Hereditary Sensory and Autonomic Neuropathy Type V: A Rare Case Report

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Abstract
Hereditary sensory and autonomic neuropathy type V (HSAN V) is a rare disorder that mainly affects the sensory neurons, which conduct the transmission of sensations such as pain, temperature, and touch. It is an autosomal recessive disorder characterized by a lack of deep pain perception. Traumatic injury and self-mutilation are persistent features of this disorder. The challenge in dentistry is to manage the self-mutilation behavior, avoiding severe damage, especially oral structures, hands, and fingers. We report a case of an 8-year-old female child with HSAN type V, having the typical clinical manifestations of pain insensitivity causing self-mutilation. We discuss the comprehensive dental treatment of the patient with HSAN V.

Keywords: Autosomal recessive, hereditary sensory and autonomic neuropathy, pain, self-mutilation

Introduction
Pain is an inborn reflex and is highly protective in nature. It guards the tissues of the body against the noxious stimuli and possible tissue destruction. It interferes with patient's quality of life and general functioning. The International Association for the Study of Pain, defines pain as "an unpleasant sensory and emotional experience associated with actual or potential tissue damage, or described in terms of such damage".[1]

HSANs are neurodegenerative disorders of the peripheral nervous system involving a different degree of sensory and autonomic disturbances, characterized by, severe ulcero-mutilations, first described in 1852, by Nelaton.[2] Based on age of onset, mode of inheritance, predominant clinical features, HSANs are classified as type I-V; recently HSAN type VI has been added to the list.[3] HSAN type V is the rarest of the five caused due to mutation in the NGFB gene, located on chromosome 1(1p13.2-p.2), coding for Nerve Growth Factor B (NGFB), that is important in the development and survival of nerve cells (neurons), including sensory neurons.[4] It is characterized by marked absence of pain and temperature sensations with intact pressure proprioception, pressure and unimpaired touch, normal intellectual abilities associated with decreased or normal sweating.[5]

Case Presentation
An 8-year-old girl reported to Pedodontics and Preventive Dentistry Department, with a chief complaint of an ulcer on the lower lip and multiple carious teeth. Family history revealed she is the second daugh-

How to cite this article: Sasalawad S, Srinath SK, Hoddetty Shivarama S. Hereditary Sensory and Autonomic Neuropathy Type V: A Rare Case Report. J Pediatr Dent 2020;6(2):53-56

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ter of the second-degree consanguineous marriage, and
the first child is normal. There was no positive family
history with HSAN. The patient’s mother noticed
delayed milestones by 3-6 months and didn't respond to
painful stimuli like vaccination and injury. At the age of
8 months, she had a total absence of blink reflex and
corneal sensation bilaterally, diagnosed as congenital
corneal anesthesia. Parents gave a history of biting lips
and tongue after the eruption of teeth.

General examination revealed the patient was pallor,
eyes showed absence of blink and corneal reflex and bilat-
eral corneal anesthesia. Ophthalmoscopic examination
showed bilateral corneal opacities with only 10% of vision
(Fig. 1a). Bilateral distal resorption of the index finger
and right great toe with spoon-shaped nails was evident
(Fig. 1b). Scars and burns over the extensor aspect of
knees, thighs, and feet were seen (Fig. 1c). Chromosome
analysis was done for karyotype results, which showed
female karyotype (Fig. 2a). Electroneuromyography
(ENMG) showed normal study.

Extra-oral examination (Fig. 2b) revealed normal
upper lip with a mild dryness and angular cheilitis with
limited mouth opening. Thin and fibrosed lower lip
with traumatic ulcer due to constant biting. Complete
loss of lower teeth caused loss of support to lower lip
resulted in lip trap. Intraoral examination (Figs. 3a and
3b) revealed complete resection of the tongue and inju-
ries over the tongue due to constant biting. Hard tissue
examination revealed presence of 16, 55, 14, 52, 11, 21,
22, 24 and 26. Dental caries with respect to 55, 11, 21,
and root stump in relation to 52. The mandibular ridge
was totally resorbed and completely edentulous. Parents
gave a history of autoextraction of the deciduous teeth.
Atrophy of the lower alveolar bone and raw bony sur-
face seen in the lower edentulous ridge. After the com-
plete examination patient was advised for orthopanto-
mogram radiograph (OPG), revealed resorbed lower
alveolar bone, embedded bilateral lower first molars
(Fig. 3c). Based on clinical findings, a histological fea-
ture, sensory and autonomic dysfunction, and karyo-
type, the case was diagnosed with HSAN Type V. The
parents were explained about the possible treatment
options and importance of soft tissues (tongue).

