CASE REPORT

Congenital insensitivity to pain - A rare case report

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Abstract

Congenital insensitivity to pain is defined as persistent occurrences of infections and inexplicable fever, anhidrosis (inability to sweat), and lack of response to deleterious stimuli, self-mutilating performance, mental abnormalities, and injuries to oral cavity. This syndrome can be diagnosed by clinical features, and genetic test is considered as a diagnostic tool. The diagnosis of this syndrome should be done earlier to provide greater lifespan to the patient, thus preventing ejections. In the present case, 3-year-old boy came to our department with the chief complaint of shaking upper front teeth for 1 day and his parents gave a history of congenitally absence of pain and temperature sensations since birth.

Keywords: Genetics, pain, self-mutilation

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Received 01 February 2018; Accepted 05 March 2018
doi: 10.15713/ins.ijmdcr.81

Introduction

Pain vastly shows shielding effect for the body as it shows a repulsive response causing removal from slightly harmful stimuli that can cause definite or possible tissue destruction. “Congenital insensitivity to pain” is an infrequent ailment, characterized by sensory experience that may be designated as an hostile responsiveness of a harmful provocation or bodily damage.[1] It shows its deleterious effect in various disorders where its late appearance shows life-threatening complications. The striking features of congenital insensitivity to pain are traumatic injury and self-mutilation, and the most frequently involved area are oral which includes teeth, lips, and surrounding structures such as lips, ears, eyes, nose, and fingers.[2,3] In few cases, self-removal of teeth has also been noted. This condition occurs due to deleterious effect on both myelinated and unmyelinated nerve fibers, which is considered by weakened or lacking sensitivity to pain, touch, and pressure on the extremities and other parts of the body without patient awareness of the pain causing burns at high temperatures seen predominantly in children.[4]

Case Report

A 3-year-old male child presented to the department of oral medicine and Radiology with the chief complaint of loose upper front teeth since 1 day, consulted dentist and got splinting done for the same [Figure 1]. In history of presenting illness patient’s parents gave history of fall while playing and got hit with cot, bleeding, and mobility was present in the upper front teeth since then they also reported that the patient is insensitive to pain as well as insensitive to hot or cold temperatures and blunt or sharp injuries since birth. Patient’s parents gave a history of consanguineous marriage and self-mutilated injuries over legs, hand, lip, tongue, head, and even self-removal of splinting done by the dentist immediately after reaching home [Figure 2]. On extraoral examination, wounds and scars were observed on upper and lower limb and on chin region [Figures 3-5]. The patient gave a history of trauma 2 years back, and middle toe of lower limb appears to be shorter than other toes and absence of nail was noticed [Figure 6]. Another history of trauma was present 1 week back with the right thumb showing healing abraded wound. On intraoral examination, lacerated wound was present on the right and left lateral border of the tongue, corner of the mouth, and on upper and lower labial mucosa. His upper lip appears to be swollen. On hard tissue examination, teeth present were 51, 52, 53, 54, 55, 61, 62, 63, 64, 65, 71, 72, 73, 74, 75, 81, 83, 84, and 85. Physiologic mobility was present in relation to 51 and 61. Ellis class IX fracture was present in relation to 52 and 62. Clinically, 82 was missing, but patient’s parents did not gave any history of self-extraction or exfoliation, only space was present in relation to 82. On investigations, intraoral periapical radiograph was taken and 51 was extracted under local anesthesia and splinting was done in relation to 52 and 62 with flowable composite and stainless steel wire.

Its treatment requires a multidisciplinary approach with the specialists involved as pediatrics, orthopedics, dentistry, ophthalmology, and dermatology. Genetic counseling for the
parents is of utmost importance. The patient should be treated with intravenous antibiotics to reduce infections, hand and foot guards to reduce injury, and referred to dentist for the oral guard and ophthalmologist for the evaluation of eyes.

Discussion

In 1932, it was firstly described by Dearborn as "congenital pure analgesia."\(^1,\text{5}\)

In 1975, McMurray suggested that diminishing of pain perception due to mental retardation, infection, and trauma is not considerable as "congenital universal insensitivity to pain."\(^1,\text{2}\)

In 1983, Dyck et al. recommended the diagnostic criteria for the
disease on the basis of histological, microscopic, and morphometric methods for the assessment of nerve fiber pathology. Its other synonyms are hereditary sensory autonomic neuropathy (HSAN), congenital general pure analgesia (Dearborn, 1932), congenital universal insensitivity to pain (Ford and Wilkins, 1938), congenital universal indifference to pain (Boyd and Nie, 1949), congenital absence of pain (Winkelmann, 1962), and Morvan syndrome. Three diagnostic criteria was proposed by Thrush for defining congenital insensitivity to pain:

1. Absence of pain congenitally.
2. Complete body affect observed.
3. Undamaged or minimally compromised other sensory modalities and tendon reflexes present.

