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ADVANCED PARRY RHOMBERG SYNDROME WITH HEMIFACIAL ATROPHY, DENTAL MALALIGNMENT AND UNILATERAL VISION LOSS: A RARE CASE REPORT

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Abstract

Parry Rhomberg syndrome, also known as Progressive Hemifacial Atrophy is a rare medical entity, characterized by progressive thinning of the skin tissues resulting in "sunken in" appearance of one half of the face with secondary neurological symptoms, most common being seizures. The disease usually presents in the second decade of life and follows a self limiting course. There are no definite diagnostic criteria for the disease; clinical features and imaging modalities remain the mainstay for the diagnosis. Treatment follows a multi-modality approach, consisting of management of complications and reconstructive therapies. We hereby describe a unique case of a 45-year-old female with left hemifacial atrophy and misaligned teeth complicated with enophthalmos and extensive keratopathy leading to vision loss in the left eye; presenting as generalized tonic clonic seizures raising high suspicion of Parry Rhomberg syndrome. Parry Romberg syndrome, though rare and diagnostically challenging, requires early recognition; timely medical and reconstructive management can improve long-term outcomes.

Keywords: Parry Romberg syndrome, hemifacial atrophy, dental malalignment, enophthalmos, keratopathy, unilateral vision loss, seizures.

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Introduction

Dr Caleb Hiller Parry and Moritiz Heinrich Rhomberg first described this rare condition characterized by progressive self limiting hemiatrophy of the face. PRS usually presents in children and young adults with female preponderance (M:F -1:3). The disease typically involves skin and underlying subcutaneous tissue which may later progress to cartilage or bone involvement. Secondary complications commonly include neurological features like seizure, headache, trigeminal neuralgia and ophthalmological features like enophthalmos and keratitis [1]. The exact pathogenesis has not been defined but multiple theories have been suggested such as an autoimmune mechanism supported by the presence of autoantibodies and response to immunosuppressive treatment, neurogenic or sympathetic dysfunction, vascular and exogenous triggers such as infection (e.g., Borrelia, herpes) or local trauma. Together, these suggest a multifactorial pathogenesis. Diagnosis is primarily based on clinical history and physical examination. Imaging modalities like MRI may be used to assess neurological involvement [2]. We hereby report a case of Parry Rhomberg syndrome presented with generalized tonic clonic seizures having left hemifacial atrophy and misaligned teeth complicated with enophthalmos and extensive keratopathy leading to vision loss in the left eye.

Case Description

A 45year female, presented in January 2025 to Swaroop Rani Nehru Hospital attached to Moti Lal Nehru Medical College Prayagraj/Allahabad in Emergency ward with complain of 2 episodes of generalized tonic clonic seizures in past 24 hours. Patient had a skin lesion in the parotid area eight years back and was in an illusion that post inflammatory scar has lead to hemiatrophy of left face otherwise there were no other co morbidities. There was no significant family history. On physical examination- left hemifacial atrophy with misaligned teeth was seen. Vitals were normal and no abnormality was found on systemic

examination except a poor communicating skill. Upon ophthalmological examination enophthalmos and keratitis were observed with vison loss in left eye (Figure 01). Retina of left eye could not be visualized due to corneal opacity and right eye was normal. Scleroderma, Rassmussen encephalitis, Hemifacial microsomia and Herpes zoster ophthalmicus with CNS involvement were considered as differentials. Diagnostic work up revealed all laboratory parameters including CBC, LFT, KFT and inflammatory markers (ESR, CRP) within normal limits. This patient had a negative autoimmune panel. Radiological investigations comprised X ray of the face, 3D CT face and NCCT head. NCCT head confirmed enophthalmos and revealed an area of gliosis in the left parietal region (Fig 2). 3D CT face demonstrated marked hemifacial atrophy indicated by hypoplastic left maxillofacial structures (Fig 3a and 3b)-findings consistent with Parry–Romberg syndrome. The patient was managed conservatively with IV levetiracetam 500mg q8h, sodium valproate 500mg q8h and ceftriaxone 1gm q12h for 7 days of hospitalization. Her condition gradually improved with no further episodes of GTCS after initiation of antiepilepticsand she was later discharged under stable condition on oral antiepileptics.

