Review Article

MYOTONIC DYSTROPHY: A RARE CAUSE OF ACQUIRED MYOGENIC PTOSIS
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Abstract
Myogenic ptosis is a rare type of acquired blepharoptosis and can be found in localized or diffuse muscular disease, such as the muscular dystrophies. The muscular dystrophies are distinguishable by their mode of inheritance and clinical features. Blepharoptosis is a clinical sign of certain muscular dystrophies, including myotonic dystrophy, Chronic Progressive External Ophthalmoplegia (CPEO), and oculopharyngeal muscular dystrophy.

We present a case report of a 55 years old lady presenting with bilateral acquired ptosis with limitation of extraocular movements. Clinical presentation, differential diagnosis and management options of acquired myogenic ptosis are discussed.

Myotonic dystrophy is a variable genetic disorder with multisystem involvement. Central nervous, cardiovascular, musculoskeletal, gastrointestinal, reproductive systems are the ones commonly affected. The ocular symptoms include bilateral acquired ptosis without ophthalmoplegia, blue dot or Christmas tree cataract and retinal pigment epithelial changes.

Key words: Myogenic acquired ptosis, myotonic dystrophy, cataract, ophthalmoplegia.

Key message:
Late onset Myotonic dystrophy, a hereditary disorder of muscular origin should be kept in mind as a cause of acquired ptosis in a patient presenting with recent onset bilateral ptosis with limitation of extraocular movements. Characteristic finding in EMG and negative neostigmine test confirms diagnosis of Myotonic dystrophy. Other ocular manifestations like cataract, RPE changes, sluggish reacting pupils along with multisystem involvement like musculoskeletal system, cardiovascular system, gastrointestinal system make the management challenging with a guarded prognosis.

Introduction:
Blepheroptosis can be of congenital or acquired type. Acquired ptosis being the less common type is further divided into neurogenic, aponeurotic, mechanical and myogenic varieties. Myogenic ptosis is a rare type of acquired blepharoptosis and can be found in localized or diffuse muscular disease, such as the muscular dystrophies.

Myotonic dystrophy is one of the most common forms of inherited muscle disease; it is estimated that one person in every 20,000 is affected with DM. [1] The ocular involvement in myotonic dystrophy occurs commonly in the form of ptosis, extraocular muscle weakness and blue dot/Christmas tree cataracts. Cataracts are useful for diagnosing myotonic dystrophy since they are present in nearly all patients with the condition. [3] Other features of myotonic dystrophy include sluggish pupillary response, low intraocular pressure, and retinal degeneration. [4]

Case report:
A 55 years old female patient presented with complaints of drooping of her eyelids and decreased visual acuity in both eyes since 6 months. On ophthalmological examination her visual acuity was 20/80 in both eyes with normal intraocular pressure. There was severe
ptosis (4mm) and 15° Exotropia in RE and Mild ptosis (2 mm) in LE. Levator function was 2 mm in RE and 5 mm in LE. Pupillary reaction was sluggish in both eyes. Extraocular movements were found to be partially restricted in medial and upward direction in RE. On slit lamp examination lens showed presence of early peripheral cortical cataract and on fundus examination no abnormality was detected.

On detailed general and systemic examination following signs were noted:

**General examination:** Drowsiness, early fatiguability, myopathic facies with frontal baldness.

**Musculoskeletal system examination:** Percussion myotonia, inability of muscles to relax, Sternocledomastoid muscle atrophy, temporalis and masseter muscle atrophy.

**Central nervous system examination:** Distal muscle weakness.

Rest systemic examination did not show any significant abnormality.

All the routine blood, urine investigations, electrocardiography, thyroid function test were within normal limits.

These findings pointed towards a muscular pathology. The EMG study carried out suggested primary muscle disease. Possible diagnosis of myesthena gravis or muscular dystrophy was made. Neostigmine test (1.5mg IM Neostigmine) and ice pack test were carried to check for Myesthena gravis. Both the tests were inconclusive, ruling out Myasthenia.
Discussion

Myotonic dystrophy is an inherited disorder accompanied by progressive wasting and weakness of the distal muscles and myotonia.\[^2\] Cases of myotonic dystrophy are generally genetically classified as one of two types: **Type 1** myotonic dystrophy has amplifications of an unstable trinucleotide CTG repeat located on chromosome 19, while **Type 2** has CCTG expansion on chromosome 3. The myotonic dystrophy is described as mild, classical or congenital according to the age of presentation and severity of symptoms.

Histologically, myotonic dystrophy is characterized by degenerative changes of skeletal muscle including disruption of myofilaments and sarcoplasmic reticulum, focal accumulation of mitochondria, and eventually atrophy and fibrosis of muscles.\[^5\] The distal musculature is usually the first to be affected. Myotonia is often the initial symptom, which results from delayed relaxation of skeletal muscle after contraction. Symptoms are exacerbated with cold, excitement, and fatigue. Patients often develop a characteristic myopathic facies, presenting with blepharoptosis, frontal balding, and wasting of the temporalis masseter muscle. Ocular findings include ptosis, blepharospasm, ophthalmoplegia (similar to CPEO), pigmentary retinopathy, “Christmas tree” cataracts, and miotic pupils.\[^6\]
Associated systemic findings include low intelligence, insulin resistance, cardiac conduction abnormalities, and testicular and uterine atrophy.

Management of such cases will include ptosis correction with frontalis sling using autogenous fascia lata. This line of management faces mainly following two difficulties:

1. Poor eye protective mechanisms as poor Bell’s phenomenon, incomplete lid closure increase risk of exposure keratopathy.[8]

2. Anaesthetic complications due to administration of muscle relaxants in such patients dictate risk of cardiorespiratory arrest. [7]

Management of cataract includes performing phacoemulsification followed by posterior chamber IOL implantation. The visual prognosis remains guarded due to possible retinal pigment epithelium changes in the fundus. [4]

Thus Myotonic dystrophy forms a rare cause of acquired ptosis. It is a hereditary disorder causing acquired myogenic ptosis, cataract and retinal pigment epithelial changes. As ophthalmologists, the cases should be treated with special attention considering the multisystem involvement. The risks and benefits for the individual patient should be kept in mind while planning for both ptosis correction and cataract extraction.

References: