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FAMILIAL OSTEOCHONDRITIS DISSECANS AND DWARFISM

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We review four generations of a short statured family in whom 12 members have osteochondritis dissecans (O.D.) affecting the knees, elbow, or both, and other minor anomalies of bony development are recorded. Four of the family, unaffected by O.D., exhibit idiopathic scoliosis.

Key words: familial; osteochondritis dissecans; scoliosis; short stature

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O.D. involving a single joint is relatively common and in some instances trauma has been implicated as the most important aetiological factor (Smillie 1960). In most patients a family history of the condition is not present, but there are 40 families reported in the literature in whom more than one member was affected (Bernstein 1925, Wagoner & Cohn 1931, Muller & Hetzar 1933, Neilsen 1933, Rahm 1934, Zellweger & Ebnöther 1951, Novotny 1952, Gardiner 1955, Pick 1955, Tobin 1957, Smillie 1960, Smith 1960, Stougaard 1961, 1964, Domack 1963, Fraser 1966, Hanley et al. 1967, Petrie 1977, Auld & Chesney 1979, Mubarak & Carroll 1979). In these families there is usually an autosomal dominant type of inheritance. O.D. has also been described in association with Osgood Schlatter's disease, Scheuermann's disease, carpal tunnel syndrome, tibia vara, endocrine disorders, tall stature, dwarfism; some of these associations have occasionally been noted to be familial (Petrie 1977).

This report describes the occurrence of familial osteochondritis dissecans with dwarfism, and traces 12 affected members of one family through four generations. In addition some members have Scheuermann's disease and minor anomalies of bony development, whilst four of the family unaffected by O.D. show an idiopathic scoliosis transmitted through four generations.

CASE REPORTS

Case 1

S.B. (IV, 1) is a 15 years 11 months old girl (height 142 cm, 3 s.d. below mean). When 11 years old she fell and damaged her right elbow. Radiographs at that time showed some loss of contour over the capitellum, with appearances suggestive of anomalous ossification, but her symptoms resolved with conservative management. Two years later she returned complaining of discomfort in the left elbow. X-rays of both elbows revealed loose bodies in both joints, with evidence of O.D. of both capitella. The loose body in the left elbow joint was removed surgically with subsequent improvement of her symptoms. She also complained of knee and back pain and further X-rays have shown O.D. affecting both patellae and marked irregularity of the end-plates of the vertebral bodies of the thoracic spine, features consistent with Scheuermann's disease. A radiological survey showed the cartilage space of the hip joints to be narrowed to 2 mm, and modelling of the lower femurs to be imperfect, but otherwise bone development was normal. Clinical examination and routine investigations showed no underlying organic cause for the short stature.

Case 2

M.B. (IV, 2) is a 13 years 6 months old boy (height 132 cm, 3½ s.d. below mean). When 11 years and 6 months old he complained of discomfort in both his knees and elbows. Clinical examination was normal, but radiographs revealed a large osteochondral defect in his right medial femoral condyle, with both patellae showing extensive involvement with O.D., and there was ano-

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malous ossification in both capitella. Recent X-rays demonstrate typical osteochondritic lesions of both capitella, but no loose bodies in the joints.

Case 3

G.B. (IV, 3), is an 11 year old boy (height 132 cm, 1½ s.d. below mean). He has never had any joint symptoms but radiological screening revealed an osteochondritic lesion of the right capitellum. In addition the first metatarsals were particularly broad and short, with the epiphyses of the first proximal phalanges forming in two parts.

Case 4

D.B. (IV, 4), is a 9 years 6 months old girl (height 124 cm, 1¾ s.d. below mean). Recently she had noticed discomfort in her back and knees and radiological survey demonstrated O.D. of both patellae and Scheuermann's disease. Both cuboid bones were abnormal and the first metatarsals were broad and short. The epiphyses of the first proximal phalanges were fragmented and there were minor anomalies of development of some of the phalanges.

Case 5

I.B. (III, 4) is a 38 year old woman (height 137 cm, 4½ s.d. below mean for adult females). When a child she had discomfort in both her knees similar to her daughters, but suffered no elbow symptoms. Over the past few years she has had markedly increasing retropatellar discomfort in both knees and radiographs confirmed the presence of bilateral patello-femoral arthritis. Radiological screening revealed no further abnormality apart from old Scheuermann's disease and mild OA changes in both hips.

Case 6

R.M., (II, 4) is a 72 year old woman (height 132 cm). She remembers clearly her knee discomfort as a child, similar to that of her daughter and grandchildren and also described episodes of knee effusion. Ten years ago she had a total hip replacement for OA and at present complains of discomfort in most joints. X-rays of her knees revealed marked OA of all compartments, with loose bodies and a marked defect in the right medial femoral condyle, features consistent with old O.D. There are also generalised OA changes in all joints examined, but no loose bodies in any other joints.

