A Case of Kennedy's Disease from India

Sir,

We report a case of an Indian patient who presented to us with progressive limb weakness and bulbar symptoms. To the best of our knowledge, it is the first case report of Kennedy's disease from India.

A 62-year-old male presented with a history of insidious onset, gradually progressive, symmetric weakness, and sensory symptoms of extremities for the past 7 years, followed by hand tremors for the last 2 years, and dysphagia since 2 weeks with a history of abnormal twitching movements (over extremities, trunk, and face), thinning of limbs, muscle cramps, fatigability, and loss of libido and diabetes mellitus for the last 3 years. Positive family history of similar complaints in two of his brothers and one maternal uncle was present [Figure 1].

He had bilateral gynecomastia with the loss of pubic hairs and testicular atrophy. Bilateral facial weakness with facial and chin fasciculations, tongue atrophy, and palatal weakness was present [Figure 2]. There was symmetric proximal as well as distal weakness of all extremities with generalized areflexia and absent plantar response. Bilateral postural hand tremors were seen. Sensory system examination revealed decreased sense of touch and pain (25%) distally in lower limbs.

Nerve conduction study (NCS) revealed asymmetric bilateral involvement of motor nerve conduction parameters, in the form of absent to decrease compound muscle action potentials (CMAPs) involving upper limb more than lower limb with normal motor distal latencies and conduction velocities. Sensory NCSs revealed nonrecordable or reduced

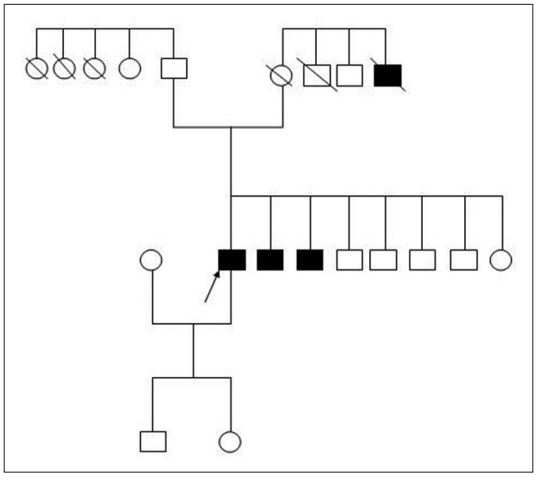

Figure 1: Pedigree chart of the patient

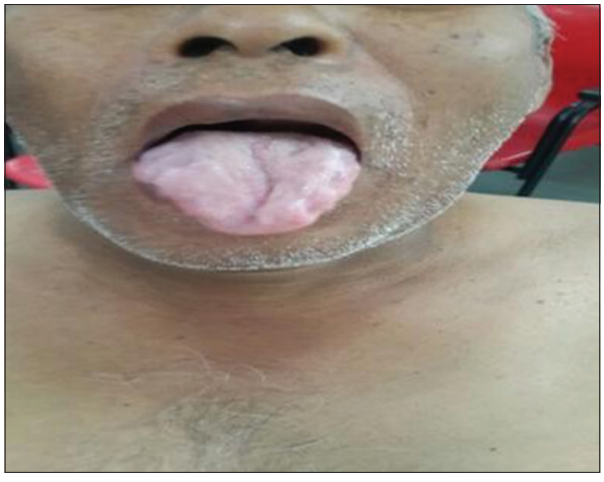

Figure 2: Tongue atrophy

sensory nerve action potentials (SNAPs) from all examined upper and lower limb nerves. F-wave studies showed normal F latencies except nonrecordable response from bilateral peroneal nerves.

Genetic analysis (Cytosine Adenine Guanine (CAG) repeat on androgen receptor gene on Xq 11-12) confirmed the diagnosis which revealed 49 ± 3 CAG repeats (normal range 9–36).

Kennedy's disease (spinobulbar muscular atrophy [SBMA]) is a rare X-linked recessive neurodegenerative disorder characterized by degeneration of lower motor neurons and is caused by CAG trinucleotide repeat expansion in the androgen receptor gene on chromosome Xq11-12.[1] It is characterized by progressive atrophy and weakness of limb and bulbar muscles with tongue atrophy and chin fasciculations and with onset in the 3rd–5th decades.[2] Patients may have endocrinological abnormalities in the form of gynecomastia, testicular atrophy, and diabetes mellitus[3] It is important to differentiate Kennedy's disease from other neuromuscular disorders as several disorders of varying severity and outcomes resemble SBMA. On electrophysiological studies, CMAP amplitudes may be low. Most patients have low amplitude or absent SNAPs, which reflect the association of Kennedy's disease with degeneration of the dorsal root ganglia. Currently, there is no cure for Kennedy's disease, and treatment is mainly symptomatic and supportive.[4]

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Conflicts of interest

There are no conflicts of interest.

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