Neurological Manifestations in Two Cases of En Coup De Sabre

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ABSTRACT

Scleroderma En Coup de Sabre (ECDS) is a form of localised scleroderma that primarily develops in the younger population, usually before the age of 18 years and occurs on the scalp or forehead. In localised scleroderma, en coup de sabre, many studies and case reports describe neurological signs and symptoms. Two patients with the disease are reported here who were noted to have brain cysts by neuroimaging. It is important to specifically inquire about neurological symptoms and signs in the history and examination, respectively, and to consider neuroimaging in patients with scleroderma en coup de sabre to diagnose and treat neurological complications.

Key Words: *Localised scleroderma, en Coup de Sabre, Neurological manifestations.*

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INTRODUCTION

Scleroderma is a disease that causes scarring of the skin. Scleroderma can be categorised into systemic sclerosis and localised scleroderma. In systemic sclerosis, in addition to the skin, various organs of the body are scarred. With localised scleroderma, the skin, with the underlying tissue is affected.

More than half of patients diagnosed with localised scleroderma were under the age of 18 years with an equal incidence in both genders.[1] When localised scleroderma occurs on the scalp or forehead, it is termed as "en coup de sabre" (ECDS). In the localised scleroderma ECDS, neurological manifestations are seen in a significant percentage of patients.[2] localised scleroderma ECDS affects one side of the scalp, or forehead and is associated with hyperpigmentation and hair loss.[3] ECDS forms a spectrum of head and neck involvement with Parry-Romberg Syndrome (PRS), which causes atrophy of half of the face and involves the subcutaneous tissue and bones of the face with or without scalp involvement and both ECDS and PRS can co-exist.[4]

Neurologic symptoms in ECDS can occur in the form of seizures, focal neurologic deficits, movement disorders, trigeminal neuralgia, and migraine.[5-10] Neuroimaging findings on computed tomography and magnetic resonance imaging include intracranial calcifications, white matter lesions, arteriovenous malformations and intracranial cysts.[8-10]

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CASE 1:

A 17-year girl presented to the neurology clinic with complaints of uncontrollable seizures and behavioural abnormalities. Upon examination, she had a dark lesion on the right forehead (Figure 1). The patient had a history of epilepsy dating back to the last five years and had used various antiepileptic drugs, including, topiramate, lamotrigine and carbamazepine, which were unable to control/treat her seizures.

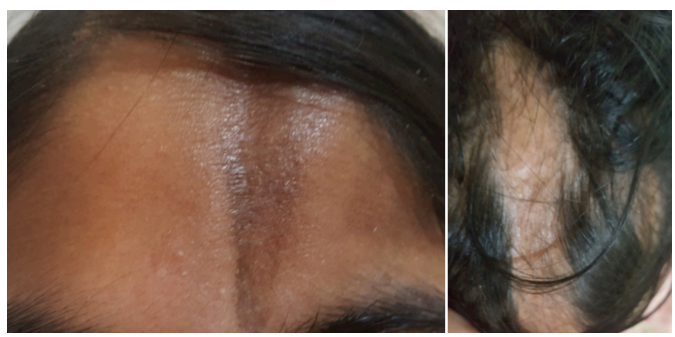

Figure 1: Sclerotic lesion on forehead with alopecia on scalp.

At the time of examination, she had been using sodium valproate and was suffering 1-2 seizures of the generalised tonic-clonic variety per month. She started experiencing behavioural abnormalities a month prior, which included episodes of crying and raging anger that were either a disproportionate response to the stimulus or had no stimulus trigger at all. Prior to the current episode, she had no behavioural problems, had a normal IQ and was a student of 9th grade. The lesion on the patient's forehead first appeared when she was 7 years old and gradually progressed to involve the scalp in continuity but stopped progressing for some time. The patient did not report any pain or discharge from the area. There was no antecedent history of trauma, infection or surgery. During examination, she

was fully conscious, oriented, following commands and had a mini-mental state examination (MMSE) of 24/30. The neurological examination did not show any focal signs. The forehead lesion was triangular in shape, depressed from the surrounding area, sclerotic, hyperpigmented and extending to the scalp with hair loss in the overlying scalp. The systemic examination did not yield any abnormal findings.

