

# **Ocular Effects and Treatments of Cystinosis**

**CRN Conference**

**San Antonio, TX**

**July 19-21, 2007**

**William A. Gahl, MD, PhD**

**National Human Genome Research Institute**

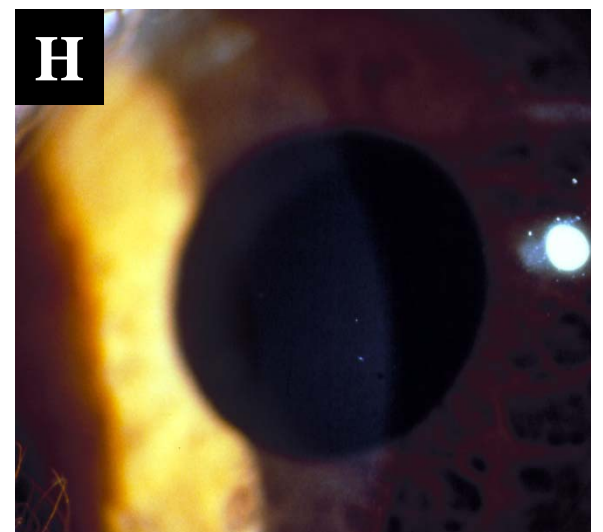
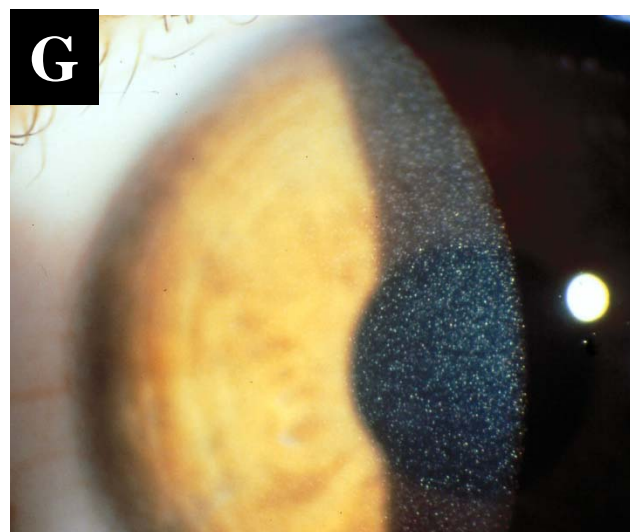
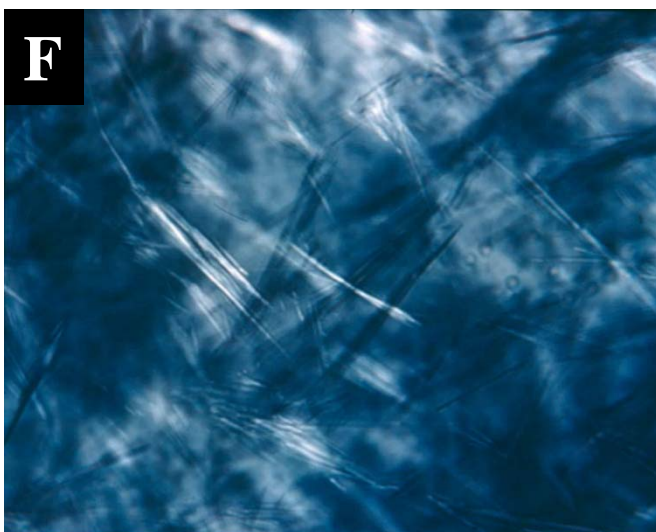
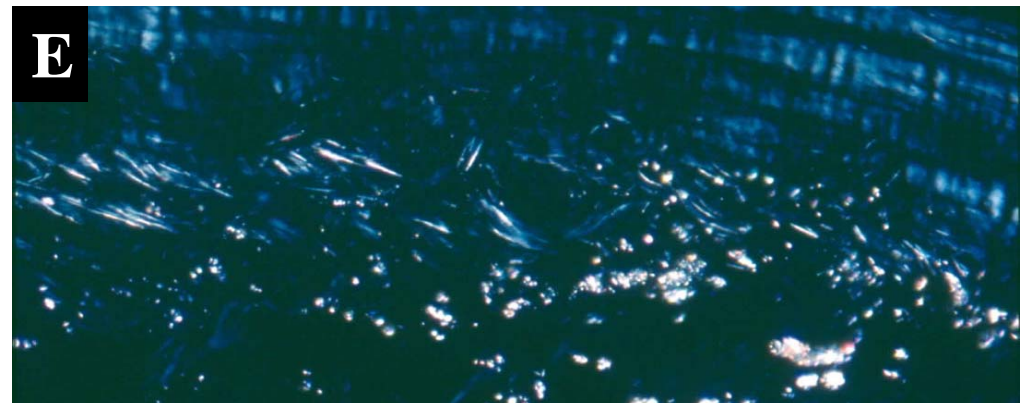
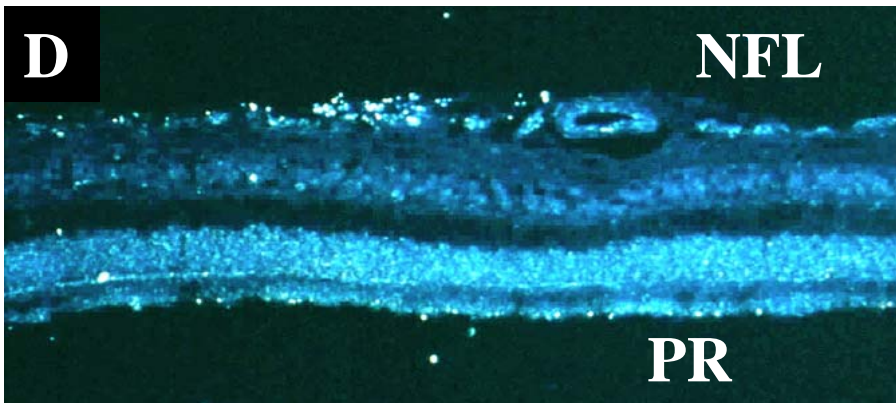
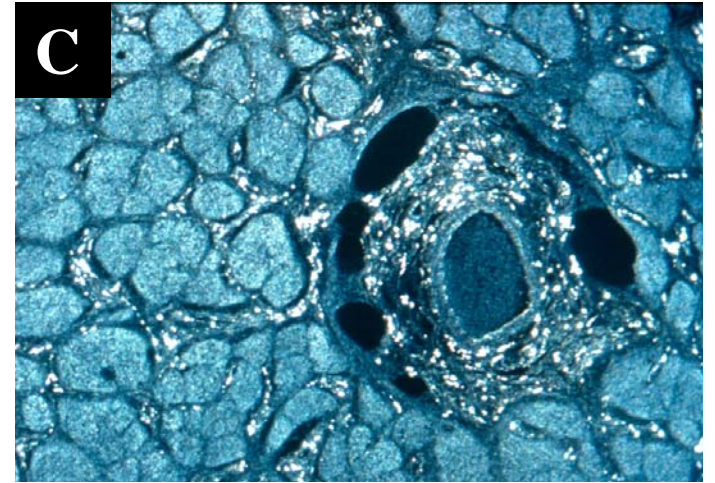
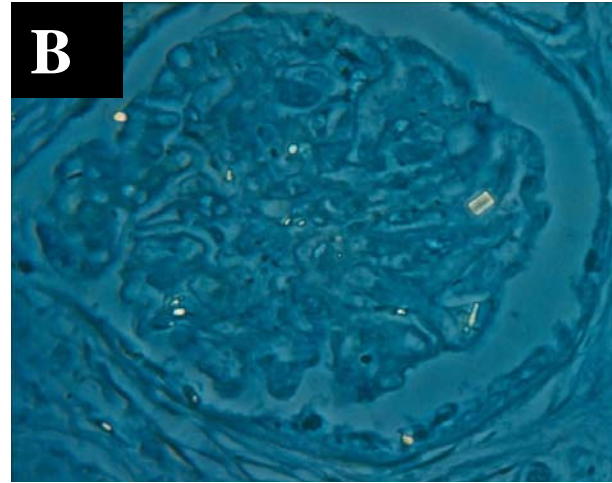
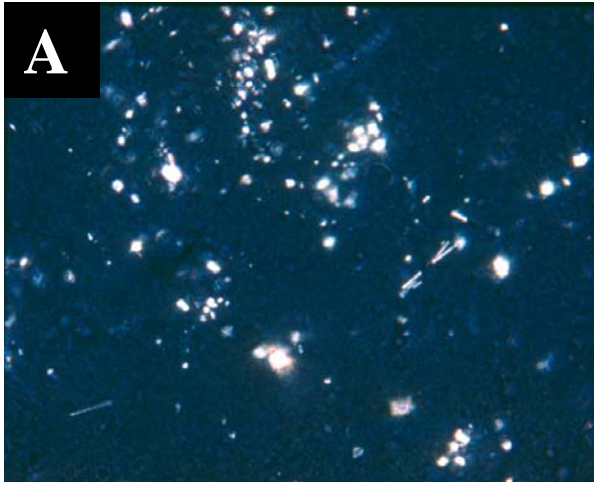
**National Institutes of Health**

