Seeing Is Not Always Believing: Congenital Insensitivity to Pain With Anhidrosis Mimicking Leprosy

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A 30-year-old woman presented with progressive history of acroamputations of both hands and feet since early childhood. Because of her insensitivity to pain, she had repeated trauma, recurrent burns, and self-mutilating behavior, ie, tongue biting, self–tooth extraction, and frequent scratching of the nasal septum, eventually leading to its complete erosion. The patient had recurrent episodes of unexplained fever, heat intolerance, especially during summer, anhidrosis, and hypolacrimia. Her parents were related by first-degree consanguinity. She had 4 living siblings, with similar complaints in her elder sister. There was history of infant death due to recurrent high-grade fever in 6 of her siblings. The patient had a depressed nose, corneal opacity in the left eye, an eroded tip of the tongue, multiple missing teeth (Figure 1, A and B), and acroamputations in both hands and feet with plantar ulcers (Figure 2, A and B). She had sluggishly reactive pupils, diminished facial and oral

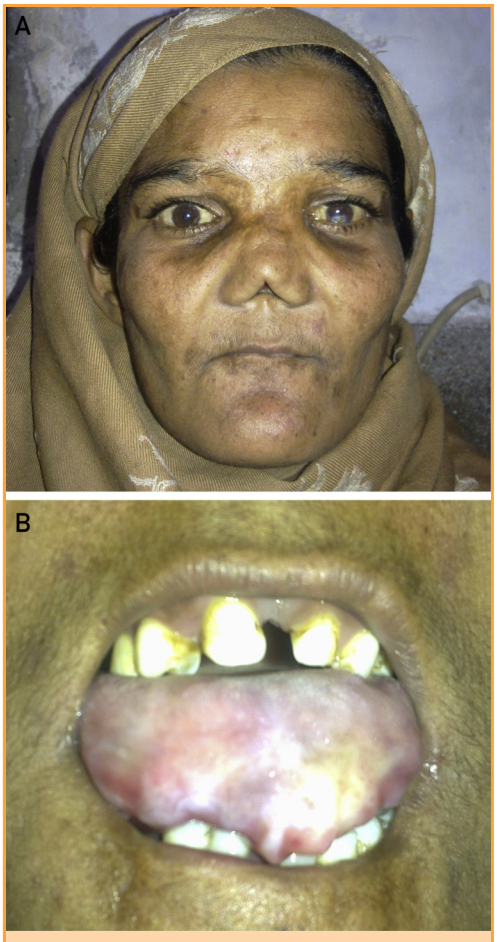

FIGURE 1. A, Depressed nose with eroded nasal septum, corneal opacity, flattened lips, and mandibular prominence. B, Multiple missing teeth, loss of tip of tongue, and fungiform papillae.

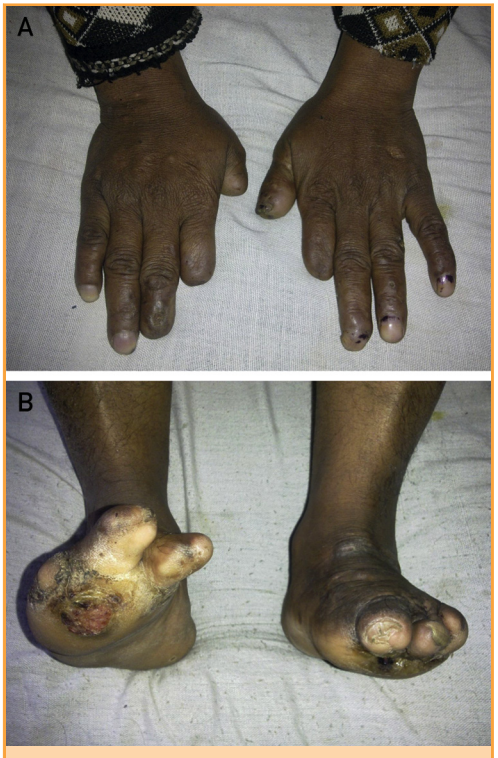

FIGURE 2. A, Acroamputations of both hands. B, Acroamputations of both feet and plantar ulcers.

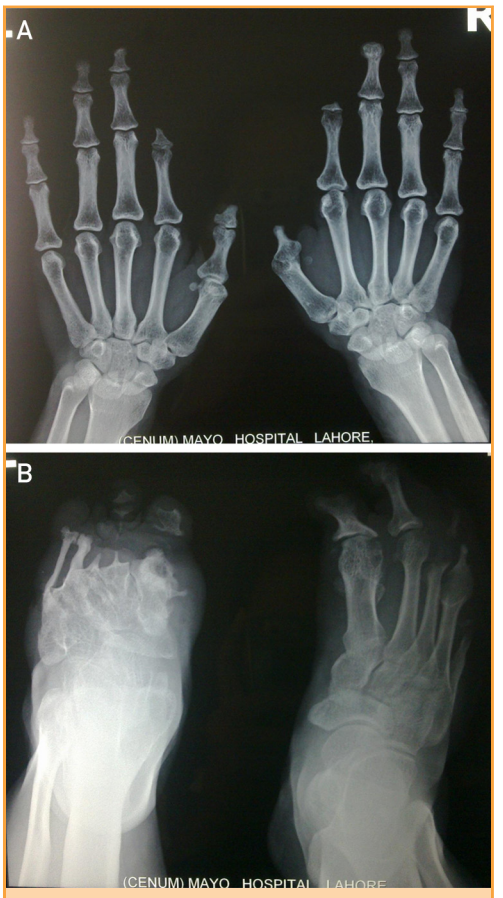

FIGURE 3. A, Radiograph of both hands showing loss of multiple phalanges. B, Radiograph of both feet showing osteolytic defects, marked porosis, and bony destructions.

sensation, and loss of pain and temperature sensation. Autonomic dysfunction was documented by abnormal results of isometric handgrip, cold pressor, starch-iodine sweat, Schirmer, and skin wrinkle testing and abnormal 30:15 ratio and E/I ratio on electrocardiography. Radiographs of the hands and feet revealed loss of multiple phalanges and osteolytic defects (Figure 3, A and B). Nerve conduction studies found normal motor function with absent sensory nerve action potentials. A smear test was negative for *Mycobacterium leprae*.

Congenital insensitivity to pain with anhidrosis is an autosomal recessive disorder caused by lack of small myelinated and unmyelinated fibers of the peripheral nerves causing absence of pain and temperature sensation. Symptoms begin early in infancy and lead to multiple injuries, fractures, osteomyelitis, self-mutilation, and acroamputation.[1] Defects in thermoregulation and anhidrosis lead to episodic hyperthermia, which may be associated with seizures. Up to 20% of patients die from hyperpyrexia by age 3. Mild to moderate mental retardation is often present. Tearing is preserved, and fungiform papillae are present on the tongue.[2]

1. Axelrod FB, Gold-von Simson G. Hereditary sensory and autonomic neuropathies: types II, III and IV. *Orphanet J Rare Dis*. 2007; 2:39.
2. Rosemberg S, Marie SK, Kliemann S. Congenital insensitivity to pain with anhidrosis (hereditary sensory and autonomic neuropathy type IV). *Pediatr Neurol*. 1994;11(1):50-56.

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