J Istanbul Univ Fac Dent 2016;50(2):49-53.
http://dx.doi.org/10.17096/jiufd.

CASE REPORT

ORAL MANIFESTATIONS AND PROSTHETIC REHABILITATION IN HEREDITARY SENSORY AND AUTONOMIC NEUROPATHY (HSAN) TYPE IV: A CASE REPORT*

Herediter Duyusal ve Otonomik Nöropati Tip IV Hastasının Oral Bulguları ve Protetik Tedavileri: Bir Olgu Nedeniyle

Duygu OFLUOĞLU [1], Nazlı ALTIN[2], Elif YAMAN [3], Elif Bahar TUNA-İNCE [3], Zeynep AYTEPE [3], Hakkı TANYERİ [1]

Received: 19/10/2015
Accepted:09/11/2015

ABSTRACT

Hereditary sensory and autonomic neuropathies (HSAN) are rare genetic syndromes of unknown etiology. They are seen in early childhood and are categorized into six different types by their symptoms. HSAN type 4 demonstrates autosomal recessive transmission pattern, with such major characteristics as loss of sense of pain, self-mutilation, anhydrosis and mental retardation. Sympathetic innervations are deficient despite the existence of sweat glands. Sufferers are hypotonic without any tendon reflexes, and neuro-motor development is retarded. In some cases tactile sensation and vibration may be intact. Biting injuries due to lack of pain sensation cause laceration, ulceration and scarring of the tongue, lips and other parts of oral mucosa. Tooth luxation and severe dental attrition have been observed. This case report presents oral and dental findings, surgical treatments and prosthetic rehabilitation of an 11-year-old boy with HSAN type 4.

Keywords: *Hereditary sensory and autonomic neuropathy; type IV; Dental needs; Removable partial prosthesis*

ÖZ

Herediter duyusal ve otonomik nöropatiler, etyolojileri bilinmeyen, nadir görülen genetik sendromlardır. Genellikle erken çocukluk döneminde görülürler ve semptomlarına göre 6 farklı tipi tanımlanmıştır. Otozomal çekinik geçiş gösterirler ve ağrı hissinin kaybolması, kendine zarar verme, terlemenin azalması ve mental gerilik karakteristik bulgularıdır. Ter bezleri mevcut olmasına rağmen sempatik inervasyon yetersizdir. Hastalar tendon refleksleri olmaksızın hipotoniklerdir ve nöro-motor gelişimde gerilik vardır, bazı vakalarda dokunma ve titreşim hissi zarar görmemiş olabilir. Ağrı hissinin olmaması nedeniyle ısırma sonucu dilde, dudaklarda ve oral mukozanın diğer bölgelerinde laserasyon, ülserasyon ve skar oluşumu görülebilir. Dişlerde sallanmalar ve şiddetli atrisyonlar oluşabilir. Bu vaka raporunda, HSAN Tip 4 teşhisi konmuş olan, 11 yaşındaki erkek hastanın oral ve dental bulguları, cerrahi ve protetik tedavileri sunulmuştur.

Anahtar kelimeler: *Herediter Duyusal ve Otonomik Nöropati; Tip IV; dental tedaviler; hareketli protez*

[1] Department of Oral and Maxillofacial Surgery Faculty of Dentistry Istanbul University
[2] Department of Oral and Maxillofacial Surgery Faculty of Dentistry Istanbul Aydın University
[3] Department of Pedodontics Faculty of Dentistry Istanbul University
* Presented as a poster in 8th ACBID International Congress, 28 May-1 June 2014, Mardan Palace Antalya,Turkey

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Introduction

Hereditary sensory and autonomic neuropathies (HSAN) are rare genetic syndromes characterized by congenital insensitivity to pain and temperature changes and are categorized under autonomic nerve formation disorders (1). These conditions are further classified into 6 types depending on the patient's natural history, population of neurons or axons affected and mode of inheritance (2, 3). Type IV HSAN is the most prevalent and is referred to as congenital insensitivity to pain with anhydrosis. It is believed that the basic pathology of the disease is the lack of superficial C group nerve fibers as well as disturbed branching of axons and dendrites. Skin biopsies performed in these patients have shown absent innervations of the epidermis and little innervations of the dermis (4).

HSAN type IV is characterized with recurrent episodes of unexplained fever, failure to thrive, absence or decline of perspiration despite the presence of normal sweat glands, insensitivity to pain, self-mutilation, finger biting, corneal injury, multiple fractures, malformations of the joints, deep ulcers, osteomyelitis and mild mental retardation (5-9).

Biting injuries cause laceration, ulceration and scarring of the tongue, lips and other parts of oral mucosa (10). Tooth luxation and severe dental attrition have been observed. Pathologic fracture of the mandible and the need for open reduction and internal fixation can also occur (11). In this paper, we present clinical and oral manifestations, dental findings, surgical treatments and prosthetic rehabilitation of an 11-year-old boy with HSAN type IV who has self-mutilation injuries in his tongue and in other parts of oral mucosa.

