Original Research Paper

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Dermatology

A RARE CASE OF SYNDROMIC ICHTHYOSIS : SJOGREN LARSSON SYNDROME : A CASE REPORT

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ABSTRACT A 15-year-old male patient presented with skin lesions associated with scaling over bilateral upper limbs, bilateral lower limbs, trunk since birth associated with severe itching. He had difficulty in walking and standing since the past 1 year and 6 months respectively. He was diagnosed as Global Developmental Delay with Intellectual Disability with history of seizure disorder in the past. There was diffuse skin thickening with prominent skin markings with excoriation marks over bilateral upper and lower limbs, and trunk. Biopsy revealed marked hyperkeratosis and papillomatous hyperplasia. Other significant findings included microcephaly, small arched eyebrows, flat nasal bridge, long philtrum, coarse facies, macroglossia, arachnodactyly, exaggerated deep tendon reflexes with extensor plantar reflex and speech difficulty. Differential diagnosis includes Sjogren Larsson Syndrome ,Trichothiodystrophy and Netherton Syndrome. Raised ALP, raised CPK, MRI of brain showing leukodystrophy, X ray both knees AP and lateral suggestive of osteopenia, IQ <30% were other significant investigation findings. Syndromic ichthyoses have phenotypes due to underlying genetic defects are seen not only in skin but also in other organs.[2] Congenital ichthyosis, spastic paralysis of the limbs(marked in the lower limbs, symmetrical) and mental weakness are three main signs of the Sjogren Larsson syndrome which were seen in our patient.[4] Gait disorder, history of convulsions, skin biopsy, MRI brain finds were concurrent with Sjogren Larsson syndrome. Speech difficulty , severe pruritis contribute towards our clinical diagnosis. Counselling of parents is of utmost importance.

KEYWORDS : Sjogren Larsson syndrome, congenital ichthyosis, Intellectual Disability, seizure disorder, hyperkeratosis, leukodystrophy

INTRODUCTION:

Ichthyosis is a heterogeneous Mendelian disorder of cornification.[1] Syndromic ichthyoses have phenotypes due to genetic defects which are not only seen in the skin but also in other organs.[2] Sjogren Larsson Syndrome is a rare autosomal recessive disease.[3] Fatty aldehyde dehydrogenase (FALDH) activity is reduced in fibroblasts cultured from the skin of these patients.[4] Although these syndromes are very rare, it is imperative to know the molecular genetics and pathomechanisms to establish an effective therapy and appropriate genetic counselling methods[4]

Case Report:

A 15-year-old male patient presented with skin lesions associated with scaling over bilateral upper limbs, bilateral lower limbs, trunk since birth associated with severe itching. He had difficulty in walking and standing since the past 1 year and 6 months respectively . No history of collodion membrane at birth. He was diagnosed as Global Developmental Delay with Intellectual Disability with history of seizure disorder in the past. History of 3rd degree consanguineous marriage in parents was present.

On Examination there was diffuse skin thickening with prominent skin markings with excoriation marks over bilateral upper limbs, bilateral lower limbs, trunk. Exfoliation was present over dorsal aspect of bilateral hands. Other findings included microcephaly, small arched eyebrows, flat nasal bridge, long philtrum, coarse facies, macroglossia, arachnodactyly , bilateral grade 2 tonsillar hypertrophy, Gowers sign positive suggestive of proximal myopathy, blepharitis in bilateral eyes, bilateral tibial swellings on medial side of bilateral knees, exaggerated deep tendon reflexes with extensor plantar reflex with speech difficulty and no hypogonadism.

The differential diagnosis included Sjogren Larsson Syndrome , Trichothiodystrophy and Netherton Syndrome.

