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Case Report:
Congenital Insensitivity to Pain

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Abstract:
Congenital Insensitivity to Pain belongs to the family of Hereditary Sensory and Autonomic Neuropathies (HSAN). It is a rare disorder of unknown etiology associated with loss of pain sensation. Cognition and sensation is otherwise normal and there is no detectable physical abnormality. We report a case of Congenital Insensitivity to Pain in a 3 year old female child.
Key Words: Congenital; Pain; Insensitivity; Nerve; HSAN

Introduction:
Pain is an unpleasant sensory and emotional experience associated with actual or potential tissue damage, or described in terms of such damage.(1) Most of us from the day we are born have this intuitive notion about pain or known to be aware of it by learning from our experiences. Imagine a life without pain, we will never feel a headache, or a toothache, or even a broken arm. This condition is called Congenital Insensitivity to Pain. Most people would think it would be great to live without pain, but one should understand, pain is an indication to our brain that our body needs something.

Case Report:
A 3 years old female patient reported to Department of Oral Medicine and Radiology with a complaint of missing lower front teeth and wound over her chin (Fig 1). Patient's mother gave a history of trauma to her daughter's chin 5-days back. Following which she noticed the wound and missing lower front teeth. Patient's mother also noticed bleeding from the lower front teeth region which subsided spontaneously after applying topical country medication. There was no history of pain, parasthesia, difficulty in opening mouth, speech and mastication.
The past history of the patient revealed that she had frequent history of trauma either unknown or self mutilated. The recent history was that of a trauma to her fingers 20 days ago and did not show any signs of discomfort. The child was generally considered normal as she rarely cries.
Family history revealed that she was a product of consanguineous marriage and did not have any complaint at the time of birth and her other sibling was normal.

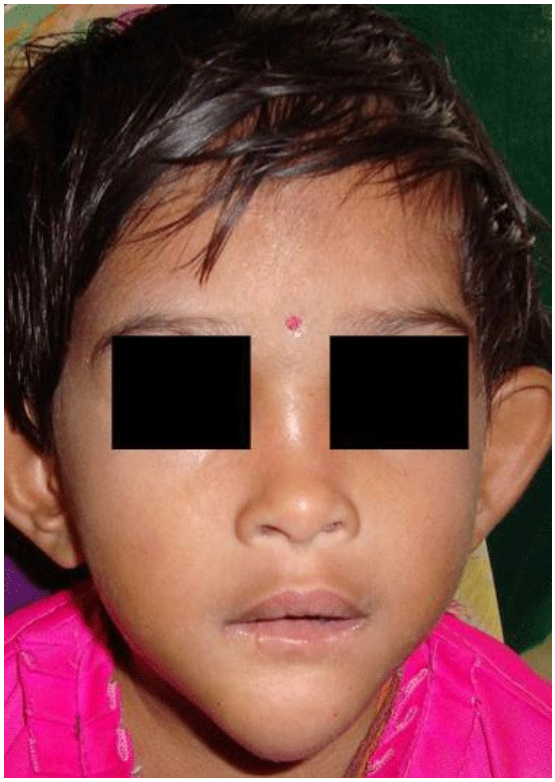

Fig 1: Patient's Photograph

General physical examination revealed a well oriented, partly cooperative, moderately built and nourished child with all her vital signs within satisfactory limits. Patient presented with multiple scars and wound over her left thumb, middle and index finger nails with shortening of fingers (Fig 2 & 3). A solitary oval shaped ulcer ranging 2x2cms in size was noted over the sole of right toe (Fig 4). The borders were well defined with an erythematosus base. It was nontender on palpation.

