PARRY ROMBERG SYNDROME- A CASE REPORT

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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 right maxillary sinus (Fig. C, D). The right submandibular and parotid glands are atrophied (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 and 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 sclerodermia, Barraquer-Simons syndrome, congenital hemiatrophy, and primary hemifacial hypertrophy. Barraquer-Simons syndrome is an acquired partial progressive cephaloarthropathy 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.5,7 Parry-Romberg syndrome is a sporadic and rare condition that has been reported to be more common in females.6,7,11,12,14 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.6,11,12,14 The progression abruptly arrests without cause and stabilizes, reaching a “burned-out” phase.7,8,13,16-18 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
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: 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, and facial hemiatrophy is typical rather than the hemifacial cutaneous sclerosis that typifies linear scleroderma. 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 peri-orbital 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


