Frontal linear scleroderma en coup de sabre associated with epileptic seizure

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SUMMARY

Linear scleroderma is a rare variant of localised scleroderma, which is usually seen in childhood and during the adolescent period, and can cause severe functional morbidity as well as cosmetic and psychological problems. Although its aetiopathogenesis is yet obscure, autoimmunity, local ischaemia and injuries, vaccination, irradiation, vitamin K injections, *Borrelia burgdorferi* and *Varicella* infections have been incriminated. A 4-year-old girl who had been followed up for about 18 months with diagnosis of epilepsy had a colour discolouration and depression that first appeared 1 year ago and then progressed on her left frontal region. Her CT scan showed a thinning in the frontal bone and depression in the frontal region. These findings are described as 'en coup de sabre' a rare form of linear scleroderma localised at the frontal region of the scalp. In this paper, we present clinical and radiological findings of a 4-year-old girl with epileptic seizures that started 1 year before the onset of the lesion of linear scleroderma.

BACKGROUND

Scleroderma is a rare disease of unknown aetiology, characterised by thickening and hardening of skin resulting from increased collagen production. The term includes a variety of diseases, from localised scleroderma (LS) to systemic sclerosis. LS is traditionally considered to be limited to skin, subcutaneous tissue, underlying bone and, in craniofacial subtype, nervous system involvement.¹

Linear scleroderma 'en coup de sabre' (LSCS) is a subtype of LS. It is usually developing in the first or second decade of life, presents as band-like sclerotic lesions with more or less marked skin discolouration of the frontoparietal area. Characteristically, LSCS manifests unilaterally and does not extend below the eyebrow. The active stage usually lasts 2–5 years. Involutionary atrophy of skin, muscle and even bone may occur. Neurological and ophthalmological abnormalities are not infrequent in LSCS.^{2,3}

In this paper, the clinical and radiological findings of a 4-year-old girl with epileptic seizures that started 1 year before the onset of the LSCS were presented and discussed with recent literature.

CASE PRESENTATION

A 4-year-old girl was admitted to our hospital owing to skin discolouration and depression on her left forehead. She had been followed by the paediatric neurology department because of her epileptic seizures, and had been receiving antiepileptic drugs for 18 months. There had been no preceding fever or trauma. Her parent noted a 6-month history of

headache and echymotic colour change and a depression becoming progressively more prominent affecting her left forehead. She was the third child of non-consanguineous healthy parents and she had a healthy brother and a healthy sister. No previous similar case was described in her family.

INVESTIGATIONS

On physical examination, her height was 103 cm, weight was 15.5 kg and blood pressure was 110/70 mm Hg. There was skin thinning and darkening of the skin colour on the left forehead. There was a mild bone depression and atrophy under these affected skin regions. Other systemic findings were unremarkable. Complete blood count, erythrocyte sedimentation rate and biochemical findings of the serum were within normal ranges. Rheumatoid factor and antinuclear antibodies were negative. Cranial multidetector CT imaging demonstrated that there was a frontal bone atrophy and thinning on left frontal calvarium (figures 1 and 2). There were no sclerotic or lytic lesions of the bone structure. Three-dimensional reconstruction images were performed using a multidetector CT scanner. A mild depression on the frontal bone was demonstrated on a three-dimensional reconstruction image (figure 3). A diagnosis of LSCS was made based on the characteristic skin changes, clinical and radiological findings.

TREATMENT

The patient was treated with prednisolone (10 mg/kg/day) and methotrexate (10 mg/m²/week). Weekly subcutaneous methotrexate was started in combination with the prednisolone. The patient's cutaneous and neurological disease has stabilised on this treatment regimen. On treatment with high-potency topical corticosteroids, there was a significant regression of her frontal lesions. Over the next 6 months, serial EEGs showed improvement in wave form, and the patient has been stable with anticonvulsant treatment.

DISCUSSION

In the present case, LS, thinning and depression of frontal bone were observed. The skin involvement did not extend below the forehead. These features were consistent with a diagnosis of LSCS. It is a rare subset of LS defined by its characteristic location on the forehead and scalp. Clinically, it is manifested by ivory coloured, sclerotic plaques with violaceous borders distributed in a band-like manner on the frontal scalp and forehead.⁴ Onset is usually during the first two decades of life. In addition to a morphea-like sclerosis of the skin, there can be depression, atrophy or calcification of

To cite: Inci R, Inci MF, Ozkan F, et al. *BMJ Case Reports* Published online: 10 December 2012
 doi:10.1136/bcr-2012-007837

Figure 1 Cranial multidetector CT imaging shows frontal bone atrophy on the left frontal bone (arrows).

underlying tissues. Unlike the gradual spontaneous remissions seen in morphea, LS tends to have a longer and occasionally progressive course.⁵