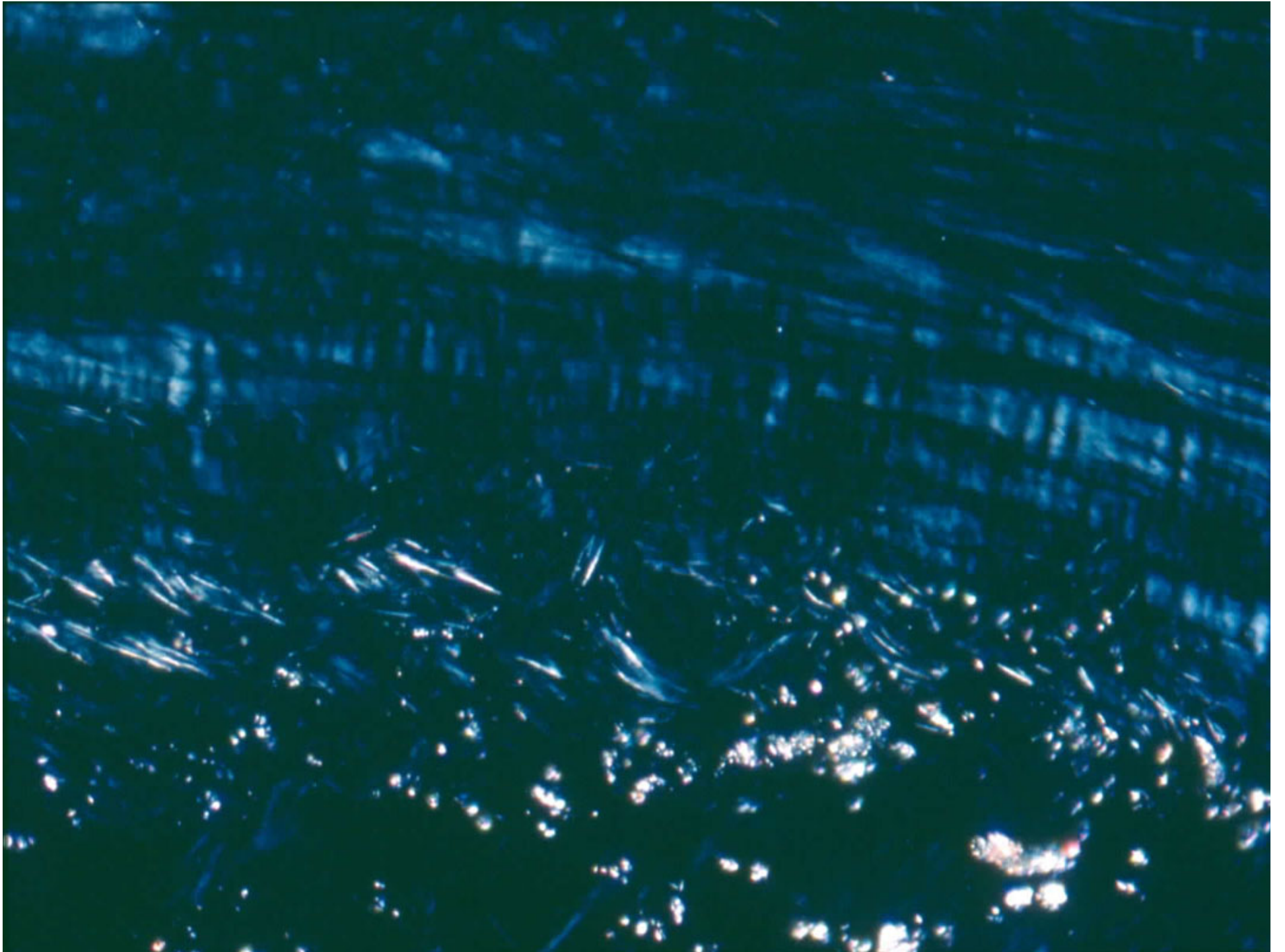
# Eye Complications of Cystinosis

- Background
- Posterior synechiae
- Anterior Segment Involvement
- Posterior Segment Involvement

# Background

- Pathology is due to cystine accumulation and crystal formation.
- The complications occur in patients not treated with cysteamine.
- Oral cysteamine helps prevent the posterior segment involvement.
- Topical cysteamine (eyedrops) prevents and removes the corneal crystals.



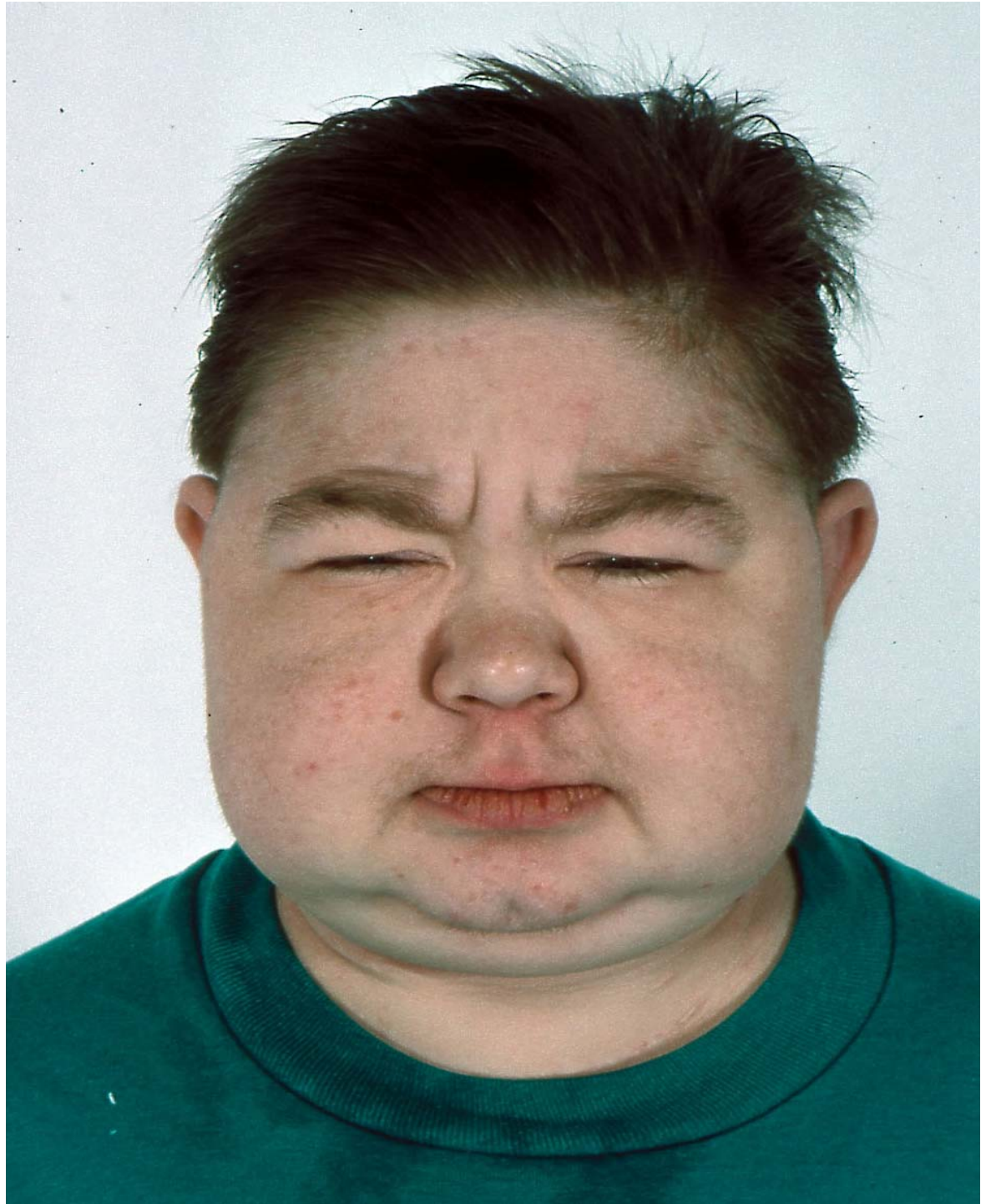


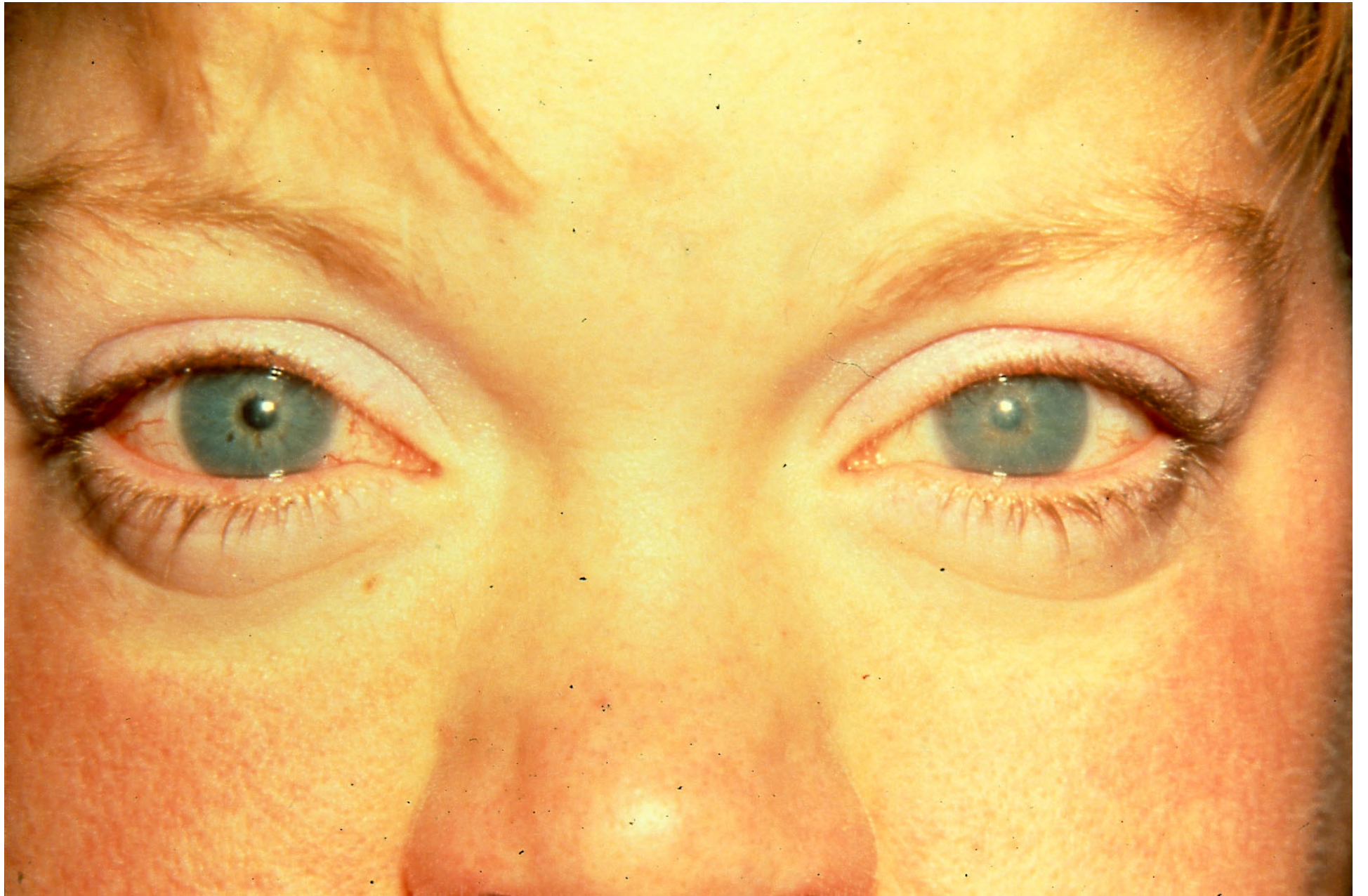
# Eye Complications of Cystinosis

- **Background**
- **Posterior synechiae**
- **Anterior Segment Involvement**
- **Posterior Segment Involvement**

# Eye Complications of Cystinosis

- Background
- Posterior synechiae
- Anterior Segment Involvement
- Posterior Segment Involvement



