Progressive hemifacial atrophy (Parry-Romberg Syndrome)

Deshingkar S. A., Barpande S. R., Bhavthankar J. D., Humbe J. G.

Abstract

Progressive hemifacial atrophy, also known as Parry-Romberg Syndrome, is an uncommon degenerative and poorly understood condition. It is characterized by a slow and progressive atrophy affecting one side of the face. The incidence and the cause of this alteration are unknown. A cerebral disturbance of fat metabolism has been proposed as a primary cause. This can be result of a trophic malformation of cerebral sympathetic nervous system. Possible factors that are involved in the pathogenesis are trauma, viral infections, heredity, endocrine disturbances, and autoimmunity, among others. Characteristically, atrophy progresses slowly for several years and, soon after, it become stable. The purpose of this work is, through the presentation of a clinical case, to accomplish a literature review concerning general characteristics, etiology, pathophysiology, differential diagnosis, and treatment of progressive hemifacial atrophy.

Keywords: Hemifacial atrophy, Parry-Romberg syndrome, scleroderma

Introduction

Progressive hemifacial atrophy, also known as *Parry-Romberg Syndrome*, is an uncommon degenerative condition characterized by a slow and progressive atrophy, generally unilateral, of facial tissues, including muscles, bones, and skin.[1] It was first described by *Caleb Hillier Parry* in 1825 and later in more detail by *Moritz Heinrich Romberg* in 1846.[2] More than an esthetic trouble, this illness brings several functional and psychological problems, when a "symmetric" face loses its identity.

Characteristically, there is regional atrophy of skin, subcutaneous tissue, and musculature. When the onset is before the second decade of life, the underlying bone and cartilage may also be involved.[3] A sharply demarcated line between normal and abnormal skin develops, so called *coup de saber*, and the involved area varies from a discrete lesion to a widespread, extensive malformation. Alopecia and pigmentation of the involved skin often appear.[3] The other important features of this pathology are the enophthalmy, the deviation of mouth and nose to the affected side, and unilateral exposition of teeth, when lips are involved.[3]

In addition to the obvious facial atrophy, a variety of accompanying conditions have been reported; ocular changes in 10% to 35% of cases; neurologic disturbances, which include focal epileptiform seizures and trigeminal neuralgia; and ipsilateral progressive body atrophy. Oral manifestations include atrophy of half of the lip and tongue, shortening of the body of the mandible and/or ramus of the mandible, retarded tooth eruption, and malformed tooth roots.[3]

The objective of this work is to report a clinical case of young male patient with emblematic features of progressive hemifacial atrophy along with discussion concerning general characteristics, etiology, pathophysiology, and treatment of progressive hemifacial atrophy.

Case Report

In 2008, a 13-year-old boy was referred to Department of Oral Pathology, Government Dental College and Hospital, Aurangabad, Maharashtra, with a complain of progressive deformity of left side of face since 5 to 6 years. During the physical examination, it was noted that this patient presented a facial asymmetry with marked hypoplasia of the left side of the face, deviation of lips and nose toward left side, and enophthalmy in left eye region [Figures 1 and 2]. A big linear dark scar (*coup de sabre*) was present in the left side of mandibular mentum region passing upward to involve lips and left ala of nose producing obvious depression [Figure 3]. The notching of the upper and lower lip leads to exposure of anterior teeth on left side. Alopecia was noticed in the region of left eyebrow toward medial side [Figure 1]. Patches of hyperpigmentation were seen on the skin of affected area. In addition, ear on left side was slightly smaller than that of right side.

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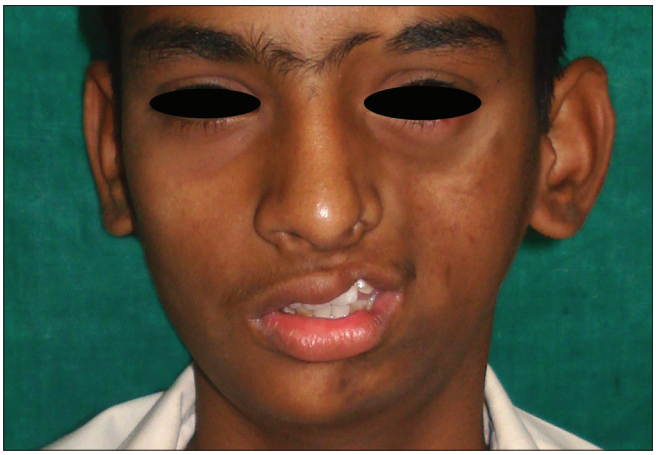

Figure 1: Marked hypoplasia of the left side of the face with deviation of lips and nose toward left side and notching of lips and nose with exposure of teeth. Alopecia in left eyebrow region

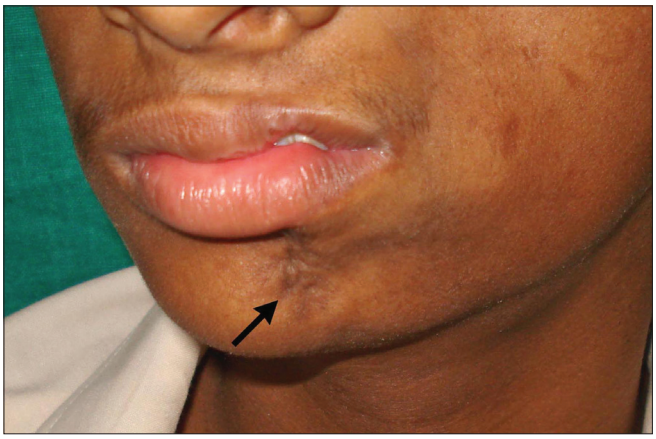

Figure 3: A big linear dark scar (coup de sabre) in the left side of mandibular mentum region

Intraorally, the most relevant alteration was the unilateral atrophy of tongue papillae of left side [Figure 4]. There was over-retained maxillary left deciduous canine with palatally erupted permanent canine leading to crowded left maxillary arch. The occlusion was disturbed with midline shifted to the left side. Also, there was hypoplasia of maxillary anterior teeth [Figure 5].

Radiographically, an abnormal root development was seen with maxillary left premolars. The over-retained root pieces of deciduous mandibular second molar of left side were seen on mesial and distal side of roots of second premolar. The overall eruption pattern of teeth on left side was retarded as compared to that of right side. The mandible showed decreased vertical height of ramus along with loss of gonial angle prominence on the affected side [Figure 6].

The patient presented with good general health condition without any history of obvious past illness that would explain the cause of facial asymmetry.

With these clinical and radiologic findings, the diagnosis of

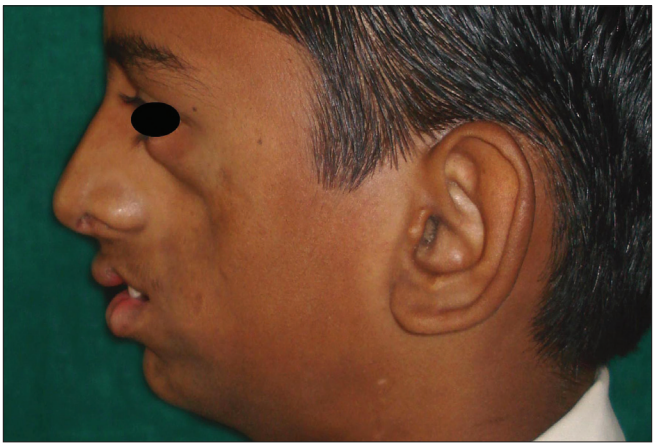

Figure 2: Hypoplasia of the left side of face with enopthalmy in left eye region

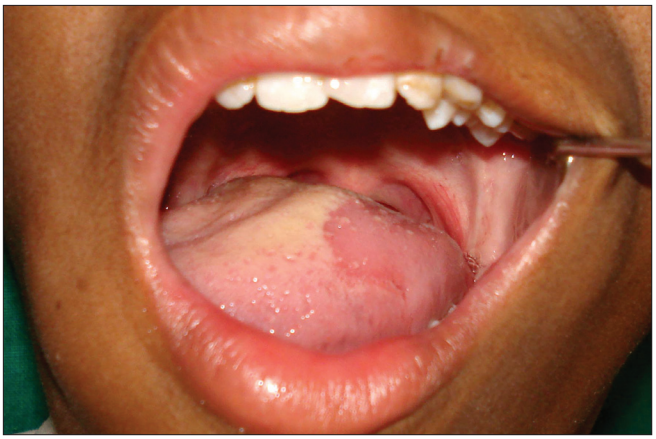

Figure 4: Unilateral atrophy of tongue papillae of left side

progressive hemifacial atrophy was made. At present, the patient is being periodically reviewed until atrophic manifestation stops and specific intervention could be accomplished.

