Case Report

Parry–Romberg syndrome

Overlap with linear morphea

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ABSTRACT

Parry-Romberg syndrome, also known as progressive hemifacial atrophy, was first described by Parry in 1825 then Romberg in 1846. It is a poorly understood rare disorder characterized by progressive hemifacial atrophy of the skin, subcutaneous tissue, and sometimes, the underlying structures including muscles, cartilages and bones. A case report of a Saudi female with this rare disorder is presented with a brief review of the literature.

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Parry-Romberg syndrome (PRS) was first described by Parry in 1825 then Romberg in 1846.[1,2] It characterized by progressive hemifacial atrophy of the skin, subcutaneous tissue, and in some cases, can extend to the underlying structures including muscles, cartilages and bones.[3] Here, we report a Saudi female with this syndrome and briefly review the literature.

Case Report. A 28-year-old Saudi female presented with facial asymmetry that started at the age of 21 years. Initially, she noticed insidious onset depression of the skin of the right side of her face, associated with scalp hair loss on the same side. She underwent liposuction of the left side of the face and chin because of false diagnosis of hypertrophy of this side. Past medical history revealed history of anxiety, depression and rheumatic valvular heart disease. On examination, she looked well, with gross facial asymmetry. Gross atrophy of the right side of the face was apparent, most prominent on the right chin (**Figure 1**). Marked atrophy and loss of the subcutaneous fat with brownish hyperpigmentation and prominent veins was seen in the right submental region. An atrophic linear band extended from the right frontal hair line to the right eyebrow (**Figure 2**). The right frontal scalp showed partial hair loss and diminished subcutaneous fat. Mouth examination revealed an atrophic right side of the tongue. Based on the clinical findings the diagnosis of PRS was made. Blood work was normal except for mildly positive antinuclear antibodies (ANA) of 17.32 units (negative is less than 20) and single strand DNA (ssDNA) of 120 units (negative is less than 10). Skin biopsy from the right submental region showed significant dermal fibrosis around the hair follicles and eccrine glands, with loss of the periadnexal adipose tissues. The scalp biopsy showed only thinning of the dermis. There was no significant inflammation in either biopsies. This was compatible with burnt out PRS. A CT scan of the brain was normal. The patient indicated that her disease had not been progressing for the last one year, thus, no active treatment was given. Referral to plastic surgery was arranged. The patient was asked to take dated facial photographs

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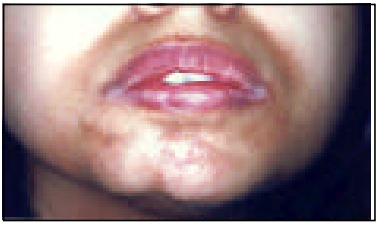

Figure 1 - The face of the patient showing gross atrophy of the right chin.

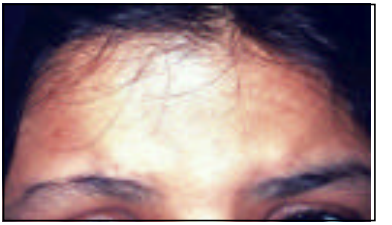

Figure 2 - The forehead of the patient showing atrophic band extending from the hairline to the right eyebrow.

every 3 months to document the stabilization of the disease. A follow-up visit at 3 months revealed no progression of the disease.

Discussion. Parry-Romberg syndrome, also known as progressive hemifacial atrophy, was first described by Parry in 1825 then Romberg in 1846.[1,2] It is a poorly understood rare disorder characterized by progressive hemifacial atrophy of the skin, subcutaneous tissue, and sometimes, the underlying structures including muscles, cartilages and bones.[3] The involved skin looks depressed, indurated, hyperpigmented, shiny and devoid of hair.[4] The skin changes occur most often in the frontoparietal region but may affect the nose, the cheeks, the mouth or ears.[5] The disease usually does not cross the midline. The eyelashes, eyebrows and scalp hair can be affected with ipsilateral poliosis or alopecia. The disease affects females more than males but there is no left/right predominance as previously thought.[4,6] The onset is usually insidious with most cases appearing during the first or early second decade of life with the median age of onset being 10 years.[3,6] Once the disease starts, it usually progresses over the first 2-20 years and then stabilizes and "burns itself out." Patients with earlier age of onset are often more severely affected.[7]

It is controversial whether PRS is considered a separate entity or a variant of LM. A clear differentiation between PRS and LM "en coupe de sabre" (LMCS) is not always possible.[8] There are no reliable histological criteria to differentiate PRS and LMCS.[8] Proponents of differentiating PRS from LM distinguish the former by more extensive involvement of the lower face and lack of induration or hyperpigmentation in PRS.[5] Blaszczyk et al[9] observed several patients with typical "en coup de sabre" in children converting within several years to PRS.[9] The finding of positive ANA in our patient and in previous reports lend support to this overlap.[10] Moreover, our patient had high titre of ssDNA antibodies, which are commonly seen in many types of morphea including LM.[11] To explain the controversy, Sakuraoka et al[12] suggested the existence of 2 types of PRS. The first is a non-sclerotic type with cutaneous atrophy not associated with other findings.[12] The second is a sclerotic type with induration and hyperpigmentation resembling morphea.[12] Our patient seems to belong to the sclerotic type as evident clinically by the hyperpigmentation and histologically by the dermal fibrosis.

Other system involvement is not uncommon in PRS. The central nervous system (CNS) involvement is the most common extracutaneous manifestation. The most common CT findings are ipsilateral atrophy of the cortical white mater and intracortical calcification.[13,14] Fry et al[5] found evident CNS abnormalities documented by MRI in 5 out of 6 patients. Blaszczyk et al[15] also found discrete MRI abnormalities in almost all patients with PRS. Hyperintense lesions in the white mater are the most common finding on T2 weighted MRI.[13,14] Other reported abnormalities include vascular malformations, cortical thickening, gyral effacement and cystic infarcts with foci of encephalomalacia and meningoencephalitis.[5,7,13,15] Despite the accompanying cerebral lesions, up to 15% of patients with PRS have symptoms in the form of epileptic seizure or migraine-like headaches.[3,7,14] Usually, the neurological abnormalities are mild or subclinical.[14,15] In a global internet survey of 205 patients with PRS, 46% suffered from anxiety and 10% had depressive symptoms.[6] Ocular findings have been reported in 10-40% of cases.[13] The most common is progressive enophthalmos secondary to orbital fat atrophy.[7,13] Other findings include deepening of the supratarsal crease, and loss of eyebrow hair.[6,7] Papillitis, retinal vasculitis, and retinal vascular changes are rarely seen.[16]

Despite the typical features of PRS, its pathogenesis remains a mystery and controversial.

This is due to the rarity and the heterogeneity of this puzzling disease. Many theories of pathogenesis have been postulated. These include neurologic dysfunction, autoimmune process, viral infection and head trauma.[6,14,15,17] The neurologic theory points to sympathetic nervous system dysfunction.[13,17] One case report of inhibition of the atrophic process of PRS by stellate ganglion blockade for 16 months supports this theory.[18] The overwhelming reports of PRS overlapping with LM support the autoimmune theory. The neurological dysfunction could explain the non-sclerotic type of PRS while the sclerotic type could be explained by autoimmune pathogenesis as morphea.[12]

At present, there is no known effective treatment for PRS.[19] Palliative reconstructive surgery is of potential benefits for patients with gross facial disfigurement.[8] These are usually deferred until the disease "burns itself out".[19] These procedures include injections of various types of fillers, autologous tissue transfer, and inorganic implants.[19]

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