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CASE STUDY

PARRY ROMBERG SYNDROME WITH EN COUPE DE SABRE: A RARE CASE REPORT

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ABSTRACT

Parry Romberg Syndrome (PRS) is relatively rare condition of debatable etiology, usually restricted to one side of the face, rarely being bilateral or involving the limbs. Characteristically, the atrophy progresses slowly for several years and become stable after certain time period. After stabilization of the disease multispeciality approach including physicians, dental surgeons, psychologists and reconstructive surgeries can be performed to correct the deformity. PRS is frequently associated with localized scleroderma, known as "en coupe de sabre" (ECDS). A debate exists whether PRS is a form of linear scleroderma or these conditions are inherently different processes or appear on a spectrum. We present a case of progressive hemifacial atrophy of left side of face in a 28 year old male with ECDS without any neurological or ophthalmic complications.

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INTRODUCTION

Progressive facial hemiatrophy (PFH) was first described by Caleb Parry in 1825 and isolated as a clinical entity by Moritz Romberg in 1846. It was referred to as "progressive facial hemiatrophy" by Eulenberg in 1871 but "progressive hemifacial atrophy" (PHA) is a more appropriate term (Gorlin and Pindborg) (Johnson *et al.,* 1969; Wartenberg *et al.,* 1945 and Yochanan Goldhammer *et al.,* 1981). Parry Romberg Syndrome (PRS) is a relatively rare, uncommon, degenerative, poorly understood condition. It is characterized by slow progressive atrophy of one side of the face affecting variably skin, subcutaneous fatty tissue, muscle, connective tissue and bone (Lakhani *et al.,* 1984).

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The apparent decrease in muscle mass appears to be due to loss of the fatty and connective tissue constituents of muscle and usually is not accompanied by weakness (Johnson *et al.,* 1969). PHA refers to hemifacial atrophy of the skin and craniofacial tissues inferior to the forehead, generally following the pattern of sensory innervations of one or all three trigeminal nerve dermatome (Duymaz *et al.,* 2009). It may be accompanied usually by contralateral Jacksonian epilepsy, trigeminal neuralgia, and changes in the eyes and hair (Jon Stone *et al.,* 2003). Linear scleroderma 'en coup de sabre' (ECDS) has a slowly progressive course and is generally limited to one side of face. It is characterized by atrophy and furrowing of the skin of the frontoparietal region. In most cases it occurs as a single, paramedian line with contraction and stiffness of the affected area forming a depressed groove and extending to the scalp developing an area of alopecia. It affects mainly children and is predominant in females (3:1). The involved skin is hyperpigmented, shiny and firm (McKenna and Benton, 1999

and Careta *et al.,* 2015). There is an age old debate regarding the relationship between PHA and ECDS because of similar clinicopathological features. PHA may be clinically very similar to linear scleroderma, and they may coexist in about 20-37% of patients, which makes it difficult to distinguish between them (Stone, 2006).

Case Report

A 28 year old male patient reported to the Private Dental clinic, at Mumbai, with complain of progressive deformity and sinking of left side of face since past 7-8 years and is stable since past 6 months. Past medical and dental history was not contributory. No history of trauma was elicited. On general physical examination patient was conscious, oriented, a febrile, general condition was fair and vital signs were stable. Extraoral examination revealed facial asymmetry with hypoplasia of the left side of face involving chin and cheek giving it a sunken appearance and thinning of lips with preserved sensibility, strength and mobility. Also seen were two paramedian linear depressions on the left side of forehead and extending onto the nose. Overlying skin was shiny, hyperpigmented, firm and displayed alopecia. Intraoral examination was not remarkable. Orthopantomogram did not reveal any abnormalities except for missing 26 and impacted 38, 48. Ocular examination and neurological examination showed no irregularity. There was no evidence of sensory or motor deficits on both sides of face. Based on history and clinical findings a diagnosis of PHA with ECDS was formulated.

DISCUSSION

Epidemiology

The prevalence of PHA is at least 1/700,000 and common in females (F:M = 3:2). The disease appears in the first or second decade of life, cases beginning in the second decade having less interference with overall growth of the facial bony skeleton (Wartenberg 1945). Both sides of the face can be affected with equal frequency (Rogers 1963) (Lakhani *et al.,* 1984 and Janowska *et al.,* 2013).

