Linear scleroderma en coup de sabre – a different clinical presentation

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ABSTRACT

Localized scleroderma (LoS) is a rare condition featuring skin and underlying tissue sclerosis not usually compromising other systems. A subtype of LoS including lesions in the head is further classified as linear scleroderma *en coup de sabre* (LSeCS). Neurological involvement in LSeCS can reach up to 4% and may include seizures. Cutaneous lesions usually emerge before neurologic symptoms and these oftentimes manifest with intracranial abnormalities.

We describe a case of an 11-year-old boy with an onset of self-limited unexplained seizures at 20-months of life. During the first year of follow-up, a midline frontoparietal lesion with alopecia and hypopigmentation was noted and a referral to dermatology and pediatric rheumatology consultation was made. A diagnosis of LEsCS was made. A 10-year follow-up of this patient is presented with favorable outcome.

LSeCS is a rare form of LoS most frequently diagnosed in children and adolescents. A meticulous examination of these patients should be performed with particular attention to the face and scalp. The mainstay therapeutical approach is based on methotrexate and corticosteroids. Neurologic abnormalities associated with skin lesions on the head should should raise clinical suspicion of LSeCS.

Keywords: Child; Neurological symptoms; Localized scleroderma.

INTRODUCTION

Localized scleroderma (LoS) is a rare chronic condition of unknown origin seen in both adults and children, with an incidence of 0.4 to 2.7 per 100.000 people[1]. Features include a typical skin and underlying tissue sclerosis and classically do not compromise extra-cutaneous systems[2]. Linear scleroderma (LS) is the predominant subtype of LoS in children and adolescents[1]. When characteristic skin lesions in the head resemble the stroke of a saber the condition is further classified as linear scleroderma *en coup de sabre* (LSeCS)[1,3]. About 90% of children that are diagnosed with LSeCS range from 2 to 14 years of age[1]. They usually develop band-like sclerotic lesions with atrophy, furrow of the skin and cranial atrophy[2], which manifest as red to violaceous patches on the skin of the forehead and evolve into ivory-colored fibrotic, hyperpigmented and hairless plaques[1].This condition usually doesn t affect other organ but is has been reported that it can affect the central nervous system as well as other body systems. Neurological involvement in LSeCS can reach up to 4%[1] and clinical presentation include seizures, headache, focal neurological deficits and neuropsychiatric abnormalities[2]. When neurologic symptoms are found they are oftentimes associated with intracranial abnormalities[4], and usually show signs of focal brain atrophy, calcifications and T2-hyperintense white-matter lesions in magnetic resonance imaging (MRI)[4]. Evidence suggests neurologic involvement occurs after initial cutaneous presentation, but a few reports stated otherwise[3]. We describe a case of an 11-year-old boy whose diagnosis of LSeCS was based on recurrent seizures followed by a characteristic skin lesion in the forehead. A 10-year follow-up of this patient is presented with a favorable outcome.

CASE-REPORT

We present a case of a 11-year-old boy, that was referred to the outpatient clinic at 20 months of age due to 2 episodes of seizures within that same year with no previous medical or family history of seizures and with normal development and growth pattern. The first