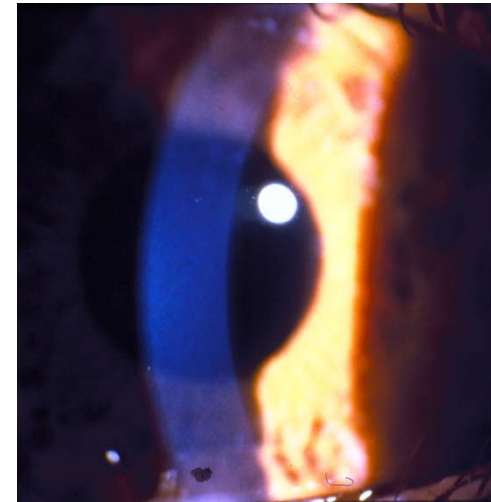
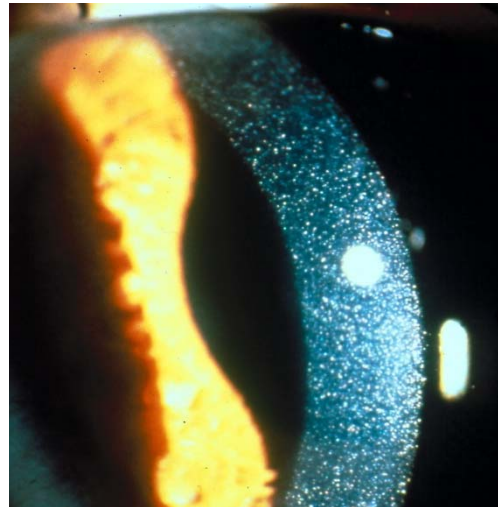


# CYSTEAMINE EYEDROPS

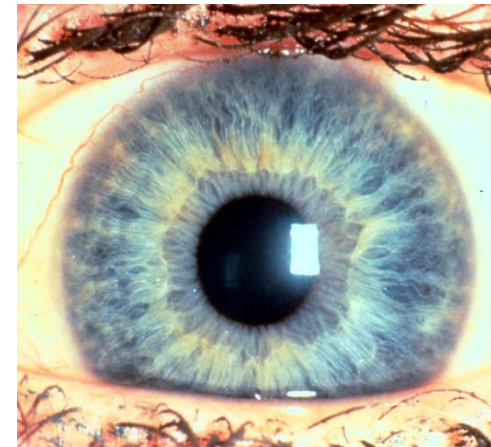
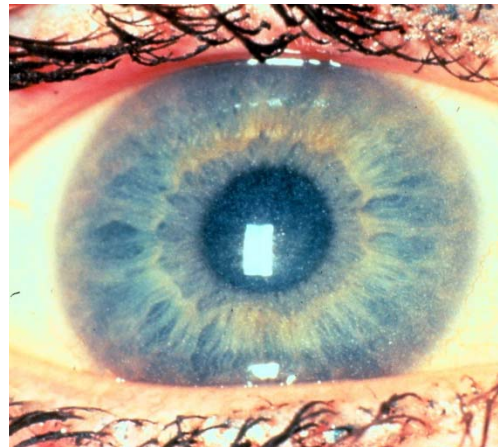
**Untreated**

