Type 1 Myotonic Dystrophy Presenting with Bulbar Weakness without Myotonia

To the Editor,

We report a case of 22-year-old male patient who presented with gradually progressive change in voice and difficulty swallowing for 2 years. He had nasal intonation of voice with occasional nasal regurgitation of liquids and coughing while swallowing. There was no history of preceding or accompanying fever, headache, vomiting, diplopia, droopy eye lids, any other focal motor or sensory deficit, involuntary muscle twitching, atrophy, easy fatigability, or impairment of balance. There was no history suggestive of myotonia. Past medical and family history were not significant. The general physical examination revealed subtle wasting of facial muscles compared to how he looked a year ago [Figure 1]. Neurological examination revealed symmetrically reduced palatal movements and reduced gag reflex; no tongue atrophy or fasciculations were seen. Motor strength and deep tendon reflexes were normal. Rest of the examination including detailed ophthalmologic workup was unremarkable. Routine blood investigations including blood counts, renal, hepatic, and thyroid function tests, glycemic and hormonal profile were normal. Creatine kinase was mildly elevated (342 U/L). Electrocardiogram and 2D-echocardiogram were normal. Magnetic resonance imaging of brain was unremarkable. Neurodiagnostic tests including nerve conduction studies, repetitive nerve stimulation testing and electromyography (EMG) per motor neuron disease protocol were within normal limits. EMG done in distal (abductor pollicis brevis, tibialis anterior) and proximal muscles (deltoid, vastus lateralis) in all four limbs and paraspinal muscles did not show any abnormal spontaneous activity; motor unit action potential and interference pattern were within normal limits. Work up for myasthenia gravis was negative. Genetic analysis for myotonic dystrophy type 1, done in view of subtle facial muscle wasting as noticed by the patient, detected an abnormal copy of dystrophia myotonica protein kinase (DMPK) gene with CTG repeats (>100) in disease range using triple repeat primed Polymerase Chain Reaction (TP-PCR) technique in patient’s tested DNA sample. Following the genetic report, detailed family history and clinical examination of family members, till two previous generations, was undertaken to no significant finding. Neurodiagnostic testing of elder sister and father did not reveal any abnormality. Patient was explained the prognosis of his illness and discharged to follow up in OPD. This case is atypical because of lack of family history, absence of clinical or electrical myotonia or any other systemic involvement.

DM type 1 is a hereditary muscular disorder with autosomal dominant inheritance. It affects skeletal, smooth muscle, and endocrine system.\[1\] Patients are predisposed to cataract formation. It is associated with a trinucleotide CTG repeat expansion in the DMPK gene (chromosome 19q13).\[1\]

DM type 1 has four distinct clinical forms: congenital, childhood-onset, adult-onset, and late-onset oligosymptomatic.\[2\] The prognosis of DM type 1 is associated with the age of onset.\[3\] Patients with childhood-onset DM have poorer prognosis compared to adult-onset DM.\[4\] Adult-onset DM type 1 (most prevalent form)
commonly presents with myotonia, muscle weakness, and amyotrophy. Myotonia is the most frequent symptom manifesting as difficulty in actively relaxing the hand muscles following contraction. Amyotrophy initially affects the hand muscles but may progress to involve the craniofacial muscles causing hatchet facies. It also leads to endocrinal involvement causing alopecia, impaired glucose tolerance, sexual dysfunction, and menstrual disorders. In view of atypical clinical presentation, we could make the diagnosis based on the genetic testing. This emphasizes the importance of genetic testing in making the diagnosis.

**Declaration of patient consent**

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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**Conflicts of interest**

There are no conflicts of interest.

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