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Case Report

A Tale of Two Diseases: Parry Romberg Syndrome and En Coup De Sabre: A Case Report

Muhammad Erfan, Ikram ullah khan, Syed Afaq Ahmed, Saqib Hasnain
Department of Dermatology Pakistan Institute of Medical Sciences Islamabad

Abstract

Background: Parry Romberg syndrome is a very rare disorder of unknown etiology. A degenerative acquired disorder characterized by hemifacial atrophy. It can present with skeletal, dental and soft tissue abnormalities with or without CNS symptoms. Majority of the affected patients have manifestations of the disease in the first two decades of life. Many cases have been documented having combination of Parry Romberg syndrome and another form of localized scleroderma- *en coup de sabre morphea*. Histological features of the disease include fibrosis with atrophy of adipose tissue and lack of any inflammatory infiltrate. Limited treatment options make the disease difficult to manage.

Case presentation: We present a case of an otherwise perfectly healthy, 12 years old girl with early features of hemi-facial atrophy and en coup de sabre morphea.

Conclusion: Both Parry Romberg syndrome and en coup de sabre morphea have many localized and systemic associations. In many case reports neurological and dental abnormalities have been described. In our case patient did not appear to have any of these.

Keywords: Parry Romberg syndrome, hemifacial atrophy, en coup de sabre morphea.

Introduction

Parry Romberg syndrome (PRS) also known as progressive hemifacial atrophy is a rare progressive but self limiting degenerative disorder characterized by the atrophy of the skin and subcutaneous tissue mainly of one half of the face.[1] The disease was initially described by Parry in 1825, and then subsequently by Romberg in 1846, in 1871 Eulenberg also provided some insight into this rare and poorly understood syndrome and labeled it as progressive hemifacial atrophy.[2]

Progressive loss of the subcutaneous fat occurs, accompanied sometimes with the atrophy of skin, muscles, bone and cartilage.[3] Hemiatrophy usually affects the maxillary region[4] and has a particular predilection for the dermatomes supplied by the branches of the fifth cranial nerve[5, 6], but any area of the face can be involved. Atypical presentation where there is bilateral involvement of the face, involvement of the ipsilateral or contralateral body has also been documented.[4]

PRS is much more common in females[7-9]. True incidence of the disease is unknown also there is an ambiguity about its cause. This might be either due to the rarity of the disease or because of lack of any standardized criterion for its diagnosis[5]. Many theories have been proposed as to the causative factor for the disease these include: Cerebral dysfunction of fat metabolism, trauma, viral infections, endocrine disturbances, autoimmunity and heredity.[8, 10-14]

PRS tends to present in the first two decades of life[15]. With the disease being more severe if it presents in the first decade of life. The disease then progresses in the following years (usually 2 -10 years) and subsequently burns out.[16]

Correspondence: Dr. Saqib Hasnain
Department of Dermatology Pakistan Institute of Medical Sciences Islamabad

Extracutaneous features include: epilepsy, migraine, trigeminal neuralgia and headaches[17, 18] with partial seizures being the most common neurological complication[19]. Ocular involvement in the form of enophthalmos is also a common finding.[20] The teeth may be small and have short roots.[21] Maxillary and mandibular hypoplasia together with atrophy of oral cavity structures like tongue, gum, lips and salivary glands can also occur.[22-26] Skull may also be involved.[27]

Diagnosis of PRS is mainly a clinical one, histopathology may help in diagnosis. Skin biopsy may reveal dermal sclerosis, fat atrophy, and decrease in adnexal structures[28]. Epidermal, dermal, adnexal structure and subcutaneous tissue atrophy has been documented.

PRS overlaps with a linear scleroderma "en coup de sabre"(ECDS).[29] At times it is very difficult to differentiate the two. ECDS usually manifests as a linear depression in the frontal or parietal scalp or the paramedian forehead[30]. It has a unilateral distribution and may extend below the forehead progressively involving the nose, cheek or rarely the upper lip[5]. The affected skin is shiny, firm and displays focal hair loss. The affected area resembles just like a "stroke from a sword". Many studies have tried to provide a differentiating clue between PRS and ECDS but it is generally believed that the two lie on the same spectrum of the disease.[31-33] Atrophy of the dermis, epidermis, subcutaneous tissue and adnexal structures has been seen in both PRS and ECDS.

Affected patients should have a multidisciplinary approach involving the physicians, neurologist, plastic surgeons, OMF surgeons, radiologist, dentists, phonoaudiologists and psychologists. Cosmetic surgery involving autogenous fat graft is the mainstay of treatment[34]. Other therapies including injections of silicone and bovine collagen, along with permanent fillers are also underway.

