Case Report and Literature Review on Parry-Romberg Syndrome Presenting Later in Life

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Introduction

Parry-Romberg syndrome (PRS), or progressive hemifacial atrophy (PHA), is a rare disorder that is characterized by slowly progressive degeneration of skin, subcutaneous tissue, muscle, cartilage and bony structures. It is typically localized to one-half of the face and follows the distribution of one or more branches of the trigeminal nerve.[1] PRS typically presents in the first two decades of life with an average onset of 13.6 years of age.[2] The onset is subtle, affecting mostly women before the age of 20 years. Patients with early onset tend to be more severely affected.[2]

Although the exact etiology of PRS is unclear, some hypotheses focus on an autoimmune origin, previous trauma, disturbance of fat metabolism, heredity and autonomic dysregulation. Treatment of PRS is challenging and requires a multidisciplinary approach with the main goals being to stop the progressive loss of tissues, improve functions of the affected structures, correct facial asymmetry, and thus, decrease the emotional burden of PRS.

Case

A 74 year old man presented to our dermatology department with an atrophic subcutaneous plaque of the left central malar cheek. A 3 mm punch biopsy of the affected region showed slightly thickened collagen bundles with loss of periadnexal adipocytes within the deep dermis. An EVG stain revealed a normal number and distribution of elastic fibers. A small erosion was noted superficially. These microscopic findings are suggestive but not diagnostic of a fibrosing process such as morphea.

The patient was referred for an MRI of the brain and orbits. A non-contrast MRI of the brain demonstrated a 2mm focus of non-specific T2 prolongation in the right cerebellum with mild to moderate involutional changes. There was no mass effect or midline shift. However, moderate right and small left-sided mastoid air cell effusions were observed. A couple sequences in the left lateral face reported a region of indentation. A relative decrease in subcutaneous fat was also noted in this region when compared to the right side. A non-contrast MRI of the orbits showed no evident abnormality associated the orbital appearance or extra-ocular muscles. No optic nerve involvement was noted.

Serum anti-topoisomerase 1, or SCL-70 antibodies tested positive at 6.7 U (positive: > or = 1.0 U), thus confirming the presence of Scleroderma in our patient. The patient was referred to a rheumatologist for a thorough work –up to rule out systemic issues. Upon his return, we plan to treat our patient with synthetic fillers and possibly fat grafting in an attempt to reconstruct the atrophied region on the left half of the face.

Discussion

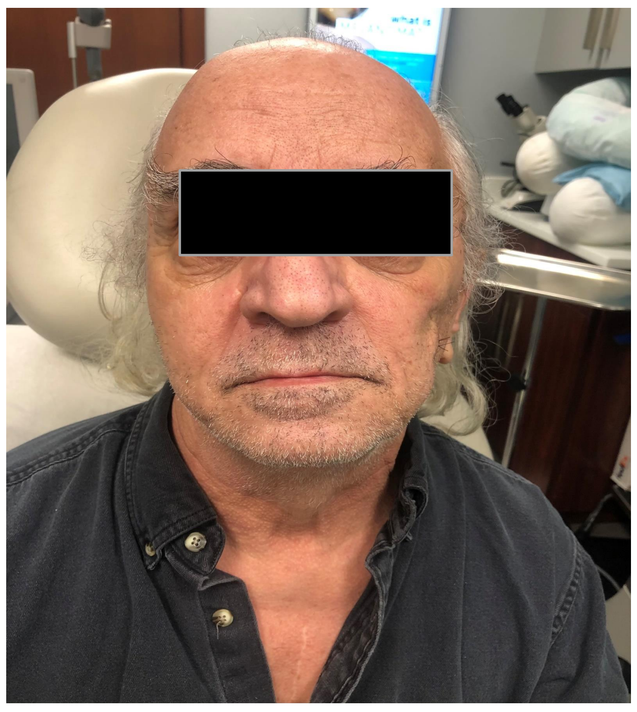
PRS has a slow progression over a period of two to twenty years that ceases without apparent cause after a highly variable period.[3] Our case is unique as it describes PRS presenting later in life in a 74-year old male patient with rapid progression.

PRS frequently overlaps and coexists with localized linear scleroderma en coup de sabre. It may be difficult to differentiate between the two conditions on initial evaluation, because both conditions have a similar age of onset and progressive course.[4] Generally, a diagnosis of PRS should be made in cases of widespread unilateral soft tissue atrophy with thin or soft overlying skin that is not preceded by inflammation or induration.[5] Localized linear scleroderma en coupe de sabre presents with cutaneous sclerosis localized to the frontoparietal region involving the forehead, which was not seen in our patient.[5]

Only a subset of patients with PRS will develop secondary neurologic or ophthalmologic symptoms, and the prognosis is highly variable.[3] Neurologic symptoms occur in 15%–20% of patients, with the most common being ipsilateral headaches, facial pain, and seizures.[6] Neurological disorders are more frequent in patients with upper face and scalp involvement.[6] The involvement of left lower face in our patient with the sparing of scalp and forehead reduced the likelihood of neurological or ocular manifestations were noted in our patient.

