

Romberg's syndrome: a series of cases

Síndrome de Romberg: uma série de casos

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ABSTRACT

Background: The Romberg's syndrome is a disease that was first described more than a century ago, and during this period had several names. It is characterized by a slow and progressive atrophy of the tissues of a hemiface, can affect all tissues and can also display neurological and ocular manifestations. The aim of this study is to report a series of patients with progressive hemifacial atrophy, addressing treatment options for each individual case. **Methods:** Was conducted a retrospective, descriptive and analytic study of the patients treated in our service, Plastic Surgery at the Hospital de Clínicas - Universidade Federal do Paraná, and the Centro de Atendimento Integral ao Fissurado Lábio Palatal (CAIF), at Curitiba, PR, Brazil. **Results:** Were analyzed 13 patients, 10 (76.9%) female and 3 (23.1%) male. The age of onset of symptoms ranged from 2 to 15 years. The first medical consultation with an specialist occurred at 11.7 years. The left hemiface was most commonly affected. Seven (53.8%) patients underwent surgery. **Conclusions:** The Romberg's syndrome is a rare disorder, devastating to facial appearance, and it has been widely studied. Despite great advances in medicine, its etiology definition and the treatment directed to its cause is still only a wish.

Keywords: Facial hemiatrophy. Face/surgery. Reconstructive surgical procedures.

RESUMO

Introdução: A síndrome de Romberg é uma moléstia descrita há mais de um século e, nesse período, recebeu várias denominações. É caracterizada por lenta e progressiva atrofia dos tecidos de uma hemiface, podendo acometer todos os tecidos e apresentar, também, manifestações neurológicas e oculares. O objetivo deste estudo é relatar série de casos de pacientes com atrofia hemifacial progressiva, abordando as opções terapêuticas individualizadas para cada caso. **Método:** Foi realizado estudo retrospectivo, descritivo e analítico de pacientes atendidos nos ambulatórios de Cirurgia Plástica no Hospital de Clínicas da Universidade Federal do Paraná e no Centro de Atendimento Integral ao Fissurado Lábio Palatal (CAIF), em Curitiba, PR, Brasil. **Resultados:** Foram analisados 13 pacientes, sendo 10 (76,9%) do sexo feminino e 3 (23,1%) do sexo masculino. A idade de início dos sintomas variou de 2 anos a 15 anos. A primeira consulta ocorreu, em média, aos 11,7 anos. A hemiface mais frequentemente afetada foi a esquerda. Sete (53,8%) pacientes foram submetidos a cirurgia. **Conclusões:** A síndrome de Romberg é uma entidade rara, devastadora na aparência facial, que tem sido muito estudada. Apesar dos grandes avanços da medicina, a definição de sua etiologia e um tratamento direcionado a sua causa ainda são apenas um desejo.

Descritores: Hemiatrofia facial. Face/cirurgia. Procedimentos cirúrgicos reconstrutivos.

This study was performed at the Hospital de Clínicas da Universidade Federal do Paraná, Curitiba, PR, Brasil.

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INTRODUCTION

The progressive hemifacial atrophy was first described by Parry, in 1825, and later by Romberg, in 1846, receiving different names such as: Parry-Romberg syndrome, proso-podismorfia and trofoneurose¹.

It is characterized by slow and progressive atrophy of the tissues of a hemiface initially affecting soft tissue, but it can affect all tissues including bone. May also occur: cerebral involvement, the carrier may have seizures², intracranial vascular malformations³, alterations in cranial nerves, peripheral and autonomic nervous system involvement⁴, as well as ocular involvement (which occurs in 10%-15% of cases)⁵.

Symptoms usually begin between the first and second decades of life, progresses for a few years and then become stationary⁶. Some authors advocate more common occurrence in females, a ratio of 3:2 men⁷, while others consider the same rate between genders⁸. The left hemiface is the most commonly affected.

Its etiology remains controversial, several theories were suggested, such as angiogenic entrapment, trauma, infection, trigeminal neuritis, among others^{6,9}.

The treatment consists in controlling the symptoms (neurological and ocular) while the disease is active. After the atrophy stabilization, the aesthetic treatment can be started. For the treatment of facial asymmetry can be used many alternatives, including dermal, bone and fat grafts, flaps rotation as well as the use of alloplastic products.

