Corneal Dystrophies

What are the four categories of corneal dystrophies?
Corneal Dystrophies

Epithelial and Subepithelial Dystrophies

Epithelial-Stromal *TGFB1* Dystrophies

What are the four categories of corneal dystrophies?

Stromal Dystrophies

Endothelial Dystrophies
Epithelial and Subepithelial Dystrophies

Epithelial-Stromal \textbf{TGFBI} Dystrophies

What does TGFBI stand for in this context?

Stromal Dystrophies

Endothelial Dystrophies
Epithelial and Subepithelial Dystrophies

Epithelial- Stromal $TGFBI$ Dystrophies

What does TGFBI stand for in this context?
‘Transforming growth factor beta induced’

Stromal Dystrophies

Endothelial Dystrophies
Epithelial and Subepithelial Dystrophies

What does TGFBI stand for in this context? ‘Transforming growth factor beta induced’

To what does the term growth factor refer?
Epithelial-Stromal Dystrophies

What does TGFBI stand for in this context?
‘Transforming growth factor beta induced’

To what does the term growth factor refer?
To any of a diverse group of protein (or steroid) molecules that promote cell growth
Epithelial and Subepithelial Dystrophies

**Corneal Dystrophies**

**Epithelial-Stromal** \(\text{TGFBI}\) Dystrophies

What does TGFBI stand for in this context? ‘Transforming growth factor beta induced’

*To what does the term growth factor refer?*
To any of a diverse group of protein (or steroid) molecules that promote cell growth

*Which growth factor is likely most familiar to ophthalmologists?*
Two words: Vascular endothelial growth factor

Endothelial Dystrophies
What does TGFBI stand for in this context? ‘Transforming growth factor beta induced’

To what does the term growth factor refer? To any of a diverse group of protein (or steroid) molecules that promote cell growth

Which growth factor is likely most familiar to ophthalmologists? Vascular endothelial growth factor
Corneal Dystrophies

Epithelial and Subepithelial Dystrophies

Epithelial-Stromal \textbf{TGFBI} Dystrophies

\textit{What does TGFBI stand for in this context?}

\textit{Transforming growth factor beta induced}

\textit{To what does the term \textbf{transforming growth factor} refer?}

Stromal Dystrophies

Endothelial Dystrophies
Corneal Dystrophies

Epithelial and Subepithelial Dystrophies

Epithelial-Stromal TGFBI Dystrophies

What does TGFBI stand for in this context? 'Transforming growth factor beta induced'

To what does the term transforming growth factor refer?
A superfamily of related growth factors

Stromal Dystrophies

Endothelial Dystrophies
What does TGFBI stand for in this context? ‘Transforming growth factor beta induced’

To what does the term transforming growth factor beta refer?
Endothelial Dystrophies

Epithelial and Subepithelial Dystrophies

**Corneal Dystrophies**

Epithelial-Stromal **TGFBI** Dystrophies

*What does TGFBI stand for in this context?*

(Transforming growth factor beta induced)

*To what does the term transforming growth factor beta refer?*

A subclass of transforming growth factors (the other subclass is, not surprisingly, transforming growth factor alpha)

Endothelial Dystrophies
Corneal Dystrophies

Epithelial and Subepithelial Dystrophies

Epithelial-Stromal \textbf{TGFBI} Dystrophies

What does TGFBI stand for in this context? 'Transforming growth factor beta induced'

To what does the term transforming growth factor beta \textbf{induced} refer?

Endothelial Dystrophies
Epithelial and Subepithelial Dystrophies

**Corneal Dystrophies**

*Epithelial-Stromal TGFBI Dystrophies*

*What does TGFBI stand for in this context?*

*Transforming growth factor beta induced*

*To what does the term transforming growth factor beta induced refer?*

It refers to a protein, the production of which is controlled by a product of the *transforming growth factor beta* family.
Epithelial and Subepithelial Dystrophies

Epithelial-Stromal TGFBI Dystrophies

What does TGFBI stand for in this context?
Transforming growth factor beta induced

To what does the term transforming growth factor beta refer?
It refers to a protein, the production of which is controlled by a product of the transforming growth factor beta family.

