CASE REPORT: CONGENITAL INSENSITIVITY TO PAIN WITHOUT ANHIDROSIS

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ABSTRACT:
Congenital Insensitivity to Pain (CIP) is a condition that manifests from birth and impairs the ability to perceive physical pain. Individuals affected by CIP are entirely devoid of the ability to feel pain in any part of their body. While they still experience discriminative touch, these patients cannot perceive sensations that would typically be described as painful by individuals with a normally functioning sensory and autonomic nervous system. Furthermore, they are unable to discern extreme temperatures, whether hot or cold. CIP is classified as a type of peripheral neuropathy because it impacts the peripheral nervous system. This intricate network connects the brain and spinal cord to muscles and sensory cells responsible for detecting various sensations, including touch, smell, and pain. In this report, we present a case of Congenital Insensitivity to Pain in a 6-year-old boy.
Keywords: CIP, SCN9A, Peripheral Nervous system, chromosome 2q24.3.

INTRODUCTION

Congenital insensitivity to pain with anhidrosis comes under the umbrella of one the rarest neurogenetic disorders in which there are mutations in the tyrosine receptor kinase 1 gene because of which loss of function of neuron occurs [1,2]. The term "congenital pure analgesia "was used for this disorder by Dearborn [3]. Studies have shown that NGF-tyrosine receptor kinase 1 is very major in the sensation of pain, itching, and inflammation [4]. The congenital insensitivity to pain without anhidrosis patient's typical characteristics is that patients have insensitivity to pain, intellectual disability, and inability to sweat. Because of insensitivity to pain, this usually results in repeated injuries that are bone fractures, oral injuries such as biting their tongue, bruises all over the body, scars, and joint dislocations. In cold weather, there is an increased risk of getting hypothermia [5]. While the other sensations such as sense of position, touch, and vibration are normal [6]. We report a 6-year-old boy with congenital insensitivity to pain with anhidrosis

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CASE REPORT

I was referred a six-year-old boy from Nangarhar Province in Afghanistan. His parents shared their concerns about his apparent inability to experience pain, a phenomenon they first noticed when he was a baby. Remarkably, he did not cry when receiving injections during infancy. As he began teething, the situation became even more apparent. He would repeatedly bite his tongue, leading to bleeding and eventually causing the tip of his tongue to take on a forked appearance, as shown in Figure 1. Additionally, the child frequently suffered injuries to his toes and fingers, some of which had occurred repeatedly. The extent of his condition became evident when he lost several upper front incisor teeth and canines, as depicted in Figure 1. Remarkably, the boy had also fractured his right femur at some point, yet he never complained of pain. It was only when his parents noticed his limping that they took him to a doctor, and an X-ray revealed the fracture, as illustrated in Figure 2A and Figure 2B.

As a result of his insensitivity to pain, the patient endured additional injuries, including burns, as he couldn't differentiate extreme temperatures from normal ones. This led to repeated burn injuries on his toes and hands, as evidenced in Figure 3, showcasing his indifference to thermal stimuli. Some studies have suggested that the ipsilateral insular cortex may serve as the primary thermal receiving center. Furthermore, the patient had a history of diarrhea and was anemic. Fortunately, aside from subnormal thermal reception, all other sensory modalities appeared normal.

Additionally, a thorough physical examination ruled out accompanying disorders such as anhidrosis. This process allowed us to rule out other conditions similar to Congenital Insensitivity to Pain, including those within the HSAN (Hereditary Sensory and Autonomic Neuropathy) groups. Based on his medical history, we arrived at the diagnosis of CIP.

Note that the patient's images were obtained and shared with the consent of his father.

