

BONE AND JOINT CHANGES IN PATIENTS WITH PORPHYRIA TURCICA

A Clinical and Roentgenologic Study

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Involvement of the bones and joints in porphyria turcica (the toxic cutaneous porphyria seen in southeastern Turkey since 1955) is now known to be one of the main features of the disease.¹⁻³ In a survey of 592 cases the incidence of bone and joint involvement was reported to be 36.1 per cent.⁴ The average duration of porphyria in these 592 cases was approximately two years. In a more recent appraisal of the course of the disease in 376 patients who had been followed for periods of from three to five years the striking fact emerged that, whereas the acute symptoms and signs had practically disappeared and porphyrinuria had ceased in the great majority of cases, the incidence of arthritic changes had increased significantly.

The purpose of this paper is to discuss the bone and joint involvement in porphyria turcica in greater detail than was possible in previous reports.

Clinical examination of 376 patients with toxic cutaneous porphyria revealed that 206 patients (54.8 per cent) showed fusiform swelling of the fingers. Three hundred and thirty-eight of all the ob-

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served cases of porphyria turcica were seen in children between the ages of seven and eighteen years. Arthritic changes were present in 196 cases (58 per cent) in this age group, whereas only 10 out of 33 patients beyond the age of eighteen (30.3 per cent) were so affected. All of the five patients below seven years of age escaped arthritic complications. Sex did not play a significant role in determining the incidence of arthritic complications in this series of cases.

The early changes in the joints are swelling and spindling of the fingers. There is some limitation of motion and later soft tissue contracture, but surprisingly little pain or tenderness. In most of these cases joint involvement is symmetrical. (Figure 1.)

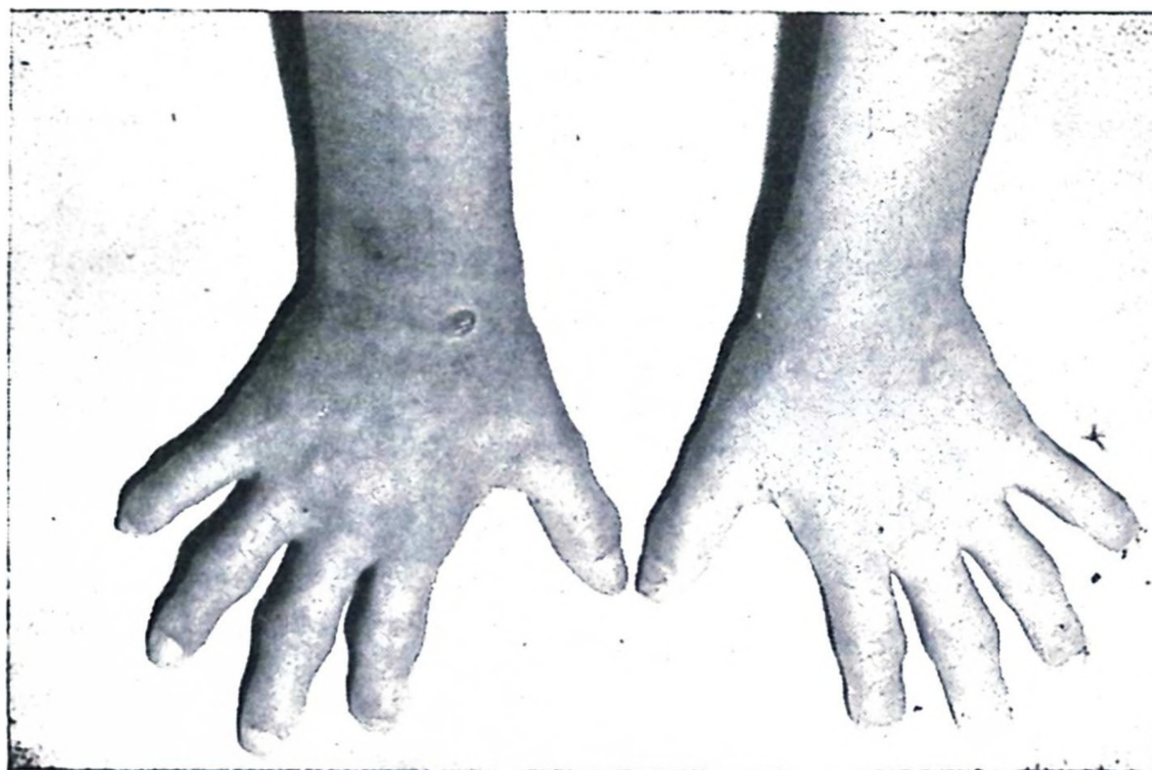


Fig. 1. S.A. (Case 2 in table 1)
Showing spindling of finger joints.

