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CASE REPORT

Parry–Romberg syndrome in association with anti-dsDNA antibodies: a case report

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ABSTRACT

Parry–Romberg syndrome (PRS) is a rare and puzzling disorder that is characterized by progressive hemifacial atrophy. It involves mainly some or all tissues of one side of the face. A case of 21-year-old Caucasian man with hemifacial atrophy in the right facial region is reported. Serological studies with anti-single-stranded DNA (anti-ssDNA), anti-double-stranded DNA (anti-dsDNA), anticentromere (ACA) and antinuclear (ANA) antibodies were done. Anti-dsDNA antibodies was found positive, but the others were negative. Rheumatoid factor (RF) was also negative. Since PRS is rare and its association with anti-dsDNA antibodies was not reported before, this case appears to be the first report.

Key words: anti-dsDNA, Parry–Romberg syndrome

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Introduction

Parry–Romberg syndrome (PRS), originally described by Parry (1825) and Henoch and Romberg (1846), consists of slowly progressive atrophy of the soft tissues of essentially half of the face, accompanied usually by neurological and ophthalmological findings.[1–3] Evidence of a mendelian basis is lacking. The presence of autoantibodies in some cases suggested that this syndrome may be a form of localized scleroderma.[4,5]

We present a 21-year-old male patient with PRS as characterized by progressive right hemifacial atrophy associated with anti-double-stranded DNA (anti-dsDNA) antibodies. To our knowledge, this association was not reported before. The clinical, immunological and pathological features of this rare entity are discussed.

Case report

A 21-year-old male patient was admitted to our hospital for evaluation of his progressive right facial atrophy of 6 years duration (fig. 1). His medical history was unremarkable. Review of his systems was essentially unremarkable. Other physical, neurological and ophthalmological examinations were normal. Radiological examination comprising plain X-rays and computed tomography (CT) scans were within normal limits. Serological studies by enzyme-linked immunosorbent assay (ELISA) revealed that anti-dsDNA antibody was positive (= 27.01 U/mL; normal value ranges 0–20 U/mL), although others including anti-single-stranded DNA (anti-ssDNA), anticentromere (ACA) and antinuclear (ANA) were negative. Rheumatoid

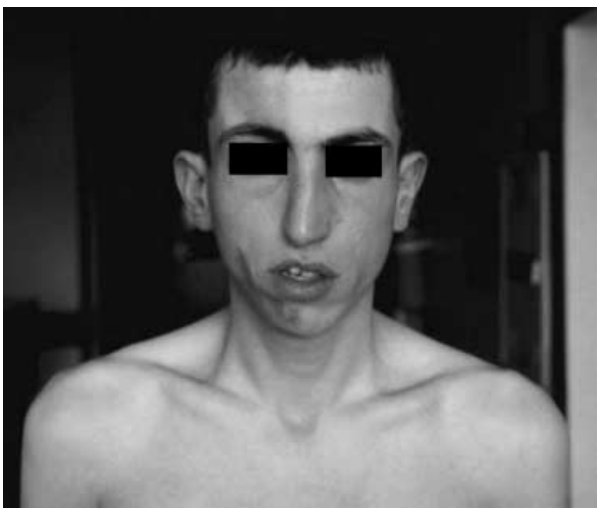
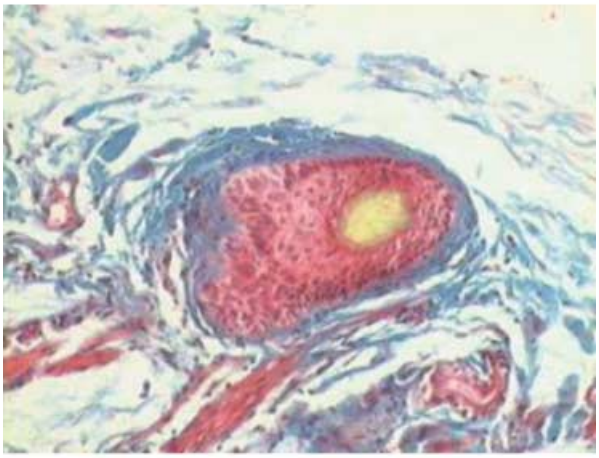


fig. 1 The photograph of the face of patient showing the right hemifacial atrophy.

a



b

fig. 2 (a) The histopathological appearance of the specimen obtained from the atrophic skin showed normal epidermis and mild mononuclear lymphocyte infiltration in the reticular dermis (H&E, ×25). (b) The histopathological appearance of the specimen obtained from the atrophic skin stained with trichrome dye showing the fibrosis and mononuclear lymphocyte infiltration around the eccrine glands (Trichrome, ×100).

factor (RF) was also negative. The pathological examination of his facial skin was made by punch biopsy obtained from atrophic region. The specimens were stained with haematoxylin and eosin (H&E) and trichrome dye. The examination revealed a sclerodermoid tissue reaction as characterized by widened collagen bundles with loss of the fibrillar architecture with a concomitant perieccrine and perifollicular lymphocytic infiltrate (fig. 2a,b).

The patient did not undergo any surgical procedure because of lesser benefits.

