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Corneal dystrophies: Differential diagnosis and management

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Abstract

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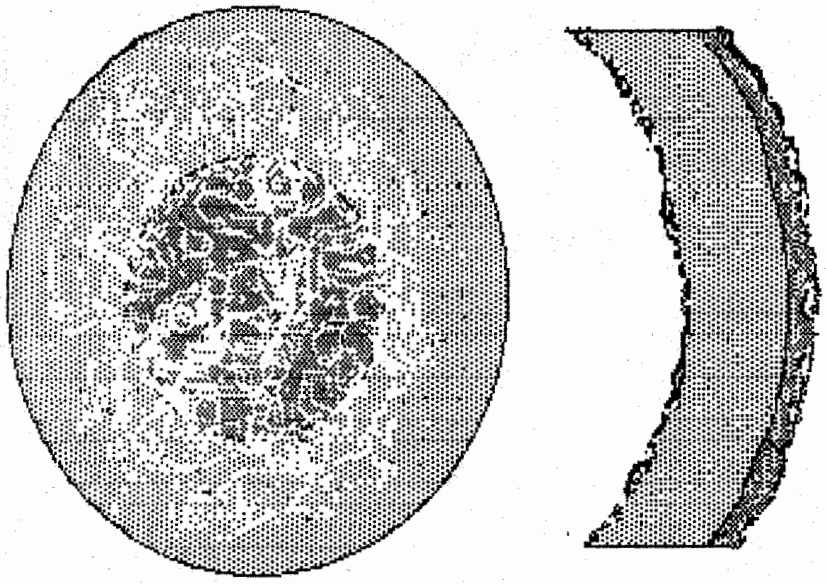

CORNEAL DYSTROPHIES
DIFFERENTIAL DIAGNOSIS AND MANAGEMENT

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INTRODUCTION

This handbook is intended as a quick-reference guide to aid the clinician in identifying the various corneal dystrophies. Because some corneal degenerations can be confused with actual dystrophies, they will also be included. But, what is the difference between the two?

Dystrophies are primary diseases of the cornea, not associated with a prior inflammation or systemic disease, though secondary inflammation and vascularization may mask them. Most are autosomal dominant, bilateral, and appear before age twenty. They usually affect one layer and are slowly progressive.

Degenerations, conversely, occur secondarily to other corneal disorders or to normal aging. They may sometimes resemble a dystrophy, especially in rare familial forms. Most degenerations appear after the age of forty.

In the interest of consistency and ease of identification, each dystrophy will be summarized on one page in a "SOAP" format along with an example drawing on the following page. Photographic slides, located in at the end of the volume, of actual cases will further aid in the identification of a dystrophy. SOAP is an acronym and is defined below:

S: Subjective - the subjective aspect of the dystrophy; what the patient experiences.

O: Objective - the objective aspect of the dystrophy; what you as the eye care specialist observe.

A: Assessment - the differential diagnosis.

P: Prognosis - the progression and management of the disease and how it will affect the patient.

TABLE OF CONTENTS

*most common vision threatening dystrophies
¥most common dystrophies that do not affect vision
†inheritance pattern not established; more likely a degeneration than a dystrophy

Bleblike Dystrophy

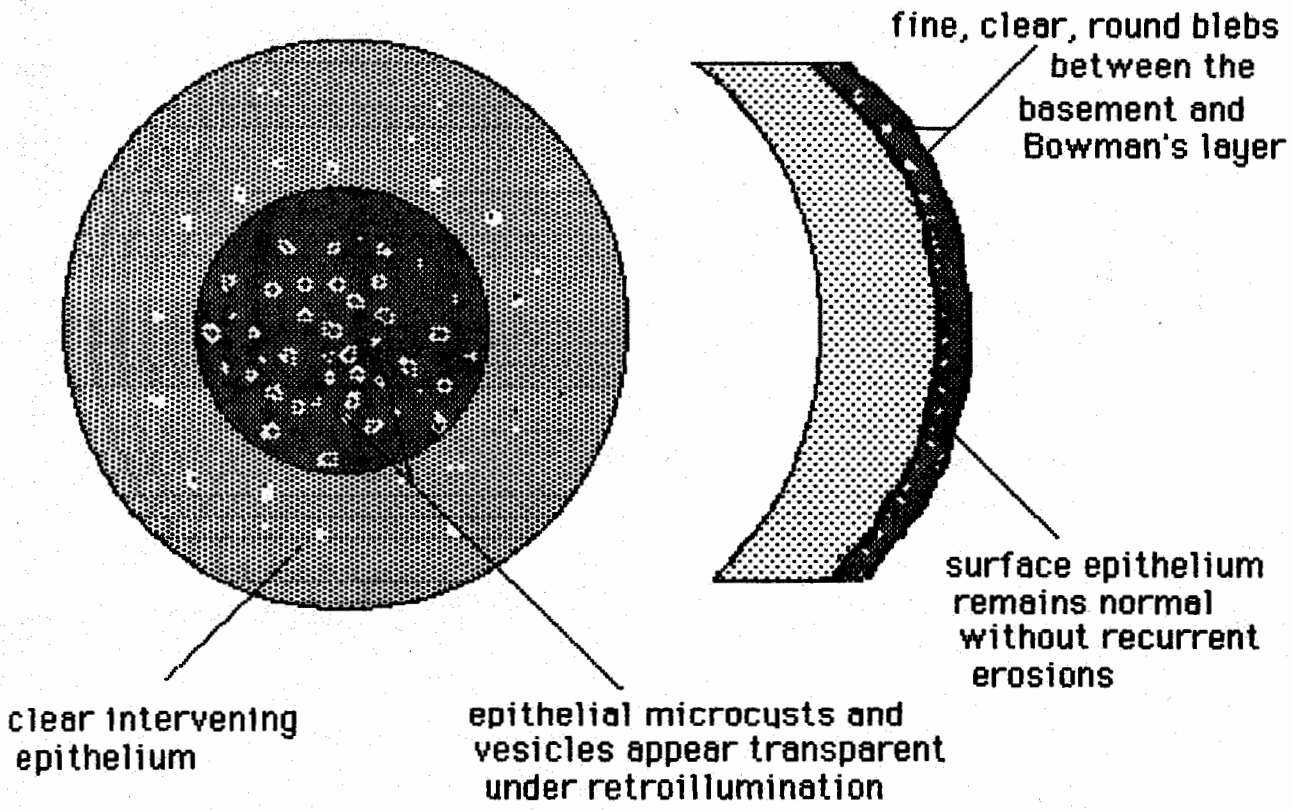
S a. **VA** - Not usually affected.
b. **Corneal Sensation** - Remains normal. Patient is usually asymptomatic.

O: a. **Part of layer in which dystrophy appears** - Bowman's layer and the basement epithelium are affected.
b. **Dystrophy Appearance** - Observed under retroilluination, the dotlike lesions appear as fine, clear, round blebs between the basement and Bowman's layer. These lesions do not result in surface epithelial erosions unless associated with Map-Dot-Fingerprint Dystrophy. The blebs are <u>not</u> psuedomicrocysts.
c. **Appearance of Other Layers** - Normal.

A: a. **Age of Onset** - Middle to late adulthood.
b. **Genetic Expression** -Autosomal Dominant. Bilaterally symmetrical.
c. **Association with Other Ocular or Systemic Diseases** - Blebs may appear with Map-Dot-Fingerprint Dystrophy.
d. **Resembles/Confused With** - Meesman's Corneal Dystrophy, which can be differentiated from Bleblike Dystrophy by it's decreased corneal sensation, reduced visual acuity, and occurence during childhood (1st year of life).

P: a. **VA Progression** - Visual acutiy remains normal throughout life.
b. **Management Strategy** -None required.
c. **Overall Prognosis** - Good. Asymptomatic patients with normal visual acuity and corneal sensation.

Bleblike Dystrophy



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MEESEMAN'S DYSTROPHY
Juvenile Corneal Epithelial Dystrophy, Stocker-Holt, Hereditary Epithelial Dystrophy

S **a. VA** - Not normally affected but may decrease with age and recurrent epithelial erosions.

b. Corneal Sensation - May be reduced. Recurrent erosions produce symptoms of lacrimation, photophobia, ocular pain, irritation, and foreign body sensation.

O: **a. Part of layer in which dystrophy appears** - Epithelium. Bowman's layer may be involved in advanced cases.

b. Dystrophy Appearance - Epithelial microcysts and vesicles appear within the epithelium during the first year of life and spread throughout the cornea by middle age. Opacities are regular in shape and size with clear intervening epithelium and may be arranged in a swirl or wedge-shaped pattern. Intraepithelial cysts appear as small clear to gray-white blebs under direct focal illumination. Tiny, fine, punctate, transparent vacuoles appear when utilizing retroillumination. Cysts that reach the surface rupture and take up fluorescein stain. The surface epithelium may be rough and irregular. Basement membrane often appears thick with finger-like projections extending into the basal epithelium. Bowman's membrane remains normal, however, in advanced cases opacification of Bowman's membrane can occur.

c. Appearance of Other Layers - Opacification of Bowman's membrane may occur. Other layers remain normal.

A: **a. Age of Onset** - Vesicles appear within the epithelium during the first year of life and patients remain asymptomatic until their 4th or 5th decade.

b. Genetic Expression - Autosomal Domininat, bilaterally symmetrical, with incomplete penetrance. The condition is observed between one and two years of age.

c. Association with Other Ocular or Systemic Diseases - None reported.

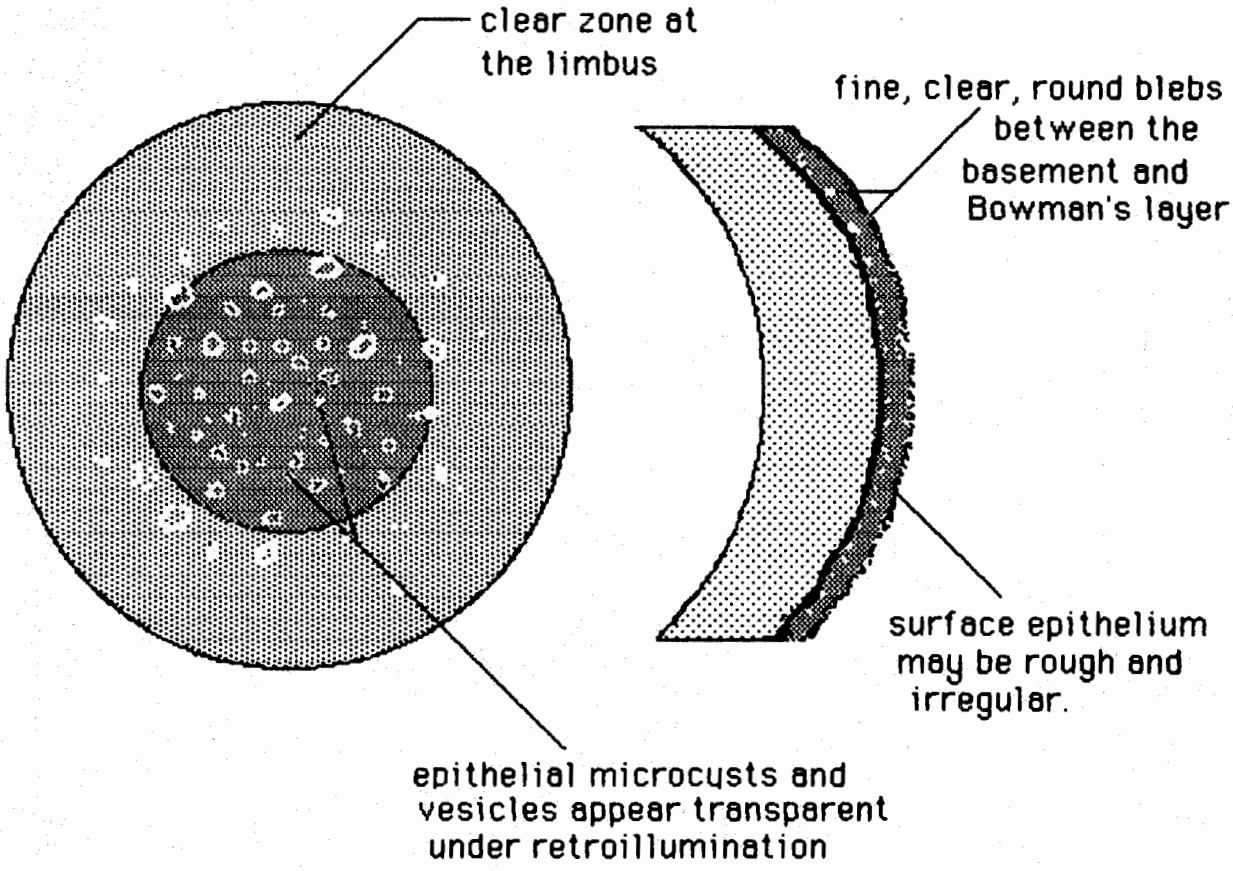
d. Resembles/Confused With - Vernal conjunctivitis, Meibomian gland disease, insensitive cornea, a reduced tear production with healing corneal erosions, mild epithelial edema, and bleb pattern dystrophy must be excluded.

