CASE REPORT

CONGENITAL INSENSITIVITY TO PAIN WITH ANHYDROSIS

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ABSTRACT

Congenital insensitivity to pain with anhydrosis (CIPA) is a rare genetic disorder characterized by inability to feel pain and temperature, and decreased or absent sweating. Familiarity to this condition is important to avoid misdiagnosing it with leprosy and other peripheral nerve diseases. An inability to feel pain may lead to repeated self-trauma (tongue, lips and finger tips) ultimately leading to self-mutilation. We report a case of CIPA who presented with a non-healing ulcer on the heel with a history of fingers being eaten by the patient himself along with multiple fractures in different bones and inability to sweat.

KEY WORDS: Congenital Pain Insensitivity; Hereditory Sensory and Autonomic Neuropathies; Pain; Sweating; Anhidrosis; Hypohydrosis; Self-mutilation.

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INTRODUCTION

Congenital insensitivity to pain with anhydrosis (CIPA) is a rare autosomal recessive genetic disorder. Its exact prevalence is not known. In this condition genetic mutation occurs at NTRK1 gene which is inherited as autosomal recessive disorder. NTRK1 gene encodes a signal which is involved in the formation of protein responsible to carry pain, touch and temperature sensation along the sensory neurons. Loss of function of sensory neurons also causes sweat glands not to function normally.[1]

CIPA commonly presents with orthopedic problems and hyperpyrexia. Its sign and symptoms usually appear at birth or early infancy, because of the inability to feel pain which is the main feature of this condition usually resulting in unintentional self-trauma. Skin injuries, bone fractures, chronic oeteomylistis and Charcot's joints are the common presentations. Anhydrosis or loss of sweating is also an important feature of this condition resulting in repeated episode of hyperpyrexia and febrile fits. This feature is the frequent cause of mortality in early life in this condition. These patients may also have behavioral, emotional and intellectual problem.[2-5]

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The management of CIPA is mostly supportive to prevent self-injury, control body temperature and treat the orthopedic problems. As the condition is autosomal recessive, genetic counseling is also important.[2] It can be fatal during the early years of life due to hyperpyrexia and fits if not properly diagnosed and treated. In spite of all these dangers, patients with CIPA can live up to the adult life if orthopedic problems and temperature control are managed properly.[6,7]

CASE REPORT

A six years old boy presented to Out Patient Department at the National Orthopedic Hospital, Bahawalpur, Pakistan with complaint of non-healing ulcer on the left heal for the last four years. (Fig. 1) Patient was treated by many local doctors for ulcer but the condition didn't improve. The patient's mother also gave the history of self-eating (auto mutilation) because of unrealized injury and on examination his fingers of both hands were smaller. (Fig. 2)

Patient also had fractures of long bones with some callus formation. (Fig. 3) There was also evidence of chronic osteomyelitis of calcaneum. (Fig. 4) The patient also had loss of most of his teeth with mandible lysis and dental deformity as seen in the skull radiograph. (Fig. 5)

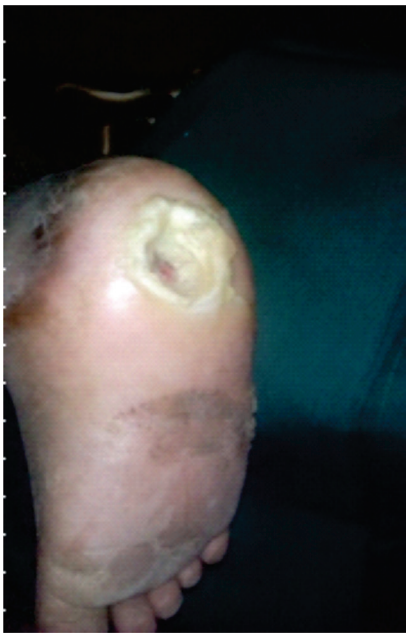

Figure 1: Non-healing ulcer on the sole of a boy with congenital insensitivity to pain with anhydrosis (CIPA).

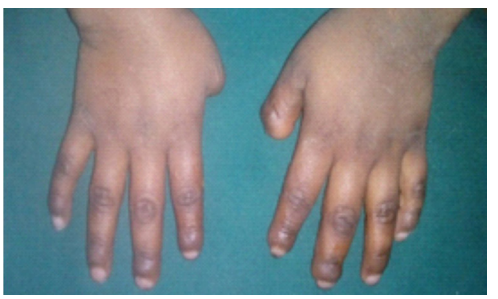

Figure 2: Showing shortening of fingers in patient with congenital insensitivity to pain with anhydrosis (CIPA).

