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DISAPPEARING BONE DISEASE, MORBUS GORHAM

Report of a Case

By

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Disappearing bone disease, Morbus Gorham, haemangiomatosis, lymphangiomatosis, massive osteolysis, spontaneous absorbtion of bone, phantom bone, primary lymphangioma and cryptogenetic progressive localized osteolysis are various names for the same skeletal disease.

The disease is characterised by osteolysis with bone disappearance and histologically pictures occur which are best interpreted as angiomatosis sometimes described as haemangiomatosis, sometimes as lymphangiomatosis.

The disease may affect only one bone but very often several adjacent bones are involved. Histologically the picture is benign. Malignant pathological tissue has never been observed. Metastases have never been seen. Deaths from the disease have been reported but then the process was localised to the rib bones or vertebrae and fatal complications arose owing to involvement of the pleurae and the spinal canal (4, 9, 10, 11, 17). As a rule the disease makes slow, irregular, local progress. The osteolysis may however cease as inexplicably as it began. No regeneration of the vanished bone has ever been observed. Generally the process does not confine itself to one bone but attacks skeletal parts. Spontaneous fractures are common. The osteolysis often commences at a young age. Boys and girls are affected about equally.

Disappearing bone disease is not accompanied by general symptoms. No endocrinal disturbance is present, nor any apparent chemical blood changes, except during acute phases when the alcaline phosphatases may show a moderate increase.

The disease may affect all skeletal parts. Only a few cases are reported involving the skull (12, 21).

The diagnosis is made with the aid of radiology and biopsy. Considerable surprise is experienced when this disease is seen for the first time for there is very little discussion of it in current textbooks.

Arteriography has not been of any diagnostic aid, but possibly an intraosseous injection of contrast-medium may be of some use, as the contrast-medium is mainly collected in the angiomatous tissue. However no idea of the drainage conditions were obstained.

The earliest known publication is that of *Jackson* 1838 (13). His casereport concerned a man whose humerus underwent complete osteolysis. During the lengthy course of the disease he experienced repeated spontaneous fractures.

In 1955 *Gorham & Stout* described 2 cases of their own and compared the cases from previous publications, a total of 24 cases. They emphasized the haemangiomatosous tissue in the bone in the osteolysis zones. In 1956 (6) *Falkmer & Tilling* described a case of osteolysis in the lower region of the spine and adjacent parts of the pelvis in a man aged 55, where the histological picture showed lymphangioma in the osteolysis zones. No progress was seen in this case. At an X-ray examination in 1966 the condition was unchanged.

After the publications of Gorham and his colleagues (7, 8, 9, 20) more new cases have been published and the total number of cases now known approaches 100.

As we have observed another case of the disease we consider this worthy of publication.

CASE HISTORY

The patient was a girl, born in 1954. There was nothing of interest hereditarily. At the age of 3 the girl sprained her left foot. The foot became swollen but the discomfort was not so great that a visit to the doctor was required. At the age of 5½ the Orthopaedic Department was consulted since the feet were of unequal growth, the left foot smaller, with a tendency of hallux valgus deformity. At the X-ray examination of that time (Figure 1) metatarsal bones 1–4 and the phalanges to no. 2 and 3 toes were attacked by a concentric atrophy. The cancellous structure in the remaining bone was coarse. The girl was examined repeatedly during the course of 6 years. She had several spontaneous fractures (Figure 2) which healed with callus but the atrophy progressed constantly and even the callus formation underwent osteolysis. Figure 3 shows the condition after almost 6 years of observation. The disease also spread to a number of tarsal bones. The left foot is clearly smaller. The calf musculature is moderately atrophied (Figure 4). The 5th metatarsal bone and phalanges of the large toe are fairly well preserved. Discolouring of the skin was not observed. During acute phases with spontaneous fractures there was moderate pain and swelling. The patient manages well with protective arch support