P: **a. VA Progression** - Acuity remains normal in young individuals but may becomes progressively worse during the fourth and fifth decade of life when a spread of vesicles throughout the cornea creats irregular astigmatism, transient blur, and discomfort when cysts break the epithelial surface.

b. Management Strategy -Often unnecessary due to minimal acuity loss. In advanced cases with cicatrization, corneal haze, opacities in Bowman's membrane, and decreased visual acuity. Lamellar or penetrating keratoplasty is attempted but does not seem to help. Epithelial debridement is of little value. Superficial keratectomy may result in reepithelization without recurrence.

c. Overall Prognosis - Good.

Meesman's Corneal Dystrophy



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Dot-Fingerprint-Map Corneal Dystrophy

Cogan's Microcystic dystrophy--"Dot," Anterior Basement Membrane Dystrophy, Lacunar Dystrophy of Bettl-- "Map," Guerry Dystrophy--"fingerprint"

S: **a. VA** - Reduced.

b. Corneal Sensation - Most patients remain asymptomatic throughout life, however, ten per cent develop recurrent erosions causing severe pain on awakening, photophobia, spontaneous irritation, and severe blepharospasm.

O: **a. Dystrophy Appearance** - The epithelium appears loose and wrinkled with eventual peeling off of areas with abnormal basement membrane. Epithelial microcysts and vesicles appear as "Map, Dot, and Fingerprint" patterns and are best seen with broad tangential and/or retroillumination through a dilated pupil against a "red" fundus reflex. A brownish granular edema occupies the underlying anterior stroma and there is rapid tear breakup over suspicious areas of abnormal basement membrane. Microcysts may coalesce with other cysts and break the surface. Maps are gray geographic patches (or sheets) with oval clear zones and white intraepithelial opacities. Fingerprints represent fine, parallel, subepithelial lines seen in retroillumination only. There are two types of Dots; large gray ameba, round, or comma-shaped opacities and/or small, clear, round blebs clustered together in the pupillary zone.

b. Appearance of Other Layers - A brownish granular edema may occupy the anterior stroma. Other corneal layers remain normal.

c. Layer in which dystrophy appears - Epithelial basement layer and Bowman's.

A: **a. Age of Onset** - Disease onset is during adulthood and may affect white women over 30 more than any other group.

b. Genetic Expression - Autosomal dominant.

c. Association with Other Ocular or Systemic Diseases - None.

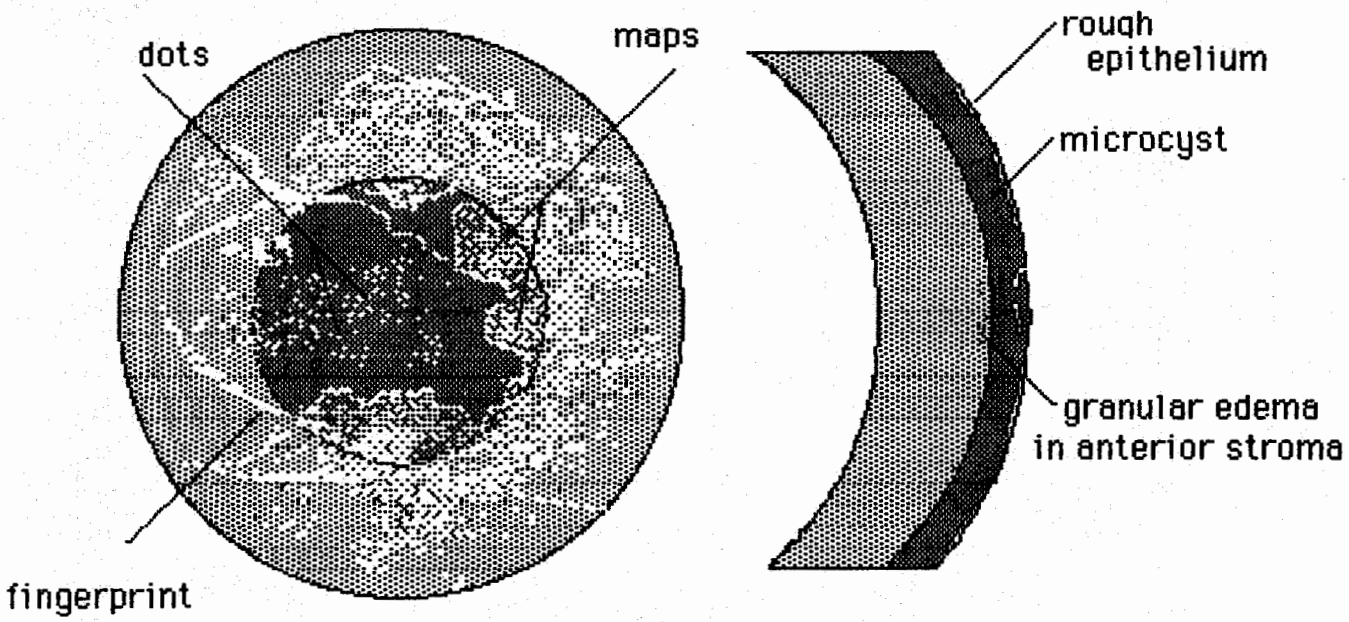
d. Resembles/Confused With - Recurrent erosions, Reis-Buckler's Dystrophy.

P: **a. VA Progression** - Most patients (90%) remain asymptomatic, however, ten percent develop blurred vision from epithelial edema and irregular astigmatism.

b. Management Strategy - Incapacitating epithelial erosions make management difficult. Topical 5 per cent sodium chloride drops administered 6 to 8 times a day and ointment at night may decrease epithelial edema and irregularity. Moderate conditions may require hypertonics, a cycloplegic agent, prophylactic antibiotic, pressure patch for 48 hours, and systemic analgesics. Severe conditions often require additional pressure patching every other day for a week. Debridement is effective when done in an eccentric pattern to prevent central corneal irregularity at the visual axis when healing is complete. See Appendix 1, erosion therapy, for detailed information.

c. Overall Prognosis - Erosions usually stop one to three years after onset of the disease, however, spontaneous recurrences are not uncommon.

Dot-fingerprint-map



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Reis-Bucklers's Corneal Dystrophy

Bucklers' Annular Dystrophy, Ring Dystrophy of Reis-Buckler

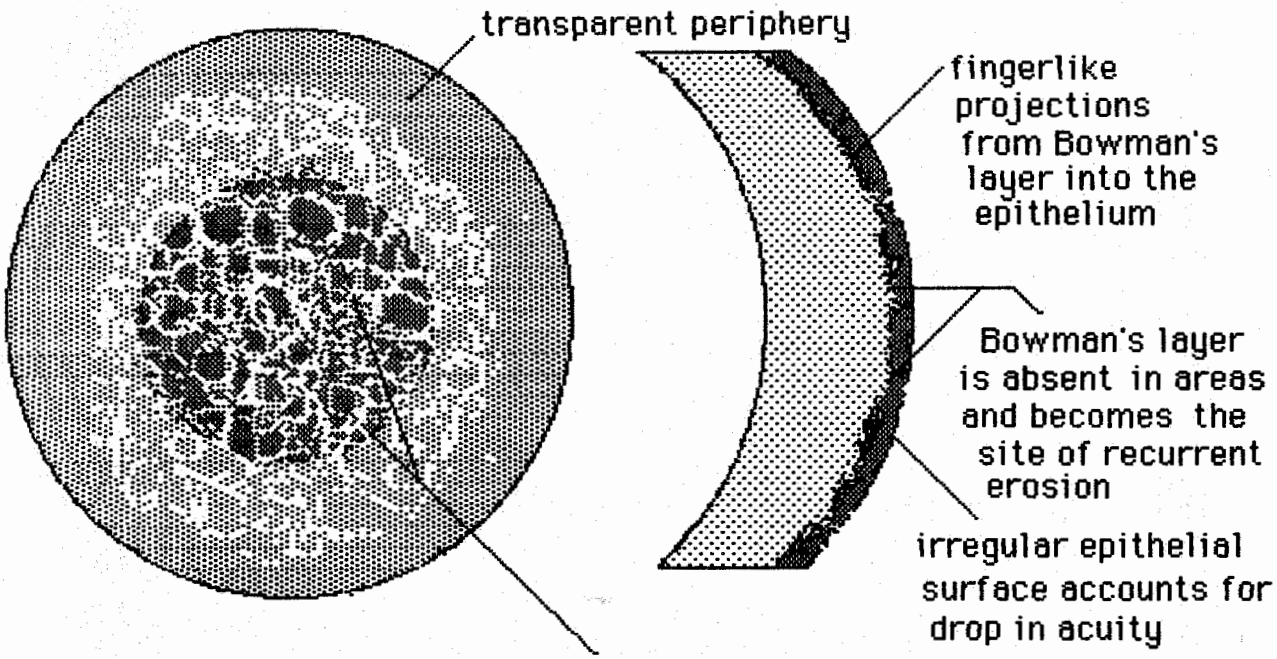
S: **a. VA** - Decreased. Deteriorated vision during twenties.
b. Corneal Sensation - Decreased. Recurring attacks of erosion resulting in photophobia, foreign body sensation, injection, and pain lasting for several weeks.

O: **a. Layer in which dystrophy appears** - Bowman's layer with fingerlike projections into the epithelial layer.
b Dystrophy Appearance - The corneal surface becomes rough and irregular with a loss of transparency. Opacities are superficial, reticulated, ring-shaped, gray-white defects of various sizes and shapes occuring in the midperiphery with the extreme periphery remaining transparent. The corneal defects are interwoven into a geographic-like surface giving the cornea a frosted glass, fishnet, or curdled milk apperance. Bowman's layer is absent in many areas and becomes the site of recurrent erosions.
c. Appearance of Other Layers - Limited to the epithelial and Bowman's layers. Other layers remain normal.

R: **a. Age of Onset** - Symptoms usually begin in childhood (age 5) with recurring erosions up to age 30. The disease condition worsens with age.
b. Genetic Expression -Autosomal dominant with strong penetrance.
c. Association with Other Ocular or Systemic Diseases -None.
d. Resembles/Confused With - Honeycomb dystrophy; Grayson-Wilbrandt (Anterior Membrane Dystrophy), which does not manifest itself until age 10 and is confined to the basement epithelial membrane in eyes with normal corneal sensation.

P: **a. VA Progression** - Progressive loss of acuity with irregular astigmatism due to recurrent attacks of erosion. Visual acuity decreases during twenties. Corneal sensation is reduced.
b. Management Strategy - Topical 5 per cent sodium chloride drops administered 6 to 8 times a day, and ointment at night, may decrease epithelial edema and irregularity. Moderate conditions may require a cycloplegic agent, hypertonics, prophylactic antibiotic, pressure patch for 48 hours, and systemic analgesics. Severe conditions often require additional pressure patching every other day for a week. With penetrating keratoplasty, superficial opacities may recur in the graft. Superficial keratoplasty may offer some help. See Appendix 1.
c. Overall Prognosis - Deteriorated vision during twenties. Corneal sensation is decreased.

Reis-Buckler's Corneal Dystrophy



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Anterior Membrane Dystrophy

Grayson-Wilbrandt, Hereditary Anterior Membranous Dystrophy

S **a. VA** - Reduced. Epithelial surface appears irregular and accounts for individuals exhibiting visual acuities of 20/200.

b. Corneal Sensation - Normal. Occasional episodes of pain and injection with spontaneous epithelial breakdown can be expected.

O: **a. Part of layer in which dystrophy appears** - Confined to the basement membrane of epithelium.

b. Dystrophy Appearance - Epithelial surface appears irregular and accounts for the drop in acuity. Discrete gray-white macular opacities are seen at the level of Bowman's layer and extend toward the epithelial surface. Cornea between lesions remains clear. The destruction of Bowman's layer distinguish it from other anterior dystrophies. Diffuse superficial haze is present in the midperipheral region of the cornea.

c. Appearance of Other Layers -Epithelium appears rough.

A: **a. Age of Onset** - Adolescence. Does not occur until 10 years of age.

b. Genetic Expression - Autosomal dominant.

c. Association with Other Ocular or Systemic Diseases - None.