Case 7

S.A. (III, 10) is a 49 year old woman (height 149 cm). She had no history of any joint problems as a child. She has recently suffered from backache, thought due to degenerative changes in an untreated moderate idiopathic thoracic scoliosis. Radiological screening revealed no abnormality of any joints apart from the thoracic scoliosis. She has a 20 year old son (IV, 5) who also has an idiopathic scoliosis, but no other joint problems.

Case 8

H.W. (II, 9) is a 69 year old woman (height 137 cm). She had no joint symptoms in her childhood, but has recently undergone total hip replacement for OA. Radiological screening revealed mild OA changes in her knees consistent with her age, and a moderate idiopathic thoracic scoliosis which dated from adolescence. There was no clinical or radiological evidence of O.D.

FAMILY PEDIGREE

It has not been possible to radiograph other members of the family, but a family tree has been constructed from interviewed relatives and postal correspondence (Figure 1). In the first generation, both D.M. and L.M. (I, 1 and 2) were of short stature (height 142, 147 cm). In addition, D.M. was known to have had a scoliosis and rib hump and L.M. had always complained of painful knees and elbows throughout life, with several episodes of knee effusion when young.

In the second generation, R.M. and H.W. (II, 4, 9) were interviewed and radiographed. Although of short stature D.M., P.M., T.M., and E.M. (II, 1, 2, 6 and 10) had no joint problems as children. D.N. (II, 3) describes symptoms in her knees and elbows since childhood similar to those of the children in the fourth generation of the family, and now apparently is severely disabled by OA, particularly of her knees. She had one daughter P.W., who never complained of joint symptoms. L.C. (II, 5) has always had knee problems, and describes several episodes of bilateral knee effusions as a child. She now has severe OA of both knees, but has never had any elbow symptoms. C.M. (II, 7) always had problems with his knees as a child and underwent an operation to remove "a piece of bone" during adolescence. He underwent a major hip operation when 50 years old, but no details of this are known, nor anything about his three children. He never had elbow symptoms. E.R. (II, 11) described

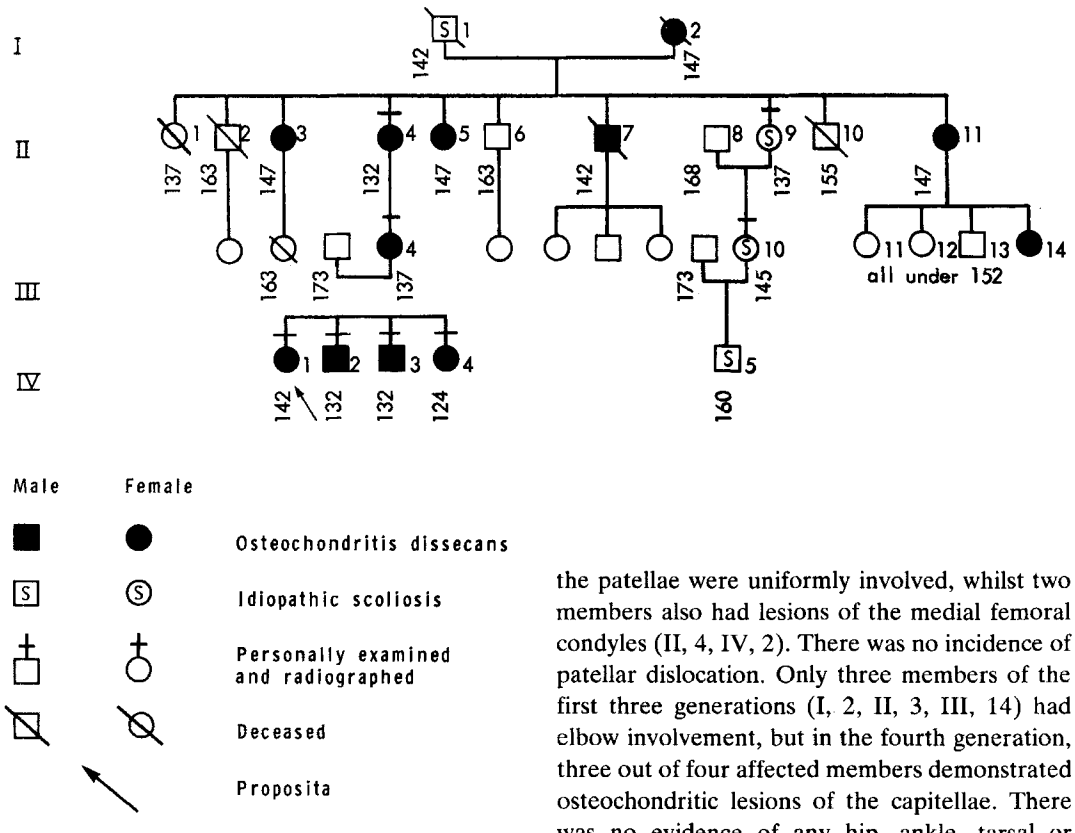

Figure 1. Family pedigree.

symptoms in both her knees during childhood, the symptoms gradually deteriorating. When 40 years old she had a patellectomy. She has four children all under 152 cm in height. One daughter (III, 14) has O.D. of both elbows proven radiologically, with loose bodies having been removed surgically from one elbow. Apparently, there are no joint symptoms in the other three children (III, 11–13).