No disturbance in other joints could be found by clinical examination of these patients. In the course of three to five years the manifestations of porphyria, including the abnormal excretion of porphyrin, have largely subsided in the great majority of the patients, but the joint involvement seems to show no clinical signs of improvement.

Biopsies of affected phalangeal joints in two of our patients (S. A. and Z. K., cases 2 and 18 respectively, in table 1) showed no pigmentation on gross examination.

The typical intense red fluorescence which was observed in the liver biopsy specimens³ was not seen in the cartilaginous tissue of these patients under fluorescent microscopy. The green fluorescence seen in the cartilage biopsy material was not significant since it was also seen in fluorescent microscopy of cartilage tissue taken from normal controls.

Eighteen of these porphyria patients with joint changes were examined roentgenographically at the Hacettepe Children's Hospital and the findings are summarized in table 1. The significant roentgenologic finding in the early stages is osteoporosis, which is notably restricted to the phalanges, the metacarpal and carpal bones, and the distal metaphysis and epiphysis of both the ulna and the radius, and to the corresponding bones of the lower extremities. This rarefaction is especially pronounced at both ends of the short bones.

Interphalangeal arthritis, leading to the narrowing of the joint spaces, is another significant feature of the disease. There is no irregularity of the joint surfaces. Occasionally erosion of the terminal digital phalanges may be encountered. This was found in two of the present series of 18 cases. The osseous structure of the long bones and vertebrae shows no remarkable change. The larger joints remain intact. (Figures 2 - 4).

The roentgenologic appearance of the soft tissues in many instances shows fusiform swelling around the affected joint. At times bullae could also be seen in the x - ray films. (Figure 4 a.)

On re - examination of some of the patients with marked bone and joint involvement at a period one to two years after they had ceased to excrete unusual amounts of porphyrins, it was found that the joint spaces remained narrow and there was no change in the periarticular swelling of the affected joints, although in some cases the bones had returned to normal density.

Discussion

Although peripheral neuritis and pains in the joints are not uncommon occurrences in some cases of porphyria, to our knowledge there have been no previous descriptions of clinical or roentgenological changes of the type observed by us in this group. It is difficult to refrain from describing these joint manifestations as «rheumatoid» but in several respects they do, in fact, differ from rheumatoid joints. First, the only joints clinically involved have been those of the hands, although we have seen some osteoporosis in the metatarsals. Second, this appears to be a painless process.

