OSTEOPATHIA STRIATA—VOORHOEVE'S DISEASE

Report of a Case Presenting the Features of Osteopathia Striata and Osteopetrosis

R. L. HURT, LONDON, ENGLAND

From the Brompton Hospital, London

Osteopathia striata is a rare developmental abnormality characterised by striation of the skeleton, especially the metaphyses of the long bones. It has been seen on only a few occasions since it was first described by Voorhoeve in 1924. The case to be reported showed well marked striation throughout the limbs and so justifies the title of the paper. The skull and ribs, however, show the appearances of osteopetrosis, and it is possible that the case may really be an atypical example of this latter disease.

FIG. 1

On the left is shown the distribution of the changes in the long bones in Voorhoeve's case and on the right the changes throughout the skeleton in the author's case.

CASE REPORT

The patient, a rather unintelligent man of fifty-one years who had previously been healthy, developed three attacks of pneumonia within three months. Empyema complicated

the third attack and a rib resection was performed for drainage. The patient improved thereafter, but a basal shadow persisted in the radiograph and bronchoscopy showed that the

FIG. 2
Right humerus showing longitudinal striation. This was much more marked at the upper (growing) end of the bone.

FIG. 3
Tibiae and fibulae, together with the lower ends of the femora, showing longitudinal striation. This is most marked at the growing ends of the bones.

FIG. 4
Figure 4—Right hand showing striation in all the metacarpals and phalanges. The carpal bones are abnormally dense and contain small translucent areas. Figure 5—Feet showing features similar to those in the hand.

left main bronchus was occluded by a mass of well differentiated squamous-celled carcinoma. Death occurred three months later but no post-mortem examination was made.

The chest radiographs had shown the ribs to be unusually dense and, to elucidate the

cause of this, radiographs were taken of the rest of the skeleton (Figs. 1 to 11). Clinical examination had revealed no abnormality apart from the chest condition. As far as could be ascertained there was no family history of bone disorder or of congenital abnormality. Radiographs of the patient's two children were normal.

FIG. 6

Pelvis showing irregular fan-like striation in the ilium. The upper ends of the femora show a rather irregular broad striation.

FIG. 7

FIG. 8

Figure 7—Skull showing thickening of the cranial vault and the projection of dense bone from the inner table. Figure 8—Skull showing marked thickening at the base and loss of differentiation of the vault into inner and outer tables.

Radiographic features—The main features were longitudinal striation in all the long bones, an irregular fan-like striation in the ilium and a considerable increase in density of the skull and ribs (Fig. 1). *Limbs* (Figs. 2 to 5)—All the long bones showed thin regular striae extending for a considerable distance into the metaphysis from the epiphysial line. This striation was

most marked at the growing ends of the bone. In some of the epiphyses there were a few small irregular opacities. In the carpus there were small translucent areas scattered among abnormally dense bone. *Pelvis* (Fig. 6)—In both ilia there were many elongated dense areas arranged so as to give an irregular fan-like striation. The ischium and pubis were normal. *Skull* (Figs. 7 and 8)—There was gross thickening of the cranial vault, with loss of differentiation into inner and outer tables, and in one or two areas there were projections of dense bone into the cranial cavity. The base of the skull was markedly thickened and the air sinuses, except for the frontals, were obscured. *Ribs* (Fig. 9)—All the ribs were dense and a few striae could be seen near the costochondral junctions. *Vertebrae* (Figs. 10 and 11)—There were scattered dense areas in the vertebral bodies but no differentiation into upper and lower bands as seen typically in osteopetrosis. In the lumbar vertebrae there was a suggestion of vertical striation.

Biochemical investigations—The serum calcium was 10.6 milligrams per cent, the plasma phosphorus 3.5 milligrams per cent, the serum cholesterol 235 milligrams per cent, the acid phosphatase 2.6 King units and the alkaline phosphatase 18.9 King units (two estimations) (normal 3–12 units). Plasma proteins were 4.8 grammes per cent (albumen-globulin ratio 1.2–1.0) (normal 6–8 grammes per cent). The Wassermann and Kahn reactions were negative. Blood examination showed Hb. 86 per cent, red blood corpuscles 5,300,000 per cubic millimetre, and white cells 18,200 per cubic millimetre (polymorphs 87 per cent, lymphocytes 7 per cent, eosinophils 4 per cent, basophils 1 per cent, monocytes 1 per cent). The urine was normal.

FIG. 9

Ribs showing uniform increased density and faint longitudinal striation anteriorly near the costochondral junctions.

