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Department of Orthopedic Surgery, Kyoto Prefectural University of Medicine, Kyoto, and Department of Orthopedic Surgery, Kyoto Saiseikai Hospital, Kyoto, Japan.

A FAMILY OF MULTIPLE OSTEOCARTILAGENOUS EXOSTOSIS

YASUSUKE HIRASAWA, HIROAKI KITADA, MASUKI KADOYA, KATSUHIKO TAKIZAWA & KOICHIRO SHINAGAWA

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The problem of hereditary multiple exostosis was taken up as early as 1814 by Boyer 1814 & Hagen 1891. Innumerable case reports have appeared since, and the pathology of cartilage-capped exostosis was described in detail by Jaffe (1943).

We encountered a case of exostosis that was observed over three generations of a family. Four members of three different generations of this family were involved in this disease. We performed an operation to remove exostoses from two of these four patients. We will report on the clinical study and discuss our observations.

CASE REPORTS

Case 1: A 51-year-old male visited our clinic with a hard and large tumor above the left knee (Figure 1). He had first noticed the mass at the age of 10, and this grew in size progressively up to the age of puberty.

A physical examination showed a poorly developed man with a mass measuring ca. $12 \times 10 \times 8$ cm at the antero-medial aspect of the left distal femur (Figure 3). The mass felt hard like a bone with an irregular surface. No tenderness was ever noted over the tumor. The range of motion of the knees was not limited.

Routine laboratory examinations, including serum calcium, phosphorus & alkaline phosphatase were within normal limits. Roentgenological examinations revealed a large pedunculated tumor with a cauliflower-like appearance at the left distal femur, which was blotchy

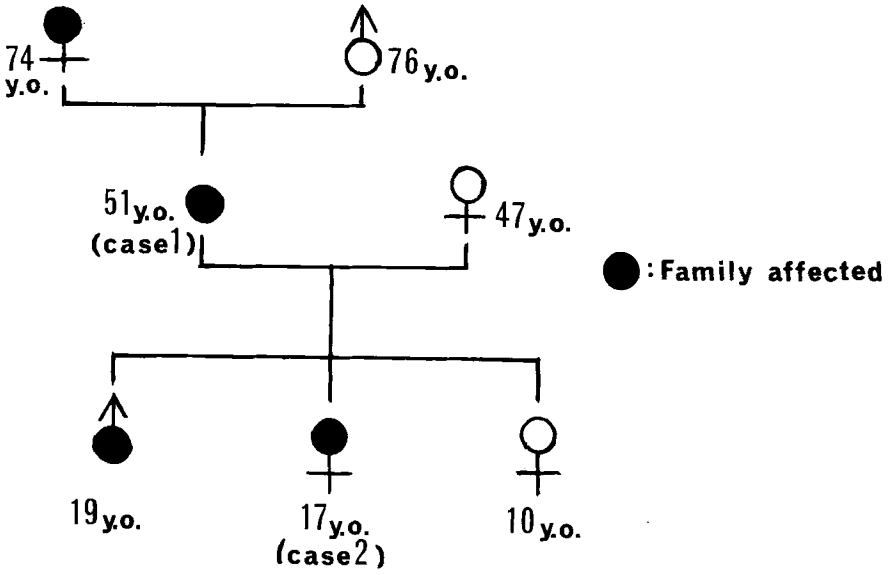
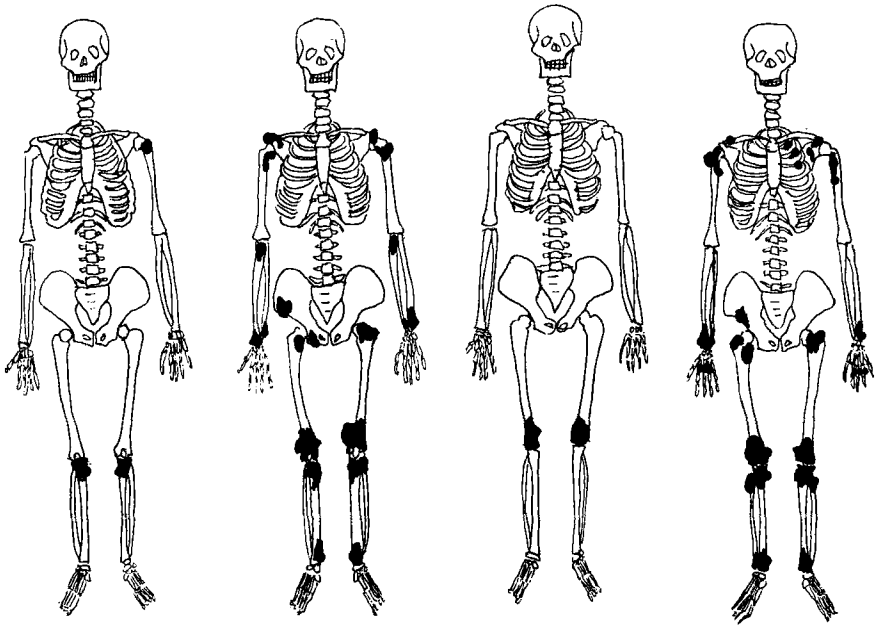


