PARRY ROMBERG SYNDROME- A CASE REPORT

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HOW TO CITE THIS ARTICLE: Bhuyan D, Sarma U, Das B, et al. Parry Romberg syndrome- a case report. J. Evolution Med. Dent. Sci. 2019;8(20):1682-1685, DOI: 10.14260/jemds/2019/370

PRESENTATION OF CASE

A 35-year-old female presented to the plastic surgery department with progressive atrophy of the right side of the face with onset in childhood. She did not have any neurological complaints. The patient was 6 months pregnant at the time of examination. There was no other significant past history or any operative history. Physical examination revealed asymmetry of her face with the right side of her face being significantly smaller. Her right eye was mildly sunken. However, there was no visual impairment. No cranial neuropathies were present. She was referred for MRI brain and face.

Imaging Findings

MR imaging examinations revealed hemiatrophy most prominently involving right-sided skin and subcutaneous fat, with lesser involvement of the muscles of mastication. Skeletal findings were most pronounced in the mandible the right zygomatic arch and the right maxillary sinus (Fig. E, F, G, H, I and J). Enophthalmos of the right globe was present with relative paucity of retrobulbar fat (Fig. C, D). The right submandibular rand parotid glands are atrophied (Fig. G, H, I, J). Few discrete T2 and FLAIR hyperintensities were noted in the white matter of bilateral frontal lobes (Fig. K). No other intracranial imaging findings were observed at the time of presentation. The clinical appearance was compatible with Parry-Romberg syndrome (Fig. A, B). Further evaluation with CT scan was not pursued as the patient was 6 months gravid at the time of presentation to our department.

RADIOLOGICAL DIAGNOSIS

Parry-Romberg Syndrome.

DIFFERENTIAL DIAGNOSIS

The differential diagnosis of our case of progressive hemifacial atrophy includes other forms of juvenile localized scleroderma, Barraquer-Simons syndrome, congenital hemiatrophy, and primary hemifacial hypertrophy. Barraquer-Simons syndrome is an acquired partial progressive cephalothoracic lipodystrophy which presents with gradual onset of symmetrical bilateral subcutaneous fat loss from the face, neck, upper extremities, thorax, and abdomen but sparing the lower extremities. Central nervous system findings of deafness, epilepsy, and intellectual disability have also been described.[1] The bilateral nature of this disease and systemic involvement of the kidneys may differentiate these processes.

Hemifacial hypertrophy is a rare asymmetric enlargement of half of the head without enlargement of other body parts.[2,3] While there is unilateral face enlargement instead of atrophy as seen in Parry-Romberg Syndrome, it may be considered in the clinical differential diagnosis of an asymmetric unilateral facial deformity.

Other mimickers of Parry-Romberg Syndrome are fat necrosis, whether from infection such as bulbar poliomyelitis,[4] trauma, or connective tissue disease, and congenital deformities such as "wry neck."[5]

DISCUSSION

Parry-Romberg syndrome (PRS), also known as progressive facial hemiatrophy, was first described by Caleb Hillier Parry in 1825 and Moritz Heinrich Romberg in 1846. It is a rare disorder of unknown aetiology characterized by unilateral wasting of the skin and subcutaneous tissue of the face with variable involvement of underlying facial muscle, cartilage, and osseous structures. 15 percent of patients have neurologic manifestations that include epilepsy, migraine headache, cranial nerve deficits, hemiplegia, cognitive abnormalities, and fixed focal neurologic deficits.[6,7]

Parry Romberg syndrome is a sporadic and rare condition that has been reported to be more common in females,[7-11,12-14] without apparent geographic or ethnic predilection.[15] Onset typically occurs during the first and second decades of life, resulting in an initially insidious but progressive hemiatrophy of the face during a span of 2–20 years, with a slight propensity for the left side.[8-11,13,14] The progression abruptly arrests without cause and stabilizes, reaching a "burned-out" phase.[7,8,14,16-18,] This peculiar disease course, along with highly variable signs and symptoms, impedes consistent understanding of the underlying pathophysiology of PRS.

