Head injury as a trigger of Parry-Romberg syndrome: A case report and review of etiology

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ABSTRACT

Introduction: This is a case of a patient with the rare entity of Parry-Romberg syndrome (PRS) which discuss the etiological association with preceding head trauma as well as the possible therapeutic role of vitamin B12, with a brief review of literature. Case Report: A 22-year-old female presented with a history of head injury preceding the progressive onset of right-sided facial pain and atrophy, evident facial asymmetry with periorbital hyperpigmentation on the right side of the face, enophthalmos and watering of the right eye. Laboratory investigations revealed a low vitamin B12 level. A final diagnosis of Parry-Romberg syndrome (“progressive hemifacial atrophy”) was based on clinical and a radiological examination. Conclusion: Parry-Romberg syndrome is an incompletely understood self-limiting degenerative condition, characterized by unilateral atrophic changes of the face. The pathophysiology of the syndrome remains unclear, but the etiologic relevance of factors like head injury and vitamin B12 deficiency needs to be investigated in further collective studies.

Keywords: Etiology, Head injury, Hemi facial atrophy, Parry-Romberg syndrome, Scleroderma

INTRODUCTION

Parry-Romberg syndrome, which is also known as progressive facial hemi-atrophy, is a rare, acquired, neurocutaneous syndrome of unknown aetiology, with a higher incidence rate in females [1, 2]. Unilateral progressive atrophy of the face was first described by an English physician, Caleb Parry as early as in 1825 and subsequently elaborated in 1846 by Moritz Romberg [3, 4]. However, it was Eulenberg in 1871 that gave the name ‘progressive facial hemi-atrophy’ [5]. It is characterized by hemi-atrophy of fat, skin and connective tissue. Muscles and/or bones are also involved in some instances. Neurological manifestations are the most frequent, seen in nearly 15% of the patients. Most commonly, seizures occur and sometimes can be attributed directly to a brain abnormality visible on MRI scan [1]. Ocular involvement is also common, and the most frequent manifestation is enophthalmos [1]. Moreover, this disease also overlaps with a condition known as linear scleroderma “en coup de sabre” [6]. The etiology of the disease is unidentified and there are no systematic studies to guide us but the most accepted theory is that it is an autoimmune condition. Although trauma has been hypothesized to be one of the triggers of PRS, there is only anecdotal evidence of Parry-
Romberg coming on after head trauma or surgery to the face. Here we report one such patient who presented with hemifacial atrophy and facial pain, after an incident of head trauma and we also propose a relationship of neurological manifestations of the disease with vitamin B12 deficiency.

CASE REPORT

A 22-year-old female presented to the neurology outpatient department with complaints of a right-sided headache and facial pain, which was intermittent, sharp and stabbing in quality for the past eight years and severe in intensity for past ten months. Alongside, she complained of progressive asymmetry of her face after she sustained blunt head trauma at the age of eight. Subsequently over the course of 10 years, a barely noticeable line appeared in the middle of her forehead, followed by progressive wasting of the right half of her face that had become static for the last two years. She developed hair loss ipsilaterally along with watering of her right eye. There was no history of similar complaints in the family. On examination, the patient was thinly built lady with a body mass index (BMI) of 19 kg/m². Right, frontal scalp showed alopecia and a line was palpable in the centre of her forehead (Figure 1). The right side of the face showed signs of Hemi atrophy of facial tissues, especially fat, with a prominent cheekbone and a slight deviation of the angle of mouth towards the right (Figure 2). There was enophthalmos of the right eye with periorbital hyperpigmentation and absent eyelashes on the right lower eyelid. Left side of the face appeared normal (Figure 3).

A diagnosis of Parry-Romberg syndrome with trigeminal neuralgia was made.

Routine blood, urine investigations and iron studies were within normal limits, however, vitamin B12 levels were found to be low. ANA, anti-dsDNA and anti Sc1-70 antibodies were negative. Magnetic resonance imaging scan of brain was unremarkable, other than a slight nasal septum deviation towards the left (Figure 4).

Differential Diagnosis

Hemifacial microsomia (first and second branchial arch syndrome), its variants such as Goldenhar syndrome and partial lipodystrophy (Barraquer-Simons syndrome) were considered. Hemifacial microsomia is usually present since birth and partial lipodystrophy usually involves both sides of the face.

Management

Supportive and conservative management was opted for initially, which included vitamin B12 supplementation with analgesics for a period of one month.

Figure 1: Right-sided facial atrophy, prominent cheekbone, ipsilateral hair loss and periorbital hyperpigmentation. A line is also visible in the center of forehead.

Figure 2: Asymmetric atrophy of face with slight deviation of angle of mouth towards right and enophthalmos. Left side of the face appears normal.
Outcome and Follow-Up:

On follow-up, her alopecia had improved markedly, while the facial pain had decreased too. An attempt to use allogeneic implant surgery to improve facial disfigurement is planned for the patient.

