CASE REPORT

Charcot's osteoarthropathy mimicking an osteosarcoma of humerus

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SUMMARY

Charcot's arthropathy or neuropathic osteoarthropathy is a form of destructive arthropathy associated with decreased sensory innervation. We present an unusual case of a patient referred to oncology clinic for further evaluation of a spindle cell neoplasm of the humerus concerning for malignancy. The mass presented as a lytic lesion associated with a right humerus fracture. This was later diagnosed as Charcot's shoulder secondary to cervical syringomyelia. A detailed case report and discussion are presented here.

BACKGROUND

Neuropathic osteoarthropathy is a chronic degenerative arthropathy associated with decreased sensory innervation resulted in destruction and impaired function of the affected joint. This rare disease has been found to be associated with syringomyelia due to traumatic injury to the cervical and/or thoracic spine. Fewer than 60 cases have been reported so far and the diagnosis is sometimes difficult to make. Neuropathic shoulder has been misdiagnosed as soft tissue sarcoma in one case, for which treatment with radiation and surgery was applied. Other conditions with similar presentations including degenerative joint disease, septic arthritis and osteomyelitis, should also be ruled out prior to treatment.

CASE PRESENTATION

A 71-year-old Caucasian man with a medical history of hypertension, coronary artery disease, atrial fibrillation, deep venous thrombosis, peripheral arterial disease and above-the-knee amputation of both lower extremities, was referred to oncology clinic for evaluation of a right proximal humerus lytic lesion causing a pathological fracture. The patient's medical history was significant for traumatic injury to the thoracic and lumbar spine 40 years prior, for which he underwent multiple back surgeries. He underwent above-the-knee amputation of the bilateral lower extremities secondary to severe peripheral arterial disease. Six months prior to his initial presentation, he noted gradually worsening numbness, tingling and swelling over the right shoulder, without any associated fevers, chills or weight loss. While lifting himself off his wheel chair, the patient experienced severe pain over the right shoulder and sudden worsening of swelling. Relevant physical examination revealed an 8×8 cm tender swelling on the outer and proximal parts of the humerus, with signs of muscular atrophy of the shoulder girdle. Shoulder

movements were limited in all directions. The patient was found to have decreased sensation over dermatomes C5 and C6. Muscle strength on both upper extremities was equal and normal. Cranial nerves II–XII were intact. Cardiopulmonary and abdominal examinations were unremarkable. Initial imaging with X-ray of the shoulder revealed a large lytic area involving the right humeral head with an associated pathological fracture through the proximal humeral shaft, with destruction of the articular surface (figure 1A). Whole body Tc-99m bone scan revealed increased focal uptake at the lytic area associated with fracture (figure 1B). CT scan revealed a right upper extremity soft tissue mass with associated lytic changes in the proximal humerus. Core biopsy of the lytic bone lesion demonstrated spindle cells with dark irregular nuclei, irregular and angulated cellular outlines with slightly increased nuclear to cytoplasmic ratio and areas of homogenous eosinophilic matrix (figure 1C). As complete imaging studies were not available at that time, there was a concern of this being osteosarcoma, and the patient was referred to oncology clinic for further evaluation.

MRI, which was carried out for evaluation of chronic neck and back pain revealed degenerative disk changes and disc herniation associated with spinal canal narrowing at C3–C5 levels. A large syrinx extending from C5 level into the upper thoracic cord was noted (figure 1D, E). MRI of the brain did not show any evidence of disease or Arnold-Chiari malformation. The patient's X-ray findings of joint destruction with periarticular bony fragmentation and deformity of the proximal humerus along with MRI findings of syringomyelia led to the clinical diagnosis of neuropathic osteoarthropathy. The patient was managed conservatively with physical therapy by orthopaedic specialty.

INVESTIGATIONS

MRI, whole body bone scan, CT, X-ray and biopsy of the lytic mass were carried out.

DIFFERENTIAL DIAGNOSIS

Degenerative joint disease, septic arthritis, osteomyelitis, soft tissue sarcoma, multiple myeloma.

TREATMENT

Orthopaedic surgery was consulted. No invasive surgical intervention, joint immobilisation, 'collar and cuff' sling, pain management and physical therapy consultation for rehabilitation.

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Figure 1 (A) X-ray of right shoulder joint. (B) Nuclear bone scan showing uptake around the fracture site. (C) H&E staining of core biopsy specimen. (D) T2-weighted MRI of cervical spine. (E) T2-weighted MRI of thoracic spine.

OUTCOME AND FOLLOW-UP

Stabilisation of the joint destruction was performed after sling placement. Follow-up was carried out with orthopaedic specialty at regular intervals.

DISCUSSION

Neuropathic osteoarthropathy, also known as Charcot's joint, is a rare chronic degenerative arthropathy associated with decreased sensory innervation. It was initially identified as a type of destructive arthropathy associated with diseases involving peripheral nerves.¹ In 1868, Charcot² provided a detailed description on the neuropathic aspect of arthropathy in a patient with *tabes dorsalis*. The estimated prevalence of Charcot's arthropathy is approximately 1% of all neuropathic patients and neuropathic arthropathy involving the shoulder joint is even less.³ In a report of 39 neuropathic joints, only two cases were found involving the shoulder.⁴ There are fewer than 60 cases of Charcot's shoulder reported in the literature so far.⁵ Most of the cases affected a unilateral shoulder joint, with only four cases involving bilateral shoulders having been reported.⁶