Many theories about PRS have emerged throughout the years, attributing this syndrome to widely varying aetiologies such as infection, trauma, sympathetic nervous system dysfunction, vascular abnormalities, inflammatory conditions, and autoimmune disorders, but at this time, a specific aetiology remains uncertain.[19,20–21]

'Financial or Other Competing Interest': None.
Submission 03-11-2019, Peer Review 01-05-2019,
Acceptance 09-05-2019, Published 20-05-2019.
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DOI: 10.14260/jemds/2019/370

Figure A, B: Clinical pictures showing normal (Figure A) and affected side (Figure B)

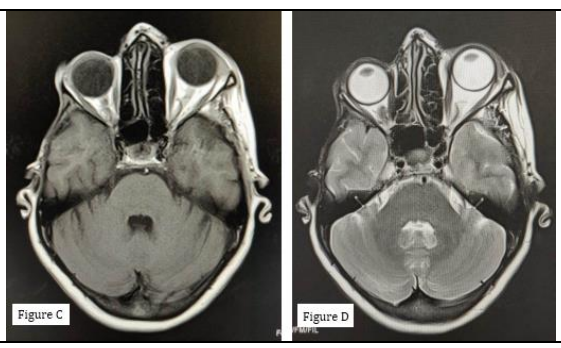

Figure C, D: Axial T1 (Figure C) and T2 (Figure D) weighted images showing gross reduction in subcutaneous fat in right hemifacial region including reduction in right retroorbital fat with resultant enophthalmos

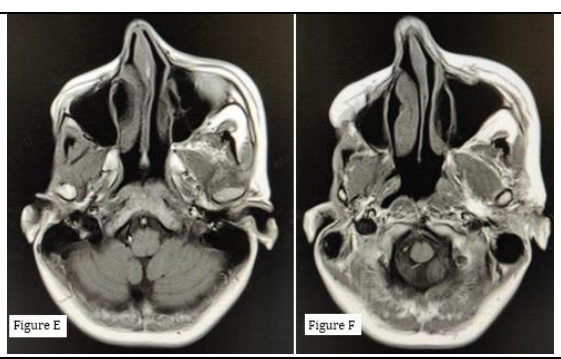

Figure E, F: Axial T1 (Figure E) and T2 (Figure F) weighted images showing gross reduction in subcutaneous fat in right hemifacial region with atrophy of right zygomatic arch

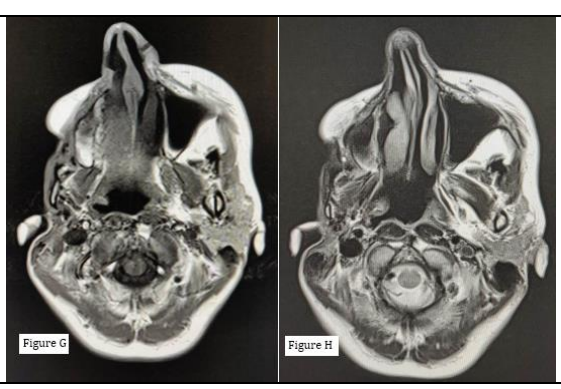

Figure G, H. Axial T1 (Figure G) and T2 (Figure H) weighted images showing atrophy of right parotid gland. The volume of right maxillary sinus is reduced

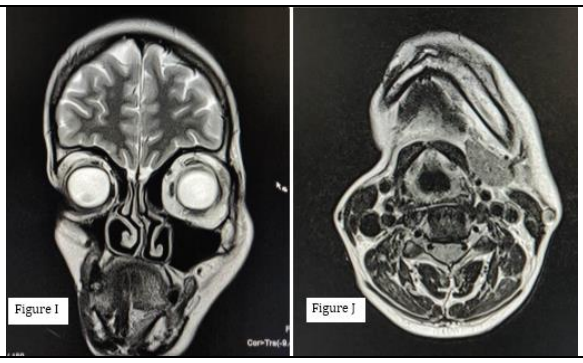

Figure G, H: Coronal (Figure G) and Axial T2 (Figure H) weighted images showing atrophy of right submandibular gland and right hemimandible

