International Journal of Science and Research (IJSR)
ISSN: 2319-7064
SJIF (2022): 7.942

Congenital Pain Insensitivity CIPA: A Comprehensive Case Report, Clinical Features, and Management Strategies in an 11-Year-Old Patient

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Abstract: ***This is a case of congenital pain insensitivity. This case projects the main presenting complaints and clinical features and psychological condition of the patient. Congenital insensitivity to pain with anhidrosis (CIPA) is characterized by recurrent episodes of infections and unexplained fever, anhidrosis (inability to sweat), and absence of reaction to noxious stimuli, self-mutilating behavior, mental retardation and damages to oral structures. Methods of early diagnosis like genetic typing can help us understand the course of disease easier.***

Keywords: Congenital Pain Insensitivity, Psychological Condition, Nociception, NGF-TRKA Signaling, Staphylococcus Aureus Infection, SCN9A and SCN11A Mutations

1. Introduction

Congenital pain insensitivity is a very rare and dangerous condition. This case report discusses the clinical features as well as diagnosis and Management a 11-year-old girl who came to the hospital with medical collateral ligament injuryin and inability to walk due to limb length discrepancy. This mendelian genetic disorder. Although only a small number of causative conditions and genes are known, most have led to profound insights into human nociception. CIP gene discovery is catalyzing the manufacture of completely new classes of analgesics, and these are needed as alternatives to synthetic highly potent opioids. The importance of nerve growth factor-tropomyosin receptor kinase A (NGF-TRKA) signaling for nociceptor genesis and subsequent pain sensing. New analgesics can be generated from knowledge of the NGF-TRKA nociceptor pathway. Increased susceptibility to Staphylococcus aureus infection is a consequence of deficient NGF-TRKA signaling. Mutations in the voltage-gated sodium channels SCN9A and SCN11A can cause congenital painlessness, and in contradistinction, other mutations can cause episodic neuropathic pain. SCN9A/Nav1.7 is an analgesic target. SCN11A/Nav1.9 is unlikely to be an analgesic target. There are further Mendelian causes of painlessness to be discovered.

2. Case Presentation

A 11 year old girl was brought to the orthopaedicout-patient department with a chief complaint of inability to walk and limb deformity noticed by her parents since one week. Self mutilating injuries like tongue biting and injuries to fingers and toes. On thorough history taking patient revealed that the problem started when she was 4 years old when she presented to the opd with a child complaint of fever and cold for which doctor assessed her and found out that she was irresponsible to pain and referred her to higher centers. On neurological examination bulk, tone and reflexes all were normal. There was no history familial history or hereditary causes in the family. But parents showed consanginous marriage making the child more susceptible to genetic disorders. Brain CT was ordered lumbar puncture was done all showed normal findings. Electro encephalogram was taken to check the activity of cortex and was normal. Further patient was advised genetic study for which she refused to get it done and so was treated for cough and cold and was discharged. Few days later patient presented with a chief complaint of limb shortening and inability to walk for which patient was underwent bone graft surgery and in her third admission patient came with a history of discharging sinus and a culture was done and identified as osteomyelitis which was treated with appropriate antibiotics. In this way patient has a history of recurrent admissions in the hospital due to pain insensitivity and underwent multiple surgeries for them. Recent admission was few days back due to medical collateral ligament injury which was diagnosed after taking an x ray and fixation of the same was done and patient was discharged with stable vitals and with appropriate post discharge advise.

Figures:

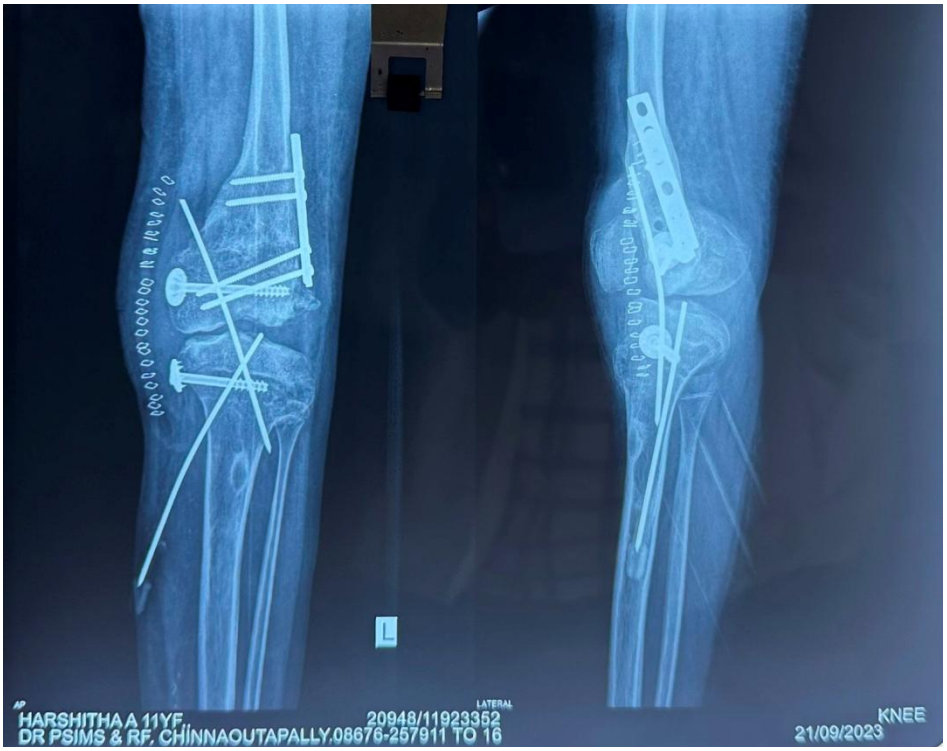

Figure 1: Postero-anterior view plain X-ray showing fixing of mcl tear with screws and plates.

3. Discussion

Congenital pain insensitivity is an autosomal recessive disease [1], all of our patients had consanguineous parents. There are few cases of insensitivity to pain described in the literature, but there is no standard treatment [2]. Patients with CIPA can live a fairly normal life [4, 5, 6]. Three of our patients had mild mental retardation and attended special schools. Difficulty in walking should be evaluated for inherited form of ataxia as some rare types of ataxia arise via family (6). However, there are certain precautions that need to be taken in order to ensure the safety of the patient. The patient must constantly check for cuts, bruises, self-mutilations, and other possible unfelt injuries. Also, they must regularly visit their doctor to receive a full work up to be sure that they do not have any insensitive internal problems that can be fatal. Understanding the main pathogenesis and characteristic of the disease will help in holistic management of the case and better prognosis for the patient.

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