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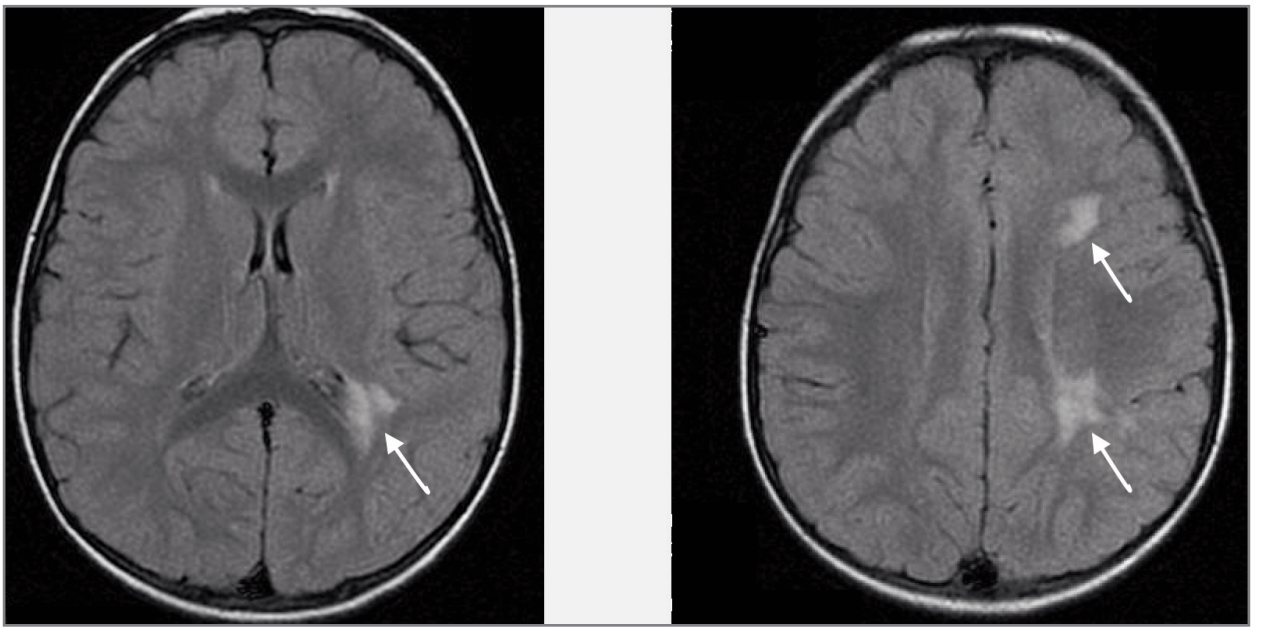

FIGURE 1. T2 Flair MRI images at diagnosis showing subcortical periventricular and frontal-parietal hyperintense lesions in the left lobe.

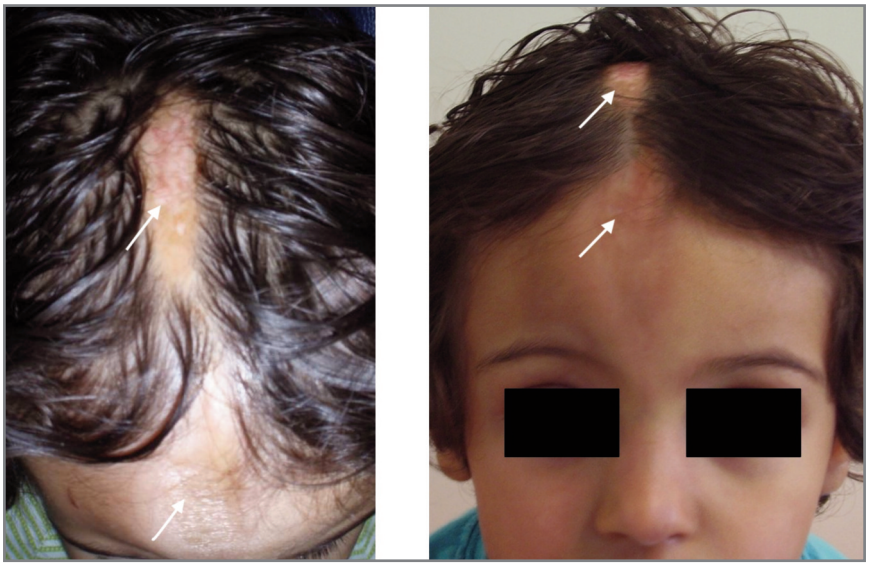

FIGURE 2. Midline frontoparietal sclerotic lesion with alopecia and hypopigmentation (see white arrows) at 1-year of follow-up.

seizure was depicted as a vague, non-responsive fixed stare, with loss of axial tone, with improvement over 5 minutes. The second episode consisted of about one-minute partial focal seizures of the upper right limb, with altered consciousness with eye revolution, sialorrhea and associated febrile state. Within the 1st year of follow-up another seizure occurred with similar characteristics as the previous one. Patient started follow-up with a neurologist and though no abnormalities were found in electroencephalogram (EEG), a cranial-MRI showed subcortical periventricular and frontal-parietal hyperintensities in the left lobe (Figure 1). TORCH perinatal infections, such as toxoplasmosis, other agents like syphilis, rubella, cytomegalovirus or herpes simplex were discarded. Since the child showed no development impairment and continued to thrive

