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Case Report

Hereditary Sensory Neuropathy: A Case Report

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Abstract

Aim: Hereditary sensory and autonomic neuropathy (HSAN) is a rare syndrome of unknown etiology that develops in early childhood. Five different types of HSAN have been described. This syndrome is characterized by the absence of pain and self-mutilation. Patients start to traumatize themselves at as young as 2–3 years of age. Subsequently, ulcers and stress fractures develop on their fingers and toes. Oral findings in HSAN patients include oral lesions after repetitive trauma, oral mucosa and tongue scars, self-dental extractions, dental infections, and premature tooth loss.
Methodology: This case report presents the oral findings of a 4-year-old HSAN patient with premature tooth loss who visited Dicle University, Faculty of Dentistry, Department of Periodontology. The extraoral examination showed wounds around the nose and bruises and burns on the forearms and legs. The intraoral inspection showed traumatic lesions on the tongue, early loss of all of the lower jaw primary dentition, but the presence of upper jaw teeth.
Results: During follow-up, the eruption of the permanent right lowe r incisors and presence of mobility were observed. A partial child pros thesis for the lower jaw and protective plaque restoration constructio ns were planned. The patient is being followed regularly.
Conclusions: It is necessary to conduct regular checkups and to ensure good communication between the dentist and family in terms of ensuring oral hygiene and preventing the development of undesired complications.
Keywords: Neuropathy, hereditary sensory and autonomic neuropathy, HSAN, oral hygiene

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Introduction

Hereditary sensory neuropathy is a rare genetic disorder of unknown etiology (1). It usually becomes apparent after birth and is characterized by varying degrees of sensory loss (2). The most common symptoms are diminished or absent senses of pain, touch, and temperature in various parts of the body and extremities (3). The pathological basis of the disease involves a reduced small myelinated nerve fiber population due to premature cell death or growth deterioration during in utero. Although the etiology of the disease is not clearly understood, a genetic component frequently exists (3). Genetic tests are not useful for all hereditary sensory neuropathy disorders; in order to distinguish the types, other methods are required. Some of the types have specific neuropathological properties that can be identified from neural tissue biopsies, which

are invasive, and it is impossible to obtain tissue in all cases. Therefore, a careful evaluation of sensory and autonomic functions and a clinical diagnosis might be preferred (4).

The clinical signs of hereditary sensory neuropathy include a burning sensation in the extremities (including the hands), joint injuries, post-traumatic recurrent oral lesions (particularly of the tongue and lower lip), serious and frequently insidious infections such as osteomyelitis, oral mucosa scars and reduced mouth opening due to scarring from cheek biting, self-extraction, tooth infections, and early tooth loss (1-3). The oral lesions are usually the initial signs of the disease.

Hereditary sensory neuropathies are divided into five types.

Type 1: This type is inherited in an autosomal dominant pattern. The symptoms first appear in the second decade. There are often slow-healing, ulcerated lesions of the foot, especially the sole, characterized by necrosis. The foot ulcers result from continuous trauma to the insensitive skin. The sensory loss in the hands is minor or absent and ulcerations do not develop in the hands. Upper extremity complications are mild and delayed (1-3, 5, 6).

Type 2 (Morvan's syndrome): The disease is similar to type 1, but is autosomal recessive. The disorder particularly affects myelinated axons. Disease symptoms begin in infancy or childhood with the loss of all of the senses in the distal extremities. Unlike type 1, the upper and lower extremities are affected equally. There is sensory loss over a wide area, including the trunk and forehead, with severe dystrophic changes and anhidrosis, tonic pupils, and joint contractures in the extremities (1, 2, 5, 6).

Type 3 (familial dysautonomia or Riley-Day syndrome): It is autosomal recessive. Symptoms begin to appear in infancy. Hypotony begins from birth. Nutrition and functional disorders are apparent. There is reduced lacrimation and fluctuating fevers. The other properties are similar to type 2. Sensory loss is not a typical sign. The prognosis is poor. Recovery is observed in the patients who reach adulthood (1, 2, 5, 6).

Type IV (hereditary anhidrotic sensory neuropathy): This rare autosomal recessive neuropathy is characterized by mental retardation, insensitivity to pain and temperature, anhidrosis, and episodic fevers. The clinical signs begin at birth. The loss of temperature and pain sensation is significant. All cases are mentally retarded (1, 2, 5, 6).

Type V: The clinical features are similar to type 4; however, the patient's mental ability is normal.

All hereditary sensory neuropathies share the absence of pain sense and patients begin to injure themselves when they are 2~3 years old. Protecting these children from trauma is difficult (1).

This paper presents the oral findings of a patient diagnosed with hereditary sensory neuropathy.

Case Report

A 4-year-old girl was referred to the Department of Periodontology, Dicle University Faculty of Dentistry, with a diagnosis of hereditary sensory neuropathy and early tooth loss. The history identified the patient as the ninth child of the family; her mother and father were relatives and there were no ailments in other siblings. The child had no feeling of pain and constantly wounded or burnt herself.

The intra-oral examination showed ulceration of the front left half of the tongue, and the loss of all mandibular deciduous teeth, with the presence of maxillary deciduous teeth, and poor oral hygiene (Figs. 1–3).

