DISAPPEARING BONE DISEASE
WITH ARTHROPATHY AND SEVERE SCARRING OF THE SKIN
A Report of Four Cases Seen in South Vietnam

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During a military sojourn in Vietnam the author served as orthopaedic surgeon to the Saint Francis Leper Colony in Qui Hoa. One clinic day a Vietnamese woman was presented by a Sister who said: “She does not have leprosy but has a problem with her hands and feet”. This was the first of four patients described in this paper. After consulting a number of doctors experienced in tropical diseases, and after a thorough review of the literature, the author formed the opinion that this disease had not been described. Although the language barrier and inadequate diagnostic facilities limited the investigations, it was considered that the clinical picture was worth recording. This report adds to the literature yet another disease in which there is “disappearing bone”.

CASE REPORTS

Case 1—A Vietnamese woman aged fifty-two presented with extensive scarring of the fingers and toes without pain. The history dated back to the age of twenty-four when the patient recalled the gradual onset of multiple infections in the feet and then the hands. This was associated with joint pain, swelling, and increased temperature in the region of the joints.

FIG. 1
Case 1. Figure 1—Photograph showing the characteristic scarring of the face, which was present in similar distribution on the opposite side. Figure 2—Scarring will be noted on the extensor surfaces of the lower extremity, the hands and feet. There is gross deformity of the hands with loss of bony architecture and subluxation of several joints. In the feet there was shortening of the toes with subluxation and gross dorsal displacement.
She also had general malaise at that time. No numbness or weakness of the extremity was noted. The patient's general medical history had been unremarkable except for an episode of coughing and fever ten years before the examination. Her mother and maternal uncle were said to have had similar problems. The patient had six siblings, none of whom had any disability.

Examination was within normal limits except for the features to be described. The skin of the face, hands and extensor surfaces of the forearm revealed extensive scarring (Figs. 1 and 2). The scars resembled old moderately hypertrophic burn scars. There was no ulceration of the hands or feet. The phalangeal joints of the hands and feet showed multiple painless subluxations and dislocations. In some instances loss of bony continuity had left the digits completely flail. Musculo-skeletal examination was otherwise normal. Sensibility was normal, as were the peripheral pulses and deep tendon reflexes. Motor function was compatible with the destruction of bony architecture and there was no evidence of muscle weakness or paralysis.

Haemoglobin level, haematocrit, sedimentation rate, white count and differential count were normal. The chest radiograph was normal. A serological test for syphilis (routine flocculation) was positive (1:2 at a 1:2 titre). Microbacterium leprae were not found in the nasal smear.

Skin biopsy from a characteristically scarred area of the wrist showed epidermal atrophy, hyperpigmentation, and hypertrophy of collagen. The histological changes were thought by the pathologist to be suggestive of latent scleroderma. He stated also that no arteries were present in the section to permit evaluation of endarteritis. Radiographs of the hands and feet showed dissolution of bones and joints with multiple dislocations of the metacarpo-phalangeal and interphalangeal joints.

Case 2—A Vietnamese man aged sixty-one was being treated for a painful amputation stump when deformities of his right hand were noted. The patient stated that at the age of twenty there had been an extensive infection involving the hand and the right lower extremity. He recalled joint pains involving the right leg and the right hand. He also recalled that he had had general malaise and fever. One and a half years before examination he had undergone above-knee amputation for a "severely deformed right lower extremity", presumably due to this same disease. (The patient was intentionally vague about exactly when, where and why this amputation had been done. The reason for this is thought to have been political.) At the time that the history was taken the patient was in good general health without pain either in his hand or in the amputation stump. He denied any family history of similar disability.

Examination was within normal limits except for the findings described below. There was moderate scarring of the right arm, hand and face. The right hand showed deformed
flail digits held to the distal radius and ulna by soft tissues only (Figs. 3 to 5). Palpation confirmed the absence of carpal and metacarpal bones. There was no evidence of vascular or neural disturbance. Movement of the fingers was present but grossly distorted due to loss of bony architecture. The left hand was normal. The left leg showed anterior bowing and prominence of the tibia. The above-knee amputation stump was satisfactory and the rest of the musculo-skeletal system normal.