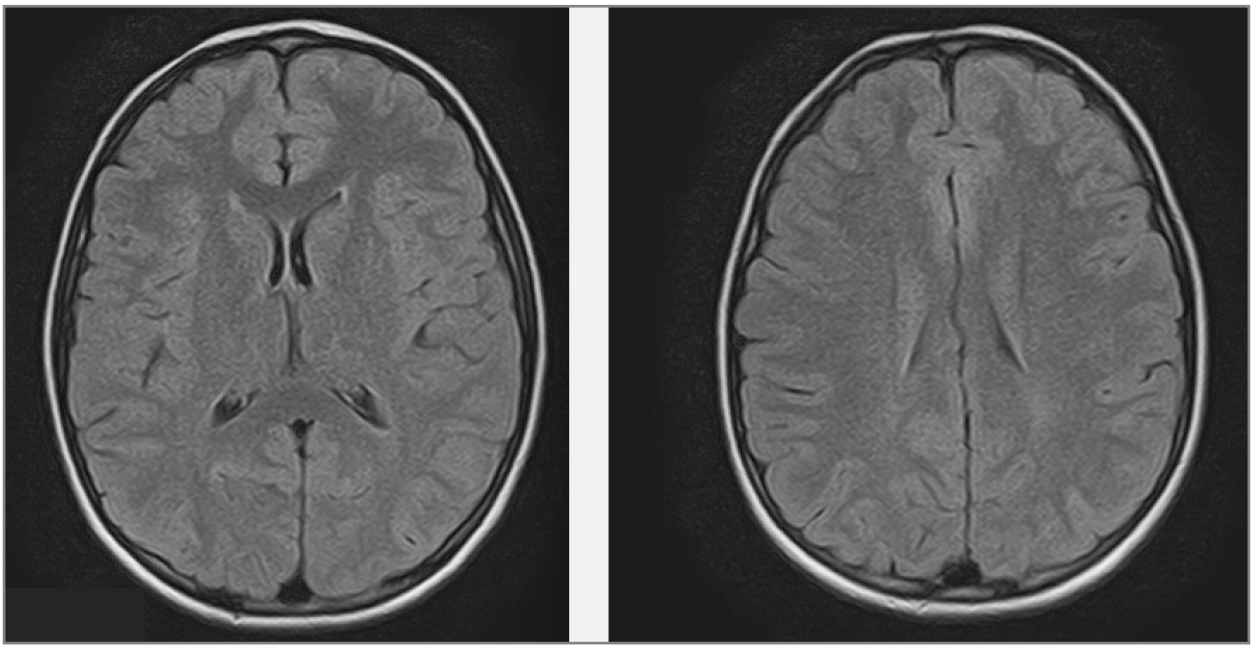

FIGURE 3. T2 Flair MRI images showing no lesions at 10-years of follow-up

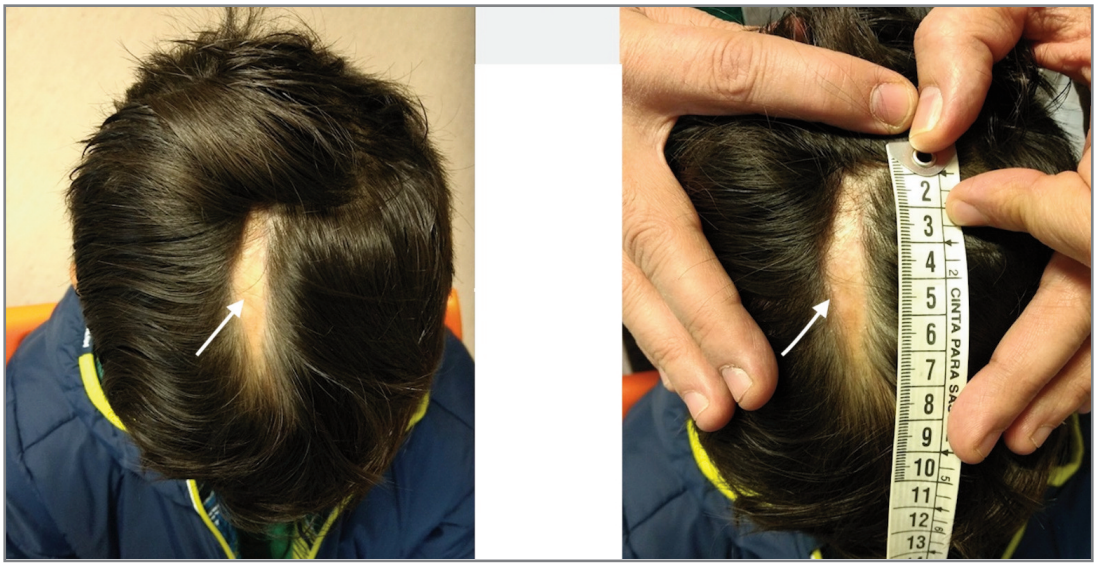

FIGURE 4. Favourable receding skin lesions surrounded by normal hair growth at 10-year follow-up.

well, a wait-and-see approach was made. At 1-year of follow-up with no more seizures, a midline frontoparietal lesion with alopecia and hypopigmentation (Figure 2) was noted and he was referred to dermatology and pediatric rheumatology consultation. A skin biopsy was performed, suggesting scleroderma and a diagnosis of LSeCS was made. Bloodwork including anti-nuclear antibodies was normal. Initial treatment regimen included oral methotrexate (MTX), oral prednisolone, supplementation with folic acid and oral and colecalciferol, skin hydration with vitamin-silicone-based lotion and facial rehabilitation. Despite initial management at year 3 of follow--up, skin lesions progressed and worsened. An inflamed frontal scalp lesion and retraction of the skin from the right supraciliary area until the upper part of the lip, together with asym-

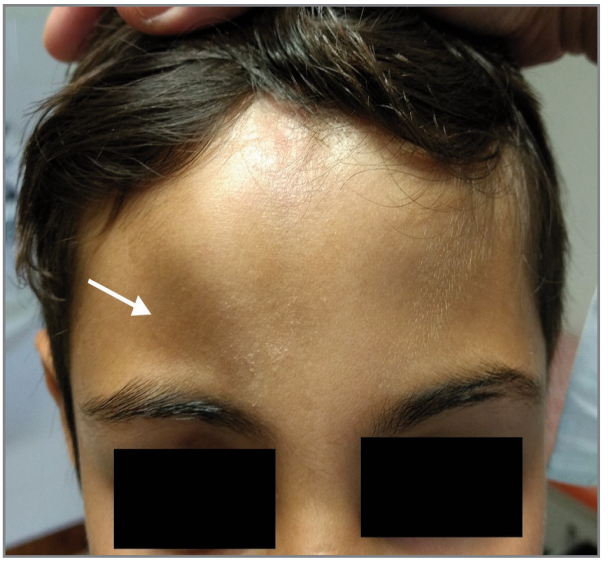

FIGURE 5. Dermatological changes of scleroderma en coup de sabre showing a hypopigmented midline frontoparietal scalp lesion with alopecia and normal surrounding hair growth. Arrow shows a groove at the frontal bone on the upper right supraciliary area

metry of the mouth was noted. Treatment regimen was switched to subcutaneous MTX (15 mg/m^2) and intravenous monthly pulses of methylprednisolone (20mg/Kg) for 9 months, with good clinical response. At year-4 of follow-up lesions had evolved favorably with less inflammation and new hair growth at the frontal scalp, together with less hypopigmentation of the remaining facial lesions. Right supraciliary area and right hemifacial atrophy remained but were less evident. Treatment was then tapered to oral prednisolone (0.5mg/Kg/day) and subcutaneous MTX up to year-8 of follow-up. All through this time although EEG evaluation showed focal epileptiform abnormality at the left frontal-central-parietal area, MRI imaging consecutively showed improvement of previously depicted brain lesions, with no further neurologic findings (Figure 3). Skin lesions stabilized and corticosteroids were progressively halted. Now, with a follow-up of 10 years, the patient continues treatment with oral MTX (10 mg/m^2), keeps on thriving well, with no development impairment. Skin lesions are progressively decreasing with normal hair growth pattern (Figure 4). Skin lesions show no inflammation and although improved, a discreet groove at the frontal bone on the upper right supraciliary area remains (Figure 5, white arrow). Oral and ophthalmological evaluation showed no abnormalities.

