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Oral Manifstations of Hereditary Sensory Autonomic Neuropathy Type IV- Case Report

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Abstract

Hereditary sensory and autonomic neuropathy type IV (HSAN-IV) is an autosomal recessive rare disorder characterized by lack of maturation of small myelinated and unmyelinated sympathetic nerves absence resulting in pain and temperature insensitivity, hypotonia, developmental delay, mental retardation and anhidrosis due to lack of innervations of sweat glands resulting in recurrent fever episodes. Self-mutilations, osteomyelitis, deformities of joints, multiple fractures and corneal scarring are the most frequent complications. We report a case of a 5-year-old girl from cousin parents with HSAN type IV

Keywords: Hereditary sensory autonomic neuropathy, Pain insensitivity, Self-mutilations, Congenital

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Introduction

Hereditary Sensory Autonomic Neuropathies (HSAN) are a group of hereditary rare neuropathies described in 1932 by Dearborn as the "Congenital pure analgesia"[1]. It affects the automatic and sensory nervous system. HSANs are categorized into five types according to the age of onset, inheritance and clinical features.

HSAN-IV, also called as congenital insensitivity to pain with anhidrosis (CIPA) or Nishida syndrome[2] is the rarest form with few hundred cases have been described[3] and it is very difficult to diagnose and treat. The symptoms of the disease start from infancy and the patients do not have the ability of sensing different sensations such as pain and temperature with preservation of other tactile sensation, in addition patients also suffer from mental problems, anhydrosis and thermos regulation disorder[3-5].

The complications of this disease are mostly orthopedic, but the prognosis can be challenged and the expectancy of life is rarely reaches adulthood in this clinical state.

Early diagnosis should be made in early childhood with lesions of the extremities, stomatological involvement begins in the first year of life with the of first teeth appear, with lips and tongue biting, frequent bone fractures, and dislocation of joints[6-11] .

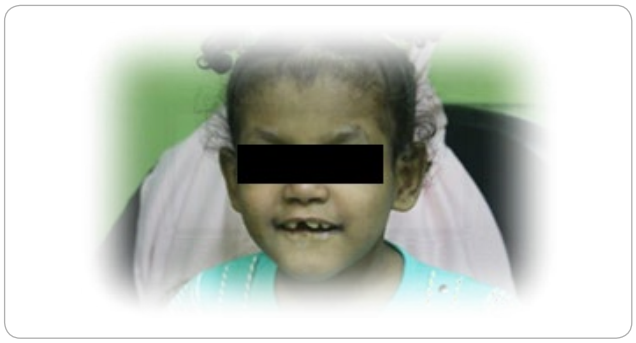
Case Report

A 5 years-old girl who was diagnosed with HSAN-IV at the age of 3 years and born to a healthy cousin parents parents, admitted to the hospital had a painless extra-oral swelling and loose teeth.

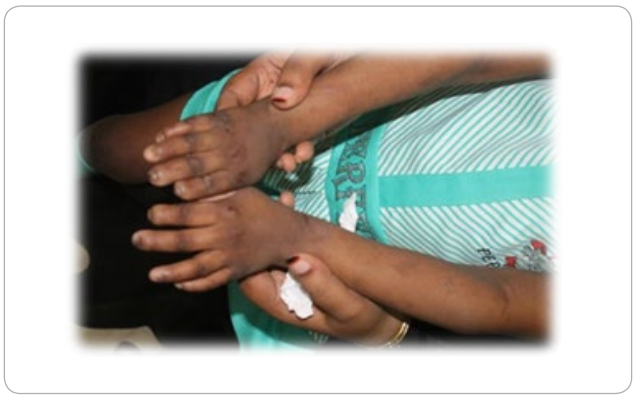
According to her mother, the girl never seemed to feel pain and there is a history of recurrent episodes of hyperpyrexia. The girl is hyperactive and had a multiple bone fractures with inadequate bone union.

On physical examination She was oriented, with normal muscle power and no deficit in the cranial nerve. There was generalized hypotonia.

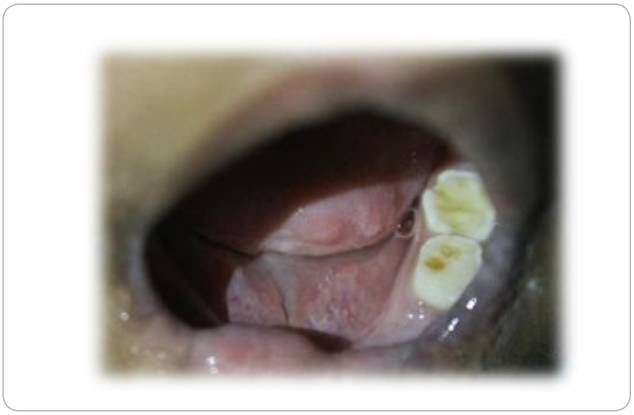
The girl's motor milestones were delayed though her mental development was appropriate for age.