A serological test for syphilis (routine flocculation) was positive (· 2 at a 1:2 titre). The chest radiograph was normal. Microbacterium leprae were not found in the nasal smear. Radiographs of the right hand showed loss of all of the carpal bones with the exception of the lunate and part of the scaphoid (Fig. 6). There was complete loss of all the metacarpal bones and the entire proximal phalanx of the ring finger with patchy deossification of the remaining bones of the hand and wrist.

**Case 3**—A Vietnamese man aged fifty-one years was first seen for non-union of a fractured left tibia. He was noted to have extensive scarring of both upper and lower extremities and the face. He had deformities of the hands and feet and absence of the nose. He reported an episode of fever and infection of the extremities at the age of seventeen. He did not remember if there had been any pain, numbness or swelling associated with the illness. No other medical history could be obtained.

Examination was otherwise normal and the patient was strong and appeared to be in good general health. The external nares were absent, and extensive, well developed scars

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**FIG. 6**
Case 2—The radiograph of the right hand shows the metacarpals absent, the phalanges all present except for the proximal phalanx of the fourth digit and the carpal bones all absent except for the lunate and part of the scaphoid.

**FIG. 7**
Case 3—Photograph showing complete loss of the nose, scarring of the face and deformities of the hands.

**FIG. 8**
Ulcers are present on the leg as well as scarring and subluxation of the right big toe.
were present on the face and the extensor surface of the forearms and legs (Fig. 7). The legs showed loss of pigmentation and very thin skin in some areas (Fig. 8). There were also several ulcers on the anterior aspects of the legs. There was considerable shortening and malposition of the digits of both hands (Figs. 9 and 10). Examination of the rest of the musculo-skeletal system was normal. There was no evidence of neural or vascular deficiency.

The serological test for syphilis (complement fixation) was positive (+2 at a 1:4 titre).

The haemoglobin level, haematocrit, white cell count, differential count, sedimentation rate and urine analysis were normal. Microbacterium leprae were not found in the nasal smear. A chest radiograph was normal. Radiographs of the hands showed partial dissolution of the proximal phalanx of the left index finger with disruption of some of the metacarpo-phalangeal and interphalangeal joints.

**Case 4**—A Vietnamese youth aged seventeen years without symptoms was presented to the author as an example of a possible early case of this disease. Several years previously he had
had infections of the hands and feet. Examination revealed a youth of normal appearance except for scarring of the right hand, the wrist, and both legs on their extensor surfaces (Figs. 11 and 12). There was moderate anterior bowing of both tibiae. Sensibility and movement of all extremities were within normal limits. There was no suggestion of impairment of blood supply in the limbs. There was some shortening of the index finger of the right hand.

The serological test for syphilis (routine flocculation) was positive (+3 with a titre of 1:8). Microbacterium leprae were not found in the nasal smear.

**DISCUSSION**

The first case of disappearing bone disease was reported 138 years ago under the title “A Boneless Arm” (Jackson 1838). The physical findings are best summarised by this vivid description: “Ordinarily the right arm swings hither and thither, like a thong with a weight at the extremity: for the forearm and hand, with reference to the division above the elbow, constitute a pendulum, oscillating according to the movements of the body. . . . To show the perfect non-resistance of the apparatus of muscles, arteries, veins and nerves in the soft, boneless space, we saw him twist the palm of the hand . . . twice round, which consequently presented the strange anomaly of having all the apparatus of the arm twisted like the strands of a rope.”

Since this classical case report a number of articles have appeared describing osteolysis in different clinical settings (Thévenard 1942; Schinz, Baensch, Friedl and Uehlinger 1951; Thévenard 1953; Gorham, Wright, Shultz and Maxon 1954; Gorham and Stout 1955; Krikler 1955; Aston 1958; Branco and Silva Horta 1958; Butler, McCance and Barrett 1958; Johnson and McClure 1958; Jones, Midgley and Smith 1958; Milner and Baker 1958; Thieffry and
Sorrel-Déjerine 1958; Mahoudeau, Dubrisay, Elissalde and Sraër 1961; Martel, Holt and Cassidy 1962; Shurtleff, Sparkes, Clawson, Guntheroth and Mottet 1964; Lagier and Rutishauser 1965; Torg and Steel 1968, 1969). While the detailed pathophysiology and etiologies have not been worked out, the different clinical pictures have been well reviewed and categorised (Gorham and Stout 1955, Johnson and McClure 1958, Torg and Steel 1969).

