

Do you know this syndrome?*

Você conhece esta Síndrome?

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CASE REPORT

Male patient, aged 20, white, with xeroderma and intense pruritus since he was born that worsened over the years. He also presented loss of visual acuity, neurologic deficit with mental retardation, psychomotor agitation and aggressiveness. There was no report of colloid membrane at birth, prolonged labor or appearing of bubbles. The previous diagnosis was Sjogren-Larsson Syndrome (SLS) and the patient was under neurological and psychiatric control and taking anxiolytics. Physical examination showed cheerful uncommunicative facies, diplegic gait pattern, with

flexed hips and knees at all stages, support with “pes equinus” and collapse of the plantar arch. The patient presented mental retardation, difficulty to understand, diffuse xeroderma with lichenification, excoriations, ichthyosis and desquamation that affected mainly neck, abdomen and folds. (Figures 1 and 2). He did not have palmoplantar keratoderma. Exam of the ocular fundus (fundus of eye) showed the presence of retinitis pigmentosa. Patient was treated with emollients, topical keratolytic and oral antihistamine.



FIGURE 1: Patient with xerotic and excoriated skin



FIGURE 2: Xerotic and excoriated skin with lichenification

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WHAT SYNDROME IS THIS?

It refers to a rare neurocutaneous disorder, autosomal recessive with an incidence of 1 to 100.000 individuals in Sweden. It was described in 1957 by Sjogren and Larsson as from a group of 28 patients with the triad of congenital ichthyosis, development of paraplegia or tetraplegia and mental retardation.^{1,2,3,4} Later, it was observed the presence of deficit in oxidation in the long-chain of fatty acids due to an inborn error in the lipid metabolism caused by deficiency of the enzyme fatty aldehyde dehydrogenase (FALDH) which generates deposit of lipid metabolites on the tissues. This alteration is caused by mutation of the ALDH3A2 gene that codifies FALDH. Most cases seem to be severely affected independent of their genotype.^{1,2,3,4}

The accumulation of lipids on the skin and nervous system is responsible for the disease outbreak. The fatty acids accumulated on the skin disorganize the transepidermal water barrier, leading to its loss and to ichthyosis. Deficiency in the oxidation of the fatty acids generates delay in myelination and demyelination of nerve fibers. Alteration in the integrity of the myelin membrane generates the neurological condition.^{1,3,5}

It is characterized by ichthyosis with pruritus, spasticity with diplegia or tetraplegia, mental retardation, ocular alterations and leukoencephalopathy.^{1,2,3,4,6,7,8}

Dermatological manifestations start at birth, with erythema. Rarely, the newborn can be surrounded by the colloid membrane and present ectropion. At

first the skin becomes dry, rough, brownish and desquamative due to a defect in keratinization. In general, the periumbilical region, neck and folds are affected while the face is spared. Pruritus is persistent and universal, generating lichenification and excoriations. There might be palmoplantar keratoderma.^{1,2,3,4,6,8}

Neurological deficit is observed in childhood with progressive retardation in motor development, pyramidal syndrome and spasticity, being the lower limbs the most affected ones. Cognitive development is slow with mild to moderate mental retardation and dysarthria.^{1,2,3,4,6,8}

Photophobia and decrease on the visual acuity are common and reported since the early years of life. Crystalline maculopathy, characterized by whitish dots arranged surrounding the fovea in eye fundus is observed in 100% of the cases. It is probably caused by deposit of lipofuscin pigment in the epithelium of the retina.^{1,3,4,7}

Diagnosis is made by the presence of the classic triad and by the alteration of the eye fundus. It is confirmed by demonstration of the deficiency of FALDH in cultures of fibroblasts or leukocytes.^{1,2,3,4,5} It must be differentiated from recessive ichthyosis linked to X, ichthyosis vulgaris, bullous and non-bullous congenital ichthyosiform erythroderma and ichthyosis lamellar. Treatment is multidisciplinary and includes the use of emollients, keratolitics as well as systemic retinoids.^{1,3,4,8} □

Abstract: We report a typical case of Sjogren-Larsson syndrome in a male patient, aged 20. The Sjogren-Larsson syndrome is a neurocutaneous, autosomal recessive and disabling condition, characterized by congenital ichthyosis, spastic paraplegia and mental retardation. It is caused by deficiency of the microsomal enzyme fatty aldehyde dehydrogenase. It has no cure, but most patients survive up to an adult age. Treatment should be multidisciplinary and dermatological therapy aims at relieving the persistent itching and ichthyosis.

Keywords: Ichthyosis; Mental retardation; Paraplegia; Sjogren-Larsson Syndrome

Resumo: Relatamos um caso típico, em um paciente masculino de 20 anos, da síndrome de Sjögren-Larsson, que é uma doença neurocutânea, autossômica recessiva e incapacitante, caracterizada por ictiose congênita, plegia espástica e retardo mental. É causada pela deficiência da enzima aldeído graxo desidrogenase. Não tem cura, porém a maioria dos pacientes sobrevive até a idade adulta. O tratamento deve ser multidisciplinar e a terapia dermatológica tem o objetivo de aliviar o prurido persistente e a ictiose.

Palavras-chave: Ictiose; Paraplegia; Retardo mental; Síndrome de Sjögren-Larsson

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