Corticosteroids and immunosuppressive drugs including azathioprine, hydroxychloroquine, methotrexate and cyclophosphamide may be considered in the active phase of disease.[7] Once the disease stabilizes, esthetic therapy consisting of augmentation of the atrophic region with autologous or synthetic fillers can be initiated. Surgical options including fat grafting, orthognathic surgery, and free tissue transfer are reserved for more severe and refractory cases.[7]

Results



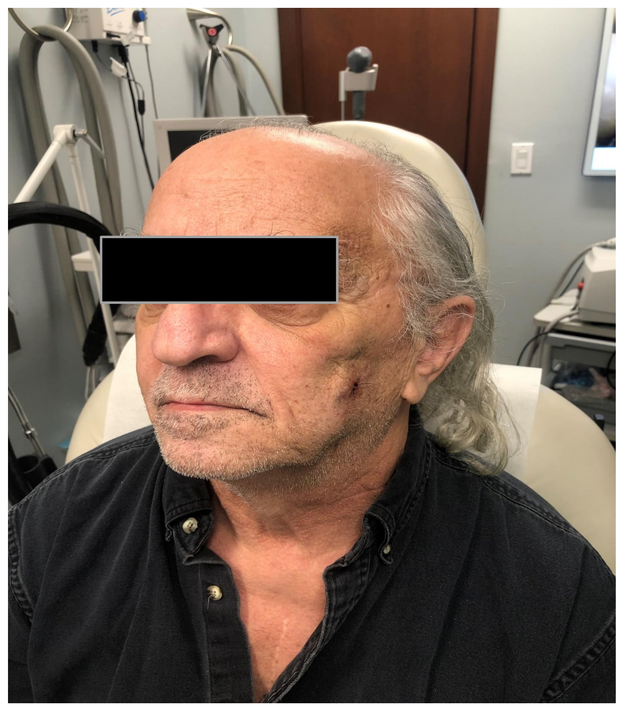


Figure 1 & 2 : Clinical photos demonstrating atrophic at left lower cheek

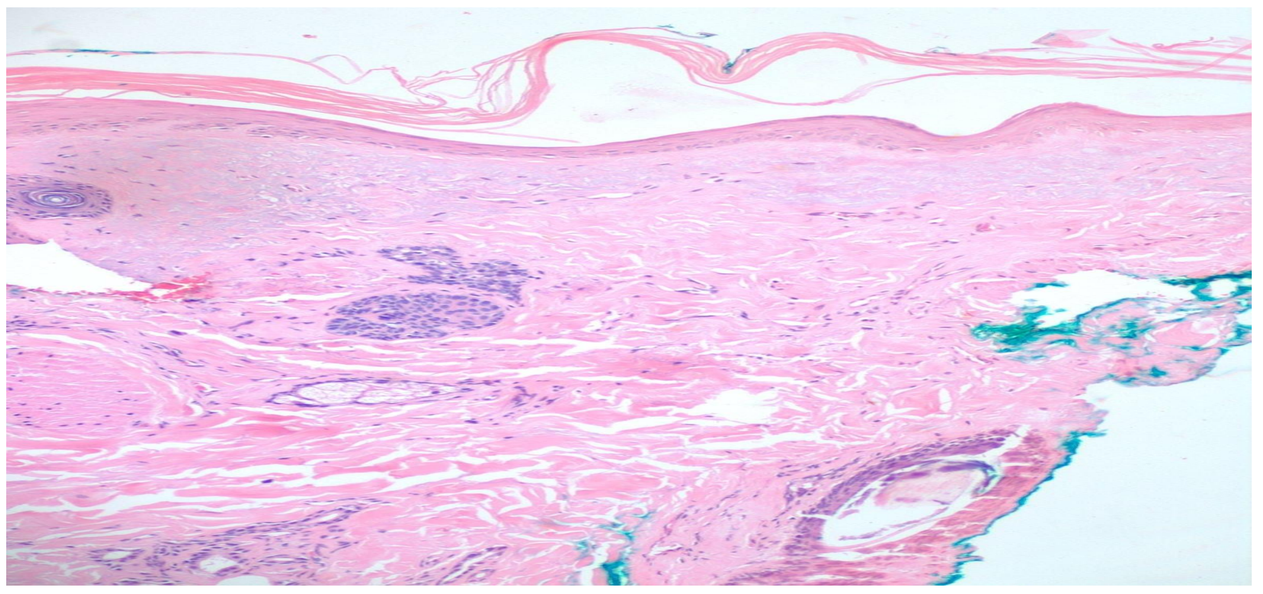


Figure 3 : Hematoxalin and eosin histology showing atrophy of the epidermis and thickening of the collagen

Conclusion

PRS frequently overlaps with localized scleroderma en coupe de sabre on initial diagnosis, therefore careful history and physical exam should be performed by dermatologists on patients presenting with this condition. It is crucial to be able to distinguish PRS from localized scleroderma before starting treatment as neurologic and opthalmologic complications may be found in a subset of patients with PRS. Many treatment options exist for PRS, however, it is generally advised to start off with medications to treat the active phase of the disease, including neurologic and/or opthalmologic symptoms. Once the condition has stabilized over the course of years, switching to esthetic and surgical management is recommended. This case report documents the classical features of this rare condition, contributing towards the need for a better understanding of the complications involved with Parry Romberg Syndrome.

References

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Disclosures: None