Figure 1. (a) General physical examination, (b) Bilateral distal
resorption of the index finger, (c) Scars and burns over the foot

Figure 2. (a) Female karyotype, (b) Extra-oral examination shows ulceration of the lower lip
Alginate impression of the lower lip was made, and the cast was poured with dental stone, splint for the lower lip was fabricated with biocryl sheet using biostar forming machine, advised to wear the splint till healing of the traumatic ulcer on the lip (Figs. 4a and 4b). After complete cure of the ulcer, bite guard with palatal covering was preferred, and the patient was advised to wear the bite guard and asked to remove it for 1-2 hours interval (Figs. 4c and 4d).

**Discussion**

Hereditary sensory, autonomic neuropathies (HSAN) are a clinically and genetically heterogeneous group of peripheral nervous system disorders. In 1983, Dyck et al recommended the diagnostic criteria for the disease based on histological, microscopic, and morphometric methods for assessing nerve fiber pathology.[6] Although the prevalence of HSAN has not been well established; they appear to be very rare, except in few countries.

Type I: Sensory radicular neuropathy.[4]

Type II: Congenital sensory neuropathy.[3-7]

Type III: Familial dysautonomia.[3-7]

Type IV: Most prevalent type is known as congenital insensitivity to pain with anhidrosis (CIPA).[8]

HSAN V: Congenital indifference to pain. It is an autosomal recessive condition similar to HSAN type IV. It is caused due to mutation in the NGF beta gene (NGFB gene at locus 1p13.2-p.2). NGF-β mutation results in a substitution of tryptophan for arginine on position 211 in a highly conserved region of the protein.[4]

Greater degree of anhidrosis and mental retardation is seen in HSAN type IV, compared HSAN type V, in which, patients have normal intelligence and motor development. In HSAN type V, there is severe reduction of unmyelinated nerve fibers and a moderate loss of thin myelinated fibers, where in HSAN type IV normal large myelinated nerve fibers with reduced small myelinated fibers.

Clinical features of HSAN V are insensitivity to pain, reduced thermal sensation, and severe loss of deep pain perception, painless fractures and joint deformities. Self-mutilation is an invariable feature of this disorder most often involving the teeth, lips, tongue, ears, eyes, nose and fingers. Oral management of these patients should focus on preventing injury to the tongue, lips, teeth, and oral mucosa. Children will require grinding of the teeth or extraction of all teeth or bite guard to prevent self-mutilation injury to the tongue and lips.[9]

**Dental management**

There is no definitive treatment for the HSAN type V, management is supportive. As self-mutilation is invariably a constant feature, oral management of these patients should focus on preventing injury to the tongue, lips, oral mucosa, and teeth. Selective grinding of sharp edges of teeth and extraction of offending teeth. Use of mouth guards or tongue guards to prevent injury to soft tissues.[10] The use of intra oral appliances may
be difficult in some patients as the self-mutilation would have begun with the eruption of primary incisors. The removal of such teeth may be unavoidable in some cases. Prevention of dental caries is crucial to intercept the spread of infection. Thus, the dental team should therefore continuously monitor these patients throughout their lifetime.[8]

Conclusion

Congenital indifference to pain is a very rare disorder. These patients suffer, even without pain. Genetic counseling and prenatal diagnosis should be done to prevent the birth of the affected child. Early diagnosis and education to mitigate the trauma to oral tissues and proper care of skin, eye, and dental tissues. The challenge to the dentist is to diagnose and treat appropriately the local infection to reduce the life-threatening complications in later life.

Financial Disclosure: Nil.
Conflict of Interest: None declared.

References


