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ARTHROPATHY IN VON WILLEBRAND'S DISEASE

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Arthropathy is a common and often severely disabling complication of haemophilia A and B. The incidence of joint lesions in a large series and its variation with the severity of haemophilia has been studied by Ahlberg (1965) and others. The corresponding correlation in von Willebrand's disease has, however, received less attention. Von Willebrand's disease is an autosomal dominant hereditary disease characterised by a low factor VIII (AHF) content and prolonged bleeding time (Nilsson & Blombäck 1963). Haemarthrosis and single cases of chronic arthropathy have been reported in patients with this disease (Nilsson et al. 1957, Sánchez et al. 1963, Corny 1965, Deshayes et al. 1967).

This paper reports the incidence and types of joint lesions in a large defined population.

MATERIAL AND METHODS

The material consisted of all known cases of von Willebrand's disease in Sweden (about 8 mill. inhabitants). At present more than 400 patients belonging to 130 families are on the register. The diagnosis in these cases has been made according to the criteria set up by Nilsson & Blombäck (1963), i.e., prolonged bleeding time, low factor VIII and an autosomal hereditary pattern. The disease is classified as *severe* if the Duke bleeding time exceeds 20 minutes and as *mild* if it is less than 20 minutes. In severe cases the AHF is 1–25 per cent of normal. The present material included 32 patients with severe von Willebrand's disease. The methods used for determining factor VIII, bleeding time, and platelet adhesiveness have been described previously (Nilsson et al. 1957, Silwer et al. 1964, Cronberg et al. 1966).

In a related investigation of all the registered patients (Silwer) notes were made of any previous haemarthrosis and of other joint symptoms. Patients who had reported such complications were subjected to orthopaedic (Ahlberg) and roent-

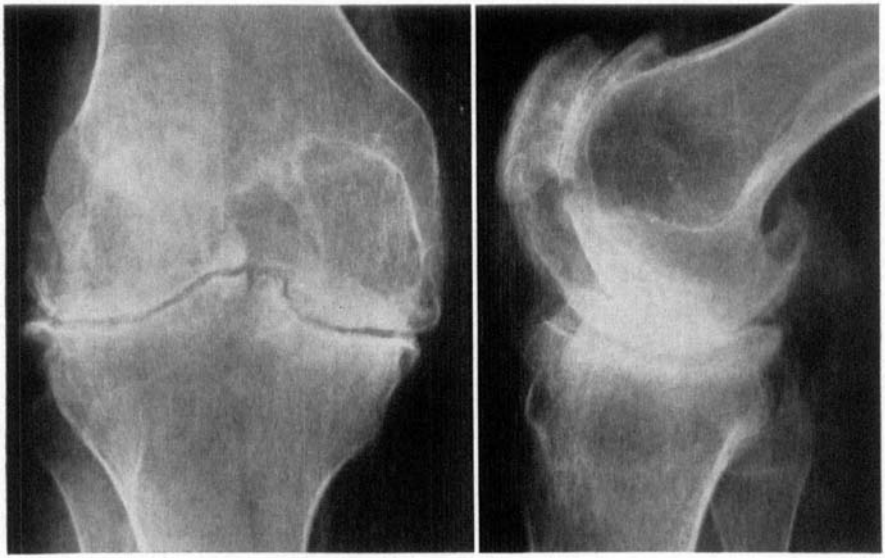

Figure 1. R.B. Right knee. Marked sclerosis of bone ends, narrowing of joint space and osteophytes.

genographic examination. The severity of the joint lesions and the degree of disability were assessed according to Ahlberg (1965).

RESULTS

14 of the 32 patients with severe von Willebrand's disease reported that they had had bleeding in one or more joints. All had had haemorrhage in the knee joint. It had also occurred in the following joints in decreasing order of frequency: ankle, elbow and hip. The haemorrhages had been spontaneous or initiated by trivial trauma. Only in a few cases of mild von Willebrand's disease had there been symptoms of haemarthrosis, and then after a more or less severe trauma.