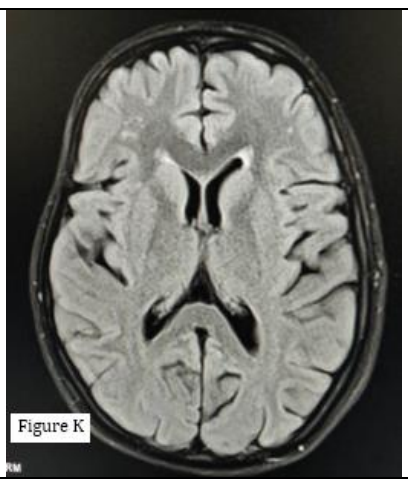

Figure J: Axial FLAIR image showing few discrete hyperintensities in white matter of bilateral frontal lobes.

It has been postulated that it actually falls within the spectrum of localized, linear scleroderma. Linear scleroderma, or "en coup de sabre," refers to focal scleroderma of the face or scalp, resulting in characteristic facies resembling a face struck on one side with a sabre sword. Parry-Romberg syndrome has similar characteristics, but there tends to be more extensive involvement of the ipsilateral subcutaneous tissue, calvarium, and orbit,[22] and facial hemiatrophy is typical rather than the hemifacial cutaneous sclerosis that typifies linear scleroderma.[23] In light of the similarity to linear scleroderma, some authors postulate chronic, progressive autoimmune neuro-vasculitis as the cause of Parry-Romberg syndrome.[22,23]

Patients characteristically experience atrophy of the skin and subcutaneous tissues and may develop atrophy of the underlying muscular, cartilaginous, osseous, and glandular structures as the disease progresses.[12,13] It typically begins in the maxillary or periorbital region and may expand to involve the forehead, perioral region, teeth, jaw, and neck to varying degrees. Involvement of the teeth may help in determining the age of onset in unclear cases, because the presence of relatively smaller teeth with short roots has been reported in PRS.[24] Neurologic symptoms occur in 15%–20% of patients, with the most common being ipsilateral headaches, facial pain, and seizures, which may be refractory to treatment.[5,6,8,9] Ophthalmologic symptoms occur in 10%–35% of patients and usually involve the ipsilateral orbit.

Atrophy of the retrobulbar fat leading to enophthalmos is common,[7,11,12] other potential orbital abnormalities include uveitis and retinal or optic nerve alterations.[7,19] Diagnosis of PRS mainly relies on the clinical history and examination and exclusion of other possibilities, supported by histopathologic and imaging studies.[21]

In summary, PRS is a rare, self-limiting, and slowly progressive hemiatrophy of the face that typically affects the skin and subcutaneous tissues and may affect deeper tissues such as the musculature, cartilage, and osseous structures. Neurologic and ophthalmologic symptoms are common, but underlying pathophysiology remains uncertain. Intracranial involvement is best evaluated with MR imaging and can range widely, with the most common findings being parenchymal calcifications, white matter abnormalities, and brain atrophy. In addition to evaluating the extent of disease, radiologic assessments may also facilitate the exclusion of other differential considerations, help monitor disease progression, and evaluate post-treatment responses.

