Dental Traumatology

Dental Traumatology 2009; **25**: 545–549; doi: 10.1111/j.1600-9657.2009.00810.x

Traumatic lesions from congenital insensitivity to pain with anhidrosis in a pediatric patient: dental management

CASE REPORT

Beatriz Gonçalves Neves, Rosemere Teixeira Roza, Gloria Fernanda Castro

Department of Pediatric Dentistry and Orthodontics, Federal University of Rio de Janeiro, Rio de Janeiro, Brazil

Correspondence to: Gloria Fernanda Castro, Travessa Alfredo Botelho, No. 66, Apt. 201 – Méier, Rio de Janeiro-RJ – Brazil, Zip code: 20720-200
Tel.: +55 21 3979 9820
Fax: +55 21 2562 2098
e-mail: gfbacastro@yahoo.com.br or beatriz_gneves@yahoo.com.br

Accepted 17 April, 2009

Abstract – Congenital insensitivity to pain with anhidrosis is a rare autosomal-recessive disorder characterized by unexplained fever episodes, anhidrosis, pain insensitivity, self-mutilating behavior, and mental retardation. The lack of sensitivity to pain results in traumatic lesions, such as ulcers, fractures, burns, bites, scars, and digital amputations. Several methods have been suggested to treat these patients; however, appropriate management is difficult, especially when the mutilation is particularly severe. This report describes the case of a 2-year-old female patient who had severe self-mutilating injuries to her tongue, hands, lips, and oral mucosa caused by biting. The patient presented digital amputation and also a premature loss of a permanent tooth germ during the treatment. The dental management is described and discussed. It is important to include the dentist on the multidisciplinary team to reduce the frequency and severity of the self-inflicted lesions in these patients, also to prevent complications.

Congenital insensitivity to pain with anhidrosis (CIPA), or hereditary sensory and autonomic neuropathy IV (HSAN IV), is an extremely rare autosomal-recessive disorder (1) that affects the peripheral and central nervous systems (2). It has been described as a rare disease; however, data regarding the incidence of this condition has not been found (3, 4). According to Bonkowsky et al. (5), the molecular diagnosis of CIPA has been confirmed in about 80 patients in the medical literature.

Clinically, HSAN IV is characterized by repeated, unexplained febrile episodes, anhidrosis, mental retardation, lack of pain and temperature sensation, and self-mutilating behavior (6–8). This condition is caused by autosomal recessive mutations and polymorphisms in the neurotrophic tyrosine receptor kinase 1 (NTRK1) gene, which encodes the receptor tyrosine kinase for nerve growth factor (4). Individuals with HSAN IV show an absence of unmyelinated fibers and loss of small myelinated fibers (7). The lack of epidermal nerve fibers and the absence of nerve fibers around sweat glands are the morphological basis of analgesia and anhidrosis in CIPA (9). However, touch and pressure sensitivity are unimpaired in these individuals (1).

The self-mutilating behavior is manifested in biting of the tongue and lips, painless fractures, burns, bruises, and cuts (1). Self-mutilation and repeated injury secondary to pain insensitivity predisposes the patient to a broad spectrum of complications including secondarily infected wound sites, osteomyelitis, fractures, joint deformities, and, less commonly, digital amputations (2).

Several methods have been suggested to treat these patients; however, appropriate management is difficult, especially when mutilation is particularly severe. In addition, information regarding this issue is scarce in the dental literature. Therefore, this report aims to describe a case of a 2-year-old girl with CIPA who exhibited self-mutilating lesions on her tongue, hands and fingers, and to describe the proposed dental treatment.

Case report

A 2-year-old girl with CIPA was referred to the Special Needs Clinic, Faculty of Dentistry, Federal University of Rio de Janeiro, by her speech therapist with the main complaint of a lesion on her tongue due to repeated bites.

Medical history revealed that the girl was a child of consanguineous parents, that is, first-degree relatives of each other (uncle–niece), and was born after a normal pregnancy of 37 weeks with a low birth weight (2.359 kg). From the first week of life, she experienced several febrile episodes, and the mother realized that the child's body temperature was influenced by room temperature. Absence of sweating and warm, dry skin was consistently noted during the febrile period.

At the age of 17 days, the patient's repeated fevers resulted in a hospital admission, where she was misdiagnosed with urinary infection. At the age of 7 months, during another hospital admission due to malnutrition

© 2009 John Wiley & Sons A/S

and continuous fever, the doctors realized that she did not cry from injections. At that time, a lesion on her tongue similar to an aphthous ulcer was observed. The patient was then referred to a dentist who chose to grind the sharp edges of her lower teeth to eliminate the self-biting. The mother reported that this treatment failed to eliminate the lesions.

By her ninth month, the patient presented with malnutrition and deep ulcers in her fingers and hands due to her self-mutilating behavior. At that point, the child was taken away from her mother based on a child-abuse report. Because of that, she temporarily lost the child's tutor. When the authorities observed that the lesions persisted, the child was referred for medical evaluation, at which time she was diagnosed with CIPA. Since then, the patient has been under the continuous care of a multidisciplinary team that includes a pediatrician, a physiotherapist, and a speech therapist.

Initial physical exam revealed the presence of wounds and deep ulcers on her fingers, hands, and knees. Oral examination was performed with difficulty as the patient was mentally retarded and agitated. The ventral surface of her tongue was extensively ulcerated with a necrotic aspect. The patient presented a complete primary dentition, except for the maxillary central incisors, which had been lost by trauma (avulsion). Her mother reported that after the avulsion, the tongue lesions diminished. Also, the child's hands were restrained with plastic wrap to prevent finger biting, but the patient removed it.

The treatment plan called for complete dental extractions under general anesthesia, which had been discussed with the mother; however, this option was countermanded by the patient's physicians because of her malnutrition. Therefore, all the extractions were carried out in the clinic under local anesthesia at weekly intervals.

Throughout treatment, regression of the lesions of the tongue, fingers, and hands was observed; however, other self-inflicted injuries appeared, such as self-extraction of fingernails, digital amputation (Fig. 1), and biting injuries of the lips and buccal mucosa (Fig. 2).

The oral ulcers were so severe that even a permanent tooth germ present in the oral cavity had to be extracted (Fig. 3) because the constant bites kept the alveolar ridge from healing (Fig. 4). These new self-mutilating behaviors took place between dental appointments as the patient learned new ways to hurt herself with the remaining teeth.

After extraction of all the teeth, the oral lesions (Fig. 4) and the scarred hands and fingers were able to heal (Fig. 1). At recall visits, the mother reported that

Fig. 1. Appearance of the patient's hands and fingers during dental treatment (a, b and c). Observe partial digital amputation of the middle finger of the right hand (c and d). The patient has almost no fingernails as the result of the self-mutilating behavior.

© 2009 John Wiley & Sons A/S

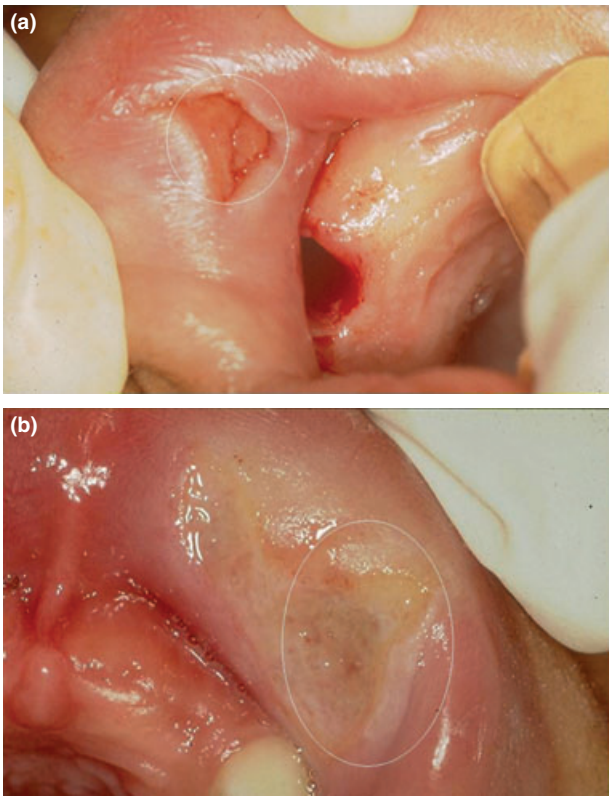

Fig. 2. Ulcerations on the upper lip resulting from biting after extraction of incisor teeth (a and b).

the child managed well without her teeth, chewing food between her alveolar ridges, with no loss in body weight.

The patient will be observed regularly in the dental department for any signs of further self-inflicted oral injuries and to monitor eruption of the permanent dentition, particularly with regard to the effect of early loss of primary teeth on the developing occlusion.

Discussion

Congenital insensitivity to pain with anhidrosis is a HSAN, in which consanguinity is frequently observed (3, 4, 8, 10). Individuals with this disorder present insensitivity to pain, a protective body mechanism essential to avoid injury (3, 11).

