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CASE REPORTS

HEREDITARY SENSORY NEUROPATHY (TYPE II) (Congenital absence of pain)

K Pavithran, P Ramachandran Nair, P S Mathew, T P Thankappan and Cheriyan Mathew

A 3-year-old girl having hereditary sensory neuropathy, type II, manifesting as congenital absence of pain sensation and trophic changes in the skin is reported. Conduction velocity study of the nerves of the hands and feet revealed a normal motor conduction, but sensory action potentials were absent. Differentiating features of this disease from other types of hereditary sensory neuropathies and congenital indifference to pain are briefly discussed.

Key words : Hereditary sensory neuropathy, Congenital absence of pain, Acrodystrophic neuropathy, Progressive sensory neuropathy.

There are a number of inherited diseases characterised by neuronal atrophy and degeneration predominently affecting the peripheral sensory neurons. The names given to these diseases were highly confusing. Recently, all of them have been grouped under the title 'Hereditary Sensory Neuropathy' (HSN) which is most appealing because it is simple, understandable and correct.[1] Hereditary sensory neuropathies have been classified into four types (Type I, II, III and IV) on the basis of clinical signs and symptoms, the type of inheritance, the type of neurons affected and the digital and cutaneous nerve action potential patterns. We are reporting a case of HSN type II in a 3-year-old female child who presented with congenital absence of the pain sensation and trophic changes in the skin.

Case Report

A girl, aged 3 years, born to non-consanguinous parents, was seen with recurrent ulcerations of the fingers and toes since two years. Mother of the patient noticed that the child was not able to appreciate the sensations of touch, pain and heat. The rats in her house used to bite her fingers and toes while she was sleeping, without causing any pain or disturbance to her sleep. She had difficulty in walking and picking up small objects. The child used to get repeated infections, whitlows and ulcers of the toes and fingers. Dis-articulation of the terminal phalanx of the little toe of the left foot was done at a local hospital for chronic infection in that area. The little toe of the ohter foot was lost spontaneously as a result of some chronic ulceration.

The child was born after a full-term normal delivery and all the developmental mile-stones were normal except for the delayed speech. She was the first of the two siblings and none in her family suffered from a similar disease. There was no history of any disease in her mother during pregnancy.

General physical examination did not reveal any abnormality. There were multiple scars and callosities on the extremities. All the fingers showed ulcers at their tips (Fig). The little toe of

From the Departments of Skin & VD, and Neurology, Medical College, Kottayam, India.

Address correspondence to : Dr. K. Pavithran.

the right foot and the terminal phalanx of the little toe of the left foot were missing. There was no perforating ulcer on the sole of the foot. The peripheral arterial pulsations were normal.

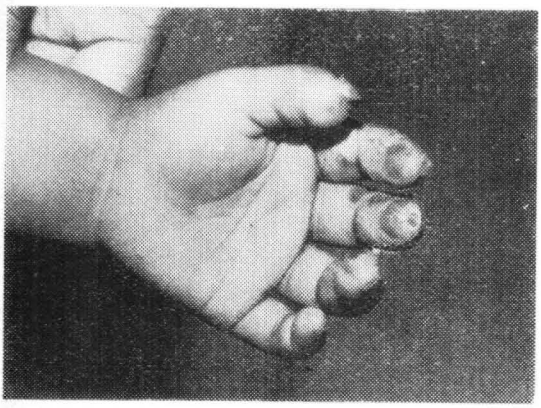

Fig. The finger-tips bitten by rats during sleep.

Higher functions of the nervous system were normal except the speech. She used to speak only mono-syllables and simple sentences. The cranial nerves were normal. Corneal sensation and the cutaneous sensations on the face were intact. There was no wasting of muscles and their power and tone were normal. Tendon reflexes (ankle, knee, biceps and radial) on both the sides were absent. Superficial reflexes (abdominal and plantar) were also absent. There was incoordination of the lower limbs resulting in unsteadiness of the gait. Bladder and bowel controls were normal. Cutaneous sensations of touch, pain and temperature were impaired all over the body except the face. The sensory loss was more dense on the limbs than on the trunk and the sensation of touch was more affected than those of pain and temperature. There was no thickening of the peripheral nerves. The sense of position and the joint sensation were retained. There were no cerebellar signs. Romberg's sign was positive. Signs of meningeal irritation were absent. Lacrimation in both the eyes was seen when the patient used to cry. Skull and spine were normal. Systemic examination did not reveal any abnormality.