Etiopathogenesis

The etiopathogenesis of PHA is obscure (Lakhani *et al.,* 1984). Autoimmune etiology is best favoured. The skin pathology if caught at the onset shows inflammatory changes. Supportive evidence for an inflammatory hypothesis includes: a higher frequency of autoantibodies than the general population; the overlap with linear scleroderma and vitiligo (Stone *et al.,* 2006 and Janowska *et al.,* 2013). The trigeminal theory considers the wasting process a result of trauma to the superior cervical ganglia (Gulati, 2006). Trauma has been implied but is an inconsistent feature. Wartenberg stated that trophism of the fat and subcutaneous tissues is under the influence of the sympathetic nervous system. Cory favors the hypothesis of hyperactivity of the sympathetic nervous system, specifically, inflammation of the superior cervical ganglion in a young patient, as the cause of progressive facial hemiatrophy. Hyperactivity of the sympathetic nervous system and its division into medial and lateral internal carotid nerve branches provide an anatomic basis for the unilateral facial and intracranial manifestations of the disease (Cory *et al.,* 1997).

Clinical Course

PHA is a craniofacial disorder with onset in the first or second decade of life in individuals who are morphologically normal at birth (Miller, 1995). Pensler evaluated 41 patients with Romberg's disease and observed that the mean age of onset in patients with skeletal involvement was 5.4 years, versus 15.4 years for patients without skeletal involvement. However, there was no correlation between the severity of soft-tissue deformity and the age of onset (Mazzeo *et al.,* 1995). PHA usually has an insidious onset but unrelenting progressive course over a period of 2-10 years often resulting in facial disfigurement, functional, esthetic and psychological complications followed by a burning out of the atrophic process with subsequent stability and varying degree of deformity (Lakhani *et al.,* 1984; Mazzeo *et al.,* 1995). In most cases the abnormality, is anatomically limited to one side of the face and cranium. Atypical presentation where there is bilateral involvement of the face, involvement of the ipsilateral or contralateral body has also been documented. Where the atrophy meets normal tissue on the other side of the face, it may produce a "line", giving rise to the ECDS (a slender rod-like mark) description often noted in these patients. Involved side demonstrates slow involution and atrophy affecting many tissues, with loss of subcutaneous fat and dermal atrophy, and resultant tissue contraction. If onset is in the first 2 decades, secondary bony changes may occur. These findings are in stark contrast to the contralateral normal side (Janowska *et al.,* 2013 and Pensler *et al.,* 1990). In addition to cutaneous atrophy, numerous ocular and neurologic complications have been reported. Migraine and facial pain are the commonest neurological symptoms (Stone *et al.,* 2006 and Pensler *et al.,* 1990). A brief summary of the manifestations is mentioned in Table 1 (with estimates of their frequency) (Stone, 2006).

Clinical features
Facial hemiatrophy of fat, skin, connective tissue, muscle, and/or bone (100%)
Hemiatrophy of contralateral or ipsilateral arm, trunk, or leg (20%)
Atrophy of tongue (25%)
Dental abnormalities (50%)
Trismus / jaw symptoms (including hemi-masticatory spasm) (35%)
Migraine/facial pain (45%)
Ocular abnormalities including globe retraction, uveitis, pupillary abnormalities, restrictive ocular myopathy (mimicking Duane's syndrome), heterochromia
Epilepsy (10%), sometimes associated with ipsilateral brain changes on MRI (5%)
Vitiligo, hair depigmentation/hyperpigmentation (20%)
Brain MRI abnormalities—usually ipsilateral but sometimes contralateral in grey and white matter

Association of PHA and ECDS

Most discussions on the relationship between PHA and ECDS eventually condense into two considerations; either two distinct disorders or a continuum. Wartenberg described ECDS as an abortive form of PHA, whereas Wolf and Ehrenclou believed that PHA is not a distinct disease but a syndrome that may coexist with linear scleroderma or occur as a sequel of various conditions. Some authors have described patients with ECDS converting with time into PHA (Careta, 2015). Tollefson and Witman reviewed 54 patients with ECDS or PRS at Mayo Clinic and observed that the condition coexisted in 28% patients and concluded that both are likely variants of morphea (Pensler, 1990). While the two may coexist in the same patient, in most cases it is possible to differentiate them based on clinicopathological findings. Orozco-Covarrubias L

