Case report

Chondrosarcoma of the calcaneum and massive soft tissue calcification in a patient with hereditary and acquired connective tissue diseases

I P WICKS AND A FLEMING

From the Prince Henry Hospital, Sydney, Australia

SUMMARY We describe the first case of the coexistence of the hereditary connective tissue disorder multiple exostoses (HME) and an acquired connective tissue disorder manifest by the overlap of dermatomyositis (DM), scleroderma (PSS), high titre speckled pattern antinuclear antibodies, and increased antibodies to double stranded deoxyribonucleic acid (DNA). Furthermore this patient developed chondrosarcoma of the calcaneum (an unusual site for this malignancy) and massive soft tissue calcification (an unusual feature of PSS, adult DM, and systemic lupus erythematosus (SLE)).

Key words: hereditary multiple exostoses, dermatomyositis, scleroderma, systemic lupus erythematosus.

Case report

A 45 year old Greek woman presented with a three month history of arthralgia, weakness, fatigue, fever, and skin rash. Examination showed muscle weakness and an erythematous rash over the face and metacarpophalangeal joints. The erythrocyte sedimentation rate and muscle enzymes were raised, and electromyogram and muscle biopsy confirmed myositis. An x ray examination showed multiple exostoses and a large calcific mass involving the right calcaneum. Treatment with high dose steroids was commenced for dermatomyositis, with little effect. Subsequently azathioprine and then cyclophosphamide were used with some clinical and laboratory improvement. One year after presentation x rays showed the first evidence of soft tissue calcification around the knees. Over the next two years bilateral hip replacements were required for avascular necrosis of the femoral heads attributed to steroids. She was then lost to follow up and was not seen until nine years after her initial hospitalisation. She had now developed Raynaud's phenomenon

and dysphagia. Examination now showed florid scleroderma of the limbs, face, and trunk, severe joint contractures, and multiple bony nodules. A previously noted mass around the right ankle had increased in size. An x ray examination of the right ankle showed a densely calcified cauliflower-like lesion arising from the calcaneum (Fig. 1) and extensive soft tissue calcification of the chest wall and limbs (Figs 2 and 3). Serology showed antinuclear antibodies (HEp₂ substrate) in a titre of 1/10 000 with a speckled pattern of immunofluorescence, double stranded DNA binding of 73% (normal <30%), and antibodies to extractable nuclear antigens (RNP 1/32, Sm 1/4, SS-A 1/512). Biopsy of the calcaneal mass showed a lobulated chondroid tumour invading normal trabecular bone with pleomorphic nuclei and occasional mitotic figures, consistent with a low grade chondrosarcoma (Fig. 4).

Discussion

HME is an autosomal dominant disorder characterised by excrescences of bone and cartilage. Our case would appear to be the first report linking this hereditary connective tissue disorder with the subsequent development of an acquired connective tissue

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Correspondence to Dr A Fleming, Department of Rheumatology, Prince Henry Hospital, Anzac Parade, Little Bay, Sydney 2036, Australia.

Fig. 1 An x ray of the right ankle showing a densely calcified cauliflower-like lesion arising from the calcaneum, soft tissue calcification and tibial exostosis en face (arrow). (From the Department of Medical Illustration, University of NSW and teaching hospitals.)

Fig. 2 Chest x ray showing soft tissue calcification in the upper arm and chest wall. (From the Department of Medical Illustration, University of NSW and teaching hospitals.)

syndrome. This was of the overlap type with clinical features of DM, florid PSS, and serological features of active SLE.

The possibility that DM was related to underlying chondrosarcoma cannot be excluded in this patient. There is one previous report of such an association.¹ It would appear more likely that DM was the presenting feature of an independent connective tissue syndrome, with evolving features of PSS and SLE.

Malignant transformation in HME is well described, with recent reports indicating a frequency of between 0.9%² and 3%.³ PSS⁴ and the use of cytotoxic agents⁵ (and probably mixed connective tissue disease⁶) have been associated with an increased risk of developing malignancy. The clinical and radiological findings at presentation of our patient, however, suggest that local proliferation and low grade malignant change preceded the onset of the acquired connective tissue syndrome. A gradual transition to malignancy over many years has been reported in HME.⁷

Chondrosarcoma occurs most commonly in the pelvis and femur.⁸ The calcaneum is a most unusual site for this malignancy.⁹ Approximately 6% of all chondrosarcomas are associated with HME.⁸

Our patient also showed widespread soft tissue calcification. Although such calcification can be a feature of juvenile DM and some degree of calcinosis is seen in PSS (particularly the CREST variant (calcinosis, Raynaud's phenomenon, oesophageal dysmotility, sclerodactyly, telangiectasia)), massive soft tissue calcification is an unusual complication of adult DM¹⁰ and is rare in PSS¹¹ and SLE.¹²

Fig. 3 An x ray of the pelvis showing extensive soft tissue calcification and bilateral hip replacements. (From the Department of Medical Illustration, University of NSW and teaching hospitals.)

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Fig. 4 Biopsy of the calcaneal mass showing a low grade chondrosarcoma with a lobulated periphery invading fibrous tissue (arrow). Most of the nuclei are small and darkly staining, while some appear larger and paler with a more open nucleus. (Haematoxylin and eosin.) (From the Department of Medical Illustration, University of NSW and teaching hospitals.)

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