Muscular heterotopic calcification with ulcerating skin lesions: a case report

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INTRODUCTION

Heterotopic calcification of muscles may arise from a variety of diseases, among which are myositis ossificans (MO) and inflammatory myopathies. MO is an umbrella term for non-neoplastic soft tissue mineralization, mostly affecting skeletal muscles. Two distinct subtypes are described: MO traumatica, resulting from trauma with repeated acute or chronic injuries, and MO progressiva (fibrodysplasia ossificans progressiva [FOP]), due to mutations in the *ACVR1* gene. Among the non-hereditary autoimmune myopathies, dermatomyositis (DM) is most common, and is accompanied by skin lesions, most typically a rash in the face, shoulders, elbows and trunk.

CASE REPORT

A 26-year-old Bangladeshi woman presented with indurated swellings on the right upper arm and elbow as well as the left thigh and knee. During childhood, she suffered repeated but minor trauma to the knee and elbows. At age 6, she developed generalized weakness with formation of globular, discharging swellings at 9 years of age. At age 11, extension of the elbows became limited. Numerous swellings on her scalp, followed by alopecia, were noted at age 20. The natural course of the disease was characterized by flare-ups of intensified swellings with localized, dull pain, lasting 3-4 days. As of age 21, she was treated with Alendronic acid, Calcitonin nasal spray, NSAIDs and recently with Ibandronic acid. Family history was negative. Physical examination revealed no congenital deformities of the toes. Biochemical analysis confirmed normal serum levels of calcium, phosphorus, muscle enzymes and inflammatory markers. Skin and muscle biopsy of the affected knee revealed calcification of the soft tissues. Radiographs revealed no bone malignancies but excessive mineralization in the muscles (Fig. 1).

