Severe Multiple Cartilaginous Exostoses
—A Case Report—

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Summary: A severe case of multiple cartilaginous exostoses (MCE) was presented. The patient, a 10-year-old girl was the affected member of a family with paternal MCE transmitted over at least 3 generations. Her growth disturbance appeared to have resulted from the severe exostoses of the long bones.

Key words: Hereditary multiple exostoses—multiple exostoses—diaphyseal aclasis—short-limbed dwarfism—secondary dwarfism

Introduction

Multiple cartilaginous exostoses (MCE) is one of the most common inherited skeletal disorders. A collection of 1124 cases was reported by Stock and Barrington (1925). Recently other large series have been published by Solomon (1963) and Sauer (1979). Different terms have been used for it such as multiple exostoses, hereditary multiple exostoses and diaphyseal aclasis. The swelling or "lump" are reported commonly to be located at the end of the long bones, in the scapula, pelvis, ribs, and rarely in the spine and skull. Growth disturbance of the bone is generally recognized but rarely results in true dwarfism. Commonly, there is little disturbance in health or activities in daily life.

Case Report

In the patient's family, the father, a paternal uncle, and the paternal grandfather have MCE. The father presented exostoses in the region close to the knee when about 10 years of age and since then there has been almost no change in the growth of the exostoses (Fig. 1). The uncle and the grandfather both had a similar

Fig. 1. Roentgenogram of her father showing MCE on the tibias.
history to that of the father, with eventual height about 155 cm. The mother and all the other maternal relatives were unaffected. The patient was delivered by Cesarean section after a normal, uncomplicated pregnancy. The birth weight was 3,820 gm, and body length was 50 cm. The circumference of the head and chest were normal.

A swelling in the PIP joint of the right middle finger was noted a few months after birth. Then appeared on the middle fingers, wrists, knees and scapulae one after another at about one year of age. She was first seen at our Orthopaedic Department at 3 years of age and diagnosed as having MCE and dwarfism. She was admitted complaining of deformity and functional disturbance of the right wrist at 4 years of age. On admission, short-limbed dwarfism was noted; body length 85 cm (-3.5 SD) with the arm span 74 cm; weight 12 kg (-2 SD), increased lordosis, and exostoses mainly on the wrists and in the regions close to the joints, on the scapulae, middle fingers and ribs. Limitation of movement due to the exostosis was noted at the wrists.

The X-ray findings revealed multiple cartilaginous exostoses located in the metaphysis of the long bones including those in the hands and feet, and on the scapulae, ribs and spine. Widening of the proximal humerus with a large exostosis was present. The lengths of the radius and ulna were bilaterally short, and there were large exostoses at the distal ends of the radii and ulnae (Fig. 2).

The blood chemistry, urinalysis, and other laboratory findings were all within normal ranges. Chromosome analysis using high resolution G-banding techniques re-

Fig. 2. Roentgenogram of the upper extremities at 4 years. There are MCE on both the proximal humeri, and distal radii and ulnae, and radio-ulnar synostosis on the right side.

Fig. 3. Photograph at 10 years. There are MCE at the end of the long bones and ribs. Dwarfism with shortening of the limbs, particularly of the forearms.
revealed normal karyotype. The resections of the tumors from the wrists and from the right scapula were performed, and pathological diagnosis confirmed cartilaginous exostosis.

At the 10 years old, she was 106 cm high (-4.5 SD) with the arm span 85 cm, a very small stature and the exostoses seriously developed (Fig. 3).

Shortness of the arm span was concluded to be due to the growth disturbance in the long bones and to the functional disturbance in the elbows. The activities of daily living was markedly impaired caused by limitation of the range of motion of the large joint and fingers; the flexion and extension of the elbows were from 70 to -35 degrees in the right and 105 to -25 degrees in the left, supination of the forearms was 0 degrees bilaterally, and in the hip joints, flexion was 50 degrees and internal rotation was 0 degrees bilaterally.

