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CASE REPORT

Mysterious Association of Systemic Lupus Erythematosis with Perry Romberg Syndrome

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INTRODUCTION

Parry Romberg syndrome (PRS), also referred to as progressive hemifacial atrophy, progressive facial hemiatrophy, or idiopathic hemifacial atrophy, was first described by C Parry and M Romberg.[1]

It manifests in the first two decades in morphologically normal-born individuals. It commonly affects one or more dermatomes in the trigeminal nerve territory. It has an early onset of ophthalmic and neurological involvement and a variable maxillo-facial or cardiac involvement. It is an idiopathic, gradually progressive craniofacial asymmetry, following the atrophy of subcutaneous tissue, muscles, osseous, and cartilaginous structures. Ophthalmic involvement occurs in up to 35% of cases[2]. Neurological symptoms manifest in 15 to 20% of cases[3].

The incidence of PRS varies from 0.3 to 2.5 cases per 100,000 population per year, with a male: female ratio of 1 to 3.[3]The disease shows a progressive course, followed by a 'burn out' within 2 to 20 years and spontaneous remission[4].

While the underlying mechanisms are unknown, the disease has been linked to Linear Scleroderma (en coup de sabre) based on the detection of antibodies in the sera of known cases[5]. The major distinguishing feature is the absence of cicatricial alopecia and skin induration in PRS cases. In contrast, LSCS shows cutaneous sclerosis of the scalp, hyperpigmentation, and loss of scalp hair and eyebrows.

The clinical criteria, as proposed by Duymaz et al favoring a diagnosis of PRS over scleroderma, are[3]

1. Unilateral facial atrophy
2. Absent/minimal signs of prior inflammation or induration
3. Skin atrophy with thin, soft skin and no sclerosis.

The presence of other autoimmune diseases like systemic lupus erythematosus, rheumatoid arthritis, inflammatory bowel disease, ankylosing spondylitis, vitiligo, and thyroid disorders have been noted in rare cases.[5]PRS is primarily a clinical diagnosis. Various investigation modalities can be employed to rule out any overlap with similarly presenting syndromes.Anti-nuclear antibody (ANA): The most commonly associated serology finding in 25 to 52% of cases.[6]Rheumatoid factor: Elevated titers in localized scleroderma and LSCS cases.[7]Other serology tests: C-reactive protein, HLA-B27 typing, extractable nuclear antigen, Anti-Scl-70, Anti-cardiolipin, and Anti-dsDNA antibodies are of limited value.

Authors herein present the case of a 21-year-old female who later on diagnosed as PRS and positive with multiple auto-antibodies. To the best of our knowledge, the relationship between PHA and the aforementioned antibodies is only loosely established in literature; thus, the purpose of this case report is to highlight this novel finding in a patient who was otherwise asymptomatic for Systemic Lupus Erythematosus.

CASE REPORT

A 21-year-old female presented to the Rheumatology clinic with the complaints of progressive wasting of left side of face leading to facial asymmetry over a period of 2 years. It was slowly progressive and she does not has same type of wasting affecting other part of the body. Upon further questioning, she does not has H/o skin thickening, raynaud's dryness of mouth and eyes, joints pain, oral or genital ulcers, malar or body rashes, injury, trauma. Examination showing significant wasting of left side of face extending from forehead to the neck, while normal over the right side. There were no sensory loss, tingling sensations, pain, or paraesthesias on left side of the face. In hope of finding any nerve-related causes or space-occupying neurological lesions, she had undergone various diagnostic investigations including MRI of the brain which was unremarkable and Nerve Conduction Studies and Electromyography which turned out to be normal. Facial CT scans were also unremarkable. To rule out the possibility of arteritis, inflammatory markers such as CRP as well as complement levels, were tested and were found to be within normal limits. A raised titer of ESR of 44 with reactive Anti ds DNA antibodies and an incidentally positive ANA profile were the only laboratory abnormalities reported for this patient.

Cementing PRS as a diagnosis of exclusion. She was subsequently started on trial of Hydroxychloroquine; however, by the time of her next visit, she had developed loss of eyebrows and eyelashes on the left side. After thorough literature search, a trial of Prednisolone along with Methotrexate given but it fails to improve the symptoms and to halt the progression of the disease. Since the patient had no SLE related symptoms, as an alternative to oral medication.Cosmetic surgery pursued as a possible treatment option for her facial symptoms, it significantly improved after undergoing plastic surgery procedure.

DISCUSSION

Parry Romberg syndrome (PRS) or progressive hemi-facial atrophy is a rare clinical entity that remains an enigma in terms of diagnostic and therapeutic approaches for physicians all over the world. It is characterized by progressive wasting and shrinkage of one side of the face, and may extend to involve the underlying osseous as well as muscular structures. The disease entity

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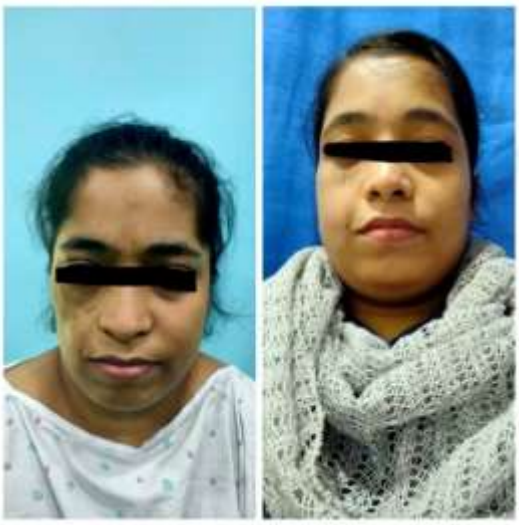
preferentially involves the left side of the face, and generally progresses rapidly over the first few years followed by a stabilization phase, all of which is coexistent with our case presentation.



Various systemic and cutaneous manifestations associated with this syndrome have been reported over the years, with Epilepsy being the most frequently observed neurological abnormality.[8] Ophthalmological defects, namely enophthalmos are also commonly observed as part of the disease spectrum.[9] Ipsilateral hair loss and dental abnormalities are some of the common presentations.[10]

No seizures, ocular or dental involvement were observed in our case, however a loss of subcutaneous tissue, loss of facial hair and scanty eyebrows were present

Although the underlying causes are largely unidentified, a largely accepted hypothesis is the autoimmune origin of this disease, with the strongest link being established between Scleroderma and PRS.[5] However very few studies have actually proven a significant correlation between positive antibody titers and PRS. In a serological study conducted on 14 patients with PRS, 57% of the patients were positive for ANA, while the second most commonly detected antibody was Rheumatoid factor[5] The only valid study illustrating a relationship between anti ds DNA antibody and PRS was perhaps the one conducted by Gonul et al[11].

Our case stands out from the previously recorded cases over Pakistan due tothe finding of positive anti dsDNA titer, as well as negative rheumatoid factor, despite the absence of any other systemic manifestations of SLE. Previous case reports have either recorded some trigger factors such as trauma or neurological findings, and have recorded remarkable response to Immunosuppressant medications such as methotrexate,[12]none of which were observed in our case.

This case report is the first of its kind, and aims to bridge the gap in knowledge that exists in literature about the association between SLE and Parry Romberg Syndrome, and hopes to highlight the importance of auto-antibodies as prognostic indicators in rare illnesses such as the above.

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