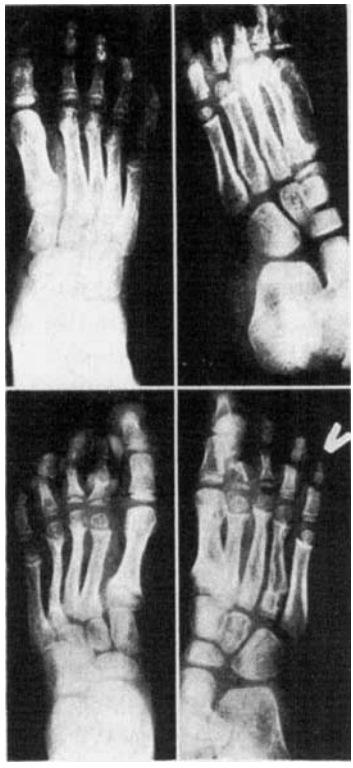

Figure 1. Appearance of unaffected right foot (above) and of left foot at first roentgen examination. 1-4 metatarsals and phalanges in second and third toes show concentric atrophy. The structure of the bone is partly loose. Moderate hallux valgus. Child 5½ years.

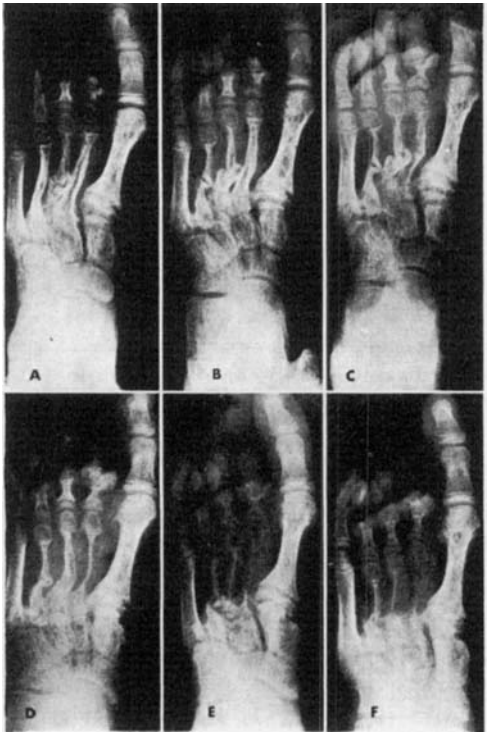

Figure 2. Frontal view of left foot during follow up A) 2 years, B) 3 years, C) 3 years 1 month, D) 3 years 8 months, E) 4 years and F) 4½ years. Slow progress. Spontaneous fratures partly healing with callus formation also affected by progressive osteolysis.

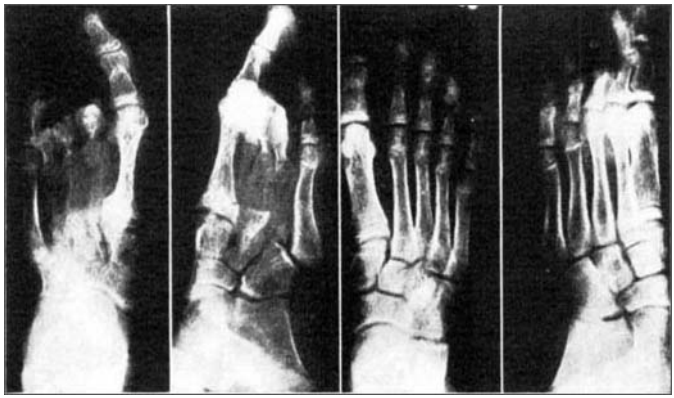

Figure 3. The affected left foot and the unaffected right foot after 6 years observation. Diaphysis of 4th metatarsal resected for biopsy. Disease also affected several tarsal bones. Phalanges of great toe as well as all 5 metatarsals are fairly well preserved.

as the sole therapy and she is able to go to school normally. Radiological therapy has been discussed but in view of the poor results previously reported we have not given such treatment.

The remaining skeletal parts were normal, the lung X-rays normal and also the kidney examination. The SR was normal, as were the blood phosphorus, serum albumin, blood calcium and calcium in the urine. Only the alcaline phosphatases were slightly raised during acute phases of the disease.

After an observation period of four years an operation was carried out exstirpating the diaphysis of the 4th metatarsal bone for the purpose of histological examination. At the operation the residual bone was roughened. The articular cartilage was intact. The tissue adjoining the bone was greyish yellow and no bleeding from this tissue could be ascertained at the operation.