The patient was advised for laboratory investigations, an EEG and neuroimaging of the brain, a consultancy with a rheumatologist and the dose of sodium valproate were increased, and blood sample was collected for serum valproate levels for dose adjustment.

The laboratory investigations showed no abnormality, an inter-ictal EEG did not show any epileptiform discharges. Non-contrast CT of the brain showed the presence of two cystic lesions that were iso-dense with CSF in left fronto parietal and left occipital lobes with minimal to no mass effect on the surrounding brain, and a hypodense lesion involving the white matter of the left frontal lobe. It also showed some thinning of the diploe underlying the scarring on the scalp. Further, MRI of the brain showed similar cystic areas which were iso-intense to CSF on T1- and T2-weighted images, and the frontal white matter lesion which was hypointense on T1- and hyperintense on T2-weighted images. Both types of lesions did not show any enhancement on post-contrast images. MRI confirmed the CT finding of diploe thinning underlying the scalp lesion. No cortical atrophy was detected in neuroimaging (Figure 2).

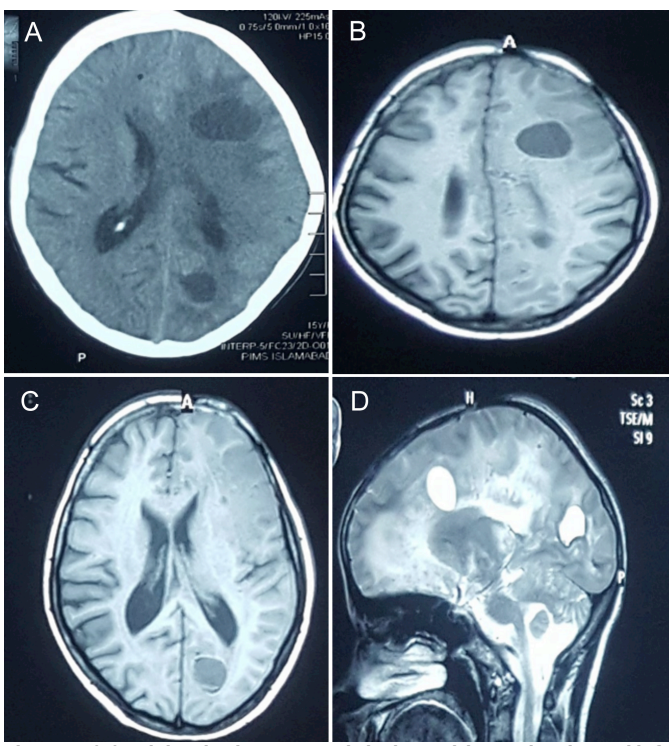

Figure 2: (A) Axial CT brain: Two cystic lesions with CSF density and left frontal lobe white matter hypodensity. (B) AND (C) Axial T1-weighted MRI showing left-sided CSF intensity cystic lesions and marrow fat thinning inside skull diploe. (D) Sagittal T2-weighted MRI showing left-sided two cystic lesions.

The laboratory results showed sodium valproate levels within normal limits and she was seizure-free on an adequate dose of sodium valproate. Serum autoantibody profiles including ANA and ENA were negative and chest x-ray and an ultrasound of the abdomen did not reveal any involvement of the lungs or abdominal organs, respectively. Opinions of a psychologist and psychiatrist were obtained for behavioural changes and she was started on a low dose of quetiapine, to which, she showed a good clinical response.

A neurosurgical consult was taken after neuroimaging and the neurosurgeon was of the opinion that since the cystic lesions were causing no mass effect and oedema and were located deep in the brain parenchyma, a surgical approach has more risks than benefits and a conservative approach with symptom control and observation is best suited to the patient.

The rheumatologist was of the opinion that since the lesion was not progressing for some time, the patient does not require any immune modulating therapy at the moment but requires a close follow-up. The patient was discharged on sodium valproate, quetiapine and was advised to regular follow-ups with neurology and rheumatology.

