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FAMILIAL ACRO-OSTEOLYSIS THROUGH THREE GENERATIONS

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Six cases of familial acro-osteolysis in 15 members of 3 generations of a family are reported. Their pedigrees traced followed an autosomal dominant pattern. Five of these patients were wrongly labelled as patients of leprosy and treated with dapsone monotherapy for variable periods without any benefit. Awareness and differentiation of acro-osteolysis from leprosy is emphasised.

Key Words : Acro-osteolysis, Leprosy

Introduction

Familial acro-osteolysis known variably as Thevenard syndrome, ulcero-mutilating acropachy or more recently as Hereditary sensory neuropathy-I (HSN-I) is an unusual familial disorder transmitted as an autosomal dominant trait. The disease refers to lytic changes in shafts of distal phalanges with preservation of tufts and bases. The process is purely osteolytic without any evidence of bone regeneration which gradually extends proximally and eventually involves other acral bones.[1] Anaesthesia and analgesia of acral parts of limbs occur with or without loss of tendon reflexes, preservation of motor functions and it is associated with nerve deafness occasionally.[2]

Six cases of acro-osteolysis are reported in 15 members of 3 generations of a family, 5 of whom were treated with dapsone monotherapy with a diagnosis of leprosy with variable periods ranging from 2 to 5 years without any response to treatment.

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Case Reports

First Case : The presenting case (Case No. 4, Fig. 1) was 20 years old male

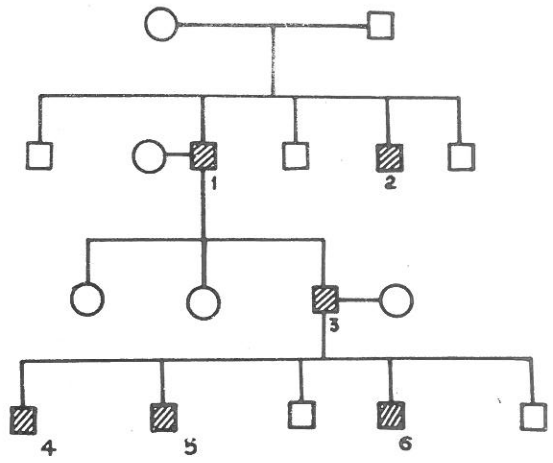

Fig.1. Dominant inheritance pattern in the acro-osteolysis family

who presented with left fore-foot amputation, partially occurring by itself and in part done by a local surgeon because of a non-healing ulcer over the planter aspect of left great toe of 6 years duration and a non-healing ulcer on the planter aspect of right great toe of 5 years duration. On examination a single ulcer of 1.5 x 2 cm size was present on plantar aspect of right great toe which was non-tender, deep, punched out with hyperkeratotic edge (Fig. 2). There was ill-

defined area of complete loss of touch, pain and temperature sensations involving all the toes and extending upto ankle dorsally and heel ventrally. Left foot with a forefoot amputation showed similar sensory loss extending upto ankle (Fig. 2).

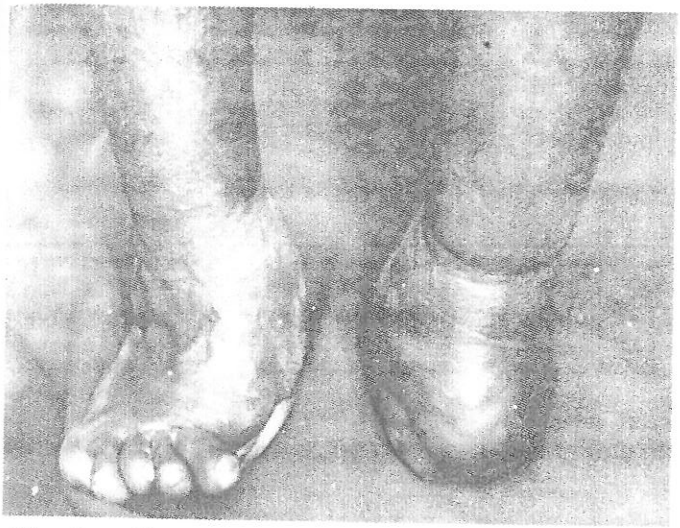

Fig. 2. Showing left forefoot amputation and an ulcer on plantar aspect of right great toe in case 4

Cutaneous examination revealed essentially normal skin, no hypopigmented or erythematous patches and no enlargement of peripheral nerve trunks. Motor functions including reflexes were normal and sensations were intact in upper limbs, hands and other parts of body. Examination of spine was within normal limits and there was no sensory deafness. Skin slit smears were normal on 2 occasions and skin biopsy was essentially within normal limits. Patient refused to undergo nerve conduction studies or nerve biopsy. Radiological examination of foot showed in the right foot decreased joint space with osteolysis of first four metatarsals and bases of 2nd to 4th phalanges with loss of phalanges of big toe and dislocation at 5th metatarsophalangeal joint (Fig. 3). Tips of all distal phalanges showed early

