

## A Patient with Progressive Shortening of the Fingers

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Hajdu-Cheney syndrome is an autosomal dominant acroosteolytic syndrome, in which clinical and radiological abnormalities develop at different stages of life.

A 43-year-old woman was seen at our rheumatology clinic for progressive shortening of her terminal phalanges. Radiographs showed acroosteolysis and classic midphalangeal band-like resorption of the terminal phalanges affecting multiple digits of both hands and feet as shown in Figures 1 and 2. The differential diagnosis for these radiological changes includes severe Raynaud's phenomenon, polyvinyl chloride exposure, thermal injury (frostbite, burns), snake or scorpion bites, hyperparathyroidism, trauma, the acroosteolytic syndromes (mandibuloacral, Hajdu-Cheney or Giacciai syndromes), Rothmund's syndrome (autosomal recessive with poikiloderma, cataracts, saddle nose, sparse hair/eyelashes, small stature, skeletal/dental abnormalities, telangiectasia, hypogonadism), or Wormian bone disorders (e.g., osteogenesis imperfecta, progeria), which can be distinguished by clinical features<sup>1</sup>.

Our patient had features of Hajdu-Cheney syndrome, an autosomal dominant disorder characterized by short stature, distinctive craniofacial changes (frontal bossing, broad nose with flared nostrils, receding chin, bushy eyebrows, coarse thick hair and low set ears), visceral abnormalities and slowly progressive acroosteolysis of the distal phalanges. The full phenotype is rarely, if ever, present in childhood, and different clinical and radiological abnormalities develop at different stages in the lifespan and often progress with age. However, acroosteolysis and concomitant digit abnormalities are present in most individuals with Hajdu-Cheney syndrome and thus, radiographic findings make the diagnosis possible before clinical signs and symptoms are fully developed<sup>2</sup>. Her father and 1 of 2 sons had similar phenotypic features.

Due to deformities noted in radiological investigations, rheumatologists should be aware of such acroosteolytic syndromes.



Figure 1. Radiograph of both hands showing classic mid-phalangeal band-like resorption of terminal phalanges.



Figure 2. Radiograph of both feet showing classic mid-phalangeal band-like resorption of terminal phalanges.

#### REFERENCES

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