OMCR

Oxford Medical Case Reports, 2020;2020,1–3

doi: 10.1093/omcr/omz127
Advance Access Publication Date: 31 January 2020
Case Report

CASE REPORT

Parry–Romberg syndrome in an adolescent: a case report on progressive hemifacial atrophy

Ranjini Srinivasan and Shasta Theresa Joy*

Department of Pediatrics, St. John's National Academy of health sciences, Sarjapur Main Rd, John Nagar, Koramangala, Bengaluru 560034, Karnataka, India

*Correspondence address. Department of Pediatrics, St. John's National Academy of Health sciences, Sarjapur Main Rd, John Nagar, Koramangala, Bengaluru 560034, Karnataka. Tel: +919886496607; E-mail: joyshasta@gmail.com

Abstract

Parry–Romberg syndrome is a rare degenerative disorder causing progressive atrophy of skin and soft tissues of the face and neck, which is usually unilateral. The mean age of onset is usually in the second decade of life and the disease causes functional, aesthetic and psychological disabilities in the affected individual. We present a 14-year-old boy with this disorder. The diagnosis was based on clinical characteristics. A multidisciplinary team approach involving rheumatologists, dermatologists, maxillofacial surgeons, dentists and psychologists is required for the management of this problem, which is mainly targeted at controlling active inflammation with the use of immunosuppressive agents in addition to possible surgical correction of repositioning of adipose tissue that is lost due to atrophy.

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, more commonly the left. This disorder was first described in 1825 by Caleb Parry and later by Moritz Romberg in 1846 [1]. The title 'PHA' was given by the German neurologist Albert Eulenburg in 1871. Being an uncommon rheumatic disease, we wish to describe a case report of an adolescent who presented to our Paediatric Department.

CASE REPORT

A 14-year-old adolescent male presented with progressive atrophy of the right side of the face of 6 months duration. He was referred by his primary care paediatrician to our tertiary hospital for evaluation and treatment. The atrophy seemed to involve the right cheek predominantly extending down to the chin. There was no history of any febrile illness, trauma or skin rash/pigmentation preceding the atrophy. The patient had no neurological symptoms before or after the onset of the atrophy. His birth and development history were normal and scholastic performance was satisfactory. His past medical history and family history was not significant. On examination, there was facial asymmetry with wasting of the muscles involving the right side of the face extending from the right cheek below the eye to the angle of the mouth. There was no hypo or hyperpigmentation of the overlying skin. His dentition was normal and there was no atrophy of the tongue. Neurological examination including cranial nerves revealed no deficits. The rest of systemic examination was normal. Computerized Tomography of the brain revealed atrophy of the soft tissues including muscles on the right side of the face. Antinuclear antibody and serological workup for other auto antibodies were negative. Blood investigations revealed normal blood counts and a normal ESR. With this, a clinical diagnosis of PRS was made and he was started on oral methotrexate six months ago and is yet to be reviewed with us. Augmentation surgery was planned after the process of atrophy completely stops.

Received: June 11, 2019. **Revised:** October 25, 2019. **Accepted:** September 16, 2019

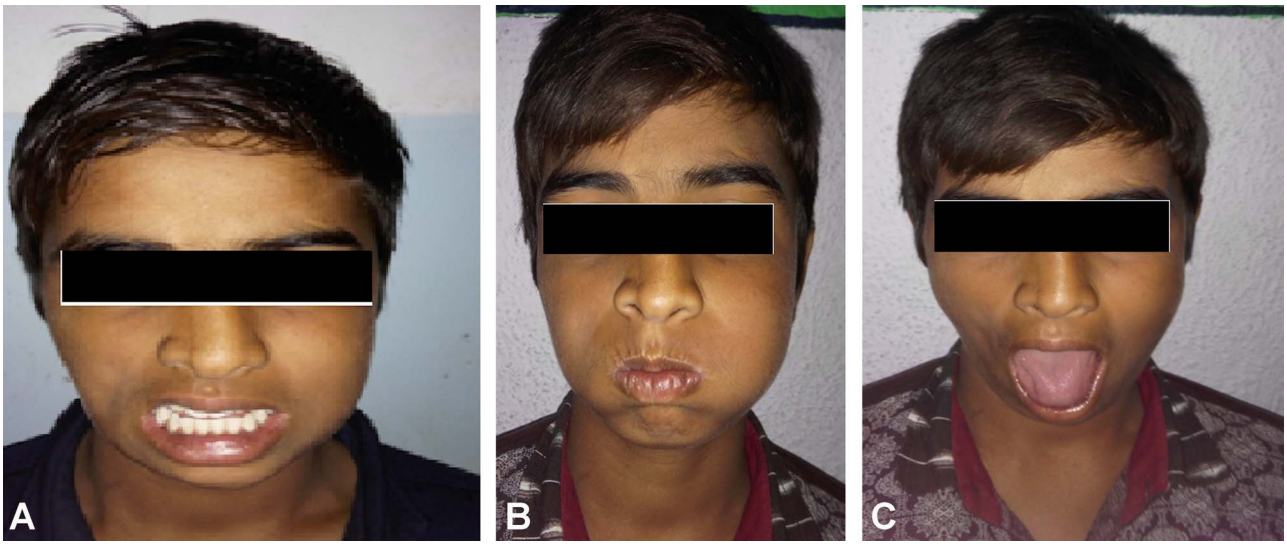

Figure 1: A 14-year male with PRS. a) Right hemifacial atrophy, b) Facial muscle atrophy and c) tongue was spared at presentation

