Sensory loss with digital autoamputation: an approach to diagnosis

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Sensory loss with digital amputation always gives a first impression of leprosy in Asian countries, but other causes need to be fully investigated. Leprosy is a chronic disease of low infectivity and clinically manifests as a wide range of skin and nerve lesions, leading to permanent damage to the skin and nerves. Mosby's Medical Dictionary 2009 defines trophic ulcers as "a pressure ulcer caused by external trauma to a part of the body that is in poor condition because of disease, vascular insufficiency or loss of afferent nerve fibres". Besides leprosy, other causes of trophic ulcers should be borne in mind. A rare case of spina bifida occulta and diastematomyelia with scoliosis, an important differential diagnosis of Hansen's disease, is presented here (Siddappa, 2013).

One of the oldest diseases, which finds its mention in the ayurvedic medical descriptions of Charaka, Sushruta and others, is leprosy (Muir, 1951). Leprosy is a debilitating illness caused by acid-fast bacilli (AFB) *Mycobacterium leprae* (*M. leprae*). Leprosy is a chronic disease of low infectivity and clinically manifests as a wide range of skin and nerve lesions, the latter sometimes causing more suffering due to permanent damage. The World Health Organization (WHO) Expert Committee on Leprosy in 2010 defined three main signs of leprosy (WHO, 2012):

1. Hypopigmented or erythematous skin lesion(s) with definite loss/impairment of sensation
2. Involvement of the peripheral nerves, as demonstrated by definite thickening with sensory impairment
3. Skin smear positive for AFB.

Its chronic, slow-growing nature and ability to invade the peripheral nervous system gives rise to neuropathy, hence the sensory loss. Further complications (e.g. amputation of digits and trophic ulceration) in the insensitive foot may follow the course if not timely intervened. India, being a developing country, is chronically plagued by a high number of leprosy cases. Trophic ulcers with sensory loss generally point towards Hansen's disease.

In this intriguing case, a young male presented with sensory loss, loss of digits and a trophic ulcer. Upon complete workup, a very close differential in such scenarios was brought to light. Spina bifida occulta is another cause that can manifest in a similar way and should be borne in mind when diagnosing.

Case report

A 15-year-old male Hindu student (non-alcoholic and non-smoker) presented to the dermatology department with chief complaints of painless autoamputation of the first and second toe of his left foot with a non-healing, slow-growing ulcer of 12 years' duration. The patient was supposedly asymptomatic when his parents first noticed repeated bleeding from his toes following trauma, but he did not cry.

Due to repeated trauma, a non-healing ulcer had developed on the great toe and second toe, which resulted in autoamputation of the great toe at the age of four and a half years old, and the second toe at the age of seven years old *[Figure 1]*. Despite taking aggressive treatment, there was no relief. Gradually, the ulcer spread to the third toe.

There was no history of urinary incontinence or retention, nocturnal enuresis, trauma or any other major illness or infection during his childhood. There was no relevant family history and birth history was normal. There was also no developmental delay of milestones and the patient's immunisation history was up-to-date.

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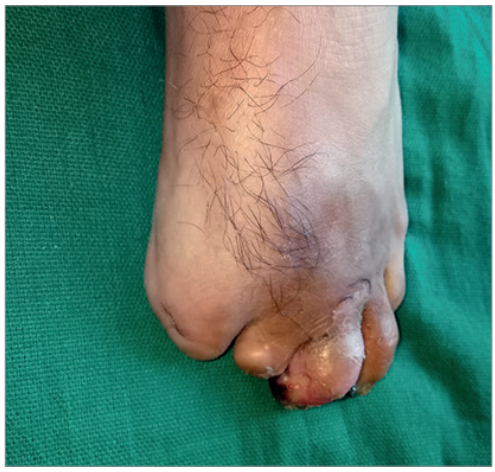

Figure 1. Right foot without great toe and second toe.

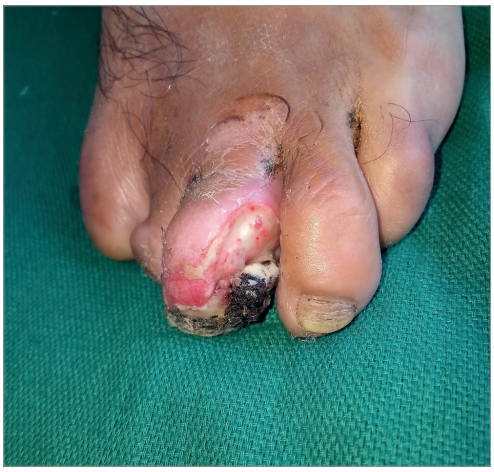

Figure 2. Non-healing ulcer (from above).

