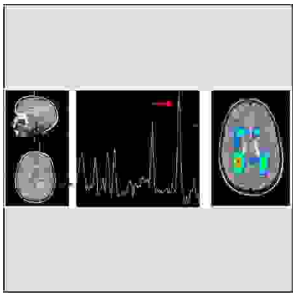
CASE REPORT PROF-968

SJOGREN-LARSSON SYNDROME



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ABSTRACT... We present a case report of two brothers suffering from Sjogren-Larsson syndrome, who were born to consanguineous parents. Sjogren-Larsson syndrome is one of the congenital icthyoses with an autosomal recessive inheritance[1]. It is characterized by the combination of congenital ichthyosis with spastic diplegia, moderate mental retardation and retinopathy.[2,3,16]. Defects in essential fatty acid metabolism have been attributed to as the cause[4,8,13,15].

CASE REPORT

Two brothers one aged 20 years and the other 18 who were amongst four siblings born to consanguineous parents, presented with the complaints of itchy skin lesions since birth and an inability to walk properly since the last 15 years. Both patients were born with normal skin at birth but developed their skin lesions at the age of 06 months. The lesions were present over the neck and both elbow and knee flexures. The lesions were intensely itchy and aggravated every winter. The elder patient had difficulty in walking since the age of 06 years and the younger one since birth.

There was no history of blistering, visual, auditory, olfactory or ejaculatory complaints. The family history revealed a female paternal first cousin also suffering from the same disease. On systemic examination, the musculoskeletal system revealed a decrease in the bulk of upper and lower limbs with an increased tone. Power was found to be 4/5, reflexes hyperactive and ankle clonus present. There was no bony deformity. A mini mental scale examination revealed mild mental retardation. Fundoscopy revealed a bilaterally absent foveal reflex and parafoveal dots. Both testes were normal in size.

On dermatological examination, the neck, elbow and knee flexures and the dorsa of both hands were found to have hyperkeratosis, accentuation of flexural creases and generalized mild scaling with adherent white scales. A skin biopsy for histo-pathology revealed epidermal hyperplasia, orthohyperkeratosis and hypergranulosis. The treatment given to the patient comprised of tab Acitretin 25mg once daily, 10% urea cream and physiotherapy which resulted in an increase in the power of the affected limbs and an almost complete clearance of the skin lesions.

DISCUSSION

Sjogren-Larsson syndrome is a disorder of autosomal recessive inheritance and therefore commoner in consanguineous marriages[1]. It is said to result from a deficiency of the enzyme fatty alcohol. NAD oxidoreductase (FAO) in leukocytes and fibroblasts resulting in defective essential fatty acid metabolism[4-8,13-15.] Congenital ichthyosis with spastic paraplegia, mental

retardation and retinal changes should lead one to suspect Sjogren-Larsson syndrome[2,16]. Patients usually have a dry and mildly erythrodermic skin at birth. Scaling develops within the first three months of life. Lichenification in the flexures, neck and peri-umbilical folds is seen. It develops, at times, as early as the first year of life. A cyclical accumulation and shedding of scales is seen especially affecting the face and limbs.

Non-progressive spastic paraplegia with upper motor neuron signs affecting the legs, and rarely arms, manifests itself in early infancy[9]. Patients are therefore, crutch-ridden from early on. This results in an altered posture and subsequent skeletal defects. Patients are often noted to have a cheerful disposition despite their physical disability along with, a mild to moderate mental retardation.

They exhibit absolutely normal vision despite the absence of the foveal reflex, the presence of foveal and para-foveal dots[10]. and a decreased macular melanin[11]. Skin biopsy for histopathology reveals orthohyperkeratosis, acanthosis and papillomatosis. A biochemical assay of fibroblasts or leukocytes reveals a deficiency of the enzyme fatty alcohol: NAD oxidoreductase (FAO)[4-8,13-15].

IN OUR CASE

* The onset of scaling was at age 06 months, whereas normally it is reported to be at 03 months of age.
* There was no history of collodion baby or erythroderma at birth[12].
* There was no cyclical accumulation and shedding of scales on the limbs or face.
* The skin on the face and periumblical folds, sites usually involved, was spared.
* The older patient did not develop spastic paraplegia of the lower limbs till the age of 06 years whereas its onset is usually in infancy.

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