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Case Report Open Access

Charcot arthropathy and acro-osteolysis in a 5-year-old girl with hereditary sensory and autonomic neuropathy type II: A case report

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Charcot neuropathic arthropathy is a destructive process that can occur in patients with neuropathy associated with medical diseases such as diabetes. A rare cause of neuropathic arthropathy is congenital insensitivity to pain (CIP) which includes varying degrees of sensory loss and lack of pain perception contributing to absent withdrawal responses. The clinical manifestations of CIP include delayed diagnosis of fractures, joint dislocations, acro-osteolysis, avascular necrosis, osteomyelitis, heterotopic ossification, and Charcot arthropathy. We herein report a case of Charcot arthropathy of ankle joints in a 5-year-old girl with CIP due to underlying hereditary sensory and autonomic neuropathy who had recurrent upper and lower limb fractures with acro-osteolysis. She had an inability to perceive pain from the first year of her life. She also had mild developmental delay. Family history was unremarkable. Blood count, liver and thyroid function tests, erythrocyte sedimentation rate, C-reactive protein, blood electrolytes, blood sugar and other laboratory tests were normal. Charcot neuropathic arthropathy in children is rare and can be a part of congenital diseases like CIP. A high degree of suspicion may lead to early detection and can prevent joint and bone destruction and deformities.

Keywords: Charcot arthropathy; Hereditary sensory and autonomic neuropathy; Congenital insensitivity to pain; Neuroarthropathy; Acro-osteolysis

Introduction

The concept of neuropathic arthropathy was first proposed by neurologist Jean-Martin Charcot in 1868 when she observed patients with tabes dorsalis [1]. Charcot neuropathic arthropathy as a degenerative disease can lead to fractures, dislocations, fragmentation, osteophytes, and loose bodies [2]. Pathophysiology of neuropathic arthropathy includes: 1. Sensory neuropathy destructs the bones as a result of numerous micro-traumas that they experience because of impaired pain sensation and proprioception, 2. Autonomic neuropathy of vasoregulation leads to increased blood flow and hyper-vascularization in the subchondral bone that can result in osteoporosis, microfractures, and dislocations [2-4]. Congenital insensitivity to pain (CIP) is an infrequent cause of neuropathic arthropathy. It includes varying degrees of sensory loss and lack of pain perception and contributes to the absence of withdrawal responses [5-7]. The hereditary sensory and autonomic neuropathies (HSANs) are characterized by a group of clinically heterogeneous manifestations [8]. The involvement of the sensory system can be presented by loss of pain and temperature perception or alterations in reflexes [8]. On the other hand, autonomic dysfunction could be presented by anhidrosis or excessive sweating, postural hypotension, and gastroesophageal reflux [8].

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Patients with HSAN can be categorized according to several systems. HSAN is classified into five subtypes based on one of the most common classification systems [9]. The most important clinical manifestation of CIP is multiple painless fractures and dislocations [10]. The healing process of these fractures can be complicated in these patients by nonunion, malunion, and osteomyelitis and can finally lead to acro-osteolysis and Charcot arthropathy [10].

Most of the publications on the topic of neuropathic arthropathy are concerned with adults and even elderly patients with a complicated course of diabetes mellitus [2]. Herein, we report a case of a 5-year-old girl who was insensitive to the pain and had recurrent upper and lower limb fractures with acro-osteolysis.

Case presentation

A 5-year-old girl was brought to our clinic for the first time with several deformities of her upper and lower limbs. She had multiple stumps of her fingers and toes due to progressive osteonecrosis and subsequent auto-amputations (Figure 1). On physical examination, she demonstrated shortening of her nails, acro-osteolysis, and progressive hindfoot valgus deformity as the typical clinical picture of Charcot arthropathy. Impairment of patellar deep tendon reflex as well as Achilles tendon were found. Moreover, there was a complete absence of bilateral dorsiflexion and plantar flexion of ankle. Similarly, great toe dorsiflexion was totally impaired. However, there was no muscle atrophy. In the inspection, chronic callosities of the upper and lower limbs were seen. Family history was unremarkable. Regarding laboratory data, there were no abnormalities in complete blood count, blood electrolytes, blood sugar, erythrocyte sedimentation rate, as well as liver, kidney, and thyroid function tests.