Neuropathic arthropathy of the shoulder often presents with rapid and extensive destruction of the proximal aspect of the humerus and the glenoid. Pathophysiology of Charcot's osteoarthropathy is unclear and is hypothesised to be secondary to complication from dysregulated neurotraumatic and the neurovascular processes. The former correlates joint damage with repetitive trauma sustained by an insensitive joint and the latter describes active bone resorption by osteoclasts, secondary to sympathetic dysfunction.^{7 8} Although many conditions have been correlated with the denervation, the most common are diabetes, syphilis and syringomyelia. Patients with diabetes tend to have involvement of the joints of the foot and ankle, whereas larger joints such as the knee are commonly affected in patients with syphilis. In patients with syringomyelia, the shoulder and elbow area seem to be affected more than other sites.⁹ It has been reported that neuropathic arthropathies develop in approximately 25% of cases of syringomyelia and of them, 80% involve the upper extremity. Neuropathic joint disease may develop at various times in the course of syringomyelia. Neuropathic arthropathy of the shoulder generally progresses slowly, although rapid progression may occur over months or even weeks after detection of syringomyelia.¹⁰

Syringomyelia is a rare disorder involving a fluid-containing cavity (syrinx) within the spinal cord, disrupting adjacent grey and white matter. Causes include congenital, trauma, vascular, tumour-related, degeneration or infection.¹¹ The nerve fibres damaged earliest are the pain and temperature sensory fibres as

they cross the midline. Their loss in the upper extremity, with intact position sense and motor function, is often the first clinical presentation. It is the loss of these sensory fibres that is thought to result in damage of the shoulder due to unprotected repetitive trauma. As the syrinx grows or propagates, the dorsal column fibres or anterior horn cells may also be affected and symptoms of areflexia, muscle weakness and atrophy may ensue.

Since neuropathic arthropathy of the shoulder is a rare condition resulting in disabling deformities, it is critical to identify such cases from various other shoulder pain symptoms. Clinical presentations of neuropathic shoulder, especially at an early stage, are non-specific. Patients often have symptoms of numbness, dull pain, weakness, stiffness and limited range of motion of the shoulder. Swelling is very common and may produce a mass at the joint due to swelling. Taking a detailed history including previous traumatic injury to the spinal cord may provide valuable clues to correlate shoulder arthropathy with possible presence of syringomyelia. Extent of the pain, association of the pain with movement, and relative duration of disease can also assist in differentiating it from other aetiologies. Musculoskeletal pain of the shoulder is usually severe and associated with activity. For example, pain from adhesive capsulitis (frozen shoulder) is associated with diminished passive range of motion, while patients with rotator cuff tendinitis typically reported pain after active motion. Compared with osteoarthritis, which may have moderate pain symptoms with slow progression over many years, neuropathic shoulder always has mild shoulder pain with rapid progression and shoulder destruction from onset of symptoms, resulting in deformity and loss of joint function within several months. It is usually sufficient to make the diagnosis with findings on conventional radiography, which include sclerosis, joint destruction, bone fragmentation, subluxation and dislocation. However, other conditions such as degenerative joint disease, septic arthritis, osteomyelitis, soft tissue sarcoma and other bone-destructive disease of unknown aetiology should be considered in the differential diagnoses.^{12 13} Further testing with MRI, nuclear medicine bone scan, fluoro-D-glucose-positron emission tomography scans and bone biopsy, may be needed to assist with the diagnosis. Management should be focused on treating the underlying neurological causes before treating the secondary complications of syringomyelia.¹⁴ Options for treating neuropathic shoulder include operative and non-operative. Total joint replacement is contraindicated due to poor bone stock and prosthetic loosening/instability, and soft tissue compromise. Arthrodesis is considered only during the quiescent stage and is not appropriate during inflammatory stage when bone erosion is still ongoing.¹⁵ The primary goal of non-operative, conservative treatment is to

reduce further articular damage by the prevention of repetitive trauma. Shoulder rest, restriction of activity and functional bracing is probably the best solution for long-term management of these patients.¹⁶

What made this case more unique is the timely recognition of the neuropathic osteoarthropathy in this patient with pathological diagnosis of spindle cell neoplasm, for which referral to oncology was made for further treatment. Gunay *et al*⁵ recently reported a case of Charcot's arthropathy caused by syringomyelia, which had been initially misdiagnosed at another institution and, unfortunately, treated with surgery and radiotherapy for a soft tissue tumour of the shoulder joint. For patients who present to orthopaedic service with pain, swelling, decreased range of motion of the shoulder and/or fracture of the proximal humerus, a differential diagnosis of neuropathic arthropathy of the shoulder should be considered. A detailed medical and surgical history (especially including previous spinal cord injuries and any surgical interventions), thorough neurological examination and X-ray of the shoulder, should be pursued. If diagnosis of a neuropathic arthropathy of the shoulder is suspected, MRI of the cervical and thoracic spine should be performed for evaluation of syringomyelia. Further evaluation with needle biopsy could be suggested to rule out septic arthritis and malignancy. However, relying solely on pathological findings, as in our case, could have resulted in delivering inappropriate and potential dangerous treatments to this patient. In summary,

Learning points

- ▶ Charcot's shoulder is a rare degenerative disease presented with decreased sensation and rapid destructive damage to affected joints.
- ▶ Charcot's osteoarthropathy should be considered as one of the differential diagnoses in patients with pathological fracture of the shoulder.
- ▶ It is critical to integrate clinical, pathological and imaging findings for accurate diagnosis and appropriate therapy.

physicians need to be more aware of this destructive joint disease and it is imperative in cases of this type, to integrate clinical, pathological and imaging findings, for accurate diagnosis and for delivering appropriate therapy.

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Competing interests None declared.

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