

Up-to Date Review And Case Report

Progressive facial hemiatrophy (Parry-Romberg syndrome): short case report

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Abstract – Introduction: The Parry Romberg syndrome (PRS) is a mosaic disease of unknown aetiology which mostly affects women. The facial hemiatrophy generally begins during the early childhood. It has a great impact on social life, and aesthetic rehabilitation is a major challenge. **Observation:** A 38 years-old-female patient presented with an enucleation of the left eye, due to multiple hamartomas and progressive facial hemiatrophy. The placement of 2 extraoral implants was proposed to bear an ocular epithesis. **Comments:** PRS develops between the 2nd and 20th year of life, and stabilizes at the adult age. Its main characteristic is a progressive facial hemiatrophy, which involves skin, fat tissues, muscles and osteocartilaginous tissues. 20% of patients have neurological symptoms, and epilepsy is often described as a side effect of the disease. **Conclusion:** Diagnosis of localized scleroderma has to be eliminated, as these 2 entities are very similar. Major facial reconstructive surgery is often proposed to restore aesthetics.

Introduction

The Parry Romberg syndrome (PRS) or progressive facial hemiatrophy has been described by Parry in 1825 and by Romberg in 1846 [1]. The main characteristic is a progressive unilateral facial atrophy of the skin of the maxillary region, subcutaneous tissue and sometimes the adjacent craniofacial bone. It mostly appears in female patients aged between 2 and 20. Neurologic involvement concerns 20% of the patients. This mosaic disease has a great impact on social life; aesthetic rehabilitation is being a major challenge for these patients' rehabilitation. This paper highlights the long-term effects of the syndrome and therapeutics proposals to overcome them.

Observation

A 38 years-old-female patient presented with an enucleation of the left eye. Her medical history began at birth with multiple benign tumors of the legs and the lacrymal foramen of the left eye. The karyotype was normal. Surgery was performed at the age of 5 months to remove the tumors. Histopathology revealed multiple hamartomas. Left endophthalmia and left

progressive facial hemiatrophy were noticed from the age of 15 months. The differential diagnosis consisted of localized sclerodermatitis. For this patient, the atrophy was more diffuse and affected the whole hemiface. Type I neurofibromatosis was also discussed but there was no NF1 gene mutation.

At 9 years old, the patient presented with evolutive epilepsy and brain tomodensitometry revealed large amounts of porencephaly in the left hemisphere. At 14 years old, a mandibular odontogenous fibroma led us to perform an interruptive mandibulectomy, mandibular reconstruction being achieved at the end of growth with a rib graft. Dental implants were placed on the graft to restore the dental arch.

Enucleation had then to be realized because of pain caused by major endophthalmia. Growth was highly affected, with an adult size of 1.32 m and major facial left atrophy. A mild mental retardation was noticed. Lipofillings were proposed to enhance facial contour.

The patient consulted at the age of 38 for the rehabilitation of her left eye with an epithesis. Clinical examination revealed facial asymmetry, decreased but sufficient bone volume on the periorbital left area, and hypodevelopment of the left hemiface. She also complained of frequent headaches.

A dental CT scan was performed to evaluate the bone amount for periocular implant placement. The CTscan also revealed an osteolysis of the sphenoid and a large hamartoma involving the soft tissues (Fig. 1).

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Fig. 1. (A and B) CT scan showing multiple intracranial hamartomas; (C) orthopantomography showing the hamartomas and the intraoral status.



Fig. 2. Extraoral implants for the bearing of the epithesis.

Two extra oral implants of 4 mm length were placed under general anaesthesia to bear an oculo-palpebral epithesis (Fig. 2). After 4 months for osseointegration, the epithesis was made.

Commentaries

This case report was first published when the patient was aged 17 [2]. Republishing this case 20 years after allows a long term follow up of the case, which is of particular interest in slowly progressive diseases such as PRS. The PRS is a rare disease, mostly affecting female patients from the age of 2 years old [3]. It develops between the 2th and 20th year of life, and stabilizes at the adult age. Late stabilization of the disease (up to 75 years) has been rarely described [4]. Various aetiologies have been proposed, of which autoimmune dysfunction [5] on the same spectrum as localized scleroderma. Serum autoantibodies, inflammatory histopathology and positive response to immunosuppressors are found. Another hypothesis is a modified migration of the neural crest cells, intracranial vascular malformation or neurotrophic viral infection [5]. Patients with early onset seem to be more severely affected, with more severe aesthetic and functional deficits [6]. This patient presented with tumors related to PRS at birth, which is early and could corroborate embryogenic ectodermic aetiology of the disease [7]. It often affects the maxillary and orbital region [4,8]. 20% of patients have neurological symptoms, such as headaches and cognitive impairment, brain atrophy, epilepsy, or hemiplegia [4,9]. In this case report, the patient presented with headaches and epilepsy which severity increased with the patient's growth.

Ocular involvement concerns 16% of PRS patients [4,10–12]. In this case report, eye involvement was early and led to enucleation of the left eye.

Alopecia is sometimes described [9]. Some authors have discussed the premium effect of soft tissues retraction on bone growth and asymmetry, but it seems that bone atrophy is also a primary phenomenon rather than a process secondary to soft tissue retraction [13].

Differential diagnosis consisting of localized scleroderma must be eliminated, as it also affects young patients. Clinical presentation includes ophthalmic and neurological complications, with a facial atrophy described as “en coup de sabre”, and a loss of skin elasticity [4]. Due to the overlapping clinical presentations of these 2 diseases, some authors consider them as clinical variants. It also has been shown that conversion of localized scleroderma to PRS has been reported, and that 30–40% patients with PRS present with typical scleroderma changes [4]. Lipoatrophy (also described as Barraquer-Simons syndrome) has also to be eliminated: the lipoatrophy is systemic and not limited to the face, and there are complement levels abnormalities and C3 seric nephritic factor [14,15]. Nonetheless, these 2 diseases are also strongly associated [16].

PRS shows various types of imaging features, and a CT scan must be performed to evaluate intracranial and facial involvement. Nonetheless, the incidence and cause of these alterations remain unknown [4,17].

Considering intra-oral status, previously to the mandibular reconstruction, the patient reported delayed teeth eruption, which is another characteristic of the PRS [5].

A few cases of bilateral PRS syndrome have been described [3,14]. It affects 2–7% of cases [17].

As facial aesthetic is a main complaint, plastic surgical procedures are often proposed. Periocular lipofilling performed before the realisation of the epithesis allowed a better esthetical result. This surgery must be performed after disease stabilization [5]. Autologous fat grafting seems to show favourable results for soft tissues deficiency [5], as well as

hyaluronic acid filling [14]. For severe diseases, free tissue transfer is preferable [5].

Conclusion

PRS is a clinical diagnosis supported by imaging and histopathology. Multimodal follow up (rheumatology, neurology, maxillofacial surgery, oral surgery, and prosthodontics) must be performed. Psychological and social problems must also be considered [6].

Conflicts of interest: The authors declare that they have no conflicts of interest in relation to this article.

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