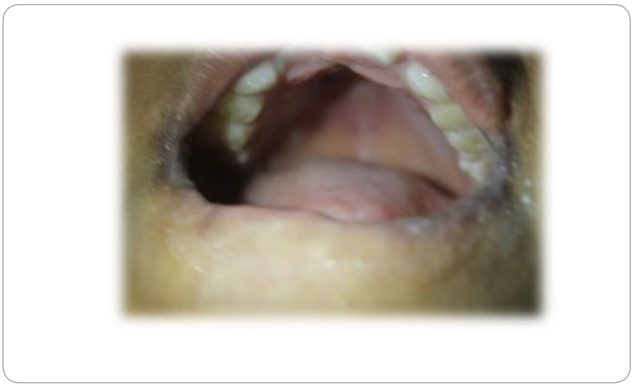
Skin, especially over the palms and soles was dry, and mildly hyperkeratotic. On sensory examination pain and temperature sensations were absent while the touch sensation was normal. On intra oral examination, the dentition was mixed -although the patient is 5 years old- with all the first permanent molars and permanent central incisors fully erupted



Teeth loss (upper right permanent central incisor, lower right and left first primary molars and lower right second primary molar) caused by the patient's manipulation herself.



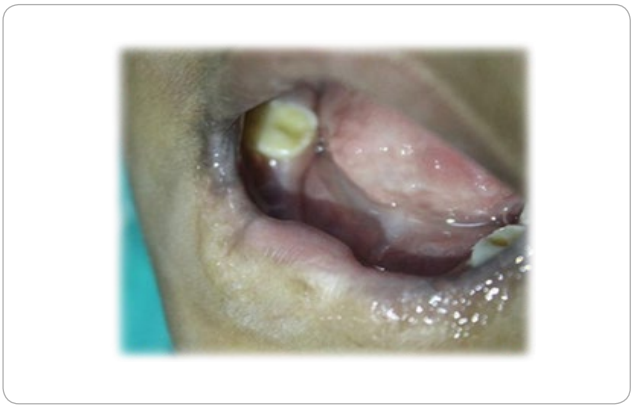
She had a remarkable tooth wear in the form of attrition due to bruxism. Her lips, tongue, fingers and toes were mutilated as a result of the repeated episodes of biting and chewing started soon after eruption of primary teeth



Investigations revealed normal hematological profile, liver and renal function tests, CT scan was done to rule out intracranial bleed and was normal.

Disscussion

HSAN is characterized by congenital pain insensitivity, temperature changes and disorders of autonomic nerve formation. It is caused by mutation in NTRK1, the neurotrophic tyrosine kinase receptor type 1 and TRKA gene. Mutations in the TRKA gene correlate with the symptoms in. This gene is located on the Chromosome 1q and contains 16 introns and 17 axons[5]. It was classified by Dyck and Ohta into five types. HSAN I is an autosomal dominantly inherited, symptoms start in the second decade or later. There is loss of temperature and pain sensation but tactile sensation preservation. Sural nerve biopsy shows more loss of unmyelinated fibers more than myelinated fibers. HSAN II is an autosomal recessive disorder; symptoms begin in infancy. There is generalized pan sensory loss.



Motor function is preserved but tendon reflexes of tendons, Autonomic disturbances included distal anhidrosis and bladder dysfunction. sural nerve biopsy shows loss of myelinated fibers. HSAN III is also an autosomal recessive. The clinical manifestations present at birth. Nerve biopsy shows reduced number of unmyelinated fibers.

HSAN IV is an autosomal recessive disorder associated with pyrexia, mental retardation and anhydrosis. Nerve biopsy reveals absent unmyelinated fibers.

HSAN V is inherited as autosomal recessive disorder with onset at birth and normal sweating.

Tendon reflexes and motor functions are normal. Sural nerve biopsy shows selective reduction in the number of smaller myelinated fibers[12].

Yagev et al[13] studied fifteen Bedouin children with anhydrosis with congenital pain insensitivity syndrome and found all them to have lack of corneal sensation. Ishii et al[6] reported a Japanese girl with high fever, pain insensitivity and anhydrosis who died at the age of twenty- one months. Rafael et al[5] described a nine-year-old girl case with pain insensitivity and the nerve biopsy suggested absence of small myelinated and unmyelinated nerve fibers. Courtney and freenberg described a patient with Hereditary sensory and autonomic neuropathy type IV but did not have development delay. Bonowsky et al[14] reported a one-year-old boy with congenital pain insensitivity and the diagnosis was confirmed by molecular analysis.

In HSAN IV, recurrent osteomyelitis, degenerative arthritis and amputation of fingers and toes, Charcot joints occur. We report a case presented with history of pain insensitivity, absence of sweating, corneal reflex absence, febrile seizures and signs of self-mutilation.

Oral and dental complications of these patients start with growing the teeth in the form of extracting the teeth and biting the lips.

Rehabilitation oral and dental tissues and training the patients and the use of dental prostheses are highlighted nowadays and can be very significant to these patients.

In our case, there were such behaviors, which had led to extraction of multiple teeth and mutation of the lip, tongue and fingers.

Conclusion

The disease is congenital and there is no specific cure for it but the early diagnosis is of paramount importance to prevent severe life threatening injuries

One of the essential factors of treatment remains reduction of complications through parent education, training, rehabilitation and supportive measures in addition, prescribing the necessary supportive tools are among the main priorities of treating those patients.

Hereditary Sensory and autonomic neuropathy type IV is a rare developmental disorder due to lack of sensory nerve fibers responsible for the sensation of pain and temperature.

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