LESSON OF THE MONTH

A young boy with deforming arthropathy

R Handa, P Aggarwal, J P Wali

Case report

A 15 year old boy presented with a three year history of painless swelling of the proximal interphalangeal (PIP) joints of both the hands. The illness started at the age of 12 years without any history of preceding trauma or cold exposure. There was no fever, morning stiffness or pain. There was no limitation of hand functions like writing skills, grasping, etc. However, for the past six months, the patient had noticed flexion deformities of the little fingers, which could not be corrected. No other joints were involved. The patient was one of three children of a non-consanguineous marriage. None of the parents or other siblings had similar illness. The patient had been treated by his doctor for juvenile chronic arthritis with diclofenac sodium and low dose prednisone for six months without any improvement. General physical and systemic examinations were entirely normal. Joint examination showed firm swelling of the PIP joints. No erythema, local rise in temperature or tenderness were noted. The fingers showed flexion deformity. No shortening was apparent. Active and passive movements were normal except in the little fingers, which had a fixed deformity (fig 1). All other joints of the hand and feet were normal.

Investigations showed haemoglobin 14.2 g/dl, leucocytes $6.1 \times 10^9/l$ (neutrophils 70%, lymphocytes 25%, monocytes 3%, eosinophils 2%) and platelets $300 \times 10^9/l$. Westergren erythrocyte sedimentation rate (ESR) was 5 mm 1st h. Rheumatoid factor and antinuclear antibodies were negative. Antistreptolysin O titres were normal. Blood urea nitrogen, serum creatinine, serum proteins, aminotransferases were within normal limits. Urine analysis was normal. Hand radiographs showed irregularity

Figure 2 Radiograph of the hands showing epiphyseal irregularity of the second phalanx of right and left ring fingers. PIP joints of little fingers are fused.

of the proximal epiphyses of the second phalanx of right and left ring fingers with fusion of PIP joints of the little fingers. No erosions were noticed (fig 2). A diagnosis of Thiemann's disease was considered and the patient reassured with withdrawal of all medications.

Discussion

Thiemann's disease or aseptic necrosis of the basal epiphyses of the phalanges of the fingers is a rare condition first described in 1909.¹ The principal clinical manifestation is progressive enlargement of PIP joints of the hands, and of interphalangeal joints of the great toes and occasionally other toes, followed by slight flexion of the enlarged joints.² The swelling is generally painless and pain, if present, is usually slight and often triggered by cold.³⁻⁵ The disease typically begins in the prepubertal age group and limitation of function is slight. Most often the disease is transmitted as an autosomal dominant disorder with virtual complete penetrance,^{3,4} although, sporadic cases are not infrequent.⁵ The familial disease shows equal sex distribution while sporadic cases show a threefold male dominance.^{5,6} Acute phase reactants like erythrocyte sedimentation rate are normal.

The proposed clinical criteria⁴ include onset before the age of 25 and swelling of PIP joints with normal laboratory tests. Radiological features include irregularity, flattening, fragmentation, and broadening of the basal epiphyses of the phalanges.^{3,5,7} These are later followed by joint space narrowing, premature epiphyseal

Department of
Medicine, All India
Institute of Medical
Sciences, New Delhi,
India

R Handa
P Aggarwal
J P Wali

Correspondence to:
Dr R Handa, Rheumatology
Division, Department of
Medicine, All India Institute
of Medical Sciences, New
Delhi 110029, India.

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Figure 1 Photograph of the hands. PIP joints are swollen and the fingers show flexion deformity.