The objective of this work is to present case report of a patient with a combination of PRS and ECDS with absence of neurological symptoms, dental anomalies or ophthalmic symptoms.

Case Report

A 12 years old girl student of class 5th presented in outpatient department of dermatology with complaints of facial and nasal deformity. History from mother revealed that the patient had been well with normal facial features till 9 years of age. Afterwards the mother noticed slow "shrinking" of the face in the area of right jaw. Two and a half year back the patient slowly started developing a longitudinal furrow on the nasal tip. There was no prior history of any trauma or fever. There was also absence of any symptoms pertaining to the nervous system. Patient didn't have any complaints regarding her vision. Systemic inquiry showed no significant abnormalities. She had no comorbids. There was no family history of similar complaints.

Examination revealed an otherwise healthy girl with normal vitals. She was alert with intact higher motor functions. There were no signs of any psychological or mental instability. Face showed a slight asymmetry in the lower half due to right mandibular area deformity. This area appeared to be atrophic as compared to the other side (figure 1a), with the underlying veins becoming visible (figure 1b).

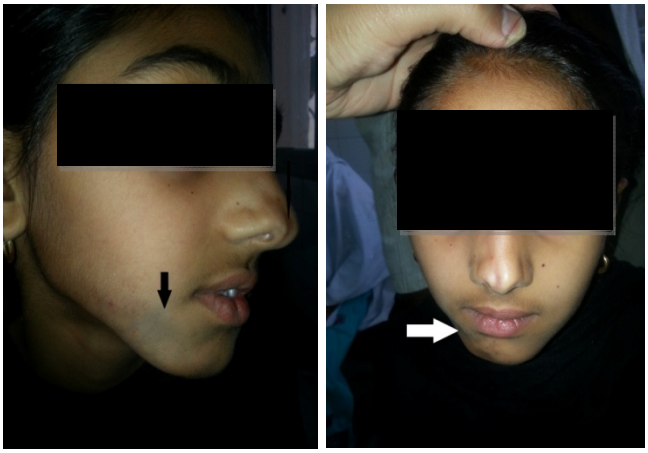

Figure 1 (a & b) Parry Romberg syndrome & en coup de sabre, a: facial asymmetry in the right madibular area (white arrow), b: visible veins in the affected area (black thick arrow) also a linear scar like depression (ECDS) is visible on the nasal tip (black thin arrow).

The lip appeared to be dragged more to the affected side. Also evident was a linear scar like defect at the nasal tip. (Figures 2 & 3)

No bony deformity was observed also there was absence of any alopecia. Eyes were symmetric and in the normal alignment. Intra-oral examination showed no teeth abnormalities and the tongue was also without any deformity. The patient was given a preliminary diagnosis of Parry Romberg syndrome and en coup de sabre and was advised baseline labs, radiographs of skull and a skin biopsy from the right mandibular area. Neurology, ophthalmology and dentistry department consultations were also sought.

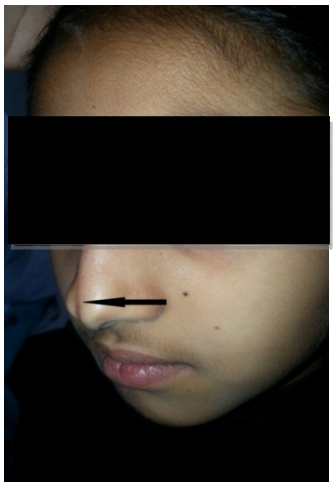

Figure 2: en coup de sabre, linear furrow on the nasal tip the "stoke from a sword"

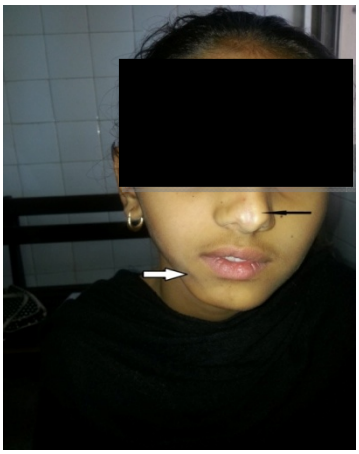

Figure 3. (Combination of PRS and ECDS marked by the white and black

The skin biopsy revealed marked epidermal, appendageal and adipose tissue atrophy. The basal cells appeared "irritated" with slight hyper pigmentation. Both papillary and reticular dermis showed significant fibrosis. (Figures 4-6)

