CASE REPORT

NOT ANOTHER CASE OF JUVENILE IDIOPATHIC ARTHRITIS: CONGENITAL INSENSITIVITY TO PAIN PRESENTING WITH JOINT PROBLEMS

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Congenital insensitivity to pain with anhidrosis (CIPA) is a rare condition with protean manifestations. Occasionally, these diseases may present with musculoskeletal problems. Here we report a case of young child who presented with joint problems in our clinic. Since the child had a number of musculoskeletal manifestations her diagnosis was difficult and delayed. She was given supportive treatment and a team of doctors from different specialties was involved in her management.

Keywords: Congenital insensitivity to pain; Neurogenic arthropathy; Charcots joints; Hereditary sensory and autonomic neuropathy

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INTRODUCTION

Congenital insensitivity to pain with anhidrosis (CIPA) is a rare hereditary sensory and autonomic neuropathy. Only a few hundred cases have been reported and the exact prevalence is not known.[1,2] The disease classically manifests as insensitivity to pain, and anhidrosis and mental retardation. The consequences of which may lead to an array of clinical findings.[3] The disease may also manifest with a number of musculoskeletal problems which are secondary to pain insensitivity.

It is caused by a defect in NTRK1 gene.[4,5] Useful investigations may include nerve conduction studies, skin and nerve biopsy.[6] Unfortunately no definitive treatment or gene therapy exists. Supportive treatment may be provided in addition to counselling and education. The prognosis is poor and lifespan rarely exceeds to adulthood.[2,7]

The rarity of the disease and diverse clinical findings may make it a difficult to diagnose condition. As a result, the patient may present in various specialties and may undergo a number of referrals before they can be finally diagnosed. Here we present a case of such a patient who remained undiagnosed and eventually presented to rheumatology clinic with complaints of joint swellings and deformities.

CASE REPORT

An 11-year-old girl presented with complaints of progressive swelling and deformities of multiple joints since early childhood. Her parents described her as completely insensitive to pain and temperature. There were unexplained episodes of high-grade fever which seldom responded to anti-pyretic. These episodes were relieved after taking baths. She had complete lack of perspiration. There was a history of self-mutilation in the form of tongue and finger biting right after her teeth eruption. She had sustained fractures to the hip in the past after repeated falls. She had learning difficulties and was not at par with her peers at school. Family also reported emotional liability. She had developed swellings and deformities in her wrists and knee joints. She also developed significant difficulty in walking and performing her daily chores. She was the second child of consanguineous couple from South Punjab. Her family history was unremarkable and the remaining siblings were healthy and unaffected. In the past she had made several consultations with orthopaedic surgeon and rheumatologists. At one occasion patient was worked up and investigated for juvenile idiopathic arthritis and given DMARDs and glucocorticoids. However, no improvement was seen with any treatment.

On examination she had a dry and thick skin at the palms. The finger tips were scarred and shortened. There were multiple ulcers and bruises over the lower limbs. Inspection of the oral cavity revealed lip wounds, tongue laceration and subluxation of multiple teeth. Her wrists and knee joints were swollen and deformed on inspection. However, palpation could not reveal joint tenderness. Range of motion was particularly decreased in knees and hips. Neurological examination revealed gait abnormality, normal muscle power, and intact reflexes. There was insensitivity to pain and temperature.

Knee joints were aspirated and fluid was sent for analysis which did not reveal any abnormality. In addition to routine labs, serum uric acid, NCS and EMG were obtained which were also normal. Radiographs of multiple joints revealed widespread destruction, disorganization and defragmentation. The family was counselled and offered genetic testing. Genetic testing was not done due to financial restraints. A clinical diagnosis of congenital insensitivity to pain with anhidrosis was made. Patient was given supportive treatment. Her management also involved care from multidisciplinary team including, orthopaedic surgeon, paediatrician and occupational therapist.

Figure-1: Grossly deformed and swollen left knee joint

Figure-2: X-ray of left knee joint showing fragmentation and widespread joint destruction

DISCUSSION

Congenital insensitivity to pain with anhidrosis is one of the hereditary sensory and autonomic neuropathies. It is classified as type IV hereditary sensory and autonomic neuropathy (HSAN-IV) according to the Dyck's classification.[8] It is an extremely rare condition with an unknown prevalence. However, some sources claim that the prevalence maybe as low as 1 in 125 million of newborns.[7]

The presence of widespread anhidrosis differentiates type IV from other types of hereditary sensory and autonomic neuropathies. As a consequence, affected individual may present with episodic fever and recurrent hyperpyrexia. This was also true for our patient and in her case the fever did not respond to anti-pyretics but was lowered by taking baths. These individuals may also have skin changes such as thick and lichenified skin, nail and hair problems. The remarkable insensitivity to pain makes these individuals vulnerable to self-mutilation in the form of tongue biting and finger biting. Children with these conditions usually have normal developmental milestones but may have significant learning disabilities and emotional liabilities.[3]

At times disease may present with deformed and swollen joints and other musculoskeletal complications such as fractures, infections and Charcot joints. Fractures in these individuals heal slowly particularly in the large joints.[9] The large joints are also susceptible to repeated trauma and infectious complications.[2,9] Our patient had presented to us with such complaints. The fact that her joints were grossly swollen and deformed had caused confusion regarding the diagnosis previously. The large joints were particularly more affected in our patient. She had fracture of the right hip and grossly deformed knees.

The NTRK1 gene is responsible for encoding the neurotrophic tyrosine kinase-1 receptor. It is one of the nerve growth factor receptors, the ligands of which play vital role in development of the peripheral and central nervous system.[4,5] Currently no gene therapy is available.

Treatment is mainly symptomatic and the management revolves around prevention of self-mutilation in order to prevent debilitating deformities and complications Parents or caretakers may be trained and educated about the disease.[3] All parents should be offered genetic counselling that includes explaining potential risks to offspring and family planning.[10]

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