Nail changes in acro-osteolysis: A case report and review of the literature

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INTRODUCTION

Acro-osteolysis (synonym *phalangeal osteolysis*) refers to the resorption of one or more of the distal phalanges of the hands or feet.[1] Two characteristic radiologic variants have been described, and may coexist in the same patient.[2] In the transverse type, linear bands of resorption are evident in the shaft of the distal phalanx, perpendicular to its long axis. In the longitudinal type, there is resorption of the tuft of the distal phalanx. Although case reports tend to emphasize these radiologic findings, little attention is paid to nail changes. We report idiopathic acro-osteolysis in an otherwise healthy 10-year-old girl presenting to our dermatology service with nail dystrophy. We performed a literature review for similar cases and for nail changes previously reported in acro-osteolysis.

CASE REPORT

A 10-year-old girl presented to a dermatology service with dystrophic changes involving all 20 nails. She had been diagnosed with nail lichen planus at 2 years of age, with no other mucocutaneous features of lichen planus and was subsequently lost to follow-up. Although at first inspection the nail changes were consistent with lichen planus, contractures of the fingers and clinical suspicion of resorption of the distal phalanges (acro-osteolysis) necessitated a careful review of the case.

The child was not known to have any other chronic medical conditions and had no history of fractures, skin fragility, or blistering. She could not recall a history of rashes and had no symptoms of Raynaud phenomenon. She did, however, admit increasing difficulty holding a pen at school. She was born of nonconsanguineous parents living in Cape Town, South Africa. Her maternal grandmother had been known to have "kort vingers" (short fingers). No other family members were affected. There was no maternal history of anticonvulsants or other substance use during pregnancy.

On physical examination, the child appeared generally well with appropriate behavior and intelligence, normal hearing, and no dysmorphic features, including normal hair and dentition and, other than the hands, had no overt musculoskeletal abnormalities. Although all nails were involved, the hands were disproportionately affected. Brachyonychia with longitudinal ridging was noted along with pterygium formation. Additional observations were dyspigmentation of the second fingers bilaterally (with no history of blisters or other preceding lesions at those sites) and shortening of distal phalanges (Figs 1 and 2). She had reduced flexion at the distal interphalangeal joints of digits 2 through 5 on both hands consistent with contractures (Fig 2). There were no other skin or mucosal findings. Radial and dorsalis pedis pulses were normal with warm peripheries.

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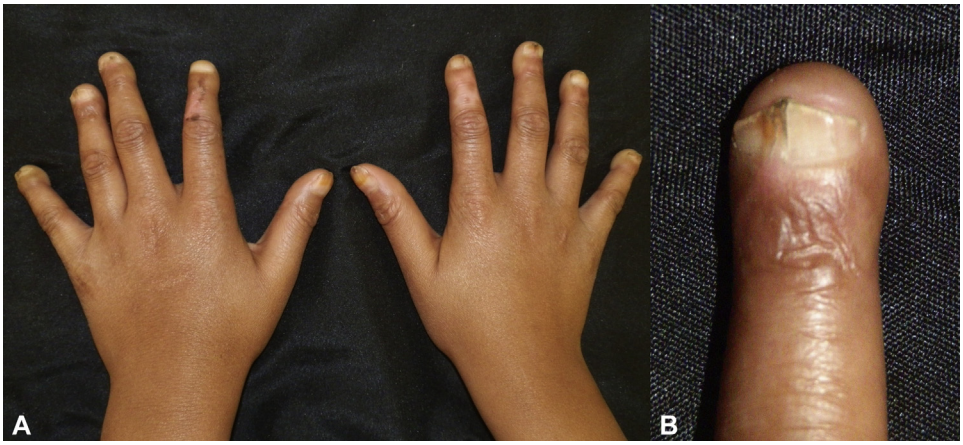

Fig 1. A, Symmetric involvement with shortening of digits, brachyonychia, longitudinal ridging, and pterygium formation. Note the dyspigmentation of the skin of the second digits bilaterally. **B**, Prominent pterygium.

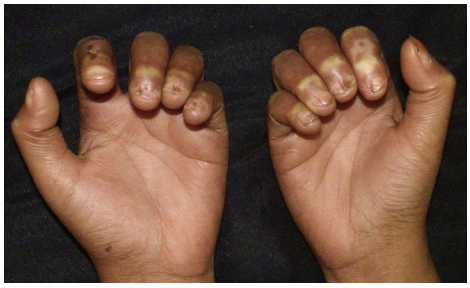

Fig 2. Contractures limiting flexion of second through fifth distal interphalangeal joints.