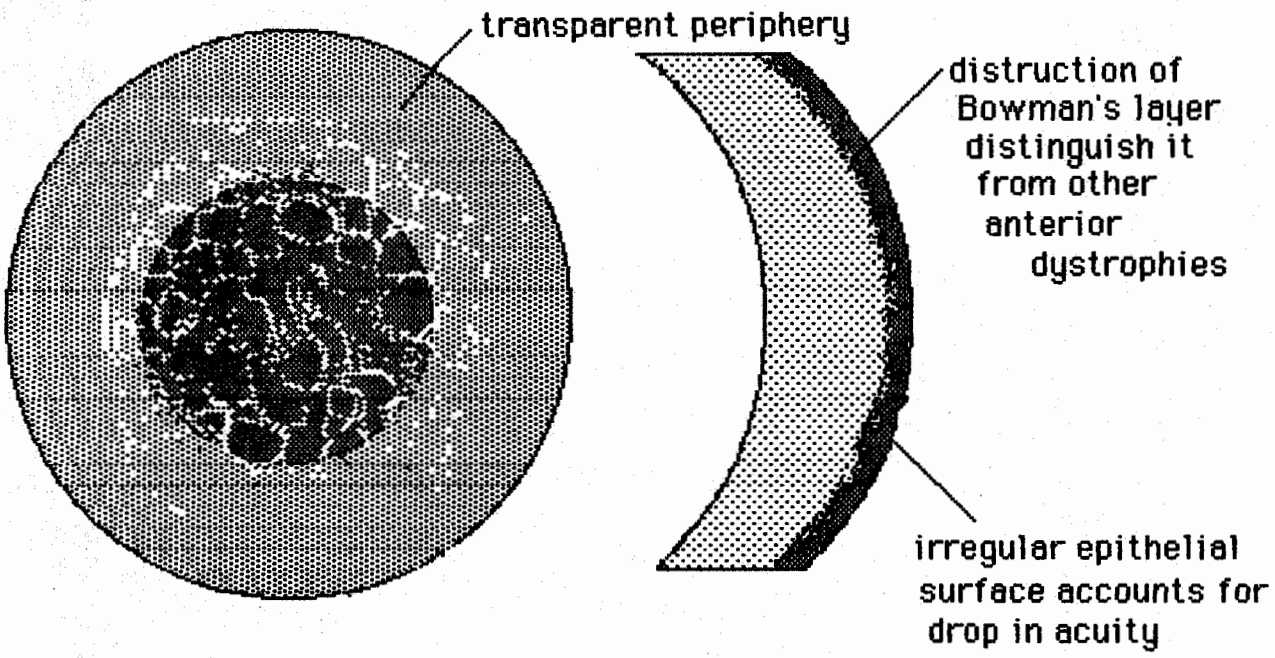
d. Resembles/Confused With - Differentiated from Reis-Buckler's by age of onset, variable visual acuity loss, normal corneal sensaiton, and only partial affection of the cornea.

P: **a. VA Progression** - Gradual decrease in vision.

b. Management Strategy - None usually required. Possible penetrating keratoplasty, although recurrences usually recur. Prescribe erosion therapy if indicated (appendix 1).

c. Overall Prognosis - Slow decrease in visual acuity (possibly down to 20/200). Recurrence of disease has been described in corneal grafts.

Anterior Membrane Dystrophy



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Duane, T. and E.A. Jaeger. Clinical ophthalmology. Harper and Row, Philadelphia, 1985. pp. 17-19.

Honeycomb Dystrophy
Thiel and Biehnke

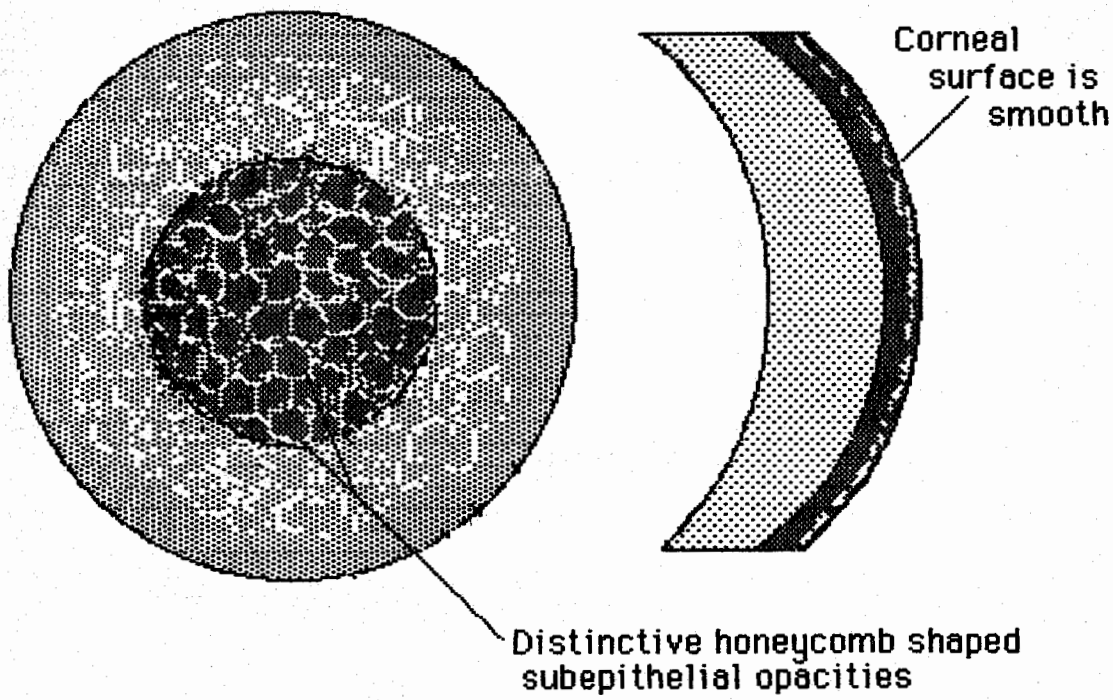
S: **a. VA** - Reduced.
b. Corneal Sensation - Is not reduced. Pain from corneal erosions.

O: **a. Layer in which dystrophy appears** -Subepithelial.
b. Dystrophy Appearance - The corneal surface is smooth. Opacities are distinctivly honeycomb shaped and are located in the subepithelial region with projections toward the surface epithelium. Progressive recurrent corneal erosions occur and are responsible for acuity loss.
c. Appearance of Other Layers - Normal.

A: **a. Age of Onset** -It begins in childhood with progressive recurrent corneal erosions that disappear by age thirty or fourty.
b. Genetic Expression - Autosomal Dominant. Bilaterally symmetrical.
c. Association with Other Ocular or Systemic Diseases -May be a variant of Reis-Bucklers' Corneal Dystrophy.
d. Resembles/Confused With - Reis-Bucklers' Dystrophy. Honeycomb Dystrophy is differentiated by the typical honeycomb appearance, the smooth corneal surface, normal corneal sensation, and less visual involvement than Reis-Bucklers' Dystrophy.

P: **a. VA Progression** -Visual acuities will range from 20/25 in young patients to 20/100 in older individuals in their 40's to 60's.
b. Management Strategy -None required. See appendix 1.
c. Overall Prognosis -Good.

Honeycomb Dystrophy



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Hereditary Calcific Band Keratopathy
Idiopathic Band Keratopathy

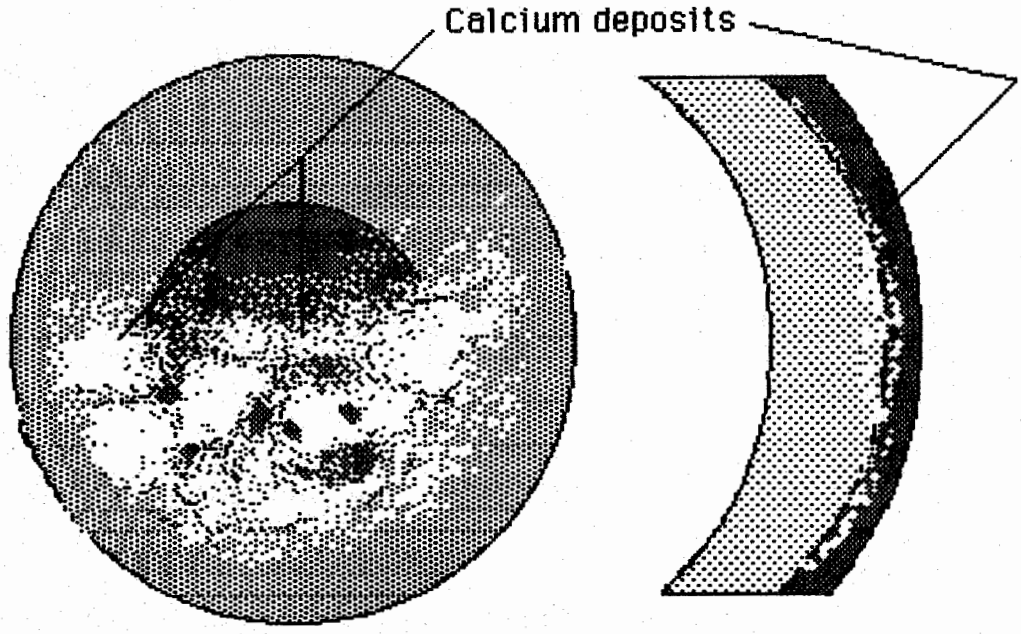
S: **a. VA** -Can be reduced.
b. Corneal Sensation -Normal.

O: **a. Layer in which dystrophy appears** -Epithelium, Bowman's layer, and superficial stroma.
b. Dystrophy Appearance - Deposits of calcium carbamate in Bowman"s layer, the basement membrane of the epithelium, and the superficial portions of the stroma are most often seen in the interpalpebral area. Cornea at the limbus is clear because Bowman's layer does not extend to the absolute limbus. Small holes in the calcific opacity are produced by corneal nerves penetrating Bowman's membrane, thus, a "Swiss-cheese" appearance is produced. Deposition of calcium usually occurs in the 3 and 9 o'clock meridians adjacent to the limbus.
c. Appearance of Other Layers -It may involve all layers of the cornea.

A: **a. Age of Onset** -Adulthood. The literature is not specific as to the mean age of this disease.
b. Genetic Expression -Autosomal Dominant.
c. Association with Other Ocular or Systemic Diseases - Sarcoidosis, Fanconi's disease, Still's Disease, hypercalcemia, multiple myeloma, discoid lupus, Hyperphosphatemia, Vitamin D toxicity, metastatic disease, Ichthyosis, chronic nongranulomatous uveitis (juvenile rheumatoid arthritis), prolonged glaucoma, degenerated globe, spheroid degeneration in band form, Norrie's disease, toxic mercury vapors, eyedrops containing mercurial drugs, and dry eyes with rapidly developing band keratopathy.
d. Resembles/Confused With - Other conditions causing band keratopathy are hypercalcemia, systemic-ocular disease, toxicity, and corneal irritations.

P: **a. VA Progression** -Can be reduced.
b. Management Strategy -If necessary, removal of the corneal epithelium utilizing 4% cocaine drops followed by ethylenediamine tetraacetic acid (EDTA) drops will act as a chelating agent so that the calcium may be scraped off. The eyes are then patched after cycloplegics and mild antibiotics are instilled.
c. Overall Prognosis - Good.

Hereditary Calcific Band Keratopathy



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Anterior Crocodile Shagreen
Mosaic Shagreen of Vogt

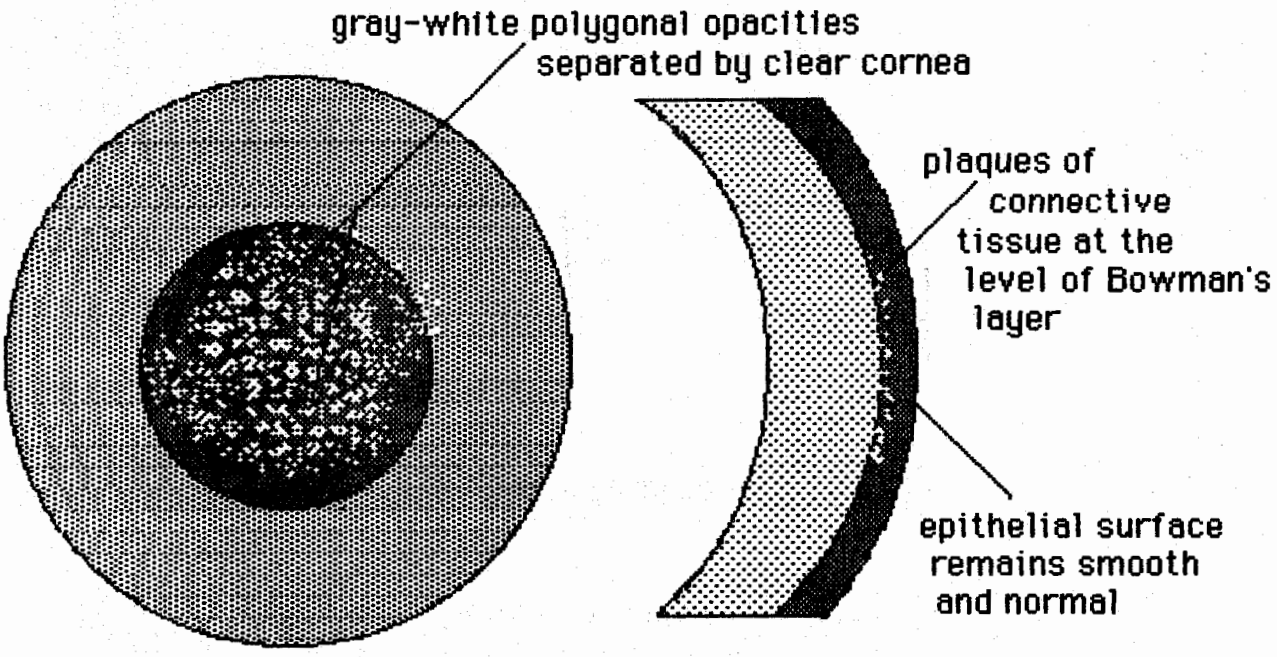
S: **a. VA** - Normal. Rarely are the opacities dense enough to cause decreased visual acuity.
b. Corneal Sensation - Normal corneal sensation.