HISTOLOGICAL EXAMINATION

Large sections of the bone tissue were replaced by a loose, partly angioma-like tissue with numerous thin-walled vessels, predominantly constructed like lymph vessels (Figures 5 and 6). In various directions an obvious inflammatory element was present in the form of lymphocyte infiltration and here and there pictures of non-specific granulation tissue were observed in association with partially necrotic bone showing osteoclastic bone resorption (Figure 7). There were no pictures of lymphangiomatous proliferation into surrounding soft tissues.

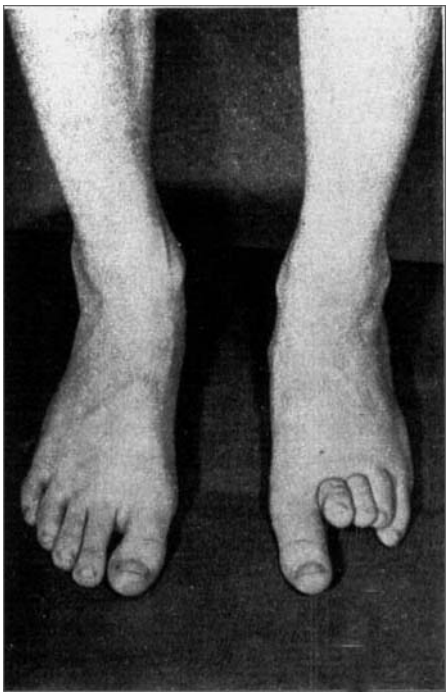

Figure 4. Photograph of both feet after 5 years' observation.

DISCUSSION

In previous published cases the angiomatous tissue demonstrated at the histological examination very often resembled the haemangioma (2, 7, 8, 9, 11, 15, 17, 20, 23). Pictures resembling the lymphangioma were also described (4, 6, 22, 23). Even a mixture of both types was found (10). Only a few authors saw osteoclasts in the histological picture (12, 18). In our present case we discovered osteoclastic resorption.

The case previously reported by *Falkmer* and *Tilling* and the present case offer similarities in the histological picture with lymphangiomatosis. In both cases the operative findings with none or only slight bleed-

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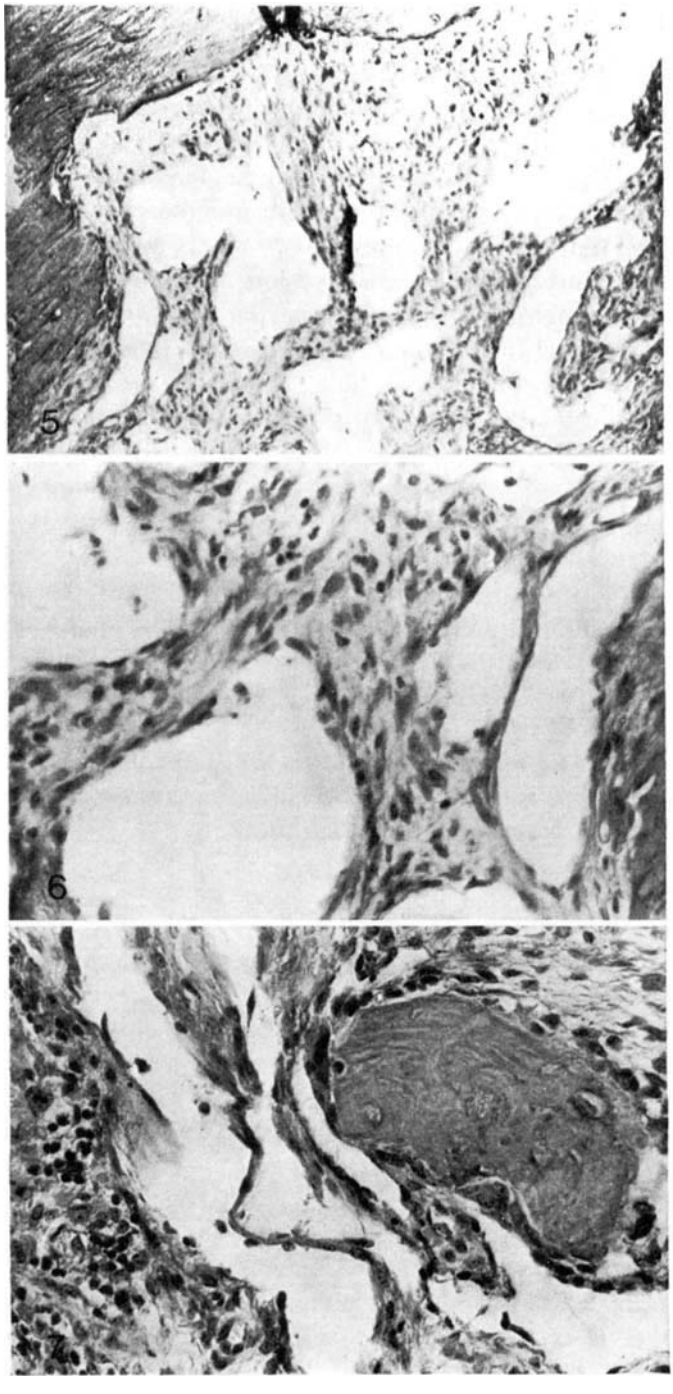
Figure 5. Bony tissue largely substituted by loose, vascular and partly angiomatous tissue with numerous thin-walled vessels of preponderantly lymphatic structure.

