An overlap case of Parry–Romberg syndrome and en coup de sabre with striking ocular involvement and anti-double-stranded DNA positivity

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Parry–Romberg syndrome (PRS) may overlap localized scleroderma (morphea) lesions with linear depression (en coup de sabre [ECDS]). Overlap case with PRS and ECDS was presented. Enophthalmos, uveitis, ocular torticollis, keratic linear precipitates, and anti-double-stranded DNA positivity were identified. Subendothelial keratic precipitates detected by an *in vivo* laser scanning confocal microscopy were the first profiled in the literature. Patients must be evaluated and followed up carefully by their clinics to prevent misdiagnosis and unnecessary procedures such as surgery of ocular torticollis as muscular torticollis.

Keywords: Anti-double-stranded DNA, en coup de sabre, keratic linear precipitates, Parry–Romberg syndrome

Progressive hemifacial atrophy, also known as Parry–Romberg syndrome (PRS), is an acquired disorder with unknown etiology. It is characterized by unilateral facial atrophy, but

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arms, trunk, or legs can also be affected. It usually begins in the first decade of life and is more common in females.[1,2] En coup de sabre (ECDS) presents as a localized scleroderma (morphea) lesion with linear depression. It generally locates on the frontoparietal scalp or paramedian forehead.[3] PRS and ECDS may overlap in the same patient. Hair and ophthalmological alterations may accompany PRS, and the most frequent finding is enophthalmos.[4] There have been many reports of progressive hemifacial atrophy with the presence of some autoantibodies. However, antibodies to double-stranded DNA (dsDNA) in PRS are rare.[5] In this report, we present a case of overlapping of PRS and ECDS associated with enophthalmos, uveitis, keratic linear precipitates, ocular torticollis, and anti-dsDNA antibody positivity.

Case Report

A 16-year-old female patient was admitted to the dermatology department with right eyebrow hair loss. Her medical history showed that she had undergone a surgical treatment for congenital muscular torticollis on the left side of her neck 4 years earlier. On dermatologic examination, a hyperpigmented, indurated, atrophic linear plaque on the right paramedian area of the forehead was found. Band-like alopecia over the right eyebrow was also detected. Cutaneous tissue on the right side of her face was evaluated as asymmetric and atrophic. Ipsilateral enophthalmos and ocular torticollis accompanied these findings [Fig. 1a]. Linear atrophic plaque on her trunk was also detected. Syphilis and borreliosis (*Borrelia burgdorferi*) serology and purified protein derivative for tuberculosis were all negative. On ophthalmologic examination, uncorrected visual acuity was found to be 10/10 in both eyes according to the standard Snellen chart. Anterior segment examination and anterior chamber depth were normal in both eyes. Intraocular pressure was measured at 14–14 mmHg with Goldmann applanation tonometry. Gonioscopic evaluation of iridocorneal angles was also narrow. Hyperreflective linear subendothelial keratic precipitates were detected in only the right eye by *in vivo* laser scanning confocal microscopy (IVCM) (Heidelberg Retina Tomograph II Rostock Cornea Module) [Fig. 1b]. Antinuclear antibodies (ANA), anti-dsDNA antibody (by immunoblotting), and rheumatoid factor (RF) (nefelometric) were all positive. Serum complement levels were within normal limits. Magnetic resonance imaging of the brain was normal, but the skin on the right side was thinner than the left. The left orbit was located in the lower region of the face than the right one [Fig. 1c and d]. Histopathological examination of the morphea lesion revealed thickened, hypocellular, and swollen dermal collagen bundles. Dermal collagen elastic fibers presented with Verhoeff-van Gieson's stain [Fig. 1e and f]. The patient was diagnosed as PRS with the help of clinical, histopathological, and imaging findings. Oral methotrexate treatment (15 mg/week) was started to prevent progression of disease by the dermatologist. Stabilization of disease was obtained.

