Hereditary Sensory and Autonomic Neuropathy Type IV

A 10-year-old boy presented with history of non-healing ulcers at base of toes with subsequent loss of toes over the last three years (*Fig. 1*). There were no draining sinuses or discharge of grains. Tone and power in limbs were normal. Sensory examination revealed generalized loss of pain and temperature sensations. Random blood sugar was normal. Nerve conduction studies showed sensorineural peripheral neuropathy. Tactile sensibility, vibration and position sense, and stereognosis were present. He had anhidrosis, habitual constipation and a low capacity overactive bladder on urodynamic study. His Intelligence Quotient was 75-80. There was no family history of similar illness. A diagnosis of Hereditary Sensory and Autonomic Neuropathy type IV (HSAN IV) was made. With bed rest, antibiotics and Eusol soaks the ulcers healed after four weeks (Fig. 2). To prevent recurrence, modified footwear for constant use was made for the child.

HSAN IV is an autosomal recessive disorder characterized by congenital insensitivity to pain, anhidrosis, and mental retardation. Affected infants are usually hypotonic and have repeated episodes of fever due to inability to sweat. Tendon reflexes are absent or hyporeflexic. Self-inflicted injuries caused by pain insensitivity include ulcers of the fingers and toes, stress factures, self-mutilation of the tongue and Charcot joints. Constant vigilance is required to prevent injuries to the skin and bones with secondary infection.

Other types of HSAN such as congenital sensory neuropathy (HSAN II), familial dysautonomia (HSAN III), and congenital insensitivity to pain (HSAN V) may be confused with HSAN IV during infancy. However mental retardation and anhidrosis are not prominent in HSAN II and not present in HSAN V; and insensitivity to pain is not prominent in HSAN III. Anhidrotic ectodermal dysplasia is another hereditary disorder that shares features of unexplained fevers and anhidrosis; however nervous system is intact and sensation to pain is present.

Other rare conditions can present with non-healing ulcers and subsequent loss of digits. Loss of pain sensibility (in long

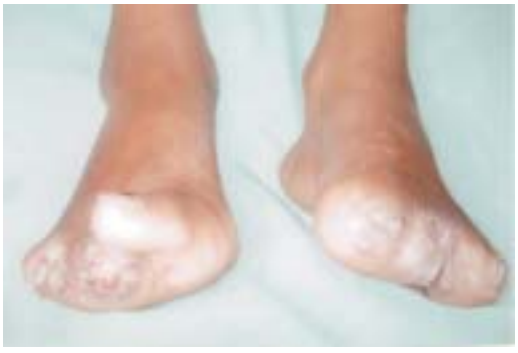

Fig. 1. Feet deformed due to loss of all toes except the right great toe. Chronic ulcers over right second and fourth metatarso-phalengeal joints with underlying bone exposed.

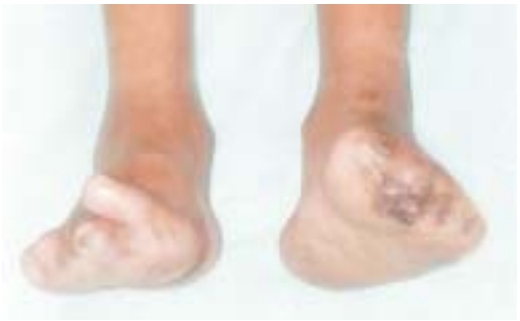

Fig. 2. Feet showing healed chronic ulcers after treatment.

standing juvenile diabetes with poor glycemic control, leprosy and syringomyelia), and self-mutilating behavior (in Lesch-Nyhan syndrome and severe mental retardation) can lead to injury, ulceration, infection, non-healing ulcers and subsequent loss of digits. Mycetoma of the foot due to chronic fungal or actinomycetes infection characterized by draining sinuses which discharge grains is another close differential diagnosis.

Acknowledgement

We thank our Dean, Dr. M.E. Yeolekar, for granting us permission to publish this clinical image.

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