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Case Presentation

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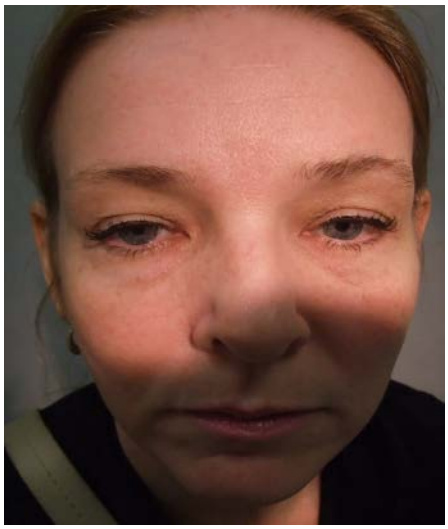
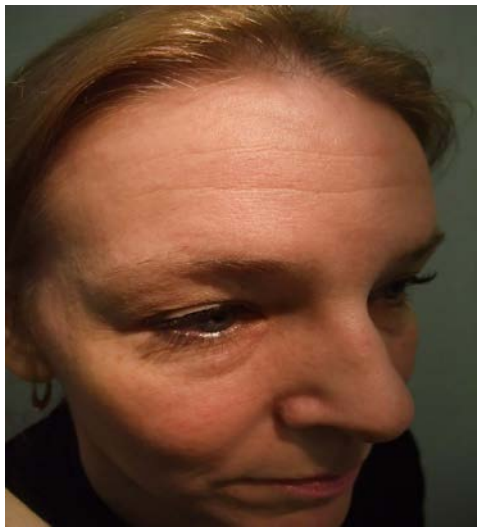
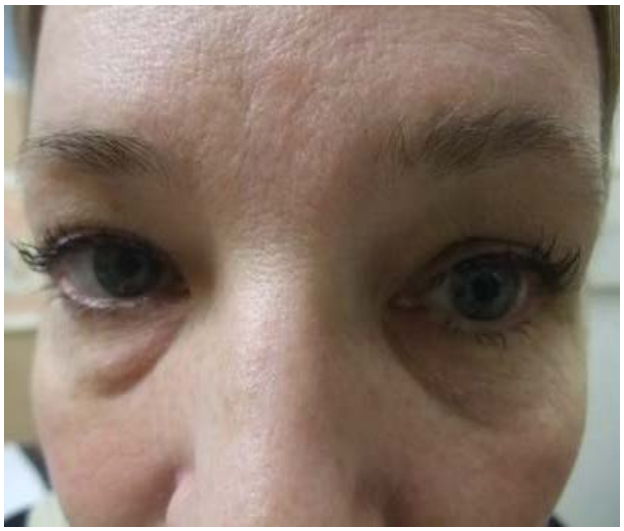
Abstract

We report the case of a 44-year-old woman with a one-year history of en coup de sabre morphea and progressive hemifacial atrophy with ipsilateral hemifacial neuralgia, migraine, and contralateral neurologic abnormalities. While rare, Parry-Romberg syndrome typically presents in the first or second decade of life; this patient's case is unusual in that the onset of her disease is demonstrated at age 43. Common clinical features, laboratory findings, and pathogenetic theories are discussed.







Case synopsis

A 44-year-old woman presented to a voluntary faculty member's private dermatology practice for evaluation of a one-year history of a linear depression on the right side of the forehead with hemifacial atrophy. These facial abnormalities were noticed concurrently with the onset of a number of neurologic symptoms, which included paroxysms of throbbing of the right cheek and eye pain, left hemifacial paralysis, numbness and weakness of the left arm and leg, and migraines. The patient has no history of seizures.

In the five months since first establishing care with her dermatologist, the patient noted a progression of the hemifacial atrophy, which had worsened periorbitally. She was referred for neurologic and ophthalmologic evaluations. Treatment with antimalarials was deferred because the patient has congenitally poor left eye vision.

Physical Examination: Extending from the right paramedian anterior aspect of the scalp towards the right medial eyebrow, there was a curvilinear, atrophic depression with modest hyperpigmentation and no induration. There was associated atrophy of the right temple and malar aspect of the cheek. Subtle right ptosis was present. No brow or forehead alopecia was noted. Exophthalmometry showed relative enophthalmos of the right eye. Fundoscopy showed mild temporal pallor of the right optic disc.

Laboratory Data: A complete blood count was normal. Anti-Ro and anti-La antibodies were negative. Lyme disease antibody screening index was 0.18 (negative). A magnetic resonance imaging study showed no abnormalities in the supratentorial gray or white matter, brainstem, or cerebellum. A magnetic resonance angiogram showed no hemodynamic stenosis, intracranial aneurysm, or dissection.

Diagnosis: Progressive hemifacial atrophy **(**Parry-Romberg syndrome)

Discussion: Progressive hemifacial atrophy, which also is known as Parry-Romberg syndrome, is a rare condition of unknown etiology that was first reported by Parry in 1825 and later recognized by Romberg as a syndrome in 1846 [1, 2].

Typically presenting within the first two decades of life, it is characterized by idiopathic, self-limited cutaneous hemifacial atrophy, usually in a dermatomal distribution. Atrophy of underlying soft tissue, muscle, and bony structures is associated. Neurologic abnormalities, which include migraine, facial pain, and seizure, are common [3, 4]. Dyspigmentation is frequently noted preceding the onset of atrophy as is alopecia in regions of scalp involvement, but neither is necessarily present [5]. A linear lesion of morphea that is classically located on the paramedian aspect of the forehead, *en coup de sabre* morphea, has been associated with Parry-Romberg syndrome as well, although these two entities are not clearly related. Some authors argue that they are distinct entities, whereas the largest cohort of patients with both *en coup de sabre* morphea and Parry-Romberg syndrome studied to date supports the assertion that these are two phenotypes on the same spectrum of disease [3-8].

Laboratory studies may show elevated antinuclear antibody titers, which has led clinicians to believe that Parry Romberg syndrome is associated with connective tissue diseases, especially when presenting in the setting of linear morphea [9]. However, several authors have reported Parry Romberg syndrome in association with borreliosis, although antibiotic treatment has yielded mixed clinical results [9-11].

The pathogenesis of Parry-Romberg syndrome remains unclear. Several hypotheses have been posited, which include neurosympathomimetic and inflammatory mechanisms. A neurosympathomimetic theory was supported by the reproduction of many of the clinical manifestations of Parry-Romberg syndrome, including hemifacial atrophy, localized alopecia, and enophthalmos, after ablation of the superior cervical sympathetic ganglion in rabbits, cats, and dogs [6, 12]. Intrathecal production of IgG and response to immunomodulatory therapy has been demonstrated in one patient, which potentially implicated an inflammatory mechanism [13].

Potentially epileptogenic ipsilateral cortical white matter atrophy and intracranial vascular malformation both have been described in association with Parry-Romberg syndrome [14]. In the absence of brain abnormalities demonstrated by this patient's essentially normal magnetic resonance imaging studies, the presence of our patient's neurologic symptoms that were temporally associated with the onset of cutaneous findings suggests an as-yet undefined pathogenesis of neurologic abnormalities in Parry-Romberg syndrome. The rarity of this disease continues to make further investigation challenging.

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