Unveiling Parry-Romberg Syndrome With Native Demyelinating Etiology as the Underlying Cause

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Abstract- Parry-Romberg Syndrome (PRS) is an atypical condition characterized by hemiatrophy of the face. Despite its rarity, the precise pathophysiological processes underlying its etiology remain elusive. While previous reports have hinted at the potential involvement of autoimmune factors, our case report aims to explore a novel perspective. Specifically, we pose the question of whether multiple sclerosis (MS) could be a plausible causative factor for PRS. Additionally, we investigate the potential association between Radiologically Isolated Syndrome (RIS) and the development of Parry-Romberg Syndrome.

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Introduction

Parry-Romberg syndrome (PRS), also referred to as progressive hemifacial atrophy, is an uncommon disorder characterized by one-sided facial atrophy. This condition typically impacts the skin, subcutaneous tissue, and underlying bone structures. The precise aetiology of PRS is still unknown, although potential contributors include trauma, infection, cerebral vascular malformation, and immune-mediated mechanisms. Pierre Robin sequence (PRS) often appears throughout infancy or adolescence, advances gradually, and may result in notable facial asymmetry and functional limitations. Treatment strategies concentrate on symptom management and may include reconstructive surgery, orthodontic therapies, and cosmetic operations. The condition is acquired, indicating that it is neither hereditary or congenital. The illness often starts during infancy or early adulthood, with facial atrophy commonly impacting the left area of the face.

Atrophy often occurs on one side (unilateral), and is commonly seen on the left side in patients. Involvement of the trunk and limb on the same side is uncommon, however 20% of cases were reported to be bilateral. Parry initially reported the disease in 1825, followed by Romberg in 1846.

The exact pathophysiology and etiology of PRS remain elusive, with various factors implicated. The condition involves the progressive breakdown and atrophy of skin, subcutaneous tissues, muscles, and underlying bone and cartilage. Several hypotheses exist regarding the etiology of PRS, including trauma, infection, cranial vascular malformation, and immune-mediated processes. Despite these associations, the specific triggers and mechanisms leading to PRS development are not well understood, contributing to the classification of the disorder as idiopathic in nature. Research on PRS remains ongoing, and the complexity of its pathophysiology suggests a multifactorial origin. Further studies are essential to unravel the precise molecular and cellular events contributing to the manifestation of Parry-Romberg Syndrome.

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Case Report

A 29-year-old woman without any previous medical conditions reported experiencing unilateral face depression in the temporal area for the last six months. No abnormalities were seen in the muscles and tissues of the nose, eyes, eyebrows, ears, and neck. There were no oral signs such as angular cheilitis or gingivitis. The history did not support trauma. She denied any familial history of autoimmune disorders.

Otorhinolaryngology and rheumatological tests (including scleroderma and vasculitis and others) were normal. Her laboratory tests for ANA, SCL70, anti-dsDNA, and TSH were negative, ruling out an underlying autoimmune illness.

A brain MRI was performed for the patient. He had some lesions with increased signal on FLAIR and T2 . Especially, the lesion in the pons could be the reason for the patient's symptoms. Suspected of MS, the patient underwent a lumbar puncture, which did not show anything in favor of infection in the tests, but OCB was reported positive and the IgG index in the CSF sample was high.

About 7 years ago, due to headaches, he had an MRI of the brain, which had exactly the same lesions.

Due to the normality of further examinations, brain MRI results, and presence of positive OCB, the patient was treated with dimethyl fumarate (Teczifuma).

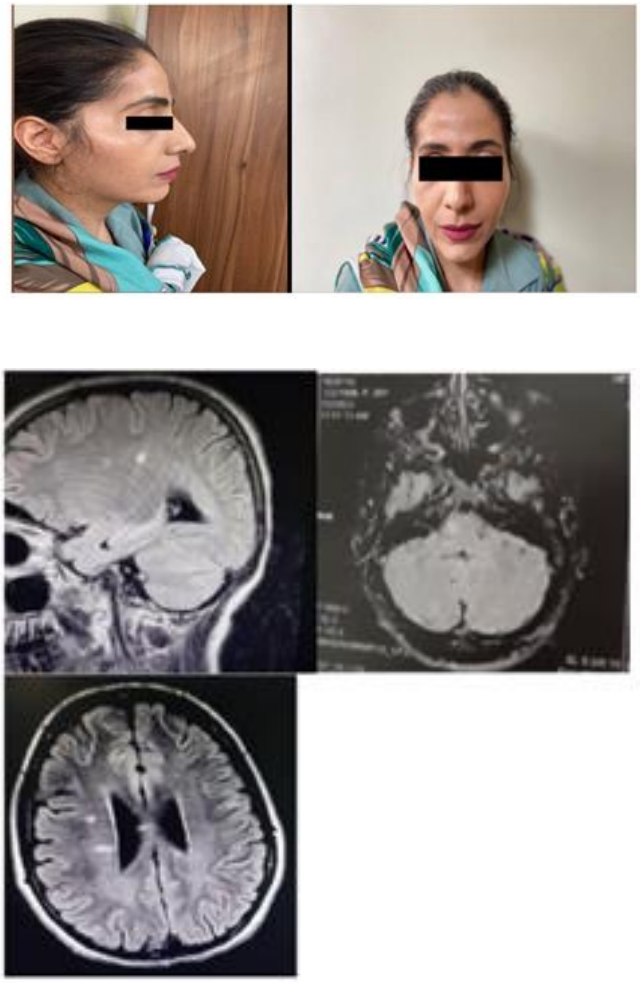

Figure 1.

Discussion

Parry-Romberg syndrome (PRS) is a relatively uncommon condition, particularly prevalent among women in the first decade of life (1-2). Neurological manifestations associated with PRS encompass cephalgia, tic douloureux, and partial epilepsy (2-4). The precise etiology and pathogenesis of PRS remain unclear, with hypotheses suggesting a link to trauma, genetics, infection, vascular malformations, endocrine

disturbances, elevated cervical sympathetic nerve activity, or autoimmune diseases (2-4). Viruses or bacteria are also considered potential contributors to PRS, with infections such as herpes zoster, Lyme disease, syphilis, rubella, tuberculosis, otitis media, and diphtheria implicated in its development (2-4). Certain PRS patients exhibit a "relapsing-remitting" course, a characteristic shared with multiple sclerosis (MS) (2-4). Diagnostic CT and MRI play a crucial role in identifying alterations in the central nervous system. These imaging modalities reveal distinctive features such as white matter hypersignal on T2-weighted and FLAIR sequences, leptomeningeal enhancement, intracranial calcifications, and brain atrophy. It is noteworthy that lesions observed in brain MRI of often manifest in regions similar to those identified in multiple sclerosis (MS) (2). Autoimmune pathogenesis is suggested to be associated with PRS, potentially implicating MS as one of the causal factors (2). Managing PRS poses a challenge, and treatment strategies aim to impede disease progression, as curative interventions are currently unavailable. Some success has been observed with immunosuppressive therapy in certain PRS patients (3-5).

The subtle nature of this rare condition often leads to its potential oversight or misdiagnosis during routine doctor-patient consultations. Recognizing the intricate manifestations requires a thorough examination, including a comprehensive analysis of the patient's medical background, scrutiny of prior images and a thorough physical examination to identify signs of progressive loss involving skin, fat, muscle, and bone. The objective of this case report is to present a distinctive instance of Parry-Romberg syndrome (PRS). Sharing such unique cases contributes to a broader understanding, and the accumulation of expert opinions and additional articles can enhance diagnostic accuracy and treatment efficacy for patients facing this challenging condition.

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