CIPA is referred to as HSAN type IV. At present, five types of HSAN have been identified. All HSANs are classified according to the different patterns of sensory and autonomic dysfunction and peripheral neuropathy, along with additional clinical features (1). However, it must be pointed out that in CIPA, pain insensitivity, and autonomic deficits are present, but touch and pressure sensitivity are unimpaired. Individuals with this disorder may also present impaired sweating with episodic hyperpyrexia associated with self-injuries (12). Self-mutilating behavior may also appear in other conditions such as Lesch–Nyhan Syndrome, so it is essential to differentiate them from CIPA (6). Furthermore, this condition can be mistaken for child abuse, as occurred in this case, as well as for those patients described in the study of Yagev et al. (10).

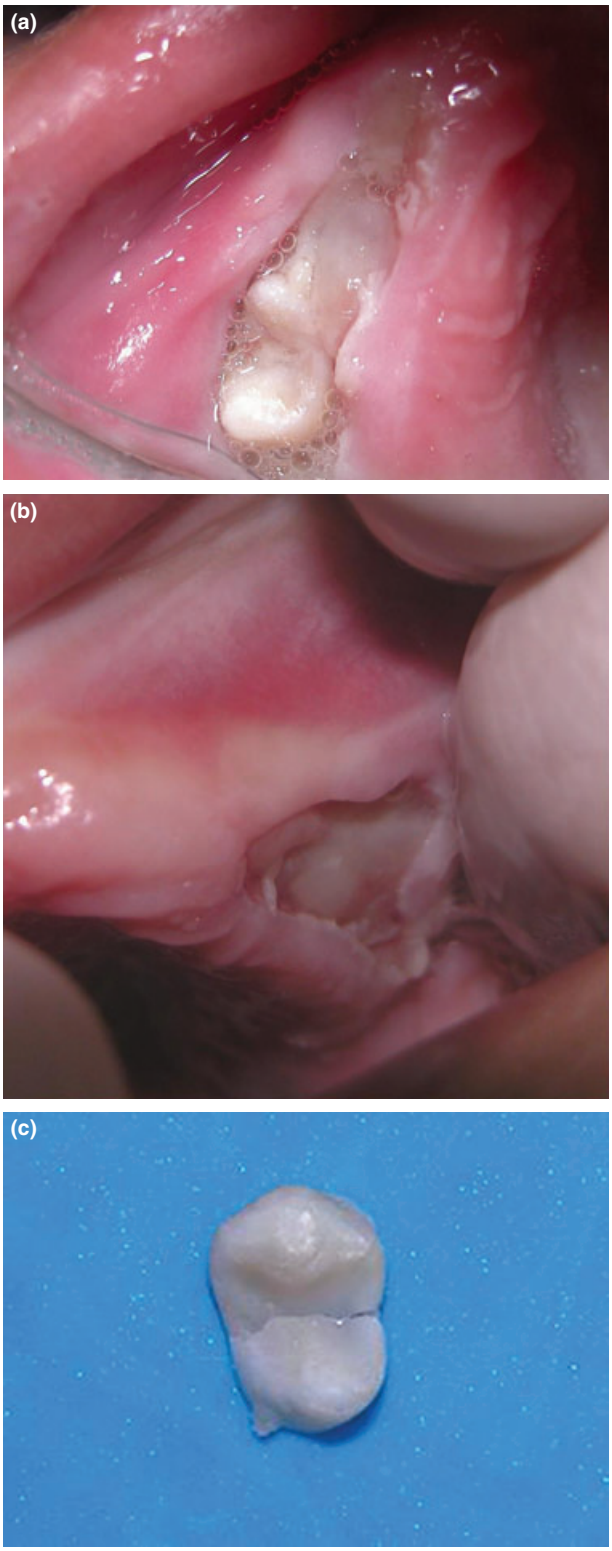

Fig. 3. Second incident of severe oro-dental self-mutilation. Healing of alveolar ridges were not complete due to biting (a and b); therefore, a permanent tooth germ had to be removed (c).