Figure 1. The family involved.



Grandmother *Father (case 1)* *Case 2.* *Elder brother*

Figure 2. The sites of bone lesions.

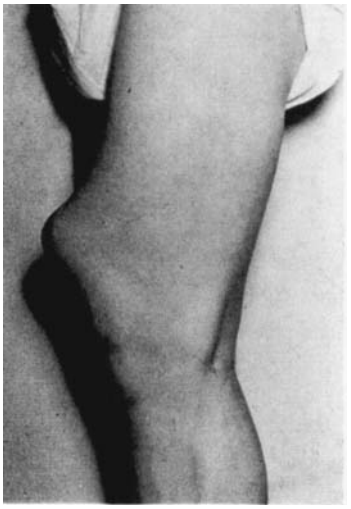


Figure 3. Case 1. Tumor in the left knee.

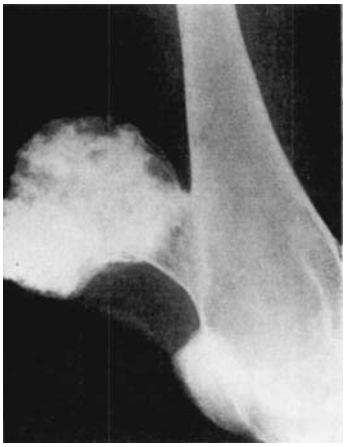


Figure 4. Case 1. Roentgenogram of the knee.

Figure 5. Case 1. Cut surface of the tumor.

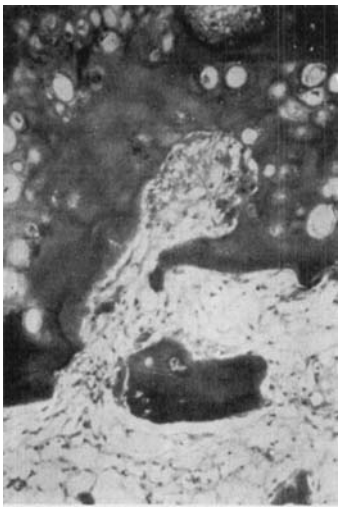


Figure 6. Case 1. The photomicrograph of the tumor tissue. The trabecular formation of the bone was evident in irregular fashion at the deep portion of the tumor. (Haematoxylin-Eosin staining. × 140).

radiopaque (Figure 4). Numerous exostotic outgrowths were also observed elsewhere (see Figure 2).

The cartilagenous tumor that was removed was 9 × 8 × 6 cm (Figure 5). The knobby surface of the tumor, which partly adhered to the connective tissue membrane, was composed of yellowish cartilages the size of rice grains, which were capped in some places and osseous in others. The periosteum covering the surface of the tumor continued to the periosteum over the unaffected portion of the bone. The cartilage was hyaline and also partly calcified. The peripheral part of the tumor was composed of osseous tissues. Most of the interior of the lesion consisted of loose-meshed spongy bone. The trabecular forma-

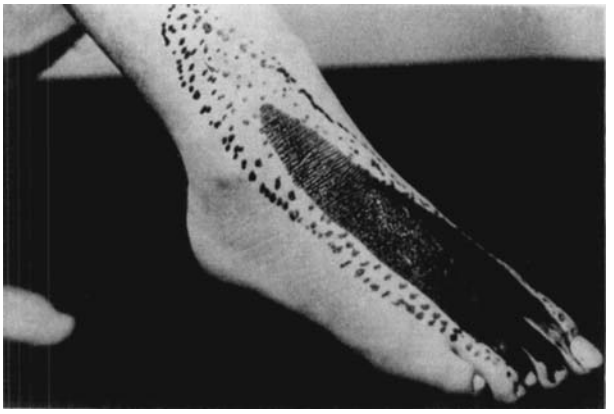


Figure 7. Case 2. Preoperative finding (right foot), showing drop foot with abnormal sensitivity. Anaesthetic area and hypoesthetic area.