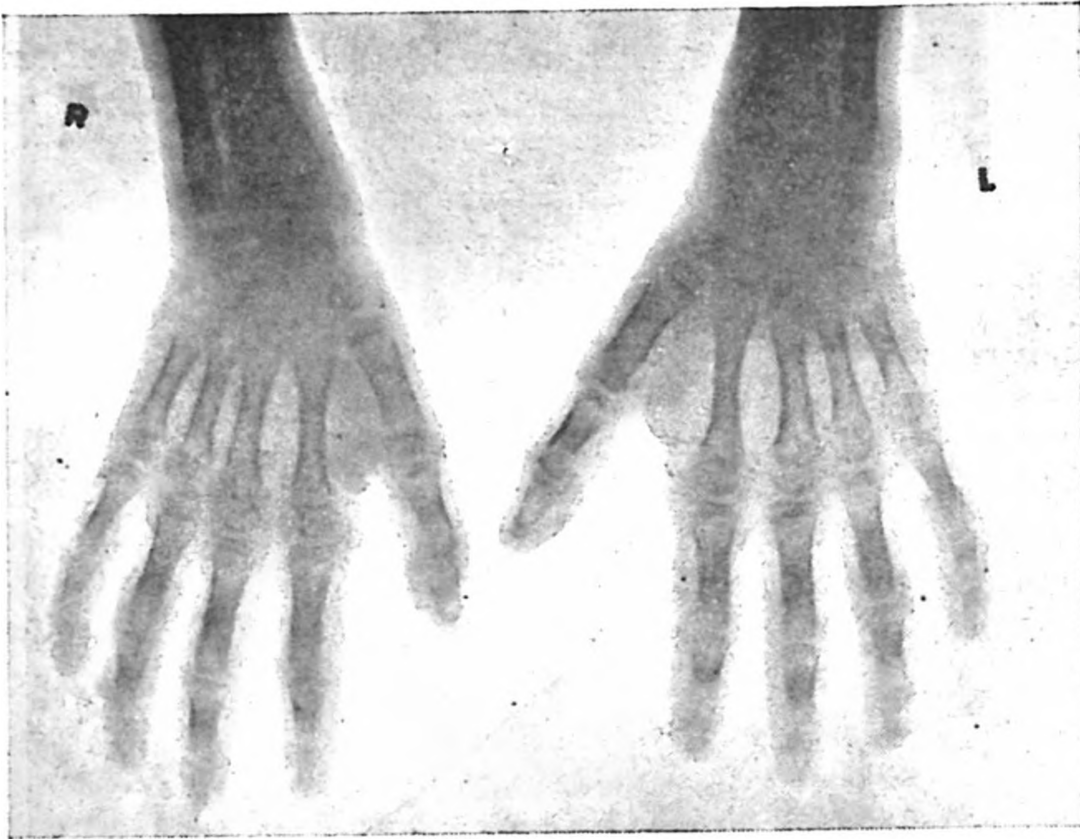


Fig. 2. B.M. (Case 3 in table 1)

Radiograph showing generalized osteoporosis with «rheumatoid» changes of the interphalangeal joints.

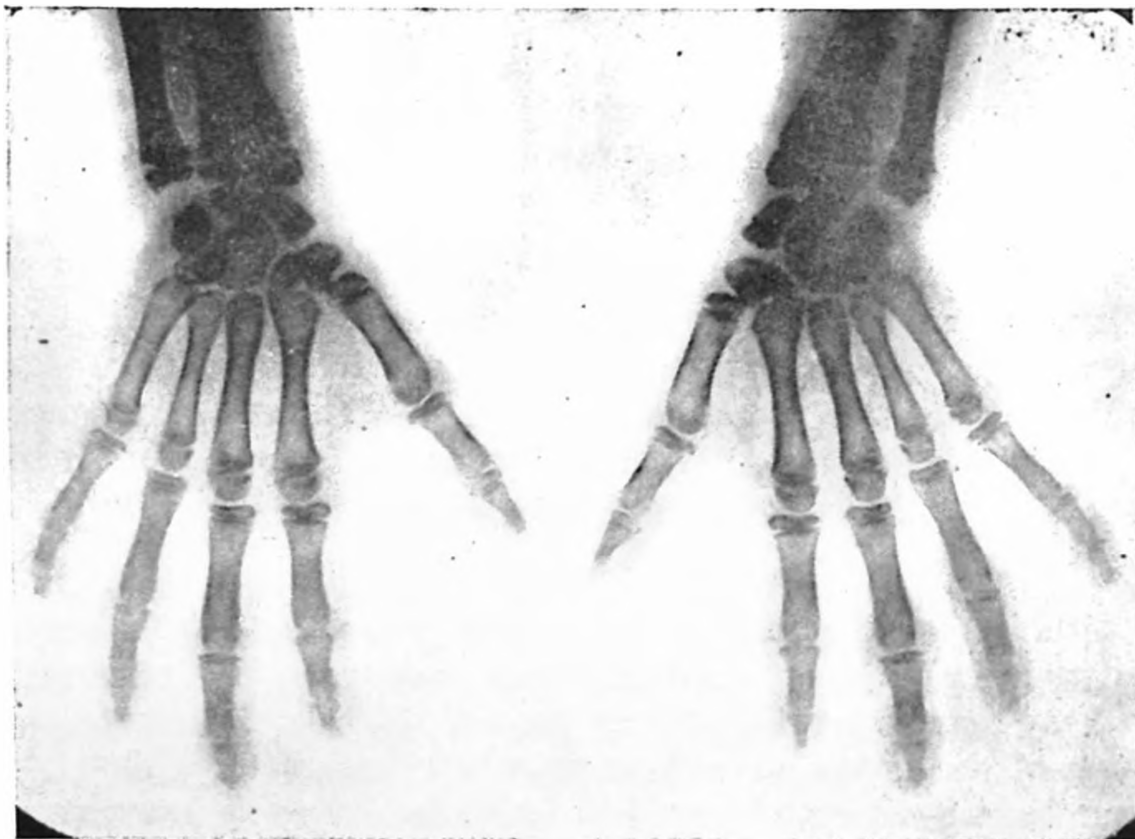


Fig. 3 a. S.A. (Case 2 in table 1)

In anterior posterior position the radiograph shows spindle formation of the digits, narrowing of the interphalangeal joint spaces and irregular bony trabeculation in the metaphyses of the phalanges. Note erosion of the tufted portion of the terminal phalanges of both index fingers and terminal phalanx of the right little finger.