Owing to the lack of pain sensation, patients often use their teeth to harm themselves, which may result in severe injuries to oral tissues, including laceration and ulceration

© 2009 John Wiley & Sons A/S

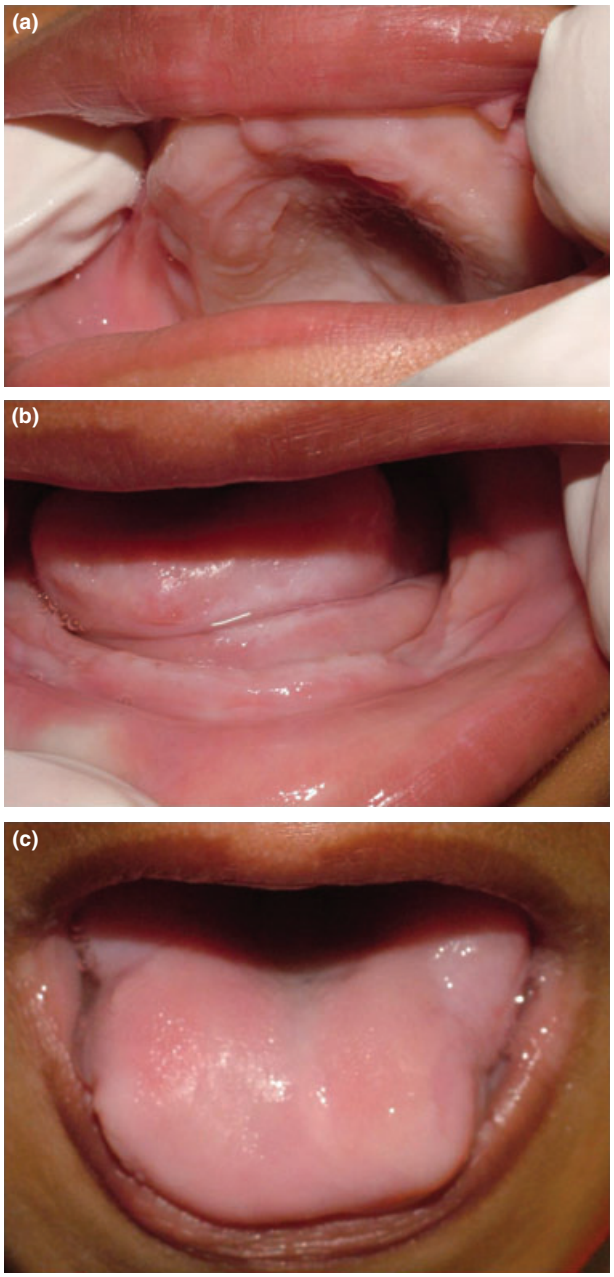

Fig. 4. Healed ulcers of the tongue (a) and upper (b) and lower (c) edentulous ridges.

of tongue, lips, and oral mucosa. In most cases, bite injuries to tongue, lips, and fingers start as the primary dentition erupts (6). Moreover, the self-mutilating behavior may result in more severe injuries, such as self-extraction of nails and self-inflicted amputation of fingers and toes (13). All these severe phenomena were observed in this case.

There are no standard techniques to prevent or treat orofacial self-inflicted injuries, so the treatment plan is predicated according to the circumstances of the individual case (14). Several methods of prevention of these lesions have been suggested, including tooth extraction (5, 10, 12, 15, 16), elimination of sharp edges of teeth by grinding or addition of composite, and the use of mouth guards and protective plates (15).

The degree of the self-injury must also be taken into account when deciding on appropriate management, and extractions may be unavoidable when the mutilation is particularly severe (12), as in this case. Therefore, the treatment chosen for this patient was full-mouth extraction. Moreover, the patient could not cooperate because of mental disability and lack of emotional maturity, so mouth guards and protective plates were ruled out. Full-mouth clearance became necessary to protect the child from further injury. It must be stressed that the extractions were performed under local anesthesia to promote hemostasis, and the treatment was carried out in short appointments, in a refrigerated room because of her characteristic hyperthermia. Although lacking pain sensation, some patients with CIPA do have tactile hyperthesia. Besides, some patients have complained of pain in the postoperative period. Thus, the use of anesthetics can prevent undue taction and are a necessity during operations (17). There are few reports regarding anesthetic management in patients with CIPA, especially concerning local anesthesia. As the procedures could not be performed under general anesthesia, local anesthesia was used to perform the extractions.

Because general anesthesia was contraindicated, dental treatment was performed at weekly intervals. However, this proved to be a disadvantage because the patient continued to bite herself between appointments, leading to a premature loss of a permanent tooth germ. It was hoped that the extractions would break her self-mutilating habit, allow time for tissue healing, and prevent further injury (14), but this was not the case. Moreover, self-inflicted trauma of the digits frequently causes wound infection, acute and chronic osteomyelitis, and digital amputation (2), which may have happened with this patient.

Prevention of dental disease is very important in these patients, as caries can progress painlessly to pulpal involvement, leading to infection and tooth loss (3, 15). Fortunately, this patient had no dental caries. Early tooth loss through trauma (5, 10) and self-extraction of teeth (15) have also been reported in patients with CIPA.