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 between two and ten years following which the disease process arrests in most patients [2]. However, in a small subset of population, the atrophy may occasionally reactivate or accelerate later in life although this is rare. In some cases, disease flare or worsening may be associated with stress including surgery [3]. Our child was an adolescent male who presented with slowly progressive hemifacial atrophy of the right side of 6 months duration. The underlying mechanism is still under debate. Infection, vasculopathy, auto immunity, cerebral fat metabolism disturbances and autonomic dysregulation are among the proposed theories [2, 4]. The destruction of skin and osseocartilaginous structures is the hallmark of this syndrome with protean systemic manifestations. [2]. There are varied neurological manifestations that accompany 20% of the patients with this disorder including headache, trigeminal neuralgia, seizures and occasionally cranial nerve palsies [5]. In severe forms, ophthalmic involvement in the form of enophthalmos, strabismus and heterochromia may also be seen [6]. The less frequent ocular findings include cataract, glaucoma, uveitis and papillitis. Dental involvement in the form of overcrowding and short crowns and roots of teeth may be observed in some patients. Cognitive and behavioural problems have also been reported [2]. The unilateral skin and soft tissue degeneration of the face was the only manifestation in this case without neurological, ophthalmic or dental involvement. However, since it is a progressive condition, other parts of the face or other systems may get involved over a period of time.

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. Duymaz *et al*. [7] proposed certain criteria to be applied when evaluating a patient with hemifacial atrophy to assess whether the patient had PHA or ECDS. Accordingly, a patient with PHA presents with unilateral atrophy without preceding induration or inflammation. On the other hand, in scleroderma there might be unilateral band like changes of sclerosis and hyperpigmentation with induration. However, 28–42% of patients can have PHA with localized scleroderma. Therefore, although there may be some clinical features that differentiate ECDS from PRS, it is now accepted that the two conditions fall on the spectrum of the same disease.

The other differentials considered are rare syndromes like Goldenhar, which manifests at birth but is non-progressive and Barraquer–Simons syndrome that resembles PRS but has bilateral involvement of the face [8].

The diagnosis of PRS is mainly clinical and investigations are used mainly to differentiate it from other entities as discussed above. Radiological studies only provide information on the severity of atrophy based on imaging and may be warranted when considering surgical options. The radiological features include maxillary mandibular hypoplasia and facial muscle atrophy as seen in our case with the more severe phenotype involving the orbit. Those with neurological symptoms may display hyperintensities on magnetic resonance imaging [9].

With regard to management, it is always a challenge and involves a multidisciplinary team. The initial goal of treatment is to slow down or halt the disease progression. Methotrexate at a dose of 15 mg/m^2 given orally or as a subcutaneous injection once a week in combination with a systemic corticosteroid at a dose of 0.3–1 mg/kg/week given orally or as an injection is recommended as first line treatment for active disease. Other drugs like hydroxychloroquine, mycophenolate mofetil, cyclosporine and cyclophosphamide may be tried in those patients who have not responded to methotrexate. [2, 10]

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 [11]. Facial augmentation procedures include dermal fat grafts, autologous fat grafts, muscle flap grafts, free silicone injections, and bone augmentations [1]. Our patient was started on methotrexate since the disease was still active and he was advised to follow up regularly. The surgical options were recommended after the condition plateaued.

CONFLICT OF INTEREST

None declared.

FUNDING

The authors report no targeted funding for this work.

ETHICAL APPROVAL

The authors declare that no formal ethical approval was needed for this work.

CONSENT

The patient was adequately informed by the corresponding author regarding the potential publication of this case and the photograph submitted here and has given his written consent.

GUARANTOR

The first author guarantees for the accuracy of the data and the article.

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. doi: 10.3174/ajnr.A4297.
2. Tolkachjov SN, Patel NG, Tollefson MM. Progressive hemifacial atrophy: a review. *Orphanet J Rare Dis* 2015;**10**:39. doi: 10.1186/s13023-015-0250-9.
3. Stone J. Parry-Romberg syndrome: a global survey of 205 patients using the internet. *Neurology* 2003;**61**:674–6. doi: 10.1212/WNL.61.5.674.
4. Bharath, Naik S, Revathy V, Shashibhushan, Kumar MSS. Parry Romberg syndrome: a case report. *J Case Rep Stud* 2016;1:104. doi: 10.15744/2348-9820.4.104.
5. Vix J, Mathis S, Lacoste M, *et al*. Neurological manifestations in Parry–Romberg syndrome: 2 case reports. *Medicine* 2015;**94**:e1147. doi: 10.1097/MD.0000000000001147.
6. Deshingkar SA, Barpande SR, Bhavthankar JD, Humbe JG. Progressive hemifacial atrophy (Parry-Romberg syndrome). *Contemp Clin Dent* 2012;**3**:S78–81. doi: 10.4103/0976-237X.95111.
7. 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.
8. Latha S, Jain S, Sur J, Khan F, Jain S, Ratna S. Parry Romberg syndrome: a case report and review. *IJADS* 2016;**2**:75–8.
9. Cory RC, Clayman DA, Faillace WJ, McKee SW, Clinical GCH. Radiologic findings in progressive facial hemiatrophy (Parry-Romberg syndrome). *Am J Neuroradiol* 1997;**18**:751–7.
10. Zulian F, Culpo R, Sperotto F, Anton J, Avcin T, Baildam EM, *et al*. Consensus-based recommendations for the management of juvenile localised scleroderma. *Ann Rheum Dis* 2019;**78**:1019–24. doi: 10.1136/annrheumdis-2018-214697.
11. 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.