The parents of the child noted that his two sisters were unaffected by the condition. However, interestingly, the child's cousin on the paternal side (the aunt's son) also presents with a similar condition. This suggests a familial component to the condition. The traumas mentioned earlier are visually evident in the figures provided below. These images capture the patient's injuries, including the instance where he bit off the tip of his tongue and sustained multiple injuries to his toes and fingers. Notably, the injury to his toes is clearly visible. For reference, the figures/images mentioned below are attached:

Photographic Documentation:

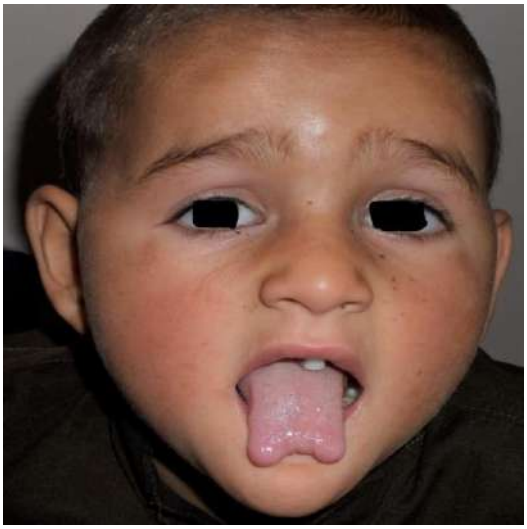

Figure 2 image shows a pronged/forked tongue due to self-biting.

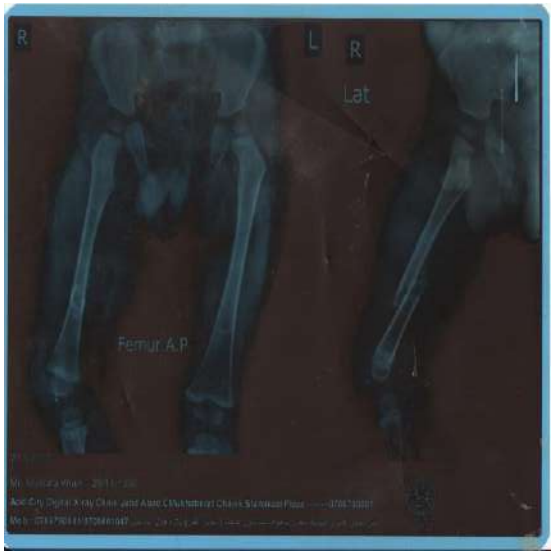

Figure 1a. Figure 2a. The fractured Femur is visible in the X-Ray, but the patient was unable to perceive pain normally associated with fractured bones.

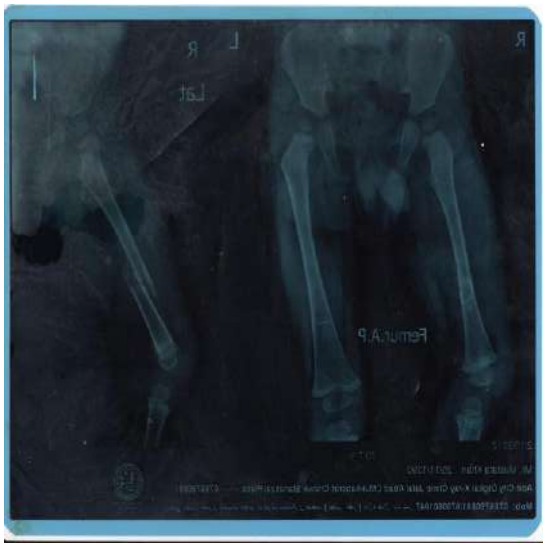

Figure 2b: A.P of the fractured femur that

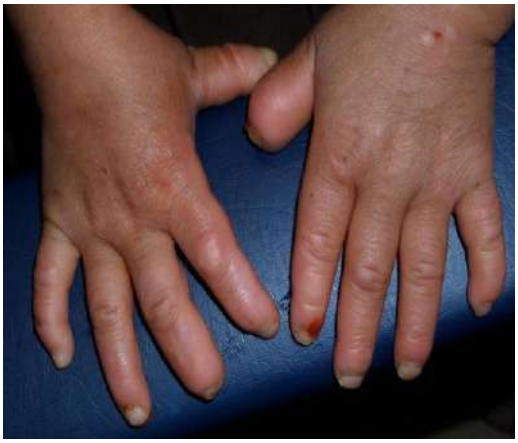

Figure 3: Swelling due to repeated trauma, ulcerations, and loss of distal tissue can be seen

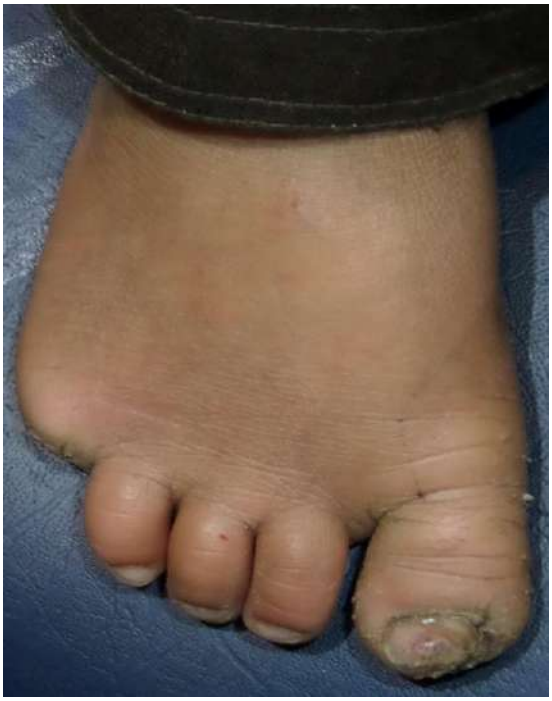

Figure 4: Loss of the fifth toe due to repeated trauma and swelling caused by trauma-induced opportunistic Infections

DISCUSSION:

CIPA, initially identified by Swanson in 1963, was first observed in two brothers who displayed a deficiency in temperature perception and an inability to feel pain [7]. Notably, consanguineous marriages have been associated with half of the recorded occurrences [8]. The tongue and lips are the body parts most frequently affected by CIPA. As the primary incisors emerge, self-injury to the tongue often begins, leading to ulcers on the ventral surface, a bifid tongue, or even the loss of the tongue tip. However, it's important to note that the functions of taste buds remain normal [9]. In orthopedics, the most prevalent symptoms include numerous fractures, particularly in long bones [10]. Dermatological symptoms encompass a multitude of scars, wounds, burns, and ulcerated fingers. Biting fingertips can lead to conditions such as phalangeal osteomyelitis, nail inadequacy, and digital shortening [9, 11]. CIPA is recognized for causing significant joint deterioration from the earliest stages of a patient's life, a phenomenon known as "Charcot's joint," which results from insensitivity to joint pain stimuli. Moreover, individuals with CIPA often experience severe spine degeneration, leading to deformities and neurological impairments [12, 13]. Due to its rarity and the absence of obvious diagnostic testing, HSAN (Hereditary Sensory and Autonomic Neuropathy) is a challenging condition to diagnose [14].

Our diagnosis of CIP was supported by a thorough examination of the patient's other sensory modalities, which were found to be functioning normally. Additionally, the absence of anosmia or anhidrosis, which are characteristic features of certain disorders within the HSAN (Hereditary Sensory and Autonomic Neuropathy) group, helped differentiate CIP from other potential conditions. Despite advancements in our understanding, CIP remains a condition about which we have limited knowledge. This lack of understanding makes both diagnosis and treatment challenging. Some potential therapeutic options, such as Naltrexone and Naloxone, have been suggested to enhance the patient's quality of life, although their effectiveness varies. These drugs, classified as opioid antagonists, work by unblocking pain receptors, as opioids typically block them. Parents can also take proactive measures to create a safer environment for their children, reducing the frequency and severity of injuries. Furthermore, by examining and comparing similar case reports, we may enhance our ability to differentiate and diagnose CIP more easily. Increasing awareness of this condition within the medical community is crucial to further our understanding and improve care for individuals affected by CIP.

CONCLUSION

Congenital insensitivity to pain is a complicated condition that involves a lot more than just a lack of pain sensitivity. The systemic effects lead to unique patterns of soft tissue presentations, joint degeneration, and reduced bone and soft tissue repair, as well as an elevated risk of infection. In this syndrome, upper limb symptoms are less frequent than lower limb ones. Early intervention in cases of digital ulceration by debridement and soft tissue cover may prevent amputation. HSANs are currently untreated. However, it is crucial to stop patients from self-mutilating. The removal of sharp tooth surfaces caused by grinding (enameloplasty), using mouth guards tongue guards, or other devices to avoid harm to the tongue.

CONFLICT OF INTEREST: None

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