Parry Romberg Syndrome: A Case Report Involving Intranasal Mucosal Atrophy and Secondary Eustachian Tube Dysfunction

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ABSTRACT

Parry Romberg syndrome is a rare disorder described as facial asymmetry. It is of unknown etiology, but trophic malfunction of the sympathetic system has been proposed as a cause. The syndrome results in specific soft tissue, skeletal, dental and skin changes in the affected side of the face, with or without neurological signs and symptoms. The case we present here is a 23-year-old female with Parry Romberg syndrome. Clinically she has left sided facial atrophy affecting both the soft tissue and the bone structure. The patient received pharmacological and surgical treatments to slow the progression and correct the contour of her face. She complained lately from autophony, which is the unusual loud hearing of a person’s own voice and own breathing. A diagnostic nasoendoscopy showed atrophied nasal mucosa and Eustachian tube dysfunction which has never been described before to be secondary to parry Romberg syndrome.

Keywords: Parry Romberg Syndrome Eustachian tube dysfunction Intranasal mucosal atrophy

1. INTRODUCTION

Parry Romberg Syndrome is a slowly progressive atrophy of the skin and subcutaneous tissue, was first reported by Caleb Parry in 1825 and then described as syndrome by Moritz Romberg in 1846 [1,2,3]. Eulenberg introduced the term 'progressive facial hemi atrophy' (PFH) in 1871 which became widely accepted [10]. It is believed to be self-limited [3]. It involves mainly the skin and subcutaneous fat, and to a lesser extent the muscles and bones. It overlaps with a condition known as scleroderma “en coup de sabre” [4].

The onset is insidious usually within the first two decades of life [5]. The disease progresses rapidly in the first 10 years after the onset then stabilizes [1]. It is uncommon and generally unilateral with a higher incidence rate in females. Hemi facial atrophy is a sporadic disorder but some familial distribution has been reported [6]. The syndrome is accompanied by neurological complications in about 15% of cases such as trigeminal neuralgia, migraine and facial paresthesia [7, 8, 9]. Patients, who manifest atrophy in early ages, have worse consequences [25].

Treatment modalities are divided into pharmacological treatment to slow the progression and relieve the symptoms, and surgical options which aim to restore the facial contour. None of these treatment options will cure the disease [11].

2. CLINICAL CASE

A 23-year-old female who is known to our team since childhood with progressive left sided facial hemiatrophy. She has no previous trauma as well as no relevant family history of such a condition or autoimmune diseases. Clinically she had an atrophy of left facial soft tissue and hard tissue, a malocclusion, vertical orbital dystopia and moderate head tilt. She had also left lower limb discrepancy (LLD) with 3 cm shortening. She was diagnosed with Parry Romberg syndrome and evaluated in a multi-disciplinary team involving a plastic, cranio-facial, maxillofacial, ENT, orthopedic and eye surgeons. A plan for her management was discussed intensively and she was prioritized for the correction of her malocclusion through pre surgical orthodontics then bimaxillary osteotomies and free groin flap to augment her left face. A right lower limb epiphesiodesis done to correct her (LLD). This was followed by several Coleman fat transfers to her left cheek, dermal fat grafts to her both lips and a cartilage graft to the left infra orbital area. She also received methotrexate and prednisolone which successfully slowed the progression of her atrophy.

A recent complaint was an annoying autophony, thought to be secondary to other causes. Later on, nasoendoscopy revealed an atrophic nasal mucosa and Eustachian tube orifice dysfunction which has never been described before in association with a Parry Romberg case.

Fig. 1: Nasoendoscopy showing atrophic nasal mucosa and Eustachian tube orifice

3. DISCUSSION

Parry Romberg syndrome or (PFH) disease is an uncommon, progressing atrophy of the face, manifests in the first or second decade of life and rarely affects the limbs. It usually stabilizes after few years of progression [1,3]. The prevalence rate is estimated to be 1 per 700,000 in the general population [4]. It involves the skin and subcutaneous fat, and on rare occasions the muscles and bones of the face [10]. Up to 5% to 10% of cases were found to be bilateral, with predominance of the left side [13,14].

