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HUMANITARIAN AND RESOURCE-LIMITED SETTING

Painless: a case of congenital insensitivity to pain in a 5-year-old male

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Abstract

Background: several genetic disorders are known to be associated with congenital insensitivity to pain (CIP), a term often used to describe an impaired ability to perceive the type, intensity and quality of noxious stimuli. Children with CIP often injure themselves severely. The injury can go unnoticed or be misdiagnosed as child abuse because it is associated with multiple and recurrent injuries which may result in permanent damage. Patient findings: we report the case of a 5-year-old boy with a history of showing no signs of pain when exposed to accidental injuries such as trauma, burns or secondary chronic lesions. Conclusion: child abuse has a much higher occurrence rate than rare neuropathies such as the one we describe. However, CIP should be considered as a diagnosis in any child presenting with a history of poor or absent responses to painful stimuli.

INTRODUCTION

Hereditary sensory and autonomic neuropathy (HSAN) is a group of genetic disorders involving varying sensory and autonomic dysfunction [1]. Several genetic disorders are known to be associated with congenital insensitivity to pain (CIP), a term often used to describe an impaired ability to perceive the type, intensity and quality of noxious stimuli [2].

Dearborn described the condition as 'congenital pure analgesia' in 1932. Swanson thoroughly studied the condition in 1963 and Mardy first reported the lack of innervations in eccrine sweat glands affecting the patient's ability to sweat. 'Painless whitlows', 'mal perforant du pied' and 'Morvan syndrome' are some of the many names used to describe a wide range of conditions that are today grouped under HSAN [3]. Van Dyck *et al.* classified HSAN into five types according to the mode of inheritance and clinical features. This classification has been modified with subtyping, addition of new types and discovery of related genes [4]. The condition is extremely rare with mere 80 cases documented, and 300 cases reported in the medical literature throughout the world [3]. However, the true prevalence of HSAN-IV and V is not well established [4] (Table 1).

CASE REPORT

A 5-year-old male living in a rural area in the Middle East visited the emergency room department with the chief complaint of hand-burning by a heater, but showing no signs of pain.

The patient was the third child of a consanguineous marriage (first degree cousins), and the only boy with two older sisters (8 and 10 years old) and one younger sister (4 years old). No other relevant health condition was reported by the family.

By the age of 6 months, the mother noticed her child being injured by a burn without crying. Similarly, the child had shown no signs of distress when he lost four of his upper teeth and dislocated his left hip.

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Table 1: Clinical features of the hereditary sensory and autonomic neuropathies

Disorder	Clinical features
HSAN1	Most are autosomal dominant
	Onset often in early adulthood but variable
	Distal sensory loss, foot ulcers
	Preservation of facial sensation
	Variable muscle wasting and weakness
	Variable neural deafness and dementia
HSAN2	Autosomal recessive
	Loss of pain, temperature and tactile sensation
	Recurrent infection and fractures of the digits
HSAN3 (familial	Autosomal recessive
dysautonomia)	Progressive sensorimotor neuropathy
	Sympathetic autonomic dysfunction
	Smooth tongue without fungiform papillae
HSAN4 (congenital	Autosomal recessive
insensitivity to pain	Profound loss of pain sensitivity
with anhidrosis)	Defects in thermoregulation
······	Anhydrosis
	Mild to moderate mental retardation
	Microcephaly
	Fungiform papillae are present
HSAN5	Autosomal recessive
	Loss of nain and temperature sensation
	Normal muscle strength
	Normal reflexes
	Normal nerve conduction
HSANG	Autosomal recessive
	Ashkanazi Jewish
	Autonomic dusfunction
	Absent fungiform papillae
	Dooth by age 2 years
HSAN7	Autosomal dominant
	Congonital inconsituity to pain
	Collemnation alow wound hading and painless
	Sen-muthation, slow would healing and painless
	Contraintentianal durature ation
	Gastrointestinai dysrunction
ICAN	Automation
HSAN and dementia	Autosomai dominant
	Dementia
TT 1'.	Autonomic dysfunction sensory loss
Hereditary sensory	Autosomai recessive
neuropatny with spastic	Spastic parapiegia
Insensitivity to pain	
Paroxysmai extreme	insensitivity to pain
pain aisoraer	
Primary erythermalgia	Autosomal dominant
	Paroxysmal extreme pain disorder
	Primary erythermalgia
Small fiber neuropathy	Small fiber neuropathy