The objective of this study is to report cases of progressive hemifacial atrophy treated in our service, Plastic Surgery at the Hospital de Clinicas - Universidade Federal do

Paraná, and the Centro de Atendimento Integral ao Fissurado Lábio Palatal (CAIF), in the period 2000 to 2012.

METHODS

A retrospective, descriptive and analytic study of patients treated in the service of Plastic Surgery at the Hospital de Clinicas - Universidade Federal do Paraná, UFPR-PR and Centro de Atendimento Integral ao Fissurado Lábio Palatal (CAIF), at Curitiba, Brazil, was performed. The medical records of patients with Romberg's syndrome who were treated in these hospitals during the period 2000 to 2012 were accessed.

The data included: age of onset of symptoms, age at first examination, gender, affected hemiface, family history, other comorbidities, number, age and type of surgical procedure.

RESULTS

Were analyzed 13 patients, 10 (76.9%) female patients and 3 (23.1%) males.

The age of onset of symptoms ranged from 2 to 15 years, with an average of 6.3 years. The first consultation with a specialist took place with 11.7 years (ranging from 4 to 36 years). The delay in seeking medical care was 5.4 years (Table 1).

The most affected hemiface was the left, corresponding to 61.5% of the total. No patient had family history of facial hemiatrophy. The associated diseases were vitiligo (n = 2), strabismus (n = 2) attention deficit disorder and hyperactivity (n = 1) and inguinal hernia (n = 1).

Table 1 – Characteristics of patients evaluated.

Patient	Gender	Affected hemiface	Symptoms onset (years)	Specialist consultation (years)	First surgery (years)	Number of surgeries
1	F	R	4	6	12	4
2	M	R	12	36	36	3
3	F	R	9	18	20	2
4	F	L	13	20	21	3
5	F	L	2	4	–	–
6	M	R	4	6	–	–
7	F	L	3	6	6	2
8	F	R	3	7	–	–
9	M	L	4	5	–	–
10	F	L	2	4	8	4
11	F	L	15	18	–	–
12	F	L	3	10	13	2
13	F	L	8	12	–	–

R = right; L = left; F = female; M = male.

In our series the patients had involvement of multiple regions simultaneously, with an average of 3.6 affected regions in each case (range 2-6). The most commonly region affected was the upper lip (n = 11), followed by: cheek (n = 9), lower lip (n = 6), jaw (n = 5), nose (n = 4), forehead (n = 4 - included 3 patients with deformity coup de sabre), maxilla (n = 2), scalp alopecia (n = 2), lower eyelid (n = 1), neck (n = 1), eyebrow alopecia (n = 1) and important localized skin atrophy (n = 1). In all cases of atrophy of the cheek, the upper lip was involved, as in all cases of nasal involvement upper lip was also affected. In 5 of 6 patients with atrophy of the lower lip, jaw also was affected.

Seven (53.8%) patients underwent surgery. Were performed 2.8 procedures per patient among these patients, ranging from 2 to 4. The surgeries performed: cartilage graft for nasal region (n = 2), temporal fascia graft (n = 1), fat grafting (n = 16), cranioplasty (n = 1), piecemeal resection of scar (n = 1), rhinoplasty (n = 1), and 3 patients submitted to associate procedure.

The volume of fat grafting ranged from 5 to 55 ml per procedure, with an average of 20 ml per session. Four (57.1%) patients showed significant absorption of the fat graft, and was necessary to repeat the procedure. Three patients had good graft integration with no significant fat resorption.

Figures 1 and 2 illustrate some cases in this series.

DISCUSSION

Romberg syndrome is a rare disease and its pathogenesis remains unknown. Despite multiple theories attempting to explain it, none is universally accepted. Clinical presentation is very inconstant notably in the onset of symptoms. The ectoscopy is the main element in the diagnosis of this disease. The facial hemiatrophy in our series affected predominantly female patients, as consistent with other studies⁷.

The initial symptoms vary: trigeminal paresthesias, seizures, hypo- or hyperchromic spots. As the disease progresses, become more apparent the alterations of skin, subcutaneous tissue and later muscle, cartilage and bone. Manifestations progress at a slow pace and tend to become stationary after some years of activity⁶.

