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A Case of Papillitis in Parry-Romberg syndrome; A Rare Presentation of a Rare Disease

CHOWDHURY A[1], ZANNAT T[2], SAHA S[3], MUZAHID MAA[4], HASAN M[5], ROY U[6], KABIR MS[7], BULBUL S[8], ZANNAT T[9],ISLAM MR[10]

Abstract:

Progressive Hemifacial Atrophy or Parry-Romberg syndrome (PRS) is a rare idiopathic disease which is characterized by mostly unilateral facial atrophy of the skin and soft tissues. PRS has been associated with various systemic manifestations, particularly neurologic, ophthalmologic and maxillofacial. In the present report, a 16-year-old female patient with hemifacial atrophy and ipsilateral papillitis, accompanied by a short review of literature.

Keywords: *Papillitis, Hemifacial Atrophy, Parry-Romberg Syndrome.*

Introduction:

Parry-Romberg syndrome (PRS) was first described by Parry in 1825 and Romberg in 1846. PRS is a rare, acquired disorder characterized by progressive hemiatrophy of the skin and soft tissue of the face and in some cases, results in atrophy of muscles, cartilage, and the underlying bony structures[1,2]. PRS usually begins in the first decade of life[1-4] although some cases with a late onset have been described[5,6]. It is more common in females and is believed to be sporadic, although some rare familial cases have been reported [7]. PRS slowly progresses over 2 to 20 years before stabilizing[1,2,8-14]. It is typically restricted to one half of the face but occasionally involves the arm, trunk, and leg[2,5,15,16]. Bilateral progressive atrophies have been described[7].

The exact pathogenesis of PRS remains unclear, but seems to be heterogeneous. There are various proposed causes for this self-limiting disease: congenital mechanisms, disturbance of fat metabolism, trauma, infectious agents, heredity, cranial vascular malformations, immune-mediated processes, hyperactivity of the brain stem sympathetic centers, cervical sympathetic dysfunction, and trigeminal neurovasculitis[17].

Parry-Romberg Syndrome/progressive hemifacial atrophy is an auto-limitable condition and has some differentials such as localized scleroderma, traumatic fat necrosis, hemifacial microsomia, atrophy secondary to facial paralysis and unilateral partial lipodystrophy[18]. Given that the most frequent systemic manifestations associated with PRS are neurologic (affecting around 15% of the PRS population), some authors have proposed that PRS be regarded as a neurocutaneous syndrome[7]. The neuro-ophthalmic examination may reveal third nerve palsy, optic neuropathy, Horner syndrome, and tonic pupil. There may be pupillary disturbances, anisocoria with mydriasis or miosis on the affected side, in some cases and only a few case reports of optic nerve disease like papillitis or neuroretinitis[17]. There is very little knowledge about the efficacy of agents used to treat this disease.

1. Dr. Ashish Chowdhury, Phase B Resident, Dept of Neurology, BSMMU
2. Dr. Tahira Zannat, Phase B Resident, Dept of Neurology, BSMMU
3. Dr. Sujan Saha, Phase B Resident, Dept of Neurology, BSMMU
4. Dr. Md. Abdullah Al Muzahid, Phase B Resident, Dept of Neurology, BSMMU
5. Dr. Mehedi Hasan, Phase B Resident, Dept of Neurology, BSMMU
6. Dr. Uttam Roy, Phase B Resident, Dept of Neurology, BSMMU
7. Dr. Md. Suman Kabir, Phase B Resident, Dept of Neurology, BSMMU
8. Dr. Shahida Bulbul, Phase B Resident, Dept of Neurology, BSMMU
9. Dr. Taposhi Zannat, Assistant Surgeon,Sadar Hospital Jhenaidah
10. Prof. Md. Rafiqul Islam, Professor and Chairman, Dept of Neurology, BSMMU

Systemic medications used to treat other forms of linear scleroderma including antimalarials, methotrexate, local or systemic steroids, tetracycline, and cyclophosphamide are commonly used for PRS[18]. Local therapeutic options include emollients, vitamin D3 analogues (+ PUVA (Psoralen plus ultraviolet A)) and phototherapy[18]. Ophthalmological treatment concentrates on stabilization or rehabilitation of (peri-) ocular complications. Ocular inflammation may require immunosuppressive therapy. After stabilization, surgical reconstruction using silicone implants, muscle flap grafts, galeal flaps, fat grafts, bone and cartilage grafts, or injectable dermal filler are used to restore natural facial contours[18]. The most promising cosmetic results recently described are autologous fat grafting with adipose-derived stem cells[17].

Case Report:

In 2019 a 16-year-old female presented to department of Neurology, BSMMU (Bangabandhu Sheikh Mujib Medical University), Shahbag, Dhaka, Bangladesh with history of wasting of right side of her face for 6 years and progressive loss of vision of right eye for 2 years. Patient stated that she noticed depression over her right forehead and orbit 6 years back, which then gradually progress to her right maxilla and finally right mandible. 2 years back she developed blurring of vision in her right eye which was slowly progressive but not painful. There was no history of trauma, skin tightening, rash, discoloration, pain, fever, joint swelling, toxin exposure or other systemic illness. On enquiry she denies presence of facial weakness and chewing difficulty. No other cases of PRS were reported in the family. On physical examination: Her vitals were within normal limit (Blood pressure 110/70 mm Hg with no postural drop, Pulse: 80/min, regular). There was wasting of right side of the face which extended from cranium to lower jaw [figure 1]. On ophthalmology examination: She had right enopthalmus. Right pupil was slightly dilated, Pupillary reflex sluggish was in right eye. Her best corrected visual acquity was finger counting 3 feet in right eye and 6/6 in left eye. Colour vison was dysmorphic in right eye. Fundoscopic examination– Right disc was hyperemic with blurred margin and engorged veins [figure 2]. All othercranial nerves were normal in both sides. There was no malocclusion of teeth or change in voice. Thorough physical examination revealed no wasting or fasciculation in any part of body and other neurological, musculoskeletal findings were within normal limit.