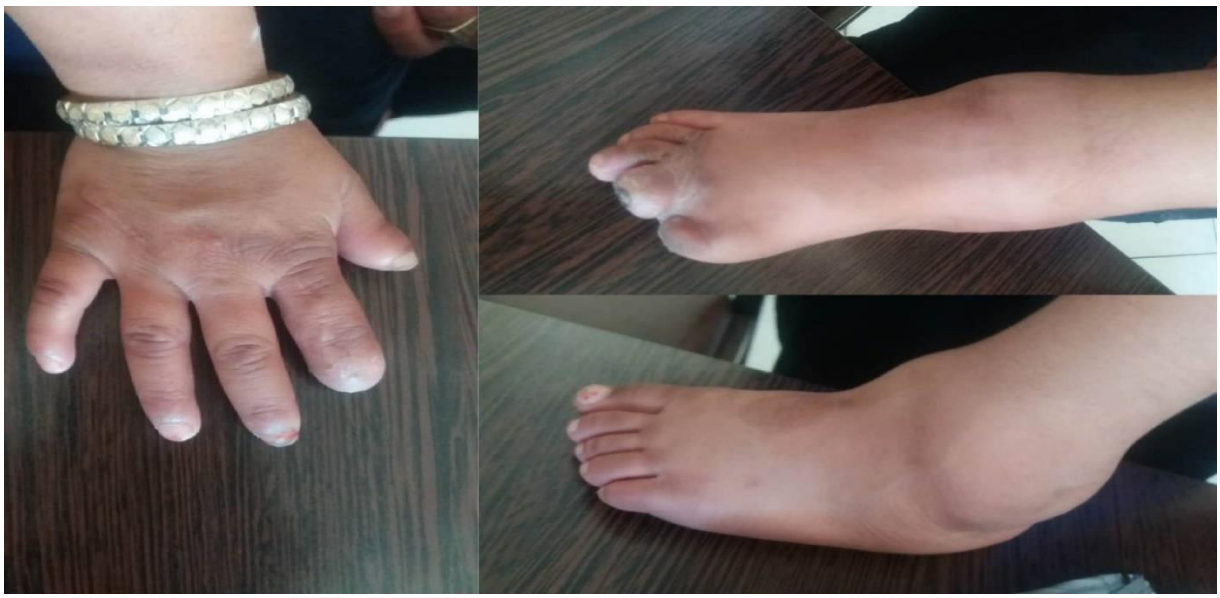

Figure1: Swollen foot due to Charcot arthropathy in ankle joint and shortening of fingers and toes due to acro-osteolysis can be seen

We made an evaluation and established the diagnosis of CIP based on her clinical presentation. Multiple fractures in addition to her abnormal pain perception and mild developmental delay were the key features that help us to establish the diagnosis. Her medical history suggests that insensitivity to pain was evident from the first year of her life and was underestimated by her parents, thus they had not followed up. A whole-body bone scan revealed an increased radiotracer uptake along the right tibial shaft and the right calcaneus, less intensity in the right talar bone, small deformity in the left talus with no remarkable abnormal uptake, and aplasia of the right toe. Radiography as our primary imaging study indicated the typical radiological evidence of Charcot foot consisting of fragmented non-united calcaneal fracture (Figure 2).

