A case report of Parry-Romberg syndrome

Kiana Babaei [1], Nora Shurvarzi [1], Ali Rahnama[1], and Ali Movahedi[1]

[1]Neyshabur University of Medical Sciences

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Introduction

Parry-Romberg syndrome (PRS) is characterized by progressive dystrophy or loss of subcutaneous tissue in one half of the face, usually beginning in childhood and often continuing with skin changes[1]. This atrophy affects the subcutaneous tissue, fat, muscle and bone-cartilaginous structures and creates a sunken appearance in the face[2]. This syndrome is often associated with linear scleroderma and is also known as En coup de sabre [3]. The clinical feature of PRS that makes it possible to diagnose is unilateral idiopathic facial atrophy[4]. This disease is self-limiting and its treatment is multidisciplinary[5]. Treatment is usually based on the replacement of adipose tissue that has been lost due to atrophy [6]. Surgical treatment for PRS often requires a multispecialty approach with repeated procedures, depending on the extent of involvement[7]. The goal of surgical treatment for PRS patients is to minimize the psychosocial effects and correct the appearance and function of the involved facial structures[5].

Case history

A 14-year-old male patient with a complaint of unilateral facial atrophy was referred to a specialist in cosmetic and plastic surgery at Hakim Hospital in Neyshabur, Iran. From the age of 7, the patient gradually experienced discoloration in the forehead area, which was gradually accompanied by atrophy in this area. Over time, the fat tissue completely disappeared and progressed to the frontal bone area. The patient had passed the stages of development normally. The mother's gestational age was full and he was born through natural delivery. There were no congenital anomalies in the family members. Additionally, there was no history of convulsions, connective tissue or systemic disease in the family. The patient had no history of underlying disease, surgery or hospitalization, drug use, or food and drug allergy. After the clinical examination, the doctor referred the patient to a rheumatology specialist, a cardiologist and a radiologist specialist.

Methods

According to the results of computed tomography (CT) scan, Magnetic resonance imaging (MRI), ultrasound and clinical examinations, the patient was diagnosed with PRS. In laboratory findings, antinuclear antibody (ANA) was normal (Table 1). All organs were normal on abdominal ultrasound. Electrocardiography and echocardiography were performed, which had no pathological findings. A CT scan of the brain did not show any abnormality in the patient's skull bone (Figure 1).

Conclusion and Results

The patient underwent surgery without systemic symptoms and in the inactive phase of the disease. In the first stage, fat injection was performed for the patient and the repair was performed with a free flap and tissue expander technique. In the second stage, after 8 months, surgery and fat injection were performed (Figure 2). Correct and timely diagnosis of this disease, as well as a multidisciplinary approach and timely and appropriate surgical treatment to minimize the psychological effects and improve the patient's appearance are of particular importance.

Discussion

PRS is a rare disease with a female predilection and ratio of 1:70,000 and is mostly seen in the left half of the face. The prevalence of PRS in boys is lower and its ratio is 1:3[8]. It is believed that brain disorders in fat metabolism, local facial trauma, endocrine disorders, autoimmunity, heredity, decreased or increased sympathetic nervous system activity, trigeminal nerve abnormalities and viral infections are related to the pathogenesis of PRS[9]. PRS has been associated with developmental and congenital abnormalities, such as neurological, ocular, cardiac, cranial, jaw, and facial abnormalities. Stress (26%) and surgery (8%) have also been identified as possible triggers for the acceleration of the disease, which usually involves the lower part of the face, and deeper involvement of bones, teeth, tongue, and gums may also be seen[10]. It should be noted that this involvement is without significant epidermal change[4], although PRS usually refers to the atrophy of dermatomes of one or more branches of the fifth cranial nerve [2], and in general, skeletal hypoplasia of the affected areas of the skull is also common [11]. In the presented case, skeletal hypoplasia was not seen in the skull and only the fat and subcutaneous tissue had atrophied. Positive ANA is the most common laboratory abnormality and approximately 25-52% of PRS patients have high antibody titers[8]. However, no positive antinuclear antibody was reported in the tests of this patient. Rheumatoid factor (RF) along with local scleroderma and extra cutaneous involvement has been shown in arthritis patients. However, it remains unclear whether any PRS patients regularly have elevated RF[12]. In imaging findings, ultrasound can be used to detect the presence of sclerosis and monitor the progress of the disease and the progress of treatment by measuring the thickness of the skin and the echogenicity of the affected areas. Color Doppler ultrasound has the additional advantage of measuring skin blood flow, the increase of which indicates active disease [13]. Considering the frequency of neurological complications in PRS, basic imaging (CT-scan, MRI), especially in patients with neurological symptoms, can be performed [8].

The timing of surgical intervention in patients with PRS has been discussed. Most experts recommend that treatment be delayed until the progression of the disease stops or gradually stops, so that the surgical site can achieve a stable skeletal base along with the progression of the defects and multiple surgeries can be avoided. For more severe atrophy, a combined approach of strengthening both skeletal and soft tissue is recommended. Bone paste cranioplasty, skin fat grafting, and facial fat flaps can also be used to correct large volume atrophy. Brow lift, Z-plasty, lip reconstruction, nose reconstruction, eyebrow reconstruction, face lift, lip augmentation, hair transplant and other auxiliary procedures can also be used to create a better cosmetic result [14-16]. However, in addition to aesthetic concerns, this syndrome creates functional and psychological problems for patients, which requires a multidisciplinary team approach to identify the treatment expectations of these patients[17].