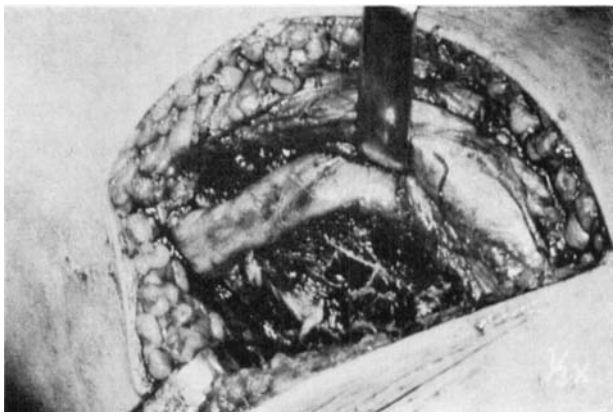


Figure 8. Case 2. Finding at operation, showing edematous peroneal nerve which had been compressed by the tumor.

Figure 9. Case 2. Dorsiflexion at the 5th month postoperatively.

tion of the bone was evidently irregular at the deep portion of the tumor. The greyish region at the central part of the tumor corresponded to the fatty bone marrow (Figure 6). There were no malignant findings.

The postoperative course was uneventful and the patient was discharged from the hospital after two weeks.

Case 2: A 17-year-old female visited our hospital with difficulty in walking due to the lack of right ankle dorsiflexion and numbness over the dorsal aspect of the right foot for several days. She was the second child of the patient of Case 1.

A physical examination showed a normally nourished and developed female with complete loss of dorsiflexion of the right foot and toes, and hypo- and anesthesia over the dorsal foot (Figure 7).

An electromyographic examination revealed fibrillation voltage at rest and no voluntary contraction in the right tibialis anterior and extensor digiti communis muscles.

Routine laboratory examination results were all within normal limits except for slight anemia. Roentgenological examination of the fibular head region showed osseous protuberance, that is, flat, broad-based exostosis.

Surgical excision of the tumor was performed. The peroneal nerve was exposed, which appeared markedly edematous with violet discoloration. The nerve was compressed by the tumor and was nicked at the top of the tumor, which developed from the fibular head in an irregular fashion (Figure 8).

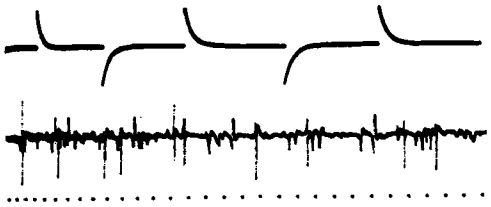


Figure 10. Case 2. Electromyographic finding at the 5th month postoperatively. Callibration: 1000 μV. Time: 10 msec.

A histological examination revealed the exostotic tumor, which was covered with thin connective tissue membrane and had a cap of hyaline cartilage. There was no obvious calcification. The interior of the tumor contained cancellous bone enclosing a small amount of the fatty bony marrow.

The 5th month after the operation, the patient showed markedly improved range of motion of the ankle (90°–130°) and of the toes (Figure 9). A manual muscle test showed 4 to 5 in the tibialis anterior and the extensor digiti communis muscles.

An electromyographic examination of the extensors of the ankle and the toes revealed silent at rest and almost normal N. M. U. voltage at voluntary contraction of the ankle and the toes (Figure 10).

At the latest follow-up, 15 months postoperatively, the patient had full range of motion of the ankle and toes, and was able to run without difficulty.

DISCUSSION

Multiple exostosis is a heritable disorder of the skeletal system and has a striking manifestation of numerous cartilage-capped exostoses which appear in different portions of the skeleton. They are usually bilateral and symmetrical and are most often encountered on the long tubular bones. The most common locations are the lower metaphysis of the femur and the upper metaphysis of the tibia. In our cases, they were found not only at the ends of the tubular bones, but also at the scapulae, the iliac crests and the ribs.

According to Japanese national statistics of registered bone tumor patients, there were 5634 patients suffering from primary bone tumor during the years from 1964 to 1969, of whom 492 were declared to be osteocartilagenous exostosis patients. The frequency of this occurrence is about 8.7 per cent. Incidentally, solitary exostosis patients numbered 1058, whose occurring rate is approximately 18.6 per cent.