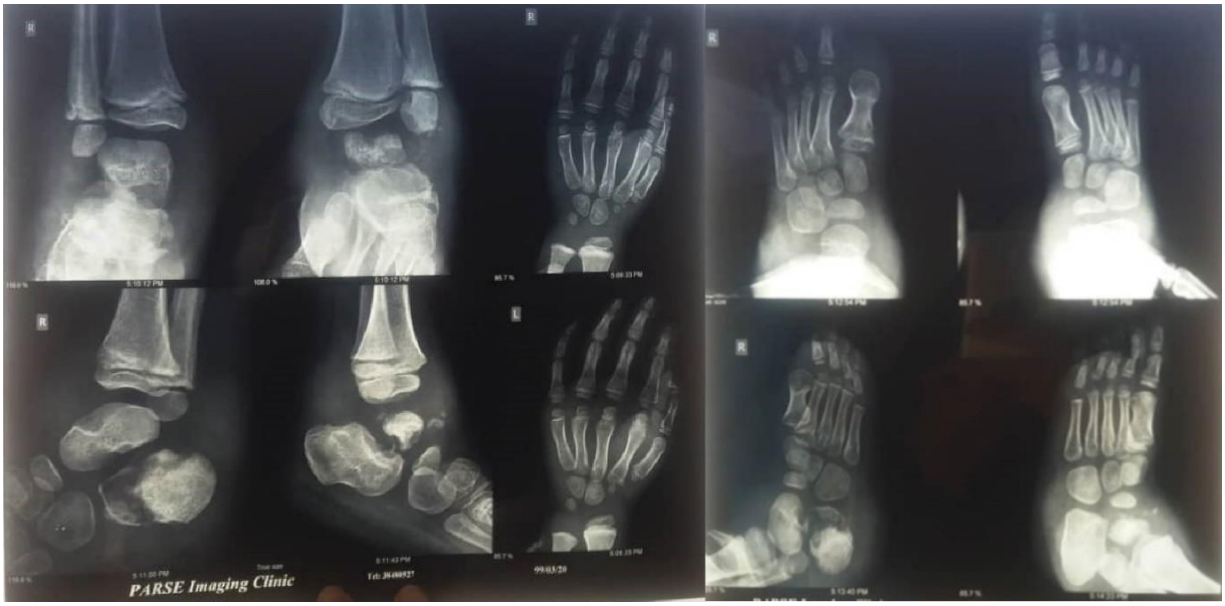

Figure 2: Radiographs of the ankles, feet and hands show acro-osteolysis of fingers and toes, and Charcot arthropathy of ankles. Destruction, loosening of the body, dislocation and density changes are evident in the ankle joints.

Discussion

Patients with neuropathy may be complicated with a bone destructive process called Charcot neuroarthropathy which can involve every joint, however, foot and ankle joints can be considered the most common joints that may be involved [11, 12].

Historically, Charcot arthropathy was the complication of neuro infections such as syphilis, tuberculosis, and leprosy. It can also be associated with medical conditions such as advanced stages of diabetes mellitus, chronic alcoholism, and hemodialysis in adult and elderly patients [11]. Recent studies revealed that neuroarthropathy may occur in patients under the age of 18 such as children with spinal dysraphism [13-15] and intra-articular neurofibroma [16]. We report a 5-year-old girl with CIP) who had Charcot neuroarthropathy due to underlying hereditary sensory neuropathy. CIP is a rare disorder in which the absence of pain perception leads to a lack of appropriate protective reflexes. Subsequently, a variety of clinical conditions including bone fractures and joint dislocations, acro-osteolysis, osteomyelitis, and eventually Charcot arthropathy may occur.

HSAN is classified into five types according to its clinical and genetic features, the pattern of inheritance and the neural system involvement. ***HSAN I*** is a mild form of HSAN that is usually autosomal dominantly inherited and symptoms begin in the second decade of life or later [4]. Sensory disturbances are key features of this type [4]. It predominantly involves lower limbs and contributes to major complications such as foot stress fractures and paronychia of the toes [4]. Additionally, plantar ulcers and bone resorption are common in this type [4]. ***HSAN II*** or Morvan's disease is recognized as the most severe form of HSAN which inherit recessively or occur sporadically [4]. Onset can be at birth or later in childhood [4]. Generalized sensory loss, significant hypotonia with the absence of tendon reflexes, and pathologic fractures are characteristics of HSAN II [4]. Autonomic involvements consisting of bladder dysfunction, impotence, and distal anhidrosis are common. Occult fractures of extremities due to repetitive unrecognized traumas as well as Charcot joints are the major manifestations of HSAN II [4]. Mental status is usually normal [4]. ***HSAN III*** is the most frequent form of the HSAN disorder and autosomal recessively inherited [4]. This progressive disorder is known as familial dysautonomia or Riley-Day syndrome and usually presents at birth [4]. Autonomic manifestations are prominent and feeding difficulties are the earliest sign of autonomic dysfunction [4]. Premature death, pulmonary infections due to vomiting, absence of tears (alacrimia), defective temperature control,

and hypertension are the common clinical features [4]. ***HSAN IV*** is an autosomal recessive disorder that predominantly involves ectodermal structures including skin, bone, and nervous system [4]. Anhidrosis can be considered the key feature of this type [4]. The onset of HSAN IV is usually in infancy [4, 8]. Mental retardation, behavioral disturbances, and defective temperature control are common presentations [4, 8]. ***HSAN V*** is an autosomal recessive disorder with onset at birth in which nociception is selectively affected [4]. As we discussed our patient is clinically diagnosed with HSAN II but genetic investigations did not perform [17]. She had skeletal manifestations of CIP due to Occult fractures caused by pain insensitivity. Frequent unrecognized injuries and fractures of hands and feet contributed to the Charcot joint [17, 18]. A high degree of clinical suspicion is required for the detection of this disorder at the early stages because the prompt treatment might preserve joints and bone from further destruction. On the other hand, late diagnosis contributes to high-level amputation and subsequent life-long morbidities [19].

Conclusion

Charcot neuropathic arthropathy in children is not a common condition and can be a part of congenital diseases like CIP. Clinicians should pay special attention to the history and physical examination of children with skeletal problems who have not reported a recent trauma. Primary radiological investigations like standard plain radiographs should be performed for suspected patients. Early detection of children with CIP can preserve joints and bones from further destruction and subsequent deformities. These data necessitate a high degree of clinical suspicion for diagnosis. It is noteworthy that short-term follow-ups should be highly recommended to patients and their family.

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Conflict of interest

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