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Case Report

Congenital Insensitivity to Pain with Anhidrosis: Dental Considerations and Management. A Case Report

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Abstract

Background: Congenital insensitivity to pain with anhidrosis (CIPA), also known as hereditary sensory and autonomic neuropathy type IV (HSAN IV) is an autosomal recessive disorder characterized by insensitivity to pain, anhidrosis, intellectual disability and unconscious self-mutilation of fingers, lips, oral mucosa and tongue. Mutations in the neurotrophic tyrosine kinase receptor type 1 gene (NTRK1), have been reported to be associated with CIPA. No specific treatment for CIPA, however, early diagnosis and management of systemic and oral complications may be useful in the reduction of frequency and severity of these complications.

Aim: To report oral manifestations associated with CIPA and its management.

Case Report: 1.5 years old Saudi girl attended with her parents to the department of pediatric dentistry at Prince Sultan Military Medical City. The parents requested extractions of the erupted teeth. Oral examination revealed multiple mouth ulcers affected her buccal mucosa, tongue and lips as a result of continuous biting her oral soft tissues. Upper central incisors were exfoliated following eruption. Primary molars erupted prematurely. The child had recurrent episodes of hyperthermia and anhidrosis. Genetic analysis confirmed diagnosis of CIPA. The parents are consanguineous. All teeth were extracted on the dental chair in subsequent visits. All ulcers healed nicely at the follow up appointment at age 2 however, the patient had hip dislocation and unable to walk.

Conclusion: There are no guidelines available to treat this rare condition. However, early diagnosis and dental management of patients with CIPA are vital for prevention of orofacial manifestations. Mouth guard is useful to prevent oral injuries in older children.

Keywords: *Congenital Insensitivity to Pain with Anhidrosis (CIPA); Hereditary Sensory and Autonomic Neuropathy Type IV (HSAN IV); Neurotrophic Tyrosine Kinase Receptor Type 1 Gene*

Background

Congenital insensitivity to pain with anhidrosis (CIPA), also known as hereditary sensory and autonomic neuropathy type IV (HSAN IV) is an autosomal recessive disorder with an estimated incidence of 1 in 25,000 [1]. It is characterized by insensitivity to pain, anhidrosis, intellectual disability and unconscious self-mutilation of fingers, lips, oral mucosa and tongue [2]. Other manifestations include: scaring and infection of the skin, multiple bone fracture and recurrent joint dislocations resulting in joint deformity [2]. One study reported dental anomalies such as hypomineralization can be seen in individuals with CIPA [3]. Recurrent febrile episodes caused by anhidrosis are the initial manifestation of CIPA [2]. Mutations in the neurotrophic tyrosine kinase receptor type 1 gene (NTRK1), have been reported to be associated with CIPA [4]. No specific treatment for CIPA, however, early diagnosis and management of systemic and oral complications

may reduce the frequency and severity of these complications. This include: dental splints, extractions in severe cases, partial dentures, finger sleeve splints and continuous wearing shoes to prevent self-mutilation injuries [5].

Aim of the Study

To report the severity of the oral and dental manifestations associated with congenital insensitivity to pain with anhidrosis (CIPA) and its management.

Case Report

1.5 years old Saudi girl attended with her parents to the department of pediatric dentistry at Prince Sultan Military Medical City. The parents of the patient requested extractions of the erupted teeth due to mouth ulcers from continuous biting her tongue and oral mucosa. The child had a history of recurrent episodes of hyperthermia, anhidrosis, hepatomegaly, pain insensitivity, peripheral neuropathy and developmental delay. The parents are consanguineous (first degree relation from both sides). The genetic diagnosis of congenital insensitivity to pain with anhidrosis (CIPA) has been confirmed. Homozygous pathogenic variant was identified in the NTRK1 gene (homo c.526ctt pathogenic class I PCG1N176). The parents said that six of their daughter's teeth were lost as a result of self-mutilation (two of them were found in her pillow, four were mobile two of them were removed by parents and the other two by pediatric dentist on a dental chair). Oral examination revealed multiple mouth ulcers affected her buccal mucosa, tongue and lips as a result of continuous biting her oral soft tissues (Figure 1a-1c). Intraoral radiographs were taken showing normal developing teeth and supporting structures (Figure 2a and 2b). Primary first and second molars erupted prematurely. All teeth were extracted on dental chair in subsequent visits (Figure 3). The extracted teeth are hypo-mineralized (Figure 4). At age 2 years, the patient came for follow up visit, her oral ulcers healed gradually with scars. The patient had right hip joint dislocated and wearing hip splint (Figure 5). In addition, there was a scar from healing burn of her left leg (Figure 6).