The ratio of males to females affected by this disease is 282 to 210, according to the above-mentioned statistics. According to Jaffe (1965), the ratio of affected males to females is 7 to 3. Solomon reported that both males and females were affected with equal frequency.

Although over 80 per cent of the patients are detected within the first decade of life, most of them come under observation between 11 and 20 years of age.

According to the above Japanese statistics, 470 cases of osteocartilagenous tumor out of 492 (95.5 per cent) suffer exostosis all over the body. In such cases it is often accompanied by the undergrowth of the bone and occurs from generation to generation of a family. Therefore, this matter will also be discussed from a genetic viewpoint.

According to Toyama & Horikoshi (1936), 16 out of 24 families are a dominant hereditary type. The inheritance rate of the offspring is 43.9 per cent and the expectancy rate is nearly 50 per cent. Tateiwa (1952) states that the disease appears in 11 families out of 29 and he distinctly admits that it tends to be inherited dominantly and cannot simply be explained by Mendelismus.

From the statistical analysis of 1124 recorded cases, Stocks & Barrington (1925) concluded that the disease was inherited by 64 per cent usually from an affected parent.

The 74-year-old grandmother of the patient of case 1 has a tumor on her left shoulder, which has never been removed. The patient of case 1 himself did not pass a physical examination because of a big

tumor in his left thigh when he tried to enlist in the army at the age of 20. Since his marriage twenty years ago, he had concealed this fact from his family as well as his children. His children, now adolescents, had wondered about their father's attitude which had caused serious family trouble. Recently the patient finally made up his mind to visit our clinic in the hope that the tumor would be removed.

Case 2 is the second child of the patient in case 1. She was uneasy about the existence of small tumors in both her knees, but she did not admit to their existence until she visited our clinic for the first time because of a sudden paralysis in her left foot.

There has been no valid explanation of the pathogenesis or the mechanism of growth of this osteocartilagenous exostosis. The hereditary nature of the disease is one of the most believable factors.

Virchow (1891) held that bits of cartilage become snared off from the lateral margins of the epiphyseal cartilage plates and, moving on to the body of the bone, they constitute the beginnings of the lesions. Müller (1913) concluded that cartilage-covered exostoses on the bones have their beginnings in small nests of cartilage formed by the osteogenic layer of the periosteum. Keith (1920) reported that the primary basis of the pathogenesis of multiple exostosis is defectiveness of the periosteal ring.

Jaffe (1965) states that the theories of defective modeling and of perverted periosteal and perichondral activity are not only mutually compatible, but reinforce each other in explaining the pathogenesis of the disorder as a whole.

Characteristic of the exostosis is that the normal cancellous bone covered by cartilage cap is encapsulated by the thin connective tissue membrane and that the tumor grows by the enchondral ossification at the deep portion of the cartilage. With the closure of the epiphyseal plate, the cartilage cap is replaced by the osseous plate and stops its growth.

In our cases, case 2 revealed more proliferated cartilagenous tissue in the tumor than did case 1, which showed scattered cartilage tissue with moderate calcification.

Jaffe (1965) reported chondrosarcoma as a complication with incidence nearer 20 than 11 per cent. In case 1, the tumor had been stationary in size for more than 30 years and showed no malignant tendency.

Solomon (1963) finds it surprising how seldom the exostoses interfere seriously with adjacent structures, despite their large number.

Instestinal obstruction, arterial aneurysm, or pressure on a peripheral nerve may call for removal of the exostosis.

In case 2, the exostosis that originated in the fibular head compressed the common peroneal nerve, which resulted in the loss of dorsiflexion of the ankle and the toes as well as anesthesia and hypoesthesia of the dorsal foot.

The compressed symptoms were completely relieved by the removal of the exostosis.

SUMMARY

We encountered a case of a multiple osteocartilagenous exostosis that was observed over three generations of a family. Four members of three different generations of this family were involved in this disease. Case 1 showed a huge tumor in the left thigh, and case 2 revealed paralysis of the right peroneal nerve.

In both these cases the symptoms were completely relieved after the removal of the exostosis.

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Correspondence to:

Yasusuke Hirasawa, M.D.
c/o Prof. C. B. Sledge, Dept. of Orthopedic Surgery, Harvard Medical School
Robert B. Brigham Hospital, Parker Hill Avenue
Boston, Mass. 02120, U.S.A.