RADIOGRAPHIC AND SKIN PHENOTYPE

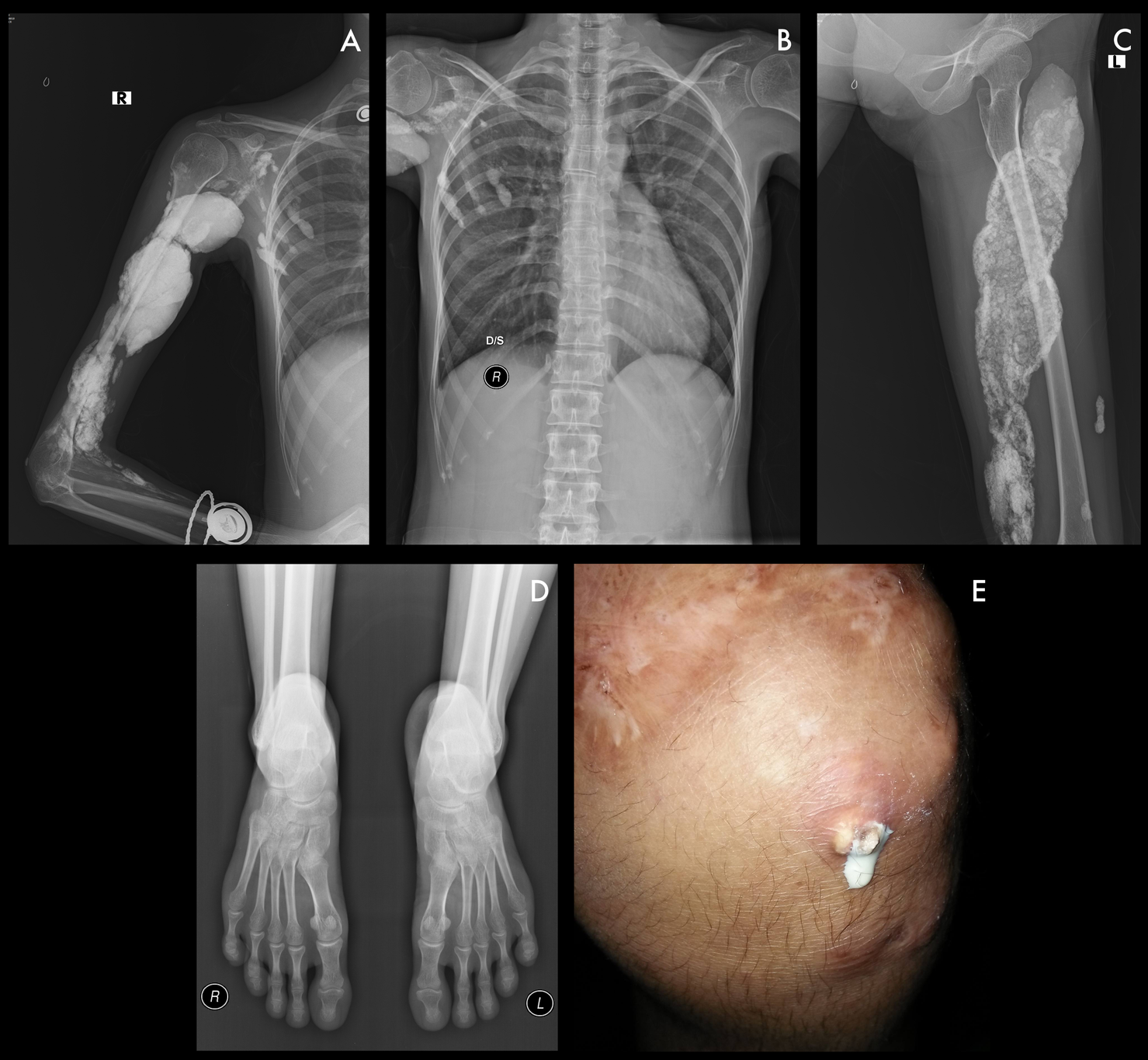

Figure 1: (A-D) Radiographs of the proband, showing extensive calcification in the soft tissue of the right upper arm (A), the pectoral and intercostal muscles (B) and left upper leg (C), separate from the underlying periosteum. Radiographs of the feet show no congenital deformation of the big toes (D). Indurations and irregular, hard, and painful skin lesions on the left knee accompanied by a white discharge (E).

DIFFERENTIAL DIAGNOSIS

Normal calcium and phosphorus serum level

Dystrophic calcification

Malignancies

⊗ No imaging evidence of osteosarcoma

Progressive osseus heteroplasia

⊗ No osteoma cutis

⊗ Calcification beyond the subcutis

Typical FOP

⊗ Normal big toes

Atypical FOP

⊗ Normal sequencing of the *ACVR1* coding region

Polymyositis

✓ Muscle weakness

⊗ Childhood onset

⊗ Asymmetric distribution

Dermatomyositis

✓ Discharging ulcerating lesions

⊗ Absent rash and symmetric distribution

⊗ CK & inflammatory parameters normal

Myositis ossificans traumatica

✓ Repetitive trauma

⊗ Discharging ulcerating lesions

DISCUSSION & CONCLUSION

Heterotopic calcification of the muscles is a severe complication of a spectrum of hereditary and non-hereditary conditions, leading to significant morbidity. Among the non-hereditary causes, traumatic MO is most common and typically affects the masticatory muscles, the arms, pectoral muscles and intercostal spaces or the hips and glutei muscles. Though usually the history of trauma is obvious, many cases of atypical MO have been described. The presented case suffered repeated but mild trauma in the past, but also presented with recurrent discharging skin lesions. These have been previously described in DM (Kaliyadan et al., 2011) but other hallmark features (skin rash, elevated CK or ANA) were absent and therefore diagnostic criteria for DM are not fulfilled. Possibly, the skin lesions are the result of the protracted natural evolution with initiation of treatment only 15 years after the first symptoms. This case demonstrates the importance of comprehensive and accurate phenotypic characterization in diagnosing heterotopic ossifications.

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