International Journal of Science and Research (IJSR)
ISSN: 2319-7064
SJIF (2022): 7.942

Rare Disease Entity - Parry Romberg Syndrome: A Case Report

Jasneet Kaur Sodhi[1], Alex Rebello[2], Dr Manmeet Singh[3]

[1]Senior Resident, Department of Neurology, AIIMS, Mangalagiri, Andhra Pradesh
Corresponding Author Email ID: *drjasneet[at]yahoo.com*

[2]Assistant Professor, Department of Neurology, AIIMS, Mangalagiri, Andhra Pradesh

[3]Fellow Cornea & Refractive Services, Sankara Academy of Vision, Sankara Eye Hospital, Guntur, Andhra Pradesh

Abstract: ***Parry-Romberg syndrome is a degenerative disorder leading to progressive, usually unilateral atrophy of skin, soft tissues and underlying structures of the face and neck. It can also have ocular and neurological involvement.***

Keywords: Parry - Romberg, progressive, Hemifacial atrophy

1. Case

We present a case report of 33 year old female who presented to us with the history of tingling sensation and progressive atrophy of right side of face since 10 years. Examination revealed facial asymmetry and muscle wasting on right side. This is a rare disorder resulting in progressive hemifacial atrophy that has multisystemic effects. Therefore, we need a multidisciplinary approach along with more studies to identify the fundamental etiology and formulate proper management guidelines.

2. Introduction

Parry–Romberg Syndrome (PRS), also known as Progressive Hemifacial Atrophy (PHA) is a rare condition characterized by progressive atrophy of the skin and soft tissues including muscles and bones of the face and neck, usually involving one side.1 The affected individuals are morphologically normal at birth, the atrophy being insidious, occurring within the first two decades of life. This is usually limited to the face, but rarely involves unilateral limbs. It exhibits similarity to localized scleroderma, with atrophy of skin and subcutaneous fat, and rarely muscles and bone. After a progressive phase which may span up to 20 years, the process stabilizes. The deformities suffered are usually permanent. Seen more often in females, it commonly affects the eyes and is sometimes (15%) associated with neurological disorders like trigeminal neuralgia, facial paresthesia, headache, and focal epilepsy.[2] In severe forms, ophthalmic involvement in the form of enophthalmos, strabismus and heterochromia may also be seen.[3]

The aetiology of PRS remains largely unknown and seems to be heterogeneous. Trauma, infection by slow viruses or bacteria, cranial vascular malformation, immune - mediated process, disturbances of fat metabolism, and sympathetic dysfunction have been proposed. The pathogenesis remains unknown.[4] The diagnosis is made by history and examination, further supported by findings of CT or MRI.

3. Case

A 33 year old female presented with the complaint of tingling over right side of face since 10 yrs. It was associated with gradually progressive atrophy of right side of face. Patient had no co - morbidities and was on no medications. Clinical examination revealed a healthy woman with normal vital signs and systemic presentation. The face was asymmetric. There was flattening of forehead on right side, enophthalmos of right eye, depressed right ala of nose, depressed cheeks and depressed chin on right side. There was also a scar - like defect prominent in the symphyseal region (*coup de sabre*) and minimal tongue atrophy on the same side. There was no hyperpigmentation, ocular movements and facial nerve were normal on examination. There were no focal neurological deficits. Routine blood investigations were within normal limits. ANA by IFA was negative. ESR and CRP were normal. MRI c spine was normal. MRI brain showed hypoplastic right orbit with normal sized globe, while there was no significant abnormality of brain parenchyma (Figure 1). Based on history, clinical and radiological findings the diagnosis of progressive hemifacial atrophy was made. Since patient was in her remarkable health, facial deformity was the major concern for the treatment. Hence, patient was given the option of trial of immunosuppresants and surgery once the disease progression is arrested. Patient was then advised for facial physiotherapy and rehabilitation as a part of palliative treatment.

4. Discussion

PRS is a rare disease with female predilection seen in 1: 70000 of the population. It is seen commonly on the left side of the face with onset usually in the second decade of life and a variable rate of progression.5 Historically, although a debate existed as to whether PRS was a form of linear scleroderma morphea en coup de sabre (ECDS) or the two conditions were clinically distinct entities, it is now well known that both PRS and ECDS lie on the same disease spectrum of localized scleroderma and may even coexist in the same patient.[6]

Although etiology is not clear, Many authors propose trophic malfunction of sympathetic nervous system as a cause, since normal development of skin, muscle, and bone require trophic stimulation.[7] The destruction of skin and osseocartilaginous structures is the hallmark of this syndrome with protean systemic manifestations, usually neurological and ophthalmic.[5] The only manifestation in this case was hemifacial atrophy without systemic involvement. However, since it is a progressive condition, systemic involvement may occur over a period of time. The diagnosis is mainly clinical. The disease is self - limiting, and has no definite cure. The active stage of the disease is usually treated with corticosteroids and immunosuppressant therapy. Once the deformities have set in, plastic and reconstructive surgeries are recommended.[8, 9] Surgical management should not be considered until after the disease is inactive to avoid the potential for triggering a flare of the disease, and worsening morbidity. The main aim of surgery is to improve the cosmetic disfigurement and minimize the psychosocial effects of the disease.[10]

5. Conclusion

Parry Romberg syndrome is a progressive, disfiguring disease of uncertain etiology. The present case was diagnosed after 10 years since the onset of clinical features and is undergoing treatment. If diagnosed earlier in life, the developing facial deformity could have been arrested. More research is needed for better treatment modalities to prevent cosmetic disfigurement, complications and psychosocial repercussions of the disease.