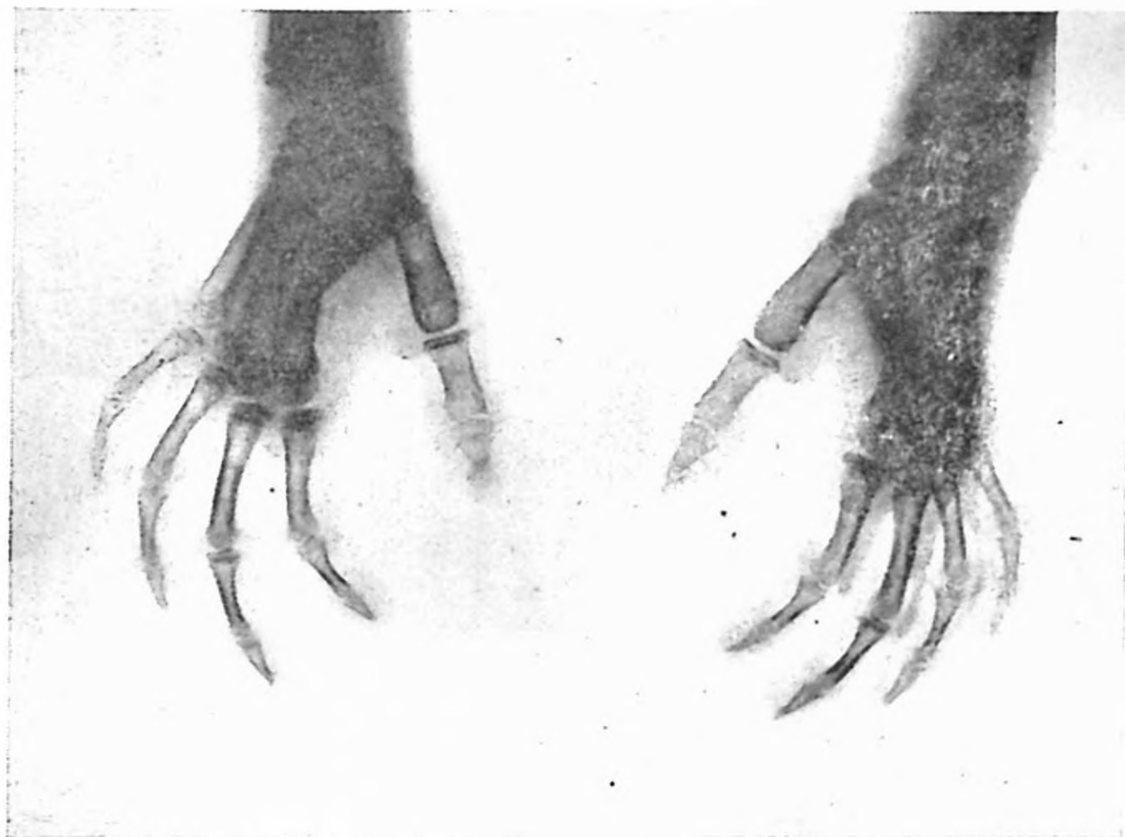


Fig. 3 b.
Lateral position same patient.



Fig. 3 c.
Same patient. Radiograph showing osteoporosis of the metatarsals and phalanges.

