Abstract

Background– Dystrophia myotonica (DM) is an autosomal dominant muscular dystrophy characterized by abnormal trinucleotide or tetranucleotide repeats. DMPK (Dystrophia myotonica protein kinase) or ZNF9 genes are affected in type 1 and type 2 forms of the disease, respectively. It is a multi-system disorder with various clinical manifestations. Patients may present with distal and proximal muscle weakness, endocrine dysfunction, cataract, cognitive impairment, abnormal liver functions, cardiomyopathy, and cardio-embolic strokes etc. Anicteric cholestasis is a rare complication of the disease with an unknown mechanism. Unfortunately, there is no specific disease-modifying therapy for DM. Therefore, the symptomatic treatment and continuous disease surveillance is the cornerstone of the management.

Case–A 60-year old male who was unable to walk for the last five years due to progressive distal limb weakness presented with progressive bilateral blindness for the last four years and osmotic symptoms for last six months. Clinical and laboratory evaluation revealed a diagnosis of DM complicated with cataract, newly diagnosed type 2 diabetes, and anicteric cholestasis. Fortunately, his vision recovered; but he remained wheelchair-bound due to the long-standing disease.

Conclusion - DM does not always present to a neurologist due to its wide array of clinical manifestations other than neurological symptoms. Therefore, a high degree of clinical suspicion is needed for the early diagnosis of the disease. Though the mainstay of therapy is supportive continuous monitoring of patients for disease's complications is vital in long-term management. Anicteric cholestasis is a rare complication of DM which requires exclusion of other common causes of cholestasis.