DISCUSSION

Scleroderma is a type of connective tissue disease that may occur at any time in life, although the clinical patterns of juvenile scleroderma differ from that of adulthood. Juvenile scleroderma can be classified as either systemic or localized. Systemic juvenile scleroderma is a chronic disorder that manifests with skin thickening and internal organ involvement, whereas localized scleroderma mostly affects skin, underlying musculature and bony structure. The predominant form of scleroderma found in children and adolescents is LoS, and although very rare, systemic involvement may occur in specific cases[3].

The European League Against Rheumatism has proposed a classification of LS based in dermatologic and rheumatologic findings that divides LoS in different subsets: 1) Linear scleroderma; 2) Progressive hemifacial atrophy; 3) Circumscribed morphoea; 4) Generalized morphoea; 5) Panclerotic morphoea; 6) Mixed juvenile LS; 7) LoS associated conditions[5]. Of these, linear scleroderma is the most frequent[3]. When craniofacial involvement is found a specific and rare type of linear scleroderma known as *en coup de sabre* is suspected. A common differential diagnosis of LSeCS is the Parry Romberg syndrome, also known as facial hemiatrophy of the face. Whether LSeCS represents a severe form of the latter is not consensual. Evidence suggests that patients with Parry-Romberg syndrome have linear lesions on the face but also on other parts of the body. There seems to be a greater involvement of the lower face and less so of the epidermis[10]. Cutaneous lesions on LSeCS often manifest as ivory-colored sclerotic plaques distributed on the frontoparietal scalp and forehead. Alopecia often accompanies the lesions probably due to excessive collagen deposition, which destroys hair follicules[4].

A diagnosis is usually based on clinical signs rather than on laboratory findings, since there are no confirmatory laboratorial tests[3,4]. Markers such as ANA may be positive in up to 50% of patients[3], but are non-specific. Though unheard of in other subsets of localized scleroderma, LSeCS as well as Parry Romberg syndrome[10] often associate with neurologic symptoms[3]. The development of cutaneous findings usually precedes neurologic ones in months to several years, but a few reports state otherwise[3,4]. The range of neurologic symptoms associated with craniofacial scleroderma can vary, but may include seizures, headaches, focal deficits and movement disorders. By far the most com-

mon symptoms are seizures[4]. In our case, recurrent seizures followed by cutaneous atrophy and alopecia led to clinical diagnosis of this condition, although not following the classical order of symptom presentation. Neuroimaging commonly depicts brain parenchyma atrophy, white matter lesions and focal subcortical calcifications. Lesions are usually ipsilateral to the skin lesions in the cerebral hemisphere. MRI scans exhibit T2 hyperintensities, mostly within the subcortical white matter, but also in the corpus callosum, deep grey nuclei and brain stem[3,4].

Oral involvement in LSeCS is rare, and it may be presented by white linear scar-like fibrotic areas, atrophic tongue papillae, gingival recession, and alveolar bone resorption[6]. Ocular abnormalities and otorhinolaryngological disorders may also be present but are not common[6].

Adequate management presents a clinical challenge, since the self-limited nature of this condition together with limited results of clinical trials are not consensual. MTX has the best evidence for systemic treatment of severe forms of linear scleroderma[7,8]. Several studies indicate that parenteral MTX may be more efficacious than the oral form and may be beneficial to mitigate gastrointestinal side effects[8,9]. Furthermore, systemic corticosteroids are usually added to therapy if disease is symptomatic or progressing[8]. A combined therapy has been proven effective[7]. Management in our patient included an initial combination of both MTX and corticosteroids, with a clear improvement in clinical symptoms. In the aftermath of discontinuing corticosteroids, the disease did not progress. Other treatment options are available such as excimer laser, calcipotriene or tacrolimus[7].

In conclusion, LSeCS is a rare form of linear scleroderma most frequently diagnosed in children and adolescents. Diagnosis is based in clinical symptoms, since laboratory findings are non-specific. This type of disease often presents with neurologic findings. A meticulous examination of these patients should be performed with particular attention to the face and scalp. Treatment is based on expert opinion but MTX and corticosteroids are considered the mainstay approach. A careful follow-up of these patients should be kept since neurologic abnormalities can develop over time. Neurologic abnormalities, especially if previously unexplained, upon the development of skin lesions should raise clinical suspicion of LSeCS.

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