O: **a. Layer in which dystrophy appears** - Bowman's membrane.
b. Dystrophy Appearance - Bilaterally symmetrical, central, gray-white polygonal opacities are separated by clear cornea at the level of Bowman's layer. This configuration gives the cornea a "crocodile-skin" appearance. The epithelial surface remains normal.
c. Appearance of Other Layers - Deep layers of the epithelium may be disrupted with connective tissue plaques.

A: **a. Age of Onset** - Late adulthood.
b. Genetic Expression - Most sources indicate there does not exist a mode of inheritance for this dystrophy, others suggest an X-linked recessive Inheritance pattern.
c. Association with Other Ocular or Systemic Diseases - Megalocornea, peripheral Calcific Band Keratopathy, and iris malformations
d. Resembles/Confused With - Often confused with "Anterior Corneal Mosaic" which is demonstrated after pressure is applied to the normal cornea.

P: **a. VA Progression** - Not usually affected. In rare instances, opacities become dense enough to produce decreased visual acuity.
b. Management Strategy - Not usually required. The need for keratoplasty is rare.
c. Overall Prognosis - Good. Normal corneal sensitivity with rare decrease of visual acuity.

Anterior Crocodile Shagreen



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Local Anterior Mucopolysaccharide Accumulation

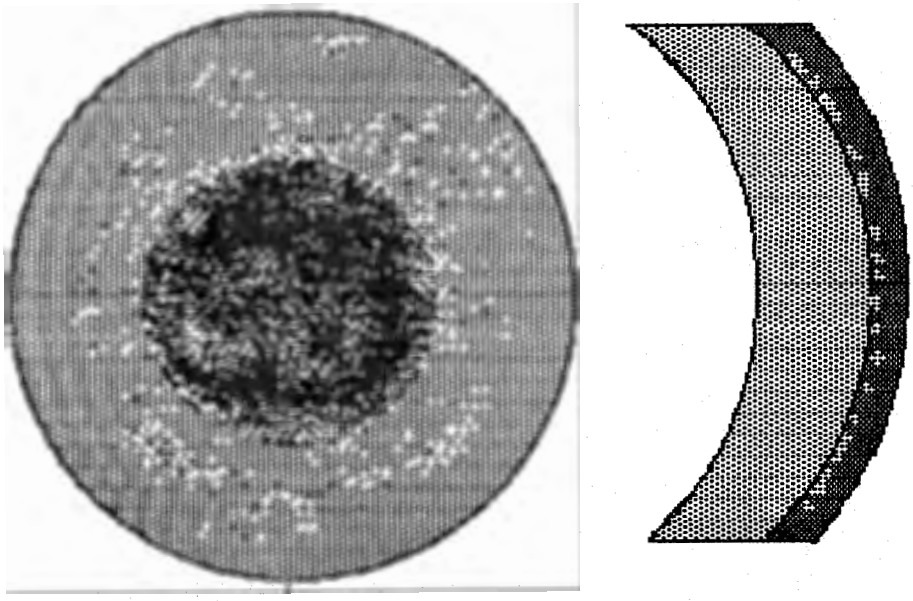
S: **a. UA** - May be reduced if opacities become dense.
b. Corneal Sensation - Decreased corneal sensitivity occurs without vascularization of the cornea.

O: **a. Layer in which dystrophy appears** - Bowman's layer.
b. Dystrophy Appearance - Diffuse corneal clouding at the level of Bowman's layer with an absence of cyst or vacuole formation. The corneal epithelium is normal. Acid mucopolysaccharide accumulation occurs in Bowman's layer without evidence of systemic mucopolysaccharidosis.
c. Appearance of Other Layers -The epithelium, endothelium, Descemet's membrane, and stroma are normal. Bowman's layer shows diffuse thickening.

A: **a. Age of Onset** -Infancy. This condition is very rare.
b. Genetic Expression -None. It is indicated in infants that have increased acid mucopolysaccharide accumulation in Bowman's layer without evidence of systemic mucoploysaccharidosis.
c. Association with Other Ocular or Systemic Diseases - None.
d. Resembles/Confused With - Systemic mucopolysaccharidosis, which is transmitted autosomal recessive and is characterized by opacities begining in the center of the cornea which progress to the proximity of the limbus, eventually affecting the endothelium in the form of corneal guttata. Patients are plagued by photophobia and irritation and have a marked decrease in visual acuity by age 30 or 40.

P: **a. UA Progression** -Vision may be affected if opacities become dense.
b. Management Strategy -None required. Penetrating keratoplasty may be necessary in individuals with dense opacities.
c. Overall Prognosis - Good.

Local Anterior Mucopolysaccharide Accumulation



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GRANULAR DYSTROPHY
GROENOUW I, BREAD CRUMB DYSTROPHY

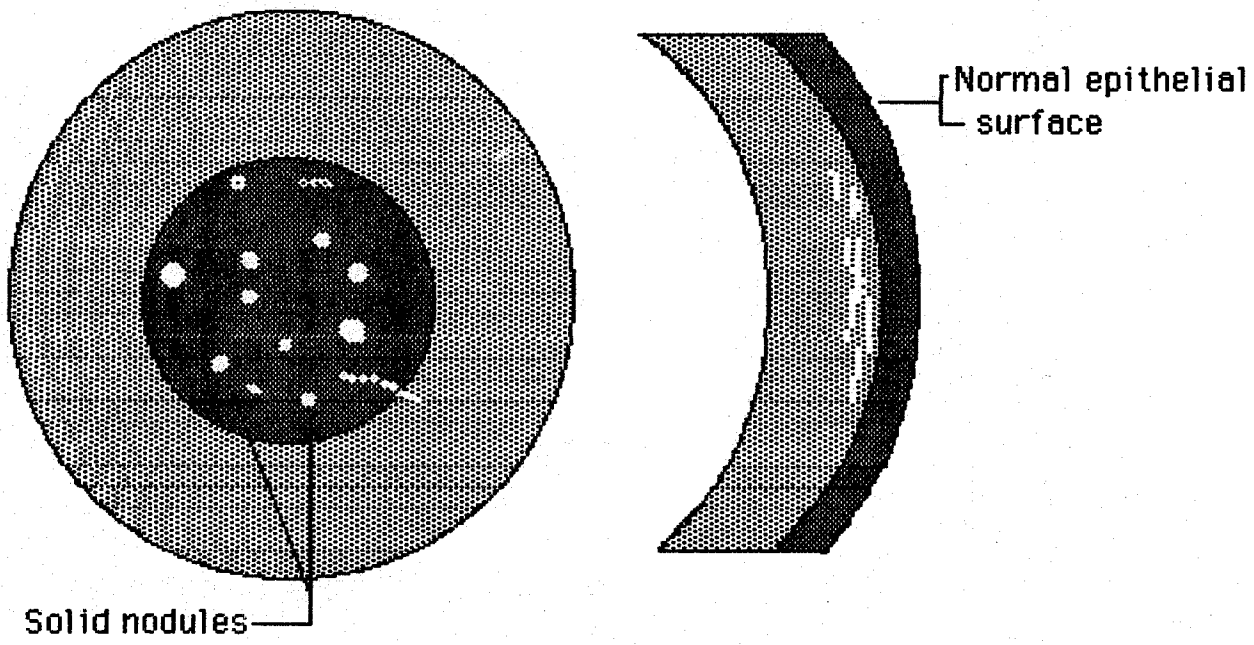
S: **a. VA** - Acuity remains normal into early adulthood, then progressively decreases, seldom past 20/200.
b. Corneal Sensation - Normal, epithelial erosions are rare - this is the least likely of the three "classical" stromal dystrophies to experience erosions.

O: **a. Part of layer in which dystrophy appears** - Axial cornea, just posterior to Bowman's layer.
b. Dystrophy Appearance - Becoming grossly visible by adolescence, this dystrophy has a variety of appearances: solid-white round or oval nodules, 0.2-0.4mm in diameter with frayed borders; semi-translucent crumb-like spots on an opaque-glass background that is snowflake-like when viewed at 40 X; white rings with punched out centers. Distribution may be random, sometimes appearing to form linear and arcuate chains. The stroma between opacities remains clear until late in the disease where upon a haze may become apparent.
c. Appearance of Other Layers - Usually appear normal. Bowman's membrane may be thinned or absent.

A: **a. Age of Onset** - First seen in early adolescence.
b. Genetic Expression - Autosomal dominant with bilateral, symmetric expression. Cases have reported where no dominance has been established.
c. Association with Other Ocular or Systemic Diseases - None.
d. Resembles/Confused With - Macular Dystrophy.

P: **a. VA Progression** - Beginning in early adult hood,visual acuity becomes progressively worse. Granular Dystrophy is one of the most common vison-threatening dystrophies.
b. Management Strategy - Early on, none is required. Later, by the fifth or sixth decade, a penetrating keratoplasty may be necessary to restore vision.
c. Overall Prognosis - Very favorable. The dystrophy may recur in the corneal graft one to nineteen years following surgery, but VA usually remains good.

Granular Dystrophy



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LATTICE DYSTROPHY
BIBER-HAAB-DIMMER DYSTROPHY

S: **a. VA** - Minimal to marked loss.
b. Corneal Sensation - Normal, to pain and photophobia from recurrent corneal erosions. The erosions and pain decrease in the third to fourth decade as well as overall corneal sensation.

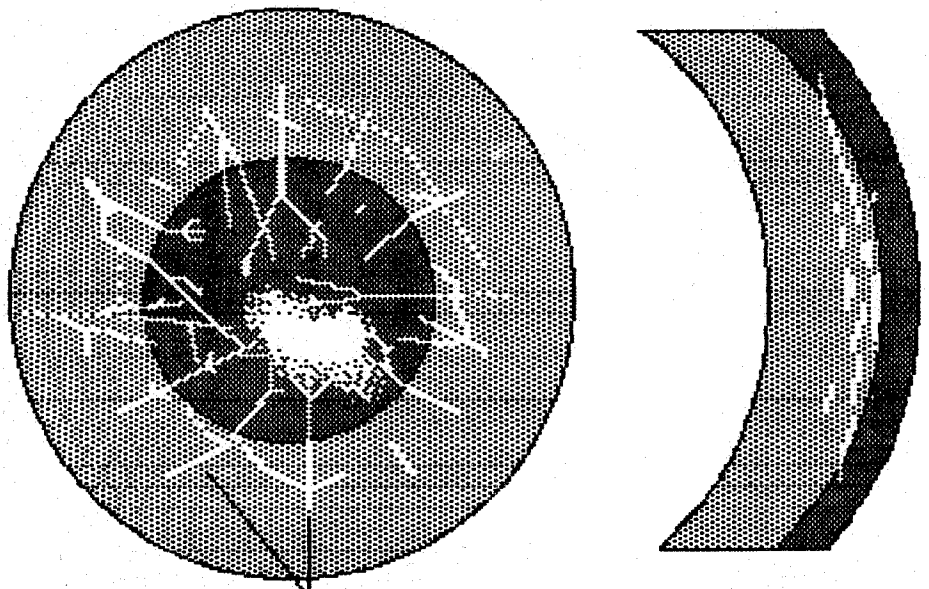
O: **a. Part of layer in which dystrophy appears** - Central, superficial stroma.
b. Dystrophy Appearance - The cornea manifests a ground glass appearance, characterized by three components, the most prominent being a central, diffuse opacity. Weaving through this are small, refractile threads that "grow" into dense cords as the dystrophy progresses. These threads may invade the epithelium, causing the above noted erosions. Small, comma-like flecks are also observed. The stroma between the threads is initially clear, but becomes progressively hazy.
c. Appearance of Other Layers - The epithelium is affected as noted above.

A: **a. Age of Onset** - is usually seven to nine years, but has been observed as early as two years. A late onset may also occur.
b. Genetic Expression - Autosomal dominant, but with subtle gene expression - only 10-25% of the cases are detected. It is generally seen bilaterally symmetrical, but asymmetric and unilateral cases do occur.
c. Association with Other Ocular or Systemic Diseases - Lattice dystrophy is often associated with systemic hypercholesterolemia, but is itself a separate entity.
d. Resembles/Confused With - Not applicable.

P: **a. VA Progression** - Marked loss by about the third decade.
b. Management Strategy - Refer to internal medicine for cholesterol problem. Artificial tears, hypertonic ointments and solutions, intermittent pressure patching and soft contact lenses can be prescribed to help with the recurrent erosions (see Appendix 1 for details); keratoplasty when indicated to restore vision.
c. Overall Prognosis - internal medicine - good with dietary control. Keratoplasty restores good VA, though opacities may recur in two to fourteen years - recurrence in the graft occurs more frequently in this dystrophy than in the other two "classic" stromal dystrophies.

