

Acute Vision Loss?

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Presentation

- 38 year old Caucasian Female in E.R.
- Acute, painless loss of vision, right eye
- Noted after occlusion of left eye
- Denies trauma

Medical/Surgical/Family History

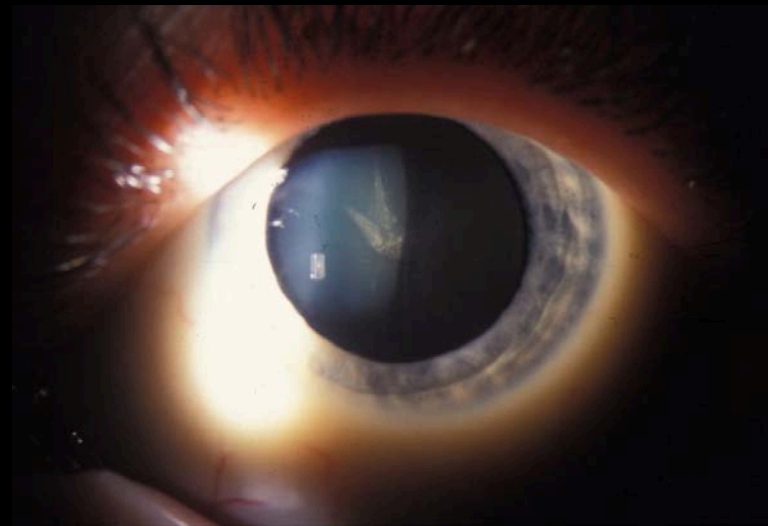
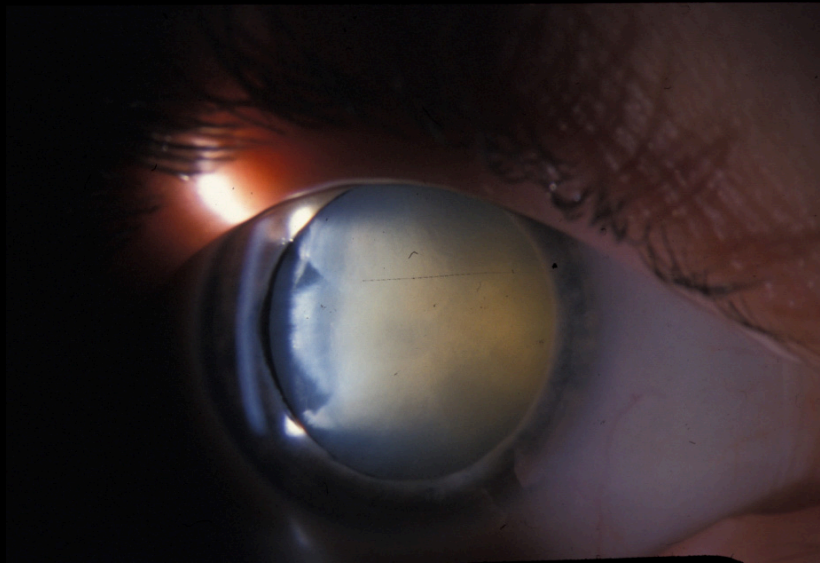
- Sleep apnea, obesity
- No surgeries
- No eye diseases or loss of vision



Examination

- Visual Acuity sc
 - CF at 3 feet, 20/40 ph 20/25
- Pupils 5→3 mm OU. No RAPD
- Extraocular Motility: full, painless
- Confrontational Fields:
 - Constricted OD
 - Full to finger count OS
- Orthophoric

Slit Lamp Examination



Dilated Fundus Exam

- B-scan OD
- DFE OS: cup/disc 0.3,
 - Macula – + foveal reflex
 - Vessels – normal
 - Vitreous – normal
 - Peripheral retina – normal

Additional History

- Difficulty releasing grasp
- Progressive difficulty with speech
- Father had a “muscle disease”

Myotonic Dystrophy (DM 1)

- Most common adult form of muscular dystrophy 1:8-10,000
- Autosomal Dominant
- Expansion of a CTG repeat (DMPK)
 - Dystrophy Protein Kinase Gene
 - Chromosome 19
 - >35 repeats - abnormal. >50 clinical expression.

CTGCTGCTGCTGCTGCTGCTGCTG
CTGCTGCTGCTGCTGCTGCTGCTG

- Chromosomes contain DNA sequences
- DNA sequences from nucleotides
 - (chemical bases: A, T, G, C)
- Different arrangements yield genes for specific proteins

(DM 1) 3 Forms

- Minimal: <150 repeats
 - Onset 20-70
- Classic: 100-1000 repeats
 - Onset 10-30 years
- Congenital: >2000 repeats
 - Onset birth – 10 years

Anticipation

- The phenomenon in genetics which allows the CTG repeats to expand from one generation to the next.
- Unpredictable.
- Does not always occur.

Myotonic Dystrophy

- Only myotonic condition affecting other organ systems
 - Eyes
 - Cardiac muscle
 - Smooth muscle
 - Brain
 - Bone

Chief Features

■ Muscles

- Weakness
- Cardiac conduction defects
- Ptosis
- Facial weakness
- Slow release of contraction

■ Cataracts

- Iridescent red & green crystals (not specific)
- Stellate posterior subcapsular
- Vacuoles
- Opacification

■ Others

- Low IOP
- EOM abnormalities

Myotonia



Diagnosis

- Complete patient and family history
- Genetic Testing:
 - Alterations in the DMPK gene
 - Prenatal testing available
- Electromyogram is Pathognomonic
 - High frequency, spontaneous action potentials
- Muscle Biopsy
 - Degenerative changes
 - Central location of nuclei and accumulations of mitochondria

Patient Follow up – 3 years

- Progressive difficulty breathing
 - Oxygen via nasal canula
- Diabetes mellitus
- Leg Cramps
- Foot weakness
- Dysarthria



Conclusions

- Most common muscular dystrophy in adults
- Genetic counseling available
- Involvement of specialist early
 - Neurologist
 - Cardiologist
 - Pulmonologist

References

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