**Treated**

**3-year old**



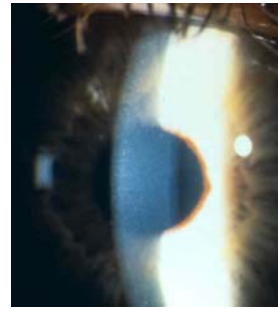
**20-year old**



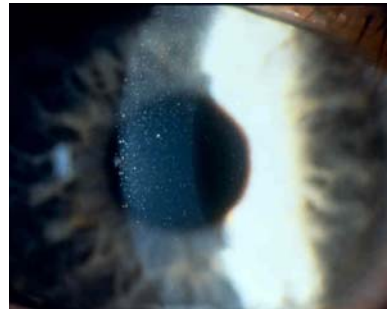
# Library of Corneal Crystal Densities



**0.00**



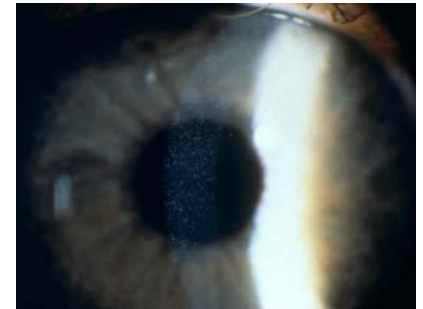
**0.25**



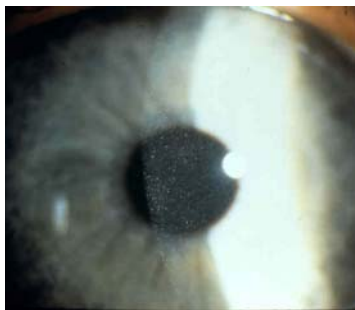
**0.50**



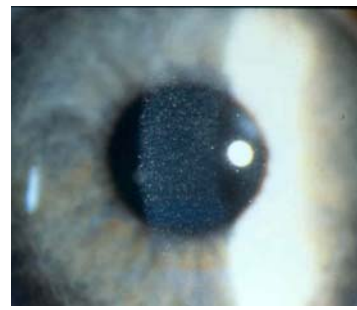
**0.75**



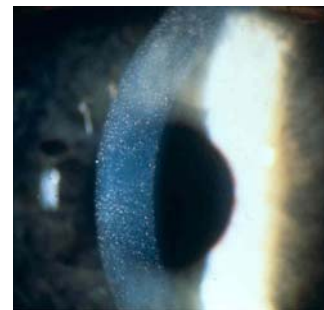
**1.00**



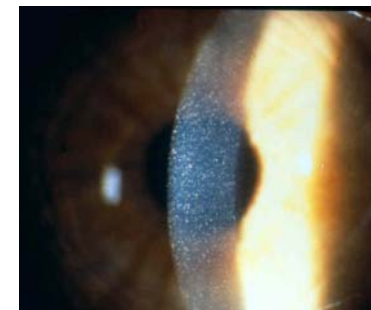
**1.25**



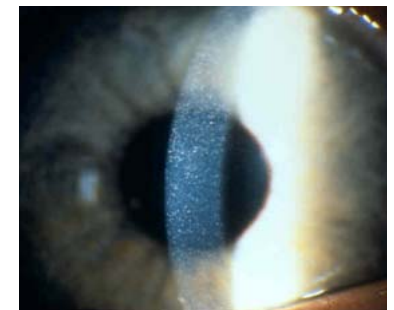
**1.50**



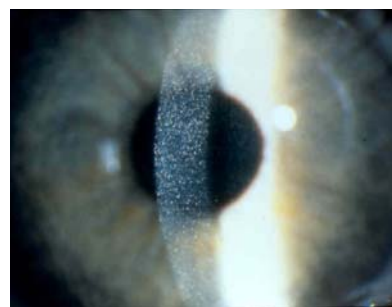
**1.75**



**2.00**



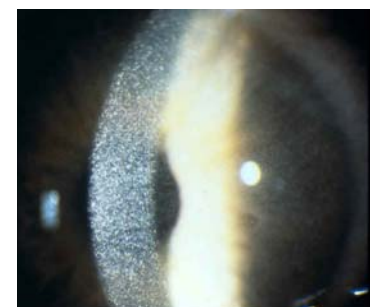
**2.25**



**2.50**

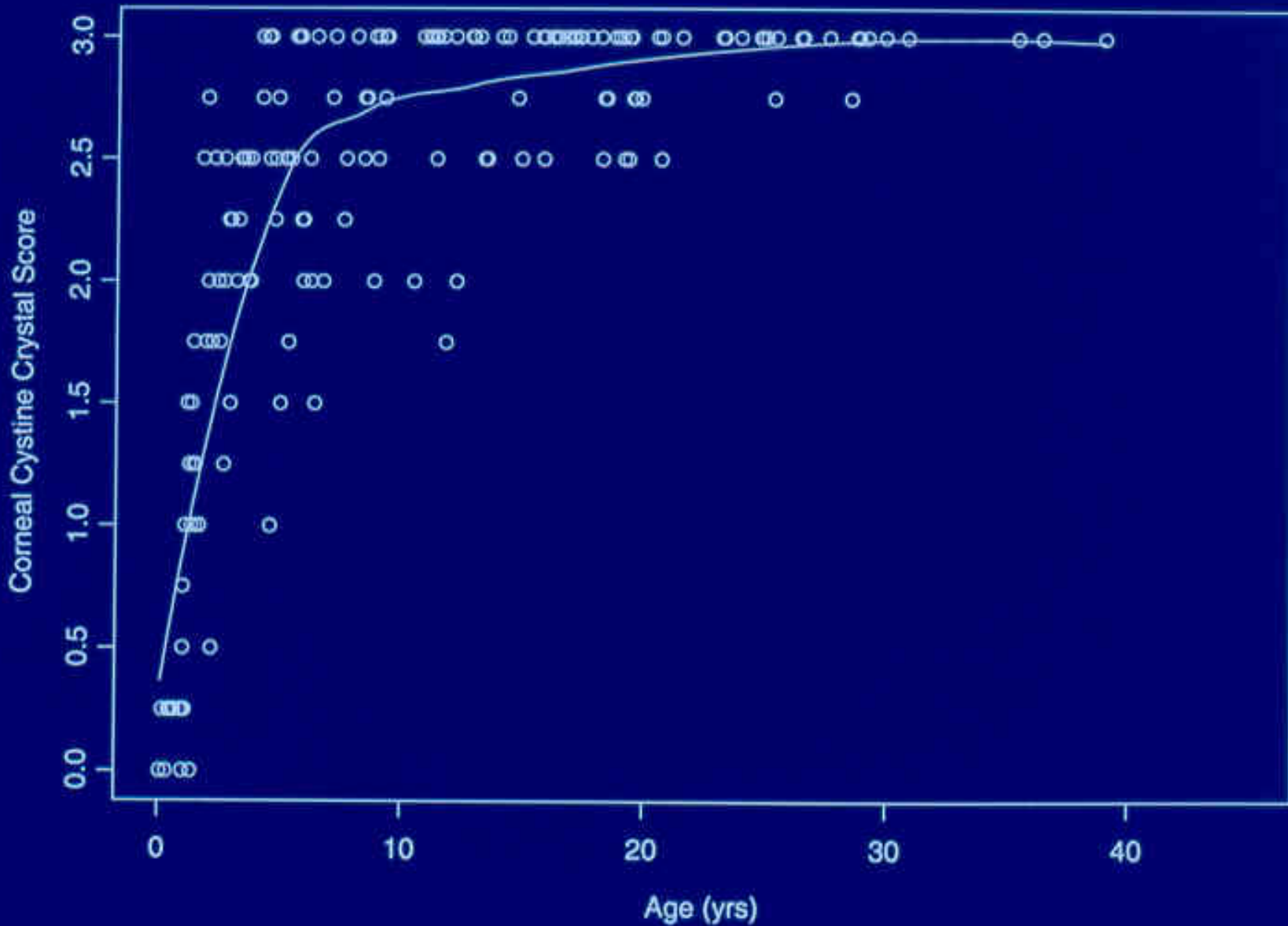


**2.75**



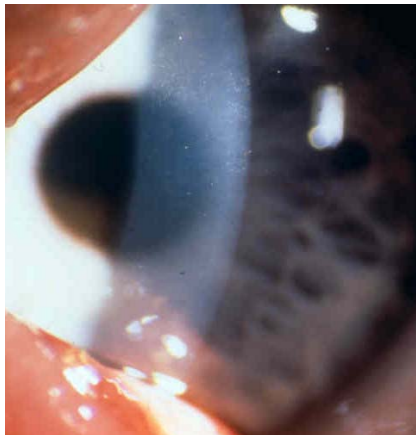
**3.00**

# Corneal Crystal Accumulation



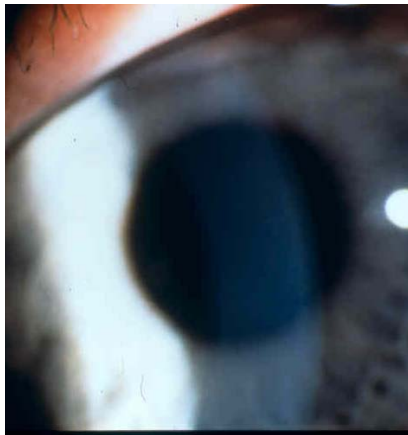
# Cysteamine Eyedrop Therapy

*12 mo*



**0.25**

*20 mo*



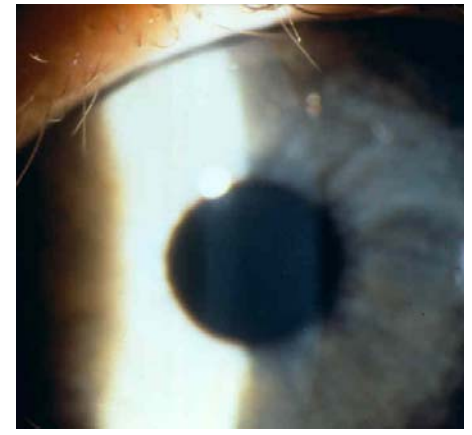
**0.00**

*32 mo*



**2.00**

*57 mo*



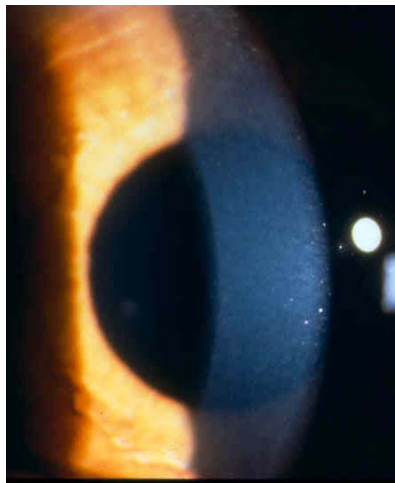
**0.00**

*15 mo*



**1.00**

*40 mo*



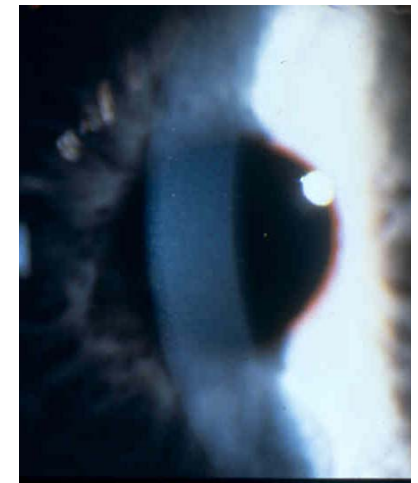
**0.00**

*43 mo*



**0.50**

*56 mo*



**0.00**

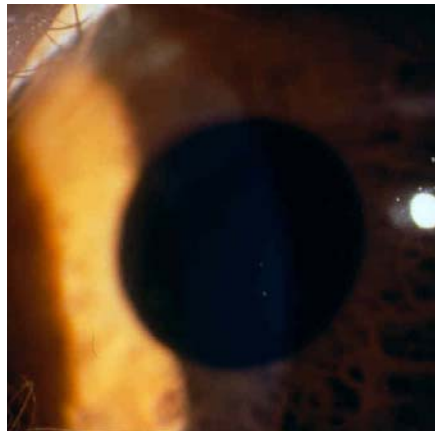
# Cysteamine Eyedrop Therapy

*69 mo*



**3.00**

*85 mo*



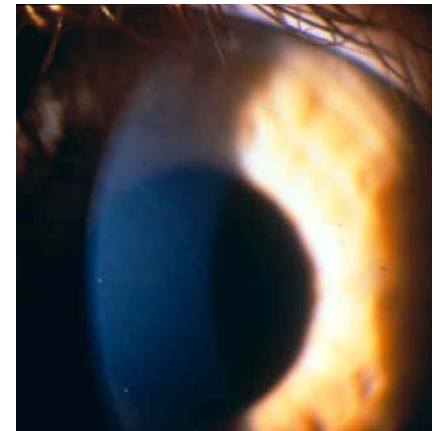
**0.00**

*133 mo*



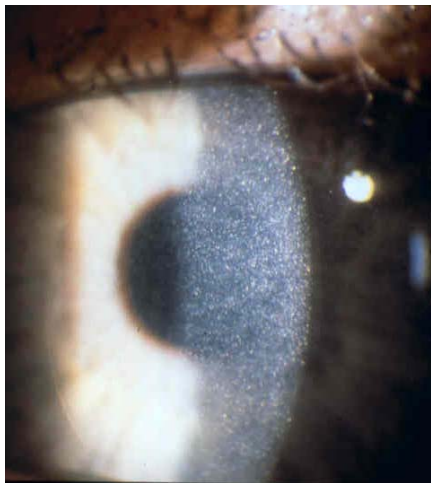
**3.00**

*157 mo*