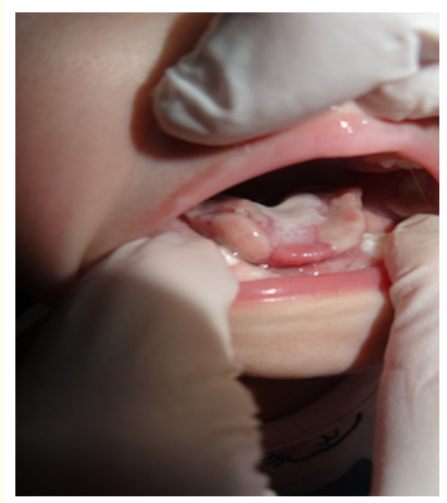

Figure 1a: *Tongue ulcers.*

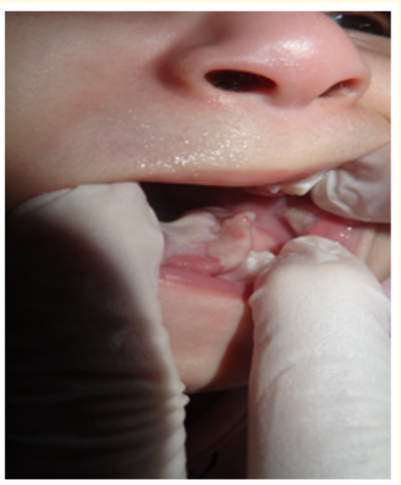

Figure 1b: *Buccal ulcers.*

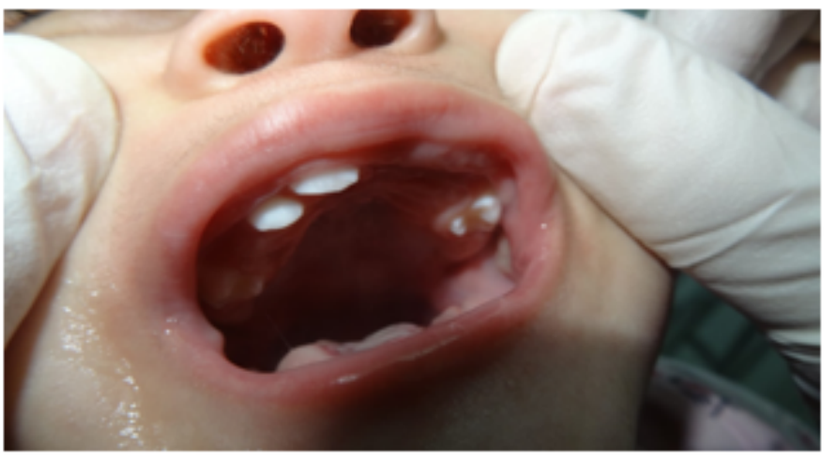

Figure 1c: *Lips ulcers.*

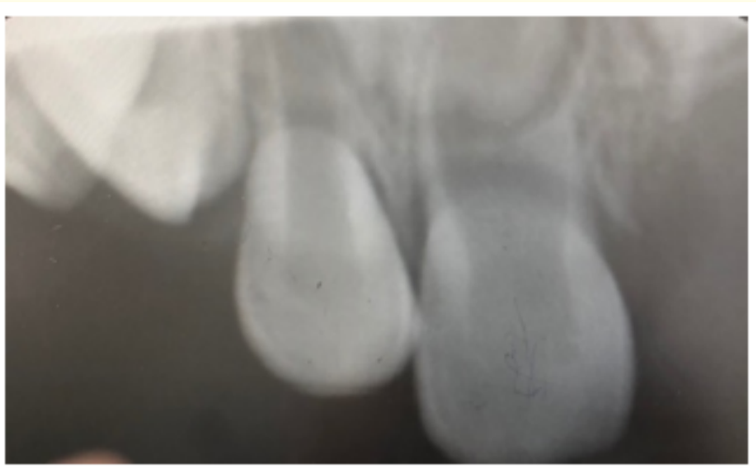

Figure 2a: *Radiograph of teeth #51 and #52.*

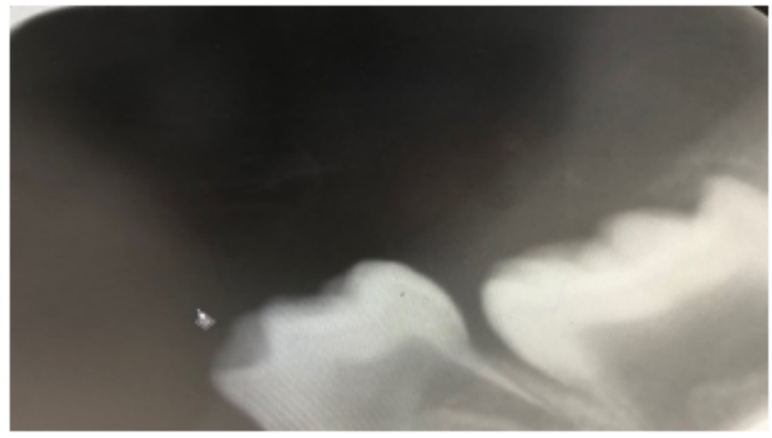

Figure 2b: *Radiograph of teeth #74 and #75.*

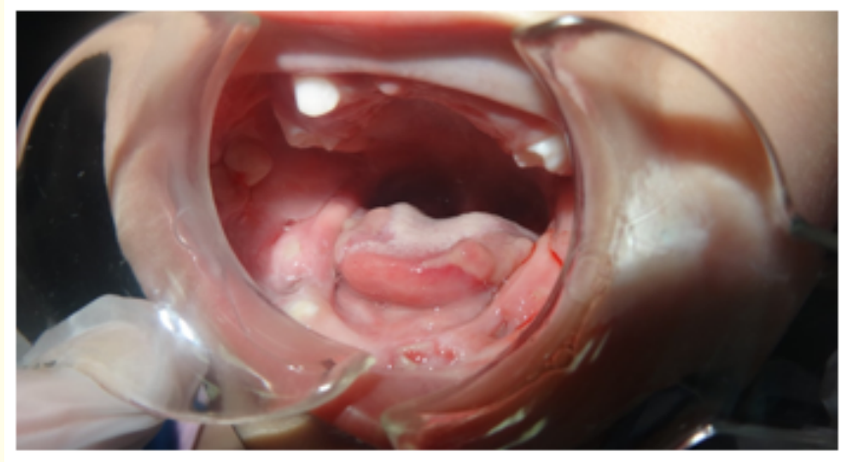

Figure 3: *Healing sockets following extraction of lower molars.*

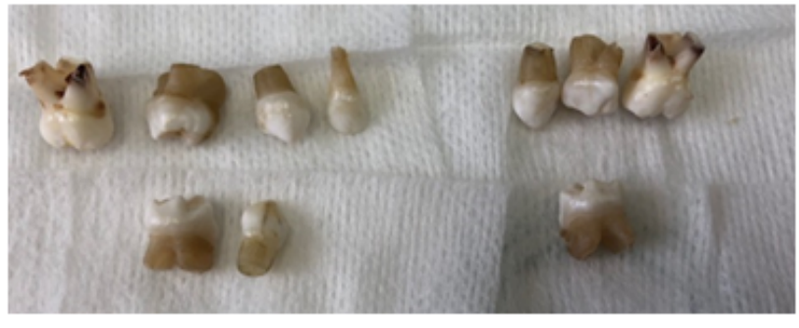

Figure 4: *Extracted teeth showing enamel hypoplasia.*

Figure 5: *Hip dislocation was managed by earing hip support.*

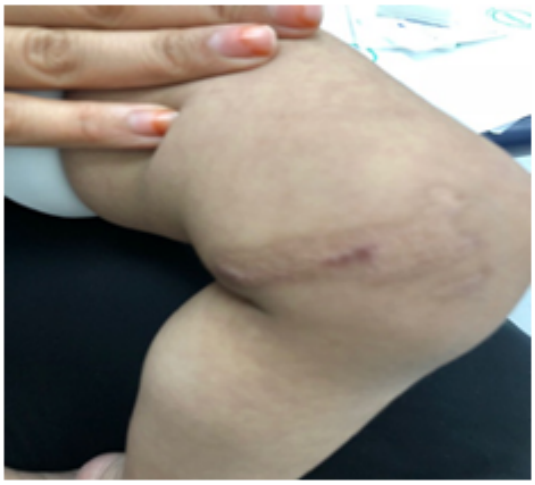

Figure 6: *Scar of healed burn.*

Discussion

This report describes the case of a 1.5-year-old Saudi female patient who had severe self-mutilating injuries to her tongue, lips, and buccal mucosa. In the present study and previous study by Abdullah., *et al*. [5] it has been noted that continues oral tissues biting during

the eruption of deciduous anterior teeth and lack of feelings pain made the child do it more until the incisors become loose and easily avulsed by itself. Interdisciplinary work of specialized medical and dental teams and development of standardized treatment protocols are essential for the management of CIPA. In addition, educate parents to ensure that the