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Case Report

Charcot Foot Arthropathy Secondary to Holocord Syringomyelia in a Skeletally Immature Patient - A Case Report

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Abstract

Charcot arthropathy, is progressive and destructive process of articular surfaces secondary to impaired nociception and proprioception of the involved joint, commonly seen in adults a long-standing history of diabetes mellitus. We report a case of a 4-year-old girl who presented to the Emergency Department complaining of right heel pain and antalgic gait. Physical examination was unremarkable, but an MRI of the right foot revealed a lytic and destructive process of the right calcaneus with associated fluid tracking. Given the absence of any biochemical evidence to favour infection, a full-body MRI subsequently revealed an Arnold-Chiari type I malformation with extensive holocord syringomyelia of the cervical and thoracic spine.

Keywords: Charcot Arthropathy; Holocord Syringomyelia; Diagnosis

Case Report

A 4-year-old girl presented to a regional Emergency Department with a one-day history of progressive atraumatic right heel pain with associated limp. The patient's caregiver noted that the patient had been complaining of acute onset localised sharp pain in the right heel precipitated by weightbearing and an altered gait pattern. The caregiver reported no antecedent trauma, infective symptoms, irritable behaviour, or joint pain prior to the patient's presentation.

The patient had no significant past medical history, was born to term at 39 weeks as a spontaneous vaginal delivery with no pre-, ante- or post-natal issues and was up to date her immunisations. There was no significant family history of medical problems.

On examination, the patient was comfortable with no signs of distress, with vital signs within normal limits. There was mild swelling of the posterior aspect of the right calcaneus which was not associated with erythema or deformity of the foot, ankle or lower limb. On palpation, there was no warmth or fluctuance of this swelling, but there was localised tenderness in this region. The right ankle had full range of dorsiflexion, plantarflexion, inversion, and eversion which was not irritable. Sensation of the superficial and deep peroneal, saphenous, sural, medial, and lateral plantar nerves were normal. Furthermore, she demonstrated 5/5 power with flexion and extension of the toes, capillary refill time of the right hallux was less than 2 seconds and the patient's dorsalis pedis and posterior tibial pulses were palpable. Examination of the ipsilateral knee and hip joints were unremarkable. The patient had an antalgic gait with pain on heel strike.

An initial non-weightbearing radiograph of the right foot demonstrated a mixed lytic and destructive process over the posterior calcaneus (Figure 1). Initial screening bloods were not concerning for infection, inflammatory or malignancy with haemoglobin, white cell count, C-reactive protein, erythrocyte sedimentation rate, calcium, alkaline phosphatase and phosphate levels within normal limits. Given the abnormal findings of the radiograph, the patient was transferred to a tertiary paediatric centre for further assessment and management.

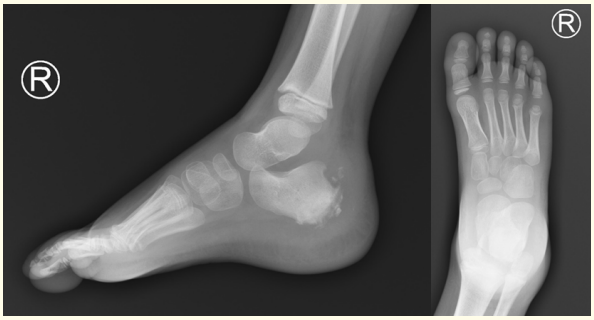

Figure 1: Lateral (A) and anteroposterior (B) non-weightbearing radiographs of the right foot demonstrating a lytic and destructive process of the posterior aspect of the calcaneus.

The patient had a magnetic resonance imaging (MRI) scan of the right foot and ankle, which demonstrated a destructive process centred on the calcaneal growth plate and the adjacent apophysis with associated curvilinear fluid signal tracts along the growth plate posterior to the calcaneal body and extending along the under surface of the posterior calcaneus deep to the plantar fascia origin (Figure 2). There was also soft tissue oedema extending into the plantar foot musculature and prominent subcutaneous oedema at the anterolateral aspect of the ankle joint but no soft tissue collection or abnormal enhancing soft tissue mass.

Given that the MRI findings were consistent with an infective process, but the patient had no significant physical or biochemical signs, initial differentials included chronic recurrent multifocal osteomyelitis (CRMO), Ewing Sarcoma, and Langerhans cell histiocytosis (LCH). Given the unclear findings and concerns of a systemic disease a joint decision between the treating team and the patient's family was made to perform a full-body MRI.

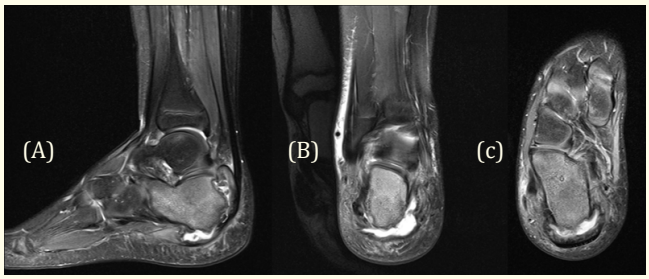

Figure 2: T2-weighted lateral (A), anteroposterior (B) and axial (C) MRI images of the right foot demonstrating an unusual destructive process centred on the calcaneal growth plate and adjacent apophysis.

