GENETICS AND GENODERMATOSES

SISTERS WITH NO PAIN, NO TEARS: A REPORT OF A NEW VARIANT OF HSAN (TYPE IX) CAUSED BY A NOVEL SCN11A MUTATION

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Background: Hereditary Sensory Autonomic Neuropathy (HSAN), is a group of very rare inherited disorders with multiple gene mutations and characterized by sensory and autonomic dysfunction with various associated features. Classification is based on clinical manifestations and genetic mutations. Here we present a case study of an entirely new variant of HSAN.

Observation: Two sisters aged 6 and 3 years, presented with history of absence of sweating since birth associated with recurrent fever, redness of skin when exposed to heat. They also had history of absence of pain sensation, dryness of mouth and absence of tears while crying. Neurological examination revealed loss of pain and temperature sensations all over the body. EMG-NCV showed absent sympathetic skin responses (SSR) on bilateral hands and feet suggestive of sensory/ autonomic neuropathy. Skin biopsy from palms showed no sweat glands with few atrophic ducts. Oro-dental, ophthalmologic, psychiatric and ENT evaluation revealed dental caries, positive Schirmer's test and no mental retardation or deafness. ANA, anti-SSA/SSB, anti-tissue transglutaminase, anti-microsomal and anti-TPO antidboides were negative. Genetic study of both sibling revealed novel missense mutation in exon of SCN11A gene on chromosome 3 which confirmed the diagnosis of a new variant of HSAN. Genetic study of father also revealed same missense mutation of SCN11A gene however he is totally asymptomatic. Both patients were managed well with symptomatic treatment along with patient education and counselling.

Key message: SCN11A gene mutation has been classically linked to HSAN type 7 which is characterized by hyperhidrosis, sensory, autonomic neuropathy and self-mutilation as against anhidrosis and absence of self-mutilation in our case with additional features of non-Sjŏgren sicca syndrome. Hence we propose this case to be a new variant of HSAN, HSAN typeIX. The case is being presented due to its extreme rarity and emergence of a new genetic variant.