CASE REPORT

Sjogren Larsson Syndrome

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Abstract
Sjogren Larsson Syndrome(SLS) is a rare neuroectodermal disorder characterized by congenital icthyosis, mental retardation and spastic diplegia along with pathognomic opthalmological findings. We report two siblings with Sjogren Larsson Syndrome from an Indian family but, without eye involvement.

Key Words
Sjogren Syndrome, Congenital icthyosis, Spastic Diplegia

Introduction

Sjogren Larsson syndrome (SLS) is a rare autosomal recessive neuroectodermal disorder, characterized clinically by congenital ichthyosis, mental retardation, and spastic diplegia or tetraplegia. It occurs in all races and its prevalence has been estimated as 0.4 per 100,000 or lowers. Over 200 cases worldwide have been reported (1). We report two cases with typical features of Sjogren Larsson syndrome.

Case Report

Two siblings of a family, an 8 year old boy and 11 years old girl born to non consanguineous parents at term by normal vaginal delivery, brought with complaints of severe icthyosis and spastic paraplegia since early childhood. There was no history of teratogenic drug intake and birth asphyxia. They had generalized erythema at birth and later developed scaling on neck, axillae, abdomen, back, cubital and popliteal fossa. They were noticed to have motor and speech delay at 18 months of age . On examination, they had mild mental retardation, spastic quadriplegia with predominantly lower limb involvement with brisk reflexes and ankle clonus. Cutaneous examination revealed generalized dryness of skin with thick blackish scales over the trunk and on the extremities. The flexures were involved and showed lichenification. Both neurological and cutaneous symptoms were more severe in boy. The face, palms, soles, nails and teeth were normal. Ophthalmologic evaluation in both children did not reveal foveal or parafoveal glistening dots. MRI of the brain done in boy showed active demyelination involving predominantly anterior bilateral cerebral white matter and posterior limbs of internal capsule,corona radiate and central semiovale. Skin biopsy confirmed the findings of icthyosis. Genetic analysis for FALDH activity could not be done because of non availability of test.

Discussion

SLS is due to a genetic block in the oxidation of fatty alcohol to fatty acid because of deficient activity of fatty aldehyde dehydrogenase (FALDH). This enzyme catalyzes the oxidation of long chain fatty aldehydes to fatty acids. Accumulation of long-chain fatty alcohols and modification of macromolecules by an excess of fatty aldehydes are thought to be the pathophysiologic mechanisms causing the manifestations of SLS (2) Clinically ichthyosis is usually present at birth in patients with SLS, but in some it may be seen only after patients are one year old. Mental retardation and spastic quadriplegia/diplegia are usually evident by the third year of life. After neurological symptoms appear, development is progressively delayed. No progression of the neurological findings or mental retardation occurs after

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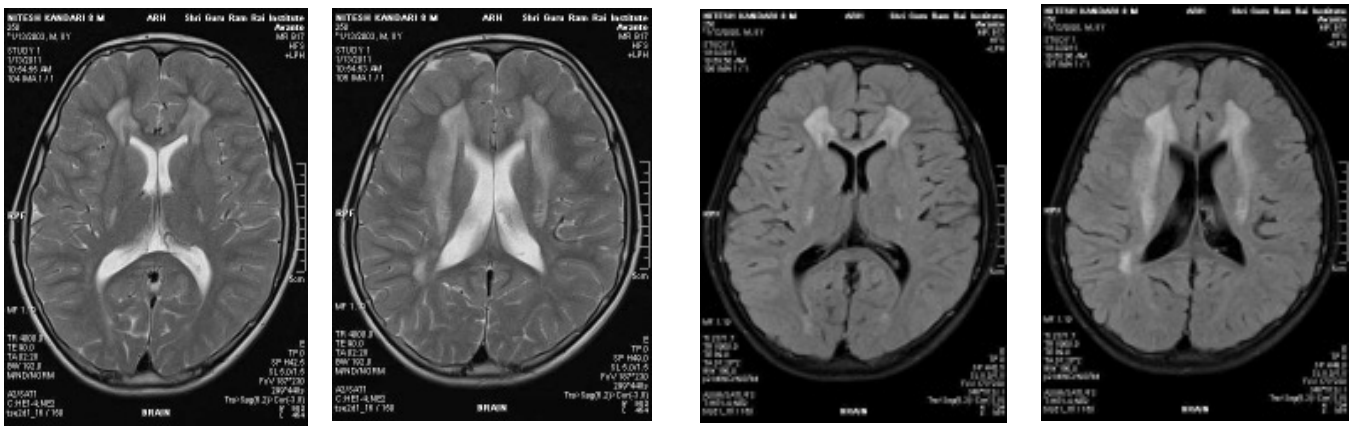

Fig.1-4 T2 and Flair Images of MRI Head Show Bilaterally Symmetrical Hyper-intense Foci Involving Bilateral Peri-ventricular and Deep White Matter Anteriorly and Posterior Limbs of Internal Capsule Suggestive of Demyelination

puberty. The disease does not show any sex preference (2). Arms are usually less severely affected than the legs (3). The diagnosis is almost always delayed till recognition of coexistence of neurological and cutaneous abnormalities. SLS should be considered in all patients when spastic para paresis is associated with unusual cutaneous signs. Only one third or about 20% of the cases exhibit a distinctive and perhaps pathognomic presence of glistening white dots surrounding the macular region of retina, which first appears after several years of age (4,5,6). However, this finding was not present in both of our patients at the time of examination and repeat followup 6 and 12 months later. Similar finding was observed by Dhanuka AK in 2002. MR imaging shows retardation of myelination and dysmyelination. The skin biopsy in SLS patients shows hyperkeratosis, focal parakeratosis, acanthosis, papillomatosis, and sparse dermal lymphocytic inflammatory infiltrate. The management is primarily of rehabilitative nature for the physical and neurological defects with early physiotherapy, and subsequent soft tissue surgery, local application of emollients, keratolytics and retinoid. Zileuton, a 5-lipooxygenase inhibitor, have been shown to decrease pain and pruritis with SLS (7).

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