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The painless eye: Neurotrophic keratitis in a child suffering from hereditary sensory autonomic neuropathy type IV

Aditya Sethi, Srikanth Ramasubramanian and **Meenakshi Swaminathan**
[1]Sankara Nethralaya, India
[2]Arunodaya Deseret Eye Hospital, India

Hereditary sensory autonomic neuropathy (HSAN) is a group of inherited disorders (total five types) that are associated with sensory dysfunction and varying degrees of autonomic dysfunction. HSAN type IV (or HSAN 4) is a rare genetic disorder inherited in an autosomal recessive manner, caused by mutations in the NTRK1 gene. It begins in infancy and is characterized by an inability to feel pain and an inability to sweat (anhidrosis). Anhidrosis can cause recurrent episodes of fever and high body temperature. Also these affected individuals cannot distinguish in temperature. An inability to feel pain can lead to unintentional self-mutation, repeated fractures, and joint damage. The sensory loss in individuals with HSAN IV is due to abnormal functioning of the sensory nerves that control responses to pain and temperature. HSAN IV is caused by mutations in the NTRK1 gene. Eye abnormalities specifically neurotrophic keratitis, a degenerative disease of corneal epithelium resulting due to impaired corneal innervation, in this case due to HSAN type IV. We report a case of this very rare genetic disease associated with neurotrophic keratitis, in a three year old girl child, born to a family in north India having clinical features of insensitivity to pain and temperature, self-mutilating behavior with multiple recurrent oral ulcers, nasal bleeds, and multiple trophic ulcers over joints and anhidrosis.

adityasethi1@gmail.com