Figure 6. The same as in Figure 5 but in higher magnification.

Figure 7. Area with slightly chronic unspecific inflammation adjacent bone destruction with osteoclasts.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ing from the pathological tissue may also support this diagnosis. However, in other reported cases with lympangioma of the bone the surrounding soft tissue was also invaded, which is obviously not so in the present case (14). In discussing the pathogenesis two views may be presented:

A. The lesion may represent a primary angiomatous process, that is, be of neoplastic type. Against this there are the characteristics mentioned above, that the surrounding soft parts are evidently not involved and that the process is not expansive but atrophying. In this respect it may be mentioned that the case reported by *Falkmer & Tilling* (6) which was reported in 1956 and followed-up in 1966, did not show expansion of the process.

B. The lesion may represent an infectious or toxic bone injury with necrosis and resorption of bone tissue and also the development of granulation tissue of a non-specific type with an abundance of vessels. The persistence of this granulation tissue would possibly give its angiomatous character to the histological picture:

The present case, which throughout the observation period showed progress clinically, is extremely interesting owing to the osteoclastic bone resorption demonstrated histologically and to the occurrence of an inflammatory process. The case would thus perhaps offer some support to alternative B above.

Typical of the radiological findings is the continuous progress of the disease over a long period, very often invading adjacent bone but also leaving adjacent bone intact at other times.

SUMMARY

A case is reported of disappearing bone disease in a girl observed for fully 6 years. The osteolytic process affected a number of bones in the left foot. As in previously described cases an angioma-like tissue was found at the site of the affected bone. In addition non-specific granulation tissue and signs of osteoclastic bone resorption were present. The last-mentioned findings are briefly discussed from a pathogenetic viewpoint.

RESUME

Il est rapporté un cas de maladie comportant chez une fille une disparition d'os. Ce cas a été suivi pendant six ans. Le processus ostéolytique touche un certain nombre d'os du pied gauche. Comme dans les

cas décrits antérieurement, un tissu similaire à un angiome a été trouvé à l'endroit de l'os atteint. De plus il y avait aussi un tissu de granulation non spécifique et des signes de résorption ostéoclastique de l'os. Il est discuté brièvement des dernières trouvailles mentionnées en partant d'un point de vue pathogénétique.

ZUSAMMENFASSUNG

Der Fall einer verschwindenden Knochenerkrankung bei einem Mädchen, das für volle 6 Jahre beobachtet wurde, wird berichtet. Der osteolytische Prozess befiel eine Anzahl von Knochen im linken Fuss. Wie in früher beschriebenen Fällen wurde ein angiom-ähnlishes Gewebe am Sitze des erkrankten Knochens gefunden. Ausserdem waren noch unspezifisches Granulationsgewebe und Zeichen von osteoklastischer Knochenresorption vorhanden. Die zuletzt erwähnten Befunde werden von einem pathogenetischen Gesichtspunkt aus kurz besprochen.