**0.00**

*86 mo*



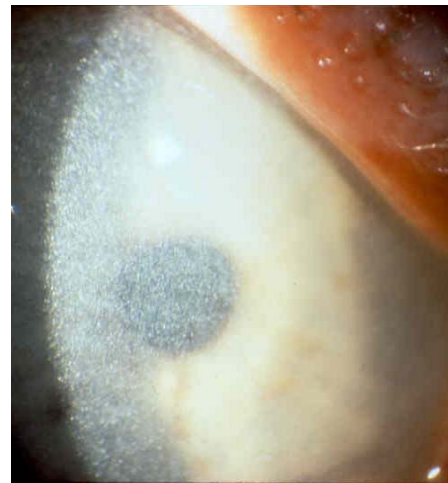
**3.00**

*109 mo*



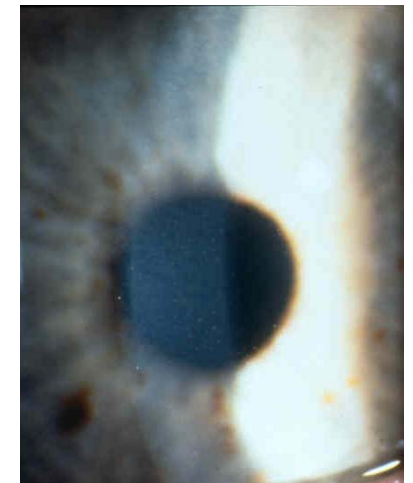
**0.00**

*237 mo*



**3.00**

*249 mo*



**0.25**

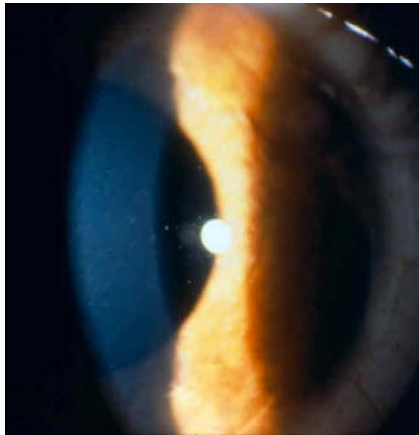
# Cysteamine Eyedrop Therapy

*262 mo*



**3.00**

*303 mo*



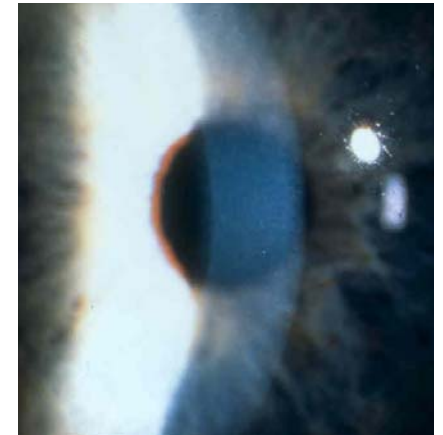
**0.25**

*342 mo*



**2.50**

*354 mo*



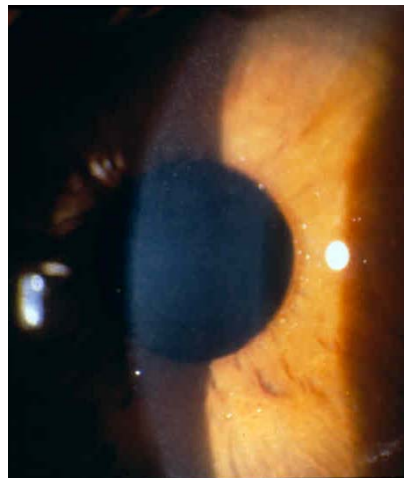
**0.25**

*304 mo*



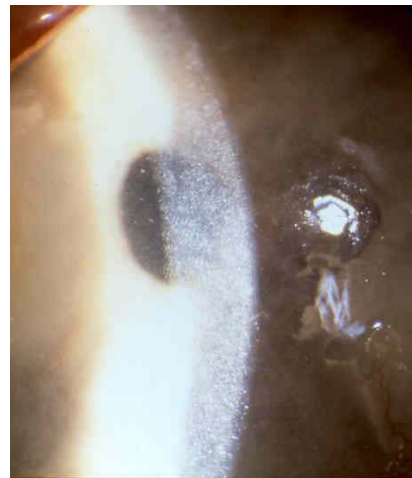
**3.00**

*316 mo*



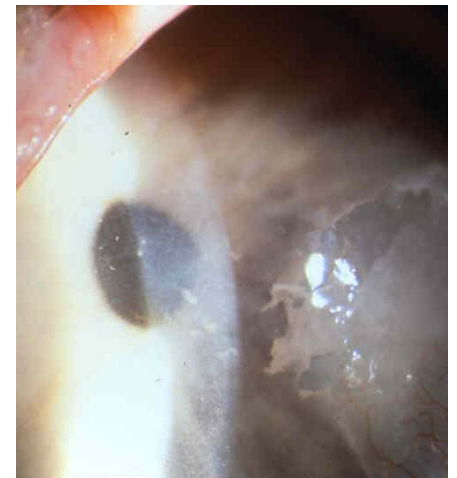
**0.25**

*394 mo*



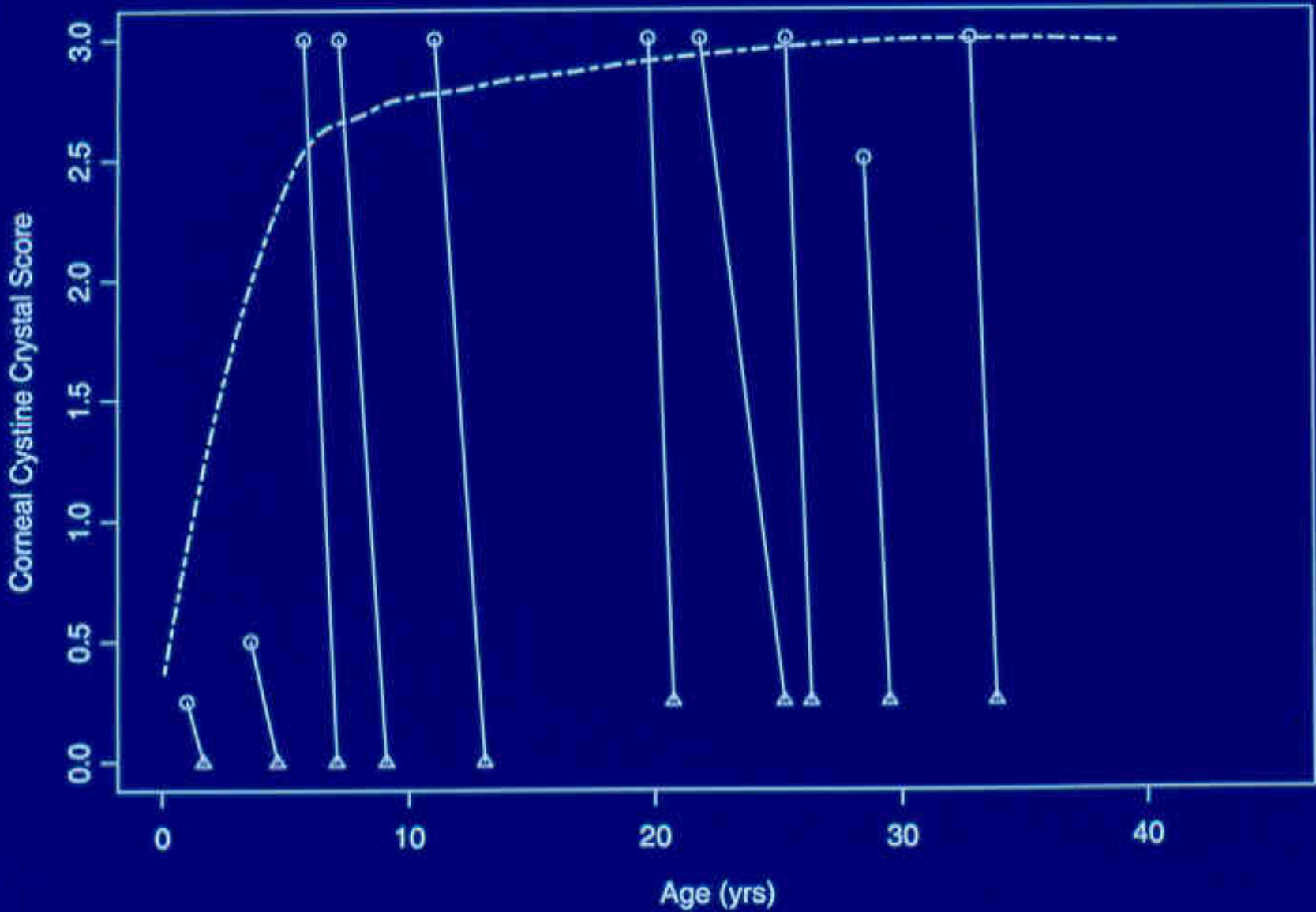
**3.00**

*406 mo*



**0.25**

# Cysteamine Eyedrop Therapy



# Cysteamine Eyedrops

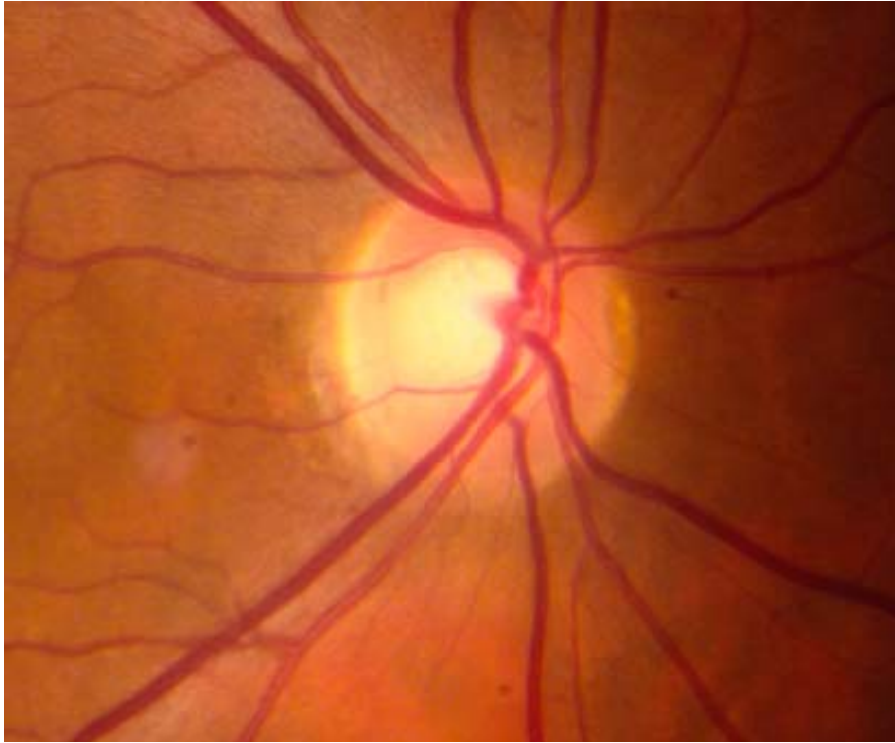
- **Dissolve corneal crystals if begun at any age.**
- **Cannot treat the band keratopathy once it occurs.**
- **Relieves the photophobia of cystinosis.**
- **Moving to New Drug Approval.**

# Eye Complications of Cystinosis

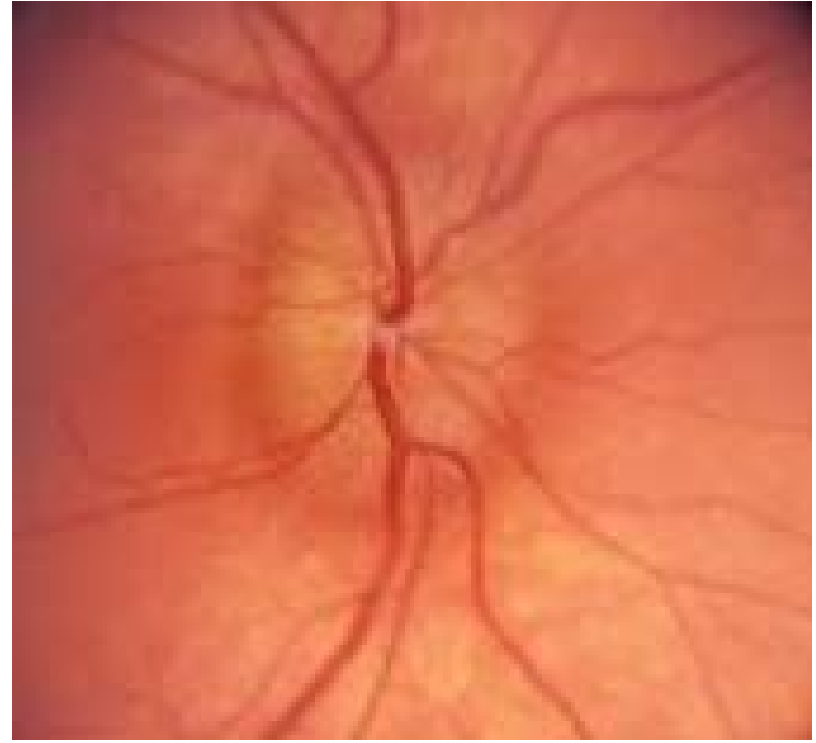
- **Background**
- **Posterior synechiae**
- **Anterior Segment Involvement**
- **Posterior Segment Involvement**

# Posterior Segment Involvement

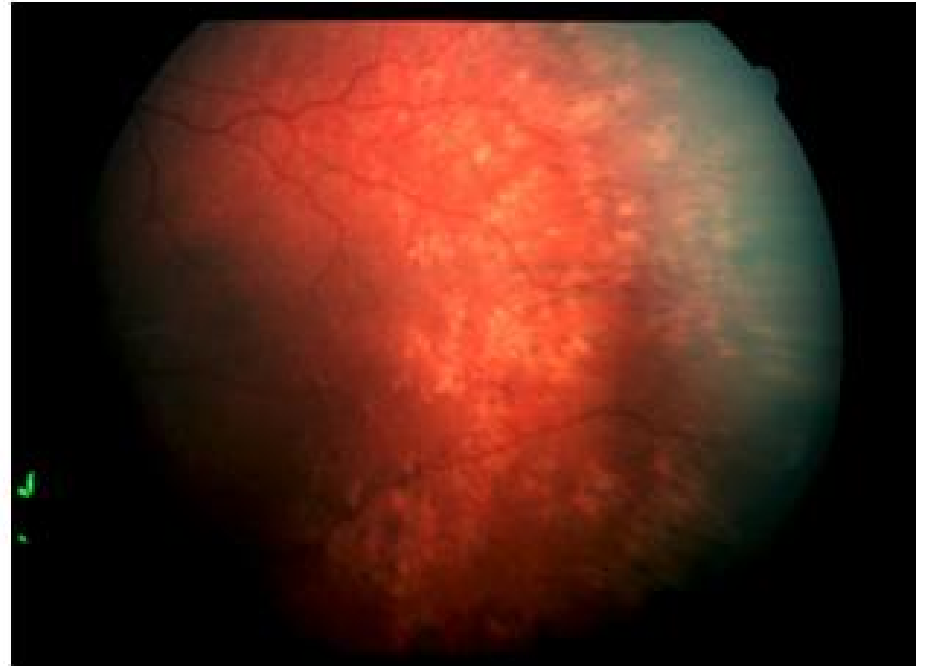
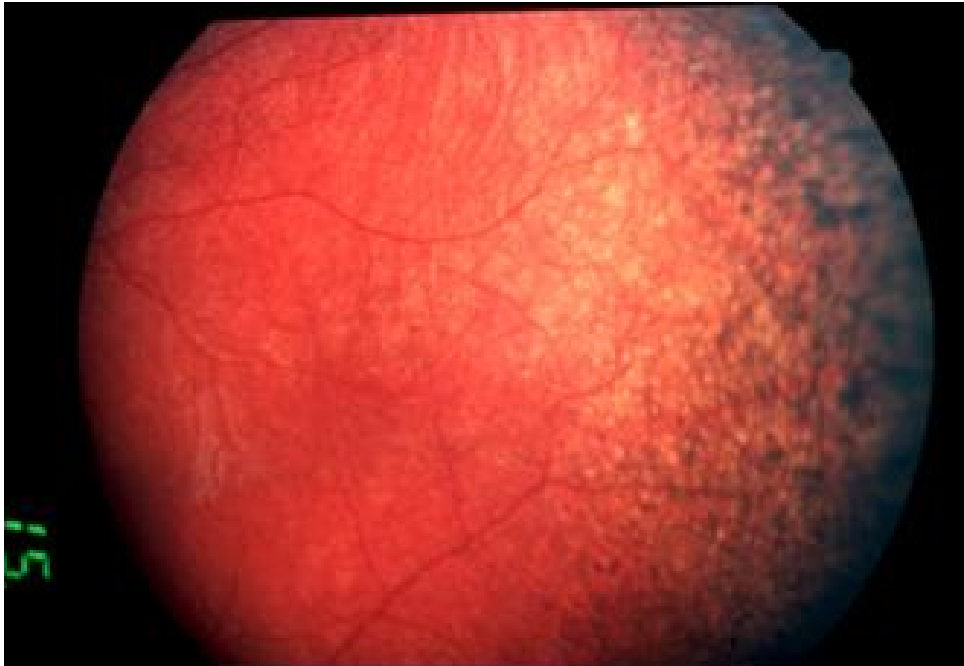
- **Optic disc (Pseudotumor Cerebri; Idiopathic Intracranial Hypertension)**
  - 8 cases
  - Related to cystinosis plus other risk factors
- **Retina**



