middle cerebral artery was normal (Figure, C). Many mechanisms are involved in maintaining the tone of the cerebral vessels with posture changes. In our patient, we consider that the separation of the 2 curves is indicative of dysfunction of the sympathetic nervous system. Moreover, myogenic mechanisms are substantially responsible for cerebral autoregulation at normal perfusion pressures.^{13,14} The contralaterality of the observed brain hemiatrophy and transcranial Doppler ultrasound evidence of sympathetic dysfunction to the facial atrophy are intriguing in that a possible causal link between sym-

pathetic dysfunction and brain atrophy was implied. Progressive facial hemiatrophy might be attributed to a diffuse rather than a unilateral process. In conclusion, we report a case of PFH with an unusual feature of contralateral brain hemiatrophy and transcranial Doppler ultrasound findings supporting a role of sympathetic dysfunction in the pathogenesis of this disorder.

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