Letters to the Editor

Sarcoidosis presenting as Heerfordt's syndrome with myopathy

Sirs,

Heerfordt's syndrome was described in a case of uveoparotid fever by Heerfordt in 1909 (1). In 1937, the syndrome was recognized as an unusual feature of sarcoidosis characterized by fever, uveitis, parotid gland swelling and facial palsy (2). Symptomatic muscle weakness in sarcoidosis is relatively uncommon and has been reported in only 2.3% of patients (3). Because of the unusual nature of this entity, we describe a patient to demonstrate the classic clinical, radiographic and pathological features, with discussion of the presenting organ involvement.

A 32-year-old African-American female presented with proximal muscle weakness of the lower extremities and peripheral right facial nerve palsy which began one month earlier. There was no history of tinnitus, hyperacusis, or vertigo. The patient also complained of headache and blurred vision in her left eye. She was afebrile with significantly swollen parotid glands. Visual acuity was decreased in both eyes but worse on the left. Bilateral cervical lymphadenopathy was detected. Neurological examination revealed mild peripheral right facial palsy without other cranial nerve involvement. Muscle strength was 3/5 in the iliopsoas muscles (both sides), and 4/5 in the quadriceps and hamstring muscles (both sides). Motor function in the upper extremities and other muscle groups as well as deep-tendon reflexes, and all modalities of sensation were normal. Laboratory examination including a complete blood count, chemistry panel, urinalysis, serum calcium, creatine phosphokinase (CPK), erythrocyte sedimentation rate, PPD skin test,HIV antibodies and antinuclear antibodies were negative or within normal limits. Angiotensin converting enzyme (ACE) was 268 (normal 8-52). Plain x-ray and computed tomography (CT) scans of the chest showed bilateral hilar lymphadenopathy. Brain CT scan showed no abnormality. Electromyography (EMG) revealed primary myopathy in the muscles of the lower extremities but nerve conduction velocity (NCV) studies were normal. Fine needle aspiration of cervical lymph nodes showed multiple non-caseating granulomas without organisms or malignant cells. Right quadriceps muscle biopsy showed granulomatous myopathy (Fig. 1). Slit lamp examination demonstrated bilateral panuveitis with uveitic glaucoma of the left eye.

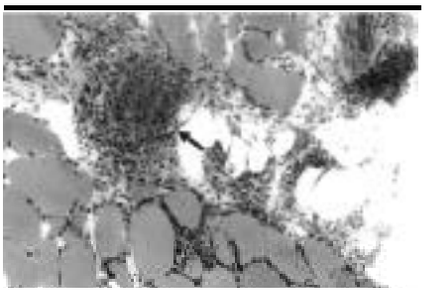

Fig. 1. A large non-caseating granuloma expands the interstitial connective tissue between muscle fibers. It consists of epithelioid cells, lymphocytes, and other chronic inflammatory cells (H&E, low power).

Sarcoidosis occurs most commonly in individuals under the age of 40 with a predominance in African-Americans (4). Women tend to have more fatal complications than men (5). Respiratory tract involvement is the most common manifestation. Common sites of extra-thoracic involvement are the eyes, skin and lymph glands (5). Heerfordt's syndrome is a rare clinical syndrome of sarcoidosis consisting of fever, uveitis, parotid gland swelling, and sometimes facial nerve palsy with female preponderance in the ratio of 2:1 (6). Although fever is one of the clinical features, some patients may have spiking fever only during the period of active disease (6). Ocular involvement occurs in 25-60% of patients with the most common manifestations being uveitis and conjunctival nodules. More than 80% of uveitic patients manifest before or within 1 year after the onset of systemic disease. Posterior ocular involvement and glaucoma indicate a poor prognosis (7). Parotid gland involvement occurs in 6% of patients (8) with 73% being bilateral (9). The swollen parotid glands are usually firm, rubbery in consistency and slightly tender to palpation (6). Neurological involvement occurs in 5-16% of patients, with manifestations ranging from peripheral or cranial neuropathy to central nervous system disease. The most common manifestation of neurosarcoidosis is facial nerve palsy of the peripheral type, presenting in 50% of patients with Heerfordt's syndrome with spontaneous resolution within days or weeks of onset (6). Electrophysiological studies of the whole facial nerve showed the lesion to begin in the cerebellopontine angle with spreading distally into the facial canal (10). Symptomatic sarcoid myopathy is found in only 2.3% of patients with a female preponderance of 3:1 (3,11). While symptomatic muscle weakness is relatively rare, asymptomatic muscle involvement is common, occurring in 50-80% of patients (3,11). CPK may be normal or elevated at some time during the course of the disease. EMG and NCV studies usually show a myopathic pattern (11).

The diagnosis of Heerfordt's syndrome can be made based on the classic clinical features, histological studies of the parotid glands, conjunctiva or other organ involvement, with the exclusion of other potential causes of granulomatous inflammation such as mycobacterial and fungal infection, metal or organic dusts, vasculitides,and malignancies (4,5).Serum ACE and gallium scanning may correlate with disease activity after treatment but have no diagnostic value because of their lack of specificity (4,5).