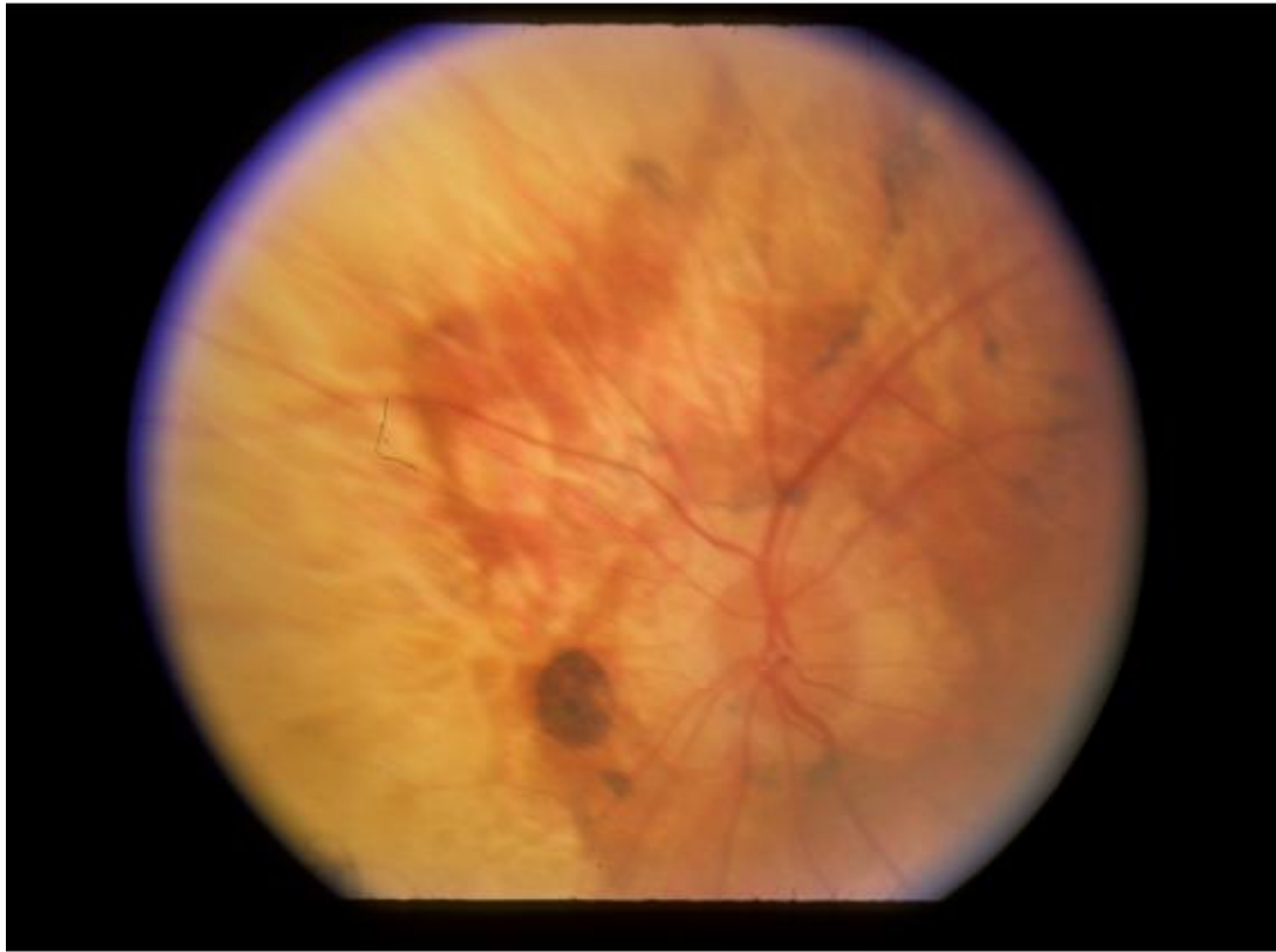
**Normal optic disc**



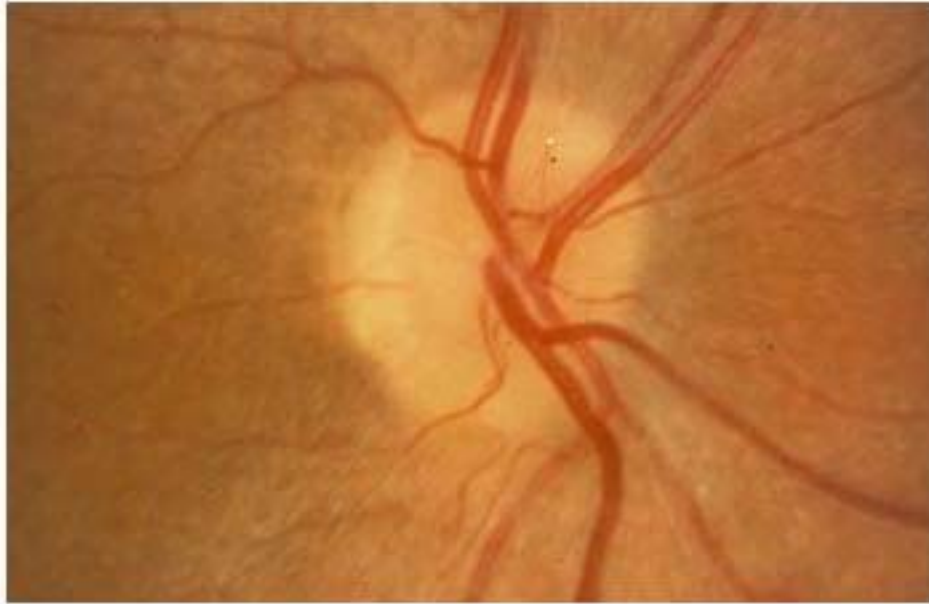
**Elevated optic disc of patient  
with pseudotumor cerebri**