Roentgenogram at 10 years of age showed there was severe involvement of the upper extremities. There were developed exostoses at the proximal humeri, and marked shortening and deformities of the ulnae and radii, with the head showing subluxation. In the hand, there were reduced lengths in the metacarpi and phalanges (Fig. 4). The exostoses of the femoral neck was considerably enlarged, and the exostosis of the iliac bone was observed. The femoral heads were flat and showed lateral displacement. There was marked shortening of the tibias and fibulas (Fig. 5).

The lengths of the extremities were measured to be 29.5 cm in the right, and

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**Fig. 4.** Roentgenogram of the upper extremities at 10 years. There are developed exostoses at the proximal humeri. Note the marked shortening and deformities of the ulnae and radii, with the head showing subluxation.

**Fig. 5.** Roentgenogram of the lower extremities. The left iliac exostosis are seen. The femoral heads are small and flattened, and show lateral displacement. There are tibio-fibular synostosis and marked shortening of the tibias and fibulas.
TABLE 1

Values of the longitudinal bone from the roentgenogram (cm)

<table>
<thead>
<tr>
<th>Age</th>
<th>Side</th>
<th>4 years</th>
<th>10 years</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Right</td>
<td></td>
<td>Left</td>
</tr>
<tr>
<td>Humerus</td>
<td>13.5</td>
<td>13.2</td>
<td>17.0</td>
</tr>
<tr>
<td>Radius</td>
<td>7.1</td>
<td>7.0</td>
<td>8.2</td>
</tr>
<tr>
<td>Ulna</td>
<td>7.8</td>
<td>8.2</td>
<td>9.0</td>
</tr>
<tr>
<td>Femur</td>
<td>19.5</td>
<td>20.8</td>
<td>26.2</td>
</tr>
<tr>
<td>Tibia</td>
<td>14.0</td>
<td>14.5</td>
<td>19.5</td>
</tr>
<tr>
<td>Fibula</td>
<td>11.8</td>
<td>12.6</td>
<td>16.7</td>
</tr>
</tbody>
</table>

29 cm in the left upper extremity, and 51 cm in the right, and 52 cm in the left lower extremity. However, we could not determine the lengths of the upper arms and forearms, the thighs and legs. This was due to obscuring effect of the exostoses in the elbows and knees being so enlarged that the condyles were not demarcated. We estimated these measurements from the bone lengths in the roentgenograms. Measurement were thereby taken from the maximal longitudinal lengths of the bone. The growth disturbance in the upper extremities was more severe than that in the lower extremities, and the values of the radii and ulnae at 10 years of age had changed only slightly in dimensions from 4 years of age (Table 1).

Discussion

It is already established that multiple cartilaginous exostoses (MCE) follows by autosomal dominant transmission. The ratio of occurence of MCE in males, to that in females is about equal and 63% of all cases had an affected parent. There had been no tendency for the lesion to increase in size in successive generations, according to Solommon (1963). In our case, however, her grandfather, paternal uncle, and father all had only mild or moderate MCE, and their height was about 155 cm. The mean height of MCE patients in the literature was 160 cm (Wynne-Davies et al. 1985), so they were not so short and they were able to live a normal family life. Our case is very different from usual cases. The disorder is rarely noted before the third year of life. Her first appearance of a bony lump was in the finger a few months after birth, and by 3 years of age she had already demonstrated dwarfism. Usually, this disorder does not result in true dwarfism. However, this case showed true dwarfism like as achondroplasia, severely with short limbs, and severe involvement of the hands and feet. Short stature was not noted at the time of the birth, though it became increasingly developed with age. The mean height of MCE girls of 10 years in the literature was 122 cm and this is equivalent to -2.3 SD in the growth chart for a Japanese girl of 10 years.

Her height was 106 cm (-4.5 SD) at 10 years of age and her arm span was only 86 cm. Such a marked discrepancy from mean values in our case is a result of severe growth disturbance in the long bones and loss of extension at the elbows.

Sauer et al. (1979) clinically examined 19 patients with exostoses to measure dimensions of the thigh, leg, upper arm and forearm. The results showed a statistically significant reduction in length of the extremities in adults aged 14 to 38 years with MCE. In our case, it seems that the growth disturbance resulted only from the very severe exostoses, especially in mesomelic part of the extremities, and was not resulting from the other symptoms that contribute to short stature.

References

Philadelphia: Lippincott. 