(2002) evaluated 13 patients diagnosed as ECDS and 9 patients as PRS clinically and histopathologically. They concluded that the most important distinguishing feature was cutaneous sclerosis present in eight of 13 patients with ECDS against none in PRS. Other clinical features more frequently found in ECDS were cutaneous hyperpigmentation and alopecia. Histopathological features were connective tissue fibrosis present in all cases with ECDS and two of nine patients with PRS; adnexal atrophy present in 11 of 13 patients with ECDS, and in three of nine with PFH, and mononuclear cell infiltrates in all patients with ECDS against six with PHA (Orozco-Covarrubias *et al.,* 2002).

Comparison of PHA with ECDS findings PFH (Careta and Romiti, 2015; Tolkachjov *et al*., 2015)

PHA	ECDS
Unilateral atrophy	Unilateral, frontoparietal sclerotic band (Usually does not extend below the eyebrow)
Minimal or absent induration or previous inflammation	Usually preceded by skin induration
Cutaneous atrophy (normal hair and absent sclerosis)	Depressed, hyperpigmented, shiny cutaneous sclerosis involving the scalp
Associated with atrophy of the subcutaneous tissue, fat, muscle, osteocartilage	Frequently causes deformity and contractures
Atrophy of tongue, teeth, gingiva	
Cranial neuropathies, vision loss, seizure disorders	

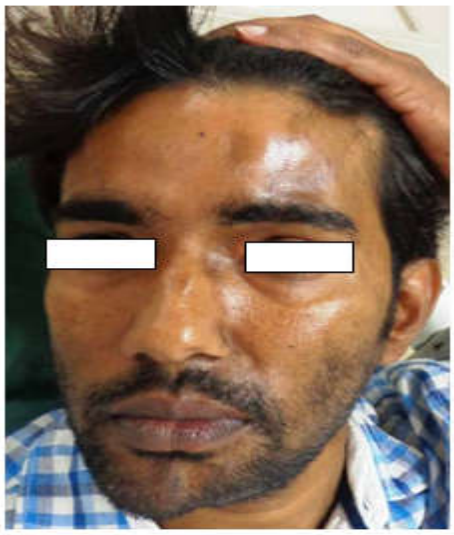

Figure 1. Left side facial changes in a 28 year old patient with PHA; marked atrophy of cheek, chin and lips and linear depression present over left forehead extending over median side of nose lip and chin

Oral Manifestations

Teeth abnormalities and involvement of the mandible and masticatory muscles is frequently seen in patients with PHA. Oral changes include maxillary and mandibular hypoplasia, unilateral atrophy of muscle of the tongue, atrophy of papillae, lips, salivary glands and gingiva. Other features include missing teeth, atrophic root development, retarded root formation, root resorption, oligodontia, microdontia, dilacerations, unilateral crossbite (due to delayed eruption and jaw hypoplasia) and pulp stones on the affected side. The teeth often appear clinically normal, have regular enamel, dentin, cementum, and pulp, and can test vital (Reddy *et al.,* 2012 and Al-Aizari *et al.,* 2015 and Deshingkar *et al.,* 2012).