**Peripheral Retinal Pigmentary Changes in  
Cystinosis**

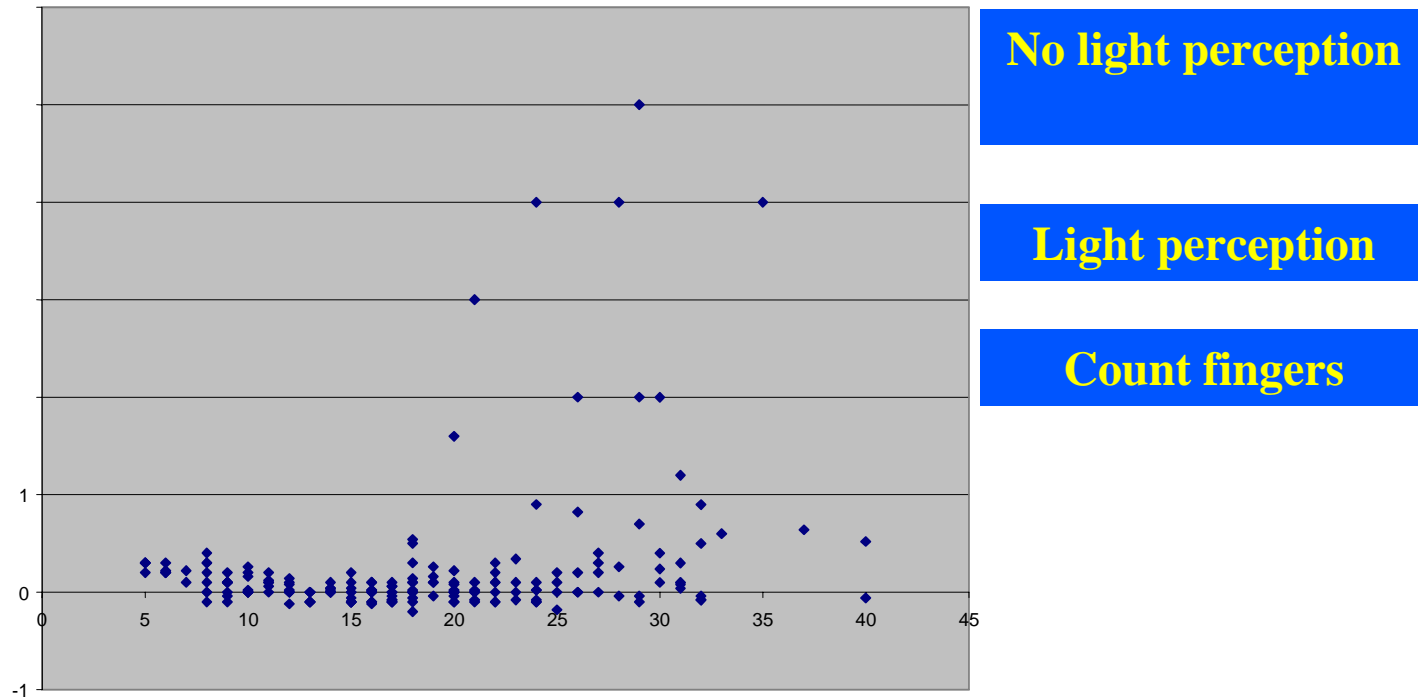


**Chorioretinal Atrophy**



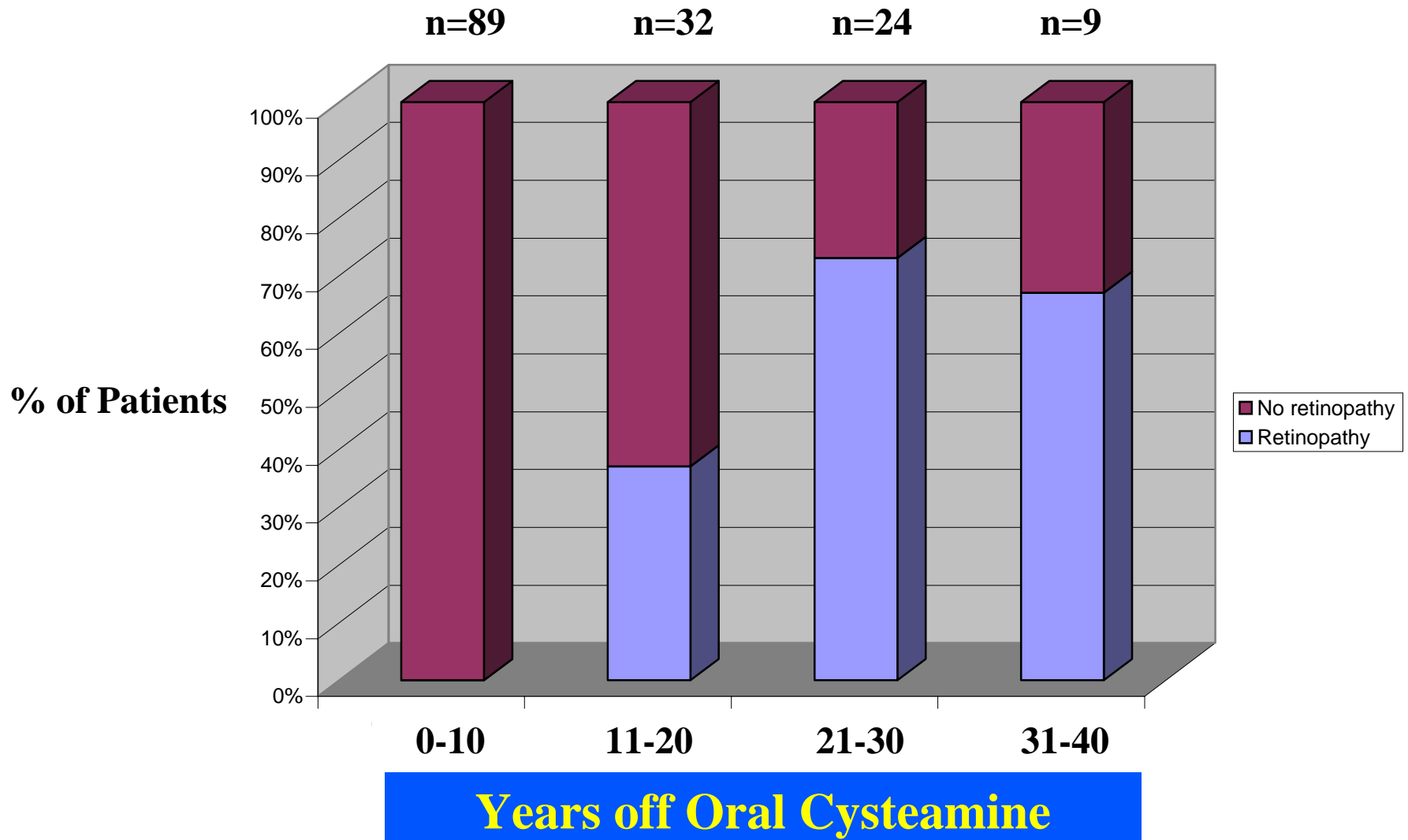
## Intraretinal Crystals

# Visual Acuity in logmar units

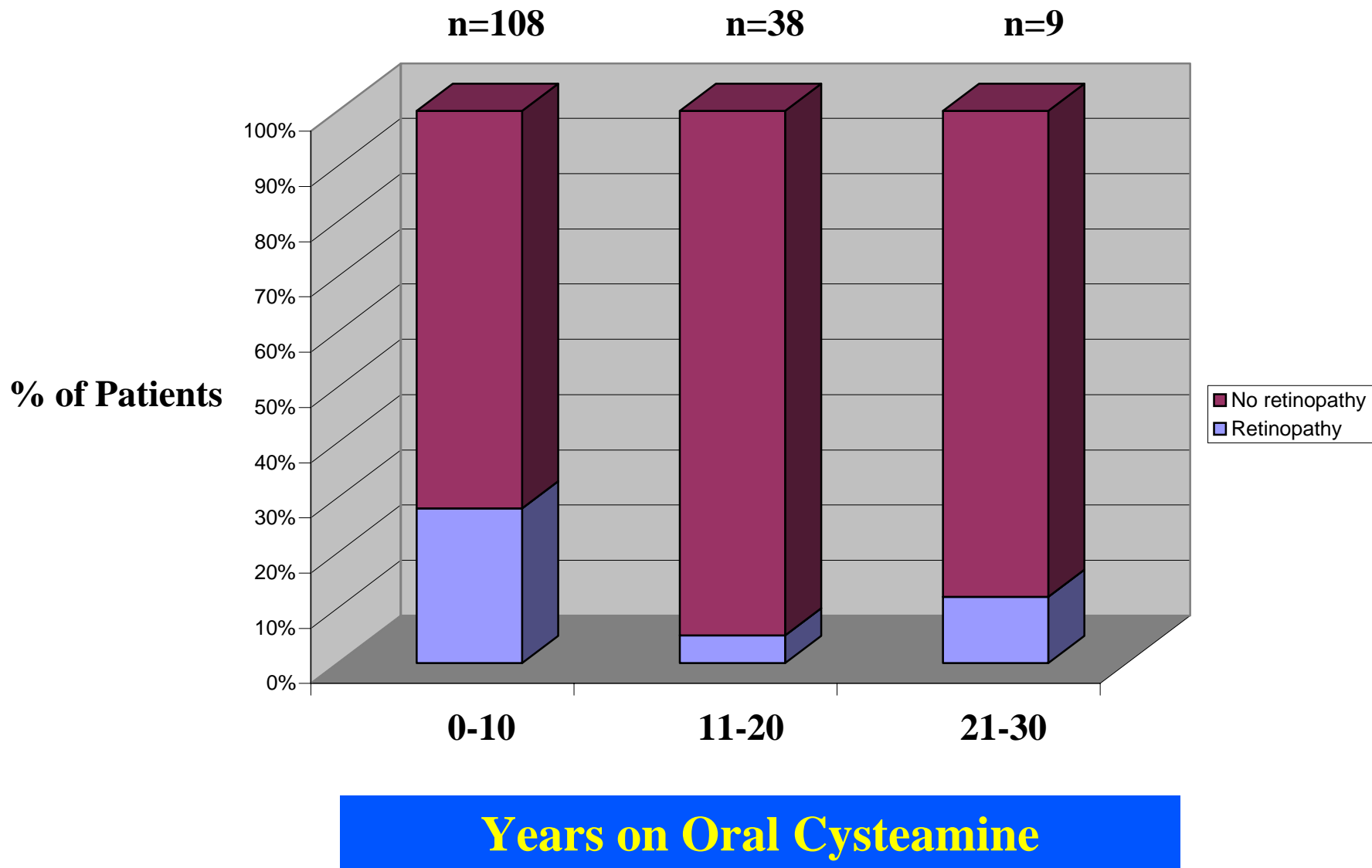


Age at last visit (y)

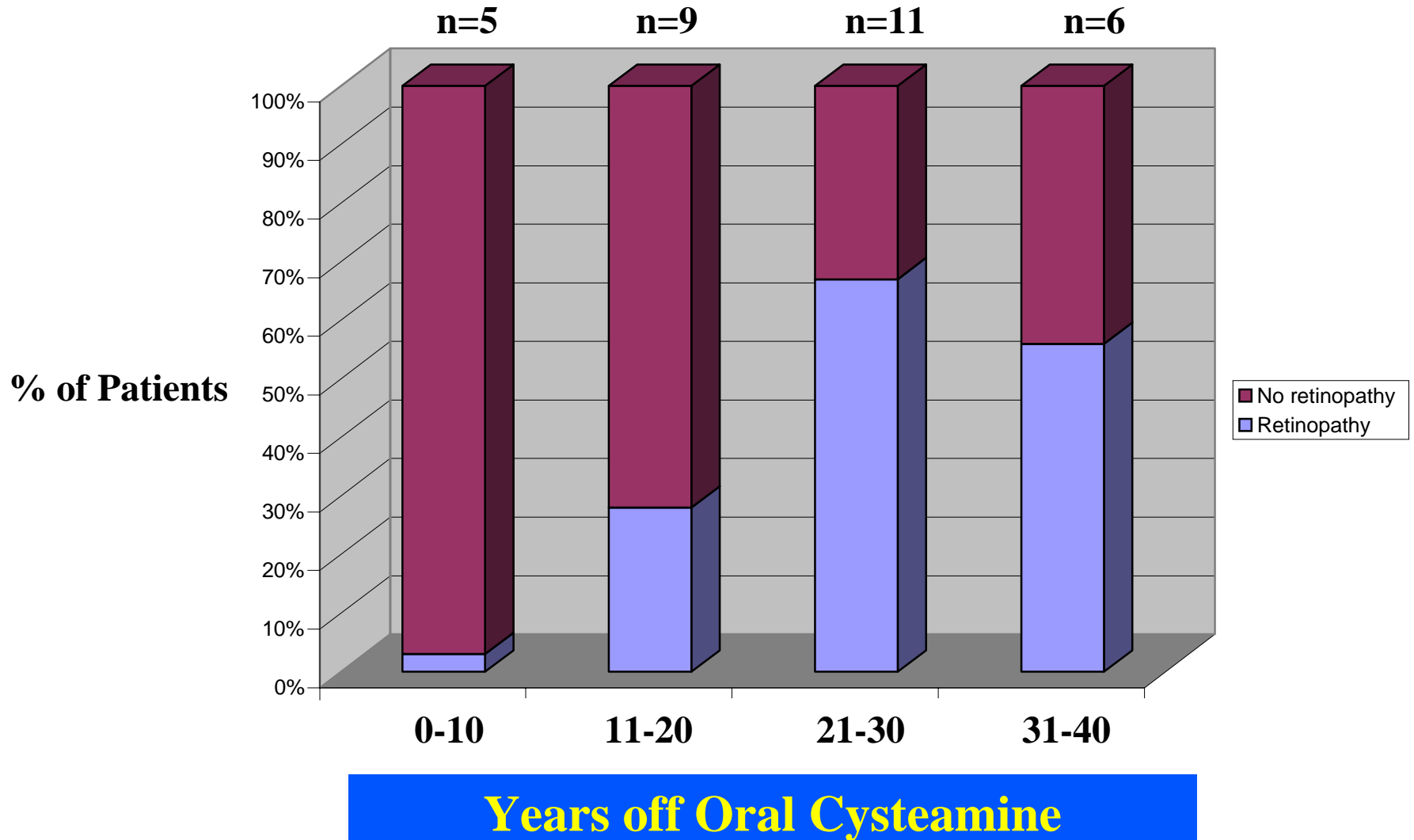
# Frequency of Clinical Retinopathy vs. Years off Cysteamine



