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J Clin Exp Dermatol Res 2018, Volume 9
DOI: 10.4172/2155-9554-C3-075

Sjogren-Larsson syndrome: A case report

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Sjogren-Larsson syndrome is an inborn lipid metabolism error inherited in autosomal recessive pattern presenting as congenital ichthyosis, spastic diplegia/tetraplegia, and mental retardation caused by deficiency of enzyme fatty aldehyde dehydrogenase[FALDH]due to mutations in ALDH3A2 gene. A 2 years old male child presented with complaints of dry scaly and thickened skin, with delayed development. On detailed history child attained neck holding at 7 months and sitting without support at 1and ½ year with tightness and stiffness in lower limbs and episodes of seizures. No history of itching, consanguinity of marriage and preterm birth. Cutaneous examination revealed dirty velvety plaques involving bilateral axillae, neck, groin and abdomen and plate like scales adherent at center and free margins involving bilateral legs with generalized dryness over whole body. Neurological examination revealed global developmental delay with mental and growth retardation with increased ton in both lover limbs. Sjogren-Larsson syndrome is a rare syndrome which classically presents as developmental delay, mental retardation, speech difficulties, seizures, spastic diplegia/tetraplegia, glistening white dots on retina and skin involvement in the form of pruritus with ichthyosis with generalized hyperkeratosis. We are presenting the case of Sjogren-Larsson syndrome for its rarity and classical clinical findings.

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