Case Report
Open Access
CrossMark

Familial hereditary acro-osteolysis with recurrent digital ulceration and autoamputation: a two-generation case report highlighting successful limb salvage via tibial transverse transport

Abstract

Acro-osteolysis—the progressive resorption of distal phalangeal bone—presents a complex diagnostic challenge, particularly in paediatric patients where genetic etiology is far more likely than acquired systemic causes. This case report documents an 11-year-old girl with a persistent, recalcitrant plantar ulcer of her right great toe and multifocal acro-osteolysis. Her father exhibited an identical clinical progression that led to bilateral autoamputation of his forefeet. Comprehensive diagnostic evaluations—including multi-modal skeletal imaging, CT angiography, and extensive autoimmune, vasculitic, and thrombophilia laboratory panels—effectively excluded classic Hajdu-Cheney syndrome, systemic vasculitis, hypercoagulable states, and length-dependent sensory neuropathies. To achieve complete wound healing and avoid progressive tissue loss, the patient underwent Tibial Transverse Transport (TTT) using the Ilizarov method. Mechanical distraction-osteogenesis promoted rapid neovascularization and tissue repair, resulting in complete, durable ulcer closure within two months. This report highlights a probable novel hereditary acro-osteolysis syndrome and demonstrates the effectiveness of TTT as a novel limb-salvage strategy for recalcitrant ischemic ulcers of non-traditional etiology.

Volume 18 Issue 5 - 2026

Prof. Emeritus Dr. Md. Mofakhkharul Bari,[1] Dr. AM Shayan R Bari,[1] Prof. Dr. Shahidul Islam,[1] Tahsin Ferdous,[2] Safwanul Aman[3]

[1]Department of Orthopaedic and Traumatology, Bari-Ilizarov Orthopaedic Centre, Bangladesh
[2]Department of Orthopaedic and Traumatology, Bangladesh Institute of Health Science (BIHS), Bangladesh
[3]Department of Orthopaedic and Traumatology, MRCP, Consultant, Omega Hospital, Bangladesh

Correspondence: Prof. Emeritus Dr. Md. Mofakhkharul Bari, Department of Orthopaedic and Traumatology, Bari-Ilizarov Orthopaedic Centre, Bangladesh, Email: bari.ilizarov31@gmail.com

Received: October 15, 2026 | **Published:** November 09, 2026

Introduction

Acro-osteolysis refers to the radiographic pattern of bone resorption affecting the distal phalanges of the hands and feet. While adult presentations are frequently secondary to acquired systemic conditions—such as scleroderma, occupational exposure to vinyl chloride, or severe thermal injury—pediatric acro-osteolysis strongly suggests an underlying genetic disorder.

Among the established hereditary causes, Hajdu-Cheney syndrome (HCS) is a key differential. Caused by truncating mutations in exon 34 of the NOTCH2 gene, HCS typically presents with band-like acro-osteolysis, severe osteoporosis, short stature, distinctive craniofacial features, and nearly pathognomonic skull abnormalities (including sutural wormian bones and a distorted sella turcica). Other primary differential considerations include Hereditary Sensory and Autonomic Neuropathies (HSAN), where impaired pain sensation leads to repetitive microtrauma, chronic infection, mutilating acro-osteolysis, and eventual autoamputation in a characteristic length-dependent distribution.[1,2]

In this report, we describe an 11-year-old female presenting with a chronic plantar toe ulcer and multifocal acro-osteolysis, with a clear vertical transmission pattern from her father, who suffered autoamputation of both forefeet. We detail the rigorous diagnostic steps taken to rule out known conditions and demonstrate the novel application of Tibial Transverse Transport (TTT)—a distraction-osteogenesis technique classically reserved for diabetic vascular ulcers—to successfully achieve limb salvage.

Case presentation

Patient profile & clinical history

An 11-year-old girl presented to the Department of Orthopaedics at BIHS General Hospital, Dhaka, Bangladesh, with a persistent, non-healing ulcer on the plantar surface of her right great toe. The lesion had been present for around the last 5 years and had failed to respond to conventional wound management and topical therapies. She had no prior history of diabetes mellitus, major trauma, fever, or systemic illness, although her medical history was notable for lichen planus.



Figure 1 Non healing Ulcer over the planter aspect of the great toe.