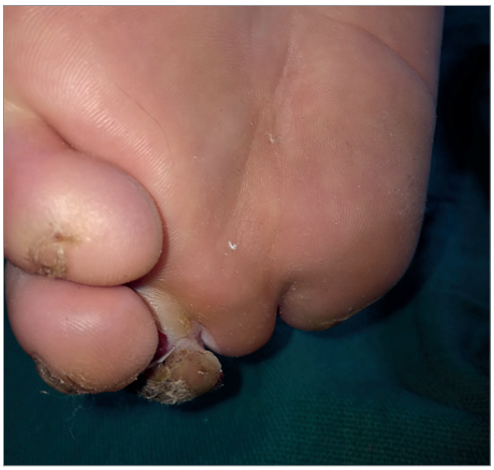

Figure 3. Non healing ulcer (from below).

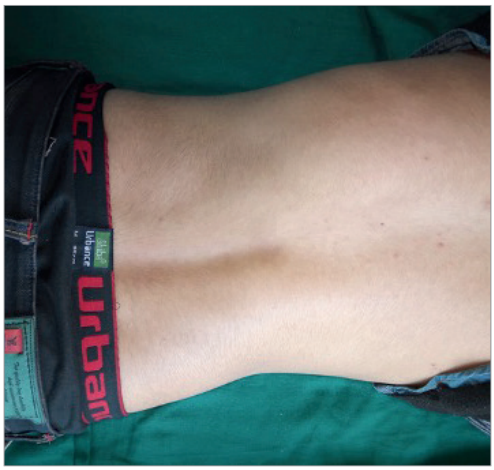

Figure 4. Scoliosis with slight convexity towards right.

The patient was of average build; his speech and gait were normal. All peripheral pulses were palpable. There was no sign of peripheral cyanosis. Localised examination revealed single moist ulcer (approx. 3 cm (length) x 2 cm (width), with irregular margin and tapering edges. The ulcer was not fixed to underlying structures and some greenish pus was noted at the erythematous base of the third toe, along with gangrenous changes. The surrounding skin appeared to be erythematous with some exfoliation.

First and second toes were amputated *[Figures 2 and 3]*. Pain sensation was absent in the left foot to the medial and lateral malleoli; all other sensations were intact. All the reflexes were present. Power was 5/5 in the left foot. No nerve was thickened. Mucosal and hair examinations were normal. Spine examination revealed scoliosis with convexity towards the right side *[Figure 4]*.

There was no tuft of hair or dimples in the sacral regions, and no bony abnormality was found on palpation. Systemic examination was normal. His routine blood investigations including vitamin B-12 level were normal and HbA_{1c} was within normal range. Colour Doppler did not reveal any vascular pathology. A punch biopsy taken from the site of the lesion was normal with no evidence of Hansen's disease and the patient was sent for a magnetic resonance imaging (MRI) of the whole spine, which revealed spina bifida occulta, diastematomyelia and scoliosis with low lying conus at the level of L4 with prominent central canal.

After ulcer debridement and proper cleansing, the patient was prescribed oral antibiotics and the ulcer was dressed with a silver antimicrobial barrier dressing containing nano-silver particles, but there was little relief. After 2 months, platelet-derived growth factor gel was tried; this gave partial relief to the patient. The patient was provided with leprosy shoes, advised to avoid strenuous activities and barefoot walks, and referred to the neurosurgery department for further management of spina bifida.

