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Magnetic Resonance Imaging and Proton Spectroscopy in Sjögren-Larsson Syndrome: Case Report

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Introduction

Sjogren Larsson Syndrome (SLS), an autosomal recessive disorder of lipid metabolism, was first described in Vasterbotten County, Sweden. The worldwide prevalence is unknown, but the estimated prevalence is only 0.4 per 100,000 population. SLS patients are deficient in the Fatty Aldehyde Dehydrogenase (FALDH) component of fatty alcohol-NAD+ oxidoreductase. Consequently, fatty alcohols and fatty aldehyde accumulate, leading to altered cell integrity and an increase in biologically active lipids.

Case report

Male, 02-years-old, born to nonconsanguineous parents, with delayed neuropsychomotor development and progressive spastic tetraplegia. The antenatal history revealed an uneventful pregnancy but premature vaginal delivery at 32 weeks gestation and complicated by meconium aspiration. She was born with 2.4 kg and exhibited dry and scaly skin, with a later diagnosis of congenital ichthyosis confirmed by skin biopsy. There was no history of seizures or similar cases in the family. He underwent audiometric testing and several serum tests during the

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diagnostic investigation, such as hemogram, liver, kidney, and thyroid function, lipid profile, calcium, phosphorus, alkaline phosphatase, uric acid and homocysteine with typical results. A Magnetic Resonance Imaging (MRI) study of the brain with proton spectroscopy (MRS) was performed, which revealed extensive signal abnormality compromising the white matter of the cerebral hemispheres (Figure 1), as well as the presence of lipid peaks at 0.9 ppm and 1.3 ppm (Figure 2) within short and long echo times. Funduscopy demonstrated the presence of yellowish-white crystals in the foveal and parafoveal areas. The triad of congenital ichthyosis, cognitive delay and spastic quadriplegia raised the suspicion for SLS, being highly suggestive when associated with characteristic imaging findings, proton spectroscopy and fundoscopy. The patient evolved with attenuation of skin lesions related to congenital ichthyosis due to treatment with emollient oils (Figure 3) but persisted with the progression of neuropsychomotor developmental delay and spastic quadriplegia.

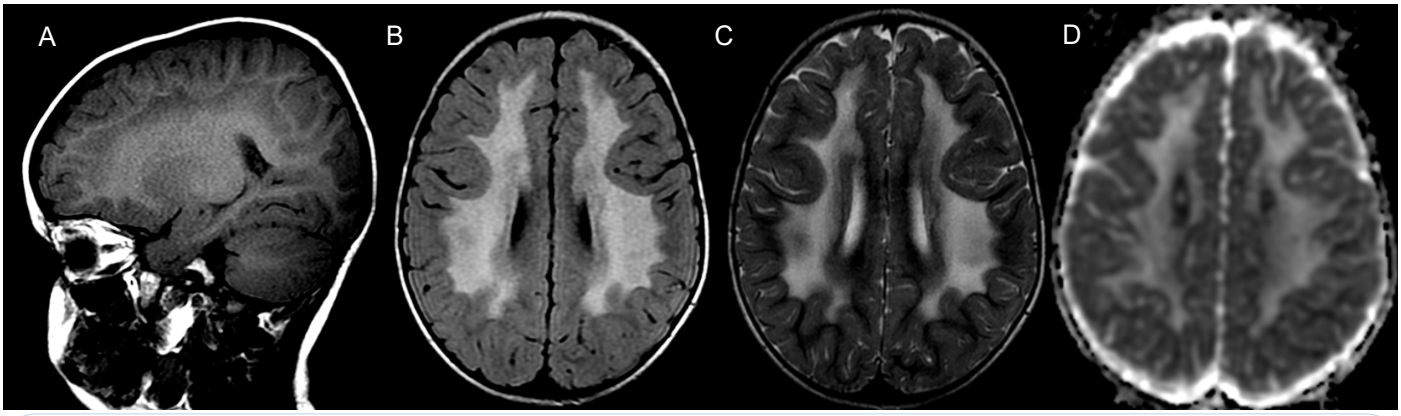

Figure 1: Sagittal T1-weighted image **(A)** shows hypointensity of the subcortical white matter. Axial FLAIR and T2-weighted images **(B&C)** reveal extensive symmetrical hyperintensity compromising profound and periventricular white matter of the cerebral hemispheres. Axial ADC map **(D)** demonstrates small areas of restricted diffusion within the signal abnormality.

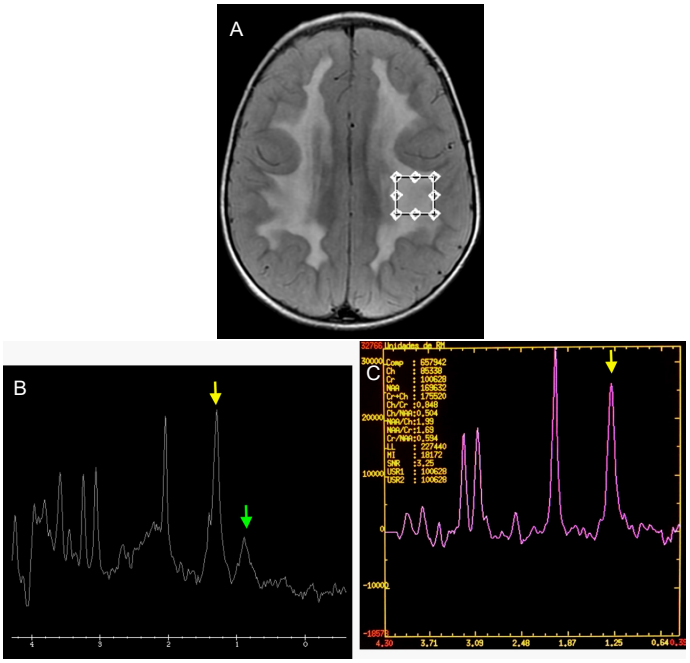

Figure 2: MR spectroscopy in the left semioval center **(A)**. The short echo time - 35ms (Figure B) demonstrates presence of lipid peaks at 0.9 ppm (green arrow) and 1.3 ppm (yellow arrow). The long echo time - 144ms (Figure C) shows the lipid peak at 1.3 ppm (yellow arrow).