The manifestation begins on the first or second decades of life⁶, from 2 to 15 years in our sample. The bone changes tend to be more severe as younger the patient is at the onset of symptoms¹⁰.

The long period between onset of symptoms and demand for the specialized center may be due to poor knowledge of this syndrome by physicians in general or for the delay to obtain an expert consultation in our health system.

The therapeutic approach of these children can be the use of medications or surgical. The clinical treatment of disease with immunosuppressant (methotrexate) or other medications, such as chloroquine and calcipotriol, is discussed by several authors; however, there is no scientific evidence for the use of this kind of medication¹¹.

Surgical treatment is delayed until stabilization of the disease. The soft tissues can be treated earlier, however the correction of the osteocartilaginous structures must await the complete facial growth.

Surgical treatment of facial defect can be performed using different techniques¹²⁻¹⁵. Structural defects can receive alloplastic materials such as silicon and porous polyethylene, while soft tissue defects can be reconstructed with autologous dermal fat grafts, sequential fat grafts, and transfer of vascularized tissue (direct or microsurgical).

Mordick et al.¹⁴ and Roddi et al.¹⁵ conducted studies comparing the two main techniques: the fat grafting and vascularized tissue transfer. These studies showed that fat grafts are simple, affordable, with low risk of complications and good cosmetic results, especially for mild to moderate

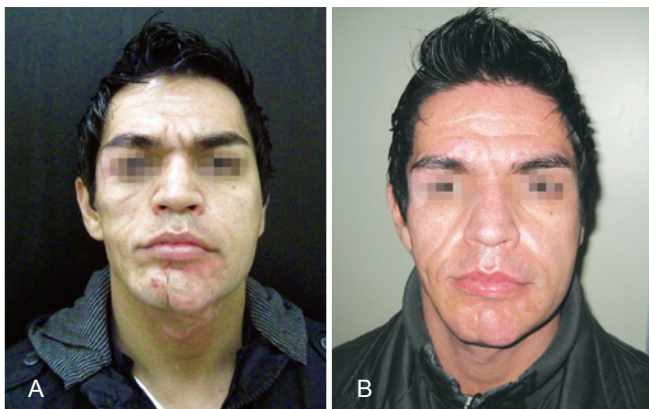


Figure 1 – In **A**, patient before the beginning of aesthetic treatment. In **B**, patient after 3 sessions of fat grafting, with aesthetic treatment in progress.



Figure 2 – In **A**, patient before the beginning of aesthetic treatment. In **B**, patient after 4 sessions of fat grafting.

defects. However, for most severe atrophy, the transfer of vascularized tissue is more effective, due to the low vascular supply. Pieces of omentum, muscle and fasciocutaneous flaps, having great divergence in indications, can be used.

We used several techniques for cosmetic improvement of our patients. The indication depends primarily on the degree and location of the defect, and its indication should be individualized. The approach to the frontal region, we chose to implant a surgical cement (n = 2) and in 1 case associated with fat grafting. There were two patients who underwent conchal cartilage graft for nasal ala, one of whom later received a temporal fascia tissue to increase coverage, and erase the edges of the graft. Other procedures were piecemeal resection of scar, and rhinoplasty.

The method of choice, cause of its easy implementation and low morbidity, regardless of the result we obtained, was the fat grafting. This technique allows increase in soft tissues, corrections of other procedures that may have been insufficient, and it's easy to be repeated. The fat grafting leads to improvement in the texture of the atrophied tissue, followed by increased local blood supply.

The fat grafting technique has recently evolved in a new research on stem cells that can be isolated from lipoaspirate material in large numbers and its easily grow under standard tissue culture conditions. Because of this, there are excellent prospects for its use in plastic surgery for the treatment of congenital and acquired deformities with aesthetic or restorative purpose¹⁶.

CONCLUSIONS

Romberg syndrome is a rare disorder, devastating in facial appearance, which has been widely studied due to the potential therapeutic improvement with the use of stem cells. Despite great advances in medicine in all its fields, the etiology and treatment directed at its cause is still only a wish. While we do not have enough knowledge to treat the cause, we improve techniques for the treatment of its consequences.

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