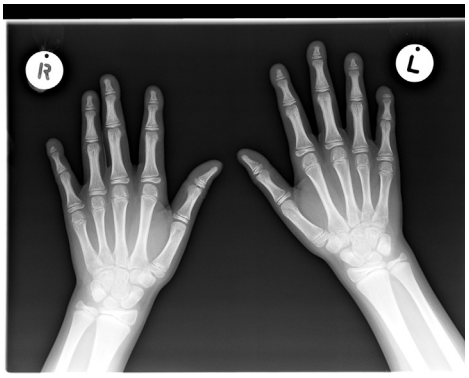

Fig 3. Radiograph of both hands confirming longitudinal acro-osteolysis.

Chest radiograph and long bone radiographs were unremarkable. An extensive blood workup was similarly unremarkable, with a normal full blood count, renal and liver function tests, normal calcium (2.41 mmol/L), magnesium (0.93 mmol/L), phosphate (1.54 mmol/L), parathyroid hormone (1.6 pmol/L), C-reactive protein (1 mg/L), erythrocyte sedimentation rate (5 mm/h), creatine kinase (90 U/L), and thyroid stimulating hormone (2.84 mIU/L). Auto-antibody tests were negative (including antinuclear, anticentromere, anti-Jo1, anti-Scleroderma-70, anti-RNP-70, anti-Sm and anti-neutrophil cytoplasmic antibodies). Rheumatoid factor was <10 IU/mL and Von Willibrand factor antigen was within the normal range (94%). Plain radiographs of the hands and feet, however, confirmed diffuse longitudinal acro-osteolysis (Fig 3). An incisional biopsy of the nail matrix and bed showed dermal fibrosis but no inflammation. Epidermal clefting along the basal layer was presumed to be caused by surgical avulsion of the nail plate rather than epidermolysis bullosa simplex. Color Doppler ultrasound scan of the hands was performed and showed normal flow in the radial and ulnar arteries bilaterally. However, digital flow could not be demonstrated in the digital arteries. The significance of this finding remains uncertain given the lack of clinical or serologic evidence of an underlying small vessel vasculopathy.

In light of the above findings, a revised diagnosis of idiopathic acro-osteolysis was made. The child was referred to occupational therapy for assistance with hand function. No medical treatment could be offered, and she was booked for multidisciplinary follow-up (dermatology, rheumatology, and occupational therapy).

DISCUSSION

Pubmed, Web of Science, and Scopus were searched for case reports using combinations of the title/abstract key words *acro-osteolysis*, *acroosteolysis*, and *phalangeal osteolysis*.

Main causes of acro-osteolysis derived from various proposed classifications are presented in Tables I and II.[1,3-5] We found no cases of acro-osteolysis

Table I. Primary acro-osteolysis

Genetic disorders
Hajdu-Cheney syndrome
Hereditary sensory neuropathies
Noggin mutations
Laminopathies
Epidermolysis bullosa
Lysosomal storage disorders
Gaucher disease
Pycnodysostosis

Table II. Secondary acro-osteolysis

Vasculopathies
Raynaud disease/syndrome
Scleroderma
Thromboangiitis obliterans (Buerger)
Inflammatory disorders
Rheumatoid arthritis
Psoriatic arthritis
Dermatomyositis
Systemic sclerosis
Granulomatosis with polyangiitis (Wegener)
Sarcoidosis
Infections
Leprosy (sensory neuropathy)
Metabolic
Porphyria
Hyperparathyroidism
Diabetes mellitus type 1
Idiopathic
Toxins
Vinyl chloride
Phenytoin
Alcohol (peripheral neuropathy)
Trauma
Repetitive mechanical injury
Frostbite

associated with lichen planus. However, 2 cases remarkably similar to ours have been reported in Cape Town.[1] This finding suggests the possibility of a locally prevalent variant of acro-osteolysis. Our case also shares similarities with a more severely affected 9-year-old Saudi boy found to have a homozygous *LMNA* missense mutation.[6]

Nail pathology is widely reported in acro-osteolysis. Brachyonychia refers to short, broad, flat nails, and is the most commonly reported nail change.[2,4,5,7-10] The term *racquet nail* is synonymous but implies associated deformity of the bone and soft tissue.[11-13] Other reports include anonychia, atrophy, transverse ridging, discoloration, thickening of the nail plate, hyperkeratosis of the cuticles, pincer nails pitting, and onycholysis.[13-20] Longitudinal ridging is evident in some published photographs.[1,2] Although pterygium formation is commonly associated with nail lichen planus and scleroderma, we did not find other published reports of pterygia in acro-osteolysis.

We suggest that nail changes may provide a useful diagnostic clue to underlying bone resorption. Although the pterygium formation in our case was unique, findings of brachyonychia with or without associated nail changes should alert the clinician to the possibility of acro-osteolysis. Correlation of nail changes to etiologic subtype could assist with future classifications of this rare and poorly understood condition.

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