DOUBLE PATELLAE IN MULTIPLE EPIPHYSIAL DYSPLASIA

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This paper describes the association of double patellae of an unusual type with multiple epiphysial dysplasia in three siblings. The patellae are of the type first described by Büttner (1925) and Haenisch (1925). Other examples have been described by Paas (1931), Schwarz (1932), Weber (1935, cited by Lodes 1949), Gorzawski (1937), Marquardt (1938) and Lodes (1949). The abnormality common to these patellae is that they possess a larger anterior part that embraces a smaller posterior part. In addition, habitual dislocation may be present, and one or both of the component layers may be multipartite. For convenience I shall refer to such patellae as double-layer patellae.

Multiple epiphysial dysplasia was first described as a clinical entity by Fairbank (1947) who reported his findings in twenty cases. Since then many examples have been recorded (Fairbank 1951; Scott 1952; Watt 1952; Waugh 1952; Jackson, Hanelin and Albright 1954; Maudsley 1955; Shephard 1956; Levy, Mazumdar and Morales 1957; and Barrie, Carter and Sutcliffe 1958) but in none were double-layer patellae found. The only abnormalities of the patella that have been noted are mottling of the patellae (Fairbank 1947), unilateral absence of the patella (Waugh 1952), thin and irregular patellae (Waugh 1952), and in one case a dense central nucleus enclosed by less dense bone (Shephard 1956).

The cases of double-layer patellae previously described, twelve in all, were found to have other skeletal abnormalities. The hips were invariably affected and the hands, feet, elbows and shoulders were often involved. The disorder was clearly familial in Lodes’s (1949) four patients. Lodes reviewed the literature and considered that all the cases could be classified as atypical chondrodystrophy (Morquio’s disease): atypical in that none of them showed the characteristic vertebral flattening. He considered the patellar changes to be an integral part of the dystrophy. However, it is surely significant that double-layer patellae have never been described in unequivocal cases of Morquio’s disease (that is, in patients showing the typical vertebral changes). I therefore regard Lodes’s (1949) conclusion that the twelve patients with double-layer patellae were examples of Morquio’s disease as doubtful. In my opinion they were more likely to have been examples of multiple epiphysial dysplasia. The published descriptions and radiographic findings are certainly compatible with this diagnosis, and they often show a striking resemblance to my own cases.

CASE REPORTS

Three of a family of eight surviving siblings were found to be dwarfed and to show multiple skeletal abnormalities. The father was short (four feet eleven and a half inches (150-5 centimetres)), though he claimed to have been five feet two inches (158 centimetres) when younger) and was the shortest of five siblings. Although his radiographs showed marked osteoarthritic changes they provided no clear evidence of skeletal dystrophy. The mother was not seen but she was said to be of normal build and about five feet two inches (158 centimetres) in height. There was no consanguinity between the two parents.

Case 1—A man of fifty-one whose height was five feet one inch (155-5 centimetres). He had no children.

Case 2—A woman aged fifty-four. She was four feet three and a half inches. Her daughter was five feet seven inches and clinically and radiographically normal. Her granddaughter was normal.

Case 3—A woman aged fifty-nine whose height was four feet seven inches. Her son was five feet eight and a half inches and of normal build.
FIG. 3
Lateral views of the knees, showing the abnormal patellae in Cases 1, 2 and 3.
All three patients were intelligent and had led normal lives until recent years, when all developed joint pain and stiffness, particularly of the hips and knees. They showed a strong family resemblance and were of similar build, with heads of normal shape which were in good proportion to their body size, stubby hands and feet with prominence of all joints, and remarkably short forearms. All had restriction of movements of hips, knees and elbows and some valgus deformity of the knees. There were no abnormalities outside the skeletal system. Laboratory investigations, including serum calcium, phosphorus and alkaline phosphatase, were normal.

**Radiographic findings**—The radiographic findings were so similar in the three cases that they can be considered together. The hands and forearms showed stunted bones with widened
and somewhat irregular ends (Fig. 4). The radii and ulnae were particularly short and the radial head showed a mushroom-shaped deformity. The shoulders showed large humeral heads and secondary degenerative changes. The hips showed deformed acetabula and very large, flattened femoral heads (Fig. 7); there was again considerable degenerative change. The knees showed valgus deformity (Figs. 1 to 3). The patellae were not dislocated but in each case were of double-layer type. In addition they were invariably multipartite. The left medial semilunar cartilage was calcified in Case 2. The ankles showed the slanting joint space characteristic of multiple epiphysial dysplasia (Fig. 5) (Fairbank 1951, Barrie et al. 1958). The feet showed changes very similar to those in the hands (Fig. 6). The skull and chest were radiographically normal.

**SUMMARY AND CONCLUSIONS**

1. The occurrence of bilateral double-layer patellae in association with multiple epiphysial dysplasia is described in three siblings.

2. Twelve cases of bilateral double-layer patellae have been reported previously, and in all there was an accompanying skeletal dystrophy which, although not diagnosed as such, is likely to have been multiple epiphysial dysplasia.

3. It is suggested that although they occur only in a minority of cases, double-layer patellae when present are a feature of considerable diagnostic value in multiple epiphysial dysplasia. Lateral radiography of the knees may therefore be of assistance in the diagnosis of multiple epiphysial dysplasia.

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**REFERENCES**