Discussion

Progressive hemifacial atrophy, as described in the case above, is a rare pathology, of unknown cause, whose degenerative condition affect not only the esthetic but also the functionality of the attained hemiface.

The etiology of hemifacial atrophy has been the subject of numerous theories, which include heredity, viral infection, trauma, endocrine disturbances, autoimmunity, sympathetic malfunctions, trigeminal neuritis, and association with a connective tissue disorder, particularly scleroderma.[1,3] A cerebral disturbance on fat metabolism has also been proposed as a primary cause.[1] However, none of the theories withstands thorough investigation, and currently the cause of hemifacial atrophy remains unresolved.

The pathogenesis of progressive hemifacial atrophy is unknown. A supposed neurotrophic pathogenesis was

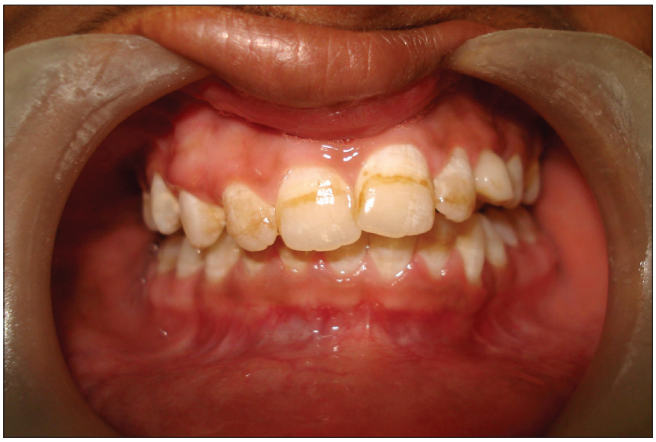

Figure 5: Hypoplasia of maxillary anterior teeth and deviation of midline to left side

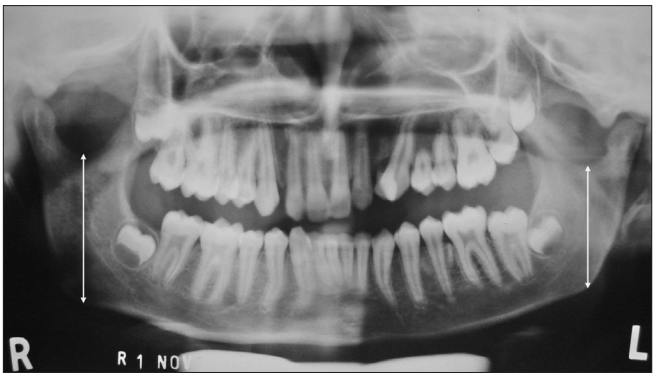

Figure 6: Retarded eruption pattern of teeth on left side compared to that of right side. Decreased vertical height of ramus along with loss of gonial angle prominence on the affected side

described by *Cassirer* in 1912.[4] He proposed that the atrophic disease process follows the pattern of trigeminal nerve innervation. Certain studies suggested that the disorder was familial one.[4] 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 tissues shrink by loss of adipose tissue.[4] Hence, atrophy that started in the second decade of life is less noticeable because facial growth is nearly complete. Early disease onset and long duration cause greater deformity.