The dental team should be involved in the management of these patients as soon as diagnosis is made, and careful monitoring should continue throughout the patient's lifetime (12), especially when there is a risk of considerable space loss for the permanent dentition, such as occurred in this case. Continuous follow-up and close control is also necessary as it is hoped that, when the patient gets older, the self-mutilating behavior will improve, as observed in the case of Kouvelas et al. (18). It would appear that older patients can learn not to self-mutilate, even though it causes them no pain (12).

Conclusions

CIPA is one of the most difficult conditions confronted in dentistry (3). To reduce the frequency and severity of self-inflicted lesions and to prevent complications, it is

© 2009 John Wiley & Sons A/S

important to include the dentist on the multidisciplinary team.

References

1. Nagasako EM, Oaklander AL, Dworkin RH. Congenital insensitivity to pain: an update. Pain 2003;101:213–9.
2. Berkovitch M, Copeliovitch L, Tauber T, Vaknin Z, Lahat E. Hereditary insensitivity to pain with anhidrosis. Pediatr Neurol 1998;19:227–9.
3. Erdem TL, Ozcan I, Ilguy D, Sirin S. Case report hereditary sensory and autonomic neuropathy: review and a case report with dental implications. J Oral Rehabil 2000;27:179–82.
4. Toscano E. Hereditary sensory and autonomic neuropathy type IV. Orphanet Encyclopedia. December 2003. http://www.orpha.net/data/patho/GB/uk-HSANIV.pdf [last accessed 28 November 2006].
5. Bonkowsky JL, Johnson J, Carey JC, Gordon Smith A, Swoboda KJ. An infant with primary tooth loss and palmar hyperketratosis: a novel mutation in the NTKR1 gene causing congenital insensitivity to pain with anhidrosis. Pediatrics 2003;112:237–41.
6. Kim J, Woo Y, Kim G, Kim C, Ma J, Hwang T et al. Congenital insensitivity to pain with anhidrosis: a case report. J Korean Med Sci 1999;14:460–4.
7. Rosemberg S, Marie SKN, Kliemann S. Congenital insensitivity to pain with anhidrosis (hereditary sensory and autonomic neuropathy type IV). Pediatr Neurol 1994;11:50–6.
8. Shorer Z, Moses SW, Hershkovitz E, Pinsk V, Levy J. Neurophysiologic studies in congenital insensitivity to pain with anhidrosis. Pediatr Neurol 2001;25:397–400.
9. Nolano M, Crisci C, Santoro L, Barbieri F, Casale R, Kennedy WR et al. Absent innervation of skin and sweat glands in congenital insensitivity to pain with anhidrosis. Clin Neurophysiol 2000;111:1596–601.
10. Yagev R, Levy J, Shorer Z, Lifshitz T. Congenital insensitivity to pain with anhidrosis: ocular and systemic manifestations. Am J Ophthalmol 1999;127:322–6.
11. Littewood SJ, Mitchell L. The dental problems and management of a patient suffering congenital insensitivity to pain. Int J Paediatr Dent 1998;8:47–50.
12. Butler JB, Fleming P, Webb D. Congenital insensitivity to pain: review and report of a case with dental implications. Oral Surg Oral Med Oral Pathol Oral Radiol Endod 2006;101:58–62.
13. Gadoth N, Mass E. Hereditary neuropathies with self-mutilation. J Pediatr Neurol 2004;2:205–11.
14. Diab M. Self-inflicted orodental injury in a child with Leigh disease. Int J Paediatr Dent 2004;14:73–7.
15. Amano A, Akiyama S, Ikda M, Morisaki I. Oral manifestations of hereditary sensory and autonomic neuropathy type IV. Congenital insensitivity to pain with anhidrosis. Oral Surg Oral Med Oral Pathol Oral Radiol Endod 1998;86:425–31.
16. Ku ASW, Rodrigo CR, To PCY. Anesthesic management of a child with congenital insensitivity to pain with anhydrosis. J Oral Maxillofac Surg 2005;63:848–51.
17. Tomioka T, Awaya Y, Nihei K, Sekiyama H, Sawamura S, Hanaoka K. Anesthesia for patients with congenital insensitivity to pain and anhidrosis: a questionnaire study in Japan. Anesth Analg 2002;94:271–4.
18. Kouvelas N, Terzoglou C. Congenital insensitivity to pain with anhidrosis: case report. Pediatr Dent 1989;11:47–51.

© 2009 John Wiley & Sons A/S

This document is a scanned copy of a printed document. No warranty is given about the accuracy of the copy. Users should refer to the original published version of the material.