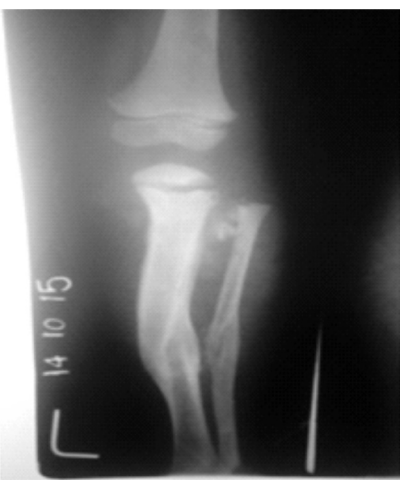

Figure 3: X-ray showing fractures of left tibia and fibula with some callus formation in a boy with congenital insensitivity to pain with anhydrosis (CIPA).

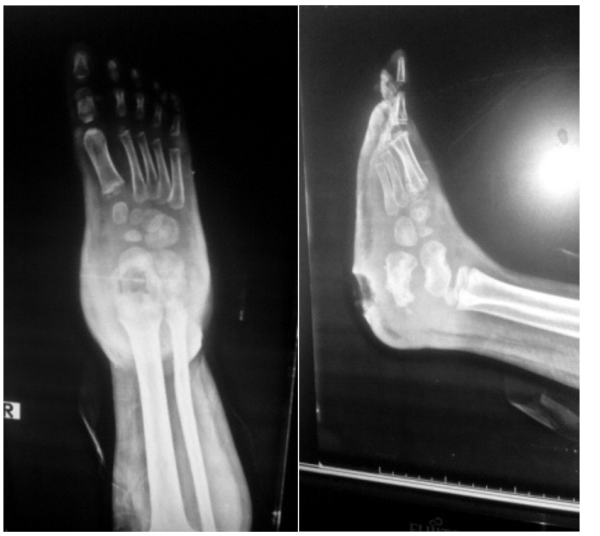

Figure 4: X-ray AP and lateral view of foot showing osteomyelitis and ulceration of the heel in a boy with congenital insensitivity to pain with anhydrosis (CIPA).

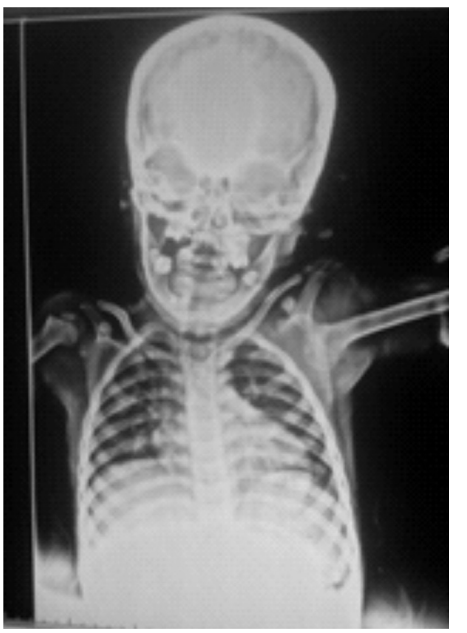

Figure 5: X-ray skull showing loss of teeth and mandible lysis in a boy with congenital insensitivity to pain with anhydrosis (CIPA).

DISCUSSION

Congenital insensitivity to pain with anhydrosis is a rare autosomal disorder. Familiarity to this condition is important among orthopedic surgeons to avoid misdiagnosing it with leprosy and other peripheral nerve diseases. An inability to feel pain may lead to repeated self-trauma (tongue, lips and finger tips) ultimately leading to self-mutilation. CIPA commonly presents with orthopedic problems and

hyperpyrexia. Skin injuries, bone fractures, chronic oeteomylistis and Charcot's joints are the common presentations.[6]

The management of CIPA is mostly supportive to prevent self-injury, control body temperature and treat the orthopedic problems. CIPA can be fatal during the early years of life due to hyperpyrexia and fits if not properly diagnosed and treated. Patients with CIPA can live up to the adult life if orthopedic problems and temperature control are managed properly. Mental issues in CIPA patients should be helped with special schooling. Patients must be checked for cuts, bruises, self-mutilation, fractures and ulcers. They must regularly visit the doctor for proper check-ups.[7]

The aim of reporting this problem was to increase the awareness among doctors about this disease. Unfortunately, in our country genetic testing is too costly and also not easily available. These tests were not performed in this patient.

CONCLUSION

The syndrome of congenital insensitivity to pain with anhydrosis (CIPA) can be diagnosed clinically and confirmed by genetic testing. It can be managed with proper orthopedic treatment and controlling temperature.

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CONFLICT OF INTEREST
Authors declare no conflict of interest.
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None declared.