Progressive facial and cerebral hemiatrophy[1]

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A 43-year-old woman with progressive facial hemiatrophy had contralateral hemianopia and sensory loss. A computed-tomographic brain scan showed ipsilateral cerebral atrophy. The neurological and neuroradiological features of progressive facial hemiatrophy are reviewed.

Index terms: Brain, pathology • Case reports • Facial muscles

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Progressive facial hemiatrophy (PFH) is a disorder of unknown etiology characterized by loss of subcutaneous tissue, sometimes accompanied by changes in skin, muscle, hair, cartilage, and bone, and confined to one side of the face and cranium.[1,2] Less commonly, the atrophy may spread to the neck, upper trunk, and arm ipsilaterally and rarely may involve the entire side of the body.[3]

Neurological disorders, including epilepsy, either partial or generalized; headache; ipsilateral facial pain; and focal deficits, may be found in up to 15% of patients with PFH.[4,5] Focal deficits usually suggest ipsilateral, but occasionally contralateral, brain involvement.[6–8]

The patient described here had typical clinical features of PFH with neurological involvement. Serial computed-tomographic (CT) scans showed brain hemiatrophy ipsilateral to the facial abnormality, correlating with the contralateral visual and limb dysfunction.

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Case report

A 43-year-old right-handed housewife was cyanotic at birth, but had no obvious congenital abnormality or developmental delay. At two years of age, she had sepsis with high fever and a generalized convulsion. She was then well until she was 21 years old when numbness of the right foot spread to the ipsilateral leg, trunk, and arm over a period of several months. From that time, she observed incoordination of the right hand, which has remained relatively stable since then. At about age 23, she noted left facial flattening and was told of Bell's palsy. At age 29, generalized tonic-clonic seizures began, occurring one to four times per month despite anticonvulsants until the last three years when only an occasional seizure occurred while she was taking phenytoin (400 mg daily). At about age 30, generalized, nonthrobbing headaches occurred two to four times per month, lasting up to one day. Three years previously, she consulted an ophthalmologist because of a "lump" on her lower eyelid. A right homonymous superior quadrantanopia and uveitis in the left eye were identified. She was referred to a neurosurgeon as well as a neurologist and underwent numerous studies, including at least two electroencephalograms (EEGs) which were normal. Interpretations of CT scans at that time were conflicting; one indicated a brain tumor; the other, brain atrophy. There was no family history of facial abnormality or neurological disease other than a recently diagnosed brain tumor in a maternal aunt.

The patient was seen at The Cleveland Clinic Foundation for differential diagnosis of her condition. Examination of this depressed-appearing woman revealed moderate left hemifacial atrophy (*Fig. 1*). The cranium, facial bones, and ears were normal. She had mild thoracic kyphoscoliosis. Her arms were symmetrical, but the right leg was slightly shorter and the right foot slightly smaller than the left. Mental function was normal. She had a congruous homonymous right upper quadrantanopia. The right optic disc was normal; the left was poorly visualized. She had heterochromia iridis, the left being lighter in color. The palpebral fissures were symmetrical. Pupils were 4 mm in diameter and normally reactive to light and near point stimulation. No oculomotor abnormalities were noted. The corneal reflex in the right eye was diminished and there was impaired sensation of the right side of the face, least prominent in the mandibular division. Despite facial asymmetry, there was