CASE 2:

An 18-year girl presented to the neurology clinic with a complaint of a continuous headache for the last one month. Headache was generalized, chronic and not associated with nausea, vomiting, photophobia, phonophobia or other features of primary headache syndromes and signs of raised intracranial pressure. The patient was using acetaminophen (over the counter) on as-required basis for headache. Her history depicted epilepsy, for which, she was taking carbamazepine that helped control her generalised tonic-clonic seizures. She was diagnosed with epilepsy 7 years prior and in the past had used sodium valproate, which affected a moderate control of seizures with one seizure every two weeks. On examination, a facial lesion was apparent on the left side of her forehead that was extending along the nasal bridge to the left side of the tip of the nose (Figure 3). There was no past history of trauma, surgery or chronic infection in the head or face. She had this lesion since the age of 5 years and had since progressed to involve more of facial area till the nose; moreover, there was no history of behavioural changes of any kind. Upon subsequent examination, no hemi-atrophy of her face was noted. There was no scalp involvement and the rest of the physical examination showed no abnormal findings. The patient was uneducated and her MMSE was consequently low at 22/30.

She was advised laboratory investigations, MRI brain and a rheumatology consultation.

The routine laboratory investigations showed no abnormality. An inter-ictal EEG did not show any epileptiform discharge. MRI brain showed a CSF signal intensity cystic area in the left frontal lobe, and hypointense white matter changes with focal cortical atrophy in the left parietal lobe with ex vacuo ventricular dilatation. There was thinning of marrow fat suggesting diploe thinning underlying the skin lesion (Figure 4).

The patient was discharged with follow-up advice including rheumatologist appointment, laboratory autoantibody tests and chest and abdomen imaging and prescribed treatment included carbamazepine and a low-dose anti-depressant for her chronic headache and advised to stop the use of NSAIDs. This patient was lost to follow-up.

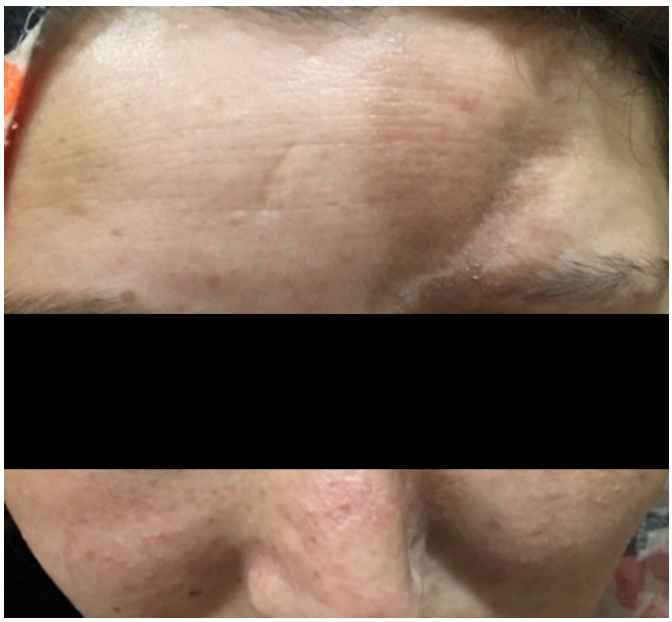

Figure 3: Sclerotic lesion on the left side of the forehead.