Special Notes - most authorities believe this to be a familial amylodosis limited to the cornea.

Lattice Dystrophy



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MACULAR DYSTROPHY
GROENOUW II, FEHR'S MACULAR DYSTROPHY

S: **a. VA** - Normal through childhood, then becomes progressively worse.
b. Corneal Sensation - Becomes decreased during the second decade. Epithelial erosions and increasingly decreased sensation may manifest late in the disease course.

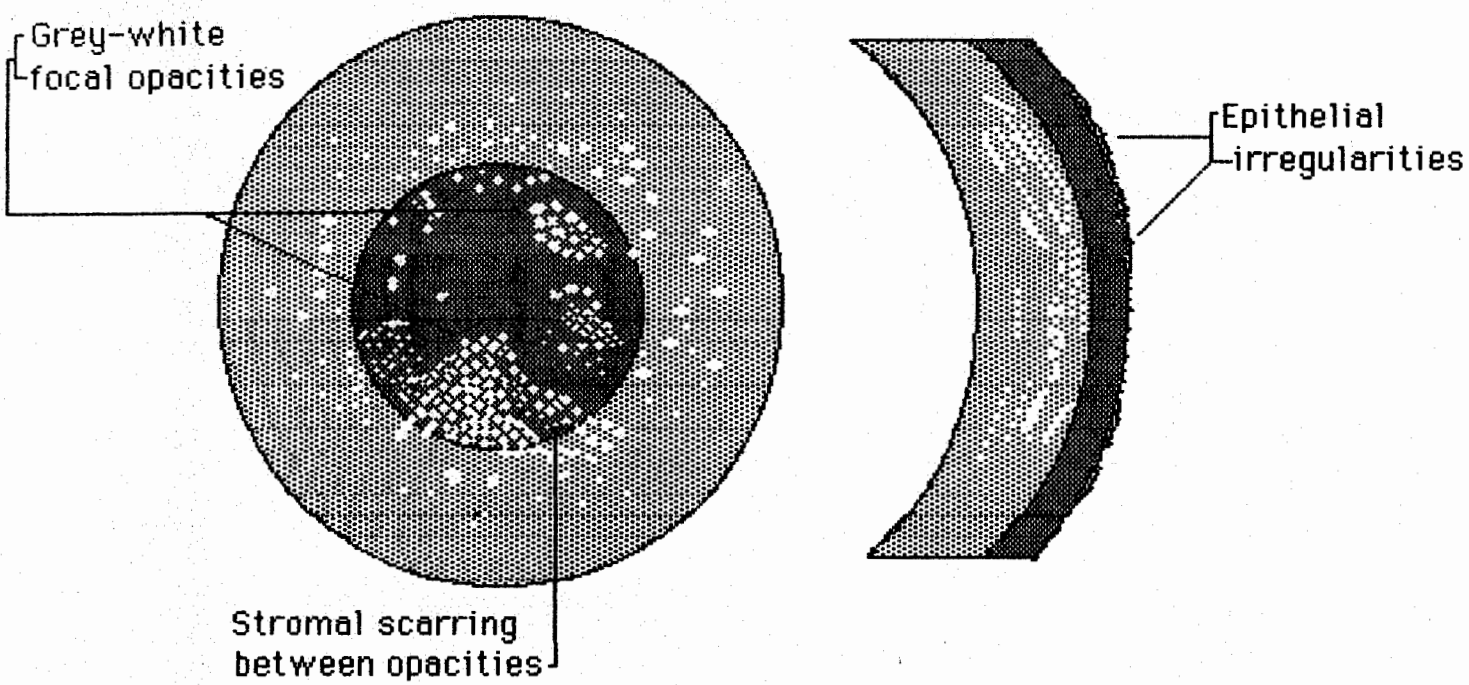
O: **a. Part of layer in which dystrophy appears** - Begins centrally and progresses to involve the entire stroma.
b. Dystrophy Appearance - is a superficial central cloudiness during the first ten years of life that progress to a diffuse stromal haze during the next ten to involve the full stromal thickness and periphery. The haze itself consists of gray-white focal opacities, minute to 0.5mm in diameter with hazy edges and hazy, scarred stroma between them.
c. Appearance of Other Layers - Epithelial irregularities appear between ten and thirty years of age. Guttata and opacification of Descemet's are observed late in the course of the disease.

A: **a. Age of Onset** - Within the first ten years of life.
b. Genetic Expression - Autosomal recessive with symmetrical, bilateral expression.
c. Association with Other Ocular or Systemic Diseases - Occurs occasionally with corneal arcus.
d. Resembles/Confused With - Granular dystrophy.

P: **a. VA Progression** - decreases by the second to fourth decade to the point of requiring a penetrating keratoplasty.
b. Management Strategy - Erosions may be managed with hypertonic ointments and solutions and soft contact lenses (see also Appendix 1). Keratoplasty may be performed when indicated.
c. Overall Prognosis - Good with keratoplasty, though the disease may recur in the graft.

Special Notes - irregular astigmatism may be present from the lifting of the epithelium by the lesions - rigid lenses can be fitted to correct for this, but may not be tolerated. Macular dystrophy is the most serious of the three classical stromal dystrophies.

Macular Dystrophy



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FLECK DYSTROPHY

SPECKLED, MOUCHETÉE, FRANCOIS/NEETENS DYSTROPHY

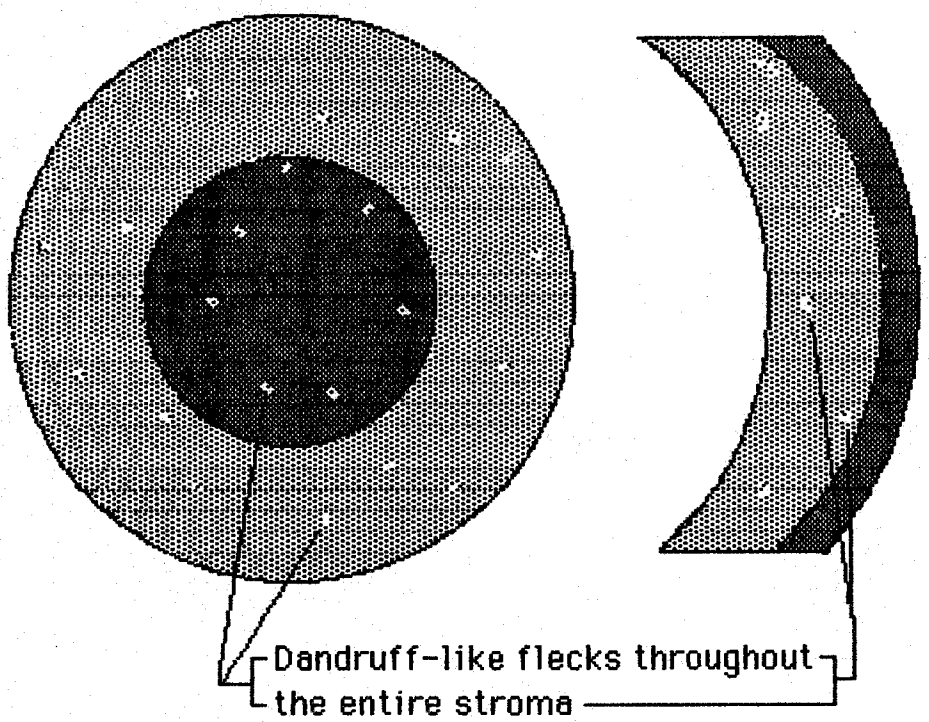
S: **a. VA** - Remains normal.
b. Corneal Sensation - Generally normal, but occasionally reduced. Photophobia of varying degrees (usually mild) may be present.

O: **a. Part of layer in which dystrophy appears** - Fleck Dystrophy usually extends throughout the entire stroma though is very occasionally seen limited to central or peripheral stroma.
b. Dystrophy Appearance - It appears as discrete, flat, grayish-white dandruff-like flecks in an otherwise clear stroma that are refractile under retro-illumination. Under high magnification the flecks are observed to be white rings surrounding a clear center; stellate and comma forms have also been observed. The number of opacities may vary from an occasional fleck to a diffuse stromal speckling.
c. Appearance of Other Layers - All are normal (Duane's reports that the flecks are apparent in all layers of the cornea).

A: **a. Age of Onset** - Usually detected at about age two, but may be congenital.
b. Genetic Expression - Autosomal dominant, generally bilateral, but may be very asymmetric or even unilateral.
c. Association with Other Ocular or Systemic Diseases - It may exist with punctate lens opacities, CCC dystrophy, keratoconnus, limbal dermoids or pseudoxantha elasticum.
d. Resembles/Confused With - Not applicable.

P: **a. VA Progression** - Remains normal.
b. Management Strategy - None specifically needed.
c. Overall Prognosis - Excellent.

Fleck Dystrophy



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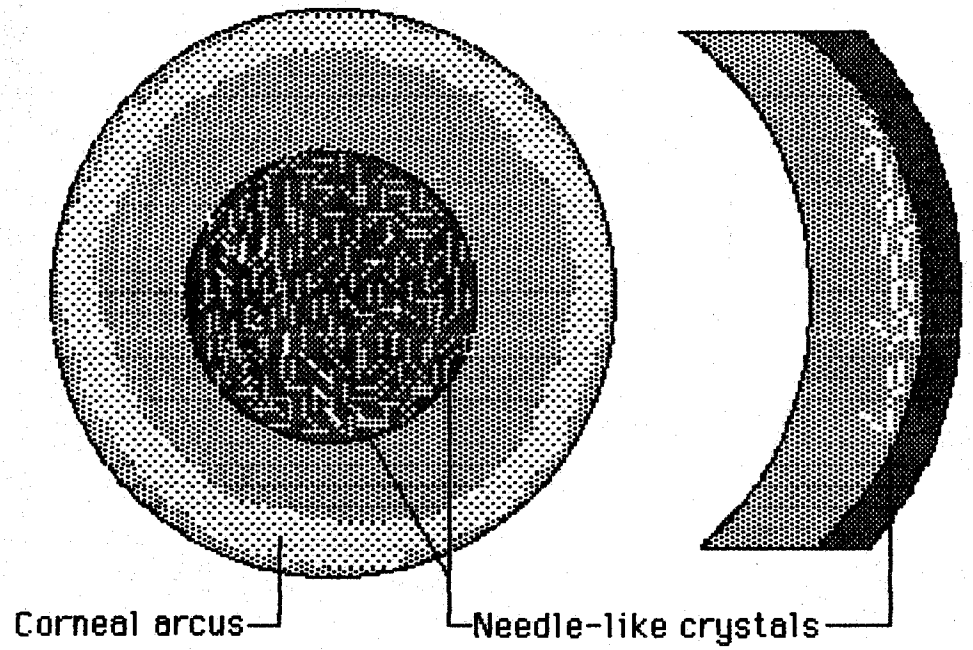
S: **a. VA** - Generally is no worse than 20/40, but occasional marked losses of acuity have been reported.
b. Corneal Sensation - Ranges from normal to decreased sensation.

O: **a. Part of layer in which dystrophy appears** - Central stroma, just beneath Bowman's layer, though sometimes deep and fusing with corneal arcus.
b. Dystrophy Appearance - Four components have been described: minute needle-like crystals that shimmer in different colors like fiberglass slivers, diffuse gray stromal haze, dense corneal arcus and limbal girdle. 80% of the cases contain the latter two. The components may occur in any combination and their clinical appearance can vary widely within families. Two distinctive patterns may occur: disc and ring shaped. Uninvolved stroma is usually clear but sometimes demonstrates white punctate opacities.
c. Appearance of Other Layers - Usually normal.

A: **a. Age of Onset** - Absent at birth, with bilateral expression by age one. The onset is very subtle and may be undetected until the 3rd to 4th decade.
b. Genetic Expression - Autosomal dominant with bilateral expression. Sporadic cases have been reported.
c. Association with Other Ocular or Systemic Diseases - This dystrophy is frequently associated with hyperlipidemia and genu valgum which are probably independently inherited. Xanthelasma and arcus have also been noted.
d. Resembles/Confused With - Not applicable.

P: **a. VA Progression** - VA usually remains quite usable as progession of the dystrophy usually halts by the third decade.
b. Management Strategy - Monitor; keratoplasty is usually not necessary, but if so, the donor cornea may be affected. Refer for hyperlipidemia screening.
c. Overall Prognosis - Usually good.