Case Report

An 11-year-old boy was referred to the Department of Oral and Maxillofacial Surgery, Faculty of Dentistry, Istanbul University by his ophthalmologist with a history of self-mutilation injuries of the tongue, oral mucosa and a chief complaint of dental problems. He was the second child of a 39-year-old healthy female and 40-year-old healthy male who are third degree relatives. He had a 14-year-old healthy brother. He was born with no complications and there was no family history of similar condition. The patient's medical history revealed that he was clinically diagnosed with HSAN type IV at the age of 15 months by his pediatrician based on the characteristic clinical features including insensitivity to pain, self-mutilation of the lips and tongue, tissue loss due to burn injury of the second right finger, corneal ulceration, scleral hyperemia and central neurotrophic keratitis, deep oral ulcers and the sural nerve biopsy. Physical appearance was normal according to his peers and he was of normal intelligence. General examination revealed the presence of normal tendon reflexes, normal reactions to touch and pressure but he did not respond to thermal stimuli and had no reaction to pain stimuli. His fingers were short and blunt. Central neurotrophic keratitis on both eyes, especially on the right eye was noted (Figure 1).



Figure 1. a) *Tissue loss on lower lip caused by biting.* ***b)*** *Short and blunt fingers and tissue loss due to burn injury of second right finger.* ***c)*** *Central neurotrophic keratitis on the right eye.*

Oral examination revealed limited mouth opening, ulcerated areas on the tongue and buccal mucosa due to biting and severe dental attrition. Maxillary central and lateral incisors, mandibular left first and second premolars were absent; the patient's family stated that there had been a spontaneous exfoliation of those teeth. The roots of maxillary right first premolar, left first molar and mandibular right and left first molars were present. Maxillary right second molar was impacted and there was a severe bone loss in the area of mandibular right second premolar (Figure 2). The treatment plan was focused on oral rehabilitation, prevention of further injuries to oral structures and restoration of oral function. Following

the consultation with Department of Pedodontics, mandibular right second premolar and the remaining tooth roots were extracted. Endodontic treatment of mandibular right first premolar was performed, fabrication of a removable partial dental prosthesis was planned for future appointments and regular follow-up of the patient was decided.

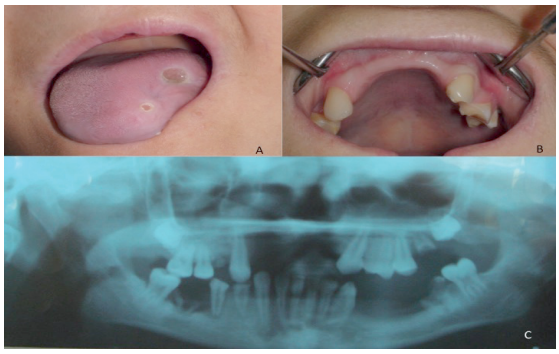


Figure 2. a) *Limited mouth opening and ulcerated areas on the tongue due to biting.* ***b)*** *Dental attrition* ***c)****Panoramic radiograph of the patient.*

Six months after the first visit, clinical examination revealed mobility of mandibular right second molar and subsequently exfoliation without any intervention. An incisional biopsy was made to examine the related area histopathologically since there was no healing observed (Figure 3).



Figure 3. *After spontaneous exfoliation of the mandibular right second molar. An incisional biopsy was made to examine the related area histopathologically, as there was no healing observed and the results confirmed osteomyelitis and osteonecrosis.*

The histopathological examination revealed a necrotic bone and chronic osteomyelitis. A full-thickness mucoperiosteal flap was elevated under local anesthesia with sedation. Necrotic bone areas were curetted under saline irrigation and closed primarily. After two months, the surgical site was completely healed without any complications and removable upper and lower partial prostheses were fabricated to improve chewing function and also, to restore aesthetic appearance (Figure 4 and 5).

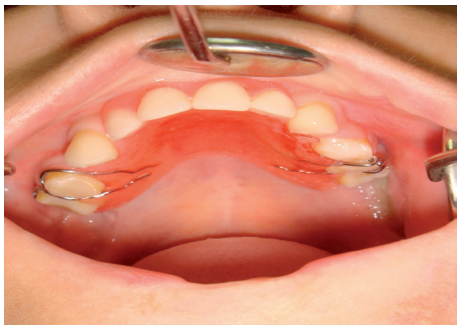


Figure 4. *Removable upper partial prosthesis.*



Figure 5. *Removable upper and lower partial prosthesis. Optimum occlusion could not provided as it should be because of bone loss.*