Investigations- Hb- 13.10, TLC- 7890 with normal eosinophil count, Platelet count – 367000/mm3 , RBS -130, RFT – WNL, **LFT (raised ALP 1751 U/L), raised CPK (347 U/L)** . ECG ,chest X ray, Hair microscopy was normal. In Skin Biopsy, epidermis shows marked **hyperkeratosis** and **papillomatous hyperplasia**. The pigmentation in the basal layer was increased . The dermis had dilated capillaries and only sparse perivascular lymphocytic infiltrate. MRI of brain showed **leukodystrophy** . MRI whole spine screening was normal . Awake EEG showed normal background activity for age with focal discharges. X ray both knees AP and lateral were suggestive of osteopenia, bowing of femur, metaphyseal flaring, lucency and irregularity suggestive of metabolic bone disease or skeletal dystrophy. Audiometry was normal. **IQ was <30%.**

Taking into consideration the clinical features and laboratory investigations the patient was diagnosed as Sjogren Larsson syndrome.

DISCUSSION:

Congenital ichthyosis, spastic paralysis of the limbs(marked in the lower limbs, symmetrical) and mental weakness as three main signs of the **Sjogren Larsson syndrome,** which were seen in our patient.[4] Gait disorder is often observed in this syndrome which were also present in our patient. Convulsion is noted in approximately 40% of these patients [4]episodes of which were seen in our patient in the past. Speech difficulty , severe pruritis contribute towards our clinical diagnosis. Skin biopsy and MRI brain findings are concurrent with the syndrome. Counselling of parents is of utmost importance.

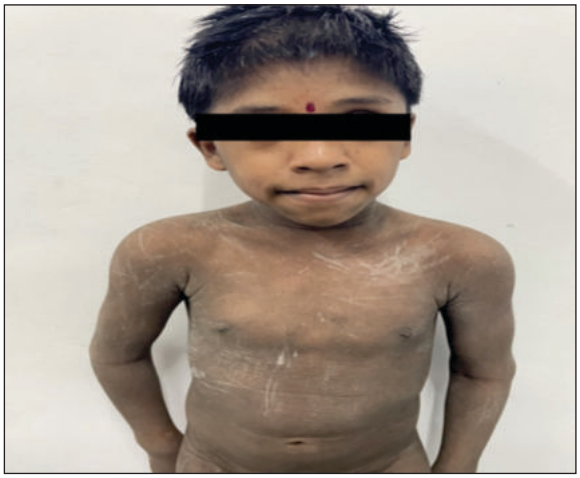


Image 1- Small Arched Eyebrows, Flat Nasal Bridge, Long Philtrum, Coarse Facies, Macroglossia

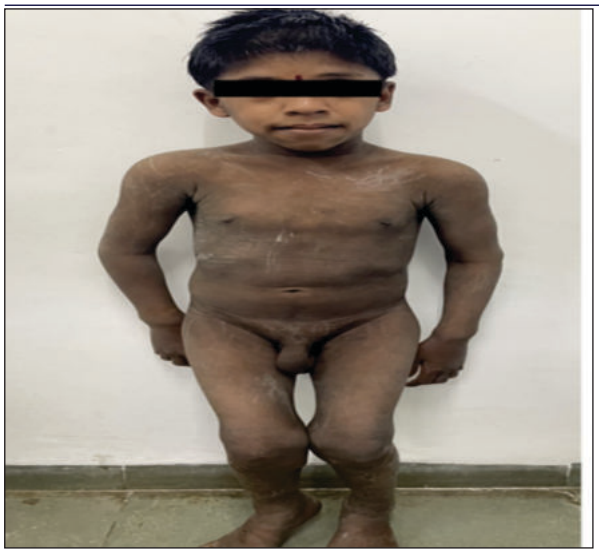


Image 2 – Tibial Swellings On Medial Aspect Of Bilateral Knees.