DISCUSSION

From the case reports described, 12 members of this short statured family had O.D. All affected members demonstrate knee involvement apart from one individual aged 12 years who may subsequently develop lesions. In those radiographed, the patellae were uniformly involved, whilst two members also had lesions of the medial femoral condyles (II, 4, IV, 2). There was no incidence of patellar dislocation. Only three members of the first three generations (I, 2, II, 3, III, 14) had elbow involvement, but in the fourth generation, three out of four affected members demonstrated osteochondritic lesions of the capitellae. There was no evidence of any hip, ankle, tarsal or shoulder lesions in this family.

The pedigree is consistent with an autosomal dominant pattern of inheritance. There is no skipping of generations, which suggests that penetrance of the gene is high. There were no consanguineous marriages and none of those who married into the family have shown any signs of the disease.

Smith (1960) suggested that there are two types of O.D., one developing in adults or older children in which trauma is the major, or even sole, aetiological agent and the other developing in children before epiphyseal closure in which the lesions are often multiple, sometimes familial and this latter type is perhaps due to a predisposition to develop avascular necrosis. Smillie (1960, 1974) believes that an underlying anomaly of ossification is responsible for this predisposition in juvenile O.D. This received support from our pedigree, where two cases of juvenile O.D. of the elbow initially manifested in anomalies of ossification of the capitellae. Duthie & Houghton

(1980) suggested that the developing epiphyseal anlage may be normal, or may have a constitutional minor or major defect, producing a spectrum of dyschondrotic epiphyses. A normal epiphysis may be exposed to extreme trauma and give rise to O.D.; however, a mildly dyschondrotic epiphysis may give rise to O.D. when it is exposed to normal mechanical forces. It is interesting that the fourth generations have elbow as well as knee lesions, which may reflect increasing stresses in the joints during adolescence.

White (1957) and Hanley et al. (1967) suggested that the underlying constitutional disturbance in their cases of familial O.D. may have been an endocrine factor. Paatsana et al. (1975) have shown that overloading of dog humeral heads alone did not give rise to any lesion, but if somatotropin and thyrotropin were added, O.D. was produced in every case. However, there was no clinical or biochemical evidence to support any underlying endocrinological abnormality in the family reported here, similar to the findings of Auld & Chesney (1979). Linden (1977) reviewing the natural history of juvenile O.D. found there were no complications in later life which could be attributed to O.D. However, two members of this pedigree developed marked patellofemoral arthritis by 40 years of age and all other affected adult members, although elderly, have symptomatic OA, and it seems likely that there is also a predisposition to OA in this family.

Idiopathic scoliosis has been recognized in four members of this family, in four successive generations. It has not been present in any members with O.D. It is rare for scoliosis of this sort to behave as a dominant trait (Wynne-Davies 1973, McKusick 1978) and were a dominant gene segregating amongst the offspring of D.M. (I, 1) it is most unlikely that it would have been passed only to H.W. (II, 9). Perhaps milder degrees of scoliosis remained unrecognized in other family members. The genetics of scoliosis in this family are not clear; nothing suggests failure to identify another independently segregating dominant trait of which scoliosis is only a part.

Short stature is known to be associated with O.D. (Zellweger & Ebnöther 1951, Pick 1955). In this family the heights of seven affected adult members range from 132 to 147 cm, with a mean of 142 cm. The four persons with idiopathic scoliosis, but without signs of O.D. had a mean height of 146 cm (range 137 to 160 cm), and those adults with neither signs of scoliosis nor of O.D. had a mean height of 156 cm, and a range from 137 to 163 cm. These figures suggest a shortening of 14 cm as a result of O.D., and a shortening of 10 cm as a result of the independently determined scoliosis, but the shortest of the individuals with scoliosis (H.W. II, 9) is the offspring of a person with O.D. and it is possible that she carries the O.D. gene and that this has produced some shortening, although she has no clinical signs of O.D. Similarly the shortest of the unaffected individuals D.M. (II, 1) is the offspring of a person with O.D. and although there have been no signs or symptoms of O.D. the gene for this disorder may be manifesting in her simply as a gene for short stature.

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