CASE REPORT

Congenital insensitivity to pain with anhidrosis: a case report from Pakistan and literature review

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ABSTRACT

Congenital insensitivity to pain with anhidrosis is a rare disease with an autosomal recessive inheritance. The patients present in early childhood with frequent episodes of fever and absence of sweating. Painless fractures, bruises and cuts are quite common. Defective lacrimation and mental retardation are strongly diagnostic. Repeated injuries often lead to a reduced life expectancy. The diagnosis depends on relevant clinical features, abnormal sensory response on nerve conduction studies and nerve biopsy showing loss of the unmyelinated and small myelinated fibers. We report here a 5 year old female child who presented for electrodiagnostic evaluation and was found to have this disease.

Key words: Congenital insensitivity to pain; Anhidrosis; Electrodiagnostic studies

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INTRODUCTION

*C*ongenital *i*nsensitivity to *p*ain (CIP) is a rare inherited disorder presenting in early childhood and characterized by insensitivity to pain resulting in unusual injuries due to repetitive self-inflicted damage. It follows an autosomal recessive inheritance.[1] The lack of pain often leads to repetitive wounds, bruises, broken bones, and rapidly degenerating joints that may go undetected. Systemic anhidrosis and mental retardation are characteristic findings but present in a severe form of CIP called the *c*ongenital *i*nsensitivity to *p*ain with *a*nhidrosis (CIPA).[2] The diagnosis depends on clinical features, biochemical, electrophysiological, histopathological and genetic testing. We report here a case of a 5 year old female child who presented to Armed Forces Institute of Rehabilitation Medicine, Rawalpindi and was diagnosed to have this disease upon electrodiagnostic (EDX) evaluation.

CASE REPORT

A 5 years old female child presented with repeated bouts of febrile illnesses due to chest infections since she was three months of age. Her parents reported dryness of her skin during repeated fevers. Parents also reported several episodes of self-biting of tongue, lip and fingers for the last 4 years. Consequently, the child had frequent scarring and ulcerations of lower lip and tongue and damaged fingers and left thumb. Her prenatal and birth histories were unremarkable. Family history revealed that her parents had consanguineous marriage. Her sister was absolutely normal.

On examination, she had a bitten tongue and some scarring of her lower lip (Figure 1). Tips of her right thumb, index and middle fingers were disfigured (Figure 2). The skin around right big toe was thick, hard and cracked around first metatarsophalangeal joint (Figure 3). She did not show withdrawal response to pinprick or hot and cold test tubes. Vibration and position sensations were intact in all limbs. The muscle tone and deep tendon reflexes were also normal.

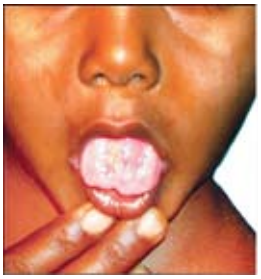

Figure 1: Showing a bitten tongue and scarring of lower lip

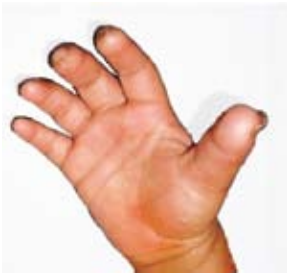

Figure 2: Disfigured tips of fingers

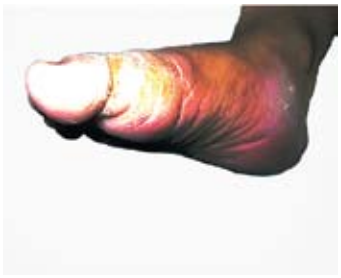

Figure 3: Thick, hard and cracked skin around big toe

Her serum uric acid levels were normal. EDX evaluation was carried out on XLTEK Neuromax 1004® electromyography (EMG) unit using surface electrodes for *n*erve *c*onduction *s*tudies (NCS) and concentric needle electrodes for EMG. Motor NCS revealed normal compound muscle action potential amplitudes, distal motor latencies, and conduction velocities in median, ulnar and radial nerves in upper limbs and common peroneal and tibial nerves in lower limbs. Her sensory NCS revealed reduced *s*ensory *n*erve *a*ction *p*otential (SNAP) amplitudes but normal conduction velocities and sensory peak latencies in median, ulnar and radial nerves in upper limbs. Sural SNAP amplitudes were absent bilaterally. The child did not respond to electrical stimulation on NCS and needle insertion during EMG. The EMG showed no involuntary activity and normal motor unit action potentials, morphology and recruitment in all sampled muscles of the four limbs.

Keeping in view the clinical history and examination, an EDX impression of chronic sensory neuropathy was noted. An advice for child's nerve biopsy to confirm CIPA was refused by parents owing to financial constraints. They were however counselled about the risks and lines of management. The genetic testing for CIPA was not done as the facility is not available in Pakistan.

DISCUSSION

It was in 1932 when Dearborn used the term "congenital general pure analgesia" to describe individuals with CIP.[3] The initial diagnostic criteria were given by Thrush in 1973 and included absence of pain sensations since birth, involvement of whole body and preservation of other sensory modalities and deep tendon reflexes.[4] Later on, the disorder was given the acronym of hereditary sensory and autonomic neuropathy (HSAN). HSAN has five subtypes based on the hereditary pattern, the clinical features and the most affected neuronal systems. HSAN type-IV is also called CIPA and is characterized by disordered pain perception and thermoregulation.[5]

The primary pathology is the abnormal neurons responsible for the transmission of pain and temperature sensations in the autonomic nerves. Sural nerve biopsy performed in these patients has shown a severe reduction of unmyelinated fibers in peripheral nerves, deficient C and A-δ fiber endings in the epidermis, and absent or hypoplastic denervated dermal sweat glands.[1,5] The cause for the disorder is not clear. However, a recent study claimed that mutation in the $Na_v1.7$ encoded by the SCN9A gene located on the chromosome 2q24.3 causes development of this syndrome.[6] Unluckily, the genetic studies could not be done in our patient.

