THE HEREDITARY FACTOR IN OSTEOCHONDRITIS DISSECAN

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Since the term osteochondritis dissecans was introduced by König in 1887 the literature has brought several examples of an inherited factor which possibly plays a part in giving rise to the disease. In certain families the incidence of osteochondritis dissecans is higher than the average, although in most patients it has been impossible to demonstrate familial occurrence. These findings might indicate that a possible inherited factor is not invariably a prerequisite or that this factor alone cannot elicit the disease.

Familial occurrence was demonstrated most convincingly by Nielsen (1933) in his series of osteochondritis dissecans affecting the elbow joint. In a study of radiographs of 1,000 normal men he found that 4.1 per cent showed evidence of osteochondritis dissecans in an elbow. Moreover, the male relatives of patients with definite radiological evidence of osteochondritis dissecans showed unmistakable signs of the disease in 14.6 per cent. His series included a family of eight brothers in which two monozygotic twins, and a third brother, had osteochondritis dissecans of the elbow. Other familial cases have been reported. Wagoner and Cohn (1931) described two families. In one family the father, son and a paternal uncle had osteochondritis dissecans of the knees; and in the other two brothers had both knees involved. Bilateral involvement was found by Bernstein (1925) in two sisters and their brother. An almost analogous case was reported by Gardiner in 1955. In the same year Pick described a family in which the mother and three of her four children had osteochondritis dissecans of either elbow or knee. Novotny (1952) found involvement of both knees in two brothers.

Multiple joint involvement in the same patient might indicate a hereditary trait—that is, reduced resistance in the joints concerned to injury. When multiple lesions exist both elbows or both knees are most often involved. Nielsen reported that in 25 per cent of his patients with osteochondritis dissecans of the elbows both were involved. Involvement of both elbows and both knees was described by Watson-Jones (1943). In the same year Wiberg reported a patient with affection of both knees, which incidentally showed spontaneous recovery. Roberts and Hughes (1950) mentioned a patient with osteochondritis dissecans of both elbows and both hips, and Hay (1950) reported one patient in whom both patellae, both capitula and radial heads were affected, and another with involvement of both elbows and one knee. Osteochondritis dissecans of both knees and one elbow was reported by King in 1935.

Thus, osteochondritis dissecans affecting several joints in one patient or in several members of the same family is not uncommon. In a follow-up study at the County Hospital, Soro, Denmark, of forty-eight patients with osteochondritis dissecans of the elbow or knee, interrogation led us to the family reported below. In this family osteochondritis dissecans of the elbow or knee, or of both, occurred in three generations. The family comprised the grandfather, aged seventy-seven, and his eighteen direct descendants. Three of the descendants have not been radiographed, but these three had a negative history and no objective signs of osteochondritis dissecans.

The grandfather had not previously been radiographed, so the radiological diagnosis was somewhat questionable. Advanced osteoarthritis was found in both knees. In the left knee there was also a defect in the medial femoral condyle, which might be ascribed to osteochondritis dissecans, and a large loose body. The patient reported that this loose body had been present since the age of twelve. During his childhood there had been several episodes of locking, pain and swelling of the left knee; so on the basis of the history the diagnosis seems fairly
certain. His deceased wife was said to have had a tuberculous lesion of the left elbow, but further details were not obtainable. Of this marriage there were four children (all grown up).

Their eldest son gave a history very suggestive of osteochondritis dissecans of both knees. From childhood there had been complaints of pain, swelling and locking. Radiography showed the effects of osteochondritis dissecans on the medial femoral condyle, and osteoarthritis of both knees. This patient has two children, the elder of whom showed radiographic evidence of osteochondritis dissecans with a loose body (asymptomatic). The younger sister showed no sign of the disease.

The second offspring, a daughter, showed the effects of osteochondritis dissecans of both knees, with moderate osteoarthritis. From childhood there had been trouble with the right knee. The eldest of her four children had osteochondritis of the right elbow, with a loose body. The other three children—two boys of sixteen and a girl of thirteen—showed osteochondritis of both elbows and both knees. Judging from external appearances, the twins are monozygotic. One of them had an operation on the right knee for removal of a loose body. The elbows were severely affected in this case and the capitulum had almost disappeared; its site was ragged, with several fragments in the joint space apart from actual loose body formation. In the other twin the changes were not so marked. Apart from moderate symptoms from the knees, this patient had approximately 10 degrees' limitation of extension of the left elbow and a tendency to locking. The capitula were flattened, but with smooth joint surfaces and with a loose body, the size of a pea, in each joint. If the disease is accepted as being hereditary in this family, and if the twins were in fact identical, the predisposition to osteochondritis dissecans must have been the same in each. The difference in the degree of manifestation must then be explained by external factors. The younger sister showed unmistakable signs of osteochondritis dissecans in both elbows and knees, but no prominence on the articular surface and no loose body. Her only complaint was of cracking in the left knee after prolonged use.

The next daughter was not radiographed, but the history and the objective findings gave no indication of osteochondritis dissecans. Her four children, three of whom were radiographed, had no joint trouble, and the films showed no evidence of osteochondritis dissecans.

The youngest daughter had no joint symptoms until the age of forty, when she had trouble with the right knee. Radiographs did not show osteochondritis dissecans or its effects. She had four children (three were radiographed), only one of whom is affected with involvement of both elbows and the left knee. The involvement of the knee was not discovered until the present study, because the patient had never complained about it.

FIG. 1
Family tree. Affected members shown in solid black.
The family tree summarises the examples of osteochondritis dissecans in its different members (Fig. 1). Whether the "tuberculous" lesion of the grandmother's elbow should be ascribed to osteochondritis dissecans is difficult to decide. However, it is likely that in this family there is an inherited factor governing osteochondritis dissecans and that it is transmitted as a dominant factor. This is supported by the occurrence of the disease in the presumably monozygotic twins and by the absence of any consanguineous marriages. The conclusion regarding the heredity factor in osteochondritis dissecans in this family and the apparently dominant transmission cannot be extended to apply to osteochondritis dissecans in general because the series is far too small. In addition, it must be considered selected, because it was the only family in the follow-up study to be subjected to a systematic investigation.

SUMMARY

Nine cases of osteochondritis dissecans of the elbow and knee in three generations of the same family are described. There was clear evidence of a dominant inherited factor.

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REFERENCES