Pathology of rib—The segment of rib which was removed was hard, so much so that there was considerable difficulty in cutting it at operation. Histological examination of the rib

showed the typical structure of osteopetrosis (Figs. 12 and 13). The lamellar structure of the bone was lost due to the obliteration of the canaliculi, and the isolated lacunae appeared small and empty.

DISCUSSION

Before the nature of this case is considered it is necessary to discuss briefly the conditions of osteopoikilosis, osteopathia striata and osteopetrosis, together with their relationship to each other.

Osteopoikilosis (sometimes called Albers-Schönberg disease, though this is unfortunate in that this latter name usually refers to osteopetrosis) is characterised by the appearance of dense areas in the cancellous part of most of the bones of the skeleton, especially the long bones in the region of their epiphyses and metaphyses. These dense areas are of two types: 1) nodular lesions which are rounded or oval; and 2) striated lesions, consisting of dense lines running in the long axis of the bone for a considerable distance into the shaft. Both these forms can occur in the same patient and in the same bone.

Osteopathia striata is also characterised by striation of the long bones. The striation is thin

and well defined, lies parallel to the long axis of an otherwise normal bone, and runs for a considerable distance into the shaft. It generally begins at the epiphysial line, but it may begin near the joint surface and cross the epiphysis. Although striation is the most important feature, other changes were noted by Voorhoeve. They were: 1) small areas of translucency in the metaphysis; 2) a localised thinning of the cortex leading to a change in the shape of the metaphysis; and 3) small exostoses. In Voorhoeve's original case the ribs were of normal density and showed faint striation at their costochondral junctions. The skull was normal. *Osteopetrosis* ("marble bones" or Albers-Schönberg disease) has as its main feature a greatly increased radiographic density of one or more bones. The structure of the bone is lost but the shape, except for the development of clubbing at the ends of the long bones, remains unchanged. The density of the bone that is formed during early development is not always

FIG. 10

Cervical vertebrae, the bodies of which contain few dense areas. There is no differentiation into upper and lower bands as seen typically in osteopetrosis.

FIG. 11

Lumbar vertebrae showing irregular dense areas.

FIG. 11

the same, so that transverse bands are produced in the shaft corresponding to the changed density of the bone that is laid down. Longitudinal bands may also be formed, and typical cases of osteopetrosis have been described by McPeak (1936) and by Sear (1927) in which broad bands of dense bone extended from the epiphysis into the shaft. Furthermore, Fairbank (1948) has published a case of osteopetrosis in which the upper end of the femur showed the typical uniform increased density whereas the lower end showed longitudinal striation almost identical with that described by Voorhoeve and with that of the present case.

Previously reported cases—As far as can be ascertained only one true case of osteopathia striata has been described since Voorhoeve's original report in 1924. This case (Fairbank 1925 and 1950) was remarkable in that the changes were predominantly unilateral—in the right arm and leg, right half of the pelvis and right half of the sacrum. The striae were regular

and well defined, but by no means so numerous as in Voorhoeve's original case or in the present case. The skull was normal.

The published cases of osteopoikilosis in which striation has been an important feature have also been examined. Eleven such cases have been found, but only three show changes that are at all comparable with the changes in the limbs in the present case. The cases of Baker and Jones (1941), Jeter and McGehee (1933), Mascherpa (1931), Schéle (1921), and Von Bernuth (1932) are all examples of osteopoikilosis in which the striation is rather irregular and does not resemble that of the present case. The cases of Batten (described by Fairbank 1950) and of Uehlinger (1941) show irregular striation in conjunction with hyperostosis, and again are not really comparable. Nor is the case of Hammer (1948) in which there is a cyst-like expansion of the lower end of the ulna. The details of the remaining three cases are as follows. In Busch's (1937) case there were a few thin striae extending across the epiphysis into the shaft of the lower end of the femur and several nodular lesions in the upper end of the tibia.

FIG. 12

FIG. 13

Transverse (Fig. 12) and longitudinal (Fig. 13) sections of the rib. The lamellar structure of the bone is lost owing to the obliteration of the canaliculi, and the isolated lacunae appear small and empty ($\times 92$).

Lindbom (1942) reported striation of most of the long bones (less regular than in the present case but otherwise similar) together with a few nodular lesions, in a brother and sister aged thirteen and fifteen years respectively. Dermatofibrosis lenticularis disseminata was also present. In Windholz's (1932) case there was fine striation extending from the articular surfaces of the lower end of the femur and upper tibia across the epiphyses into their respective shafts, with many nodular lesions. Dermatofibrosis lenticularis disseminata was also present.

In none of these cases did the changes in the whole skeleton resemble those of the present case—that is, there was no appearance of osteopetrosis. The occurrence of dermatofibrosis in two of these cases is of interest; examples of the nodular type of osteopoikilosis also occurring in association with dermatofibrosis have been reported by Curth (1934) and by Von Bernuth (1932). It will be recalled that fibrosis of the bone marrow is an important change in osteopetrosis.