Central Crystalline Dystrophy



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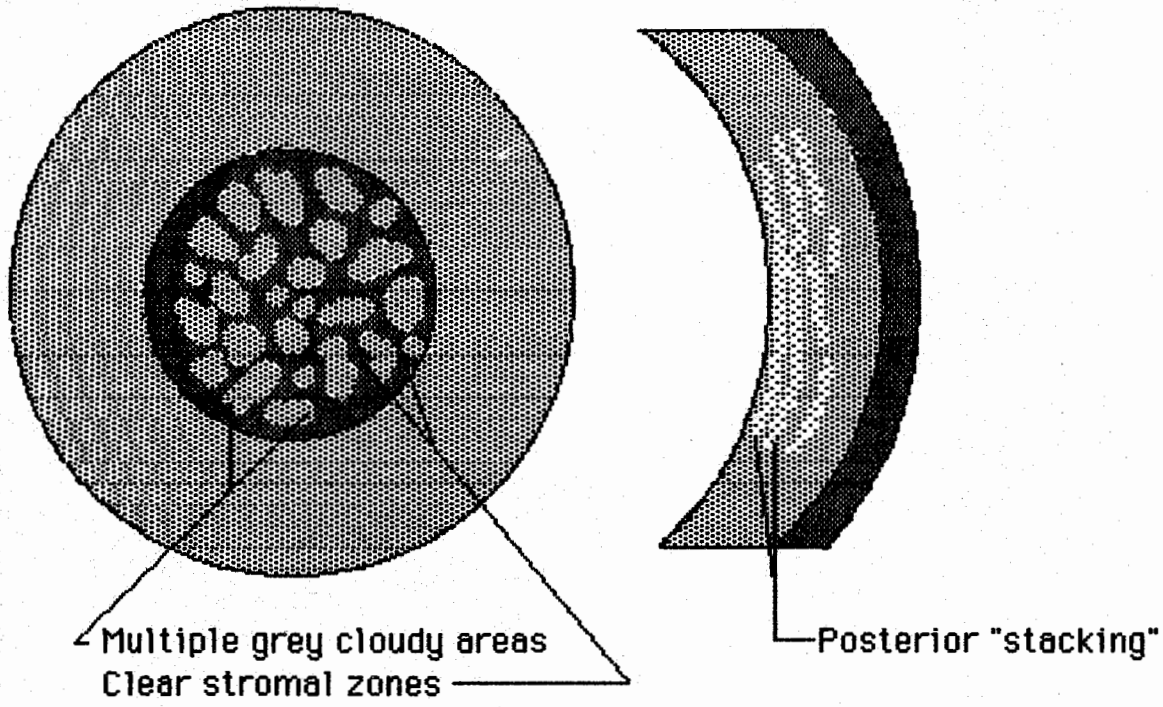
S: **a. VA** - Remains normal.
b. Corneal Sensation - Remains normal.

O: **a. Part of layer in which dystrophy appears** - This dystrophy is located in the central third of the stroma and is densest toward the posterior, leaving a clear periphery. At times, the entire stroma may be involved though Bowman's layer is spared.
b. Dystrophy Appearance - Observed under oblique or direct illumination, it appears as three to five stacks of small, multiple, grey, cloudy areas, somewhat polygonal in shape, separated by clearer fracture-like zones.
c. Appearance of Other Layers - Normal.

A: **a. Age of Onset** - This dystrophy has been observed from ages eight to eighty.
b. Genetic Expression - Autosomal dominant; expression may be sporadic.
c. Association with Other Ocular or Systemic Diseases - Central Cloudy Dystrophy has been noted to occur with Fleck and Pre-Decemet's Dystrophies, and also pseudoxanthoma elasticum.
d. Resembles/Confused With - Mosaic pattern dystrophy. Can be differentiated from this by its posterior location in the stroma.

P: **a. VA Progression** - Remains normal; the dystrophy is non-progressive.
b. Management Strategy - None required.
c. Overall Prognosis - Excellent.

Central Cloudy Dystrophy



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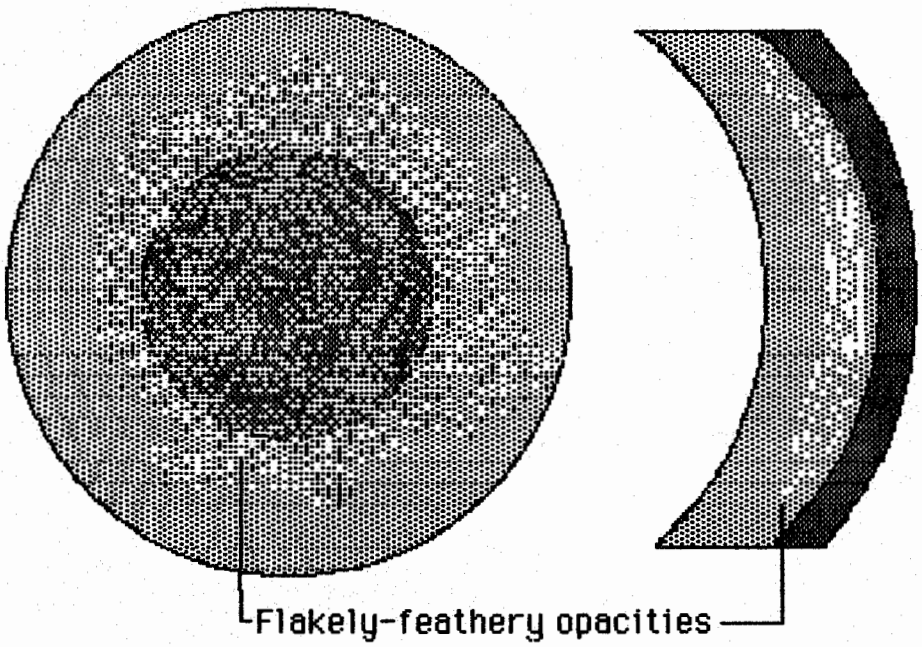
S: a. **VA** - Greatly decreased.
b. **Corneal Sensation** - Normal.

O: a. **Part of layer in which dystrophy appears** - Central, anterior stroma.
b. **Dystrophy Appearance** - Central, flakey-feathery opacities that fade toward the periphery. Corneal thickness is normal. Early visual deprivation leads to nystagmus and esotropia. The dystrophy is non-progressive.
c. **Appearance of Other Layers** - All normal, except that the anterior banded portion of Descemet's is poorly developed.

A: a. **Age of Onset** - Present at birth.
b. **Genetic Expression** - Autosomal dominant with symmetrical, bilateral expression.
c. **Association with Other Ocular or Systemic Diseases** - None.
d. **Resembles/Confused With** - Congenital Hereditary Endothelial Dystrophy (CHED). CHSD may be differentiated from CHED by the normal thickness ofboth the overall cornea and of Decemet's membrane.

P: a. **VA Progression** - VA is poor, usually no better than 20/200, even following keratoplasty, though acuity levels to 20/30 have been reported.
b. **Management Strategy** - Early keratoplasty.
c. **Overall Prognosis** - Poor, because amblyopia usually limits BVA to 20/200. The keratoplasty itself has a good prognosis.

Congenital Hereditary Stromal Dystrophy



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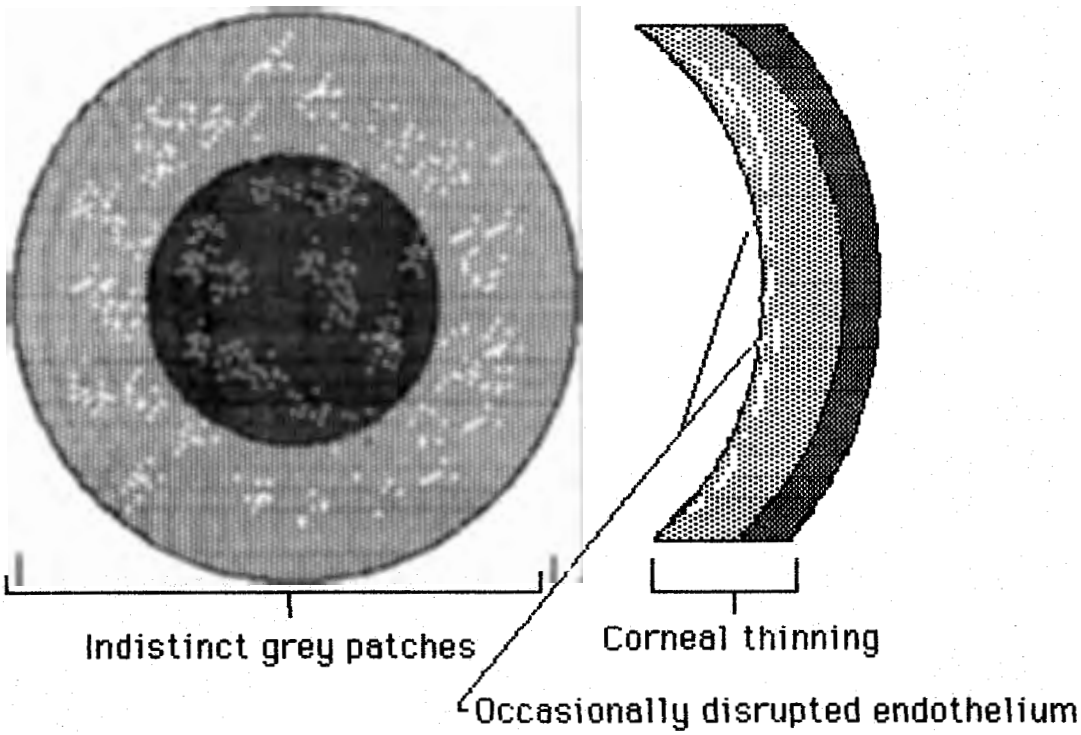
S: a. **VA loss** - Remains better than 20/40.
b. **Corneal Sensation** - Remains normal.

O: a. **Part of layer in which dystrophy appears** - Posterior Amorphous Dystrophy occurs in the deep stromal layers across the entire cornea.
b. **Dystrophy Appearance** - It appears as gray sheets in indistinct patches. Corneal thinning to 0.3mm also takes place. Both these processes occur slowly and gradually.
c. **Appearance of Other Layers** - The opacities extend into Descemet's membrane; occasional disruption of the endothelium may be seen.

A: a. **Age of Onset** - First decade of life.
b. **Genetic Expression** - Autosomal dominant, bilaterally symmetrical.
c. **Association with Other Ocular or Systemic Diseases** - None.
d. **Resembles/Confused With** - Posterior Amorphous Dystrophy may be differentiated in the following manner: it does not have the punctate opacities of pre-Descemet's "Dystrophy", nor the refractile changes in Descemet's membrane as observed in Posterior Polymorphic Dystrophy, nor the diffuse edema seen in Congenital Heriditary Endothelial Dystrophy, and lacks the vessels as seen in interstitial keratitis.

P: a. **VA Progression** - Doesn't decrease below 20/40.
b. **Management Strategy** - None required.
c. **Overall Prognosis** - Good.

Posterior Amorphous Dystrophy



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POLYMORPHIC STROMAL "DYSTROPHY"
POLYMORPHIC AMYLOID DEGENERATION

S: **a. VA loss** - Remains normal.
b. Corneal Sensation - Remains normal.

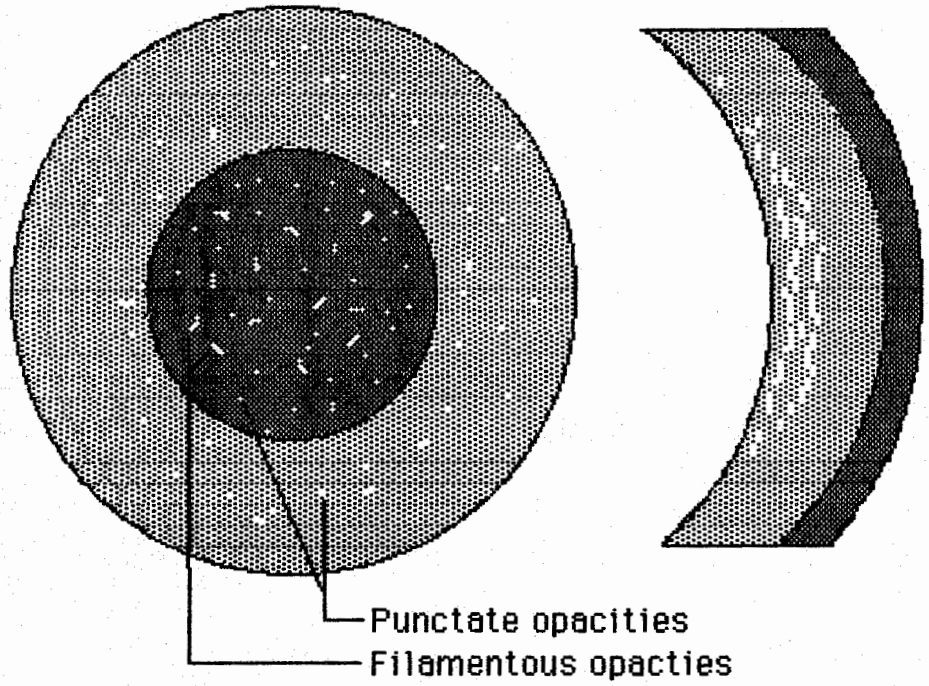
O: **a. Part of layer in which dystrophy appears** - Opacities are largest in the deep central stroma and become smaller towards the anterior and periphery.
b. Dystrophy Appearance - Two types of opacities are demonstrated: punctate and filamentous. The punctate opacities appear as discrete grayish-white stellate flecks within clear stroma. The filamentous opacities appear grey in focal illumination and refractile in retro-illumination and are irregularily beaded. The filaments are similar to the small lines in lattice dystrophy and can occur together with the punctate opacities; either one may predominate.
c. Appearance of Other Layers - Normal.