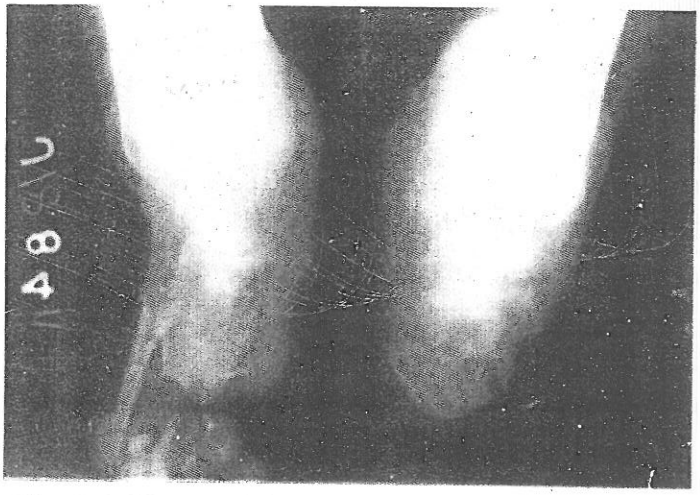

Fig. 3. X-ray of feet demonstration complete loss of left forefoot at metatarsales which are disorganised and osteolysis changes of metatarsals and phalanges of right foot in case 4

osteolysis. Left foot showed complete less of forefoot at metatarsals which were disorganised. Osteopenia was seen in all bones. Overlying soft tissue was smooth and regular. X-ray of hands revealed no bony changes. Patient was explained about the nature of his disease, care of his feet and daily dressing and is on follow up for ulcer and specialized foot wear.

Second Case : 18-year-old male (Case No. 5, Fig. 1) younger brother of the presenting case showed similar pattern of disease with amputation of left great toe and subluxation of matatarsophalangeal joints of 4 years duration with a trophic ulcer over plantar aspect of right great toe of 2 years duration. Pattern of loss of sensation, motor functions and cutaneous examination was similar to as seen in the presenting case. Skin slit smears were negative on 2 occasions and skin biopsy was normal. He toe refused to undergo nerve conduction studies and a nerve biopsy.

Radiological examination of foot showed in the right foot resorption of 2/3rd of 1st and 4th metatarsal and complete reserption of 5th matatarsal. Phalanges of great toe and 5th toe and proximal parts of 2nd and 3rd toes showed resorption. Left foot showed decreased intratarsal joint space and resorption of distal half of first matatarsal and phalanges of big toe. X-rays of the hands were normal.

Third Cases : 12 years old male (Case No. 6, Fig. 1) youngest brother of the presenting case showed a trophic ulcer over plantar aspect of right great toe which was present since the last 9 months only. Loss of sensation was limited to right great toe and head of first matatarsal. No amputations or deformities were present. General physical examination and sensory and motor functions were within normal limits. Skin slit smears were negative and skin biopsy was normal. Nerve conduction studies and nerve biopsy was refused by the patient. Radiological examination of foot showed acro-astoolysis in tips of distal phalanges of all toes, soft tissue and X-ray of hands were normal.

Forth, Fifth and Sixth Cases : The patients were aware of similar disease in their father who is 60 years old (Case No. 3, Fig. 1) who also had a left great toe amputation and sensory deficit of left foot upto ankle. Their grandfather (case 1) and grandfather's younger brother (case 2) had similar disease and they both died of natural causes, at the age of 72 years and 68 years respectively. There was no further pedigree of case 2 as he remained unmarried. Presenting cases (No. 4,5,6) are also unmarried as yet partially because of their skin diserder.

Discussion

Nelton[3] was the first to describe an unusual disease of bones of foot observed in several members of one family. Similar cases were reported by Smith[4] and Heller et al[5] and word neuropathic was added to familial osseous atrophy. Begaert[6] concluded that ulcers and mutilating lesions were due to affliction of spinal ganglia, spinal nerve roots and to a certain extent due to peripheral nerve trunks and their network. The disease was hereditary in most of the patients but sporadic cases were also reported. Three types of acro-osteolysis have new been documented, familial, non-familial (idiopathic/sporadic) and occupational type (in polyvinylchoride worker).[7] The present cases correspond to familial type which is probably same as hereditary sensory neuropathy-I (HSN-I) characterized by familial occurrence, dominant inheritance, male preponderance, less of finger nails, plantar ulceration, sensory disturbances and occassional association with sensory deafness.

Although the exact aetiology is not known, Cheney[8] postulated a process of vascular bone resorption associated with ingrowing of capillaries, the bone being resorbed ahead of advancing vessels producing ischaemic necrosis of bone. The similarity of symptomatology, character of multilation of peripheral parts of limbs and familial occurrence of this disorder can mislead the unwary to the diagnosis of leprosy which is especially true in areas where leprosy is endemic. Five patients in the present family of acro-osteelysis have undergone this social stigma and have received anti-leprosy treatment for many years without any benefit. Although the

psychosocial aspect of this disease are to some extent similar to that of leprosy but the social implications of their wrong diagnosis as leprosy patients cannot be overemphasised. The clinical presentation, skin slit smears, skin biopsy and radiologic changes of osteolysis without any avidence of bone regeneration in acral bones in acro-osteolysis and that of concentric bone atrophy and pencilling as seen in leprosy, are all helpful in the differentiation between acro-osteolysis and leprosy.

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