Case Report

A rare case report on Chiari malformation and syringomyelia-related Charcot arthropathy

Shasthara Paneyala[1], Soofia Naaz[2], Pallavita Chirravuri[2], C V Thejesh[3]

From [1]Associate Professor, Department of Neurology, [2]MBBS Student, [3]Senior Resident, Department of Neurology, JSS Medical College and Hospital, Mysore, Karnataka, India

ABSTRACT

Charcot arthropathy is a progressive and often underrecognized joint condition typically resulting from impaired sensory innervation. Although commonly associated with diabetes, it may also occur in the context of less frequent neurological disorders. This report discusses the case of a 27-year-old woman who presented with chronic swelling and restricted movement of the right elbow, without a history of trauma or systemic symptoms. Imaging studies eventually revealed syringomyelia and Chiari malformation as the underlying causes. This case underscores the need for clinicians to consider neuropathic arthropathy in the differential diagnosis of unexplained joint dysfunction, particularly in the upper limbs, and to use advanced imaging techniques like magnetic resonance imaging for timely identification and intervention before irreversible joint damage occurs.

Key words: Charcot arthropathy, Chiari malformation, Elbow joint, Syringomyelia

Charcot arthropathy, also known as neuropathic arthropathy, was originally described by Jean-Martin in 1868. This condition is most frequently seen in individuals with diabetes mellitus, but it may also occur in association with syringomyelia, a fluid-filled cavity or syrinx within the spinal cord [1]. The overall prevalence of Charcot arthropathy in the general population is <0.1%, whereas in the diabetic population, the prevalence is about 0.1–0.4% and among those with diabetic neuropathy, the prevalence rises to 10–30% [2]. Syringomyelia is commonly linked with Chiari malformation type I, a congenital disorder involving downward displacement of the cerebellar tonsils through the foramen magnum [3]. While Charcot arthropathy most often affects weight-bearing joints, syringomyelia-associated cases typically involve the shoulder or elbow. The rarity of Charcot arthropathy of the elbow, especially due to syringomyelia and Chiari malformation, presents a diagnostic challenge.

Here, we describe the case of a young woman who presented with progressive swelling and restricted movement in her right elbow, ultimately diagnosed as Charcot arthropathy secondary to syringomyelia and Chiari malformation. This report highlights the importance of early imaging and clinical suspicion in atypical presentations to prevent irreversible joint damage. Only a few case reports are available that describe Charcot arthropathy brought on by Chiari malformation and syringomyelia [4]. This underscores the relevance of presenting our case to contribute to the limited literature and enhance clinical awareness of such atypical manifestations. As a result, in this case report, a patient with this rare condition with an unusual presentation has been described.

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CASE PRESENTATION

A 27-year-old female presented to the outpatient clinic with an 8-month history of progressively worsening swelling and restricted range of motion in her right elbow (Fig. 1a). The swelling was insidious in onset, not associated with pain, fever, or any history of trauma. She also denied experiencing any constitutional symptoms such as weight loss or fatigue. The patient had no significant past medical or surgical history, and her family history was non-contributory.

On general examination, the patient was conscious, cooperative, and well oriented to time, place, and person. She was moderately built and nourished. No pallor, icterus, cyanosis, clubbing, generalized lymphadenopathy, or bipedal edema was observed. At presentation, the patient's vitals were within normal limits. On physical examination, diffuse swelling of the right elbow joint was observed. The joint exhibited a marked limitation in movement but was not tender to palpation. There was no local rise in temperature, skin discoloration, or fluctuation over the joint. Sensory examination revealed hypoesthesia over

Correspondence to: Soofia Naaz, 778 Bannimantap B Layout, SS Nagar, Mysuru - 570015, Karnataka, India. E-mail: soofnaaz@gmail.com

the right upper limb, particularly over the elbow region. A trophic ulcer was also noted on the lateral aspect of the right elbow, suggestive of underlying neuropathy (Fig. 1b).

Laboratory investigations were ordered to rule out inflammatory and infectious causes (Table 1). Given the neurological findings, a magnetic resonance imaging (MRI) of the brain and cervical spine was performed. Imaging revealed a large inferiorly displaced cerebellar tonsil herniation (Chiari malformation) extending 5 cm below the foramen magnum (Fig. 2). Furthermore, a syrinx was observed from the cervical segment C1 extending down to T2, with the signal intensity matching that of cerebrospinal fluid on T2-weighted images (Fig. 3). These findings confirmed the presence of syringomyelia associated with a type I Chiari malformation.

