SJOGREN LARSSON SYNDROME

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INTRODUCTION

The Sjogren - Larsson Syndrome (SLS) is an autosomal recessive disorder. It is characterized by three cardinal symptoms: congenital ichthyosis, spastic diplegia or tetraplegia and mental retardation.[1] SLS was first described by Sjogren in 1958, Sjogren and Larsson in 1957 and independently by Soderhjelm and Enell in 1957. The majority of patients have been reported from Sweden where the prevalence in certain regions is as high as 1 in 12,000, although the disease occurs world wide at a much lower frequency.[2]

CASE REPORT

We report a eight years old male child whose mother had full term and uneventful pregnancy. Birth weight was 5 kg. Their was no history of icterus during the first few weeks. Blood groups were identical in both mother and child. At birth the patient had diffuse redness of the skin, which gradually faded during childhood, where after the skin became dry, thick and scaly. Occasionally pruritus was experienced, but there was no heat intolerance. Early motor development was normal. The patient began to sit at seven months, had normal speech at ten months and could walk at fifteen months. It was not until later in childhood, at about four years of age, that the parents noted a disturbance of gait. At the age of 7 years he also started having tonic clinic fits.

Clinical examination revealed a boy who appeared to be his chronological age. He was 55 inches tall and 100 pounds in weight. The blood pressure was 110/80 mmHg and the heart, lungs, abdominal organs and genitalia appeared normal. Vision was 6/6 in both eyes and fundi were normal. The head was normocephalic. Separation of the teeth was slightly increased. Hearing was impaired and otologic examination revealed sclerosis of both eardrums.

The skin exhibited a rough exfoliative hyperkeratosis, especially marked on the back, knees, inner surface of hands, palm and soles. Nails were dystrophic. Hypertonicity with increased reflexes was observed in the lower limbs. The Bibinski was bilaterally positive. Gait was spastic with scissoring of lower limbs. The IQ was around 40. Rest of the CNS examination was unremarkable.

The peripheral blood picture, biochemistory and urine analysis were normal. Radiologic examination of the skeleton showed no abnormalities. The bone age was appropriate to the chronologic

Fig. 1

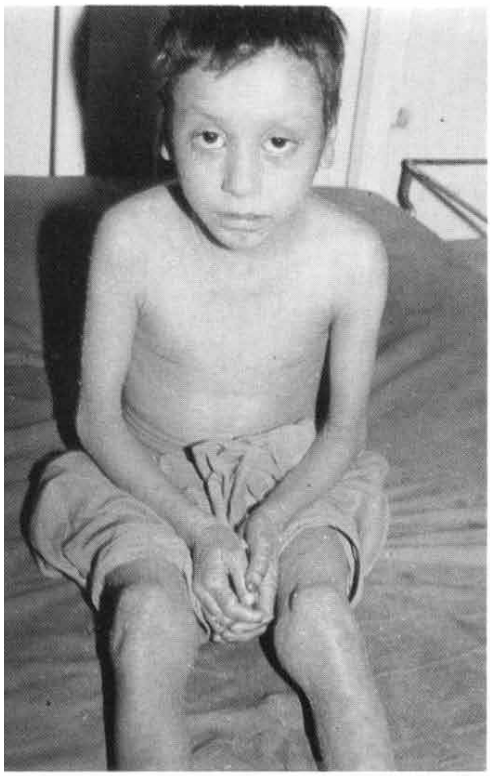
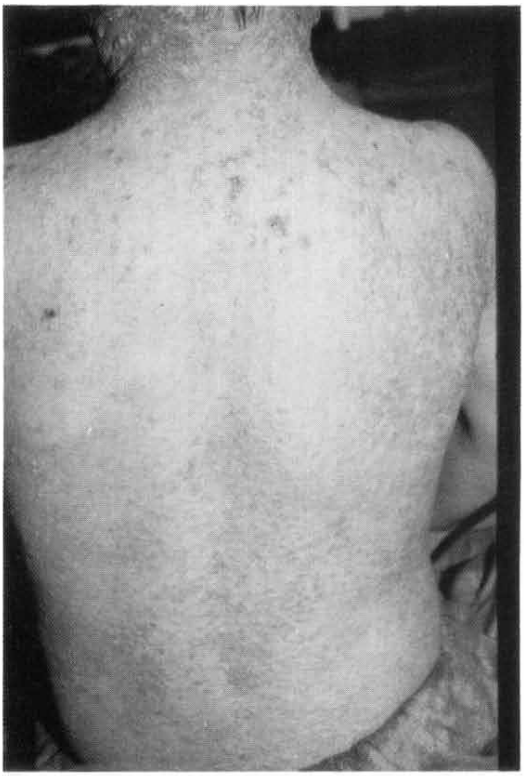
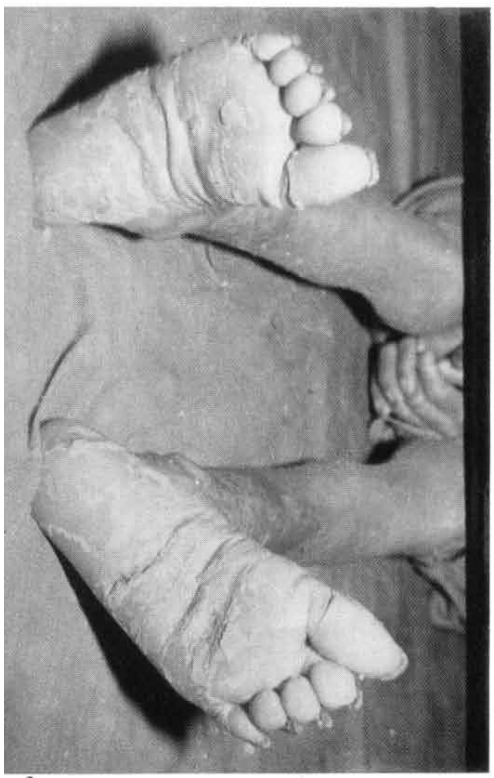