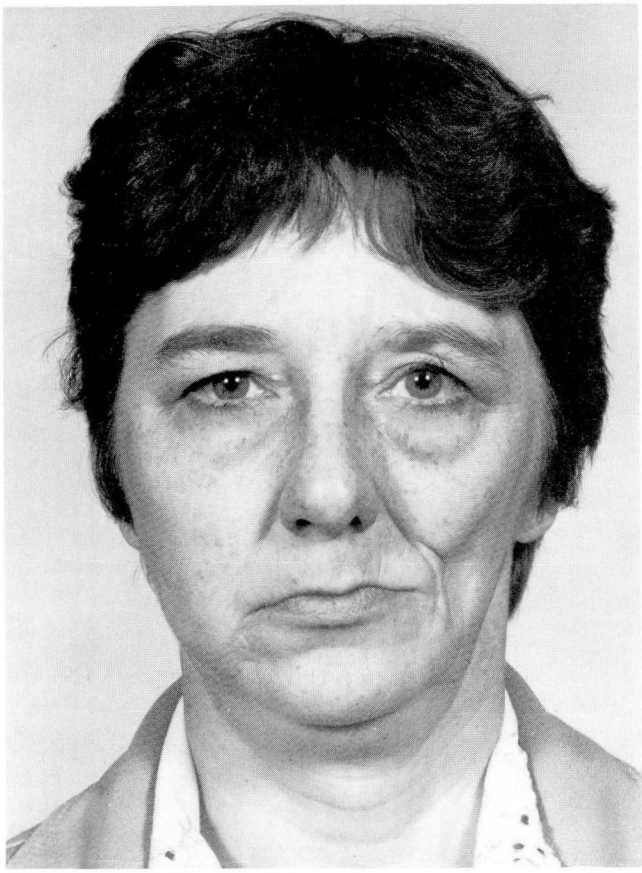

Fig. 1. Left hemifacial atrophy.

neither facial weakness nor synkinesis. The left nasolabial fold was deeper than the right. The lower cranial nerves were normal. Muscle tone, bulk, and strength in the trunk and limbs were symmetrical. There was mild slowing of finger tapping on the right and marked ataxia of the right limbs, particularly with eyes closed. Sensation was markedly impaired for all modalities on the right. Pseudoathetotic movements of the right hand were noted with eyes closed and upon walking. Tendon reflexes were symmetrical and normal. Plantar responses were flexor. A CT scan is shown (*Fig. 2*). The patient's condition was diagnosed as PFH.

Discussion

PFH was first described by Parry[9] in 1825, although credit is generally given to Romberg[10] for the initial recognition of this entity in 1846. Eulenburg[11] named this disorder PFH in 1871. The neurological aspects of PFH have been extensively reviewed by Archambault and Fromm[6] and Wartenberg.[7] Wolf and Verity[8] have more recently enumerated the neurological complications of the disorder. The prevalence of neurological abnormalities in PFH is difficult to ascertain, but probably is in the range of 15%.[4,5] Seizures are the most common form of neurological involvement, including generalized tonic-clonic,[6–8,12,13] simple partial (primarily affecting contralateral limbs), and complex partial types.[7,12,14,15] Headaches, both hemicranial and generalized, have also been frequently described.[7,12,15,16] Facial pain ipsilateral to the hemiatrophy was reported by Wolff.[13] Asher and Berg[16] described a patient with bifacial paresis, more pronounced on the atrophic side, and ipsilateral tongue atrophy has also been noted.[7,16]

Signs of cerebral hemispheric dysfunction, usually ipsilateral to the facial atrophy, occasionally occur. These include hemiparesis and hemisensory loss,[7,8,14,15] hemianopia,[14] and aphasia.[14] A variety of neuro-ophthalmologic disturbances has also been recorded, including pupillary dilatation,[6,8,13] with or without reactivity, ocular palsy or conjugate-gaze paresis,[6,8,15,17] ptosis,[6,8,15,17] and optic atrophy,[15] as well as heterochromia iridis as in the patient described here. Archambault and Fromm[6] reviewed several cases showing signs of Horner syndrome, including facial anhidrosis.

EEG abnormalities have been reported by Rischbieth,[15] Eadie et al[12] (bilateral or unilateral slowing, focal contralateral slowing, diffuse dysrhythmia), and Asher and Berg[16] (ipsilateral focal slowing). Air encephalography has demonstrated ventricular dilatation, both ipsilaterally[12,14] and contralaterally,[12] as well as diffusely.[8,15] Abnormalities on the CT scan, as reported by Asher and Berg,[16] included intracerebral calcification, ipsilateral hypodense areas, and a contralateral area of enhancement with no angiographic lesion demonstrable. Brain hemiatrophy, as noted on the CT scan of the patient described here, has not been previously reported to my knowledge.

The etiology of PFH remains unknown. Wartenberg[7] considered it to be a heredodegenerative disorder of the nervous system, although familial cases remain distinctly rare and only in a minority can neurological abnormalities be detected. A primary disorder of the sympathetic nervous system has been suspected by some,[2,6] and there are a few experimental studies linking localized atrophy to lesions of sympathetic ganglia. Moss and Crikelair[18] produced variable ipsilateral decrease in subcutaneous facial fat in rats by severing the cervical sympathetic chain. No bony, skin, or neurological changes were noted. The sympathectomy was done when the animal was one month old—a time at which