Routine investigations of the blood and urine were normal. Blood sugar (fasting) was 74 mg%, blood VDRL was non-reactive, serum uric acid 5.3 mg%, X-ray of the chest, skull and spine were also normal. X-rays of the hands and feet revealed that the little toe of the right foot and terminal phalanx of the little toe of the left foot were missing. Skin smear taken from the ear, did not show any acid-fast bacilli. Skin biopsy from the leg showed no abnormality. Intradermal pilocarpine test showed normal sweat response. Nerve conduction velocity study showed that sensory action potentials could not be elicited on the nerves of the hands and feet. Motor conduction and EMG were normal. Needle EMG showed no evidence of active denervation or myopathies. Motor conduction velocity of the left lateral popliteal nerve was 40 meters per second and that of the right median nerve was 52 meters per second. Nerve biopsy was not done since the consent was denied.

Comments

Highly confusing terminologies were used in the past to describe this group of diseases. The term hereditary sensory neuropathy (HSN)[2,3] is preferred now and it has been grouped into type I, II, III and IV.[1,4] Type-II HSN had been reported earlier as congenital sensory neuropathy,[5] progressive sensory neuropathy,[6] and acrodystrophic neuropathy.[7] The hall marks of HSN type II are autosomal recessive mode of inheritance, infantile onset and mutilating acropathy characterized by paronychia, whitlows and ulcers of the toes and fingers. All modalities of cutaneous and sometimes kinaesthetic sensations are lost all over the body, especially on the distal parts of all the four limbs. The sensations of touch and pressure are affected more severely than those of pain and temperature. The tendon reflexes are diminished or absent. The nerve action potentials of afferent fibres are undetectable in conduction

velocity studies. Complete absence of the myelinated fibres in the cutaneous nerves of distal parts of the limbs, is seen under electron microscopy. The present case has all the signs and symptoms as described for HSN type II. Though, the nerve biopsy could not be done, the conduction study revealed absence of sensory action potentials in the nerves of the hands and feet. Onset in early childhood also favours the diagnosis of HSN type II.

It is unlikely to be HSN type I because it usually manifests in the second or the third decade of life with autosomal dominant mode of inheritance. The lower limbs alone get affected and the sensations of pain and temperature are more grossly impaired than touch. The trophic ulcers are seen on the plantar surface rather than on the fingers and toes. There may be associated wasting of muscles also. The Achillis reflex alone is absent, the other deep tendon reflexes being uneffected. Sensory loss is less generalised and electron microscopy reveals a decrease in the unmyelinated fibres in the nerves. All these features of HSN type I were absent in our case. Cases reported as hereditary sensory radicular neuropathy by Denny-Brown[8] and ulcerative and mutilating acropathy by Thevenard[9] are examples of HSN type I. Normal lacrimation in the present case excludes the possibility of type III HSN which affects the peripheral autonomic neurons in addition to the sensory and motor neurons. Cases with type IV HSN have anhidrosis and mild mental retardation in addition to the insensitivity to pain. Our case had no mental retardation and the sweat response to intradermal pilocarpine was positive.

HSN type II must also be differentiated from the congenital indifference to pain which is characterised by the absence of normal appreciation and reaction to pain unassociated with other neurological deficits.[10,11] It manifests since birth and sensations other than pain are well appreciated. Sensory nerves in the skin are normal and there is no underlying abnormality of the nervous system. In the present case, the sensory action potentials of the digital cutaneous nerves were undetectable and in addition to pain, other sensations also were lost. All these features in our case exclude the possibility of congenital indifference to pain.

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