The treatment of sarcoidosis depends upon the major organ involvement and disease activity, because spontaneous improvement can occur. Oral corticosteroids remain the mainstay of therapy and are indicated for severe ocular, neurologic or cardiac sarcoidosis, malignant hypercalcemia, and symptomatic or severe pulmonary involvement (5). Our patient was treated with 60 mg/day prednisone initially and showed improvement of weakness and visual impairment, and resolution of facial nerve palsy within 1 month. Patients refractory to corticosteroids should be considered for alternative drugs,including methotrexate, hydroxychloroquine, azathioprine and cyclophosphamide (5). Methotrexate was added in our patient after 2 months of high dose prednisone for its steroid sparing effect.

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Spondyloepiphyseal dysplasia tarda in a child with severe and an adult with mild clinical features

Sirs,

Spondyloepiphyseal dysplasia (SED) refers to heritable abnormalities in which the primary skeletal disturbance occurs in the spine and in the epiphyses of the long bones. SED may be categorised in 3 broad groups as follows:the congenital type (SED congenita), with abnormalities recognisable at birth; the pseudoachondroplastic type, which presents in infancy and early childhood; and the tarda type, recognised later in childhood (1). Two cases of spondyloepiphyseal dysplasia tarda (SEDT),one a child with severe and one an adult with mild clinical features, are presented here. The radiologic progression of the disease and the differential diagnosis are discussed.

The first case was a 12-year-old boy referred with progressive dorsal kyphosis, joint deformities, leg pain with activity, and gait disturbance. His physical development was normal up to age 8. His parents were first-degree cousins with no family history of a skeletal disease. His posture was hyperlordotic with an increased base width during gait. His dorsal kyphosis, lumbar lordosis and the antero-posterior diameter of the chest were increased. Moderate limitation of motion was present in both shoulders and hips. There were bony deformities in the medial aspects of the elbow joints, knees and ankles. A comparative radiographic examination at age 8 and 12 revealed progression and expansion of the platyspondyly towards the dorsal region with increasing anterior wedging, irregularities, subchondral sclerosis and Schmorl's nodes at the end-plates of the T8-11 vertebrae. The humeral and femoral heads were flattened and irregularities in the proximal epiphyses were noted.

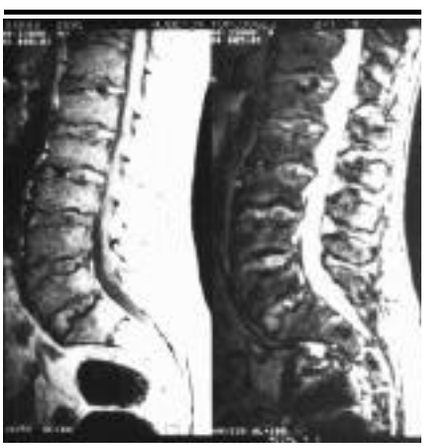

Fig. 1. T1 and T2-weighted sagittal magnetic resonance images of the lumbosacral spine in case 2, demonstrating platyspondyly, Schmorl's nodes and irregularity at the end-plates of the lumbar vertebrae with narrowing of the intervertebral disc spaces, emphasizing the findings in the plain radiographs.

The second case was a 29-year-old male who played soccer as a recreational activity, who was admitted with the complaint of pain in the right hip which became evident on physical activity. On examination he had a relatively short trunk with mild scoliosis and increased dorsal kyphosis. Paravertebral muscle spasm and pain in motion of the lumbar spine were observed, although the spinal mobility was not limited. The range of motion in the hips was limited and painful. The radiographic findings resembled those of the first case with increased degenerative changes in the spine (Fig. 1) and hips.

Spondyloepiphyseal dysplasia tarda, first described by Nilsonne in 1927 (2,3), is a developmental skeletal disorder that usually presents clinically late in the first or early in the second decade of life with progressive involvement of the spine and epiphyses. The diagnosis of SEDT is difficult in a sporadic case without a positive family history, but when suspected clinically radiographic findings are usually sufficient for the diagnosis. Hyperostotic new bone formation on the posterior two-thirds of the articular surfaces of the vertebral bodies is usually distinctive for the X-linked type (3, 4). Degenerative spine and hip disease characterize the later stages of the disorder (3). Dysplastic changes may be observed in other major joints (1,3).

Scheuermann's disease, juvenile lumbar osteochondrosis and spinal manifestations of the mucopolysaccharidoses resemble the spinal findings of SEDT (5). A subtype of pauciarticular juvenile chronic arthritis (JCA) with late onset may also affect the hips (6). JCA and ankylosing spondylitis might have been considered in the differential diagnosis of our cases. The results of the extensive laboratory workup were within normal ranges for the two cases.

We believe that case 1 represents a case of SEDT with severe and progressive features, beginning at an early age. The second case was a fairly mild one clinically. There was no back pain despite the striking radiographic findings. The late clinical presentation and the delay in diagnosis (at the age of 28),despite his physically active lifestyle seems unusual.

We would like to emphasize that SEDT should be considered in the differential diagnosis in patients who have complaints of back and hip pain and gait disturbances even in adulthood. After the diagnosis of SEDT, patients should be advised to engage in appropriate exercises and moderate activity and should be informed about the prognosis and genetic aspects. Overtreatment should be avoided.

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Pachydermodactily as a cause of painful swelling of the knuckles: Successful treatment with intralesional steroids

Sirs,

Pachydermodactily is a fibrosing condition characterized by symmetrical, diffuse swelling of the proximal interphalangeal (PIP) joints of the fingers (1-3). The lesions are usually painless, but may cause pain and stiffness of the PIP joints, which has in