Case report

Parry-Romberg Syndrome: Case Report of a Rare Type of Linear Scleroderma.

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Abstract

Parry–Romberg syndrome (PRS), which is also known as Progressive hemifacial atrophy, is a form of linear scleroderma of the face and head. The hallmark of this presentation is varying degrees of atrophy at one side of face, affecting the subcutaneous fat, muscle, and underlying bone structures, with mild or absent epidermal and dermal changes. Here we report a 10-year-old girl presented with right hemifacial atrophy and convulsion. Considering the clinical features and laboratory workup diagnosis was consistent with Parry–Romberg syndrome (PRS).

Keywords: Parry-Romberg syndrome, Hemifacial atrophy.

Received on: 12.05.2024; Accepted on 30.05.2024

Introduction:

Linear scleroderma is the most common subtype of localized scleroderma in children and adolescents, and is characterized by one or more longitudinal, band-like lesions that typically involve upper or lower extremities and the face.[1] Parry–Romberg syndrome (PRS), which is also known as Progressive hemifacial atrophy is a form of linear scleroderma of the face and head. The hallmark of this presentation is varying degrees of atrophy at one side of face, affecting the subcutaneous fat, muscle, and underlying bone structures, with mild or absent epidermal and dermal changes.[2] PRS may occur with or without En coup de sabre (ECDS) where linear induration of the skin that affects the face and/or the scalp.[3] Extensive cases usually cause marked hemifacial atrophy resulting in severe, permanent facial asymmetry and disfigurement.

The etiology and pathogenesis of PRS are still not well understood. a combination of genetic predisposition, autoimmunity, and environmental factors are thought to trigger local inflammation that leads to persistent increased collagen synthesis and deposition in the skin.[4] Neurological, oral, and ocular problems more commonly associated with Parry–Romberg syndrome (PRS).[5,6] There is no diagnostic or characteristic laboratory findings of PRS, so diagnosis is essentially based on clinical findings. Histological documentation is helpful but not essential in confirming the diagnosis.[7] Single Hub and Access point for pediatric rheumatology in Europe (SHARE), recommended all patients, with or without signs of neurological involvement, have an MRI of the head at the time of the diagnosis.[8] Also they recommended orthodontic and maxillofacial evaluation, ophthalmological and musculoskeletal assessment at diagnosis and during follow-up. Management depends on the stage of the disease and extent, presence and severity of ECDS manifestations and potential for functional or cosmetic sequelae. We are reporting here, a case of PRS in a 10-year old girl, for proper understanding of this rare disease related to its presentations, diagnosis and treatment issues.

Case report

An 10-year-old girl presented with gradual onset progressive unilateral facial atrophy and facial disfigurement for the last 7 years. Mother informed that until the age of 3 years, her daughter's face was quite normal after that, a hyperpigmented area over right side of forehead was observed. This lesion gradually increased in size and extended in linear pattern from forehead to scalp followed by progressive tightening of skin and atrophy of right side of face but left side was quite normal. She had history of headache and convulsion. There was no history of joint pain, facial pain, visual problem, head trauma, delayed tooth eruption. Clinical examination revealed a healthy girl with normal vital signs and systemic presentation. She was alert with no signs of mental or psychological instability. Speech, hearing and vision were normal. The face was asymmetric as there was wasting of the entire right half of the face. A central ridge on the forehead was

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present, with right side underdeveloped compared with left side. Hyperpigmented, indurated lesion was present over right forehead and extended in linear pattern from forehead to scalp. Linear band across the forehead, with a waxy infiltration toward the eyelid causing dermal and subcutaneous atrophy, eyebrow, and eyelash losses. The right eye was at a upper level compared to left. The right cheek, right side of nose, zygomatic areas, right side of upper lip and jaw were atrophic compared with left side. Nose and chin were deviated to the right side. Ears were symmetric and normal. Intraoral examination revealed normal dentition and tongue.

Routine laboratory investigations including CBC and other biochemical profile were normal except elevated ESR, CRP. Patient was negative for antinuclear antibodies and ENA (Extracted nuclear antigen) profiles. X-ray PNS revealed atrophy of the right maxillary bone. MRI of the brain revealed focal thinning of frontal bone and lacunar infract at right paraventicular region. Based on the clinical and radiographic findings, the diagnosis of PRS was made. After diagnosis, facial deformity and convulsion became the major concern for her treatment, so the patient started treatment with methotrexate (15mg/m^2/week SC) and oral prednisone (1 mg/kg/day), anticonvulsant (Phenobarbitone: bolus dose followed by maintenance dose). She was on regular follow-up in our clinic and remain stable.

