

## Case report

# A case of hereditary sensory and autonomic neuropathy (HSAN) type II

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## Summary

We describe a case of hereditary sensory and autonomic neuropathy (HSAN) type II in a child with a penetrating foot ulcer, acral sensory impairment, and anhidrosis. This is the first documentation of HSAN in Sri Lanka.

## Introduction

Hereditary sensory neuropathies were recognised to be associated with autonomic manifestations, and they were subsequently termed HSAN (1). Classification of HSAN (Table) is based on the pattern of inheritance, natural history and histopathological features on nerve biopsy (1,2).

HSAN type II is an autosomal recessive condition presenting in early life with a pansensory neuropathy. Static and slowly progressive subtypes have been described (3). As most of the reported cases refer to young patients, it can be inferred that the life span of these patients is reduced.

## Case report

An 8-year old boy with a painless non-healing ulcer of the left foot of 6 months' duration. He was the second of two children from a non-consanguineous marriage. His birth history and developmental milestones were normal. At 5 years of age, his parents noticed recurrent ulcers and callosities in his feet, from which he did not complain. He was also noted not to sweat as much as his sibling. There was no history of self-mutilation or recurrent pyrexia. His parents and elder brother were healthy.

Mental functions, cranial nerves and motor system were normal. All tendon reflexes were absent. The skin over the limbs was dry, hyperkeratotic, and fissured. A penetrating ulcer was noted at the left heel. All modalities of sensation were absent in the distal extremities but normal on the trunk and face. The peripheral nerves were not thickened. There was no postural drop in blood pressure. Examination of the patient's parents and elder brother was normal.

Table. Classification of HSAN

HSAN type I	- Autosomal dominant sensory neuropathy
HSAN type II	- Autosomal recessive sensory neuropathy
HSAN type III	- Familial dysautonomia (Riley-Day syndrome)
HSAN type IV	- Hereditary anhidrotic sensory neuropathy
HSAN type V	- Congenital sensory neuropathy with selective loss of small myelinated fibres.

The complete blood count, ESR, serum proteins, urinalysis, blood urea, serum electrolytes, and chest radiograph were all normal. VDRL was negative. Motor nerve conduction studies of median, ulnar, common peroneal and posterior tibial nerves were within normal limits. The sensory action potentials of the median, ulnar, and sural nerves were not recordable, consistent with a pure sensory neuropathy. Electron microscopy of the sural nerve revealed a striking absence of myelinated fibres, supporting the diagnosis of HSAN type II. Anhidrosis was demonstrated over the region of sensory loss. Cardiovascular changes during Valsalva manoeuvre and the tilt test were normal. Lacrimation was normal, as well as the pupillary response to 0.125% pilocarpine and 0.1% adrenaline eye drops.

## Discussion

HSAN type II presents in infancy or childhood with impairment of all modalities of sensation affecting the limbs more than the trunk or face (4). It may present with recurrent acral skin ulcers, painless long bone fractures and Charcot's neuropathic joints. Tendon reflexes are usually lost (4).

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Autonomic disturbances are relatively mild in HSAN type II. The main autonomic abnormality is anhidrosis in the extremities; erectile dysfunction and bladder disturbances may also occur (3). Cardiovascular autonomic disturbances have not been reported. Nerve conduction studies show the absence of sensory action potentials, but the motor are normal. Reduced cutaneous nerve calibre results in difficulty of finding the sural nerve at biopsy. Nerve biopsy shows a striking loss of myelinated nerve fibres and relative preservation of unmyelinated fibres (4).

HSAN type I distinguished by its dominant inheritance and adult age of onset. Other clinical features may be similar to type II (3). Nerve biopsy reveals loss of myelinated fibres, especially of small diameter, and unmyelinated fibres (4,5). HSAN type III is dominated by autonomic manifestations and is a clinically distinct condition (6). HSAN type IV presents in infancy with recurrent pyrexia, failure to thrive and marked anhidrosis. Self-mutilation is a feature and mental retardation is common. Although pain and temperature sensation is lost touch is preserved (7). Sensory nerve conduction is usually normal and nerve biopsy shows a virtual absence of unmyelinated axons. HSAN type V is characterised by defective pain sensation whereas other sensory modalities are normal or only minimally impaired. Only a detailed analysis of nerve biopsy shows an absence of small myelinated nerve fibres (1).

There is no definitive treatment available for these patients. They should avoid burns and trauma to the limbs, while local infection and injuries will require aggressive treatment. Genetic counselling is required in this recessively

inherited condition, although sporadic cases have also been reported (3). Thus early diagnosis of HSAN, and early implementation of preventive measures are necessary to reduce morbidity in these patients.

## References

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## Ageing in the millennium

What then will be the role of the doctor and prescriber in this ageing society in the future? While advances will reduce the proportion of disability due to disease, it is likely that there will be a significant (and probably increasing) group of vulnerable elderly people who are dependent, especially on their doctors. These people may not be 'curable', but their lives will be enriched immensely by an empathetic relationship with their doctor which allows them to maintain their autonomy and dignity, have access to wisely applied scientific advances and retain their position as valued members of society.

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