DISCUSSION OF MANAGEMENT

The primary goal is to stop the active disease process. Methotrexate (MTX) is the standard therapy for active disease. The MTX is often combined with oral prednisone over the first three months due to the fact that the methotrexate has a delayed effect on inflammation and fibrosis. A long course of therapy is typically required as relapse is frequently seen with shorter courses of therapy. The specific length of therapy required to reduce relapse is unknown, and likely varies from patient to patient. Current evidence supports a 12-24-month course of methotrexate being most effective in inducing prolonged remission.[25-30]

Isolated case reports of other immunosuppressive agents such as mycophenolate mofetil, cyclosporine, and cyclophosphamide have shown variable success in patients who have failed treatment with MTX.[29,31,32] PUVA has been reported to arrest disease activity in isolated reports of PHA.[33,34]

Surgical treatment for PHA often requires a multi-specialty approach with repeated procedures, depending on degree of involvement. The therapeutic goal of surgery for PHA patients is to minimize psychosocial effects, and to correct the appearance and function of involved facial structures.[35,36]

REFERENCES

[1] Simsek-Kiper PO, Roach E, Utine GE, et al. Barraquer-simons syndrome: a rare clinical entity. Am J Med Genet A 2014;164(7):1756-60.

[2] Bergman JA. Primary hemifacial hypertrophy. Review and report of a case. Arch Otolaryngol 1973;97(6):490-4.

[3] Gorlin RJ, Meskin LH. Congenital hemihypertrophy. Review of the literature and report of a case with special emphasis on oral manifestations. J Pediatr 1962;61:870-9.

[4] Antoniades K, Giannouli T, Vahtsevanos K. Hemifacial atrophy secondary to poliomyelitis. Int J Oral Maxillofac Surg 1997;26(3):215-6.

[5] Tolkachjov SN, Patel NG, Tollefson MM. Progressive hemifacial atrophy: a review. Orphanet J Rare Dis 2015;10:39.

[6] Buonaccorsi S, Leonardi A, Covelli E, et al. Parry-Romberg syndrome. J Craniofac Surg 2005;16(6):1132-5.

[7] Aynaci FM, Şen Y, Erdöl H, et al. Parry-Romberg syndrome associated with Adie's pupil and radiologic findings. Pediatr Neurol 2001;25(5):416-8.

[8] Pichiecchio A, Uggetti C, Egitto MG, et al. Parry–Romberg syndrome with migraine and intracranial aneurysm. Neurology 2002;59(4):606-8.

[9] Shah JR, Juhasz C, Kupsky WJ, et al. Rasmussen encephalitis associated with Parry–Romberg syndrome. Neurology 2003;61(3):395-7.

[10] Stone J. Parry–Romberg syndrome: a global survey of 205 patients using the Internet. Neurology 2003;61(5):674-6.

[11] Moko SB, Mistry Y, de Chalain TM. Parry–Romberg syndrome: intracranial MRI appearances. J Craniomaxillofac Surg 2003;31(5):321-4.

[12] El-Kehdy J, Abbas O, Rubeiz N. A review of Parry-Romberg syndrome. J Am Acad Dermatol 2012;67(4):769-84.

[13] Dalla Costa G, Colombo B, Dalla Libera D, et al. Parry Romberg syndrome associated with chronic facial pain. J Clin Neurosci 2013;20(9):1320-2.

[14] Kister I, Inglese M, Laxer RM, et al. Neurologic manifestations of localized scleroderma: a case report and literature review. Neurology 2008;71(19):1538-45.

[15] Sharma M, Bharatha A, Antonyshyn OM, et al. Case 178: Parry-Romberg syndrome. Radiology 2012;262(2):721-5.

[16] Paprocka J, Jamroz E, Adamek D, et al. Difficulties in differentiation of Parry-Romberg syndrome, unilateral facial sclerodermia, and Rasmussen syndrome. Child's Nerv Syst 2006;22(4):409-15.

[17] Okumura A, Ikuta T, Tsuji T, et al. Parry-Romberg syndrome with a clinically silent white matter lesion. Am J Neuroradiol 2006;27(8):1729-31.

[18] Tollefson MM, Witman PM. En coup de sabre morphea and Parry-Romberg syndrome: a retrospective review of 54 patients. J Am Acad Dermatol 2007;56(2):257-63.