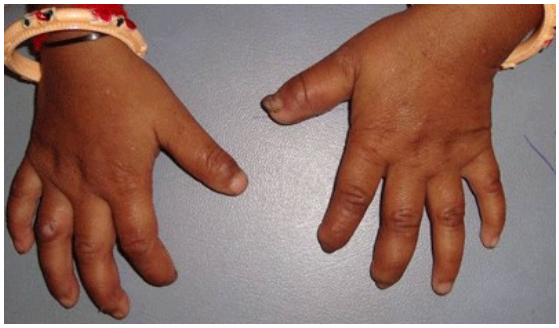

Fig 2: Patient's hands showing dystrophic finger nails

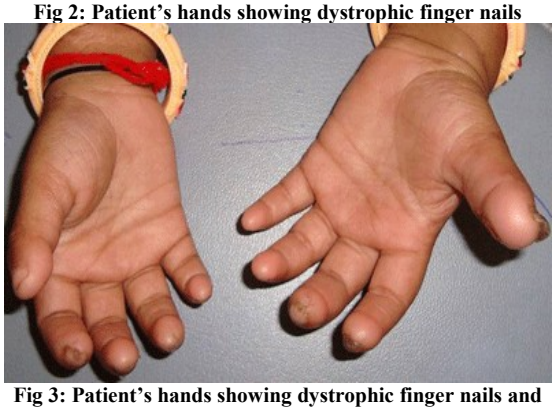

Fig 3: Patient's hands showing dystrophic finger nails and wounded left thumb

Fig 4: Ulcer over the sole of the right big toe

Fig 5: Traumatic erosion over the chin

Extra oral examination showed presence of abraded wound over the symphysis region ranging 3x3cms in size (Fig 5). The surface appeared erythematous and was non tender on palpation. The facial bones appeared normal. Intra oral examination revealed missing 71, 72, 73, 81, 82 and mobility of 61. Decayed 51, 52, 54, 55, 64, 65, 74, 75, 84 and 85 were noted.

Based on the history and clinical findings a provisional diagnosis of avulsed 71, 72, 73, 81, 82; traumatic erosion over the chin and congenital insensitivity to pain were considered with the following differential diagnoses: Lesch-Nyhan syndrome, familial dysautonomia and familial amyloidosis.

Patient was subjected to radiological investigations, to rule out any fractures involving the facial skeleton. As she was uncooperative for Orthopantamogram (OPG), it was decided to take a Posteroanterior (PA) view of skull with the patients head stabilized by her attender. The PA view did not show any pathological changes (Fig 6).

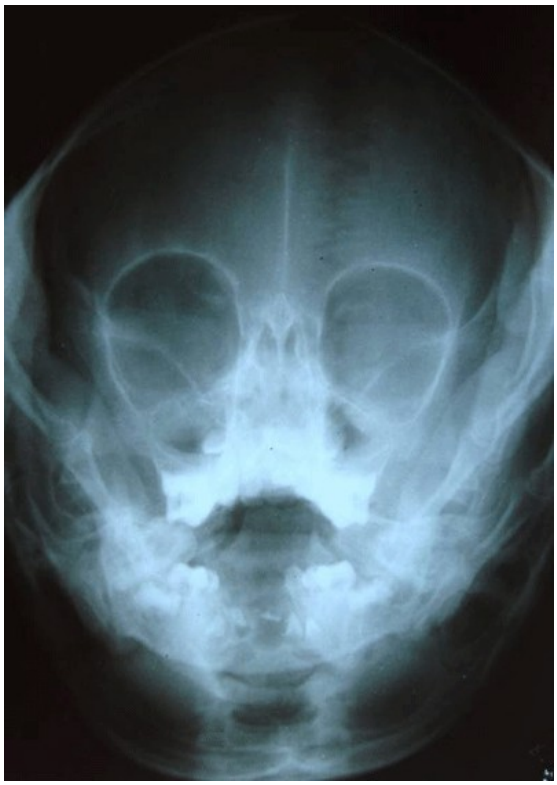

Fig 6: Posterioanterior view of skull

Then the patient was referred to a general physician and neurologist. The patient was tested for measurement of pain intensity, hot and cold. The response was noted in a visual analog scale. The testing materials included a pin (of varying diameter- which was pricked on the sole of the feet), ice cubes and hot instrument. Patient responded immediately to hot and cold and for tactile sensation but not for pain. Sensory examinations including light torch, proprioception and vibration were intact. Deep tendon and corneal reflexes were present. There were no autonomic disturbances – she sweated normally and produced tears. The uric acid levels were normal.

The history, clinical and investigatory findings confirmed our diagnosis of congenital insensitivity to pain.

The patient parents and relatives were educated and cautioned about the condition. For the traumatic erosion- topical antibacterial application thrice daily (to prevent secondary infection) was advised and the patient was referred to department of pedodontics for further dental management and complete dental rehabilitation.

Discussion:

Congenital insensitivity to pain is a rare, autosomal recessive sensory neuropathy first reported by Dearborn in 1932.(2) The disorder is characterized by absence of reaction to painful stimuli, self-mutilating behavior. It is diagnosed early in the childhood, as the affected child rarely cry of pain.

There are atleast two common forms of congenital insensitivity to pain - Congenital insensitivity to pain (HSAN - V) and congenital insensitivity to pain with anhidrosis (HSAN - IV). Our case falls under the former category which is also called as Congenital analgia or Congenital analgesia, congenital asymbolia and even some prefer the term indifference instead of insensitivity.(3) The type V is characterized by disorder in pain perception with no other neurologic deficit, whereas HSAN – IV first reported by Nishida in 1951, is a severe form which is characterized by disorders of pain perception and thermoregulation.(4)

The cause for the disorder is not clear. There are various studies available in the literature concern to CIPA, while studies related to congenital insensitivity to pain are meager in the literature. There are many questions and assumptions prevail regarding this condition. More recently, a study claims mutation in the $Na_v1.7$ encoded by the *SCN9A* gene located on the chromosome 2q24.3 causes inability to experience pain.(5)

Congenital insensitivity to pain is an Autosomal recessive disorder with no ethnic distribution. As seen in our case half of the cases are reported in children born to consanguineous marriages.(6) The clinical features of congenital insensitivity to pain vary markedly. The insensitivity to pain is profound and may lead to repeated trauma and self-mutilation as noted in the present case. An additional finding that may be noted in these patients may include fractures that are slow to heal and frequently go on to develop osteomyelitis and charcot joints.(7)

Diagnosis of congenital insensitivity to pain should be done with atmost caution as the clinical findings can resemble CIPA, which is a severe entity. In association with pain the cardinal feature of CIPA is absent or markedly decreased sweating causing episodic fevers and extreme hyperpyrexia which is usually the earliest sign. Anhidrosis also contributes to the thick and calloused appearance of the skin with lichenification of palms, dystrophic nails, and areas of hypotrichosis on the scalp.(8) The individuals affected with CIPA usually do not live past 3 years of age; if they make it many do not past 25 years. The reason behind this is probably due to anhidrosis leading to recurrent fevers that are unexplained and can be fatal due to hyperthermia. Patients who survive their infant years have a high chance of acquiring mental retardation. This may be due to the fact that higher the body temperature goes the more harmful bacteria and virus attacks can occur leading to swelling of blood vessels causing aneurysms, all of which a CIPA patient will not even feel.

The other condition that can resemble CIP is familial dysautonomia (Riley- Day syndrome). The clinical features include absence of pain, lacrimation and fungiform papillae. The condition also comprise of postural hypotension, hyperhidrosis, nystagmus, poor muscle tone, co-ordination, and lack of reflexes. (3) Apart from insensitivity to pain, the present case did not have any other features.

Self mutilating behavior can also be a part of Lesch-Nyhan syndrome, which is a rare, inherited disorder caused by a deficiency of the enzyme hypoxanthine-guanine phosphoribosyltransferase (HPRT). The diagnosis of Lesch-Nyhan syndrome is based initially on the self-mutilating behaviors that begin in the second year of life.(9) In some cases the first symptom is related to overproduction of uric acid; the parents notice "orange sand" in the child's diapers. The "sand" is actually crystals of uric acid tinged with blood.(10) Abnormal high uric acid levels can cause sodium urate crystals to form in the joints, kidneys, central nervous system leading to gout-like swelling in the joints, failure to crawl and walk at the usual ages. Measuring the amount of uric acid in a person's blood or urine can be helpful in diagnosis of Lesch-Nyhan syndrome. The present case did not have any aforementioned symptoms and the uric acid level was normal.

Andrade type of familial amyloidosis should also be considered while evaluating CIP, as affected patient's present with preferential loss of pain. However, this neuropathy can be readily differentiated as these patients pose loss of other types of sensations and the tendon reflexes.(3)

The history and clinical findings of CIP usually leads to diagnosis. Neurological studies such as electroencephalogram, cerebrospinal fluid, and sensory and motor nerve conduction studies were tried and found normal in majority of patients.(11) When a nerve is biopsied, the histological findings may include a complete absence of non-myelinated and small myelinated nerve fibers in the dorsal root ganglia.(12)

There is no single gold standard treatment available for this condition. Reports suggest naloxone and naltrexone can be used to reverse the analgesia.(6) The above treatment option however lacks evidence and further support. Hence, most treatments are narrow down to other associated conditions.

Dentists who encounter such patients should bear in mind, that although they lack the sensation of pain some may have tactile hyperesthesia. Hence anesthesia is needed to reduce the apprehension, for relaxation of the muscle thereby to avoid any accidental fractures.

Conclusion:

Congenital insensitivity to pain is a rare group of neuropathic disorder. The varied manifestations, the lack of validate investigation and treatment modalities make it a distinctive entity. This necessitates the oral physician to be cognizable with it and ample steps should be taken to reduce the paranoia and post traumatic complications associated with it.

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