A middle-aged man with monoclonal gammopathy and osteopetrosis

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Introduction

Osteopetrosis is a rare disorder with generalised increase in bone density, and impairment of osteoclast mediated bone resorption [1], with a reported prevalence of 1 in 100 000 – 500 000 adults for the mild (benign) osteopetrosis and 1 in 200 000 – 500 000 for the infantile (malignant) osteopetrosis. Mild osteopetrosis may cause no symptoms. Serious forms can result in stunted growth, deformity, fractures, and anaemia, and nerve compression by extra bone causing blindness, facial palsy and deafness. There is no known cure. Monoclonal gammopathy is not a recognised association of osteopetrosis, and a Medline search did not show previous reports.

Case report

A 53-year old man presented with sudden onset loss of vision of his right eye. He had noticed gradual impairment of vision in the left eye also over a 6-month period. He also had dyspnoea on moderate exertion for 2 months. There was no history of bone pain, recurrent infections, bleeding manifestations or fractures. He was pale, with bilateral axillary and inguinal lympadenopathy and moderate hepatosplenomegaly. Both optic fundi had Roth spots and deep retinal haemorrhages with normal vessel calibre and a macular haemorrhage was seen in the right eye.

His haemoglobin was 5.2g/dl, and the platelet count 35×10^9/l. Blood picture was leucoerythroblastic with marked rouleaux formation. ESR was 155 mm in the first hour. Renal function, liver function and serum calcium were normal. Urine Bence-Jones proteins and serum cryoglobulin were negative. Plasma electrophoresis showed a monoclonal gammopathy of 80g/l.

A skeletal survey showed generalised bone sclerosis (figure 1). Right axillary lymph node biopsy was compatible with extensive extramedullary haemopoesis (figure 2). A bone biopsy from the right tibia confirmed histological evidence of osteopetrosis.

His anaemia and low platelets were corrected by blood and platelet transfusions, and vision in the right eye gradually improved with the regression of macular haemorrhage. He defaulted and died subsequently after emergency hospital admission from heart failure complicating severe anaemia.

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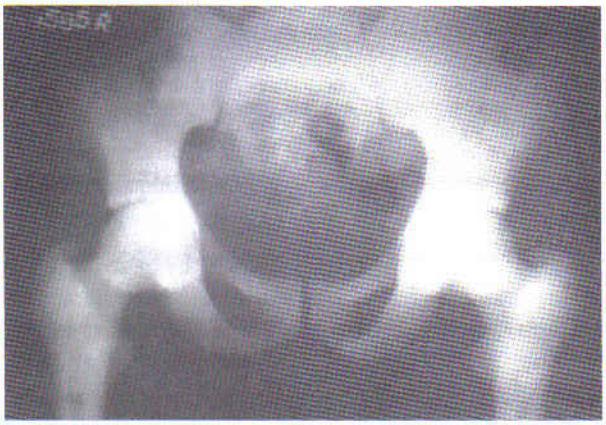

Figure 1. **Xray of pelvis showing bone sclerosis.**

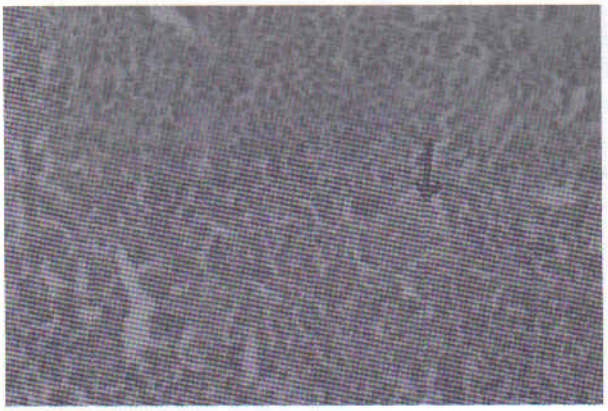

Figure 2. **Lymph node biopsy showing extra-medullary haemopoesis with 2 megakaryocytes (arrowed) H and E 40×10.**

Discussion

Diagnosis of osteopetrosis is usually by skeletal survey showing unusual high density of bone with a chalky white appearance. Bone density tests such as quantitative computed tomography, and dual energy xray absorptiometry and bone biopsy can confirm the diagnosis.

Two types of osteopetrosis are described. The autosomal recessively inherited malignant or infantile osteopetrosis and the autosomal dominantly inherited mild or benign osteopetrosis of adults. A third autosomal recessively inherited form of osteopetrosis known as the intermediate form is now considered to accommodate patients with features that do not clearly fit into the other two types. It is likely that the patient we describe belongs to this category.

In smouldering multiple myeloma the IgM concentration is more than 30g/l but there are no other complications such as anaemia, renal insufficiency or skeletal lesions. Smouldering multiple myeloma could therefore explain this patient's monoclonal gammopathy, and osteopetrosis could explain the other clinical findings. Bone marrow cytological analysis was not possible as the marrow cells were distorted during bone demineralisation, and so the origin of the monoclonal gammopathy and the association with osteopetrosis remains unexplained.

References

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