

Congenital Insensitivity to Pain: A Case Report with Dental Implications

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Abstract

Congenital insensitivity to pain is a rare disorder seen in early childhood. Five different types of hereditary sensory and autonomic neuropathy have been identified, to date, with different patterns of sensory and autonomic dysfunction, peripheral neuropathy, clinical features, and genetic abnormalities. Absence of pain and self-mutilation are characteristic findings of this syndrome. Teeth in the oral cavity can cause damage to the oral tissues and tongue. When diagnosed, there should be cooperation between the dentist and neurologist. Using an oral shield prevents biting, and thus tissue trauma can be prevented. Here, we present the case of a 6-month-old boy with congenital insensitivity to pain (hereditary sensory and autonomic neuropathies; HSAN type V) with self-mutilation injuries to his tongue and fingers caused by biting, along with a discussion of treatment strategies. The results of this report suggest that early diagnosis and specific dental management for patients with congenital insensitivity to pain are important for prevention of the characteristic oral and dental problems accompanying this disorder.

Key words

Congenital insensitivity; Dental implications; Neuropathy; Pedodontics

Introduction

Congenital insensitivity to pain is a rare autosomal recessive disease, which was first described in 1932 by Dearborn as "Congenital pure analgesia".¹ Absence of pain sensation is a symptom of several disorders, which may be congenital or acquired. The majority of the congenital types are diagnosed in early infancy. Collectively termed "hereditary sensory and autonomic neuropathies" (HSAN),

these disorders affect the number and distribution of small myelinated and non-myelinated nerve fibers. They are also characterised by diminished or absence of sensitivity to pain, touch, and pressure on the extremities and varying parts of the trunk.² To date, five types of HSAN have been identified.³

Type I, hereditary sensory radicular neuropathy, is the most common type of HSAN, which is transmitted as an autosomal dominant trait. This disorder is characterised by a sensory deficit in the distal portion of the lower extremities, chronic perforating ulcerations of the feet, and progressive destruction of the underlying bones. Symptoms appear in late childhood or early adolescence. Many patients have accompanying nerve deafness, atrophy of the peroneal muscles, and a reduced number of unmyelinated nerve fibers. Motor nerve conduction velocities are normal, but sensory nerve action potentials are absent.

Type II, congenital sensory neuropathy, presents with onset of symptoms in early infancy or childhood. Upper and lower extremities are affected, with chronic ulcerations and multiple injuries to the fingers and feet. Pain sensation is predominantly affected, and deep tendon reflexes are reduced. Autoamputation of distal phalanges and neuropathic joint degeneration are quiet common. There is

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a total loss of myelinated fibers and a reduced number of unmyelinated fibers.

Type III, familial dysautonomia or Riley-Day syndrome, is an autosomal recessive disorder seen mostly in Jews of eastern European descent. Patients present with sensory and autonomic disturbances, such as a weak suck reflex, hypotonia, hypothermia, retarded physical development, poor motor coordination, reduced tears, depressed deep tendon reflexes, absent corneal reflexes, postural hypotension, and scoliosis. Peripheral nerves show reduced numbers of myelinated and unmyelinated axons. The prognosis of this type is very poor, with most patients dying in early infancy or childhood.

Type IV, congenital insensitivity to pain and anhidrosis (CIPA), is a condition in which infants present with hyperthermia unrelated to the environment, anhidrosis, and insensitivity to pain. Palmar skin is thickened, and Charcot's joints are commonly present. Peripheral nerve biopsy reveals the absence of small, unmyelinated fibers with abnormally enlarged mitochondria.

Type V, congenital insensitivity to pain, is an autosomal recessive condition with onset in infancy. There is congenital loss of sense of pain, which affects the extremities. Features of this condition include marked insensitivity to pain with tactile, vibratory, and thermal sensations usually intact and a severe reduction in unmyelinated fiber number.

The various disorders within this group are classified according to the different patterns of sensory and autonomic dysfunction and peripheral neuropathy, and the presence of additional clinical features, such as learning disabilities. Within this classification, congenital insensitivity to pain is referred to as HSAN type V^{3,4} and it is associated with marked insensitivity to pain, but with normal tactile, vibratory, and thermal sensations. Motor and tendon reflexes are intact in most cases.^{3,5-12} Self-mutilation is an almost invariable feature of this disorder, involving the teeth, tongue, lips, fingers, ears, eyes, and nose.^{13,14}

Here, we present a case report of congenital insensitivity to pain (HSAN type V) combined with self-mutilation injuries to the tongue and fingers, caused by biting, and discuss preventative treatment. The purpose of this paper is to present a case of congenital insensitivity to pain and to emphasize the oral implications of this condition.

Case Description

A 6-month-old boy was brought to the Department of Pedodontics, Dental Faculty, Dicle University, Diyarbakır,

Turkey, with a chief complaint of tissue loss on the tongue due to biting. He was born of a consanguineous marriage and was the only child. He was born with no complications and there was no family history of similar illness. The patient's history revealed an absence of normal reaction to painful stimuli, such as falls and cuts and no alteration in taste. The mother also reported that the infant did not cry during vaccinations.