CIPA becomes evident in early childhood with frequent episodes of elevated body temperature and absence of sweating as in our patient.[1] Self-mutilation starts at the time of initial tooth eruption with biting of the tongue and digits.[4] Painless fractures, bruises, and cuts are quite common. Defective or absent lacrimation and mental retardation are strongly supportive of diagnosis when present.[2] Charcot's joints may result from a major or repeated minor trauma.[4] Loss of sweating also leads to a thick and calloused appearance of the skin particularly in the palms, areas of reduced hair on the scalp and dystrophic nails.[6] Repeated injuries often lead to a reduced life expectancy in people with CIP.[1] Diagnosis depends on relevant clinical features, abnormal sensory response on NCS and characteristic findings on peripheral nerve biopsy.

There are few conditions that may be confused with CIPA due to pain insensitivity at initial presentation. The congenital indifference to pain presents with loss of pain sensitivity but without dysautonomic symptoms. The NCS and nerve biopsy are normal.[1] Familial dysautonomia (HSAN-Type-III) also comprises of nystagmus, hypotonia, areflexia and poor coordination.[6] Asymbolia for pain usually has damaged insular cortex evident on magnetic resonance imaging of brain and NCS/EMG are always normal.[7] CIPA does not have a cure at present. Reports suggest that naloxone and naltrexone can be used to reverse the analgesia but lack evidence and further support.[6] Counseling of the family and school personnel for the prevention of injuries should be the priority. Early diagnosis is very important for the prevention and treatment of various complications.

Conflict of interest: Nil

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'My Most Memorable Patient'®

'Can't ventilate, can't intubate' in a patient with tracheal stenosis

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Can't ventilate, can't intubate (CVCI) situation is a rare anaesthetic emergency requiring rapid and decisive management. This condition obviously has life-threatening implications and must be resolved within minutes, to avoid hypoxic brain damage or death. Repeated attempts at airway manipulations are common cause of airway deterioration and morbidity. The situation becomes worst when trachea is already compromised with a pre-existing pathology. The incidence is difficult to estimate; however, recent work has suggested an incidence during all anaesthetics of one in 50 000.[1] American Society of Anesthesiologists (ASA)[2] and Difficult Airway Society (DAS)[3] have published guidelines on the management of CVCI situations. The DAS has produced a management algorithm[3] which suggests rescue techniques like cricothyroidotomy for the CVCI situation. Emergency invasive airway access e.g. surgical airway, jet ventilation, and retrograde intubation must be kept in mind.

My patient was just an ordinary one in any way; a 4 years old girl, weighing 10 kg, average built with stable hemodynamics, who presented to the ENT department with progressively increasing difficulty in breathing with noisy sounds for the last one month. No history of fever, cough, trauma, asthma, tuberculosis found. Patient had had appendectomy one month ago under GA with endotracheal intubation. Previous anesthetic management record was not available, and there was no complaint of any postop complication. ENT surgeon planned for emergency laryngotracheoscopy under GA. Expected procedure duration was of 10 minutes. Difficult airway was not anticipated, so a routine general anesthesia management plan was chalked out; premedication with inj. glycopyrrolate 0.04 mg and inj. fentanyl 20 μg, preoxygenation with 100% oxygen, induction with ketamine and maintenance with sevoflurane in oxygen with assisted BMV. Inj. succinylcholine 20 mg was given just before laryngotracheoscopy with ventilation in between apneic phases. Laryngotracheoscopy revealed tracheal stenosis about 2 to 2.5 cm below vocal cords and planned for dilatation on elective basis.

The real drama was unleashed during recovery when spontaneous breathing was found to be inadequate and laborious and patient desaturated progressively despite BMV with 100% oxygen. Treatment on the line of laryngospasm started immediately but not effective and SpO_2 rapidly fell. Help was called and two senior anesthesiologists arrived, BMV with CPAP could not raise saturation >85% with 100% oxygen. Anesthesia deepened and intubation tried. Surprisingly, the glottis was seen wide open but even the smallest sized ETT (2.5 mm ID) could not be negotiated beyond stenosed part.

Patient's condition deteriorated and saturation could not be corrected. LMA was inserted after laryngospasm was ruled out but ventilation was still inadequate leaving the patient in the face of impending death. Options of cricothyroidotomy and conventional tracheostomy were considered of doubtful benefit in view of the anatomic location of stenosis and its unknown lower level. But what other options were available to us? The child was rapidly drifting towards death. So after with good wish and best hope emergency low tracheostomy was done by the ENT surgeon. It turned the tide and saturation started improving to the relief of us all. Patient reversed uneventfully and was shifted to Pediatric Critical Care Unit after a prolonged period of observation on the table.

The patient was saved, but not the anesthesia and the operating team. The parents of the child refused to accept any explanations from surgical and anesthesia colleagues and compelled us to remove the tracheostomy tube. All reasoning fell flat on them. In fact during the battle for life for the child, there was no time to inform them or take consent regarding possible tracheostomy. Ultimately, this battle was also won with gentle handling and perseverance.

Patient recovered well with no neurological deficit noted clinically. Tracheal dilatation was done the following week under GA with tracheostomy tube in situ. Decannulation was done subsequently and she was discharged.

Low tracheostomy saved our patient and us, and the parents went home with a smile on face; but this incidence taught us few lessons; no case is a small case in anesthetic practice; the consent is all very necessary; and finally, documentation is very, very important.

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