Physical examination revealed a chronic plantar ulcer with surrounding dusky discoloration but no active purulent discharge. Neurological examination demonstrated an intriguing non-length-dependent pattern: she had gradually progressive sensory loss over her right index finger, whereas tactile and pain sensation across both lower extremities—including the ulcerated foot itself—remained fully intact. Distal peripheral pulses were strong and symmetrical bilaterally.

©2026 Bari et al. This is an open access article distributed under the terms of the Creative Commons Attribution License , which permits unrestricted use, distribution, and build upon your work non-commercially.

Familial inheritance pattern

A detailed family history revealed a striking dominant pattern. The patient's father experienced an almost identical clinical disease course beginning in his early life. His condition started with recurrent, slow-healing distal extremity ulcers and progressive digit resorption, ultimately culminating in complete bilateral autoamputation at the forefoot level. On examination, his lower limbs terminated in smooth, rounded, bulbous stumps.[2,3,8]



Figure 2 Auto amputation of the forefoot due to Acro-osteolysis (Father of the patient).

He also exhibited multiple discrete, well-healed hyperpigmented scars and active superficial ulcers scattered along both shins and knees, separate from the terminal stumps, as well as distinct scarred lesions on his forearms. His sensory loss was strictly localized to the amputation stumps themselves—while sensation over his proximal legs remained completely preserved. Patient's 2-year-old younger brother was clinically healthy with no skin or skeletal abnormalities.[1,2]

Diagnostic work-up

To establish a definitive diagnosis, a systematic multidisciplinary evaluation was performed:

Radiological & vascular studies

Plain Radiography (Feet & Hands): Dorsoplantar radiographs of the feet demonstrated pronounced multifocal acro-osteolysis with extensive osteolysis of the phalanges of the 5th toes, distal phalanx of the 4th toe, and a lucent destructive lesion at the interphalangeal joint of the great toe. Hand radiographs revealed localized acro-osteolysis restricted to the distal phalanx of the right index finger.

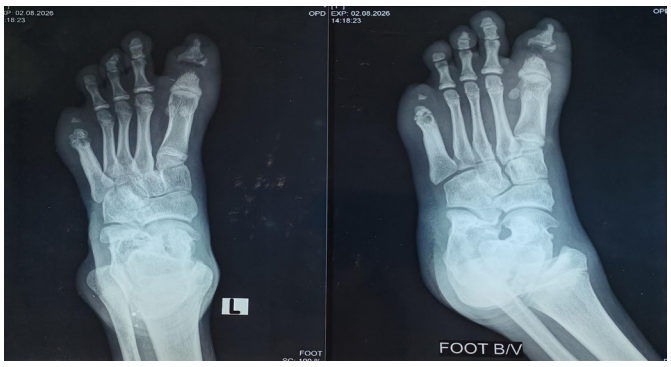


Figure 3



Figure 4

Figure 3 & 4 Acro-osteolysis with extensive osteolysis of the phalanges of the 5th toes, distal phalanx of the 4th toe, and a lucent destructive lesion at the interphalangeal joint of the great toe. Hyper calcification of the Calcaneum.

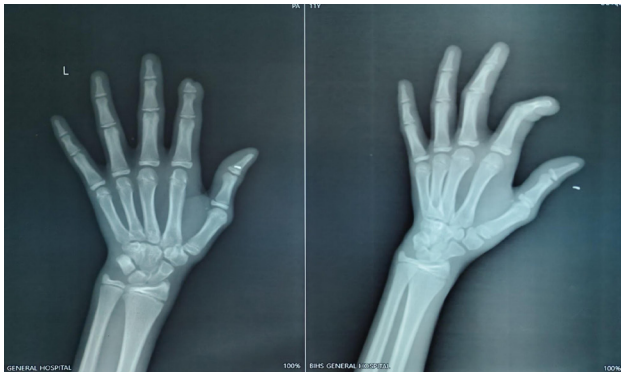


Figure 5



Figure 6

Figure 5 & 6 Acro osteolysis of the distal phalanx of the left index finger, with normal finding in the right hand

Skull radiography: Posteroanterior and lateral skull radiographs were completely normal. There was no evidence of sutural diastasis, wormian (sutural) bones, or sella turcica enlargement—effectively excluding classic Hajdu-Cheney syndrome.[1]