Declaration of patient consent: The patient has given consent for his images and other clinical information to be reported in the journal. The patient understands that his name and initials will not be published and due efforts will be made to conceal his identity but anonymity cannot be guaranteed.

Financial support and sponsorship: Nil

Conflicts of Interest: There are no conflicts of interest

References

[1] Wong M, Philips CD, Hagiwara M, Shatzkes DR. Parry Romberg syndrome: 7 cases and literature review. AJNR Am J Neuroradiol 2015; 36: 1355–61.
[2] Buonaccorsi S, Leonardi A, Covelli E, Indrizzi E, Perdicchi A, Fini G. Parry - Romberg syndrome. J Craniofac Surg 2005; 16: 1132 - 5.
[3] Deshingkar SA, Barpande SR, Bhavthankar JD, Humbe JG. Progressive hemifacial atrophy (Parry - Romberg syndrome). Contemp Clin Dent 2012; **3**: S78–81.
[4] El - Kehdy J, Abbas O, Rubeiz N. A review of Parry - Romberg syndrome. J Am Acad Dermatol 2012; 67: 769 - 84.
[5] Tolkachjov SN, Patel NG, Tollefson MM. Progressive hemifacial atrophy: a review. Orphanet J Rare Dis 2015; **10**: 39.
[6] Duymaz A, Karabekmez FE, Keskin M, Tosun Z. Parry - Romberg syndrome: facial atrophy and its relationship with other regions of the body. Ann Plast Surg 2009 Oct 1; **63**: 457–61.
[7] Cory RC, Clayman DA, Faillace WJ, McKee SW, Gama CH. Clinical and radiologic findings in progressive facial hemiatrophy (Parry - Romberg syndrome). AJNR Am J Neuroradiol 1997; 18: 751 - 7.
[8] Padmavathy J, Rao LL. Unilateral linear pansclerotic morphea affecting face and limbs. Indian J Dermatol Venereol Leprol 2005; 71: 192 - 4.
[9] Sudarshan R, Annigeri RG. Parry Romberg syndrome. Wien Klin Wochensch 2010; 122: 606.
[10] Arnold N, Shareef S, Sikorski LA. Unique case of progressive hemi - facial atrophy successfully treated with methotrexate. Spartan Med Res J 2017; **1**: 63–72.

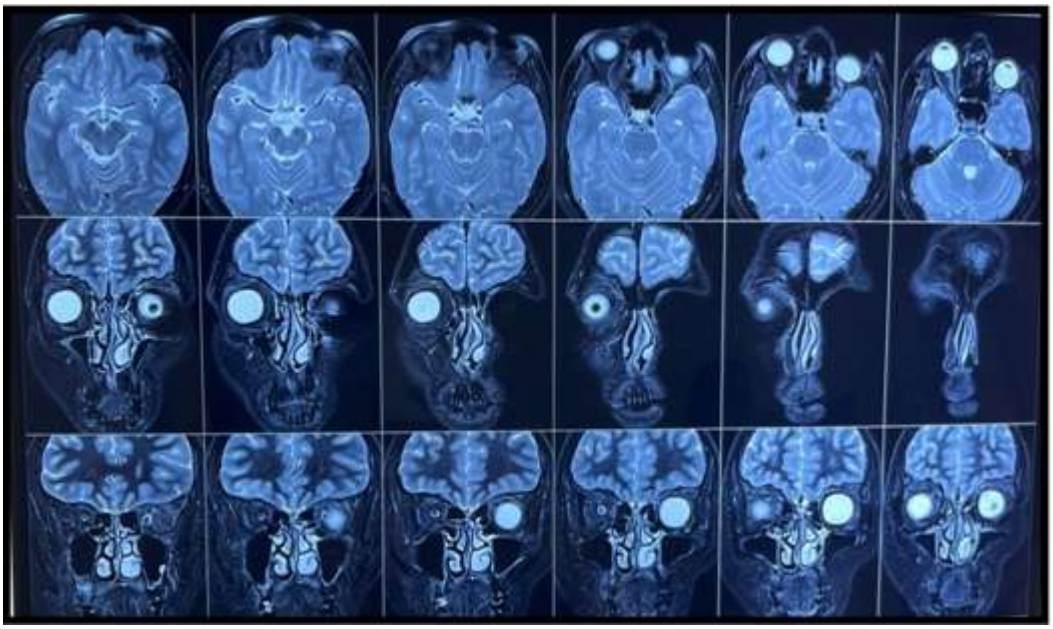

Figure 1: MRI brain with orbits (showing hypoplastic right orbit)