DYSPLASIA EPIPHYSIALIS MULTIPLEX IN THREE SISTERS

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Dysplasia epiphysialis multiplex, first described by Fairbank (1947), is characterised by dwarfism, stubby digits and mottling or irregularity in density and outline of the developing epiphyses. The title was introduced in order to bring into one group a number of cases which had previously been recorded under a variety of names. Fairbank's paper was based on a study of twenty patients. The purpose of this communication is to add a further three cases occurring in one generation. This in itself is hardly sufficient reason for publication, but a study of these three sisters enables comparison of the changes found at different ages to be made, and the recording of some unusual and atypical changes.

The condition is said not to be familial, but in Fairbank's series are included two sisters; also quoted are two brothers (cases of Mr J. A. Cholmeley). Other cases occurring in one family were described under different titles but were considered by Fairbank to fall into his group—two sisters (Gardiner-Hill 1937), brother and sister (Barrington-Ward 1912) and a mother and twin boys (Resnick 1943).

The sisters about to be described are from a family of eight children. The father, who is seventy-five years old and five feet three inches high, suffers from chronic arthritis of the rheumatoid type, but there is no radiological evidence of epiphysial dysplasia. The mother is five feet two inches high and also shows no evidence of the condition. The age and height of the present generation of eight children are shown in Table I.

<table>
<thead>
<tr>
<th>Sibling</th>
<th>Age</th>
<th>Height</th>
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<tbody>
<tr>
<td>Theresa</td>
<td>4½ years</td>
<td>3 feet 5 inches</td>
</tr>
<tr>
<td>Anne</td>
<td>9 years</td>
<td>4 feet 5½ inches</td>
</tr>
<tr>
<td>Keith</td>
<td>12 years</td>
<td>5 feet 2 inches</td>
</tr>
<tr>
<td>Malcolm</td>
<td>14 years</td>
<td>5 feet 4 inches</td>
</tr>
<tr>
<td>Sheila</td>
<td>15 years</td>
<td>4 feet 7 inches</td>
</tr>
<tr>
<td>Robert</td>
<td>16½ years</td>
<td>5 feet 9 inches</td>
</tr>
<tr>
<td>Mary</td>
<td>18 years</td>
<td>5 feet 4 inches</td>
</tr>
<tr>
<td>Roberta</td>
<td>19 years</td>
<td>5 feet 0 inches</td>
</tr>
</tbody>
</table>

It will be seen that the affected sisters are short compared with the other members of their family. This shortness does not amount to dwarfism, but is sufficiently obvious to enable them to be picked out from their brothers and sisters (Fig. 1). The other obvious features which distinguish them are the short stubby hands and feet (Figs. 2 and 3) and the fact that they all have cleft palates. It is difficult to relate the last finding to their general condition and it is possibly only an interesting coincidence.
Group of four sisters. Left to right: Anne (nine years) a normal sister for comparison, Sheila (fifteen years), Roberta (nineteen years) and Theresa (four and a half years).

Figure 2—Sheila’s hands showing the characteristic short and stubby fingers. Figure 3—Sheila’s legs. The genu valgum was later corrected by a supracondylar osteotomy of the right femur.
Of the three, Sheila has the severest deformities, and she was brought to hospital because of right genu valgum (Fig. 3). This was satisfactorily corrected by a supracondylar osteotomy of the femur in 1949. When Sheila was under observation the opportunity was taken to investigate the other members of the family.

**RADIOGRAPHIC FINDINGS**

While epiphysial changes characteristic of the condition are found to a varying degree in all three sisters, there is one striking feature which differs from the description given by Fairbank. He states that "the centres of ossification for the epiphyses may be late in appearing and backward in development and fusion with the shaft may be delayed." In our cases the epiphyses have appeared early and fused early, and a study of the bone detail in the radiographs shows a remarkable degree of maturity for their age. For example, the radiograph of Theresa's hand at four and a half years (Fig. 8) shows a bone age of at least seven years, as indicated by the appearances of the carpal centres. And the radiographs of Sheila's hips at the age of fourteen years (Fig. 5) show fusion of the capital and trochanteric epiphyses, which normally does not occur until the age of eighteen years. This suggests that the relative shortness of these girls may be due to early epiphysial fusion rather than delay in development.

Fairbank, however, emphasised that "the principal change is irregularity in ossification," and these three sisters do show almost all the typical features in this respect.

