Hereditary Sensory and Autonomic Neuropathy Type VIII: Congenital Insensitivity to Pain with Anhidrosis

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Hereditary sensory autonomic neuropathy type VIII (HSAN VIII) is a rare genetic disorder that characterized by insensitivity to pain, anhidrosis, self-mutilation, and absence of corneal reflex. We here report a 2-year-old female child presented with corneal opacity and was diagnosed HSAN VIII by its typical clinical features confirmed by gene analysis revealing the presence of homozygous mutation in the PRDM12 gene.

Case Summary

A 2-year-old female child was presented with complaint of corneal opacity in both eyes noticed 4 months ago [Figure 1a]. The mother also complained of missing both upper and lower front teeth [Figure 1b]. Frequent history of trauma either unknown or self-mutilated without any sign of discomfort was revealed. No lacrimation during cry, lack of sweating with dry skin, and recurrent episodes of unexplained fever were also noticed [Figure 1c]. She was the only child of consanguineous marriage with no similar disease in the family. General examination revealed evidence of mutilation such as multiple scars, well-defined nontender ulcers over face and limbs, and shortening of distal phalanges [Figure 1d–f]. Ocular examination showed absence of corneal sensation with corneal scar. Neurologic examination found generalized absence of pain and temperature perception with loss of reflexes. Gene analysis revealed the presence of homozygous mutation in the PRDM12 gene confirming the diagnosis of HSAN VIII.

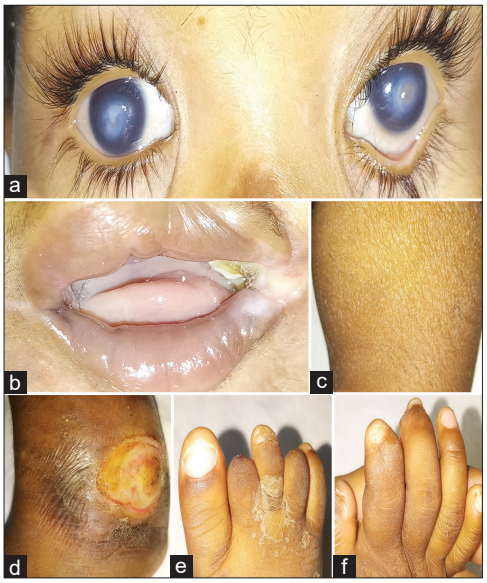

Figure 1: Composite picture depicting various manifestations of a child with hereditary sensory and autonomic neuropathy type VIII: (a) bilateral corneal scarring, (b) missing upper and lower front teeth, (c) dry skin, (d) painless ulcer on the right ankle, and (e and f) mutilated distal phalanges of hand and feet

HSAN type VIII comprises a group of genetic disorders involving sensory and autonomic dysfunctions, described as a rare autosomal recessive disorder caused by homozygous mutation in the PRDM12 gene on chromosome 9q34.[1,2] It is characterized by insensitivity to pain and thermal stimuli, altered sweat and tear formation, self-mutilation behavior, absence of the corneal reflex resulting in corneal scarring and presence of repeated infections of the skin and bone that are also common [Table 1].[3,4] The diagnosis of HSAN is challenging due to its rarity and lack of simple diagnostic tests. The affected persons are vulnerable to severe complications and self-mutilation. Therefore, early identification and appropriate management are necessary to avoid severe injuries and its consequences.

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Table 1: Reported cases of hereditary sensory autonomic neuropathy type VIII (HSAN VIII)

Year of publication	Authors	Country	Age at presentation	Sex	Presenting complaint	Parental consanguinity	HSAN (type/gene)
2016	Zhang *et al*.[2]	United Kingdom	NA	Male: 4 Female: 1	Insensitivity to pain	Yes	VIII/PRDM12
2017	Elhennawy K *et al*.[3]	Germany	8 months	Male	Premature loss of incisors	Yes	VIII/PRDM12
2018	Gaur *et al*.[4]	India	1 year	Male	Corneal opacity	No	VIII/PRDM12
2020	Hasanuddin *et al*.[5]	India	7 months	Female	Ulcerations on the tongue	No	VIII/PRDM12
Present study	Kusumesh *et al*.	India	2 years	Female	Corneal opacity	Yes	VIII/PRDM12

Conflicts of interest

There are no conflicts of interest.

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