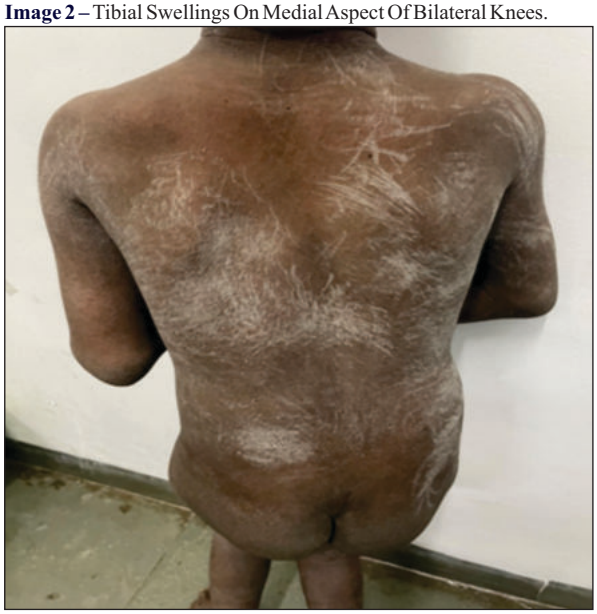


Image 3- Multiple Excoriation Marks Seen Over Back.

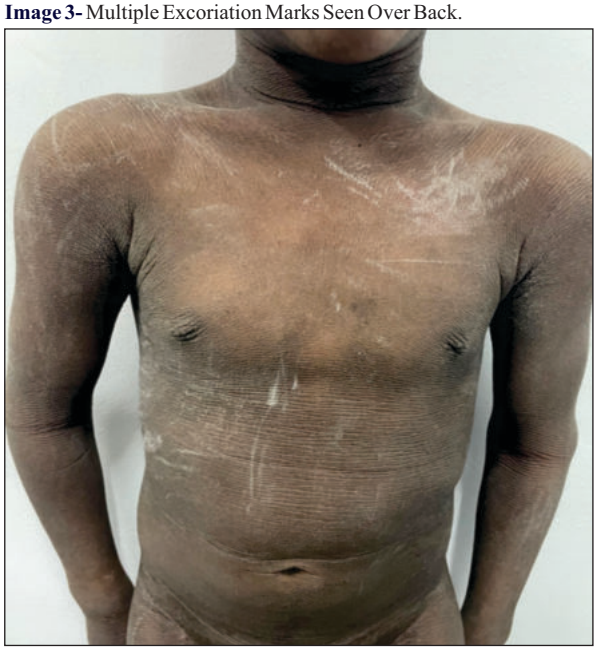


Image 4 – Diffuse Skin Thickening With Prominent Skin Markings, Multiple Excoriation Marks Are Seen.

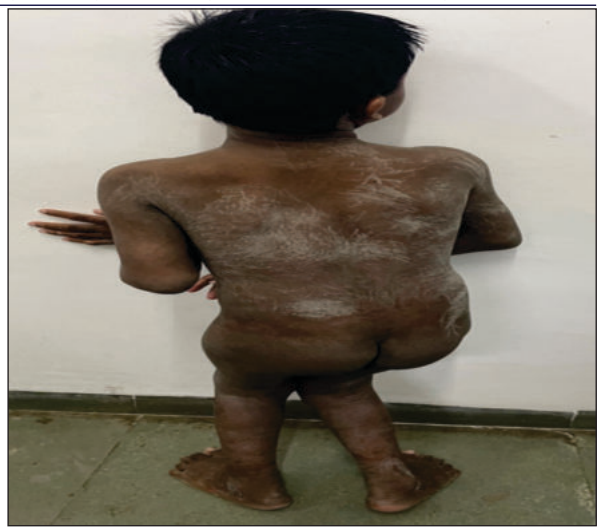


Image 5- Unable To Stand Erect Wall Support Taken To Stand Due To Proximal Myopathy.