The MRI revealed an incidental finding of a Type I Arnold-Chiari malformation with approximately 40mm of tonsillar descent to the superior end plate of T4 (Figure 3). There was a cystic encephalomalcia involving the right cerebellar tonsil which further compresses the cervical cord. There was an extensive holocord syringomyelia extending from C4 to the level of the conus. There was no abnormal enhancement or mass seen within the spinal canal. Despite the tonsillar descent filling the foramen magnum with obliteration of the CSF signal, there was no hydrocephalus. Otherwise, the brain appeared structurally normal and no other abnormal osseus lesions were detected.

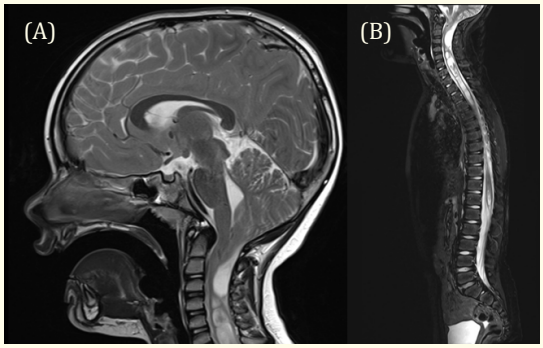

Figure 2: T2-weighted MRI images of the lateral brain (A) and spine (B) demonstrating Arnold-Chiari type I malformation with extensive syringomyelia from C4 to T12.

Following this, a decision was made to perform an incisional biopsy of the calcaneal lesion which was performed via an extensile incision of the lateral heel with imaging guidance. The procedure was uncomplicated. Histopathology of bony samples revealed periosteal fibrous tissue and physeal cartilage with areas of non-inflamed granulation tissue. No evidence of malignancy or suppurative or granulomatous inflammation was noted.

Given the combination of a destructive and lytic process of the right foot and detected syringomyelia, a sensory mapping study was performed. Given the patient's age and compliance, it was a difficult study to perform. It was revealed that right heel had no response to deep and protective pressure, and the toes of the right foot had a diminished response to deep pressure.

Differential diagnosis

The presentation of a limping child encompasses multiple differential diagnoses. Common differentials include infection, trauma, transient synovitis, acute myositis, Toddler's fracture, developmental dysplasia of the hip, and non-accidental injury. Rarer differentials include rheumatological, immunological and haematological disorders and malignancy.

Our patient's imaging findings were consistent with an infective process; however, they had no significant findings on history, examination, biochemistry and histopathology to support this diagnosis. Initial differentials included chronic recurrent multifocal osteomyelitis (CRMO), Ewing Sarcoma, and Langerhans cell histiocytosis (LCH) which prompted the full-body MRI study, which subsequently revealed the extensive holocord syringomyelia. This raised Charcot arthropathy potentially being a diagnosis given previously established association with syringomyelia, which was later supported by the sensory mapping study results.

Treatment

The MRI findings of a Type I Arnold-Chiari malformation and tonsillar cyst prompted neurosurgical input. The patient had a full cranial nerve, upper and lower neurological examination. It was reported that the patient's cranial nerves were intact and demonstrated normal tone, power, reflexes, co-ordination, sensation of their upper and lower limbs. The patient underwent an uncomplicated suboccipital craniectomy and C1 laminectomy and decompression for the Type I Arnold-Chiari malformation. They were admitted to the intensive care unit post-operatively and had a repeat MRI brain and spine demonstrating suboccipital decompression and cerebellar tonsil resection with reduced mass effect on the upper cervical cord. The patient was subsequently stepped down to ward-base care, had physiotherapy input until mobilising independently without aids, and subsequently discharged from hospital.

Outcome and follow-up

The patient has an uncomplicated recovery following discharge home from hospital. She has been weightbearing as tolerated post-operatively, but continues to have pain on palpation of the right calcaneus, an antalgic gait but no residual neurological deficit.

Discussion

To our knowledge, this is the first report of Charcot arthropathy (CA) in a skeletally immature patient secondary to holocord syringomyelia which warrants sharing with colleagues. Our case presented with heel pain and antalgic gait, with imaging subsequently demonstrating a destructive and lytic process of the calcaneus and a C4-T12 holocord syringomyelia, with the diagnosis of CA confirmed through a sensory mapping study. Previous reports of CA secondary to syringomyelia have been seen exclusively in skeletally mature patients, affecting the upper extremity, and involving the glenohumeral and elbow joints [1-5].