A: **a. Age of Onset** - After 50.
b. Genetic Expression - Not familial.
c. Association with Other Ocular or Systemic Diseases - None.
d. Resembles/Confused With - May be confused with lattice, however it is non-progressive, non-familal, does not affect the epithelium, and occurs later in life.

P: **a. VA Progression** - Remains normal.
b. Management Strategy - None required.
c. Overall Prognosis - Excellent.

Special Notes - As defined in the introduction, this "dystrophy" is more likely an age-related corneal degeneration.

Polymorphic Stromal "Dystrophy"



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Pre-Descemet's Dystrophy
Deep Filiform/Deep Punctiform Dystrophy

S: **a. VA** - Remains normal.
b. Corneal Sensation - Remains normal.

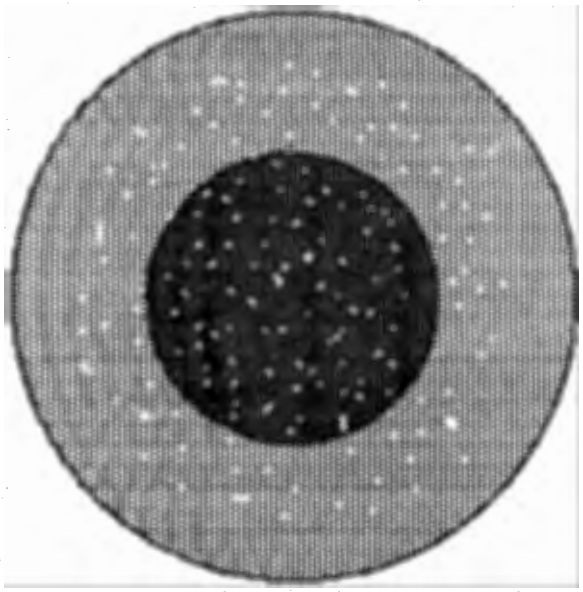
O: **a. Part of layer in which dystrophy appears** - Can occur centrally, peripherally, or both.
b. Dystrophy Appearance - is tiny, discrete punctate or linear specks. Larger forms may take on a variety of shapes including boomerang, comma, worm and circular. The opacities are refractile upon retro illumination.
c. Appearance of Other Layers - Normal, but see association below.

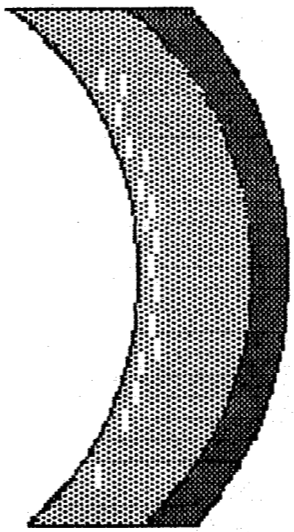
A: **a. Age of Onset** - Apparent after age thirty.
b. Genetic Expression - Thought to be degenerative; seen bilaterally symmetrical and without "antecedent" disease.
c. Association With Other Ocular or Systemic Diseases - May appear in conjunction with dot-map-fingerprint dystrophy, central cloudy dystrophy, posterior polymorphous dystrophy or keratoconnus.
d. Resembles/Confused With - Not applicable.

P: **a. VA Progression** - Acuity remains normal.
b. Management Strategy - Rule out the presence of other dystrophies.
c. Overall Prognosis - Excellent.

Special Notes - A rare degeneration.

Pre-Descemet's Dystrophy





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Fuch's Dystrophy

Epithelial-Endothelial Dystrophy, Combined dystrophy of Fuch's, Endothelial Dystrophy of the Cornea, Epithelial Dystrophy of Fuch's.

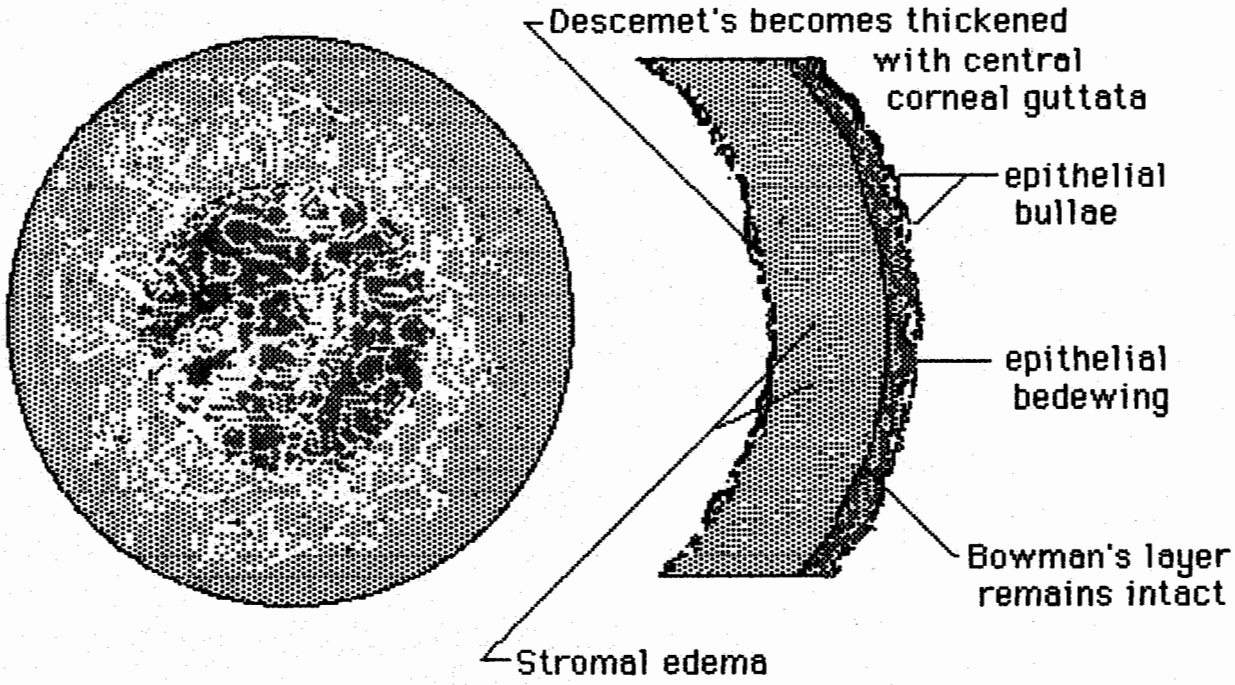
S: **a. VA** - Progressive visual loss.
b. Corneal Sensation - Reduced or absent. Patient is asymptomatic at first. Ruptured corneal bullae cause severe pain, especially in the morning.

O: **a. Layer in which dystrophy appears** -The disease affects the corneal endothelium but later involves all layers of the cornea.
b. Dystrophy Appearance - The disease begins with bilateral, often asymmetric, excresences of the central cornea that resemble Hassal-Henle warts. Later Descemet's membrane becomes opaque and thickened followed by stromal edema. Epithelial edema then ocurs as small clear cysts in the epithelium with roughing of its surface (called bedewing). Eventually, large epithelial bullae rupture, causing severe pain. Bowman's layer remains intact with occasional breaks filled with connective tissue.
c. Appearance of Other Layers - All areas may become affected.

A: **a. Age of Onset** -Adult. Women between 30 and 50 are mostly affected.
b. Genetic Expression - The inheritance pattern has not been clearly defined but is occasionally autosomal dominant and occurs 3 to 4 times more often in women than in men.
c. Association with Other Ocular or Systemic Diseases - None.
d. Resembles/Confused With -Conditions that may be etiologic factors in producing corneal edema; Macular corneal dystrophy, Latice corneal dystrophy, Posterior polymorphous dystrophy, surgical trama, epithelial downgrowth, hemorrhage into the anterior chamber, corneal graft failure, herpes simplex keratitis, herpes zoster keratitis, severe uveitis and iritis, corneal ulcers, epinephrine and pilocarpine instilled into the anterior chamber, acute narrow angle, and acute hydropes caused by keratoconus.

P: **a. VA Progression** - The patient experiences glare and hazy vision as edema worsens.
b. Management Strategy -See erosion therapy, Appendix 1. Advanced cases may require penetrating keratoplasty and is the treatment of choice. Intraocular pressure is often reduced to prevent stromal edema with 1% pilocarpine 3 or 4 times daily or 0.25% Timolol twice daily or 250 mg of acetazolamide 3 times daily.
c. Overall Prognosis -Short term prognosis is good with 80 per cent of transplanted corneas remaining clear for two years. Eventually, bullous keratopathy, cicatrization, endothelial degeneration, epithelial and stromal edema, folds in Descemet's membrane, and vascularization degrade acuity. Ocular pain, secondary glaucoma, and irritation are common symptoms.

Fuch's Dystrophy



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POSTERIOR POLYMORPHOUS DYSTROPHY (OF SCHLICHTING)

PPD, HEREDITARY DEEP DYSTROPHY, GROUPED VESICLES, CONGENITAL CORNEAL EDEMA, SCHNYDER'S POSTERIOR HERPES

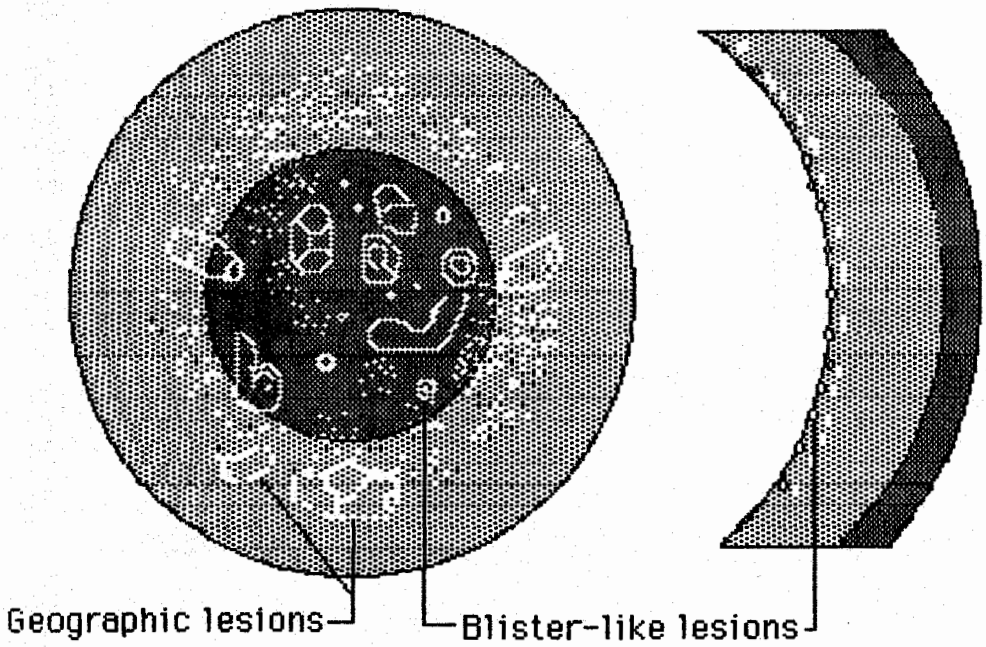
S: **a. VA** - Normal in most cases but can be decreased due to edema.
b. Corneal Sensation - Normal.

O: **a. Part of layer in which dystrophy appears** - endothelium.
b. Dystrophy Appearance - PPD is observed as small round vesicles or blisters in groups of two to twenty under retro-illumination, "beaten metal" under specular reflection. Larger, less defined grayish geographic lesions may also be present. Corneal thickness remains normal.
c. Appearance of Other Layers - Broad bands of grey thickened areas can be observed in Descemet's membrane. Grouped, blister-like lesions, at one time thought to be caused by herpes, are also seen in Descemet's membrane. Occasionally, there is mild stromal edema that can progress to epitehelial edema with a secondary band keratopathy.

A: **a. Age of Onset** - Most likely congenital.
b. Genetic Expression - Usually autosomal dominant with wide, bilateral expression. Some autosomal recessive cases have been noted.
c. Association with Other Ocular or Systemic Diseases - May be seen with abnormal iris processes, band keratopathy, displaced pupil, epithelialization of the endothelium, glaucoma, peripheral irido-corneal lesions, and posterior circumscribed keratoconnus.
d. Resembles/Confused With - Differentiate from CHED by family history of PPD, normal corneal thickness, and the relatively mild edema.