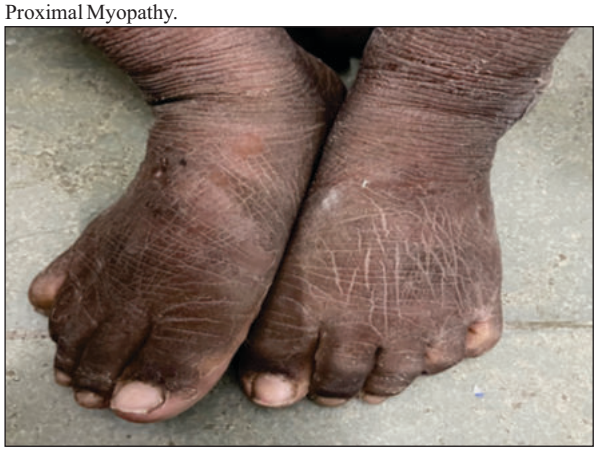


Image 6- Prominent Skin Markings With Keratotic Lichenification .

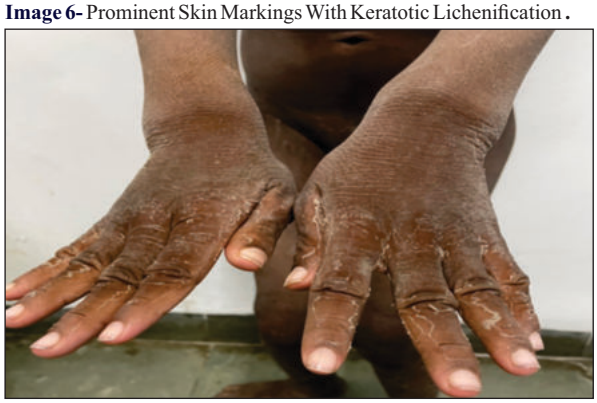


Image 7-Arachnodactyly With Exfoliation Over Dorsum Of Hands.

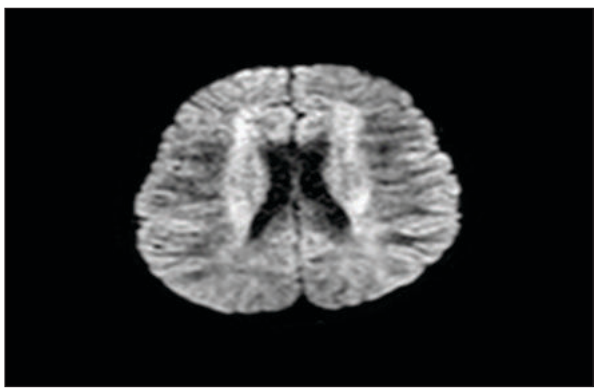


Image 8- MRI Brain Showed Hyperintensity Involving Frontal And High Parietal Periventricular White Matter And Deep White Matter Of

Bilateral Fronto-parietal Lobes Suggsestive Of Metabolic Etiology Likely Leukodystrophy.

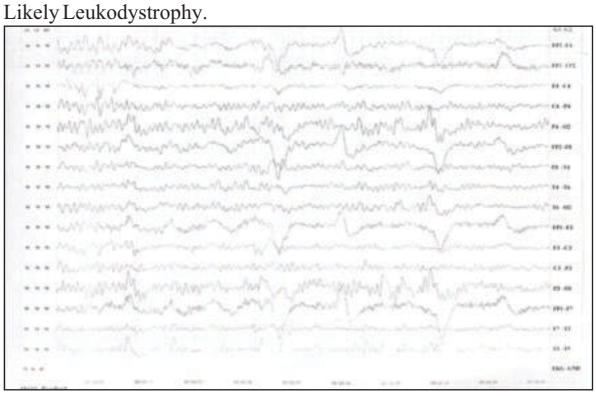


Image 9- Awake EEG Showed Normal Background Activity For Age With Focal Discharges.