Table 1 Hand deforming arthropathies in children

Condition	Differential diagnosis
Juvenile chronic arthritis	Pain, swelling, erythema, warmth present in the affected joints. ESR and CRP usually raised in active disease.
Systemic lupus erythematosus	Arthropathy is inflammatory, non-erosive, and most often non-deforming, although reversible or fixed deformities may occur. One of the osteochondroses with painless enlargement of PIP joints of hands and feet. Clinical and laboratory features of inflammation absent, ESR normal. Characteristic radiological picture (described in text).
Thiemann's disease	Syndrome of insulin dependent diabetes mellitus, short stature and flexion contractures of finger joints. Pain is typically absent. Functional disability is common. Nodular thickening of the palmar fascia with flexion contractures of the digits.
Diabetic cheiroarthropathy (Rosenbloom syndrome)	Congenital contractures of knees, elbows, and PIP joints, which tend to improve with age. Typically hand and feet are long. Progressive kyphoscoliosis may develop. Obvious history of trauma present. History of cold injury present. Distal phalanges more severely affected. Sparing of thumb characteristic.
Dupuytren's contracture	History of electrical or thermal injury with prominent skin and soft tissue involvement. Multiple organ involvement seen in addition to stiffness and deformity of hands, elbows, and knees.
Congenital contractural arachnodactyly	Transient swelling and tenderness of hands and feet seen in children with sickle cell anaemia below 4 years of age. Radiography reveals periosteal elevation, new subperiosteal bone formation, radiolucent areas intermingled with areas of increased density ("moth eaten appearance")
Traumatic arthropathy	Degenerative arthropathy involving MP, PIP, DIP joints, hips and knees seen in 20–40% cases. Rare in children because significant iron overload takes several years.
Frostbite arthropathy	Occurs most often in the setting of pulmonary tuberculosis. Tuberculous arthritis typically involves large joints of lower limbs. Dactylitis uncommon.
Burns	
Storage diseases, for example, mucopolysaccharidoses	
Sickle cell dactylitis	
Haemochromatosis	
TB dactylitis	

fusion, and phalangeal shortening. In some cases, radiographic improvement may be seen because of resorption of bone fragments, but more frequent is the later development of interphalangeal joint osteoarthritis.⁵ Most often, the disease has a benign course and is not functionally disabling, although interphalangeal joint osteoarthritis may develop.

Thiemann's disease is now thought to be one of the osteochondroses. Osteochondroses are a group of disorders that share certain features: predilection for the immature skeleton; involvement of an epiphysis, apophysis, or epiphysoid bone; a radiographic picture that is dominated by fragmentation, collapse, sclerosis and frequently reossification with reconstitution of the ossification contour.⁸ Osteochondroses characteristically affect children between the ages of 3 and 12 years and are much more common in boys than in girls. It is presumed that trauma, and in some instances, avascular necrosis unrelated to trauma are the pathogenetic mechanisms.⁹ The clinically important members of this heterogeneous group of disorders include: Legg-Calve-Perthe's dis-

ease, Kohler's disease, Osgood-Schlatter's disease, Thiemann's disease, and Scheuermann's disease.⁹

The most important differential diagnosis of Thiemann's disease is from other inflammatory paediatric arthritides like juvenile chronic arthritis and systemic lupus erythematosus. Acute phase reactants are usually, but by no means invariably, raised in juvenile chronic arthritis while the articular involvement in systemic lupus erythematosus is usually non-deforming. The characteristic age of onset in Thiemann's disease with painless bony enlargement of PIP joints without any redness or warmth, conspicuous lack of systemic features, and absence of acute phase reactants coupled with a benign course make clinical differentiation possible. Primary generalised osteoarthritis begins later in life and usually shows less symmetrical affection, without shortening of the phalanges.⁴ Other conditions that may mimic Thiemann's disease like trauma, infection, thermal injury such as frostbite or burns, storage diseases, for example, mucopolipidosis III, diabetic cheiroarthropathy, contractural arachnodactyly, etc, can be easily distinguished on clinical or radiological grounds^{5 10 11} (table 1). Awareness of this rare disease is important not only for greater clinical recognition but also avoidance of unnecessary treatment with anti-inflammatory or disease modifying agents.

The lesson

- Deforming arthropathies in children could be caused by juvenile chronic arthritis, systemic lupus erythematosus, trauma or thermal injury, diabetic cheiroarthropathy, storage diseases, contractural arachnodactyly, sickle cell disease, and Dupuytren's contracture.
- Painless swelling of PIP joints in adolescent children, with preserved function and normal erythrocyte sedimentation rate, should arouse suspicion of Thiemann's disease.

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