CORRESPONDENCE

Sensory Ataxic Neuropathy with Dysarthria and Ophthalmoplegia (SANDO): A Multisystem Mitochondrial Disorder

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Mitochondrial diseases are clinically heterogeneous and have complex inheritance patterns resulting in incorrect or delayed diagnosis.1 We report a case of mitochondrial cytopathy with ataxic sensory neuropathy, dysarthria, and ophthalmoplegia in a middle-aged female.

A 36-year-old lady presented with insidious onset, gradually progressive neurological illness in the form of distal, symmetrical, flail weakness of both upper limbs and lower limbs (lower limbs > upper limbs) of 8 years duration, associated with numbness and paresthesias in all four limbs of 6 years duration.

Neurological examination revealed mild dysarthria, mild ptosis in the left eye, restriction of eye movements in all directions, without double vision, absent deep tendon reflexes in lower limbs, and glove and stocking pattern of graded sensory loss for all modalities of sensations in all four limbs. Romberg’s sign was positive.

Laboratory investigations revealed normal blood parameters without muscle/liver enzyme elevation. Vitamin B12 levels were normal. The vasculitic workup was negative. Nerve conduction test demonstrated severe sensory motor axonal neuropathy of all four limbs. Needle electromyography showed features of chronic denervation. Magnetic resonance imaging brain was normal. Muscle biopsy of peroneus brevis muscle, demonstrated ragged red fibers and COX-negative fibers, features suggestive of mitochondrial myopathy. Superficial peroneal nerve biopsy shows features of severe axonal neuropathy.

The term “sensory ataxic neuropathy with dysarthria and ophthalmoplegia” syndrome is characterized by an adult-onset severe form of ataxic sensory neuropathy, dysarthria, and chronic progressive external ophthalmoplegia,2 results from mitochondrial dysfunction and is due to mitochondrial deoxyribonucleic acid depletion in muscle and peripheral nerve. The phenotype is largely variable.3 Mitochondrial disorders should always be considered in the differential diagnosis of neurological diseases with multiaxial involvement (Fig. 1).

REFERENCES


Fig. 1: Restriction of eye movements in all directions (external ophthalmoplegia)