Case Report:

Sjögren-Larsson syndrome- a case report

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Abstract:

The present article represents a case report on a 10 year old male child with generalized dryness since infancy as well as difficulty in walking since a young age. the child presented with recurrent episodes of falling over and pruritis. the authors have concluded that a diagnosis of SLS must be considered in any neonate or infant with congenital icthyosis and emerging neurological features.

Keywords : Sjögren-Larsson syndrome

Introduction:

Sjögren-Larsson syndrome (SLS) is inherited as an autosomal recessive disease and is associated with congenital ichthyosis, spastic diplegia or tetraplegia, and mental retardation, caused by a deficiency of fatty aldehyde dehydrogenase[1]. SLS is an inborn error of lipid metabolism that is caused by a deficiency of the microsomal enzyme fatty aldehyde dehydrogenase (FALDH), a component of fatty alcohol: NAD-oxidoreductase enzyme complex. FALDH deficiency has been found to lead to an accumulation of long-chain fatty alcohols with structural consequences for cell membrane integrity which do disrupt the barrier function of skin and the white matter of the brain[2]. It occurs in all races and its prevalence has been estimated as 0.4 per 100,000 or lower. Over 200 cases worldwide have been reported[3]. The case report is as follows.

Case report:

A 10 year old male child born of second degree consagnuity, presented to the pediatric out -patient department with generalized dryness of skin since infancy and difficulty in walking since the age of 3-4 years. He had recurrent episodes of falling over and there was waxing and waning of pruritis, the episodes lasting 20 days every 2-3 months. Stiffness in lower limbs started in later part of the first year of life with progressive increase up to the time of presentation. The child was delivered at full term by normal vaginal delivery and he had an uneventful post natal period. Further, it was found that the child had attained head holding at 6 months, sitting without support at 1 year age and walking with support at 2 years. Neurological examination revealed mental retardation, increased tone in both lower limbs, brisk deep tendon reflexes, and bilateral extensor plantar response suggestive of spastic diplegia. The upper limbs did not show any tone or deep tendon reflex abnormalities. There was also evidence of conduction aphasia in the child. Sensory system was normal. Skeletal, dental, eye/ fundus examination and eye or limb movements were normal. Child also had

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microcephaly. Scholastic performance was poor. Skin examination showed scaly ichthyotic lesions with severe pruritus affecting all body parts and which started in late infancy on the face, neck and abdomen. Chest radiograph and all routine hematological investigations were normal. Skin biopsy revealed hyperkeratosis dermis with non-specific peridermal inflammation. MRI revealed cerebral atrophy. BERA revealed sensory neural hearing loss. The patient showed progressive worsening of ataxia and diplopia and eventually became bed ridden. However, the icthyosis was seen to improve. The patient is currently on regular follow up. The discussion is as follows.

Discussion:

In 1957, Sjögren and Larsson described a rare syndrome that was characterized by congenital ichthyosis associated with spastic diplegia or tetraplegia (spastic pyramidal symptoms), aphasia, and low-grade dementia[4]. SLS is an inherited neurocutaneous disorder and it is caused by mutations in the ALDH3A2 gene that is found to encode fatty aldehyde dehydrogenase (FALDH). Over 70 mutations in ALDH3A2 have been discovered in SLS patients including amino acid substitutions, deletions, insertions, and splicing errors[5]. Unfortunately, the diagnosis of SLS is almost always delayed because usually only cutaneous symptoms are present at birth.

Newborns may manifest symptoms and signs of the disease (first ichthyosis, subsequently neurologic symptoms).Spasticity may be apparent before age 3 years and is more severe in the lower limbs than in other parts of the body[6]. The diagnosis must be considered in all patients with generalized hyperkeratosis and central nervous system dysfunction. Glistening dots in the macular region is considered pathognomonic, although it is not constant. The nature of the crystalline deposits in the retina is unclear. It is postulated that they might represent accumulations of long-chain fatty alcohols or fatty aldehydes[7]. There is also spastic diplegia, occasionally tetraplegia, with mental retardation, epilepsy, speech defects, dental, dermatological, skeletal, and retinal changes[8]. Skin manifestations are in the form of icthyosis which is a generalized hyperkeratosis of the trunk, joints, and the dorsal aspects of the hands and the feet. Neuropathologically, the hallmark of SLS is demyelination of the cerebral white matter and corticospinal and vestibulospinal tracts that is responsible for spasticity[9]. Recently, few reports have described the use of proton MRI spectroscopy in Sjogren–Larsson syndrome which shows abnormal lipid peak at 1.3 ppm at both TE-30 and 135 ms in the area of T2 white matter abnormalities[2]. Management in SLS is supportive [10]and mainly involves topical moisturizing lotion, keratolytic agents and oral retinoids. Training to provide home-based physical therapy is useful to prevent contractures.

Conclusion:

In conclusion, the diagnosis of SLS must be considered in any neonate or infant with congenital icthyosis and emerging neurological features. Ocular features and pruritus must be looked for. Cerebral MRI reveals arrested myelination or demyelination in white matter and lipid peak on spectroscopy helps in making the diagnosis. However, spectroscopy is a rare resource and is not readily available to be made use of in a country such as ours, which is why a high degree of suspicion based on the clinical findings alone will go a long way in reducing the morbidity and mortality due to this condition.

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