Citation: Bari EM, Bari AMSR, Islam S, et al. Familial hereditary acro-osteolysis with recurrent digital ulceration and autoamputation: a two-generation case report highlighting successful limb salvage via tibial transverse transports. *MOJ Orthop Rheumatol.* 2026;18(5):187–191. DOI: 10.15406/mojor.2026.18.00764



Figure 7

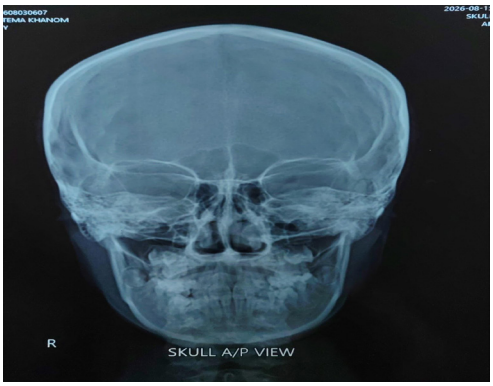


Figure 8

Figure 8 & 9 Normal skull x-ray findings in both view.

CT peripheral angiography: Contrast-enhanced CT angiography confirmed complete patency of the lower limb arterial tree from the aortoiliac bifurcation down to the pedal arches bilaterally. No arterial stenosis, occlusion, or vasculitic wall thickening was identified. Incidental increased distal pedal vascularity and a dilated great saphenous vein were noted on the left side.[3]

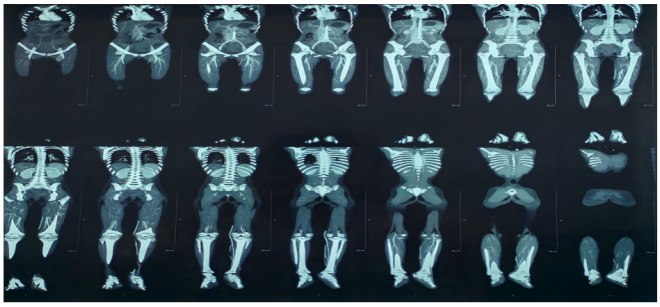


Figure 9

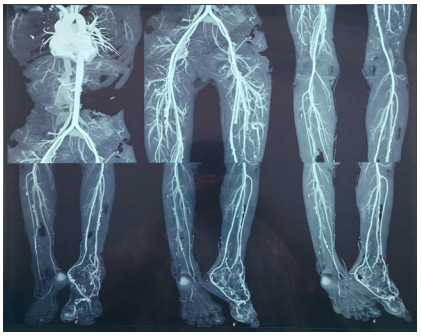


Figure 10

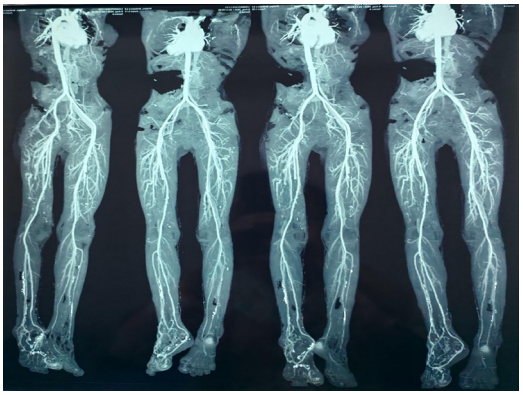


Figure 11

Figure 9-11 No arterial stenosis, occlusion, or vasculitic wall thickening was identified. Incidental increased distal pedal vascularity and a dilated great saphenous vein were noted on the left side.

Laboratory & serological findings

A comprehensive blood panel was conducted to screen for autoimmune, vasculitic, and hereditary hypercoagulable states. All key laboratory findings were within normal ranges, as detailed in Table 1 below:

Table 1

Diagnostic test	Patient result	Clinical interpretation
Anti-Phospholipid IgG	2.0 U/mL	Negative (Excludes APS)
Anti-Phospholipid IgM	2.5 U/mL	Negative (Excludes APS)
P-ANCA (anti-MPO)	2.52 AU/mL	Negative (Excludes ANCA vasculitis)
C-ANCA (anti-PR3)	< 2.00 AU/mL	Negative (Excludes Wegener's/GPA)
ENA Profile (dsDNA, Sm, RNP, Scl-70, Jo-1)	Negative Panel	No connective tissue disease / scleroderma
Antithrombin III Level	140%	Normal functional capacity
Protein C & Protein S Activity	125% / 87.6%	Normal (Excludes thrombophilia)

Surgical management: Tibial Transverse Transport (TTT)

Because the plantar ulcer proved entirely refractory to standard conservative care and local debridement, and given the high risk of progressive distal autoamputation seen in her father, the surgical team elected to perform Tibial Transverse Transport (TTT) using the Ilizarov method.

