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Case Report

Myotonic Dystrophy; Diagnosed based on Various Ophthalmic Findings

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Abstract

Ocular manifestations are often the presenting signs or symptoms for multiple systemic diseases. Herein, we present a case of myotonic dystrophy where the sole presentations were ophthalmic examination findings. A 75-year-old male presented with excessive tearing and was found to have bilateral severe blepharoptosis, lash ptosis, sluggish pupillary response associated with unilateral Christmas tree iridescent cataract.

The presence of christmas tree in conjunction with external ophthalmoplegia is almost pathognomonic for myotonic dystrophy. The patient was referred for genetic evaluation to confirm the diagnosis of myotonic dystrophy. In conclusion, ophthalmologists may play a major role in the diagnosis of myotonic dystrophy based on clinical ophthalmic findings, before the manifestation of systemic abnormalities.

Keywords: *Myotonic Dystrophy; Ophthalmic Findings; Ocular Manifestations*

Introduction

Myotonic dystrophy is the most common muscular dystrophy in adults. It is a multi-systemic, progressive, autosomal dominant inherited disease. Highly variable in its presentation, symptoms can either be muscular or extramuscular and that may result in diagnosis delay [1].

The diagnosis of the disease is based on clinical signs and symptoms and are confirmed by genetic analysis looking for the origin of this clinical abnormality [2].

Cataract is the most common ocular finding in these patients, almost all myotonic dystrophy patients will develop bilateral iridescent cataracts. Nevertheless, ocular complications can be the first clinical sign or symptom in myotonic dystrophy patients and can lead the patient to seek medical care [3].

This case report describes a patient with myotonic dystrophy based on his ocular findings.

Case Report

A 75 year old male patient visited our clinic complaining of tearing of both eyes, he is a known case of glaucoma, controlled with eye drops. He has no known underlying diagnosed systemic conditions.

His best corrected visual acuity was 0.6 in the right eye and 0.4 in the left eye, and intraocular pressure was 18 mmHg in the right eye and 22 mmHg in the left eye.

He has severe bilateral blepharoptosis in his both eyes and lash ptosis (Figure 1), with margin reflex distance -1 mm in the right eye and 0 mm in the left eye, and levator function test of 13 mm in the right eye and 12 mm in the left eye, palpebral fissure height was 8 mm in the right eye and 9 mm in the left eye. Bilateral brow elevation was also noticed (Figure 2).

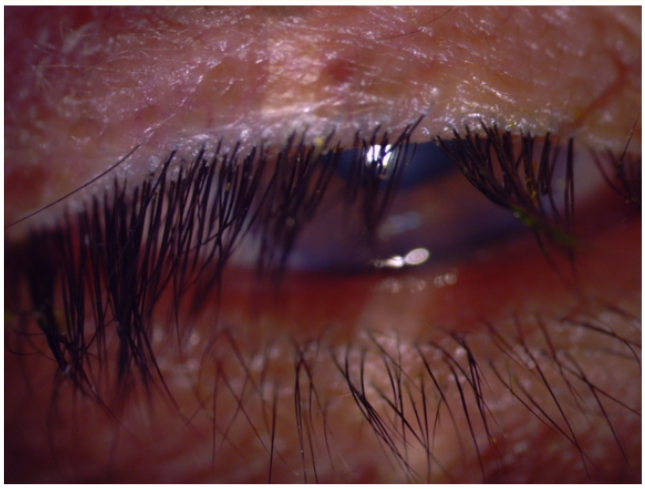

Figure 1

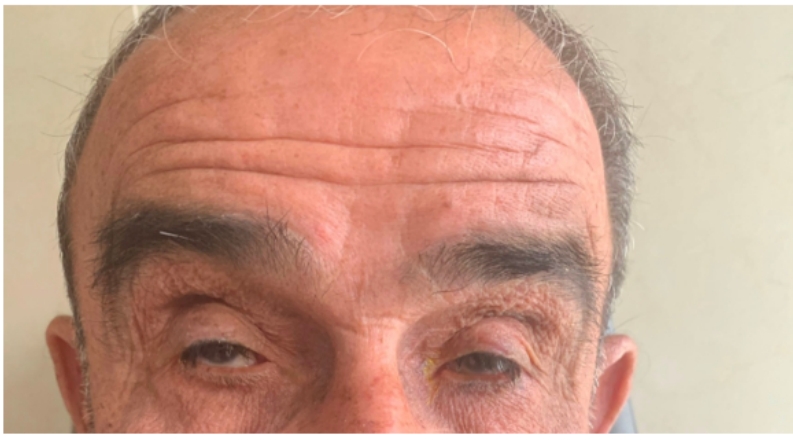

Figure 2

Ocular motility was restricted in all directions. On slit lamp examination, a characteristic fine dust like opacity with tiny iridescent spots in cortex; Christmas tree like cataract in the right eye (Figure 3A) and nuclear sclerosis cataract stage 1 in the left eye (Figure 3B).

Patent punctum bilaterally, corneas were clear, anterior chamber of both eyes were deep and quiet, pupils were regular round reactive and no abnormality detected in the iris bilaterally.