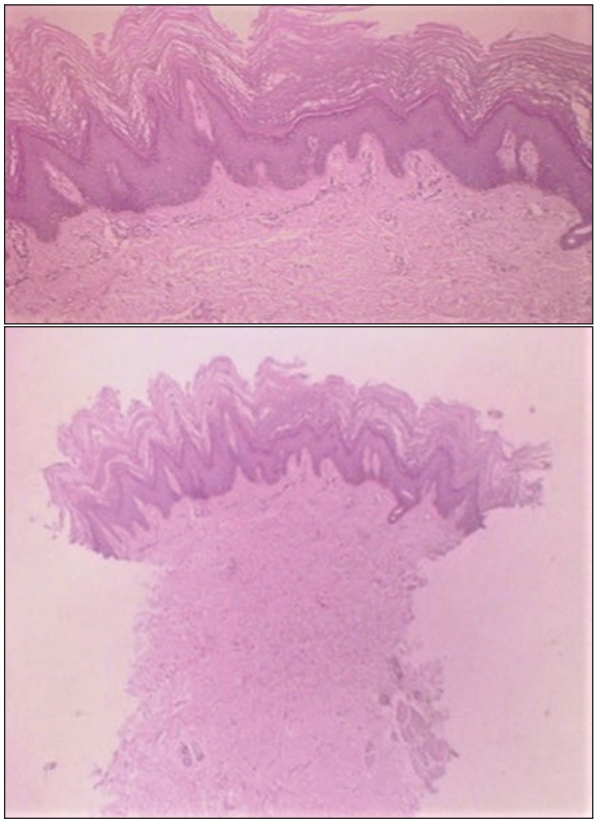
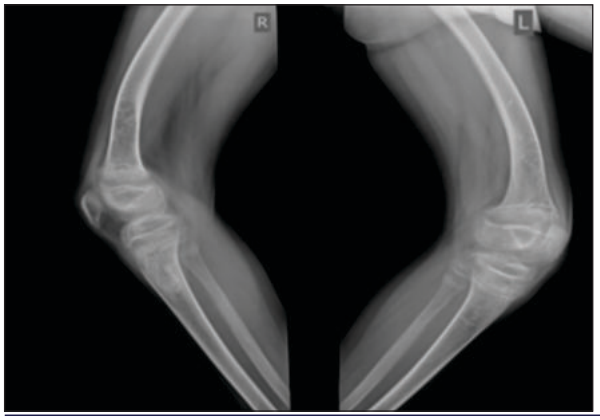


Image 10 And Image 11-
Histopathology :
The epidermis shows marked hyperkeratosis and papillomatous hyperplasia. The pigmentation in the basal layer is increased . The dermis has dilated capillaries and only sparse perivascular lymphocytic infiltrate.



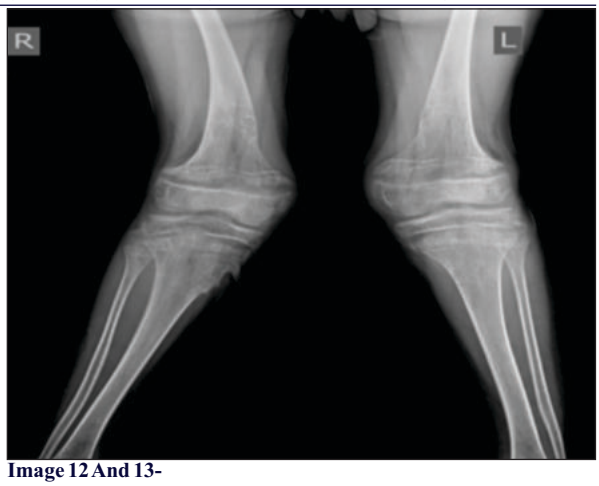


Image 12 And 13-
X-RAY BOTH KNEE AP/LATERAL –
Metaphysial flaring, lucency, irregularity is noted, osteopenia , bowing of femur is noted suggestive of metabolic bone disease/skeletal dystrophy.

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