Seven of these 14 patients had chronic arthropathy of varying severity with involvement of all together 7 knees, 5 ankles, 4 elbows, and 1 hip.

The joint lesions consisted of increased trabeculation, sclerosis of the bone ends, subchondral cysts, narrowed joint space, and osteophytes (Figures 1, 2, and 3). The surfaces of the affected joints were sometimes deformed (Figures 4 and 5).

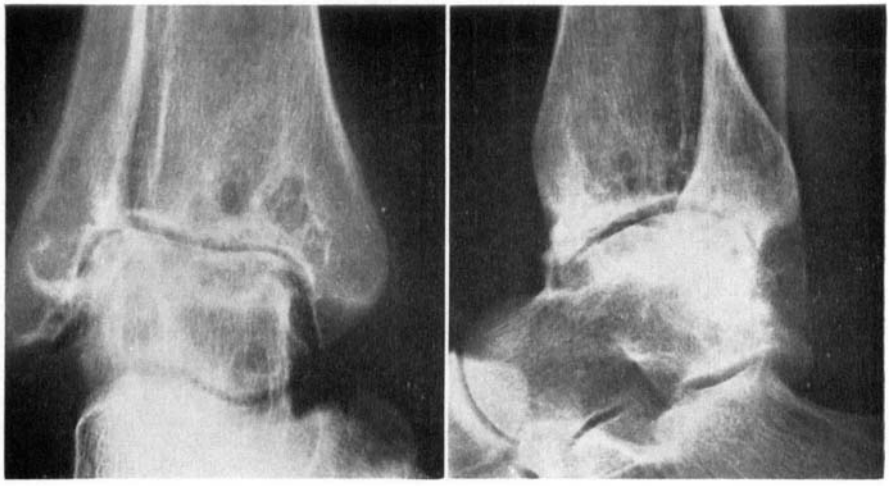

Figure 2. R.B. Right ankle. Marked subchondral cyst formation and sclerosis.

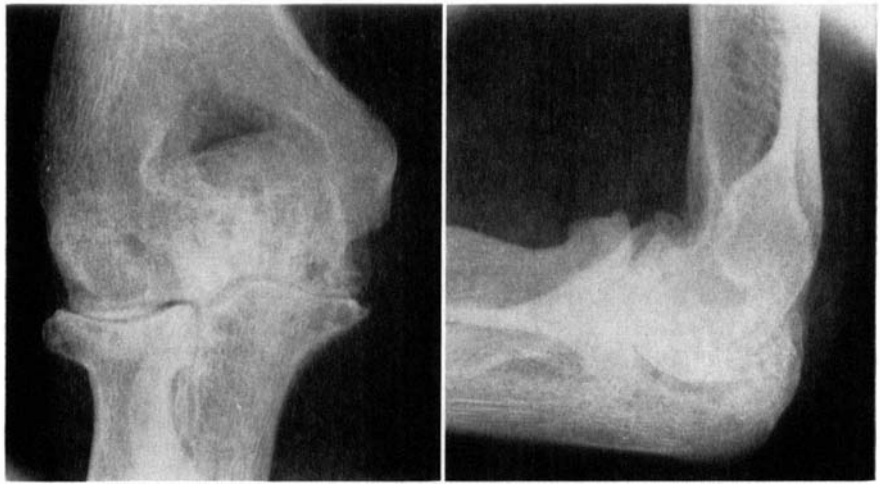

Figure 3. R.B. Right elbow. Sclerosis, subchondral cyst formation, narrowing of joint space and enlargement of radial head.

