LETTER TO EDITOR (VIEWERS CHOICE)

CONGENITAL INSENSITIVITY TO PAIN WITH ANHIDROSIS

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A 2½ years old girl born of a third degree consanguineous marriage presented with a non healing ulcer on the left great toe since last 3 months in spite of having received treatment for the same at various hospitals. The parents had noticed high fever only during summer poorly responding to medications and also absence of sweating in the child. She also had global developmental delay. On examination she was irritable, pale and febrile. She had an ulcer with gangrenous changes on the left great toe with fissuring and surrounding hyperkeratotic changes. There was also a small chronic ulcer on the right knee and some fissuring noticed at the lower lips. On central nervous system (CNS) examination, the child had preserved reflexes with normal tone and power.On sensory system examination, her sense of touch was intact however she was unable to perceive painful stimuli. Hemogram revealed anemia (hemoglobin of 7.6gm/dL).Skin biopsy revealed sweat glands without any innervation. Nerve conduction velocity (NCV) study was normal. A pilocarpine skin testing induced no sweating in the child. Considering all of the above, we reached a diagnosis of Hereditary Sensory and autonomic neuropathy type IV.Wound debridement was done and osteomyelitis was ruled out; she was started on IV antibiotics for the ulcer and transfused twice with packed cells. The parents were counseled regarding the genetic nature of the disease.

Hereditary sensory and autonomic neuropathies (HSAN) include a number of genetic disorders associated with sensory and autonomic disturbances. Originally 4 types were described by Dyck and Ohta. (1) HSAN IV is a rare autosomal recessive disorder described by Gillespie et al in 1960. (2) The commonest mutations are seen in the NTRK1(TRKA) gene located on chromosome 1(1q1-q22) which results in impeded signal transduction at the NGF(Nerve growth factor) receptor, leading to failure of survival of small sensory and sympathetic neurons.(3)It is characterized by triad of anhidrosis (leading to hyperpyrexia), decreased pain perception and mental retardation. (4) Impaired thoracolumbar sympathetic outflow is responsible for anhidrosis. Decreased sensation leads to self mutilation once teeth erupt and they present with refractory ulcers and osteomyelitis. There is loss of superficial, deep and visceral pain. Mild to moderate mental retardation and delayed motor development are usually seen. Ultrastructural examination of peripheral nerve reveals complete lack of unmyelinated fibres(slow pain and temperature) and reduction in number of small myelinated fibres(fast pain). Electron microscopy of skin shows absence of C and Aδ fibres in epidermis and demonstrable sweat glands without the surrounding unmyelinated fibres. (5) Management is supportive; prevention of hyperthermia with cooling bath, careful inspection for self mutilation daily, smoothening/extraction of teeth, use of antipyretics, use of braces to prevent injury and early aggressive treatment of the wounds. With early diagnosis and increased awareness, the prognosis seems to be slightly better than earlier thought.

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