[19] Sommer A, Gambichler T, Bacharach-Buhles M, et al. Clinical and serological characteristics of progressive facial hemiatrophy: a case series of 12 patients. J Am Acad Dermatol 2006;54(2):227-33.

[20] Longo D, Paonessa A, Specchio N, et al. Parry-Romberg syndrome and rasmussen encephalitis: possible association. Clinical and neuroimaging features. J Neuroimaging 2011;21(2):188-93.

[21] Madasamy R, Jayanandan M, Adhavan UR, et al. Parry Romberg syndrome: a case report and discussion. J Oral Maxillofac Pathol 2012;16(3):406-410.

[22] Grosso S, Fioravanti A, Biasi G, et al. Linear scleroderma associated with progressive brain atrophy. Brain Dev 2003;25(1):57-61.

[23] Orozco-Covarrubias L, Guzman-Meza A, Ridaura-Sanz C, et al. Scleroderma 'en coup de sabre' and progressive facial hemiatrophy. Is it possible to differentiate them? J Europ Acad Dermatol Venereol 2002;16(4):361-6.

[24] Khan M, Khan M, Negi R, et al. Parry Romberg syndrome with localized scleroderma: a case report. J Clin Exp Dent 2014;6(3):e313-6.
[25] Tollefson MM, Witman PM. En coup de sabre morphea and Parry-Romberg syndrome: a retrospective review of 54 patients. J Am Acad Dermatol 2007;56(2):257-63.
[26] Zulian F, Athreya BH, Laxer R, et al. Juvenile localized scleroderma: clinical and epidemiological features in 750 children. An international study. Rheumatology 2005;45(5):614-20.
[27] Sartori S, Martini G, Calderone M, et al. Severe epilepsy preceding by four months the onset of scleroderma en coup de sabre. Clin Exp Rheumatol 2009;27(3 Suppl 54):64-7.
[28] Kreuter A, Gambichler T, Breuckmann F, et al. Pulsed high-dose corticosteroids combined with low-dose methotrexate in severe localized scleroderma. Arch Dermatol 2005;141(7):847-52.
[29] Korkmaz C, Adapinar B, Uysal S. Beneficial effect of immunosuppressive drugs on Parry-Romberg syndrome: a case report and review of the literature. South Med J 2005;98(9):940-3.
[30] Hashkes PJ, Becker ML, Cabral DA, et al. Methotrexate: new uses for an old drug. J Pediatr 2014;164(2):231-6.
[31] Martini G, Ramanan AV, Falcini F, et al. Successful treatment of severe or methotrexate-resistant juvenile localized scleroderma with mycophenolate mofetil. Rheumatology 2009;48(11):1410-3.
[32] Bergler-Czop B, Lis-Święty A, Brzezińska-Wcisło L. Scleroderma linearis: hemiatrophia faciei progressiva (Parry-Romberg syndrom) without any changes in CNS and linear scleroderma "en coup de sabre" with CNS tumor. BMC Neurol 2009;9:39.
[33] Baskan EB, Kaçar SD, Turan A, et al. Parry–Romberg syndrome associated with borreliosis: could photochemotherapy halt the progression of the disease? Photodermatol Photoimmunol Photomed 2006;22(5):259-61.
[34] Gambichler T, Kreuter A, Rotterdam S, et al. Linear scleroderma 'en coup de sabre' treated with topical calcipotriol and cream psoralen plus ultraviolet A. J Eur Acad Dermatol Venereol 2003;17(5):601-2.
[35] You KH, Baik HS. Orthopedic and orthodontic treatment of Parry-Romberg syndrome. J Craniofac Surg 2011;22(3):970-3.
[36] De Vasconcelos Carvalho M, do Nascimento GJ, Andrade E, et al. Association of aesthetic and orthodontic treatment in Parry-Romberg syndrome. J Craniofac Surg 2010;21(2):436-9.