Ocular manifestations of adult-onset myotonic muscular dystrophy.
Frank J. Kuchera, O.D. & Gregory S. Wolfe, O.D., M.P.H., F.A.A.O.

Abstract:

A 55 year-old Caucasian male presents for examination with rare macular changes as well as classic lenticular and peripheral retinal findings characteristic of myotonic dystrophy. Additionally, longevity adds to the uniqueness of his clinical presentation.

I. Case History

A. Patient demographic: 55 year old Caucasian male

B. Chief complaint: Constant blur at distance and near with current spectacles

C. Ocular history: History of mild blepharitis OU

D. Medical history: Myotonic Muscular Dystrophy
   • Left proximal arm weakness
   • Dysphagia, Oropharyngeal Phase
   • Electrocardiogram abnormal
     o NSR with ant-lat T wave inversion suggestive of ischemia
     o PR interval is within normal limits
   SOB/Wheezing
   Mild dysarthria
   Halitosis
   Calculus of gallbladder
   H/O Cocaine abuse, episodic use

E. Meds/Allergy: None

II. Pertinent findings

A. Gross: Locking hand grasp when shaking hands
   Short stature
   Slow gait
   Mournful facial presentation
   No ptosis

B. BCVA: OD: 20/25+2
       OS: 20/25-1

C. EOMS: Full range of motion

D. Conjunctiva: OU Few petechial hemes around limbal area

E. Lens: OU 1-2+ NSC, anterior and posterior cortical punctate polychromatic opacities, 1-2+ PSC
F. Macula: OD: Flat, RPE pigment clumping in arcuate shape inferior to fovea  
OS: Flat, Mild ERM inferiorly  
G. Periphery: OU: area of RPE hypertrophy nasal and superior

III. Differential Diagnoses  
A. Differential Diagnoses of Ocular Findings  
   a. Lens  
      i. Diabetes Mellitus-related cataract  
      ii. Hypocalcemia  
   b. Retinal pigmentary clumping  
      i. ARMD  
      ii. Choroidal nevus  
      iii. CHRPE  
B. Differential Diagnoses of Systemic Findings  
   i. Muscular Dystrophy  
   ii. Kearns-Sayre Syndrome  
   iii. Occulopharyngeal Muscular Dystrophy

IV. Diagnosis and discussion  
   a. Autosomal dominant disorder mapped on chromosome 19q  
      i. Affects 1/8000 Caucasians  
   b. Systemic manifestations:  
      i. Myotonia (often presenting symptom)  
         1. skeletal  
            a. EMG findings  
            b. specific muscle groups  
               i. myopathic facies  
            c. warm-up phenomenon  
         2. respiratory  
      ii. endocrine involvement  
         1. Diabetes Mellitus  
         2. other endocrine involvement  
   iii. cardiovascular involvement  
      1. conduction deficit  
      2. electrocardiographic abnormalities  
   iv. intellectual impairment  
   v. life span  
   c. Ocular manifestations:  
      i. polychromatic cataract  
         1. Lens epithelial changes  
            a. Related to mutated allele of protein kinase gene  
            b. Expression of DMPK gene (Myotonic Dystrophy Protein Kinase)  
            c. Lens epithelial cell death leads to significantly reduced number of cells  
            d. High Ca$^{2+}$ levels found to lead to cell death  
            e. Cytoplasm adjacent to retractile bodies shows high Calcium concentration
ii. ptosis
iii. extraocular myopathy
iv. wasting of temporalis muscle
v. miotic pupils
vi. retinal pigment change
   1. peripheral
   2. macular

V. Treatment, management
a. Dilated fundus examination yearly
b. Home amsler grid to monitor macular changes
c. Fundus photography for progression analysis
d. Patient education of findings
e. Considerations for cataract excision
   i. Recurrent posterior capsular opacities are very common
      manifestations of cataract excision in myotonic dystrophy patients
   ii. Remaining epithelial cells have large propensity to undergo
      fibroblastic, proliferative reaction after cataract removal
   iii. Ciliary body atrophy leading to unopposed centripetal force at the
      capsulorhexis margin may lead to capsulorhexis contracture

VI. Conclusion:
A. This case represents both uncommon and classic ocular presentations of
   Myotonic Dystrophy.
B. Due to the minor impairment of vision, cataract excision was not deemed
   necessary. Yearly exams with regular monitoring of the progression of
   symptoms including OCT Cirrus Macular Cube scans has been recommended.

VII. Clinical Pearls:
   o Abnormal polychromatic retractile lenticular bodies warrant workup for
     Calcium-related abnormalities
   o Lid ptosis and mournful facial expression can be pathognomonic for myotonic
     dystrophy and require additional systemic evaluation
   o The greatest prevalence of myotonic dystrophy has been found in the
     Caucasian population

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