TTT utilizes the biological principle of distraction osteogenesis developed by Gavriil Ilizarov. Controlled, slow mechanical distraction of a corticotomized bone segment creates local tension-stress, triggering robust expression of physiological growth factors (such as HIF-1α and VEGF). This process induces microvascular neovascularization and dramatic perfusion enhancement in the distal microcirculatory bed, accelerating tissue repair even in angiographically patent limbs.[4–6]

Citation: Bari EM, Bari AMSR, Islam S, et al. Familial hereditary acro-osteolysis with recurrent digital ulceration and autoamputation: a two-generation case report highlighting successful limb salvage via tibial transverse transports. *MOJ Orthop Rheumatol.* 2026;18(5):187–191. DOI: 10.15406/mojor.2026.18.00764

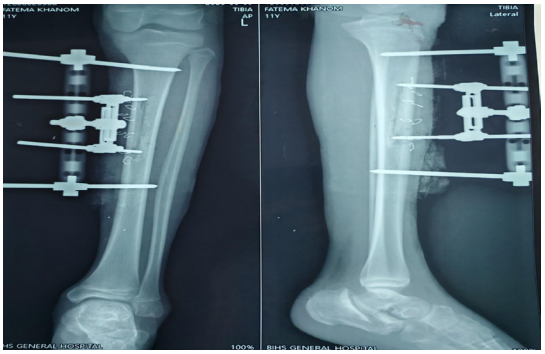


Figure 12 X-ray shows Tibial Transverse transport by using the Ilizarov method.

Surgical procedure & treatment protocol

Corticotomy & frame mounting: Under spinal regional anaesthesia (subarachnoid block), a rectangular medial tibial bone flap was created via a precise corticotomy. A device composed of two 4.5 mm Schanz screws and two 2.5 mm Schanz screws was set up with a dynamic bar and a static bar. The dynamic bar, with two 2.5 mm Schanz screws, was placed over the corticotomy site (single-cortex penetration), and the 4.5 mm Schanz screws stabilised the frame through dual -cortex penetration into the tibia.[7,8]

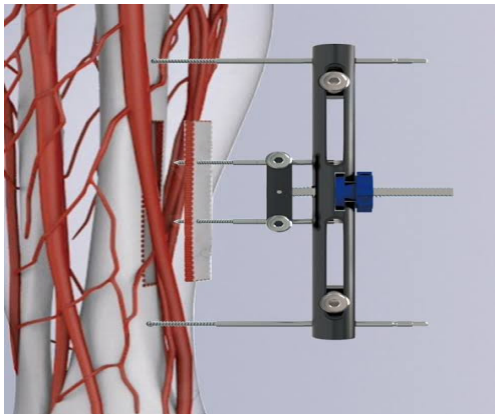


Figure 13 New model TTT device established on Ilizarov method.

i. **Pre-Distraction Latency:** An 8-day initial post-operative latency period was allowed for initial callus formation.

ii. **Active distraction phase:** Distraction was conducted at a rate of 1.0 mm/day (divided into four daily increments) for 10 consecutive days. Active lower-limb weight-bearing and joint range-of-motion exercises were encouraged throughout to stimulate mechanical loading.[4,7]

iii. **Consolidation & frame removal:** The frame was maintained in static fixation for 42 days during consolidation. Complete frame removal was accomplished at 60 days post-surgery, followed by temporary protection in a below-knee posterior splint.[1,5,7]

Results & clinical outcome

The biological response to distraction was rapid and dramatic. Within 30 days of initiating TTT, the chronic plantar ulcer demonstrated robust granulation tissue and significant perimeter contraction. By the time of frame removal at day 60, complete epithelial healing of the plantar toe ulcer was achieved.