P: **a. VA Progression** - Remains normal in most cases. Patients with sufficient edema will have decreased VA.
b. Management Strategy - Uncomplicated cases require no treatment. Cases with mild edema can be managed with hair dryers, hypertonic ointments and solutions, or therapeutic soft contact lenses (see also Appendix 1). Penetrating keratoplasties may be performed if the edema is severe enough.
c. Overall Prognosis - generally good. The dystrophy may be slowly progressive.

Posterior Polymorphous Dystrophy



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CONGENITAL HEREDITARY ENDOTHELIAL DYSTROPHY (CHED)

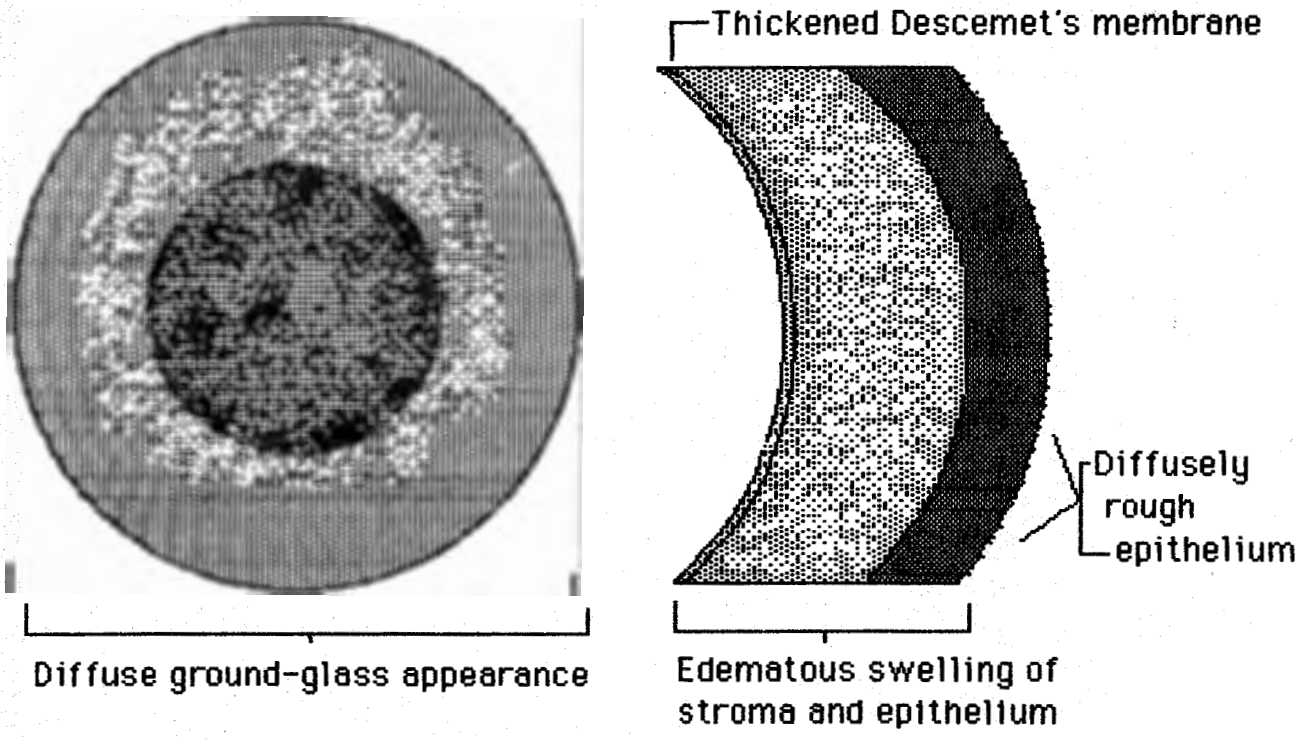
S: a. VA - Moderate (20/70) to severe (<20/200) acuity loss.
b. Corneal Sensation - normal, to increasing photophobia as the dystrophy progresses.

O: a. Layer in which dystrophy appears - See (b) below.
b. Dystrophy Appearance - Epithelial and stromal edema extending to the limbus are major hallmarks with the corneas swelling to 2-3 times their normal size. They (the corneas) have a diffuse grey-blue ground glass appearance that may be dotted with focal grey spots. The endothelial mosaic may be present or appear to be absent. Ocular inflammation and corneal vascularization can develop. Nystagmus is present in the more severe but non-progressive recessive form.
c. Appearance of Other Layers - Epithelium: diffusely rough. Stroma: exhibits different densities from one area to another, but is the same anterior to posterior. Desemet's membrane: grey and thickened and may be poorly formed. No guttatae are present.

A: a. Age of Onset - The dominant form appears within the first few years of life; the recessive form is congenital.
b. Genetic Expression - CHED expresses itself in one of two modes: 1) a rarer autosomal dominant mode that has a widely variable bilateral, symmetrical expression, or 2) an autosomal recessive one that is more "common".
c. Association with Other Ocular or Systemic Diseases - Possibly associated with a progressive autosomal dominant high-tone sensory neural deafness.
d. Resembles/Confused With - CHED cand be differentially diagnosed from macular dystrophy by time of onset, from PPD by corneal thickness, and from congenital glaucoma without bupthalmos by lack of inflammation, photophobia, progressive corneal enlargement and increased IOP. The presence of anterior staphyloma, birth trauma, interstitial keratitis, Peter's anomaly and sclerocornea also need to be ruled out.

P: a. VA Progression - VA in the dominant form may be reduced to light perception over a 5-10 year period. The recessive form is non-progressive.
b. Management Strategy - The use of a hair dryer, hypertonic ointments and solutions and soft contact lenses may be employed in mild cases. Keratoplasties are called for in severe cases, though these often opacify within months or years of the surgery. New surgical techniques offer hope of improved long-term prognosis.
c. Overall Prognosis - guarded to poor.

Congenital Hereditary Endothelial Dystrophy



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Keratoconus

Conical cornea

S: **a. VA** - Often decreased due to high astigmatism. Most patients are myopes.
b. Corneal Sensation - Decreased corneal sensation.

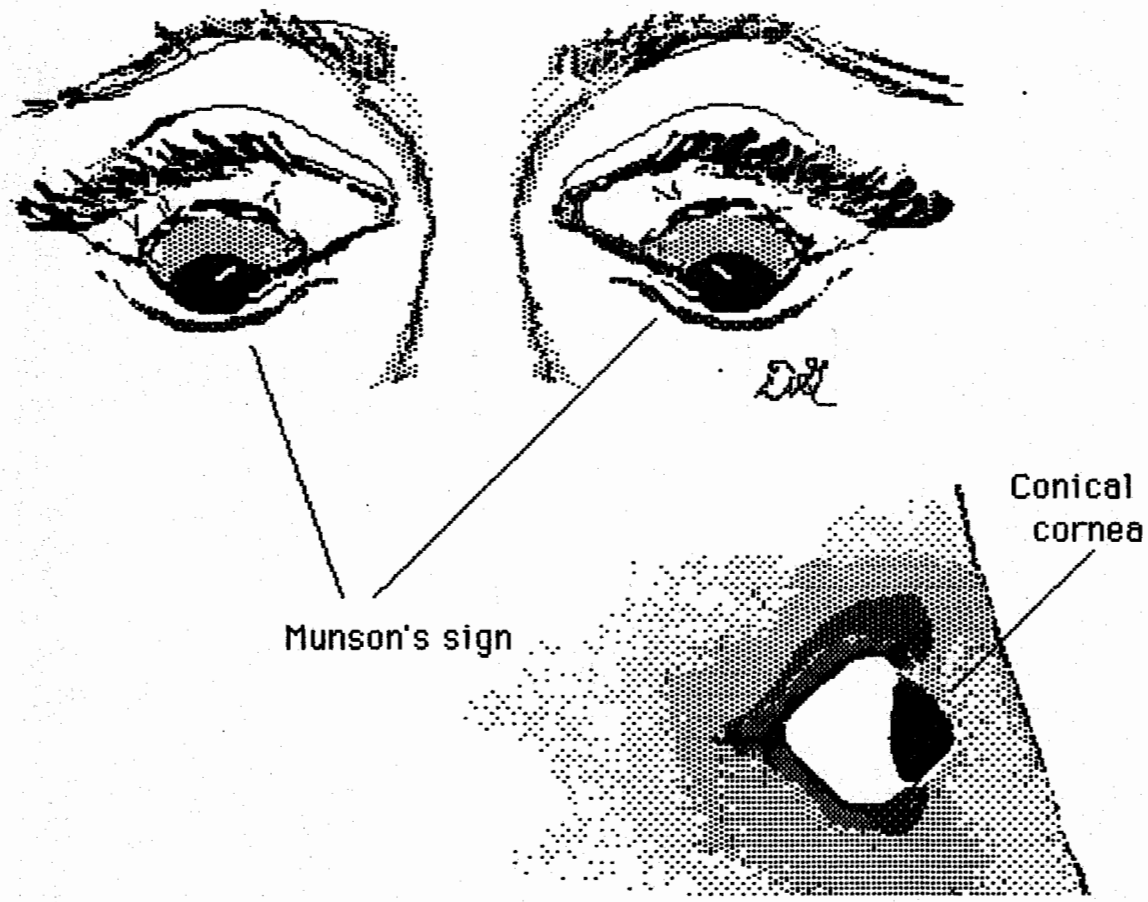
O: **a. Dystrophy Appearance** - There is an oblique conical deformation of the cone apex which is usually displaced inferior-nasally. Cones can be either nipple or oval shaped. Apical thinning of the cornea occurs with rare perforation. the deformation causes a convexity of the lower lid on downward gaze called Munson's sign. Vertical stress lines are present in the stroma. There is increased visibility of the corneal nerves with occasional rupture and scaring of Bowman's layer. When Descemet's membrane ruptures, acute hydropes occur which increase corneal clouding as a result of edema. Fleischer's ring and Munson's sign are diagnostic features of this disease.

A: **a. Age of Onset** - Keratoconus becomes evident around puberty and most often affects girls 10 to 16 years of age.

b. Genetic Expression - The mode of transmission is not clear. Dominant, recessive, and irregular transmission have all been documented.
c. Association with Other Ocular or Systemic Diseases -Keratoconus may be associated with retinitis pigmentosa, ectopia lentis, congenital cataract, aniridia, microcornea, and blue sclera. Systemic association includes; vernal catarrh, atopy, hypothyroidism osterogenesis imperfecta, Down's syndrome, and Crouzon's syndrome. In Ehlers-Danlos and Laurence-Moon-Biedl syndromes keratoconus has been reported with microcornea.

P: **a. VA Progression** -High astigmatism will require spectacle correction or the use of contact lenses to correct refractive error. Irregular astigmatism may decrease visual acuity. Visual acuity may be markedly reduced with acute hydropes but usually clears over an 8 to 10 week period.
b. Management Strategy -Spectacles are the initial treatment for astigmatism. The use of hard, soft, or piggyback lenses are very useful. Conditions that cannot be corrected by conventional means may require thermokeratoplasty if there is absence of corneal scaring. Penetrating keratoplasty is indicated if corneal scaring or vision cannot be corrected with contact lenses. If the patient is unable to wear contact lenses, ie; ectasia without scaring, retarded patients, or thoses with Down's syndrome, lamellar grafts are the treatment of choice. There is no need to consider acute hydropes as cause for an emergency keratoplasty procedure. The patient should be assured that the condition will usually improve with possible residual scarring.
c. Overall Prognosis - The condition progresses for 7 to 8 years and usually becomes arrested. Relapse can occur.

Keratoconus



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Appendix 1

Management of corneal erosions and referal criterion for Keratoplasty

Management of epithelial edema and erosions- For mild epithelial edema, topical 5 per cent sodium chloride drops administered 6 to 8 times a day, and ointment at night, may decrease epithelial edema and irregularity. Moderate conditions may require hypertonics, a cycloplegic agent, prophylactic antibiotics, pressure patch for 48 hours, and systemic analgesics. Severe conditions often require additional pressure patching every other day for a week.

Management of stromal edema - Hypertonic ointments and solutions do little in reducing stromal edema which must be dealt with in a different manner than epithelial edema. Decrease stromal edema by reducing the intraocular pressure with one of the following drugs; 1% pilocarpine, 3 or 4 times daily ; 0.25% timolol, twice daily; acetazolamide, 250 mg, three times daily.

Surgical referral criteria - Visual acuities of less than 20/70, in both eyes, are usually present before a keratoplasty is attempted. Monocular patients usually reach acuities of less than 20/400 and have severe bullous keratopathy before surgical intervention is recommended.

Corticosteroids prevent immune reactions in corneal grafts. One per cent prednisolone solution or ointment, 3 to 6 times per day, is usually administered postoperatively.

Failing corneal grafts manifest the following complications; bullous keratopathy; cicatrization; endothelial degeneration; epithelial and stromal edema; folds in Descemet's membrane; pigment on the posterior corneal surface; striae; corneal vascularization; vesicle formation; ocular pain, secondary glaucoma, and irritation.

Further Readings

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*most informative, suggest reading for detailed information

¥ good pictorials and photographs (viewmaster slides, Donaldson)

† good clinical reference manual

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