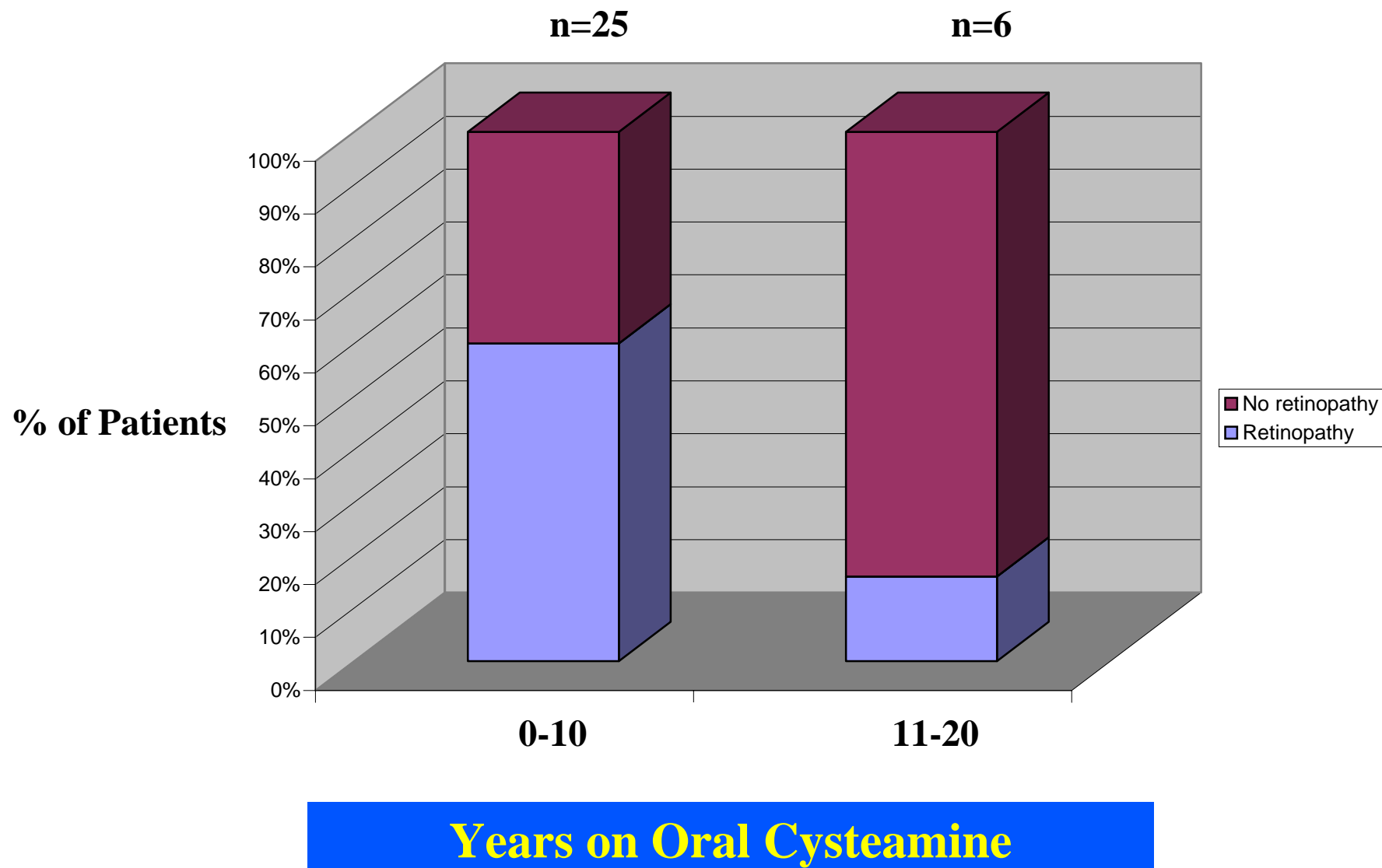
# Frequency of Clinical Retinopathy vs. Years on Cysteamine



# Frequency of ERG-proven Retinopathy vs. Years off Cysteamine



# Frequency of ERG-proven Retinopathy vs. Years on Cysteamine

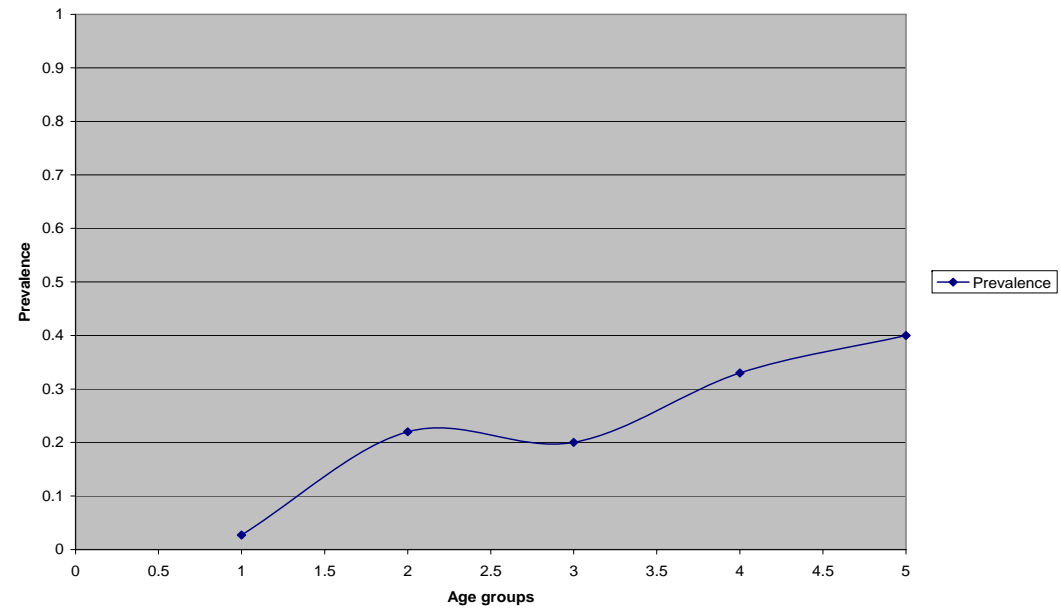


# Conclusions

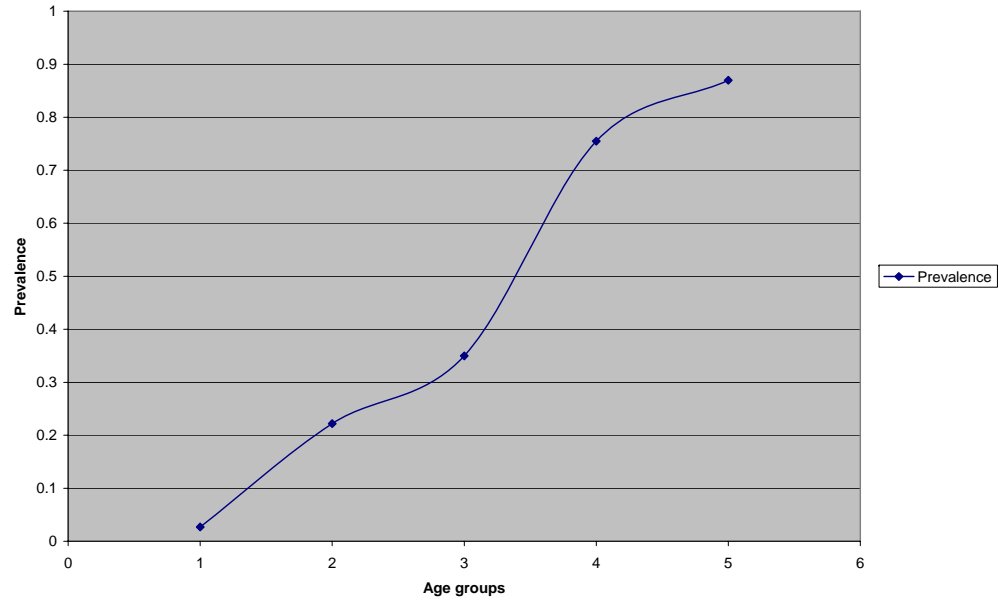
- **Take oral cysteamine to prevent the retinal complications of cystinosis.**
- **Take cysteamine eyedrops to dissolve the corneal crystals of cystinosis.**



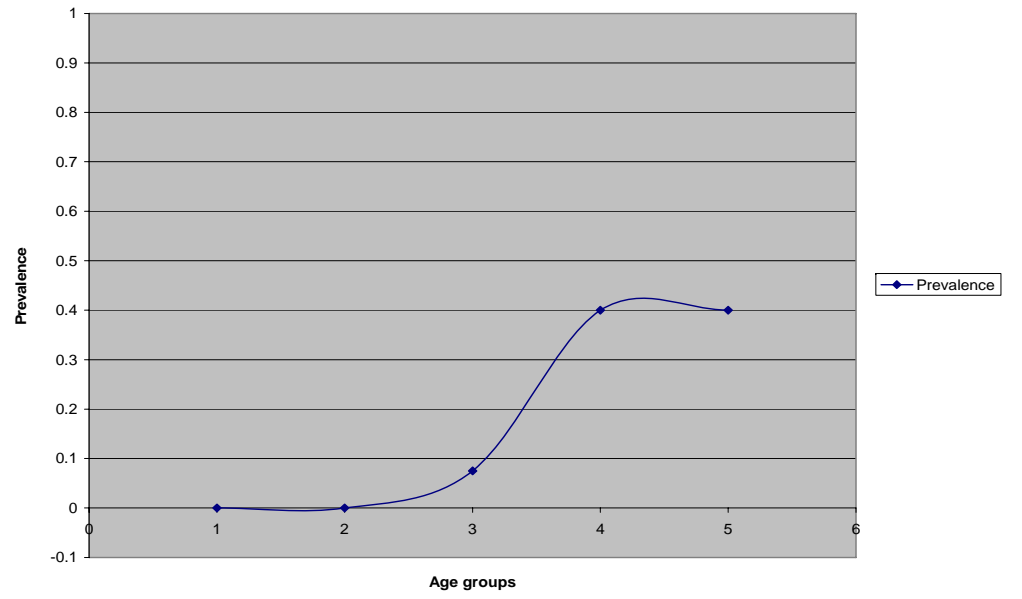
### Superficial punctate keratopathy



Anterior segment complications



Iris abnormalities



Peripheral neovascularization