What is the name of the protein involved in the TGFBI dystrophies?
Keratoepithelin (the Cornea book refers in passing to the TGFBI conditions as keratoepithelin dystrophies)

What is the clinical hallmark of the keratoepithelin dystrophies?
Recurrent epithelial erosions
Corneal Dystrophies

Epithelial and Subepithelial Dystrophies

**Epithelial-Stromal TGFBI Dystrophies**

What does TGFBI stand for in this context?
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To what does the term transforming growth factor beta induced refer?
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Endothelial Dystrophies
Corneal Dystrophies

Epithelial and Subepithelial Dystrophies

**Epithelial-Stromal** $TGFBI$ Dystrophies

*What does** $TGFBI$ **stand for in this context?*  
*Transforming growth factor beta induced*

*To what does the term transforming growth factor beta refer?*  
*It refers to a protein, the production of which is controlled by a product of the transforming growth factor beta family.*

*What is the name of the protein involved in the $TGFBI$ dystrophies?*  
*Keratoepithelin* (the *Cornea* book refers in passing to the $TGFBI$ conditions as *keratoepithelin dystrophies*)

*What is the clinical hallmark of the keratoepithelin dystrophies?*
What does TGFBI stand for in this context? *Transforming growth factor beta induced*

To what does the term transforming growth factor beta refer? It refers to a protein, the production of which is controlled by a product of the transforming growth factor beta family.

What is the name of the protein involved in the TGFBI dystrophies? *Keratoepithelin* (the *Cornea* book refers in passing to the TGFBI conditions as *keratoepithelin dystrophies*).

What is the clinical hallmark of the keratoepithelin dystrophies? Recurrent epithelial erosions.
Corneal Dystrophies

Epithelial and Subepithelial Dystrophies

What does TGFBI stand for in this context? 
Transforming growth factor beta induced

What is the name of the protein involved in the TGFBI dystrophies? 
Keratoepithelin (the Cornea book refers in passing to the TGFBI conditions as keratoepithelin dystrophies)

What is the clinical hallmark of the keratoepithelin dystrophies? 
Recurrent epithelial erosions

The Cornea book considered this factoid important enough to make it one of three ‘Highlights’ for the Dystrophy chapter—take note of it!
Corneal Dystrophies

Epithelial and Subepithelial Dystrophies

What does TGFBI stand for in this context? ‘Transforming growth factor beta induced’

What is TGFBI’s chromosomal location?

Stromal Dystrophies

Endothelial Dystrophies
Corneal Dystrophies

Epithelial and Subepithelial Dystrophies

Epithelial-Stromal $\text{TGFBI}$ Dystrophies

*What does TGFBI stand for in this context?*
‘Transforming growth factor beta induced’

*What is TGFBI’s chromosomal location?*
5q31

Stromal Dystrophies

Endothelial Dystrophies
Epithelial-Stromal Dystrophies

What does TGFBI stand for in this context?
‘Transforming growth factor beta induced’

What is TGFBI’s chromosomal location?
5q31

The TGFBI gene was formerly known as what?
Epithelial and Subepithelial Dystrophies

What does TGFBI stand for in this context?
‘Transforming growth factor beta induced’

What is TGFBI’s chromosomal location?
5q31

The TGFBI gene was formerly known as what?
BIGH3 (this factoid is important because you might encounter this name in the older literature)
Epithelial and Subepithelial Dystrophies

Epithelial-Stromal TGFBI Dystrophies

1) ?
2) ?
3) ?
4) ?
5) ?
6) ?

What are the six epithelial-stromal TGFBI corneal dystrophies?

Stromal Dystrophies

Endothelial Dystrophies
Epithelial and Subepithelial Dystrophies

**Epithelial-Stromal TGFBI Dystrophies**
1) Reis-Bücklers corneal dystrophy
2) Thiel-Behnke corneal dystrophy
3) Lattice, type 1
4) Lattice, variant types (III, IIIA, I/IIIA, IV)
5) Granular type 1
6) Granular type 2

What are the six epithelial-stromal TGFBI corneal dystrophies?