DISCUSSION

Haemorrhages in the joints and arthropathy are not predominant symptoms of von Willebrand's disease. As expected, they were found to vary in frequency and severity with the severity of the disease, as judged from the bleeding time and the AHF content. Haemarthrosis had occurred in about 40 per cent of the patients with severe von

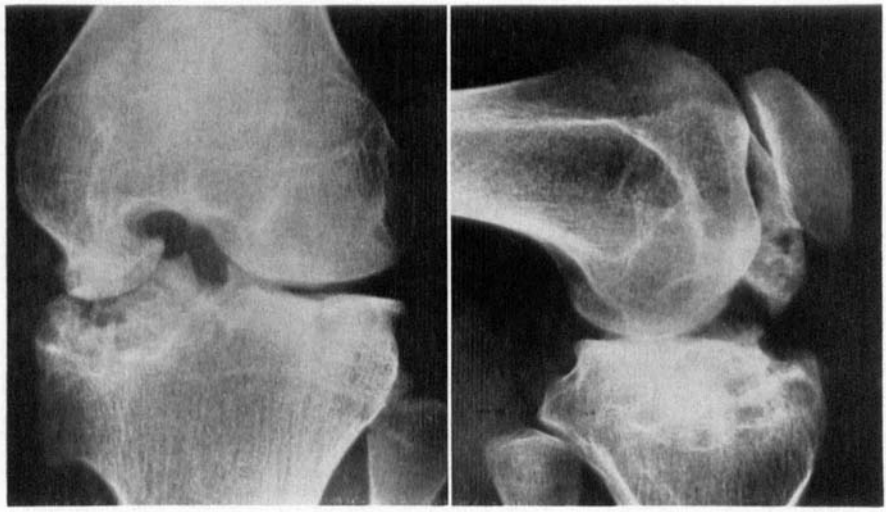

Figure 4. K.A. Left knee. Deformity of the medial tibial and femoral condyles.

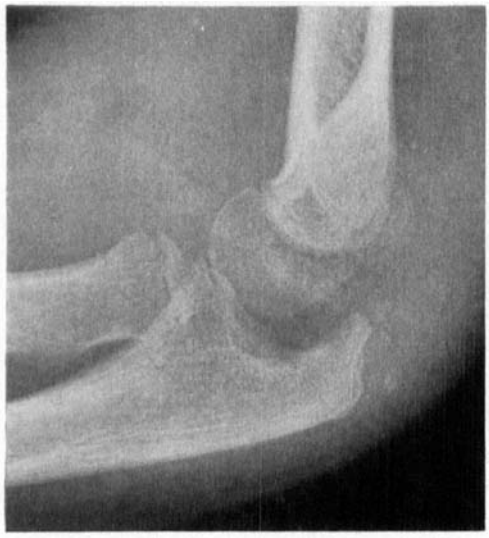

Figure 5. B.H. Left elbow. Deepening of incisura semilunaris.

Willebrand's disease and arthropathy in about 20 per cent. These figures are not strictly comparable with those in haemophilia, because in von Willebrand's disease the bleeding time and the AHF factor content vary more widely from one occasion to another and tend to improve with increasing age. To enable comparison with patients

with haemophilia, the 32 patients with severe von Willebrand's disease were divided into two groups with an AHF of 1–4 per cent and 5–25 per cent, respectively. The former group is comparable with patients with moderate haemophilia and contained 12 patients; the latter, comparable with patients with mild haemophilia, contained 20 patients. Five of the seven patients with arthropathy belonged to the former group and two to the latter. On comparison of joint score as a function of AHF level, mean number of affected joints per patient in various age classes and degree of disability, good agreement was found with the results obtained in Ahlberg's (1965) investigation of patients with haemophilia.

As in haemophilia, the changes were dominated by early and advanced arthrosis. Subchondral cyst formation, trabeculation, and sclerosis were often severe. There was no demonstrable broadening of the epiphysis of the knee and no genu valgum or varum. Neither did the patients show any squaring of the patella, i.e., a lesion typical of haemophilia. In affected elbows the head of the radius was enlarged (Figure 3) and the semilunar incisure was abnormally deep, as in haemophilia (Figure 5).

CONCLUSION

The incidence and severity of joint disease in von Willebrand's disease vary with the severity of coagulopathy. Such lesions are rare in mild von Willebrand's disease. The joint changes are comparable in type, site, and severity with those in cases of haemophilia with corresponding degree of coagulation defects.

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