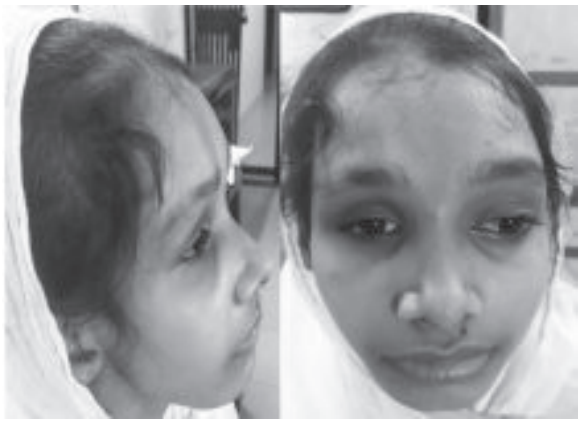

Fig.-1: *wasting of right side of the face*

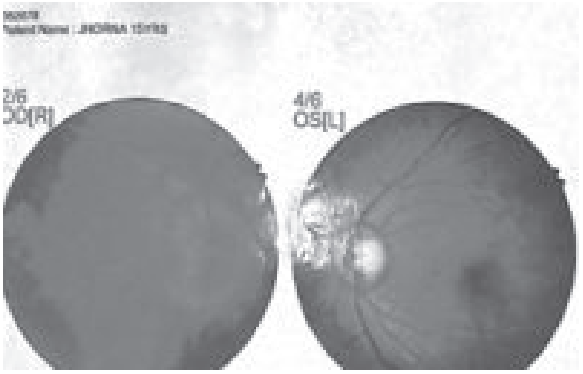

Fig.-2: *Right fundus shows disc swelling with hyperemia end engorged veins*

On laboratory investigations she had normal complete blood count with normal ESR. Perimetry reveals moderate loss of both central and peripheral vision of right eye. CT scan of brain, MRI of brain with MRV reveals only enopthalmus in right side. Findings of X-ray Chest P/A view, X ray skull, serum electrolytes including Calcium, serum TSH, ANA, P-ANCA, Anti-Scl 70, ACTH, Growth hormone was within normal limit. Patient was later properly counseled about the condition

and treated with prednisolone 1mg/day. Patient was reviewed 3 months later and her condition was static.

Discussion:

Parry-Romberg Syndrome (PRS) itself is a rare disease. The exact etiology is unknown. Generally, it is slow and progressive unilateral facial atrophy affecting the skin, subcutaneous tissue, muscles, and sometimes extending to the osteocartilaginous structure[17]. The condition is more often found in female population and has predilection for the left side of the face[16]. Though our patient is female, her affected side was right. Clinically, the skin can be dry, thin and with a dark pigmentation. PRS has been associated with various systemic manifestations, particularly neurologic, ophthalmologic and maxillofacial. Seizures are the most common neurologic manifestation. Other associations include migraines, hemiplegia, aneurysms, brain atrophy, limb atrophy, intracranial vascular malformations[7]. But no such clinical or radiological findings are available in our case. PRS has been associated with multiple ophthalmologic manifestations. The most common are enophthalmos due to loss of orbital fat which is present in our case. Other are uveitis, retinal vasculitis, ipsilateral and contralateral third nerve paresis, glaucoma, eyelid atrophy[7]. Rarely there may be papillitis and neuroretinitis[17].Which is present in our case. Some patients present a demarcation line between normal and abnormal skin, resembling a big linear scar, known as coup de sabre (French term which means “cut of the sword”)[19,20] . This finding was not noticed in this case reported. which were not found in our case. The intraoral soft tissue and muscles of mastication’s are usually normal, and there is no difficulty in jaw movement, speech or deglutition limitations[21]. The dental ûndings may include delayed ipsilateral tooth eruption, atrophic root development, missing teeth, retarded root formation, oligodontia, microdontia, dilacerations, unilateral crossbite, and pulp stones on the affected side[22]. No such findings were manifested in our patient on clinical observation.

Treatment of PRS aims first at halting the disease process and improving some symptoms of the disease. Immunosuppressive drugs might be considered in some patients especially in the case of cerebral involvement. Once the disease has stabilized, aesthetic treatments of the hemifacial atrophy consisting of augmentation of the atrophic region and restoration of the symmetry of the face can be initiated with a recommended pause of 1 to 2 years before proceeding with the reconstruction[7] .Ophthalmological treatment concentrates on stabilization or rehabilitation of (peri-) ocular complications. In one case with panuveitis and papillitis was treated with local and systemic steroids[23] . With treatment of systemic corticosteroid disease progression of this case seems to be halted.

Conclusion:

Though Parry-Romberg Syndrome isvery rare disease with no definitive treatment but usually it is self-stabilizing. Further work is required to establish its exact underlying pathophysiology. Suspected patient should be evaluated for other systemic manifestations, specially ophthalmological evaluation because glucocorticoid may have a role in halting the progression.

Conflict of interests:

The authors declare that they have no conflict of interest.

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