Haemophilia

Both haemophilia A (factor VIII) and haemophilia B (factor IX) are inherited as X-linked recessive diseases and in both the functional defect is in the intrinsic pathway, the extrinsic tissue-factor-dependent pathway being the only mechanism left intact to subserve haemostasis. The most important consequence of haemophilia is arthritis and it is interesting that synovial tissue is deficient in tissue factor, which may explain this association. Arthritis occurs only in those patients who are severely affected, with concentrations of circulating antihemophilic globulin of less than 5%, and onset is usually in childhood. The knee and the elbow joints are most commonly involved and may become swollen acutely either spontaneously or after only the most minor trauma. Range of movement, usually preserved after the first episode, is progressively lost with subsequent attacks and severe joint destruction and deformity may follow. Extra-articular bleeding is also common.

Pathologically there is a chronic proliferative non-specific synovitis with extensive haemosiderin deposition and fibrosis. Haemorrhages occur both into the joint and in subchondral bone. Radiologically soft tissue swelling proceeds to local osteopenia, the development of large subchondral and juxta-articular bone cysts, cartilage destruction and secondary osteoarthritis. Fibrous bony ankylosis may follow but true bony ankylosis is uncommon.

The diagnosis is usually perfectly straightforward and frequently will have been made in the past. The usual presentation is a child with a single swollen peripheral joint. The most troublesome differential diagnosis is from infection and the haemophiliac joint is itself at risk from this complication. The other group of conditions which may be confused with this presentation in this age group is juvenile chronic arthritis. Bleeding into muscles and non-articular soft tissues may be the presenting complaint. Bleeding into the forearm may produce Volkmann’s ischaemic contracture, into the gastrocnemius a talipes equinus deformity, and into the iliopsoas a flexed hip with pain in the groin. The last may be distinguished from an intra-articular process in the normal
classical way by demonstrating that passive rotation of the hip is preserved. Once suspected, the diagnosis is confirmed by demonstration of the specific coagulopathy. The family history may of course provide the diagnostic clue and may permit initiation of treatment in hitherto unaffected children at the earliest possible moment.

The limiting factor in treatment is prompt diagnosis, both initially and with each successive bleeding episode, and the recent improvement in prognosis represents a synthesis of prompt recognition coupled with the immediate institution of effective treatment. Home programmes incorporating prophylaxis have contributed in large measure to the improved prognosis. Severely affected patients will require replacement as often as weekly at some stages in their disease, and it is particularly important in such patients that their requirements are properly calculated and that circulating anti-factor VIII or IX antibodies are detected if present. It seems that there may be a familial predisposition to the development of these acquired inhibitors.

The aim, therefore, is prevention rather than treatment, and the combination of specialty hospital units working together with the family doctor and an informed family has improved the outlook for these children dramatically. The most important adverse reaction to cryoprecipitate or factor concentrates apart from allergic reactions is hepatitis, which may be of the A, B or non-A, non-B variety. The indications for elective surgical intervention in those patients who do not possess inhibitor antibodies are in accord with general principles and the management of local articular or extra-articular bleeding likewise should follow routine lines. Once bleeding has been controlled by the prompt elevation of the circulating factor concentration to at least 50% of normal then there may be a case for aspiration of a haemarthrosis. The presence of blood in the articular cavity is all that is required to produce the histopathological abnormalities of the disease in dogs. The importance of maintaining local muscle power and tone around an affected joint and of avoiding the development of a flexion deformity needs no further emphasis. The management of those unfortunate children who do develop circulating antibodies may tax the resources of the best team and is an area which only the specialist should direct.

**Haemoglobinopathies**

Arthritis and musculoskeletal complications occur commonly in patients with haemoglobinopathies. Many of these manifestations are common to the process of chronic haemolysis rather than to the underlying specific metabolic defect. In all there is a tendency to marrow expansion with trabecular changes in the axial skeleton, and in HbS this process extends to the peripheral long bones and even to the phalanges, producing the “hand-foot” syndrome which may be the presenting complaint. The extremities are inflamed and swollen and the child may be toxic and pyrexial. Other features which may be common