CASE REPORT

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The Case of a Melting Face: Parry–Romberg Syndrome

ABSTRACT

Background: Parry–Romberg syndrome (PRS), or progressive hemifacial atrophy, is a rare acquired disorder characterized by unilateral facial tissue loss of uncertain etiology. It is often associated with neurological, ocular, dental, and autoimmune manifestations.

Case Report: We report a 38-year-old woman with 12–13 years of progressive right-sided facial atrophy associated with enophthalmos and hypothyroidism. Clinical examination showed marked hemifacial atrophy involving skin, soft tissue, and jaw with no neurological deficits.

Conclusion: This case highlights the classical presentation of PRS with ocular involvement and autoimmune association. Early recognition and multidisciplinary management are essential to optimize functional and cosmetic outcomes.

Key words: Parry–Romberg syndrome, progressive hemifacial atrophy, enophthalmos, hypothyroidism, autoimmune

INTRODUCTION

Parry–Romberg syndrome (PRS), also known as progressive hemifacial atrophy, is a rare acquired disorder characterized by slowly progressive unilateral atrophy of facial tissues including skin, subcutaneous fat, muscle, cartilage, and bone.[1,2] The disease usually begins in the first two decades of life, shows a female predominance, and follows a progressive phase lasting 2–10 years before stabilizing.[1,2]

The etiopathogenesis remains unclear, with proposed mechanisms including autoimmune inflammation, neurogenic dysfunction, vascular abnormalities, genetic susceptibility, and sympathetic dysregulation.[2,3] PRS may be associated with neurological manifestations, ocular abnormalities, dental deformities, and autoimmune disorders such as hypothyroidism.[1,2,4] We report a case of long-standing hemifacial atrophy with ocular and maxillofacial involvement in a middle-aged woman with hypothyroidism.

CASE REPORT

A 38-year-old woman, a known case of hypothyroidism on regular thyroxine, presented with gradually progressive right-sided facial asymmetry for the past 12–13 years. The illness began with a sensation of tightness over the right jaw region, followed by progressive wasting of the right side of the face. This was associated with watering of the right eye and gradual sinking of the eyeball (enophthalmos).

There was no history of facial trauma, connective tissue disease, seizures, focal neurological deficits, or similar illness in the family. Systemic review was unremarkable.

On examination, marked facial asymmetry was noted with atrophy of the right cheek, temporal hollowing, and thinning of overlying skin (Figure 1). Right-sided enophthalmos with mild eyelid atrophy was present. The right jaw appeared smaller, with subtle deviation of the facial midline toward the affected side. No active inflammatory lesions or features of linear scleroderma were seen. Neurological examination was normal. Based on the characteristic clinical features, a diagnosis of Parry–Romberg syndrome was made. The patient was counseled regarding the disease course, need for follow-up, and reconstructive options after stabilization.

Figure 1: Marked facial asymmetry with atrophy of the right cheek, temporal hollowing, and thinning of overlying skin.

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DISCUSSION

Parry–Romberg syndrome is an uncommon disorder with an estimated prevalence of about 1 in 250,000.[1,2] It predominantly affects females and usually begins in childhood or adolescence, though adult-onset cases are described.[2]

Several mechanisms have been proposed. The neurogenic hypothesis suggests dysfunction of the trigeminal nerve or sympathetic chain leading to trophic changes.[2] Autoimmune mechanisms are supported by its association with morphea and the presence of autoantibodies in some patients.[2,4] Vascular abnormalities, including intracranial white-matter lesions, suggest an underlying vasculopathy.[2] Genetic susceptibility has also been proposed, with variants in MTOR and DHX37 genes reported.[3]

The presence of hypothyroidism in our patient supports the autoimmune overlap described in literature.[4]

PRS shows wide variability. Facial changes often begin with pigmentary alterations followed by progressive loss of subcutaneous fat and muscle, resulting in sunken cheeks and facial asymmetry.[1,2] Maxillofacial involvement may cause mandibular hypoplasia, dental root resorption, malocclusion, and temporomandibular joint dysfunction.[2]

Ocular involvement includes enophthalmos, ptosis, uveitis, retinal vascular abnormalities, and optic nerve involvement. Neurological manifestations such as headache, trigeminal neuralgia, seizures, and intracranial imaging abnormalities may occur. Our patient had classical facial atrophy with ocular involvement but no neurological manifestations.

Diagnosis is mainly clinical, supported by imaging when needed. Important differentials include linear scleroderma "en coup de sabre," morphea, hemifacial microsomia, and Rasmussen encephalitis.

There is no definitive curative therapy. Management is multidisciplinary involving neurology, dermatology, ophthalmology, and maxillofacial surgery. During the active phase, immunosuppressive therapy such as corticosteroids and methotrexate may be considered. After stabilization, reconstructive procedures like fat grafting, fillers, and orthognathic surgery can improve cosmetic and functional outcomes.[2]

CONCLUSION

Parry–Romberg syndrome is a rare progressive disorder with significant cosmetic, functional, and psychological impact. This case demonstrates classical hemifacial atrophy with ocular involvement and associated hypothyroidism, supporting a probable autoimmune link. Early diagnosis, regular follow-up, and multidisciplinary care are essential to optimize outcomes.

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