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REFERENCES

1. Abell, J. M., Jr. & Badgley, C. E. (1961) Disappearing Bone Disease. J.A.M.A. **177**, 771–773.
2. Aston, J. N. (1958) A Case of Massive Osteolysis of the Femur. *J. Bone Jt Surg.* **40** B, 514–518.
3. Bickel, W. H. & Broders, A. C. (1947) Primary Lymphangioma of the Ileum: Report of a Case. *J. Bone Jt Surg.* **29**, 517–522.
4. Blundell, G., Jones, R. L. & Stewart, G. (1958) Massive osteolysis—Disappearing Bones. *J. Bone Jt Surg.* 494–501.
5. Branco, F. & Horta, J. Da-S. (1958) Notes on a Rare Case of Essential Osteolysis. *J. Bone Jt Surg.* **40** B, 519–527.
6. Falkmer, S. & Tilling, G. (1956) Primary lymphangioma of Bone. *Acta orthop. scand.* **26**, 99–110.
7. Gorham, L. W. & Stout, A. P. (1954) Hemangiomatosis and its relation to massive osteolysis. *Tr. A. Am. Physicians* **67**, 302–307.
8. Gorham, L. W. & Stout, A. P. (1955) Massive osteolysis (acute spontaneous absorbtion of bone, phantom bone, diseappearing bone): its relation to hemangiomatosis. *J. Bone Jt Surg.* **37** A, 985–1004.
9. Gorham. L. W., Wright, A. W., Schultz, H. H. & Maxon, F. C., Jr. (1958) Disappearing bones: rare form of massive osteolysis; report of two cases, one with Autopsy. *Radiology* **71**, 43–47.

10. Halliday, D. R., Dahlin, D. C., Pugh, D. G. & Yong, H. H. (1964) Massive Osteolysis and Angiomatosis. *Radiology* **82**, 637–644.
11. Hambach, R. (1958) Zur Morphologie der massiven Osteolyse. *Zentralblatt für allg. Pathologi und Path. Anatomie* **98**, 298–306, Jena.
12. Iancu, R. & Vargha, A. (1962) Ein Fall von progressiver kryptogenetischer Osteolyse des Unterkiefers. *Radiologia diagnostica. Int. Zeitschrift für das Gebiet der Röntgendiagnostik* **3**, 521–525.
13. Jackson, J. B. S. (1838) A singular case of Absorbtion of Bone (a Boneless Arm). *Editorial Boston M. and S J* **18**, 366–369.
14. Jaffe, H. L. (1959) Tumors and Tumorous Conditions of the Bones and Joints. S 238. Lea and Febiger, Philadelphia.
15. Johnson, P. M. & McClure, J. G. (1958) Observations on massive osteolysis: review of literature and report of case. *Radiology* **71**, 28–42.
16. Jones, G. B., Midgley, R. L. & Smith, G. S. (1958) Massive osteolysis: disappearing bones. *J. Bone Jt Surg.* **40** B, 494–501.
17. Lavrishcheva, G. I. (1960) On spontaneous bone absorbtion. *Ortopedija travmatologija i protezirowanie*, Moskva and Charkov, **21**, 14–18.
18. Milner, S. M. & Baker, S. L. (1958) Disappearing bones. *J. Bone Jt Surg.* **40** B, 502–513.
19. Shopfner, C. E. & Allen, R. P. (1961) Lymphangioma of bone. *Radiology* **76**, 449–453.
20. Stout, A. P. (1959) Massive osteolysis. *Radiology* **73**, 435–436.
21. Thoma, K. H. (1933) A case of Progressive Atrophy of the Facial Bones with complete Atrophy of the Mandible. *J. Bone Jt Surg.* **15**, 494.
22. Tucker, A. S. (1964) Lymphangiectasis, Benign and Malignant. *The American Journal of Roentgenology, Radium Therapy and Nuclear Medicine* **91**, 1104–1113.
23. Van de Weijer (1960) So called essential osteolysis. *Nederlandsch tijdschrift voor Geneeskunde*, 545–547.