These neuropathies was firstly described by in 1966 by Pinsky and DiGeorge and was later on revised by Dyck in 1984 who categorized them into five types: [4,5]

Type 1: It is a result of autosomal dominant inheritance which occurs due to missense mutations in the human gene SPTLC1 present at the locus 9q22.1-q22.3. Serine palmitoyltransferase is a pyridoxal-5'-phosphate-dependent enzyme. The disease occurs between 2nd and 4th era of lifetime. Initial signs often involve the absence of pain and temperature sensation principally in the distal parts of the lower limbs with further progression to more proximal parts and hands. Later on, there may be a loss of pinprick and surface sensation with the preservation of vibration sense. [1]

Type 2: It follows autosomal recessive trait which due to mutations in the HSN2 gene (hereditary sensory neuropathy) presents at locus 12p13.33. The condition is presenting in infancy or early childhood. Initially, the patient complains of numbness in distal extremities. Later on, pain, temperature, and touch sensations were also affected, and in severe cases apart from the limbs, it may affect trunk also. There was no effect in patient’s mental status. Absence of sensory nerve action potentials with normal functioning of motor nerves. The absence of sweating will be absent with no effect on the regulation of temperature and blood pressure. [4,5]

Type 3: It is recognized as familial dysautonomia or Riley–Day syndrome affecting predominantly Ashkenazi Jews. It is an autosomal-recessive inheritance, and IKB kinase complex-associated protein (KAP) gene IKBKAP is responsible which is located at locus 9q31 in the site of mutation. [6,5] Initially, signs are nourishing problems due to deprived oral harmonization and hypotonia. Clinically, there will be reduction in pain perception, unknown fevers, hypertension, and postural hypotension which is triggered due to defective control mechanisms of temperature and blood pressure. Patients suffer from oropharyngeal incoordination and feeding complications due to abnormal gastroesophageal motility, vomiting, and recurrent aspiration pneumonia initiating prolonged lung disease. Musculoskeletal manifestations include ataxia gait, severe kyphoscoliosis, and deformities of the feet leading to increased incidence of fractures. [1,4]

Type 4: It is the most widespread type with the striking feature of anhidrosis and hence known as congenital insensitivity to pain with anhidrosis. It is an autosomal recessive inheritance affecting “NGF-tyrosine kinase system.” It is characterized by persistent incidences of unknown fever, and despite the presence of normal sweat glands, it affects the normal functioning of perspiration, insensitivity to pain, self-mutilation, and mild mental retardation. Intraorally, laceration and ulceration of the tongue, lips, and other oral mucosa were found due to the presence of bite wounds as there was a total absence of pain and temperature sensations. In many cases, self-extractions along with severe dental attrition have also been reported. Another distinctive feature in Type IV was found to be decubital ulcer on the ventral surface of the tongue which may cause tongue injury during sucking or nursing after the eruption of incisal edges of erupting mandibular primary incisors as seen in case of Riga–Fede disease, which is caused by mechanical irritation by the incisal edge of the natal or neonatal mandibular incisors. Further, oral trauma, such as tongue or lip biting, is observed with the eruption of the upper and mandibular primary. One of the diagnostic signs of HSAN IV is unrestrained tongue biting with the primary incisors leading to the severely injured tongue with the presence of tissue laceration and excessive bleeding along with infection, fever, or malnutrition. [5,7]

Type 5: It is an autosomal recessive disorder due to NGF beta gene mutation. Clinically, pain and temperature insensitivity are apparent in childhood, with the manifestations of painless fractures, ulcers, and burns. Classically, self-mutilation was observed as biting of the lips, and tongue was present. In this type, pain and temperature sensitivity are lacking, whereas proprioception and sensitivity to touch, pressure, and vibration are unaffected. The autonomic manifestations are variable, with minimal autonomic abnormalities and blotching, abnormal sweating, difficulties with feeding, and elevated temperatures. [1,4]

**Oral manifestations of HSAN**

Innumerable oral indicators that are frequently appreciated in HSAN patients are as follows: Trauma to the tongue leading to chronic non-healing ulcers, trauma to lower lip, severe cheek biting leading to the formation of thick fibrous scars owing to reduced mouth opening, missing teeth due to self-extraction or dental sepsis, insensitivity to dental pain leading to space infections or dental sepsis, increased incidence of fractures and osteomyelitis of jaws, increased incidence of traumatic ulcers orally, dryness or fissuring of vermilion border of lip due to anhidrosis, severe dental attrition and cervical abrasions, erosion of teeth due to regurgitation of fluids because of defective gastroesophageal motility, abnormal gastroesophageal motility may cause feeding worries and recurrent vomiting in children, xerostomia and increased incidence of dental caries, and oral infections such as candidiasis. Although patients are sensitive to thermal stimuli, they may be unaware of the pain associated with high temperatures, thus leading to burns of oral mucosa, especially palate, due to hot food or beverages, etc. [6]

**Role of a Dentist**

By taking into consideration the degree of self-injury, there are several methods that can be imparted for the prevention of the oral mucosal injuries in these patients, such as follows:
• Contouring the sharp surfaces of teeth (enameloplasty) or addition of composite restorations.
• Usage of mouth guards or tongue guards and other appliances for the prevention of injury to the tongue.
• Extraction of offending teeth.

However, the usage of intraoral appliances is often problematic to implement in cases where eruption of the primary incisors is leading to self-mutilation. Therefore, extractions of obvious teeth should be done as soon as possible.[4]

Thus, the role of dental team is of utmost importance so that the condition of such patients should be diagnosed as early as possible and then regular follow-up is necessary to avoid further consequences.

In the present case, the affected child was an infant with no mental retardation, but his parents gave a history of impairment of pain and temperature since his childhood, and self-mutilated injuries were observed over legs, hand, lip, tongue, and head and even he tried for self-removal of splinting, and hence, we diagnosed it as type II congenital analgesia patient.

Conclusion

Congenital insensitivity to pain is an uncommon disorder categorized by self-mutilating behavior leading to the child to oral ulcerations on lips, tongue, and cheeks and also finger and hand biting. This ailment characterizes a challenge to the dentist and should be diagnosed and treated appropriately as early as possible to avoid life-threatening complications.

References