DISCUSSION

Parry-Romberg syndrome (PRS) is an infrequent, acquired and incompletely understood pathology, which involves progressive shrinking and deformation of one side of the face, resulting in unilateral atrophy. The PRS is associated with various systemic manifestations.

Usually, the onset of the disease is in the first decade of life, although there are reported cases of later onset of PRS. It is more often seen in females [1] and is believed to be sporadic. There are very few cases (<2%) with a possible hereditary etiology [2].

Clinically, cutaneous manifestations are seen, involving dermatomes of one or multiple branches of the trigeminal nerve. Unilateral facial atrophy of the skin, soft tissues, muscles, and/or underlying bony structures is seen in almost all the cases [2]. Skin hyperpigmentation or depigmentation and alopecia may also be observed in the affected areas [7, 8]. Around 25% of patients with facial hemi atrophy have a more definite vertical or diagonal “line” on their forehead as a result of cutaneous sclerosis, linear scleroderma “en coup de sabre” (LSCS) [2]. There is a close relationship between Parry-Romberg syndrome and linear scleroderma en coup de sabre. In fact, differentiating PRS from LSCS is very challenging, as both diseases have a similar age of onset (mean age of 11 years), predominantly affect women, and present with similar lesions [9]. Intraoral involvement with ipsilateral tongue hemi atrophy may also occur.

Neurological manifestations are the most common, accounting for nearly 15% of PRS patients, with seizures being the most common [10]. Psychologically, the disfigurement is often the most debilitating symptom, particularly, since this is an acquired condition.

Multiple ophthalmological manifestations are associated with PRS, enophthalmos being the most common [2]. Nearly half the patients, diagnosed with Parry-Romberg syndrome, also have dental anomalies [2]. Teeth of affected side may present some deficiency in root development and, consequently, delayed eruption and dental crowding [11], as seen in our patient. Mandibular involvement may also be seen. However, final degree of deformity may depend upon the duration of the disease.

The pathogenesis of PRS is not well understood with only case studies to guide us, however, it appears to have a heterogeneous etiology; trauma, immune-mediated processes, infection, disturbance of fat metabolism, sympathetic dysfunction and cranial vascular malformation have been proposed. Till date, no nutritional deficiency has been reported as a contributory factor in the symptomatology of PRS.
The anatomic changes of Parry-Romberg syndrome impact the growth potential of hard tissue, preventing an increase in size during active growth periods. The associated soft tissue shrinkage by the loss of adipose tissue [12]; hence, atrophy that starts in the second decade of life is less noticeable because facial growth is nearly complete. Early disease onset and longer duration cause greater deformity. Trauma has been thought to be a trigger of PRS in 24–34% of patients [13], whether accidental trauma (as seen in our patient), operative traumas or obstetric traumas. However, the mechanism whereby it might produce this condition remains to be identified.

The autoimmune hypothesis is based on the frequent association of PRS with autoimmune diseases [14] supported by the occasional finding of autoantibodies in the sera of patients with PRS. In addition, infections by slow viruses or bacteria have also been hypothesized as a possible causative factor in PRS; however, a clear relation has not yet been proven [15].

A cerebral disturbance of fat metabolism has been proposed as a primary cause, as a result of trophic malformation of cervical sympathetic nervous system [16]. This hypothesis has found some support because ablation of the superior cervical ganglion in animals has reproduced the principal clinical manifestations of PHA: hemifacial atrophy, enophthalmos, and bone atrophy on the side of the sympathectomy [17]. However, autonomic dysfunction testing has proved to be inconclusive, casting a shadow over this proposed theory [18].

CONCLUSION

Parry-Romberg syndrome is an incompletely understood self-limiting degenerative condition, characterized by unilateral atrophic changes of the face. The pathophysiology of the syndrome remains unclear, but the etiologic relevance of factors like head injury needs to be investigated in further collective studies. Also, the role of vitamin B12 in our case report might only be an incident finding due to nutritional deficiency but the temporal relationship between replacement of B12 and patients’ subjective improvement emphasizes that it should be a subject of research in future.

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Author Contributions

Saad Azim – Substantial contributions to conception and design, Acquisition of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

Jamal Janjua – Substantial contributions to conception and design, Acquisition of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

Azer Majeed – Substantial contributions to conception and design, Acquisition of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

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Guarantor

The corresponding author is the guarantor of submission.

Conflict of Interest

The authors certify that they have no affiliations with or involvement in any organization or entity with any financial interest (such as honoraria; educational grants; participation in speakers’ bureaus; membership, employment, consultancies, stock ownership, or other equity interest; and expert testimony or patent-licensing arrangements), or non-financial interest (such as personal or professional relationships, affiliations, knowledge or beliefs) in the subject matter or materials discussed in this manuscript.

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REFERENCES