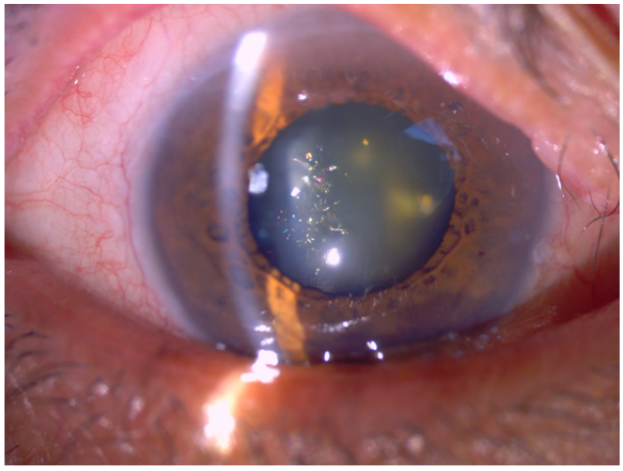

Figure 3A

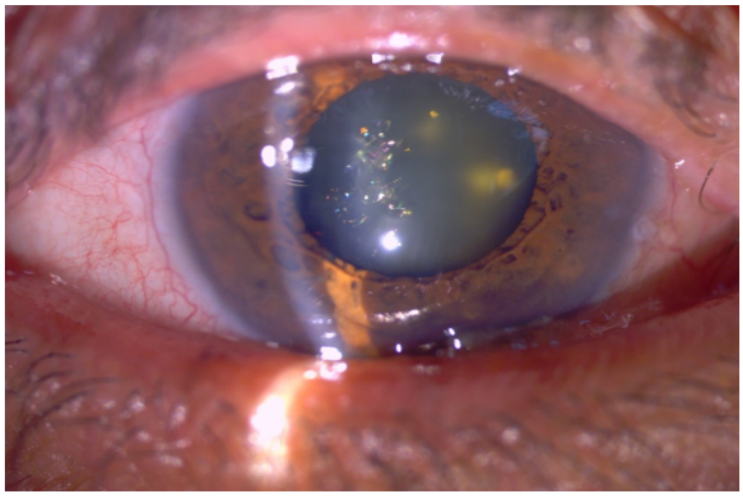

Figure 3B

Dilated fundus examination showed vertical cup to disc ratio of 0.7/0.6 in his right and left optic nerve respectively. Retina was flat bilaterally and foveal reflex was normal in both eyes. Optical Coherence tomography of the macula and the optic nerve head was done, revealed normal foveal contour bilaterally, mild inferior and superior retinal nerve fiber layer thinning bilaterally, and ganglion cell damage in the left eye.

General systemic review and other myotonic dystrophy symptoms were reviewed; his general appearance showed frontal balding and muscle wasting, no delayed relaxation upon hand shaking, absence of general weakness or fatigue.

Discussion

Myotonic dystrophy is classified into two types: type1 results from CTG expansion in the 3′ untranslated region of the dystrophia myotonic protein kinase (DMPK) gene on chromosome 19q13 and type 2 results from CCTG expansion located within intron 1 of the cellular nucleic-acid-binding protein (CNBP) gene on chromosome 3q21 [4].

These multi-organ diseases mainly present clinically as myotonia due to myopathy, cardiac conduction defects, cerebral involvement, endocrine disorders and ocular involvement such as ptosis, ophthalmoplegia, extra-ocular myotonia, and decreased visual acuity due to cataract or retinal degeneration, most commonly posterior iridescent cataracts [5,6].

Therefore, a multicolored iridescent cataract finding on slit lamp examination in a patient with muscular symptoms and family history of muscular symptoms or premature cataract, is highly suggestive of myotonic dystrophy [7].

Upon ophthalmic examination, our case was found to have unilateral iridescent Christmas tree cataract, bilateral blepharoptosis, bilateral lash ptosis, normal intraocular pressure, increased CDRs in both eyes, and retinal nerve fiber layer defect in the left eye.

Glaucoma is not one of the ocular manifestations of myotonic dystrophy, further studies should be done to reveal the relation between these disorders, and the underlying mechanism of the association [3].

Based on these ophthalmic findings, the patient was referred to the National Center for Diabetes, Endocrinology and Genetics, Department of Laboratory Medicine for genetic evaluation for myotonic dystrophy. Based on ophthalmic indications, the patient was possibly diagnosed with myotonic dystrophy and could be confirmed by genetic examination before the manifestation of other systemic changes.

Although this disease cannot be cured, proper diagnosis is essential for management of symptoms and complications thus effective control of the development of the disease, decreasing the associated comorbidities. As a result, this will enhance the quality of life of these patients.

Conclusion

We conclude that characteristic ocular findings that usually have associations with underlying systemic diseases could open the door for the diagnosis of a multi-organ disabling disease far beyond a diagnosis of a cataract. This leads to proper diagnosis of patients and management of the resulted complications and symptoms. Our case showed ocular involvement of myotonic dystrophy, which can be the first manifestation of a disease and most commonly presents as cataracts.

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