Fig. 2





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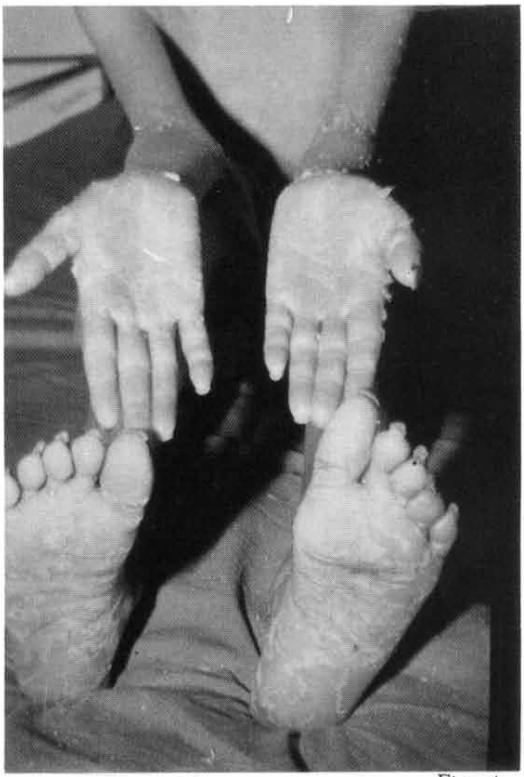

Fig. 4

age. The EEG revealed a poorly organized basic activity and bilateral eplieptic discharges. Skin biopsy revealed hyperkeratosis, hypertrophy of the granular layer and stratum mucosum, long and irregular papillary bodies and a chronic inflammatory infiltrate in the upper dermis.

The diagnosis of Sjogren Larsson Syndrome was made and the patient was discharged on multivitamins, antiepileptics and advised daily oil application.

DISCUSSION

Sjogren Larsson Syndrome have a variety of presentations. Besides the ichthyosiform erythroderma, pyramidal spasticity and mental retardation, speech defects, epileptic type convulsions, pigmentary degenerative changes of the chorioretina, dental osseous dysplasias, ocular hypertelorism, dermatolyphic anomalies and defective sweating have also been reported.[3] The primary biochemical defect of Sjogren Larsson Syndrome is still unknown. Altered lipid metabolism has been demonstrated in certain inherited forms of ichthyosis such as X. linked ichthysosis,neural lipid storage disease, multiple sulfatase deficiency and Refsum disease. In SLS also it has been proposed that their is impaired fatty acid oxidation in cultured fibroblasts due to deficient fatty alcohol nictoimamide adenine dinuoeotide oxidoreductase activity.[4] The impaired fatty acid oxidation in SLS has further been supported by case reports from Hoft et al and Christian G J P Harpey who have demonstrated that the ichthyosiform skin condition in SLS improves if all the lipids in diet are replaced by medium chain triglycerides.[5,6] There is no specific treatment of the disease, the skin problem is less disabling than the neurologic defects and can best be managed by lubrication. Life expectancy is about 50% of normal and despite mental retardation these patients are good natured and sociable.

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