Author's case—It is difficult to determine whether the case reported here should be classified as a case of osteopathia striata or as an atypical example of osteopetrosis. The striation in

the long bones is identical with that described by Voorhoeve, and the rather irregular fan-like appearance in the ilium, although a little different from that in Voorhoeve's case, is quite comparable. But the dense appearance in the skull and ribs has no counterpart and is typical of osteopetrosis, a fact that is confirmed by the histological examination of the rib. Is it possible that this increased density is due to an exaggeration of the process which caused the striation in the bones of the limbs? If this is so, then the present case may represent a link between osteopathia striata and osteopetrosis, both conditions being due to the same developmental abnormality.

A possible relationship between osteopathia striata and osteopoikilosis—The suggestion that there is a relationship between the two conditions is plausible, for it is easy to postulate that osteopathia striata is a type of osteopoikilosis in which the striated form of lesion predominates to the exclusion of the nodular form. The oval nodular areas invariably lie in the long axis of the bone, and if the stimulus which leads to the formation of these nodular areas were accentuated, typical striation would be produced. Alternatively, if the tendency to produce striation were intermittently overcome, nodular areas would be formed. Voorhoeve, at a time when relatively few cases of osteopoikilosis had been described, was of the opinion that the two conditions were related, and that they were both manifestations of the same abnormal process. Other authors (Baker and Jones 1941, Erbsen 1936, Hirsch 1935, Jeter and McGehee 1933, Mascherpa 1931, Wilcox 1932, Windholz 1932) have held the same opinion, and Lindbom (1942) has published two cases under the title of "Striated Osteopoikile (Voorhoeve)." This view is not accepted by Fairbank (1950), however, who maintains that the two conditions are separate entities. It is true that the nodular areas of osteopoikilosis are prolonged in places into streaks, but these are much shorter and much less regular than the dense lines seen in osteopathia striata.

It will not be possible to give a final answer to the question of the inter-relationship of osteopoikilosis, osteopathia striata and osteopetrosis until the nature of the underlying error of development is known. It is possible that one fundamental error is common to them all. Until such a time, many cases must occur that show the features of what are at present considered to be separate disease processes.

DIFFERENTIAL DIAGNOSIS

The diagnosis of osteopathia striata rests entirely on the radiographic appearances, for the physical examination and biochemical findings are normal. The striation is characteristic and appears in bones that are normal in other respects. Osteopetrosis and osteopoikilosis are likewise entirely radiological diagnoses and their main features have been described. Other conditions to be considered when increased density or striation is a feature are as follows.

Dyschondroplasia (Ollier's disease)—In this condition the columns of unossified cartilage may give an appearance of striation at the ends of the long bones, but the striation is not so regular as in osteopathia striata. Moreover, the masses of cartilage produce a translucent appearance and may also cause broadening of the metaphysis. In the ilium an appearance slightly resembling osteopathia striata may occur, as in cases described by Bentzon (1924) and by Hunter and Wiles (1935). Voorhoeve (1924) considered that osteopathia striata represented a link between osteopoikilosis and dyschondroplasia.

Paget's disease—In Paget's disease new bone formation leads to thickening of the bones, the outlines of which are rather woolly. In the present case there is no suggestion of this, except perhaps in the vault of the skull where there is some thickening of the inner table. Paget's disease, however, usually affects the outer table first and the inner table only in the later stages.

Secondary carcinoma—Especially in the present case, an osteoblastic form of secondary carcinoma must be considered, but the widespread changes and the regularity of the striation make this unlikely. Moreover, the pathological report on the bone is conclusive.

Leri's melorheostosis—In this condition an irregular deposit of dense bone gives the appearance

of having run down the outer surface of the bone (" candle drippings ") causing thickening of the shaft. Usually only one limb is affected.

Fluorine poisoning—In fluorine poisoning the bones are normal in shape, are of increased density and show excrescences at the points where ligaments are attached, especially in the pelvis. There is a history of exposure to fluorine-containing dust, and usually the patient complains of gastro-intestinal symptoms (Møller 1939).

Syphilis—In syphilis there is evidence of a periostitis and the Wassermann and Kahn reactions are usually positive.

SUMMARY

1. A case is reported which shows the typical features of osteopathia striata in all the long bones and probably in the pelvis, with the features of osteopetrosis in the skull and ribs.
2. The occurrence of longitudinal striation in osteopoikilosis and osteopetrosis is described, and the possible relationship between these two diseases and osteopathia striata is discussed.
3. The differential diagnosis is indicated.

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