

Images in Rheumatology

# Solitary Band Acro-osteolysis in Hajdu-Cheney Syndrome

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Hajdu-Cheney syndrome (HCS) is a rare autosomal dominant manifestation of a genetic disorder caused by a mutation in the *NOTCH2* gene, resulting in acro-osteolysis and generalized osteoporosis, accompanied by many developmental skeletal disorders and multiple clinical and radiological manifestations.<sup>1</sup>

Radiographs of the hands of an 11-year-old boy with shorter left thumb and lower back pain show solitary band acro-osteolysis and shortening of the distal phalange of the left thumb (Figure 1A, with Figure 1B showing enlarged images of both thumbs). Moreover, radiographs of the spine and skull showing (C) multiple compression fractures of thoracolumbar spine and (D) multiple wormian bones. (E) 13-month follow-up showing progressive acro-osteolysis of the distal phalange of the left thumb.



*Figure 1.* (A) Solitary band acro-osteolysis and shortening of the distal phalange of the left thumb. (B) Enlarged images of both thumbs. Radiographs of the spine and skull showing (C) multiple compression fractures of thoracolumbar spine and (D) multiple wormian bones. (E) 13-month follow-up showing progressive acro-osteolysis of the distal phalange of the left thumb.

showed multiple compression fractures of thoracolumbar spine (Figure 1C) and multiple wormian bones (Figure 1D). All serological tests, including antinuclear antibody and rheumatoid factor levels, were within normal limits. Then, molecular analysis confirmed the diagnosis of HCS with the identification of a mutation in the *NOTCH2* gene. Biphosphonate therapy was given, and 13 months later, a follow-up radiograph showed progressive acroosteolysis of the distal phalange of the left thumb (Figure 1E).

Notably, although band acro-osteolysis is one of the typical radiological features of HCS, solitary band acro-osteolysis of the distal phalange of the left thumb is very rare. Thus, the differential diagnosis for the solitary band acro-osteolysis should be consid-

ered, including in systemic sclerosis, sarcoidosis, polyvinyl chloride exposure, and thermal injury (eg, frostbite, burns).<sup>2</sup> Further, aside from band acro-osteolysis appearing as a central resorptive band in the distal phalanx shaft with an intact terminal tuft and base, acro-osteolysis had the terminal tuft type, which exhibits distal to proximal tuft resorption and is seen more commonly in rheumatic diseases.

## REFERENCES

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