The patient was regularly followed-up for mutilation that can be caused by prosthesis but contrary to expectations, traumatic biting lesions occurred when the patient did not wear his prosthesis. A large ulceration area occurred in the retromolar area during the period in which the pediatric dentist was fabricating a new lower partial prosthesis. After a while, the lesion healed. Since a long lasting, large ulcerated area was observed at the left buccal mucosa close to commissures, an incisional biopsy was taken to eliminate the unexpected histopathological changes such as carcinoma *in situ* or dysplastic changes. The histopathological examination revealed the inflammatory granulation tissue (Figure 6). When the patient started to wear his new prosthesis, the ulcerated area healed over time. The mandibular partial dental prosthesis has been renewed three times and maxillary partial dental prosthesis has been renewed once because the patient was in growth period. He was followed up in 3 months intervals for

4 years in the Department of Oral and Maxillofacial Surgery and Pedodontics for dental problems and further mutilations.



Figure 6. *Long lasting, large ulcerated area.*

Discussion

Hereditary sensory and autonomic neuropathies (HSAN) are rare genetic syndromes of unknown etiology in which oral manifestations may be the presenting complaint. Injuries are due to congenital absence of pain sensation which results in a functional deficit of the feedback mechanism for pain (2). These injuries often begin as the primary dentition erupts and are self-inflicted. Frequently, the lips, tongue and buccal mucosa are affected with resultant scarring and deformation (12, 13). In this case, multiple ulcerations of tongue, buccal mucosa and scarring of lower lip due to biting were detected. Several methods including the elimination of sharp surfaces of teeth by grinding or addition of composite, the use of mouth-guards and other appliances and extraction of teeth have been suggested. Since the mutilation may begin in infancy with the eruption of primary incisors, the use of intraoral appliances is often difficult or impossible to implement (14). When deciding on the appropriate management, the degree of self-injuries is the most important factor; if the mutilation is severe, conservative treatment options could be ineffective and the extractions can be unavoidable.

Yet, older patients sometimes learn not to self-mutilate even though it causes them no pain (15). In our case, approximately 6 months after the first visit, an upper partial removable prosthesis was fabricated to check whether the patient can tolerate it or not. Unexpectedly, oral ulcerations caused by biting decreased. Because of the positive effects in oral cavity, dental team decided to fabricate the lower partial removable prosthesis. Additionally, severe dental attrition, early loss of primary teeth and some of the permanent teeth had led to bone loss in this patient; therefore, we could not provide an optimum occlusion.

Self-extraction or autoextraction of teeth has been previously reported (13). Self-mutilated tooth luxation was found to be as frequent as tongue bite in a case series study. Multiple missing teeth due to autoextraction were reported as a characteristic finding in more than one half of the subjects, being particularly observed in the mandibular anterior region of the infants. Amano *et al*. (14) suggested that children with HSAN type IV might induce oral self-mutilation through excessive bruxism or autoextraction in response to malaise or discomfort accompanying tooth eruption. There were four permanent missing teeth in this case and his parents declared that almost all primary teeth and the other four permanent teeth had been lost due to autoextraction. The incidence of mandibular osteomyelitis has been found high among patients with HSAN type IV but the reason for this phenomenon was not clear and could result in pathologic fracture and cause a need for open reduction and internal fixation (16). In this case, after autoextraction of mandibular right second molar, healing was not achieved and the biopsy of this region confirmed osteomyelitis and osteonecrosis. The necrotic bone was curetted up to healthy bone under local anesthesia and oral sedation, subsequently irrigated with saline solution and was closed primarily in the end.

The expected complete healing was achieved within two months without any complications. Provision of oral hygiene and prevention of dental disease is very important for these patients as caries can progress to pulpal involvement without causing pain and may lead to infection and tooth loss. Early loss of primary dentition, even if it results from extractions or autoextractions, can lead to loss of arch length in both jaws and significantly increase the orthodontic treatment needs (17). In our case, early loss of maxillary incisors and mandibular premolars due to autoextraction led to a significant decrease in bone volume and limited the provision of optimal occlusal relationships with partial removable prosthesis.

Conclusion

For the management of dental treatments, such patients should be carefully monitored throughout the lifetime by a dental team as soon as the diagnosis is made and the parents should be made conscious about dental treatment needs of their children to maintain the patients' social, psychological and behavioral rehabilitation. Training of the family regarding dental and gingival hygiene is very important. Dental team including a pedodontist and oral surgeon should plan a regular follow-up procedure and the consecutive prosthetic rehabilitation aiming to achieve a better occlusal harmony.

Source of funding
None declared

Conflicts of interest
None declared

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Corresponding Author:
Duygu OFLUOĞLU
Department of Oral and Maxillofacial Surgery
Faculty of Dentistry Istanbul University
34093 - Çapa Fatih, Istanbul / Turkey
Phone: +90 212 414 20 20/30353
e-mail: : duyguofluoglu@gmail.com