Discussion

Parry–Romberg syndrome is a rare disorder characterized by an atrophic dysplasia of the superficial facial structures including the dermal and subcutaneous tissues on one side of the face, occasionally extending to the other parts of the body. Tissues involved are the skin, tongue, gingiva, soft palate, the cartilage of the nose, ear, subcutaneous, larynx, muscle and bone. But the muscles and bones are seldom involved. It usually begins in the first two decades and is slowly progressive. In its advanced form, the face is gaunt and the skin is thin, wrinkled and rather brown.[3] Rarely there is a positive family history.

It is an age-old argument that there is a relationship between linear scleroderma (LSc) and PRS. PRS and LSc are considered parts of a clinicopathological spectrum that also includes cases with features of both diseases. But in morphea, the lesions usually are limited to the skin and to the subcutaneous tissue beneath the cutaneous lesions; rarely, however, that the underlying muscles and bones are also affected. In PRS, the atrophy is deeper than that seen in LSc. The skin is less often bound down. More extensive involvement of the lower face is another feature of PRS.[6,7]

There have been many reports of progressive hemifacial atrophy and LSc with the presence of autoantibodies and RF. The most common antibodies encountered in scleroderma are anti-Scl-70 and ACA antibodies. The intriguing finding of autoimmune antibodies directed at cellular targets in scleroderma point to a role for antibodies as clinically valuable prognostic indicators. ACA and anti-Scl-70 found in scleroderma have a 50–80% prevalence with greater than 90% specificity for CREST (Calsinosis cutis, Raynaud's phenomenon, Esophageal dysmotility, Sclerodactyly, Telangiectasia) syndrome and diffuse systemic sclerosis (dSSc), respectively.[8] Adebajo *et al.* reported two cases of LSc and hemiatrophy in association with antibodies to dsDNA in 1992.[9] Ruffatti *et al.* reported 52 patients with localized scleroderma and revealed a significant prevalence of anti-ssDNA in morphea.[10] But the autoantibody profile of PRS was different from that in localized scleroderma in one study. Garcia-de la Torre *et al.* reported ANA positivity as 57%, RF positivity as 36%, antihistone antibody positivity as 21% and ACA positivity as 14% in their PRS series, and they did not find any anti-DNA antibody.[5] Kayanuma and Oguchi also reported a case of progressive hemifacial atrophy associated with the presence of anti-DNA antibody and RF in 1994, but the atrophy in this case was not limited on one side of the face and disseminated to the arm.[4]

Anti-dsDNA was positive and anti-ssDNA, anti-Scl-70, ACA and RF were negative in our patient. Although anti-dsDNA is a characteristic finding for systemic lupus erythematosus, we did not found any clinical sign of this autoimmune disorder in our patient. The reports mentioned above suggest that there is still not a clear-cut difference of the autoantibodies between localized scleroderma and PRS, even though, to our knowledge, there is not another report about PRS associated with anti-dsDNA antibodies as in our case.

Skin biopsy of PRS is indistinguishable from that of LSc. The histopathological characteristics of LSc consist of two stages

including, early inflammatory and late sclerosis stages. There are no inflammatory changes in PRS, even in its early stages. The epidermis is normal. The collagen bundles in the reticular dermis often appear thickened and closely packed and stain more deeply eosinophilic than in normal skin. The eccrine glands appears markedly atrophic.[11,12] In our patient, we obtained punch biopsy from the right side of frontal region of the face and histopathological examination was performed with haematoxylin and eosin (H&E) and trichrome stains. Mild mononuclear lymphocyte infiltration was observed in the reticular dermis and around the eccrine glands and hair follicles obtained from the atrophic site. The collagen bundles were thickened in the reticular dermis. We used trichrome stain to demonstrate the collagen fibres because the main value of this stain is in the evaluation of the type and amount of the extracellular material. The histopathological examination of our patient's specimens complied with the PRS.

The associated lesions of PRS vary from the neurological disorders to ocular complications. Rasmussen syndrome, the chronic focal encephalitis, is one of the similar syndromes of Parry–Romberg, which is reported by Straube *et al.* in 2001.[13] The age of onset, unilateral manifestation and occurrence of focal seizures are the clinical similarities of these two syndromes. Seizure, migraine and intracranial aneurysm are the most reported neurological conditions in association with PRS.[13,14] Adie's pupil, enophthalmus, blepharoptosis, loss of cilia, retinal telangiectasis, shrinkage of the eyeball and thinning of extraocular muscles are the ophthalmological abnormalities which may be present with PRS.[7,15,16] We examined our patient in detail and not found any neurological and ophthalmological findings similar with literature.

In conclusion, PRS is a rare and poorly understood disease that is difficult to differentiate from the localized scleroderma with histopathological examination, especially in the late stage. And nowadays it is also not possible to differentiate two diseases with the help of autoantibodies because of having common ones such as anti-dsDNA and limited number of studies.[5,9] The coexistence of these antibodies with PRS may confirm that PRS and LSc *en coup de sabre* represent overlapping conditions. In the view of the fact that observing serological abnormalities in PRS, it can be offered that this should be an autoimmune disease that is stimulated by a variety of factors including trauma and neurological abnormalities in genetically predisposed individuals. But if someone thinks that PRS is a severe localized scleroderma, this would not be wrong for the time being.

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