At follow-up, the patient maintained full, painless weight-bearing ambulation without shoe modifications. There was no wound breakdown, recurrent ulceration, pin-site infection, or bone transport-related complication. This outcome successfully halted the cascade of tissue loss, achieving complete limb salvage and preserving extremity function.[3,4,7]



Figure 14 Non healing Ulcer cured by distraction angiogenesis by Ilizarov method.

Discussion

This case presents a unique clinical dilemma: a father-daughter pairing with autosomal dominant-appearing acro-osteolysis and recurrent digital ulceration that does not match any established genetic syndrome.

Hajdu-Cheney syndrome was initially considered due to autosomal dominant transmission and acro-osteolysis. However, the complete absence of sutural wormian bones, normal skull architecture, normal stature, and lack of characteristic dysmorphic facies strongly argue against HCS. Similarly, while Hereditary Sensory and Autonomic Neuropathy (HSAN) leads to autoamputation, classic HSAN is length-dependent—affecting distal feet sensation first. In our patient, foot sensation was fully intact while sensory loss appeared in an isolated hand digit, pointing away from standard HSAN types.[1,3]



Figure 15 Family history of Acro osteolysis in both daughter and father.

From a therapeutic standpoint, Tibial Transverse Transport proved to be a powerful surgical intervention. While TTT is primarily utilized in diabetic microvascular disease, its fundamental action—mechanically induced distraction angiogenesis—functions independently of the underlying pathology. By stimulating capillary sprouting and local tissue regeneration, TTT provided the necessary microcirculatory boost to heal a recalcitrant ulcer in an angiographically patent limb.[7,8]

Citation: Bari EM, Bari AMSR, Islam S, et al. Familial hereditary acro-osteolysis with recurrent digital ulceration and autoamputation: a two-generation case report highlighting successful limb salvage via tibial transverse transports. *MOJ Orthop Rheumatol.* 2026;18(5):187–191. DOI: 10.15406/mojor.2026.18.00764

Conclusion

We report a novel two-generation family affected by hereditary acro-osteolysis and recurrent ulceration resulting in autoamputation. Systematic investigation excluded Hajdu-Cheney syndrome, systemic vasculitis, hypercoagulability, and standard length-dependent sensory neuropathies. Tibial Transverse Transport achieved rapid, complete, and durable healing of a recalcitrant plantar ulcer, demonstrating that Ilizarov-based distraction osteogenesis is an effective limb-salvage option for non-traditional, complex ischemic ulcers.[8]

Acknowledgements

None.

Conflicts of interest

The authors declare that there are no conflicts of interest.

References

1. Cortés-Martín J, Díaz-Rodríguez L, Piqueras-Sola B, et al. Hajdu–Cheney Syndrome: A Systematic Review of the Literature. *Int J Environ Res Public Health*. 2020;17(17):6174.
2. Hajdu N, Kauntze R. Hajdu-Cheney syndrome: a review. *Orphanet J Rare Dis*. 2014;9:200.
3. Limenis E, Stimec J, Kannu P, et al. Lost bones: differential diagnosis of acro-osteolysis seen by the pediatric rheumatologist. *Pediatr Rheumatol Online J*. 2021;19(1):113.
4. Bari MM. A Colour Atlas of Limb Lengthening, Surgical Reconstruction and Deformity Correction by Ilizarov Technique. 1st ed. Dhaka, Bangladesh: Bari-Ilizarov Orthopaedic Centre; 2013.
5. Ilizarov GA. Basic principles of transosseous compression and distraction osteosynthesis. *Orthop Travmatol Protez*. 1971;32(11):7–15.
6. Liu J, Huang X, Su H, et al. Tibial cortex transverse transport facilitates severe diabetic foot wound healing via hif-1α-induced angiogenesis. *J Inflamm Res*. 2024;17:2681–2696.
7. Ilizarov GA. The tension-stress effect on the genesis and growth of tissues. Part I: the influence of stability of fixation and soft-tissue preservation. *Clin Orthop Relat Res*. 1989;(238):249–281.
8. Qin W, Liu K, Su H, et al. Tibial cortex transverse transport promotes ischemic diabetic foot ulcer healing via enhanced angiogenesis and inflammation modulation. *Eur J Med Res*. 2024;29(1):155.