Torg and Steel (1968) suggested three basic categories as follows.

**Essential osteolysis with nephropathy**—These cases are characterised by progressive and ultimately total resorption of the carpus and tarsus, with partial resorption of adjacent tubular bones. There is an associated renal disturbance that eventually leads to hypertension, azotemia, and death in early adult life (Mahoudeau, Dubrisay, Elissalde and Sraër 1961; Shurtleff, Sparkes, Clawson, Guntheroth and Mottet 1964).

**Idiopathic hereditary osteolysis**—This disease is thought to be transmitted as an autosomal dominant. There is multicentric involvement of bone with sclerosis, collapse and resorption of the carpus and tarsus but no involvement of adjacent tubular bones. There is no renal disease, skin involvement, arthropathy or neurological involvement (Thieffry and Sorrel-Déjerine 1958).

**Gorham's massive osteolysis**—This unifocal involvement of bone is associated with histological evidence of replacement of bone by haemangiomatous or lymphangiomatous tissue. The skin is not involved and renal function is normal. There is no family history (Gorham, Wright, Shultz and Maxon 1954; Gorham and Stout 1955; Aston 1958; Branco and Silva Horta 1958; Butler, McCance and Barrett 1958; Johnson and McClure 1958; Jones, Midgley and Smith 1958; Milner and Baker 1958; Lagier and Rutishauser 1965).

Another well described clinical picture in which bone disappears is *l'acropathie ulcero-mutilante familiale*. This is a familial disease characterised by dissolution of bone, arthropathy, ulceration of the skin, and is associated with motor and sensory neuropathy (Thévenard 1942, 1953).

Trauma, Sudeck's atrophy, various diseases of the nervous system, gout, scleroderma, reticulosis, rheumatoid arthritis (*la main en longnette*), malum perforans, and congenital pseudarthrosis have been described as sometimes associated with disappearing bones. It is obvious from the cases that none of these diseases is involved here.

Several diseases, however, do merit consideration and discussion in relation to these case reports. The patients were living in a leper colony and had positive serological reactions. The patients did not have leprosy however. There was no neurological deficit and skin changes were not characteristic of leprosy. The patients did not have positive nasal smears for microbacterium leprae.

It is tempting to theorise that these cases represent the end-result of some active infection that had run its course and left sequelae. During the acute episode when there was pain, fever and malaise, the joints were destroyed along with either extensive softening or dissolution of bone. There was at the same time extensive destruction or ulceration of skin which healed with extensive scarring. While the positive serological tests and anterior tibial bowing suggest a treponemal agent, it should be pointed out that the clinical picture does not fit that of syphilis, pinta or yaws. The gumma of syphilis, for example, is a radiopaque lesion (Middlemiss 1961). It is well known that a variety of conditions can give a positive test for syphilis.

The syndrome described in these cases is thought thus to represent a distinct clinical entity not previously described. It was not possible to record and investigate these patients sufficiently to describe the disease completely. More tissue for histologic examination, more specific laboratory examination for treponemal disease and more detailed and reliable family histories would have been desirable. The cases are presented because the information available does strongly suggest a distinct previously undescribed disease entity. It is hoped that a record of these cases will stimulate the recognition of similar cases and lead ultimately to a more complete description of the disease.
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SUMMARY

Four cases are presented of patients seen in a leper colony in South Vietnam. The patients presented with evidence of osteolysis, arthropathies and severe scarring of the skin. They were without pain and in generally good health. There were no neurologic or vascular deficit. They had positive serological tests for syphilis. Other known types of osteolysis are discussed including leprosy and the treponemal diseases, and the cases are presented as a previously undescribed entity.

REFERENCES