Other important features of this disease are enophthalmos due to fat loss around the orbit, the deviation of mouth and nose to the affected side, with unilateral exposition of the dentition (when lips are involved in atrophy) [12]. The oral mucosa, salivary glands and the tongue can be also affected. There may be a unilateral reduction in tongue size [16]. No functional ear abnormalities were described.

Bony defects are usually seen when the disease manifests before the age 15. In cases the onset is before the age of 5, Fronto-maxillary defects are seen. Between the age 5 and 15, mandibular de-fects are seen while with later onset (>15 years) almost exclusively soft tissue changes are present. The mandibular atrophy is unilateral and, in all dimensions, (height, length and width). As a result, the midline shifted towards the affected side. Bony regeneration is altered after exodontia in the affected side [18].

Parry Romberg syndrome is found to be more common in females [7,8,9]. The etiology of the disease is not clear. Many authors claim a sympathetic nervous system malfunction as a cause of skin, muscle, and bone abnormalities. This was shown by experimental studies in animals (Cory et al) [24].

Another proposed aetiologyas are cerebral disturbance on fat metabolism [8,10,11], Trauma, viral infections, endocrine disturbances, auto-immune and hereditary causes are believed to be also associated with the pathogenesis of the disease [12,15,17]. Lyme disease was also described as a cause (Borreli burgdorferi) [16, 23]. Occasionally, patients develop neurological complications, such as trigeminal neuralgia, facial paraesthesia, severe headache and contra lateral epilepsy which is the most common complication as reported by Chbicheb M et al [8].

Histopathological examination of skin reveals atrophy of the epithelium and dermal tissue and hair follicles with fibrosis (if clinically resembling linear
scleroderma). Perivascular chronic inflammation is a very important distinguishing feature between PFH and scleroderma. Perivascular inflammation is variable in the former and massively present in the latter, especially in the early stages. The elasticity of dermal tissue is preserved in PFH but lost in scleroderma [23,24].

The diagnosis of Parry Romberg syndrome is based on patient history and clinical examination and is supported by imaging and histopathological studies. The severity of the condition depends on the age of onset. Patients, who manifest atrophy in early ages, might have a better outcome [4].

Different treatment modalities are tried and divided into pharmacological treatment, mainly immunosuppressive drugs such as corticosteroids, methotrexate, azathioprine, and cyclophosphamide. Other medications are given to treat symptoms like anti-seizure and migraine for pain relief. The other modalities are the surgical ones which advised to be started after the deformities stabilized. The aim is to restore the facial contour using loco-regional flaps, fat transfer, orthognatic surgeries and bone distractions [19,20].

None of these treatment options will cure the disease itself but believed to slow its progression, relieve the symptoms and partially correct the deformities [11].

Cases associated with Lyme disease have been treated with antibiotics like parenteral penicillin and ceftriaxone. As an adjunct, phototherapy with UV-A radiation (340–400 nm) has been used to re-verse fibrosis since it is known to induce matrix metalloproteinase 1 (MMP-1).

It has never been found that patients with parry Romberg syndrome suffer from hearing abnormalities or even soft tissue involvement extends to involve nasal mucosa. The patient we present in this report complained lately from hearing hear own voice and breath sounds, a condition called an autophony. It was first thought to be secondary to other causes. Later on, nasoendoscopy revealed an atrophic nasal mucosa and Eustachian tube orifice dysfunction (figure 1) which believed to be an extension to the severity of her (PFH). It has never been described before in association with a Parry Romberg case.

4. CONCLUSION

Parry Romberg syndrome is a progressive disease of all tissues kind with more preference to skin and subcutaneous fat. It can extend beyond what is usually described. Autophony, secondary to nasal mucosal atrophy and Eustachian tube dysfunction is a possible complaint and a late manifestation of the disease. Affected patients should have multidisciplinary attendance for proper diagnosis and treatment.

REFERENCES