Charcot arthropathy is an irreversible, destructive condition characterised by acute fractures, dislocations, and joint destruction in the weight-bearing neuropathic foot [6]. In the Western World, it is predominantly seen in people with diabetes mellitus, however other risks reported include alcoholism, leprosy, myelomeningocele, syphilis infection and syringomyelia [7,8]. The true prevalence of CA is unknown, due to a high incidence of incorrect or delayed initial diagnosis [9]. The two primary explanations of the developing of CA are the neurotraumatic and neurovascular theories. The neurotraumatic theory hypothesises that trauma is the primary driver in the absence of protective sensation. Trauma results in the release of pro-inflammatory cytokines which stimulate osteoclastogenesis and excessive bone turnover [10,11]. The neurovascular theory as described by Charcot, hypothesises that autonomic dysfunction increases blood flow through arteriovenous shunting [10,11]. Increased blood flow enhances fluid filtration via capillary leakage, resulting in increased compartmental pressure and ischemia to ligaments and tendons which can result

in joint instability [10,11]. Anatomic-based radiological classifications for CA have been developed, with Brodsky's classification system the most widely used, which is based on the most common regions affected (Table 1) [12]. This system was further revised by Trepman., *et al.* (2005) to include Type 4 (multiple areas) and Type 5 (forefoot) involvement [13]. Our case demonstrated radiographic findings consistent with Type 3B of the Brodsky classification system as it solely involved the calcaneus. When identified early, non-surgical management in the form of contact casting, orthotics, shoe modifications and pharmacotherapy is preferred to halt the progression of disease. In cases identified late or those which have exhausted operative measures, management is surgical and may involve exostoectomy, tendo-achilles lengthening, arthrodesis, osteotomies and amputation [14,15].

Type	Location	Involved Joints
1	Midfoot	Tarsometatarsal, naviculocuneiform
2	Hindfoot	Subtalar, talonavicular, calcaneocuboid
3A	Ankle	Tibiotalar
3B	Calcaneus	Tuberosity fracture
4	Multiple regions	Sequential, concurrent
5	Forefoot	Metatarsophalangeal

Table 1: Brodsky's Anatomic Classification system of Charcot arthropathy (Types 1-3B) with Trepman., *et al*. modification (Types 4 and 5) [12,13].

A syringomyelia is a fluid-filled cyst called a 'syrinx' within the spinal cord that can cause cord compression and disruption of nerve fibres. The prevalence is thought to be around 8.4 per 100000 cases, and usually presents around 30 to 40 years of age [16]. The pathogenesis of syringomyelia is unclear with multiple theories hypothesises, but it is generally thought that herniated cerebellar tonsils act as 'pistons' which obstruct the subarachnoid space at the foramen magnum which then generates a pressure wave in the subarachnoid space within each heartbeat [17-19]. This can interrupt sensory innervation and lead to peripheral neuropathy [5]. This peripheral neuropathy can result in a patient not realising a fracture or joint dislocation which can result in repetitive release of inflammatory cytokines and increase osteoclastic activity as described above. Syringomyelia predominantly affects the upper limbs is due to involvement of the spinothalamic tract fibres, which are essential for pain and temperature sensation, that supply the shoulders and elbows [20].

Our literature search yielded a single case report of syringomyelia affecting the lower limb. Memarpour., *et al.* (2015) described a 36-year-old man presenting with hip pain with associated upper limb pain and thermal sensory deficits in a cape-like distribution. Their hip imaging demonstrated articular surface destruction of the hip joint, with a subsequent MRI of the spine revealing an Arnold-Chiari type I malformation with extensive syringomyelia of the C- and T-spine [21]. They demonstrated similar radiological findings to our case, however the upper limb pain and thermal sensory deficits made such a diagnosis more straightforward. In the setting of a paediatric patient, eliciting such details can be difficult on history and examination, which prompted our whole-body MRI scan. Deng., *et al*. (2013) prospective cohort study of 12 patients with diagnosed CA secondary to syringomyelia remains the largest report in the literature [22]. Their cohort had a mean age of 45.8 years, ranging from 13 to 76 years of age. Of the 12 cases, 8 involved the elbow joint exclusively, two involved the shoulder exclusively, with the last remaining cases involving multiple articulations of the upper limb. All cases albeit one, had no history of preceding trauma. The paediatric patient, like our case had no evidence of pain or sensory changes at diagnosis. In this cohort, 5 patients underwent neurosurgical treatment with improvement in neurological dysfunction and no deterioration in symptoms.

Conclusion

Charcot foot arthropathy secondary to holocord syringomyelia is an extremely rare diagnosis. The cardinal signs and symptoms associated with this syndrome can be difficult to elicit, particularly in the paediatric patient. There should be a low threshold for multidisciplinary input and full-body imaging in the case of inconsistent clinical, biochemical and radiological findings. Early recognition and interventional is critical to prevent the progression of joint destruction, as treatment relies on the early diagnosis and management of the underlying neurological disease.

Learning Points

- A child presenting with a limp requires careful evaluation, as serious causes can be missed in otherwise healthy patients.
- Charcot arthropathy should be considered when imaging demonstrates a mixed lytic and destructive process, irrespective of the patient's age.
- Neuropathic changes associated with syringomyelia are predominantly upper limb but can occur in the lower limb as well.

- A low threshold for full-body imaging is warranted when imaging demonstrates an infective process in the absence of clinical and biochemical markers.

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