On general examination, the boy measured 110 cm and weighed 18 kg, with a head circumference of 49 cm. He had normal gait and posture and was conscious, alert and oriented with no fever. He presented multiple scars on his hands, feet and both knee joints due to previous unintentional trauma. Misalignment of his left big toe had been caused by a previous fracture. Swelling of both feet and ankle joints had appeared after left hip joint dislocation at 3 years.

The patient had normal developmental milestones for his age but suffered from stress incontinence and was still wearing a diaper. Corneal reflexes were absent. The child did not react to pinprick or hot bodies. The family had not noticed sweating with physical activity, or reaction to odour. The child did not react to the salty and spice test nor to hot and cold drinks. The rest of clinical examination was normal. And infectious disease is ruled out.

We treated the child's injuries, ensured rehabilitation and provided mental health support to him and his family, mostly using support techniques developed by the Japanese organization 'Tomorrow', which focus on daily life techniques to keep children safe at home and in the outside environment.

Box 1: Patient perspective

The family frequently mentioned the futility of their search for treatment prior to coming to our clinic—a hurdle they had to undertake with no financial support. Despite this, they stated that they were willing to pay with their lives to help their son. The true suffering that they experienced was how to keep their child away from danger and how to teach him to take care of himself.

DISCUSSION

Child abuse has a much higher occurrence rate than rare neuropathies such as the one we describe. However, CIP should be considered as a diagnosis in any child presenting with a history of poor or absent responses to painful stimuli. CIP often presents with unexplained oral injuries (especially NTRK1 and PRMD12 CIP), burns, bruises, fractures and joint injuries [5].

This condition can be easily missed because it is not well known by the medical community, especially in situations such as war, when knowledge of new or rare conditions may be limited. Children with CIP often injure themselves severely, and the injury may go unnoticed, resulting in permanent damage. Diagnosis is primarily clinical, based on impaired pain and temperature perception. Usually diagnosis is made around the age of three, when the family notices the lack of pain. In our case, the family had noticed the problem very early—at 6 months—because the environment was propitious to injury. The parents had been seeking medical help for their child for over 4 years and though many laboratory tests had been carried out such as: virological (HIV, HBs Ag, HCV) and immunological (immunoglobulin levels: IgA, IgG, IgM) tests; skin lesions revealed benign ulcers; endoscopy reported gastric erosion only; numerous blood tests and blood counting, serology, C-RP fluorescence, X-rays, computed tomography scans, and finally a nerve conduction study were carried out and the findings were all suggestive of hereditary sensory neuropathy, with lower limbs more severely affected; given the possibility of congenital loss of pain ('Type C sensory fibre'); they had not had a clear diagnosis nor any support until they came to our clinic.

There is no single gold standard treatment available for this condition. Reports suggest naloxone and naltrexone can be used to reverse the analgesia [6]. Therapeutic options are restricted to treatment of symptoms and protection from self-mutilation, fractures and wound infections, which may lead to amputation. Such limited treatment options imply potentially catastrophic consequences of the natural pathologic evolution of the disease [4]. The treatment and care for patients with HSAN types IV and V require a wide range of knowledge and experience, and a multidisciplinary team approach [7].

The sensation of pain is a precursor for a large variety of pathological conditions, but its absence for any reason may lead to potentially life-threatening situations [3]. For this reason, it is important that the medical world not view these cases from a research perspective only, but also develop strategies to support affected patients and their families with education and care guidelines [8] (Figs 1–4).



Figure 1: Multiple scars and normal appearance.



Figure 2: Loss of teeth.

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Figure 3: (a) Fracture of the big left first toe and (b) left hip dislocation.



Figure 4: Charcot's joint.

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CONFLICT OF INTEREST STATEMENT

None declared.

INFORMED CONSENT

The patient's parents provided written informed consent for the publication of this case and the accompanying pictures.

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