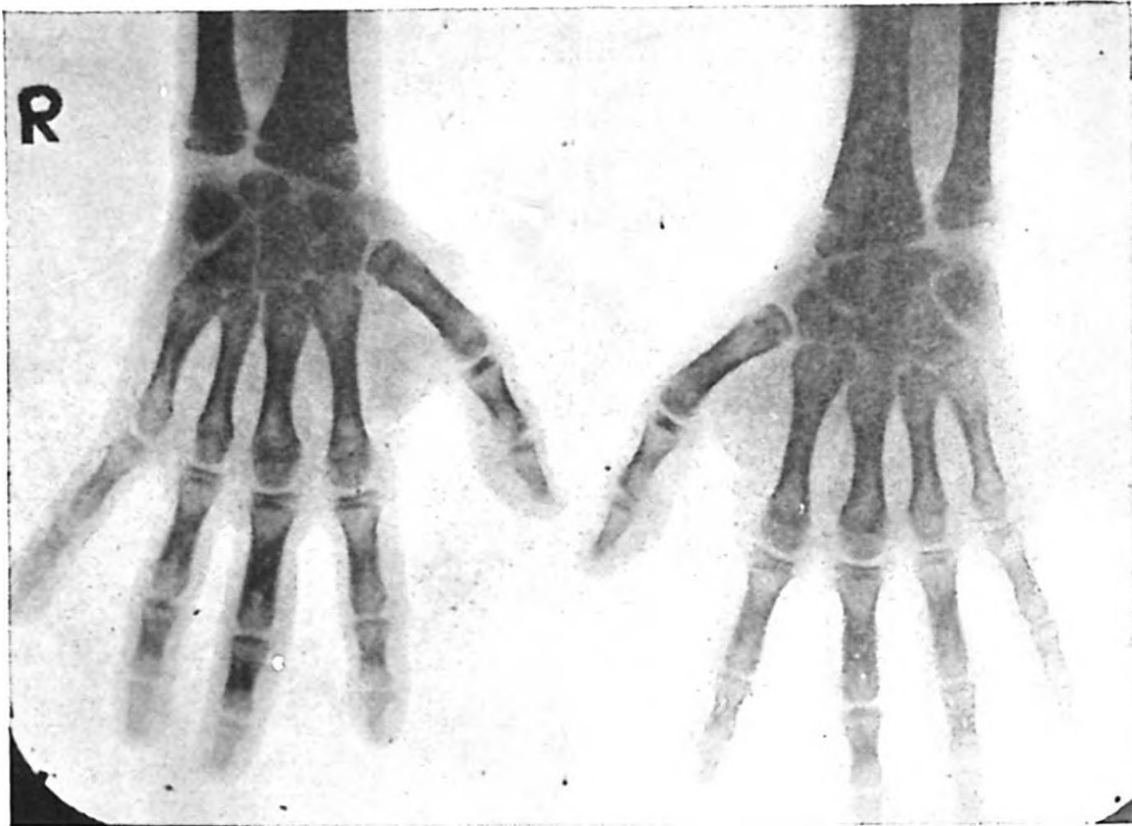


Fig. 4 a. S.A. (Case 4 in table 1)

The radiograph shows osteoporosis of the carpal, metacarpal and phalangeal bones with swelling of the periarticular soft tissues and narrowing of the interphalangeal joints. The swelling of the soft tissue in the right hand is due to bullae.