child is always in a safe environment [5]. Children at proper age of understanding can be educated to stop injure their oral tissues and fingers by showing them photographs of similar condition to avoid harming themselves. Soft or hard dental splints may be used for the upper or lower teeth to break the habit. This is only possible when the deciduous molars are present and able to retain the splints [5]. However, in the present case, the child was too young to accept such treatment and her condition was so severe that necessitate full mouth clearance. Similar observation has been reported previously as the choice of conservative treatment can be ineffective and the extractions can be unavoidable in severe cases [6]. In the present case, all extractions have been done in the dental surgery by applying 20% benzocaine topical ointment on the surgical site. The reasons behind this option was to avoid unnecessary risk associated with general anesthesia in individual diagnosed with CIPA and because the extractions have been done in subsequent visits due to different times of teeth eruption. At older age, the patient can have removable dentures to improve chewing function, speech and to restore aesthetic appearance. This depends on patient's motivation and acceptance of the prostheses.

Conclusion

There are no guidelines available to treat this rare condition. However, early diagnosis and dental management of patients with CIPA are vital for prevention of orofacial manifestations. Mouth guard is useful to prevent oral injuries in older children.

Bibliography

1. Algahtani H., *et al*. "Congenital insensitivity to pain with anhidrosis: A report of two siblings with a novel mutation in (TrkA) NTRK1 gene in a Saudi family". *Journal of the Neurological Sciences* 370 (2016): 35-38.

2. Indo Y. "Congenital Insensitivity to Pain with Anhidrosis". GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2019 (2008).

3. Gao L., *et al*. "Oral and craniofacial manifestations and two novel missense mutations of the NTRK1 gene identified in the patient with congenital insensitivity to pain with anhidrosis". *PLoS One* 8.6 (2013): e66863.

4. Wang WB., *et al*. "Identification of a novel mutation of the NTRK1 gene in patients with congenital insensitivity to pain with anhidrosis (CIPA)". *Gene* 679 (2018): 253-259.

5. Abdullah N., *et al*. "Congenital Insensitivity to Pain without Anhidrosis: Orodental Problems and Management". *Case Reports in Dentistry* (2015): 179892.

6. Ofluoglu D., *et al*. "Oral manifestations and prosthetic rehabilitation in hereditary sensory and autonomic neuropathy (HSAN)type IV: a case report". *Journal of Istanbul University Faculty of Dentistry* 50.2 (2016): 49-53.

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