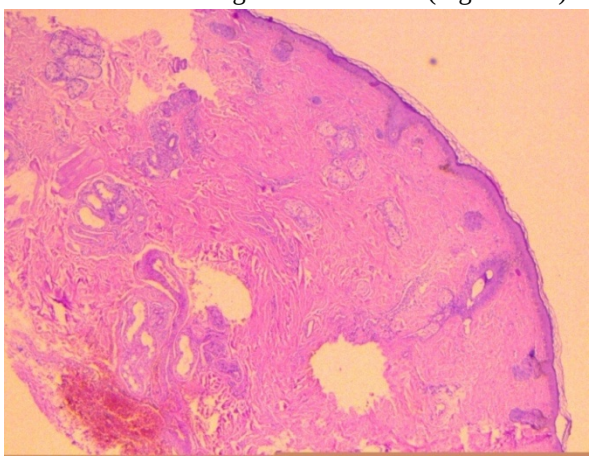

Figure 4. Skin biopsy showing epidermal atrophy with loss of rete ridges extensive dermal fibrosis and loss of adipose tissue (H&E, ×40)

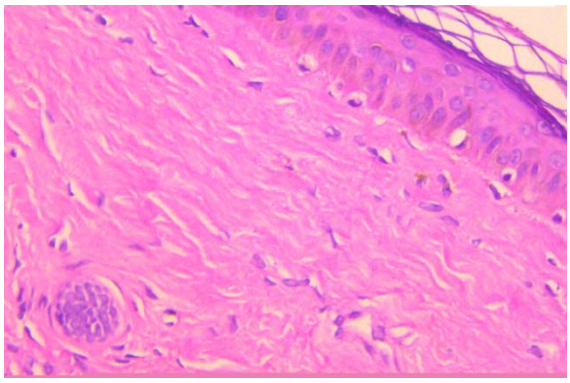

Figure 5. Epidermal atrophy (H&E, ×400)

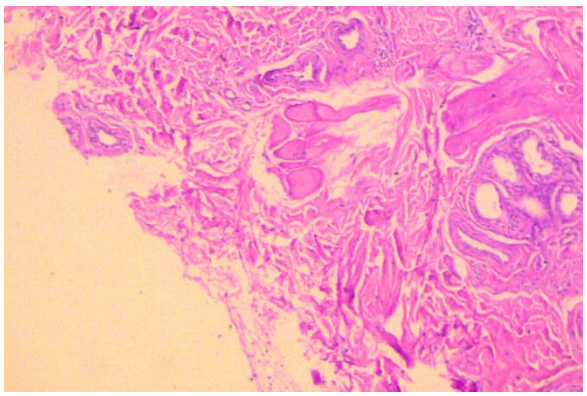

Figure 6. Reticular dermis with dermal fibrosis and atrophy of adipose tissue(H&E, ×100)

Based on these finding a diagnosis of PRS with ECDS was made, the family was counseled about the nature of the disease, its course and the possible treatment options. They were also advised strict follow-ups in

order to assess the progression of the disease and to make a final decision on the surgical correction of the problem once the major growth takes place and the disease burns out.

Discussion

PRS is an uncommon, slowly progressive, generally unilateral atrophic disorder of unknown etiology[7]. Cerebral dysfunction of fat metabolism, trauma, viral infection, endocrine, autoimmunity and hereditary have been proposed as the possible pathologies[8, 10-14]. However, there was no such cause in our patient. PRS is believed to have its onset in the first or second decade of life. Patients that have early onset disease especially in the first decade have more severe disease manifestation. However, this was not the case in our patient. The disease slowly progresses over next 2-10 years and then burns out.[27] In some patients the disease can re-activate. The duration of the disease is the most important factor deciding the degree of deformity[35]. PRS is frequently associated with CNS, ophthalmic symptoms and dental problems, however in our case the patient did not complain of any of these. Inigo et al., proposed a classification for PRS and divided it into mild, moderate and severe.

1. **Mild**: Atrophy of the skin and subcutaneous tissue affecting the territory of only one of the sensory branches of the trigeminal nerve; no bone involvement.
   2. **Moderate**: two trigeminal territories affected; no bone involvement.
2. 3. **Severe**: all three trigeminal territories affected or bone involvement.[36]

PRS frequently overlaps with linear scleroderma-"*en coup de sabre*". ECDS usually manifests as a linear furrow in the frontoparietal scalp or the forehead, in our patient ECDS linear furrow started in the nasal region with a solitary mark at the nasal tip. There was absence of hair loss, scalp or forehead lesion.

Histology showed atrophy of epidermis with loss of Rete ridges, fibrosis of dermis and loss of adipose tissue consistent with PRS.

Methotrexate, steroids, immunosuppressive agents have been tried for the treatment of PRS in order to halt the disease process. Surgical transposition of fat, collagen and fillers are used to correct the facial defect when the disease burns out.

PRS, in short can present in a number of ways and is frequently associated with ECDS. Further work up is required to ascertain the definite etiology of the disease.

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