Discussion

PRS is typically restricted to one half of the face but occasionally also involves the arms, trunk, and legs.[2,6,7] It slowly progresses over 2–20 years before stabilizing. An early sign of PRS is a frontal hypo- or hyperpigmented skin lesion known as frontal linear scleroderma (ECDS). It is a localized form of scleroderma, as in our case.[8] While some clinical features may distinguish ECDS from PRS, clinical and histopathologic findings often overlap in the same patient. Thus, overlapping clinical manifestation supports the belief that ECDS and PRS are on the same spectrum of disease.[8]

Laboratory work on patients with PRS as well as other types of localized scleroderma largely tends to be unrevealing. While ANA positivity is the most common laboratory abnormality, serology for RF, anti-dsDNA antibody,[5] can rarely be abnormal. Garcia-de la Torre *et al.* reported ANA positivity as 57%, RF positivity as 36%, anti-histone antibody positivity as 21%, and anticentromere antibody positivity as 14% in their PRS series, and they did not find any anti-dsDNA antibody.[9] ANA, anti-dsDNA, and RF were positive in our patient. Although anti-dsDNA is a characteristic finding for systemic lupus erythematosus (SLE), we did not found any clinical sign of SLE in our patient. Difference in autoantibodies between localized scleroderma and PRS is not clear. However, coexistence of these antibodies with PRS may confirm that this disease and ECDS represent overlapping conditions. In case of observing serological abnormalities in PRS, it can be offered that this should be an autoimmune disease.

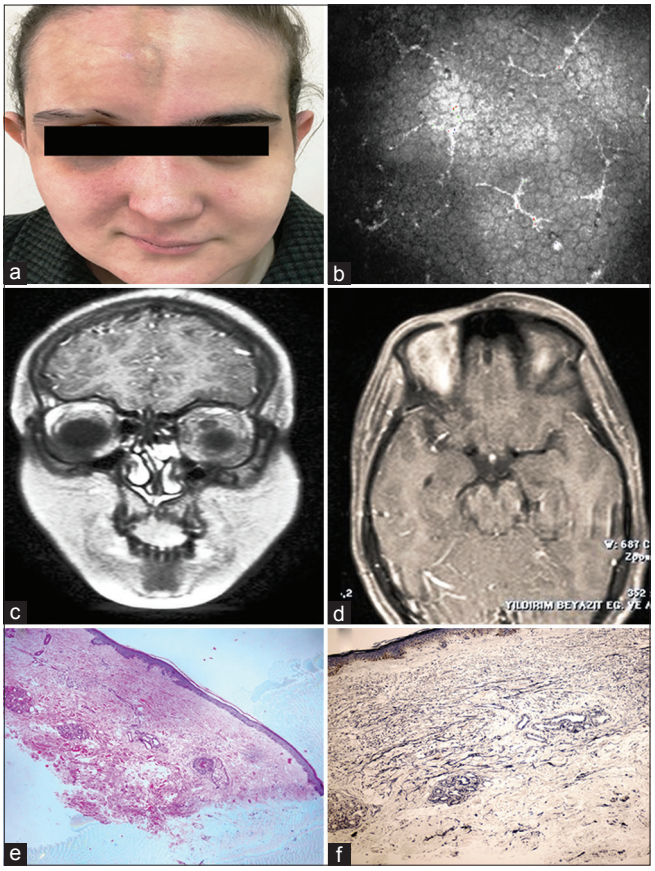

Figure 1: Unilateral facial hemiatrophy, central madarosis, ipsilateral enophthalmos, and ocular torticollis (a). Hyperreflective linear subendothelial keratic precipitates with in vivo laser scanning confocal microscopy (b). Magnetic resonance imaging of the brain was normal, but skin on the right side was thinner than the left side and the left orbit was localized on more below than the right orbit (c and d). Dermal collagen bundles thickened hypocellular and swollen (H and E, ×40) (e). Dermal collagen elastic fibers (Verhoeff-van Gieson, ×100) (f)

Ocular problems may be seen in patients with PRS.[4,6] Because of unilateral facial hemiatrophy and ipsilateral enophthalmos, abnormal head posture (ocular torticollis) is adopted to improve visual acuity and maintain binocular single vision. Keratic linear precipitates in our case were probably associated with fuchs heterochromic iridocyclitis (FHIC). FHIC was previously reported in a case of PRS in 1970. Subendothelial keratic precipitates detected by IVCM in our patient with PRS were the first report in literature.

Conclusion

The treatment's aim is to stop the disease's progress through immunosuppression such as prednisone, methotrexate, and cyclosporine and to repair the deformities after stabilization.[4,6] Subdermal fat grafting can be used for atrophic sites;[10] however, there was no surgical indication for this patient in terms of dermatology and ophthalmology. Stabilization of disease was obtained with methotrexate. Autologous lipid grafting and esthetic surgery might be useful cosmetically.

Consequently, our patient had several exceptional features such as ocular torticollis, anti-dsDNA positivity, and presence of keratic precipitates. However, ocular torticollis may be misdiagnosed as muscular torticollis in some cases for a long time. Thus, patients must be evaluated and followed up more carefully with their clinical and laboratory findings.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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