Frequently, the onset of *Parry-Romberg Syndrome* occurs along first and second decades of life.[5] This syndrome seems to have higher incidence among women and affect left side of face most often.[5] Characteristically, the atrophy progresses slowly over many years and, then, it become stable.[1] Alternatively, the condition may "burn" itself out at a very early stage and result in minimal deformity.[3] Alterations concerning involvement, duration, and deformity can stabilize in any stage of growing and development. The extension of the atrophy is frequently limited to on one side of the face, and the ipsilateral involvement of body is rare.[1]

Clinically, the skin can be dry and with a dark pigmentation. Some patients present a demarcation line between normal and abnormal skin, reminding a big linear scar, known as *"coup de saber,"* as could be noticed in this patient.[1,6] Ocular involvement is common, and the most frequent manifestation is the enophthalmy, due to fat loss around the orbit, as was observed in the present case. The eye usually works normally. There may be localized areas of alopecia as was observed in present case in the region of left eyebrow. Occasionally, there may be some neurological complications, such as trigeminal neuralgia, facial paresthesia, severe headache, and contralateral epilepsy.[1,6] These complications were not discerned in the present case. Mouth and nose are deviated to the affected side, deviating also facial and dental midlines. Atrophy of superior lip led the anterior teeth to be exposed, and there may be also unilateral atrophy of tongue.[1,6]

The present case showed clearly those features of facial asymmetry, atrophy of lingual papillae; however, atrophy of the tongue was absent.

Radiographically, teeth on affected side can present some deficiency in root development and, consequently, delayed eruption.[7] However, the affected teeth are normal and vital clinically.[1,8] This situation occurred with the reported case, as two of the teeth on the affected side presented a malformation of their roots. Also, there was over-retention of deciduous canine causing delayed eruption of permanent successor and dental crowding. Very often, there is unilateral posterior crossbite, as a result of jaw hypoplasia and delayed teeth eruption.[8]

The intraoral soft tissue and chewing muscles are usually normal without any movement, speech, or deglutition implications.[1] Histologically, atrophy of epidermis, dermis, and subcutaneous tissue is observed.[1,5] Variable infiltrate of lymphocytes and monocytes at dermis and lack of subcutaneous fat in the affected tissue are also characteristic.[1] Besides, degenerative alterations can be identified on vascular endothelium in electron microscopy.[9]

Differential diagnoses include hemifacial microsomia (first and second branchial arch syndrome) and its variants, such as *Goldenhar syndrome*, but these are congenital and essentially non-progressive conditions. Post-traumatic atrophy and partial lipodystrophy (*Barraquer-Simon Syndrome*) are also included in the differential diagnosis. However, partial lipodystrophy is usually bilateral and involves primarily the adipose tissue.[9]

The relationship between *Parry-Romberg Syndrome* and localized scleroderma is controversial. It has been suggested that the term *Parry-Romberg syndrome* should be used for progressive hemifacial atrophy without features of cutaneous scleroderma.[10] However, in patients with *Parry-Romberg Syndrome,* cutaneous changes are also reported.[10] Therefore, localized scleroderma may be preceding lesion of progressive

hemifacial atrophy, and in patients with localized scleroderma, hemifacial atrophy may develop in several years.[10] Hence, regarding the clinical findings and clinical course, localized scleroderma and *Parry-Romberg Syndrome* may represent differential spectra of same disease process.[10,11] Though the relationship between scleroderma and *Parry-Romberg Syndrome* remains unclear, the former usually responds to drug therapy whereas later is progressive.[9]

Parry-Romberg Syndrome is an auto-limitable condition and there is no cure. Affected patients should have multi-disciplinary attendance of physicians, dentists, phonoaudiologists, and psychologists. The treatment is usually based on reposition of adipose tissue that was lost due to atrophy.[12] Autogenous fat grafts, cartilage grafts, silicon injections and prostheses, bovine collagen, and inorganic implants are some alternatives to esthetic correction of the atrophy.[12] Besides esthetic improvement, symptomatic treatment for neurological disorders is indicated.[12] The cosmetic treatment is just recommended when the illness stops its evolution, so this is the reason why this patient has not been submitted to any surgical intervention yet.

Conclusion

In conclusion, a case of progressive hemifacial atrophy, with its archetypal feature, was discussed. Till recently, the exact etiology and pathogenesis of this degenerative condition have not been elucidated. Many patients present with classic clinical features and there is little intricacy in diagnosis of progressive hemifacial atrophy. Still, some clinical conditions, especially scleroderma, should be kept in mind while dealing with this disorder. Proper diagnosis and multidisciplinary treatment approach is essential for management of progressive hemifacial atrophy.

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