Figure 1: Clinical photograph of the patient showing hemiatrophic change on the right side of face

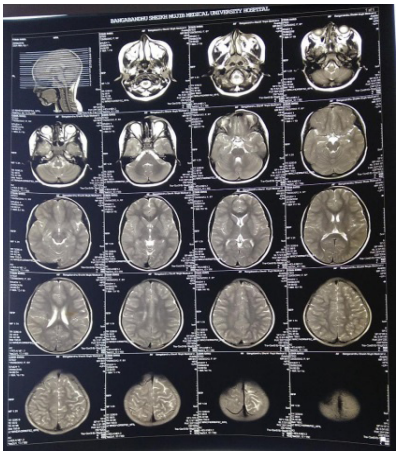

Figure 2: MRI of brain showing focal thinning of frontal bone & scalp soft tissue with lacunar infract at right paraventicular region

Discussion:

Parry Romberg syndrome is a rare disorder of unknown etiology. First described by Parry in 1825 and Romberg in 1846, the term progressive facial hemiatrophy (PFH) was coined by Eulenberg in 1871.[9] The syndrome is characterized by slowly progressive atrophy involving one side of the face. PRS is a sporadic illness that has been shown to be more prevalent in females.[10] Our index case was a girl and presented at 10 year of age. The condition is observed on the left side of the face about as often as on the right side.[11] Trauma, viral infection, endocrine disturbance, autoimmunity and hereditary are also believed to be associated to the pathogenesis of the disease.[12] The onset of this syndrome often occurs during the first and second decades of life, after which the atrophy slowly progresses over several years, eventually becoming stable.[13] Beginning in the maxillary or periorbital region, the disease can spread to the forehead, perioral region, teeth, jaw, and neck.[11] The condition has overlapping features with 'en coup de sabre', a type of linear scleroderma affecting the head. However, recent studies suggest that PRS is a clinical subtype of linear scleroderma.[14] In our case (Fig1) atrophied right side of upper lip, nose, eye lid along with right side of fore head and scalp is seen.

Neurological, oral, and ocular problems more commonly associated withPRS.[5,6] In oral involvement, potentially affecting all structures, including lips, gingiva, and bony structures, resulting in various degrees of atrophy and misalignment.[15] In Our patient right side of upper lip was atrophied. Neurological involvement has been reported in 25% to 50% of patient with linear scleroderma .[16] The most frequent neurological conditions are seizures and headaches.[17] Our patient also presented with convulsion

and headache in this report. Other Neurological involvement includes cranial nerve palsies, trigeminal and peripheral neuropathy, autoimmune encephalitis , neuropsychiatric problems, movement disorders, slurred speech, cognitive problems, and central nervous system vasculitis.[18,19] The most common ocular complications are fibrotic changes in eyelids, eyelashes (often lost where lesions cross the eye), uveitis, and episcleritis.[20] In our patient initial ophthalmological evaluations found only fibrotic changes in eyelids. Hemiatrophy of the tongue may occur in half of the children with PRS.[21] Dental issues include ectopic or delayed tooth eruption, tooth crowding, root deficiencies or resorption, and gingival and periodontal ligament defects.[22] As SHARE have recommended Linear scleroderma patients involving face and head, with or without signs of neurological involvement, to do a MRI of the brain at the time of the diagnosis. In this index case , MRI finding was focal thinning of frontal bone & scalp soft tissue with lacunar infract at right paraventicular region. The most common MRI abnormalities in the brain are T2 hyperintense white matter lesions that can be ipsilateral or contralateral to the skin lesion; they are most commonly subcortical but also found in the periventricular and central white matter.[23]

Current pharmacological treatments are focused on controlling the active, inflammatory phase. Because controlling inflammation reduces the risk for damage progression such as dyspigmentation and tissue atrophy, as well as more serious morbidity of arthropathy, growth disorders (limb and facial hemiatrophy), ocular issues, neurological symptoms and other EC morbidity.[24,25]Standardized treatment regimens for Linear scleroderma developed by SHARE and CARRA. The CARRA recommended MTX dose is 1 mg/kg/week (maximum 25 mg) administered subcutaneously. The MTX-based standardized regimens are (1) MTX alone, (2) MTX plus intravenous CS (methylprednisolone), and (3) MTX plus oral CS (prednisone, prednisolone).[26] The SHARE recommended MTX dosing of 15 mg/m^2/week (maximum of 25 mg MTX/dose) and a minimum of 3 months of CS treatment.[8]

Methotrexate (MTX) in combination with oral corticosteroids along with supportive therapy were administered in our case . Different surgical strategies have been used to correct atrophy and other deformities associated with PRS.[27] Surgery should ideally only be considered when the disease is inactive and possibly when growth is complete.

Recovery period for overall prognosis of Parry-Romberg syndrome is unpredictable. In some cases, the atrophy ends before the entire face is affected. In mild cases, the disorder usually causes no disability other than cosmetic effects. Usually atrophy slowly progresses over several years, eventually becoming stable.

Conclusion

Parry–Romberg syndrome or progressive hemifacial atrophy is very rare, uncommon, degenerative, condition. Early diagnosis and effective treatment of these conditions is essential to reduce the risk for functional impairment and other damage. More study is needed to advance our understanding and to identify effective therapies to achieve sustained remission so that we can improve the long-term outcome for these children.

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