Table 1. Causes of trophic ulcers in foot.	
Neurogenic	Hansens disease, syringomyelia, pressure ulcer, paraplegics, diabetic neuropathy, spina bifida, alcoholic polyneuropathy
Vascular (arterial)	Poor arteria supply (e.g. arteriosclerosis, diabetic microangiopathy, peripheral vascular disease)
Vascular (venous)	Venous disease (e.g. venous stasis ulcer)
Systemic causes or malnutrition	Vitamin B12 deficiency, severe avitaminosis, ulcer over deposits of gout

Discussion

India is a tropical country heavily affected by leprosy and, as dermatologists, the first thing that always strikes us in a case of ulcer with sensory loss is leprosy. Cutaneous nerves are affected in all types of leprosy, leading to anaesthetic or hypoaesthetic lesions. Neural predilection of *M. leprae* is promoted by immune suppression and relatively low temperature of the peripheral nerves (Hastings et al, 1968), and is prone to trauma due to insensitivity, resulting in an ulcer. Infections, dry cracked and fissured skin (due to loss of sweating) or the weight of the patient themselves leads to development of trophic ulcers in leprosy, most commonly at the metatarsophalangeal joint (Hastings et al, 1968). In a person with normal sensations, fatigue or injury is sensed by the person and allows it to heal by taking rest, whereas in patients with sensory loss, the situation is not realised, hence with continuous use, vascular supply is compromised. This ultimately results in a trophic ulcer or amputation of digits.

Spina bifida (split spine) is a neurodevelopmental congenital disorder caused by incomplete closure of the embryonic neural tube (Fletcher and Brei, 2010). Some vertebrae overlying the spine are not fully formed and remain unfused during neurulation. It is one of the most common birth defects seen in the lumber and sacral region. There are various causes, such as folic acid deficiency during pregnancy, maternal diabetes and use of anticonvulsants (e.g. valproic acid) during pregnancy in the period of organogenesis.

Spina bifida is divided into two categories (Del Bigio, 2010):

1. Spina bifida occulta
2. Spina bifida cystica with meningocele, myelomeningocele or myelochisis.

Clinically, spina bifida cystica manifests at birth whereas spina bifida occulta is diagnosed incidentally (Waller et al, 2000). Patients may present with abnormalities like partial or complete loss of sensation, ulcers, club foot or hip dislocation, amputation of digits, bladder bowel incontinence, poor renal function, urinary tract infection, paralysis, scoliosis, pressure sores, meningitis, backache, dimpling of skin, tuft of hair in presacral area, hoarseness, apnoea attacks, low intelligent quotient and Arnold Chiari 2 malformation.

Spina bifida should be managed surgically within 48 hours of birth and a VP shunt conducted for cerebro spinal fluid outflow. Spina bifida occulta is managed as per symptoms. Dietry supplements of folic acid (whole grains, fortified, dried beans, green leafy vegetables) reduces the incidence (Centers for Disease Control, 2008).

In this case, the patient presented with a moist ulcer with auto-amputation of two toes with all intact sensations, except pain sensation in his left foot. There are various causes *[Table 1]* of trophic ulcers with sensory loss, but after excluding other possibilities, two differential diagnoses were considered: Hansen's Disease and spina bifida.

The patient was initially sent for a MRI of the whole spine since there was no thickened nerve with all intact sensations except pain, and then a biopsy was taken. MRI findings suggested spina bifida occulta with diastematomyelia (vertically bifid spine) with scoliosis that helped in clinching the diagnosis. Such trophic ulcers should be handled carefully with collaborative approach of neurosurgery and medicine departments.

From a dermatology point of view, there are various ways of managing a non-healing ulcer and oral vitamin C (Taylor et al, 1974) can help to speed up recovery. Various dressings that have been described are gauze and impregnated gauze, hydrocolloid, hydrogel, alginate, oxidised regenerated cellulose and collagen, silicone, foam, hydrofibres, hydroconductive transparent and cloth dressings (Taylor et al, 1974). Negative pressure dressings yield good results. Nano silver particle dressings, platelet-derived growth factor gel, platelet-rich plasma gel, aloe vera gel and allantoin are the upcoming modalities.

The patient was given platelet rich plasma gel that resulted in partial relief, probably because primary cause was still there.

Although the sensory loss is difficult to recover from, precautions can prevent grievous outcomes. Avoidance of trauma (no strenuous sports activities) and barefoot walking, wearing supportive shoes in the correct size and precise wound care with antibiotic coverage can drastically improve outcomes and enhance the patient's quality of life.

Conclusion

Spina bifida occulta is an important differential diagnosis of Hansen's disease. Patients with chronic slow-spreading trophic ulcers with loss of sensation should be thoroughly examined for neurologic or musculoskeletal abnormalities. WME

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