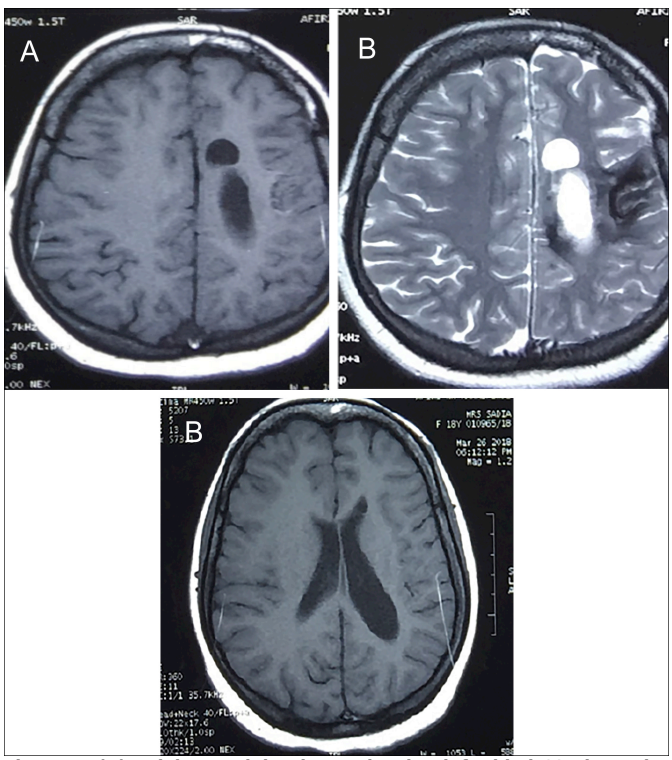

Figure 4: (A) Axial T1-weighted MRI showing left-sided CSF intensity cystic lesion adjacent to the frontal horn of ventricle, and focal cortical atrophy of parietal lobe. (B) Axial T2-weighted MRI showing left-sided CSF intensity cystic lesion adjacent to the frontal horn of ventricle, and focal cortical atrophy of parietal lobe with white matter changes. (C) Ex vacuo dilatation of left lateral ventricle compared to the right due to atrophy.

DISCUSSION

Scleroderma results from increased collagen production in the skin resulting in hardening, thickening and scarring of the skin, *i.e.*, sclerosis, hence the name scleroderma. It has an estimated prevalence of less than 3 per 100,000.[1] Localised scleroderma is traditionally considered to be limited to skin, and its underlying tissues.[1-3] Apart from the skin involvement, up to 20% of patients with localised scleroderma show rheumatologic, ophthalmologic and neurologic symptoms and signs.[3] Limited scleroderma needs to be differentiated from systemic sclerosis by the presence or absence of clinical signs like sclerodactyly. Raynaud's phenomenon and presence or absence of nail bed capillary abnormalities.

ECDS is a subtype of localised scleroderma. It usually involves fronto-parietal region, and 90% of cases are diagnosed between 2-14 years of age. The mean age of onset is 13 years.[1] Linear scleroderma ECDS presents like a band on the frontoparietal scalp and forehead, and with scalp involvement, hair loss is common. Skin lesions can extend to the nose, cheek, chin, and neck. If atrophy of hemi-face with involvement of muscle, bone and cartilage is found, a diagnosis of PRS should be considered.[7-10] Linear scleroderma ECDS and PRS should both be considered as a part of a spectrum of a single disease.

Multiple studies and case reports have documented neurological involvement in Linear scleroderma ECDS. In most cases, skin lesions predate neurological manifestations; although in up to 16% of cases, neurological manifestations may occur before skin lesions.[10] In both these cases, neurological findings developed after skin lesions, five years in the first case and six years in the second. Neurological manifestations described in studies include epilepsy, focal neurologic symptoms, migraines and trigeminal neuralgias, and neuropsychiatric manifestations.[2] A review of the literature of 54 patients, with linear scleroderma ECDS showed the following findings: 11% had normal neuroimaging, 37% had calcifications, 63% had more than one lesion on CT or MRI brain (T2W), while 31 % had a single lesion.[3] Eight patients had enhancing lesions on CT or MRI brain, six patients had abnormal MRA or cerebral angiogram, and 19 patients showed new lesions at follow-up. A literature review showed two case reports of cysts on neuroimaging in patients with PRS, but no case of intra-cerebral cysts with linear scleroderma ECDS.[7,8] Thus, the two cases reported here, are novel in nature and therefore are the first of their kind in known literature.

COMPETING INTEREST:

The authors declared no competing interest.

AUTHORS' CONTRIBUTION:

ZW, HM: Patient data collection, literature, review critical review, revision of the manuscript.

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