Fig 01: Left hemiatrophy with keratitis and enophthalmos with misaligned teeth

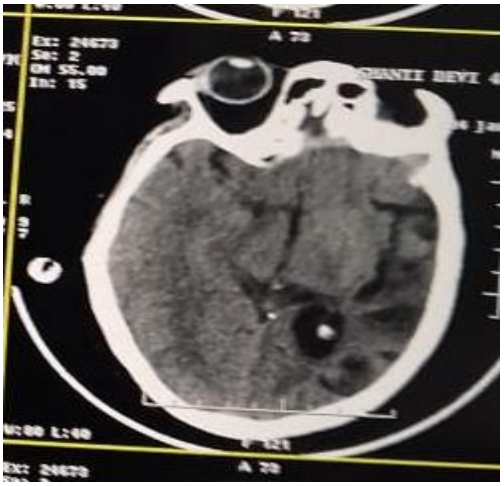

Fig 2: Gliosis in left parietal region with marked Left enophthalmos

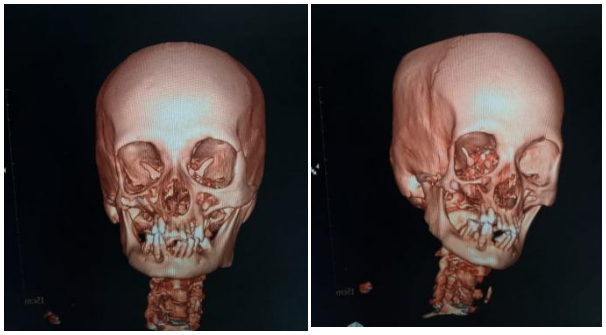

Fig 3a Fig 3b

Fig (3a, 3b): 3D CT Face showing left hypoplastic maxillofacial structures.

Discussion

Parry–Romberg syndrome (PRS) is a raredisorder marked by progressive hemifacial atrophy with incidence ranging from 0.3 to 2.5 cases per 100,000 population per year. It typically begins in childhood or adolescence and then gradually evolves over a course of 2 to 20 years. Patients exhibit unilateral atrophy of skin, subcutaneous fat, muscle, cartilage, and bone-usually along trigeminal dermatomes [3]. Common ocular findings include enophthalmos, keratitis, and corneal disease. Neurological symptoms such as seizures (usually refractory), migraines, trigeminal neuralgia and hemiparesis of contralateral side occur in a significant number of cases. Opthalmic involvement occurs in 10-30% of patients of PRS on the ipsilateral atrophic side common being enophthalmos, uvieitis and retinal damage etcetera [4]. Imaging studies like MRI or CT often show cortical atrophy, white matter lesions, or calcifications. The disease usually stabilizes within several years, but relapses can occur [5]. The pathogenesis of PRS remains poorly understood and is thought to be multifactorial. Proposed mechanisms include autoimmune processes, sympathetic nervous system dysfunction, neurovascular inflammation, mechanical injury or infection, and developmental anomalies. Histologic examination often shows fat loss, fibrosis, collagen deposition, and varying inflammation [6]. Clinically, PRS is categorized in 4 types based on tissue involvement: Type 1 and 2 affect only the skin and soft tissue, while Type 3 and 4 are more advanced and involve deeper structures like cartilage and bone, requiring more extensive reconstructive approaches [7].

Progressive hemiatrophy of the face, as seen in PRS must be distinguished from several conditions like linear scleroderma- en coup de sabre, which also causes progressive skin and soft-tissue atrophy but typically lacks deeper tissue or bony involvement with positive autoimmune panel [8]. Another differential is Rasmussen encephalitis, characterized by unilateral cerebral inflammation, intractable seizures, and cortical atrophy, but cutaneous changes are usually absent [9]. Other considerations include hemifacial microsomia and Herpes zoster ophthalmicus with CNS involvement. Hemifacial microsomia is a congenital disorder characterised by unilateral facial hypoplasia with non progressive course

unlike PRS [10]. While Herpes zoster ophthalmicus can be ruled out on the basis of absence of typical vesicular rash in trigeminal distribution. Cutaneous signs, pattern of progression, and neuroimaging findings will help in differentiation and accurate diagnosis.

There is no standard treatment guideline for PRS, however a multidisciplinary approachis needed. Active disease phase can be managed with immunosuppressive therapies like methotrexate (0.3-1mg/kg/week), oral steroids such as prednisolone(1mg/kg/day), cyclosporine, mycophenolate mofetil and cyclophosphamide with variable results [8]. Neurological symptoms, like seizures, are treated with antiepileptic medications and, in refractory cases, neuromodulation techniques like vagus nerve stimulation can be used. Ocular complications such as keratitis, uveitis, and enophthalmos should be managedwith topical or systemic therapies, and surgery when required. Various strategies are being used currently for cosmetic reconstruction such as autologous fat grafting, dermal grafts, silicone implants, and tissue flaps [11].

Our case was unique as it has the worst debility of vision loss of left eye due to enophthalmos and keratopathy as a complication of PRS having the full blown hemifacial atrophy and misaligned teeth. Most cases of PRS suffer mental trauma due to face disfigurement but this patient had this add on visionary challenge.

Conclusion

Parry Rhomberg Syndrome is a rare entity with no definite diagnostic criteria and varied symptomatology including neurological, ophthalmological and cutaneous manifestations. This case highlights the need of high index of clinical suspicion for PRS. This case presenting with new onset seizures was unique regarding visionary loss of left eye due to enophthalmos and keratopathy besides severe hemifacial atrophy and dental malalignment. Early recognition and comprehensive approach consisting of pharmacological and reconstructive therapies can optimize long term outcomes and form the cornerstone of management of PRS.

Funding

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Acknowledgement

None

Conflict of Interest

None

Informed Consent

Yes

Ethical Statement

Not applicable as it is a case report and identity of the case reported was not disclosed.

Author Contribution

Ashish Rai and Ayushi Krishna Evaluated the case. All managed the case. All wrote manuscript. Ayushi Krishna and Ashish Rai collected the pictures. All reviewed the manuscript.

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