Correlating the clinical, radiological, and laboratory findings, a final diagnosis of Charcot arthropathy of the right elbow joint secondary to syringomyelia and Chiari malformation was established.

A multidisciplinary treatment plan was initiated, focusing on neurosurgical consultation for potential decompression, conservative management of the elbow joint, protection against further trauma, and physical therapy to preserve joint function. The patient was educated about the chronic nature of her condition, the risk of progression, and the importance of regular neurological and orthopedic follow-up.

DISCUSSION

Chiari malformation is a collective term for a group of congenital abnormalities involving the upper spinal cord, brainstem, cerebellum, and adjacent bone structures. It is of four types, type I being the most common, which is characterized by herniation of the cerebellar tonsils through the foramen magnum into the cervical spinal canal (cerebellar ectopia). This condition may not produce any symptoms. In symptomatic patients, the associated conditions are syringomyelia and hydromyelia (50%) and hydrocephalus (10%). The pathophysiology behind Chiari 1 malformation associated with syringomyelia is controversial, as there are different theories proposed for it, such as fluid mechanics theory [5], separated pressure

Table 1: Laboratory investigations of the patient

Laboratory investigations	Values	Reference range	Inference
Leukocyte count	9.9×10^9/L	4-11×10^9/L	Normal
C-reactive protein	80 mg/L	<10 mg/L	Increased
Erythrocyte sedimentation rate	100 mm/h	0–20 mm/h	Increased
Alkaline phosphatase	Normal	44–147 U/L	Normal
Acid phosphatase	Normal	0.1–0.6 U/L	Normal

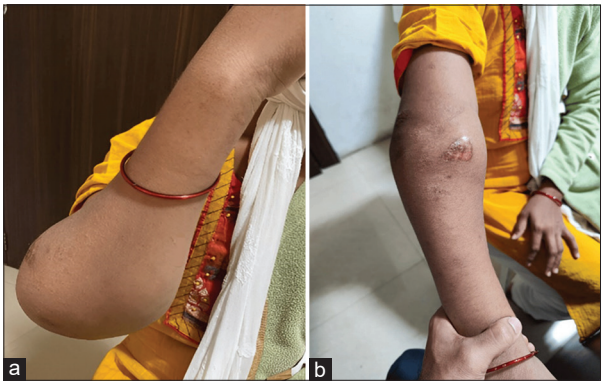

Figure 1: (a) Charcot arthritis of the right elbow joint; (b) Trophic ulcer on the overlying skin of the right elbow joint

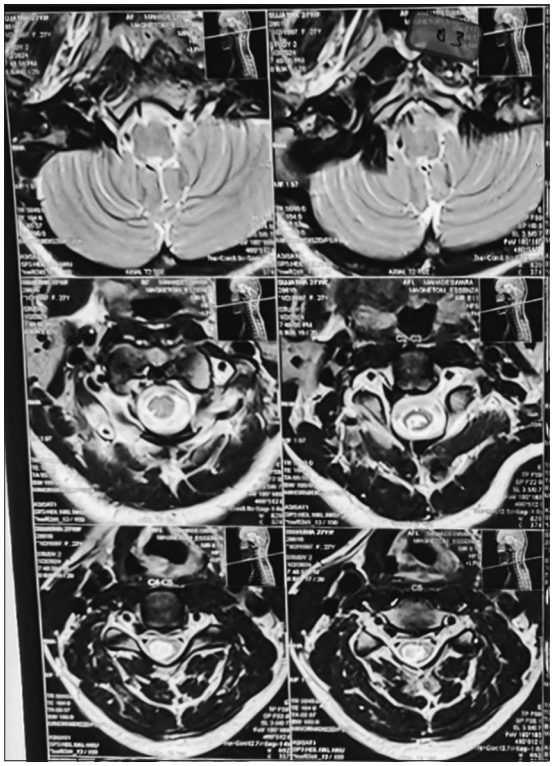

Figure 2: Magnetic resonance imaging of the base of the brain suggestive of syringomyelia and Chiari malformation