Discussion

SLS is a rare autosomal recessive disorder characterized by the clinical triad of congenital ichthyosis, cognitive delay and spastic tetraplegia or diplegia [1-8].In addition to exhibiting the complete triad, the patient reported in this study also demonstrated characteristic findings on fundoscopy and imaging examinations.

The clinical manifestations are likely related to abnormal accumulation of alcohol and fatty aldehydes secondary to FALDH deficiency, and it is recognized that FALDH plays a role in LeukoTriene B4 (LTB4) metabolism, so it is hypothesized that increased urinary excretion of LTB4 by the fetus in the amniotic fluid may be the trigger for the induction of an inflammatory response, however, there is no definitive evidence for this theory [1].

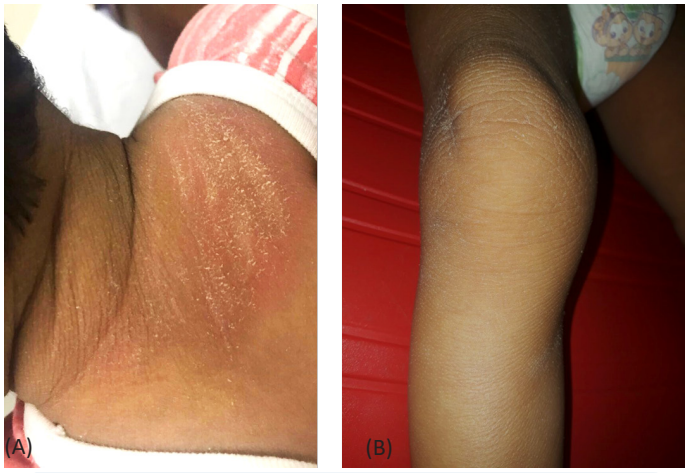

Figure 3: Photos of the patient's back **(A)** and knee region **(B)** demonstrate ichthyosis.

Ichthyosis in SLS is identified in the neonatal period and is characterized by diffuse skin involvement with a lichenified appearance and brownish coloration, particularly in the flexures [1].

Ocular abnormalities are common and usually manifest after two years of age, including flickering, yellowish-white dots in the macular area of the retina, of undetermined etiology. The presence of these foveal and parafoveal crystals is of cardinal importance and probably a pathognomonic sign of this syndrome [1,8].

The neurological picture manifests with spasticity, usually progressive, intellectual disability and sometimes seizure.

Diagnosis begins by identifying the clinical phenotype of the early onset neurocutaneous disorder. While premature birth and congenital ichthyosis are considered early findings, delayed motor function, cognitive deficits, and retinal abnormalities develop later. Diagnostic suspicion is strengthened by characteristic MRI findings and confirmed by targeted sequencing of the ALDH3A2 gene [1-7].

MRI may demonstrate T1 hypointensity and T2 hyperintensity of the periventricular white matter, with preservation of U-shaped fibers. The corpus callosum and cerebellum are usually not compromised, and anomalous contrast enhancement or diffusion restriction areas are not usually characterized. The extent of involvement and signal change on MRI may be progressive or stable and does not correlate with age or clinical picture [1].

MRS can reveal characteristic lipid peaks at 0.9 and 1.3 ppm, the latter being more expressive, observed in the short (35ms) and long (144ms) echo times, denoting a defect in fatty acid metabolism [1-7]. Therefore, some authors propose that proton spectroscopy can be used as a reference for therapeutic monitoring [5].

The differential diagnosis of SSL should be made with cerebral palsy, Rud syndrome, and Desantis-Cacchione syndrome and Refsum syndrome [1-3,8].

Importantly, abnormal lipid peaks are also seen in multiple sclerosis, Niemann-Pick disease type C and other peroxisomal disorders, however, in these diseases, lipid peaks are broad and more minor, found only in studies with short echo time, in contrast to SLS [1,4].

The disease has no cure and its treatment is palliative, affected patients should preferably be followed by a multidisciplinary team of neurologists, ophthalmologists, dermatologists and rehabilitation specialists. Some authors advocate the theory of dietary modification by reducing total fat intake and increasing the ratio linoleic acid / linolenic acid, but with still limited benefits. Symptomatic treatment of ichthyosis consists of applying emollients, keratolytics and topical calcipotriol [1]. The prognosis depends on the neurological abnormalities and the survival rate described in the literature is around 20 to 30 years [2,8].

Conclusion

SLS is a rare entity with imaging findings poorly described in the literature, however they can be suggestive and aid in the diagnosis and therapeutic management. Therefore, the diagnostic hypothesis should be considered in patients with congenital ichthyosis, cognitive delay and characteristic MRI findings consisting of signal abnormality of the white matter of the cerebral hemispheres and presence of lipid peaks on spectroscopy.

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