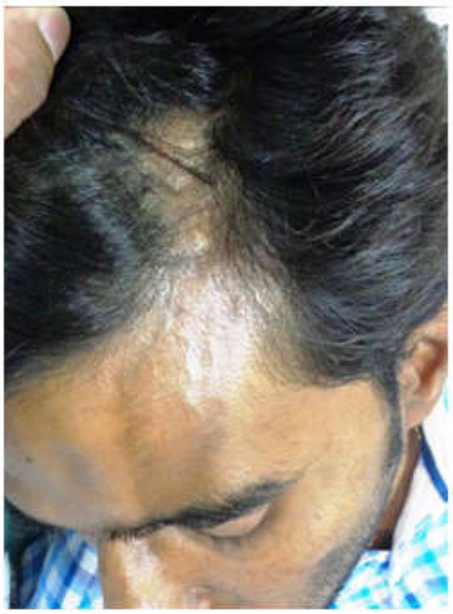

Figure 2. Shiny, hyperpigmented skin and alopecia of frontoparietal scalp

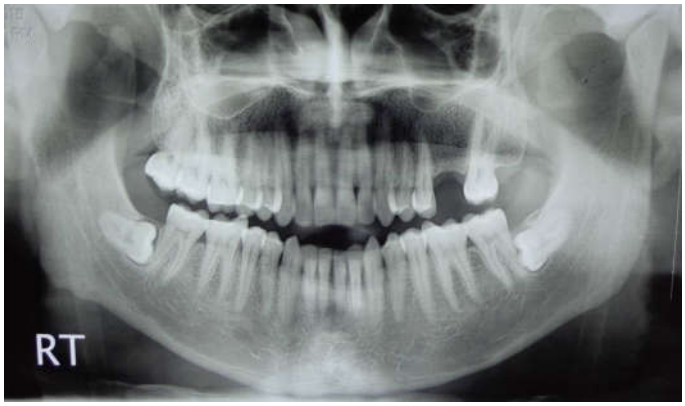

Figure 3. Orthopantomogram showing missing 26 and horizontally impacted 38, 48 without any bony abnormality

Diagnosis

No universal diagnostic criteria are accepted. For a patient who only has facial asymmetry, a clinical diagnosis can be made without investigations. MRI is the brain imaging of choice for patients with neurological symptoms (Stone, 2006). Positive anti-nuclear antibody is the most common laboratory abnormality, with approximately 25-52% of patients having an elevated titer (Tolkachjov *et al.,* 2015) Histopathology of affected areas in PHA, with or without clinical findings of a sclerotic process, shows homogenized dermal sclerosis, fat atrophy, decrease in adnexal structures, and perivascular plasma cells and lymphocytes and abnormalities of vascular endothelium and basement membranes (Careta *et al.,* 2015 and Orozco-Covarrubias *et al.,* 2002).

Differntial Diagnosis

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. Also it shoud be carefully distinguished from fat necrosis, localised scleroderma, Bell's palsy, unilateral ankylosis and trauma (Miller *et al.,* 1995).

Treatment

Treatment for PHA can be challenging. The primary aim is to stop the active disease process. Methotrexate is the standard therapy for active disease and is often combined with oral prednisone. The treatment often is based on the replacement of tissue that was lost due to atrophy (Tolkachjov *et al.,* 2015). Autologus fat tissue grafts are the most frequently used due to a lower risk of tissue rejection and, consequently, smaller local and systemic inflammatory reaction. Alternative methods are application of dermal fillers agents such as bovine collagen implants, silicone implants and injections or filling with hyaluronic acid. However, fat injections may simply be resorbed if the disease is still active. Proper mandibular development and parallelism of facial planes can be guided with orthodontic rehabilitation and orthodontic appliances (Tolkachjov *et al.,* 2015; Alencar *et al.,* 2011). American Society of Plastic and Reconstructive Surgery concluded that only 30% of the transferred adipose tissue remains viable after one year and recommended overcorrection (Alencar *et al.,* 2011).

Conclusion

No universal diagnostic criteria are accepted in the diagnosis of PHA. The diagnosis is largely based on the characteristic clinical features including presence of unilateral idiopathic facial atrophy, typically involving the lower face, without significant epidermal change. Patients are typically classified as having ECDS if they demonstrate linear scleroderma of the front parietal scalp with involvement of medial or paramedian forehead with cutaneous sclerosis. Thus we are presenting a very interesting case of PHA with overlapping ECSD. Close association and coexistence of scleroderma and PHA warrants thorough clinical examination and sound knowledge to identify these conditions.

REFERENCES

Al-Aizari, N.A., Azzeghaiby, S.N., Al-Shamiri, H. M., Darwish, S., Tarakji, B. 2015. Oral manifestations of Parry–Romberg syndrome: A review of literature. *Avicenna J Med.,* 5(2):25-28.

Alencar, J.C.G., Andrade, S.H.C., Pessoa, S.G.P., Dias, I.S. 2011. Autologous fat transplantation for the treatment of progressive hemifacial atrophy (Parry-Romberg syndrome: a case report and review of medical literatute). *An Bras Dermatol,* 86(4 Supl 1):S85-8.

Careta, M.F., Romiti, R. 2015. Localized scleroderma: clinical spectrum and therapeutic update. *An Bras Dermatol* 90(1):62-7.