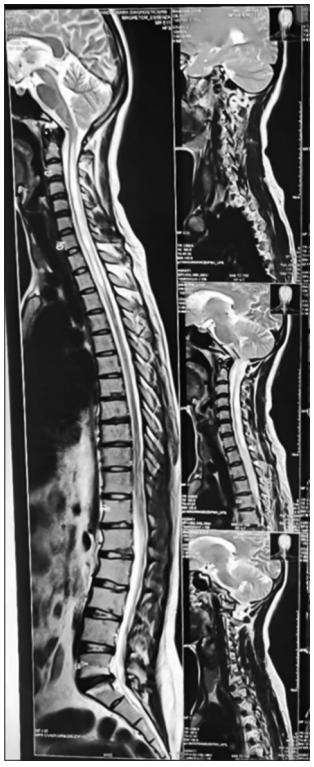

Figure 3: Magnetic resonance imaging of the whole spine suggestive of syringomyelia and Chiari malformation

theory [6], and cerebrospinal fluid infiltration hypothesis of spinal cord parenchyma [7].

Syringomyelia is a chronic, progressive, degenerative, or developmental disorder of the spinal cord. It is characterized by painless weakness and wasting of the hands and arms and loss of thermal and painful sensations with sparing of tactile, joint position, and vibratory sensations. This is caused by cavitation of the central parts of the spinal cord, usually in the cervical region, which might extend upward into the medulla and pons or downward into the thoracic and lumbar segments. Syringomyelia can rarely cause neuropathic arthropathy, which may develop early or late in the course of the disease and is commonly misdiagnosed.

Charcot arthropathy is a progressive degenerative condition involving the joints that are mostly associated with neurosensory loss. Although it often affects weight-bearing joints such as the ankle, knee, and hip, the position of the affected joint varies according to the underlying neurological disease [8]. Patients with diabetic neuropathy have the highest prevalence of the illness [2], with the ankle joint being the most commonly involved joint. In addition, Charcot arthropathy can occur in people with syringomyelia. The joints that are most frequently impacted by syringomyelia are the elbow and the shoulder. Single joints are typically affected in this condition; multiple joint involvement is uncommon [9,10]. The genesis of Charcot arthropathy has been explained by several theories, including osteopenia due to increased blood flow linked to autonomic neuropathy, bone failure due to abnormal stress on bones and joints resulting from muscle imbalances caused by sensorimotor neuropathy, joint subluxation due to ligamentous stretching linked to joint effusion, and the loss of protective mechanisms in the body due to sensory disturbance brought on by neuropathy [11].

The patient came to the hospital complaining of painless swelling and limited range of movement in her right elbow joint for 8 months. At the presentation, she denied having any pain, injuries, or constitutional symptoms. After an MRI, it was found that she had comorbid syringomyelia and Chiari malformation, which was thought to be the reason behind the joint's degradation (Charcot arthropathy). When Charcot arthropathy was discovered, it was typically in an advanced stage and was difficult to treat. The outcomes of complete replacement arthroplasty for Charcot arthropathy have been unsatisfactory, even though recent reports reveal good middle and long-term results [12], so early diagnosis is important. It might be difficult to differentiate Charcot arthropathy from other disorders, as there are currently no distinguishable clinical, radiological, and laboratory findings that can establish the diagnosis of Charcot arthropathy in its early stages. Similarly, in this case, the patient did not undergo any examination that could describe her condition. As a result, her condition progressed to an advanced stage, which was difficult to cure. Early detection of this rare condition would have been possible if an MRI had been done in the early stages. This case emphasizes the importance of early diagnosis of Charcot arthropathy secondary to Chiari malformation and syringomyelia.

CONCLUSION

This case emphasizes the necessity of maintaining a high index of clinical suspicion for rare neurological etiologies in patients presenting with painless joint swelling and dysfunction. Charcot arthropathy of the elbow, though infrequent, may arise secondary to syringomyelia associated with Chiari malformation that often remains undiagnosed until significant joint degeneration has occurred. The insidious onset and atypical joint involvement contribute to diagnostic delays, resulting in progressive structural damage and functional decline. Early deployment of advanced imaging modalities, particularly MRI, alongside a multidisciplinary management strategy, is essential to identify these conditions promptly and initiate appropriate interventions. Timely recognition and treatment are critical to preserving joint integrity and optimizing patient outcomes. Awareness of such uncommon associations is imperative for clinicians to facilitate early diagnosis and improve long-term prognosis.

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