He was diagnosed as having HSAN type V at 6 months old at the Department of Neurology, Faculty of Medicine, Dicle University. A general examination revealed the presence of normal deep tendon reflexes and normal reactions to light touch, tickling, and pressure. He also responded normally to thermal stimuli, although there was no reaction to painful stimuli. There was no history of flushing, temperature lability, or altered lacrimation, suggesting normal autonomic function.

His physical appearance was normal, with no dysmorphic features. Oral examination revealed a loss of tissue on the tongue caused by biting, although he had only two teeth (mandibular right and left incisors; Figure 1).

Tooth extraction is an extremely radical treatment, and should be the last option. First, we eliminated sharp surfaces of the two teeth by grinding. However, the treatment was unsuccessful. When the child reached 1 year old, intraoral examination revealed eight teeth consisting of four maxillary incisors and four mandibular incisors. Physical examination showed a loss of tissue with ulceration on the fingers of both hands, caused by biting (Figure 2).

Among the available treatment options, selective grinding would no longer be useful in this case. We felt that extraction was too aggressive for this patient, due to his young age. The focus was on oral rehabilitation, prevention of further injury to oral structures and other body parts, and restoring oral function. A management strategy was formulated, and his parents were consulted. We constructed acrylic plates to cover the incisal edges to prevent damage by the teeth. Initially, alginate impressions were taken from both jaws. Midazolam was given orally (0.2 mg/kg) together with ketamine (3 mg/kg) to facilitate impression taking due to the patient's management issues. The teeth on the impressions taken were blocked out, and acrylic plates were constructed using hot acrylic (Figure 3). After polishing, the plates were bonded to the patient's teeth with glass ionomer cement. The patient's parents were then instructed regarding oral hygiene.

The patient has been followed for 3 months using the preventive protocol and improvements in the condition of his fingers have been observed (Figure 4).



Figure 1 Extra-oral appearance of a 6-month-old infant with congenital insensitivity to pain. Note the loss of tissue on the ventral aspect of the tongue caused by biting.



Figure 2 Appearance of the fingers in a 12-month-old infant with congenital insensitivity to pain. Note the loss of tissue on the index finger.

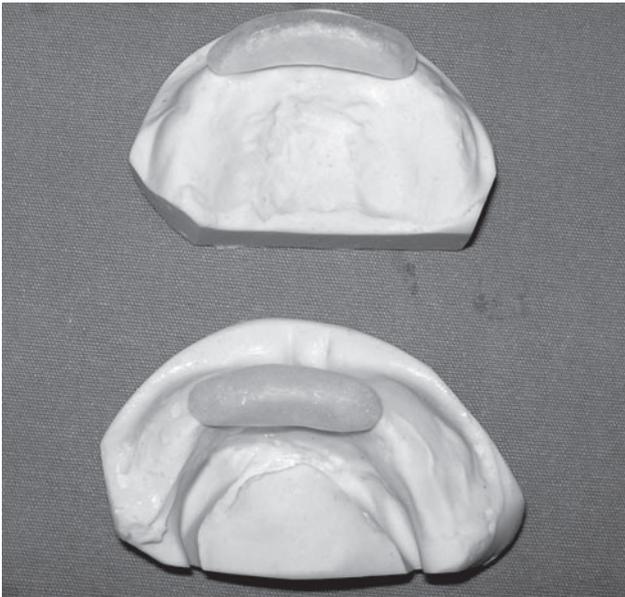


Figure 3 Maxillary and mandibular study casts used for the fabrication of acrylic plates.



Figure 4 Post-treatment appearance of the fingers in patient with congenital insensitivity to pain.

Discussion

Not only is the sense of pain the precursor of a large variety of pathological conditions, but its elimination for any reason may cause a number of adverse conditions and result in injury. This is a rare disorder caused by loss of the sense of pain, and the teeth represent one means by which these patients can harm themselves. These injuries often

begin as the primary dentition erupts, and are self-inflicted. Frequently, the tongue and lips are affected, with resultant scarring and deformation,¹⁵ as in the present case. Self-extraction of the teeth has also been reported.¹⁶

Presently, there is no specific treatment for congenital insensitivity to pain or the other hereditary sensory and autonomic neuropathies. Several methods for prevention of these injuries have been suggested, including elimination

of sharp surfaces of the teeth by grinding or addition of composite, the use of mouth guards and other appliances, and extraction of teeth. The use of intraoral appliances is often difficult or impossible to implement, because the mutilation may begin in infancy with the eruption of the primary incisors.¹⁷ Thus, extractions may be unavoidable in cases in which the mutilation is particularly severe.^{18,19} Where extractions in the primary dentition have been carried out, the effects must be monitored and addressed. Early loss of primary teeth for any reason is known to lead to a significant increase in need for orthodontic treatment. Loss of the entire primary dentition at such an early stage may result in loss of arch length in both the maxilla and the mandible, with marked mesial tipping of the permanent first molars, particularly in the mandible.²⁰ We considered use of an oral appliance to be the best option in this case. Training of the family regarding dental-gingival hygiene is also important. The dental team should be involved in the management of these patients as soon as a diagnosis is made. Moreover, careful monitoring should continue throughout the patient's lifetime, along with comprehensive dental care to maintain the patient's social, psychological, and behavioural rehabilitation. It is important for clinicians to be aware of the potential complications caused by hereditary sensory and autonomic neuropathies so that the appropriate treatment can be provided promptly following early diagnosis, thus preventing the development of untoward complications.

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