Abbreviations

ANA: Antinuclear antibody; CT: Computed Tomography; MRI: Magnetic resonance imaging; PRS: Parry-Romberg syndrome; RF: Rheumatoid factor

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Consent for publication

Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

Availability of supporting data

The authors declare that all the data supporting the findings of this study are available within the article.

Authors' contributions

KB, NS, AR and AM, took full history and performed full examination. AR, was consulted about the case and requested investigations needed. AM and NS, major contributors in writing the manuscript. All authors read and approved the final manuscript.

Key Clinical Message

Parry-Romberg syndrome is characterized by progressive dystrophy in one half of the face, which usually begins in childhood. Correct and timely diagnosis of this disease, as well as a multidisciplinary approach and timely surgical treatment to minimize the psychological effects and improve the patient's appearance are of particular importance.

References:

1. Ong B, Chong P, Yeo P. Progressive hemifacial atrophy–a report of 2 cases. *Singapore medical journal* . 1990;31(5):497-499.

2. Duymaz A, Karabekmez FE, Keskin M, Tosun Z. Parry-Romberg syndrome: facial atrophy and its relationship with other regions of the body. *Annals of plastic surgery* . 2009;63(4):457-461. doi:10.1097/SAP.0b013e31818bed6d

3. Jun JH, Kim HY, Jung HJ, et al. Parry-Romberg syndrome with en coup de sabre. *Annals of dermatology* . 2011;23(3):342-347. doi:10.5021/ad.2011.23.3.342

4. Tollefson MM, Witman PM. En coup de sabre morphea and Parry-Romberg syndrome: a retrospective review of 54 patients. *Journal of the American Academy of Dermatology* . 2007;56(2):257-263. doi:10.1016/j.jaad.2006.10.959

5. de Vasconcelos Carvalho M, do Nascimento GJF, Andrade E, Andrade M, Sobral APV. Association of aesthetic and orthodontic treatment in Parry-Romberg syndrome.*Journal of Craniofacial Surgery* . 2010;21(2):436-439. doi:10.1097/SCS.0b013e3181cfe917

6. Roddi R, Riggio E, Gilbert PM, Hovius SE, Vaandrager JM, van der Meulen JC. Clinical evaluation of techniques used in the surgical treatment of progressive hemifacial atrophy. *Journal of Cranio-Maxillofacial Surgery* . 1994;22(1):23-32.

7. Abdelnour JGW, Abdelnour YGW, Kerollos R-MAB, Mahmoud ZIT. Parry–Romberg syndrome associated with en coup de sabre in a patient from South Sudan–a rare entity from East Africa: a case report. *Journal of Medical Case Reports* . 2019;13(1):1-5. doi:10.1186/s13256-019-2063-2

8. Tolkachjov SN, Patel NG, Tollefson MM. Progressive hemifacial atrophy: a review. *Orphanet Journal of Rare Diseases* . 2015;10(1):1-13. doi:10.1186/s13023-015-0250-9

9. El-Kehdy J, Abbas O, Rubeiz N. A review of Parry-Romberg syndrome. *Journal of the American Academy of Dermatology* . 2012;67(4):769-784. doi:10.1016/j.jaad.2012.01.019

10. Stone J. Parry–Romberg syndrome: a global survey of 205 patients using the Internet. *Neurology* . 2003;61(5):674-676. doi:10.1212/wnl.61.5.674

11. Balan P, Gogineni SB, Shetty SR, D'souza D. Three-dimensional imaging of progressive facial hemiatrophy (Parry-Romberg syndrome) with unusual conjunctival findings.*Imaging science in dentistry* . 2011;41(4):183-187. doi:10.5624/isd.2011.41.4.183

12. Zulian F, Athreya B, Laxer R, et al. Juvenile localized scleroderma: clinical and epidemiological features in 750 children. An international study. *Rheumatology* . 2006;45(5):614-620. doi:10.1093/rheumatology/kei251

13. Wortsman X, Wortsman J, Sazunic I, Carreño L. Activity assessment in morphea using color Doppler ultrasound. *Journal of the American Academy of Dermatology* . 2011;65(5):942-948. doi:10.1016/j.jaad.2010.08.027

14. Palmero MLH, Uziel Y, Laxer RM, Forrest CR, Pope E. En coup de sabre scleroderma and Parry-Romberg syndrome in adolescents: surgical options and patient-related outcomes. *The Journal of Rheumatology* . 2010;37(10):2174-2179. doi:10.3899/jrheum.100062

15. Slack GC, Tabit CJ, Allam KA, Kawamoto HK, Bradley JP. Parry-Romberg reconstruction: optimal timing for hard and soft tissue procedures. *Journal of Craniofacial Surgery* . 2012;23(7):1969-73. doi:10.1097/SCS.0b013e318258bd11

16. Yu-Feng L, Lai G, Zhi-Yong Z. Combined treatments of facial contour deformities resulting from Parry-Romberg syndrome. *Journal of reconstructive microsurgery* . 2008;24(05):333-342. doi:10.1055/s-2008-1080536

17. Al-Aizari NA, Azzeghaiby SN, Al-Shamiri HM, Darwish S, Tarakji B. Oral manifestations of Parry-Romberg syndrome: a review of literature. *Avicenna Journal of Medicine* . 2015;5(2):25-28. doi:10.4103/2231-0770.154193

Hematology	**Biochemistry**
WBC 108000 uL PB˜ 5.53Mιλ/ μΛ Hb 14 mg/dl HTC 41.8 % Plt 410000 uL	**Akl P 1315 U/L Gama GT**

Table 1: Laboratory Test Results

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