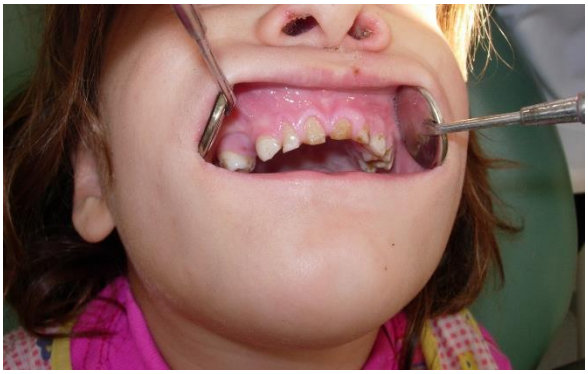

Figure 1. Maxillary deciduous teeth

Figure 2. Mandibular deciduous teeth

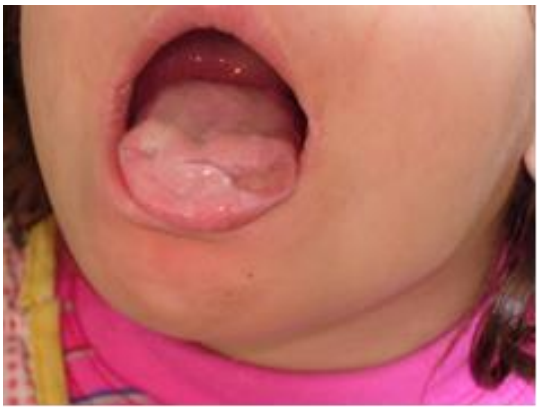

Figure 3. An ulceration in ½ front left section of the tongue.

The family reported that the wound to her tongue had existed for a long time and had not healed. The extra-oral examination showed wounds around the nose and burn injuries on her left forearm and legs (Figs. 4–6).

Figure 4. Wounds around the nose

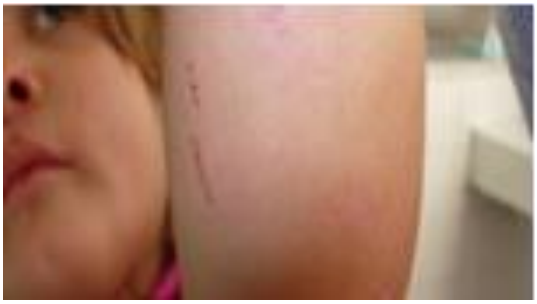

Figure 5. Burn injuries on her left forearm

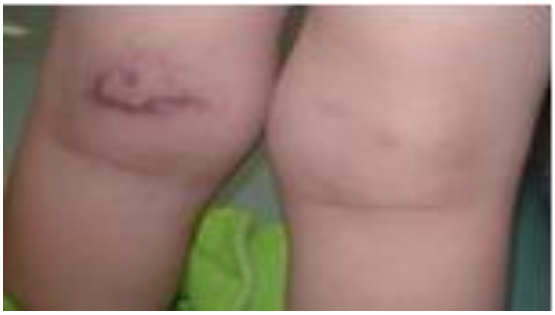

Figure 6. Burn injuries on her legs

During follow-up, the permanent mandibular right incisor began to erupt and was mobile (Fig. 7). The patient and her family were taught about oral hygiene. A partial denture for the lower arch and a protective plate for the upper arch are planned and the patient will continue to be followed periodically.

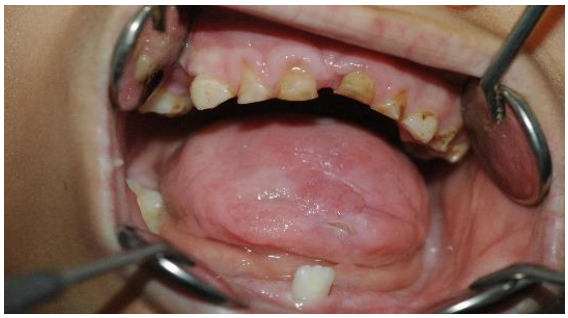

Figure 7. Permanent mandibular right incisor

Discussion

There are numerous hereditary sensory and autonomic neuropathies. The identification and classification of these diseases are ongoing processes. The clinical examination, autonomic tests, pathological examination, and molecular assessment allow precise identification. Axelrod et al. reported that many patients now reach adulthood with the identification of the various disease types and the development of treatment programs. At present, the clinical goal is to help these patients achieve independent function with minimal physical limitations (4).

Hereditary sensory neuropathy is classified into five types based on the clinical findings and nerve biopsy results (1, 2, 5, 6). The type could not be determined in our case because the patient's parents refused a biopsy. However, it is thought to be type V based on the clinical findings.

The most characteristic symptoms of this syndrome are the absence of pain sensation and self-inflicted wounds. When the patients reach 2~3 years of age, they begin to injure themselves because of the lack of pain sensation. This leads to ulcers on the hands and feet and traumatic fractures (1). In addition, the patients can wound their tongues or burn their arms and legs, as seen in our case.

The oral findings of the disease include oral lesions caused by repetitive trauma, scar formation in the oral mucosa and tongue, self-dental extraction, dental infections, and early tooth loss (2). Our patient had early loss of teeth.

In the 5 months after the patient first attended our clinic, she has been followed monthly. At the end of the first month, eruption of the mandibular right permanent central incisor and first molar was identified. Unfortunately, at the next visit, the right mandibular central incisor was missing. We plan to follow the patient regularly and make a child's prosthesis.

In these patients, we believe that it is necessary to conduct regular checkups and to ensure good communication between the dentist and family in terms of ensuring oral hygiene and preventing the development of undesired complications.

Acknowledgments

The authors deny any conflicts of interest related to this study.

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