The aetiology of LS is unknown. Environmental exposures, immune alterations, microchimerism with autoimmunity, trauma and *Borrelia* infection have been suggested as contributing to the pathogenesis of the disease.^{5–6} The vast majority of cases are sporadic. However, a small number of familial LS cases have been reported.^{6–8} Epidemiological, genetic, familial and twin studies suggest that scleroderma is not a heritable disease but may arise in genetically susceptible individuals when exposed to environmental triggers.^{6–7}

Multiple case studies have been published describing neurological dysfunction associated with LS. Typically, the skin lesion develops prior to the onset of neurological symptoms though cases of the reverse have certainly been described.⁹ Many case studies have described the association between LS and neurological abnormalities. A case study in 2002 described a patient with LS and progressive atrophy of the ipsilateral cerebral hemisphere, not associated with overlying facial lesions.¹⁰ Another in 2009 described a patient with refractory partial seizures prior to the development of a skin lesion on his head.¹¹ A study of 55 neurologically symptomatic patients with LS showed that 78% had central nervous system (CNS) lesions only on the side of the skin lesion.¹² Two similar case studies of neurologically

Figure 2 Cranial multidetector CT imaging demonstrates frontal bone thinning on the left frontal calvarium (arrow).

Figure 3 Three-dimensional reconstruction of multidetector CT image shows a mild depression on the frontal bone in a 4-year-old girl.

asymptomatic patients with LS who received CNS imaging showed that 82–100% (n=13) had exclusively ipsilateral lesions in the brain.^{13–14}

The diagnosis is a clinical one that relies on the characteristic cutaneous and soft tissue findings because there are currently no diagnostic laboratory tests. Antinuclear antibody with homogeneous and speckled patterns may be positive in 37–50% of patients with LS, and antisingle-stranded-DNA antibodies may be present. More specific autoantibodies such as Scl-70, anticentromere, Ro/La and U1RNP may precede the development of systemic disease, and patients with these markers should be followed closely for several years.¹⁵

CT and MRI studies have shown central nervous system abnormalities in LSCS patients. Neurological findings are more frequently ipsilateral to the skin lesions, but contralateral involvement has been described.¹⁶ Neurological symptoms should not be used as a predictor for MRI abnormalities because neurological lesions have been discovered in asymptomatic patients.¹⁷ Moreover, symptomatic patients were sometimes proven to have normal radiological examinations. Outer diploe thinning, cerebral atrophy, white matter lesions, focal subcortical calcifications and meningocortical alterations have been described.¹⁷

The management of LS is still unsatisfactory. Various therapeutic modalities (eg, topical and pharmacological agents, immunosuppression, physiotherapy and phototherapy) have been suggested.¹⁸ In a retrospective study of LSCS and/or Parry-Romberg syndrome (PRS) patients at a tertiary care centre, anti-malarials, methotrexate, topical and oral steroids, and tetracycline were used for cutaneous disease, but no definite conclusions could be drawn owing to the small sample size and the absence of a control group.¹¹ D-penicillamine, methylprednisolone, mycophenolate mofetil and methotrexate might be considered in the treatment of neurological involvement of LSCS.⁹ In reported cases, association of methotrexate or mycophenolate mofetil and steroids appeared to have impact in controlling intractable seizures and stabilising central nervous system damage.^{9–12–18} However, management of frontal and facial atrophy is a challenge and palliative reconstruction surgery is potentially beneficial to patients with disfiguring facial atrophy, and the application of three-dimensional reconstruction imaging may be helpful in preoperative scheduling of bony reconstruction surgery.¹⁵

In our patient, neurological manifestations occurred before the diagnosis of LSCS. The neurological symptoms were previously unexplained when they presented to dermatology for an evaluation of their skin lesions. This case illustrates the need for placing craniofacial linear scleroderma on the differential of any patient with neurological abnormalities such as partial seizures. A thorough examination of the skin should be performed in these patients, with particular attention to the face, forehead and scalp. Long and careful follow-up of these patients is prudent based on evidence that neurological abnormalities may develop at any time during the course of the disease.

Learning points

- ▶ Linear scleroderma 'en coup de sabre' is a subtype of linear scleroderma and usually develops in the first or second decade of life, presents as band-like sclerotic lesions with more or less marked skin discolouration of the frontoparietal area.
- ▶ Linear scleroderma 'en coup de sabre' has been associated with a variety of neurological abnormalities and, typically, is preceded by the development of cutaneous disease by months to years.
- ▶ Neurological abnormalities may accompany with linear scleroderma 'en coup de sabre' and CT and MRI studies have shown central nervous system abnormalities.

Competing interests None.

Patient consent Obtained.

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