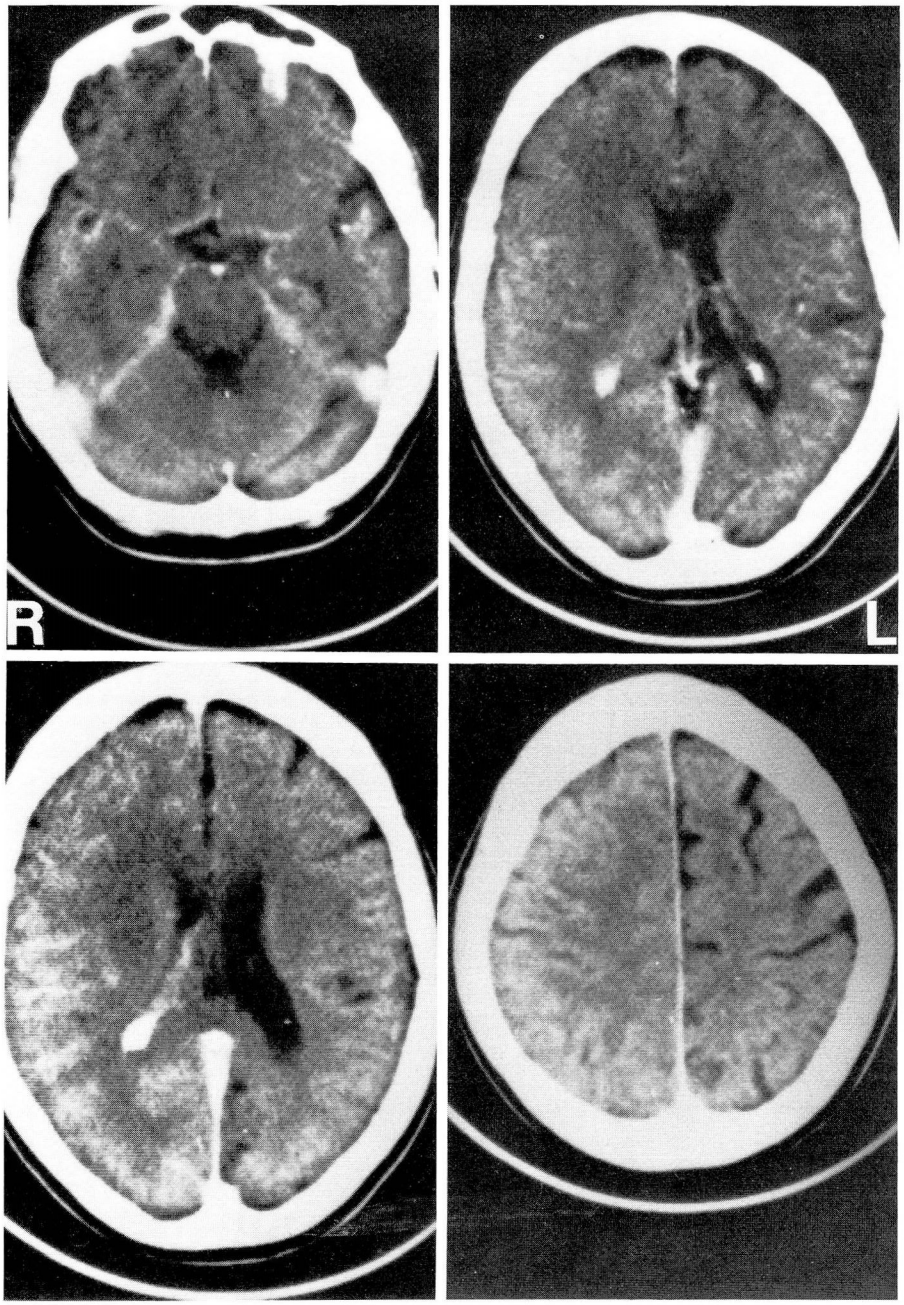

Fig. 2. Contrast-enhanced CT scan. The left cerebral atrophy involves both cortical and subcortical structures.

neurocranial growth in the rat is virtually complete. Certainly, atrophic changes are well recognized in limbs following nerve injury, and the autonomic nervous system presumably plays at least some role in this, whether directly or via effects on nutrient blood vessels. Probably little has been learned in this regard since the classic and extensive monograph on trophism by Wyburn-Mason.[19] Other suspected causes of PFH have included localized lipodystrophy, localized scleroderma, trauma, and infection.[2]

Treatment of PFH has largely been symptomatic. Seizures and headaches will generally respond to the usual measures. Early attempts to manage the cosmetic abnormalities included subcutaneous injections of paraffin—a procedure

now considered hazardous and obsolete.[1,2] A variety of grafting techniques has been suggested and some are highly successful in improving the appearance of the more severely disfigured patients,[20,21] thus reducing the psychological burden of the disease.

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