INTRODUCTION

Myotonic Dystrophy (MD) is the most common inherited neuromuscular disease with a worldwide prevalence of 2.1 to 14.3 per 100,000 inhabitants and incidence of 1 in 8000 births [1]. MD exhibits anticipation and variable expression from generation to another. It is a multi-system disorder affecting muscles, central nervous system, eye, endocrine system, gastrointestinal system, and most importantly heart. Features that allow early diagnosis are myotonia and muscle weakness. However, the diagnosis can be under recognized when only cataract and or dementia are the only presenting features. Increased awareness is important as prompt diagnosis allows prevention of complications, detection of other family members and genetic counseling.

CASE REPORT

We report a 65-year-old gentleman presented with two-year history of progressive memory impairment. Medical history includes cataract at age of 50 and ischemic heart disease. His mother, who died aged 68, had cataracts and had been diagnosed with Alzheimer’s Disease. Two of his sisters also had cataracts; one died suddenly aged 50 from a presumed cardiac cause. His physical examination was unremarkable apart from cognitive impairment consistent with dementia. 18 months after presentation, he died from myocardial infarction.

A year later his 30-year-old son presented with weakness and myotonia of hand muscles. At the age of 26 he had undergone cataract extraction. Genetic testing confirmed the diagnosis of myotonic dystrophy.

It is very likely that our patient had MD when he presented with dementia. The clues that point toward that are his presenile cataract, strong family history of cataract and the confirmed diagnosis in his son.

DISCUSSION

In one study, 5% of cataract patients without precipitating factor, other than diabetes, with or without neurological manifestations had MD mutation [2]. Authors also found other affected family members of those patients. Several other studies have detected MD by screening cataract patients [3, 4]. In our case, the son, with strong family history of cataract, had bilateral cataract extraction at age 26.

Age-related decline of frontal and temporal cognitive functions were documented among 60 patients with myotonic dystrophy in one study [5]. According to that study, cognitive decline can be the sole manifestation of myotonic dystrophy in otherwise asymptomatic individuals. Memory impairment is suggested to be associated with mild MD [6]. In our case, the father presented with dementia and had past and family history of cataract extraction. It is likely that the father also had MD.

CONCLUSION

We therefore strongly recommend that clinicians should consider myotonic dystrophy in the differential diagnosis of dementia and or cataract. Careful attention needs to be paid to family history of cataract.

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