**Hips**—In Theresa's case the capital epiphyses are flattened, narrow and irregularly developed (Fig. 4). The neck of the femur is widened and there is coxa vara. The general bony development in advance of her age (four and a half years) can be noted. Sheila's hips (Fig. 5) show a similar abnormality of the femoral head occurring at a later period of growth. Roberta shows the changes to a lesser degree (Fig. 6). The short neck and the beaking of the greater trochanter are seen. **Knees**—Sheila alone has an abnormality of the lower femoral epiphysis causing the knock-knee for which she was treated. The patella on this side is absent. Abnormality in the growth of this bone is also found in the other two sisters. In Figure 7 (Theresa) the precocious and irregular development of the patellar epiphysis can be seen.

**Ankles**—The characteristic obliquity of the joint line is not present, but a flattening of the superior articular surface of the talus in the lateral view is seen in all three sisters. **Feet**—The abnormalities of the tarsus and the stunted metatarsals are well shown in Figure 8. **Hands**—The metacarpals are broad and short, especially in Sheila's left hand (Fig. 11). The fourth metacarpal is stunted; it is interesting to note that this finger is not relatively short owing to an increase in length in the proximal phalanx. In Theresa's hand (Fig. 9) the developing epiphyses of the proximal phalanges are narrow and dense. Roberta's hand (Fig. 10) shows less obvious changes than her sisters', but there is a relative overgrowth of the lower end of the ulna. **Elbows**—Sheila's elbows (Fig. 12) show the unusual shape of the lower end of the humerus and head of the radius. In Theresa's elbows at the age of four and a half years the epiphysis of the capitulum, which normally appears at two years, is not yet present. This suggests that the forward subluxation of the radial head is due to lack of development of the capitulum in Sheila's case as well. It may be noted that it is only around the elbow that epiphysial development is delayed; elsewhere it is premature. **Shoulders**—The distinctive abnormality in the shape of the head of the humerus is not present in any of the sisters.

Apart from some flattening of the lumbar vertebral bodies in Sheila, a full radiographic survey of the three sisters showed no abnormalities other than those described.

**DISCUSSION**

These three sisters show changes in the epiphyses of the long bones which are sufficiently typical to allow a diagnosis of dysplasia epiphysialis multiplex to be made. It is, however, important to note that there is some variation from Fairbank's described pattern in that the
Figure 4—Theresa’s left hip. The capital epiphysis is flattened, narrow and irregularly developed. The neck is widened and there is coxa vara. Note the maturity of bony development. Figure 5—Sheila’s left hip showing an abnormality like Theresa’s, but at a later period of growth. Figure 6—Roberta’s left hip which shows some flattening of the femoral head, but the changes are less marked than in the younger sisters.

Figure 7—Theresa’s left knee, showing the thin and irregularly developed patella. Figure 8—Sheila’s right foot, showing the short, stunted metatarsals. The broadness of the first metatarsal and proximal phalanx is striking.
Figure 9—Theresa’s right hand. The typical shape of the metacarpals is shown. The epiphyses of the proximal phalanges are narrowed and dense. The development of the osseous centres for the carpus is in advance of her age (four and a half years). Figure 10—Roberta’s left hand. The stunted metacarpals are typical, but the changes are not so advanced as in the younger sisters. There is overgrowth of the lower end of the ulna.

Figure 11—Sheila’s left hand. The stunting of the metacarpals is present, the changes being most obvious in the fourth. The middle phalanges are short and broad. Figure 12—Sheila’s right elbow, showing the abnormal shape of the capitulum and the consequent subluxation of the radial head.
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epiphyses have appeared early and fused early. This is a substantial difference, but since it is impossible to make sharp distinctions in classifications of growth abnormalities it seems correct to include them under this title.

A point of practical importance in consideration of this condition is the suggestion that the changes in the femoral heads may be misinterpreted as pseudocoxalgia. Although there may be radiological similarities at certain stages of the disease, confusion should not arise on clinical grounds. In no case of dysplasia epiphysialis multiplex have there been noted any symptoms referable to these changes. This is confirmed by the three sisters who have never had pain in the hips or limp and who, at their various ages, have full hip movements. It may also be reasonably deduced that in these hips there is a tendency towards improvement without treatment. It is, of course, possible that the sisters may have been affected in varying degrees from the onset, but on the other hand a study of the radiographs suggests that they show serial changes which are most obvious during the period of active growth. In any event, it is submitted that treatment, in the form of relief from weight-bearing, is unnecessary and should not be contemplated because of the radiographic appearances. Moreover, if dysplasia epiphysialis multiplex is diagnosed early in life there should be no need to give the parents a dismal prognosis, for dwarfism is seldom severe, deformity has been troublesome only in a small proportion of the reported cases, and function is invariably good.

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REFERENCES


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