GENETICS AND GENODERMATOSES

SOLVING THE JIGSAW PUZZLE OF A RARE NEUROICHTHYOTIC SYNDROME

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Background: Neuroichthyotic Syndrome encompass a wide variety of clinical syndromes each with its constellation of mucocutaneous, neurological and other systemic symptoms. However, overlapping clinical features occasionally necessitate genetic studies to arrive at a complete diagnosis.

Observation: 20 months old female child born of a third degree consanguineous marriage presented with fever, cold and cough since 5 days and peeling of the skin since childhood. Gross developmental delay was present with no h/o epilepsy. Past H/o of recurrent episodes of pneumonia present. Cutaneous examination showed diffuse scaling over the body with large plate like scales. Systemic examination revealed b/l rhonchi. Motor examination revealed increased tone in all four limbs with reduced power <3/5 and b/l extensor plantar reflexes. Cranial nerve and sensory examination were normal. Fundoscopic examination showed multiple pigmented dots. BERA studies showed moderately severe hearing loss in bilateral ears. Dental examination showed enamel hypoplasia. Provisional diagnosis of Sjogren Larsson Syndrome was kept. Routine investigations revealed anaemia. MRI Brain with Spectroscopy revealed paucity of white matter in B/L occipital region and hypoplastic corpus callosum with no lipid peak. Genetic Study revealed homozygous nonsense variation in exon 2 of the SNAP29 gene (Synaptosomal-associated protein 29). Thus after correlating clinical, radiological examination and genetic studies, a final diagnosis of CEDNIK Syndrome was made.

Key message: CEDNIK syndrome is caused by mutations in SNAP29 that leads to decreased soluble n-ethylmaleimide sensitive factor receptor (SNARE) protein SNAP29 that mediates endocytic vesicle trafficking between organelles and plasma membrane Affected patients are managed by a multidisciplinary team of neurologists, ophthalmologists, dermatologists, and rehabilitation specialists, among others. This case is being presented to emphasize the importance of early diagnosis and timely management of such a rare disorder to improve the overall development of the child.