Cory, R.C., Clayman, D.A., Faillace, W.J., McKee, S.W., Gama, C.H. 1997. Clinical and radiologic findings in progressive facial hemiatrophy (Parry-Romberg syndrome). *Am J Neuroradiol.,* 18(4):751–7.

Deshingkar, S.A., Barpande, S.R., Bhavthankar, J.D., Humbe, J.G. 2012. Progressive hemifacial atrophy (Parry-Romberg Syndrome). *Contemp Clin Dent*., 3:78-81.

Duymaz, A., Karabekmez, F.E., Keskin, M., Tosun, Z. 2009. Parry-Romberg syndrome: facial atrophy and its relationship with other regions of the body. *Ann Plast Surg* 63(4):457–61.

Gulati, S., Jain, V., Garg, G. 2006. Parry Romberg syndrome. *Indian J Pediatr.,* 73(5):448-449.

Janowska, M., Podolec, K., Lipko-Godlewska, S., Wojas-Pelc, A. 2013. Coexistence of Parry-Romberg syndrome with homolateral segmental vitiligo. *Postępy Dermatol Alergol,* 30 (6):409-411.

Johnson, R.V., Kennedy, W.R. 1969. Progressive facial hemiatrophy (Parry-Romberg syndrome): Contralateral extraocular muscle impairment. *AmJ Ophthalmol*., 67: 561-564.

Jon Stone. 2003. Parry Romberg Syndrome, A global survey of 205 patients using the internet. *Neurology*, 61:674-76.

Kodama, Y., Miyazawa, M., Fukuda, J.I., Iida, A., Ono, K., Takagi, R. 2005. Progressive hemifacial atrophy treated by orthodontic surgery. *Oral Science International*, 2 (2):131-135.

Lakhani, P.K., David, T.J. 1984. Progressive hemifacial atrophy with scleroderma and ipsilateral limb wasting (Parry-Romberg syndrome). *J R Soc Med*., 77(2):138–9.

Mazzeo, N., Fisher, J.G., Mayer, M.H., Mathieu, G.P. 1995. Progressive hemifacial atrophy (Parry-Romberg syndrome). *Case report. Oral Surg Oral Med Oral Pathol Oral Radiol Endod,* 79 (1):30-5.

McKenna and Benton. 1999. A tri-linear pattern of scleroderma 'en coup de sabre' following Blaschko's lines. Clinical and Experimental Dermatology, 24:467–468.

Miller, M.T., Spencer, M.A. 1995. Progressive hemifacial atrophy. A natural history study. *Trans Am Ophthalmol Soc.,* 93: 203–15.

Orozco-Covarrubias, L., Guzman-Meza, A., Ridaura-Sanz, C., Carrasco Daza, D., Sosa-de-Martinez, C., Ruiz-Maldonado, R. 2002. Scleroderma 'en coup de sabre' and progressive facial hemiatrophy. Is it possible to differentiate them? *J Eur Acad Dermatol Venereol,* 16(4):361–6.

Pensler, J.M., Murphy, G.F., Mulliken, J.B. 1990. Clinical and ultrastructural studies of Romberg's hemifacial atrophy. *Plast Reconstr Surg.,* 85(5):669-74.

Reddy, S.S., Devaraju, D., Kunal, A. 2012. Parry Romberg syndrome: Report of two cases with rare dental and skeletal findings. *EJ Dent.,* 2: 212-5.

Stone, J. 2006. Parry-Romberg syndrome. *Practical Neurology* 6: 185-188.

Tolkachjov, S.N., Patel, N.G., Tollefson. 2015. Progressive hemifacial atrophy: a review. *Orphanet Journal of Rare Diseases*, 10: 39.

Tollefson, M.M. and Witman, P.M. 2007. En coup de sabre morphea and Parry-Romberg syndrome: A retrospective review of 54 patients. *J Am Acad Dermatol.,* 56: 257-63.

Wartenberg, R. 1945. Progressive facial hemiatrophy. Arch Neurol Psychiatr, 54: 75-96.

Yochanan Goldhammer, Jona Kronenberg, Rina Tadmor, Jackson Braham and George Leventon. Progressive hemifacial atrophy (Parry-Romberg's disease), principally involving bone. *The Journal of Laryngology & Otology* 1981; 95: 643-647.

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