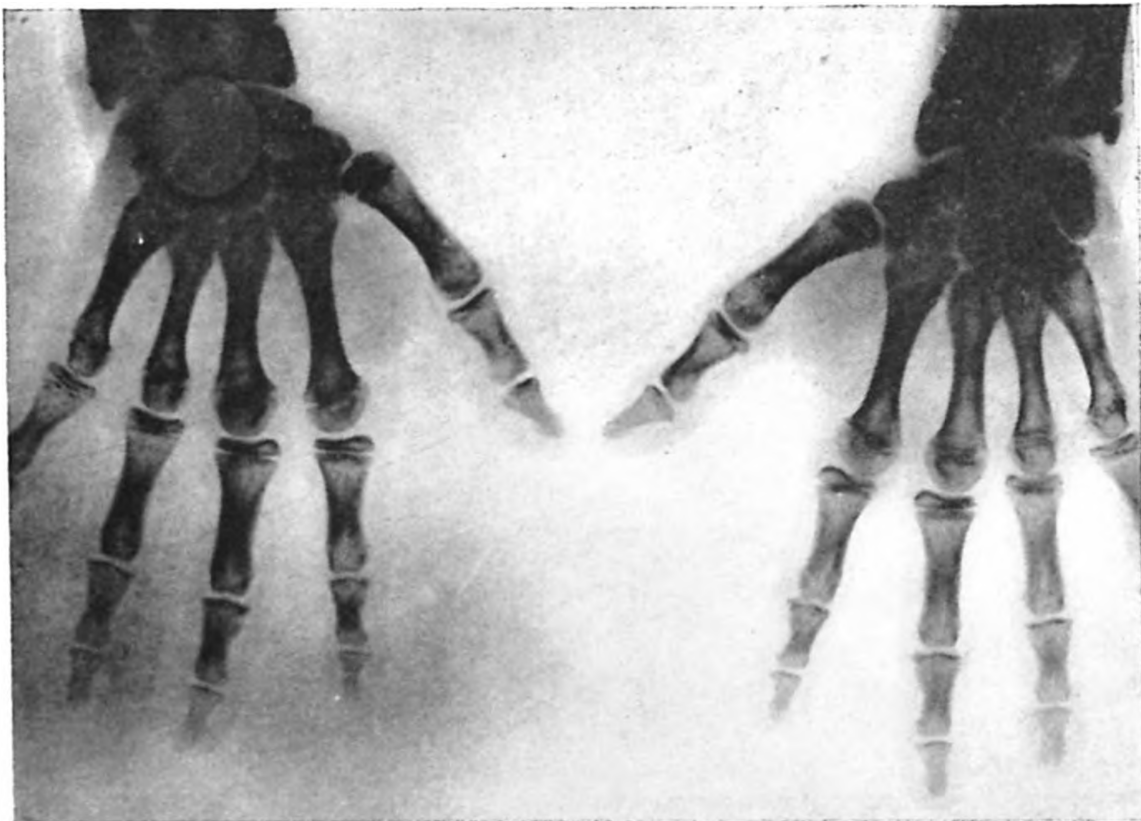


Fig. 4 b.

Same case 18 months later. Osteoporosis has disappeared but the spindle form of the interphalangeal joints and narrowing of the joint spaces persist.

TABLE 1.
ROENTGENOLOGIC FINDINGS IN EIGHTEEN CASES OF PORPHYRIA TURCICA

CASE	AGE IN YEARS	SEX	NARROWING OF THE JOINT SPACES	PERIARTICULAR SOFT TISSUE SWELLING	OSTEOPOROTIC CHANGES	COARSENING OF BONY TRABECULATION	EROSION OF THE TERMINAL PHALANGES
1 V. C.	13	F	0	0	0	0	0
2 S. A.	10	F	+	+	+	+	+
3 B. M.	12	M	+	+	+	+	0
4 S. A.	13	M	+	+	+	0	0
5 S. G.	13	M	+	+	+	+	+
6 M. T.	11	M	+	+	+	+	0
7 A. H.	13	M	+	+	+	+	0
8 C. T.	9	M	0	0	+	0	0
9 S. G.	11	F	0	0	+	+	0
10 A. T.	7	F	+	+	+	+	0
11 A. E.	14	F	+	+	+	0	0
12 H. S.	12	F	0	0	0	0	0
13 I. K.	11	M	0	0	0	0	0
14 A. O.	14	M	+	+	+	+	0
15 S. C.	7	M	0	0	0	0	0
16 N. T.	12	F	0	0	0	0	0
17 B. K.	35	F	+	+	0	+	0
18 Z. K.	18	F	+	+	+	+	0

In another condition known to be due to an enzymatic error of metabolism, alkaptonuria, a black polymer of homogentisic acid accumulates in the cartilaginous surfaces of joints producing destruction of the surface and subsequent arthritic changes. However, since fluorescent microscopy failed to demonstrate typical red fluorescence in cartilage biopsy specimens from our affected patients, it cannot conclusively be said that the arthritic changes observed in these patients are the result of porphyrin deposits in the joints.

Summary

In over half of three hundred and seventy-six patients with porphyria turcica, secondary to consumption of hexachlorobenzene treated wheat, joint involvement was observed.

The striking feature was swelling and spindling of the fingers, usually in a symmetrical fashion. Roentgenologic examination of eighteen patients showed osteoporosis which was notably restricted to the phalanges, the metacarpal and carpal bones, and the distal metaphysis and epiphysis of both the ulna and the radius and to the corresponding bones of the lower extremities.

Interphalangeal arthritis leading to the narrowing of the joint spaces was striking and resembled in a way the changes of rheumatoid arthritis.

In two cases erosion of the terminal phalanges was seen.

Despite the lapse of one to two years after the termination of other porphyria symptoms, including cessation of abnormal excretion of porphyrins, the joint lesions were found to be still present.

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