Stromal Dystrophies

Endothelial Dystrophies
Epithelial and Subepithelial Dystrophies

**Corneal Dystrophies**

Epithelial-Stromal *TGFBI* Dystrophies

1. **Reis-Bücklers** corneal dystrophy
2. **Thiel-Behnke** corneal dystrophy
3. Lattice, type 1
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Stromal Dystrophies

Endothelial Dystrophies
Corneal Dystrophies

Epithelial and Subepithelial Dystrophies

Epithelial-Stromal $\text{TGFBI}$ Dystrophies
1) Reis-Bücklers corneal dystrophy
2) Thiel-Behnke corneal dystrophy
3) Lattice, type 1
4) Lattice, variant types (III, IIIA, IIIIA, IV)
5) Granular type 1
6) Granular type 2

How do you pronounce this?
RICE BOO-klerz

How do you pronounce this?
TEAL BEN-key

Stromal Dystrophies

Endothelial Dystrophies
Epithelial and Subepithelial Dystrophies

Epithelial-Stromal $TGFBI$ Dystrophies
1) **Reis-Bücklers** corneal dystrophy
2) **Thiel-Behnke** corneal dystrophy
3) Lattice, type 1

What is the histologic hallmark of Reis-Bücklers and Thiel-Behnke?
Disruption/fragmentation of Bowman’s layer

Stromal Dystrophies

Endothelial Dystrophies
Epithelial-Stromal $TGFBI$ Dystrophies

1) Reis-Bücklers corneal dystrophy
2) Thiel-Behnke corneal dystrophy
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What is the histologic hallmark of Reis-Bücklers and Thiel-Behnke?
Disruption/fragmentation of Bowman's layer

Stromal Dystrophies

Endothelial Dystrophies
Epithelial-Stromal TGFBI Dystrophies

1) Reis-Bücklers corneal dystrophy
2) Thiel-Behnke corneal dystrophy
3) Lattice, type 1

What is the histologic hallmark of Reis-Bücklers and Thiel-Behnke?
Disruption/fragmentation of Bowman’s layer

Granular type 2 (Avellino dystrophy)

The corneal-dystrophy section of the Cornea book underwent a major revision recently. In what ‘category of corneal dystrophy’ were Reis-Bücklers and Thiel-Behnke in previous editions?
Endothelial Dystrophies

Epithelial-Stromal \textit{TGFB1} Dystrophies

1) \textbf{Reis-Bücklers} corneal dystrophy
2) \textbf{Thiel-Behnke} corneal dystrophy
3) Lattice, type 1
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What is the histologic hallmark of Reis-Bücklers and Thiel-Behnke?

\textbf{Disruption/fragmentation of Bowman’s layer}

The corneal-dystrophy section of the Cornea book underwent a major revision recently. In what ‘category of corneal dystrophy’ were Reis-Bücklers and Thiel-Behnke in previous editions? The \textbf{’Corneal Dystrophies of Bowmans’}
Corneal Dystrophies

Epithelial and Subepithelial Dystrophies

Epithelial-Stromal TGFBI Dystrophies
1) Reis-Bücklers corneal dystrophy
2) Thiel-Behnke corneal dystrophy
3) Lattice, type 1
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6) Granular type 2 (Avellino dystrophy)

What is the histologic hallmark of Reis-Bücklers and Thiel-Behnke?

Disruption/fragmentation of Bowman’s layer

Important aside: When you hear ‘disruption/fragmentation of Bowman’s layer,’ a specific corneal ectatic disorder should come immediately to mind. Which one?

The ’Corneal Dystrophies of Bowmans’

Endothelial Dystrophies
Corneal Dystrophies

Epithelial and Subepithelial Dystrophies

Epithelial-Stromal TGFBI Dystrophies
1) Reis-Bücklers corneal dystrophy
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Disruption/fragmentation of Bowman’s layer

Important aside: When you hear ‘disruption/fragmentation of Bowman’s layer,’ a specific corneal ectatic disorder should come immediately to mind. Which one? Keratoconus

The ‘Corneal Dystrophies of Bowmans’

Endothelial Dystrophies
Epithelial-Stromal \textit{TGFB1} Dystrophies

1) \textbf{Reis-Bücklers} corneal dystrophy
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\textbf{What is the histologic hallmark of Reis-Bücklers and Thiel-Behnke?}

\textit{Disruption/fragmentation of Bowman's layer}

\textbf{Important aside:} When you hear ‘disruption/fragmentation of Bowman’s layer,’ a specific corneal \textit{ectatic} disorder should come immediately to mind. \textit{Which one?} Keratoconus

\textit{To finish off this aside: The Cornea book addresses four ectatic conditions. What are the other three?}

--Keratoconus
--?
--?
--?
**Corneal Dystrophies**

**Epithelial and Subepithelial Dystrophies**

**Epithelial-Stromal TGFBI Dystrophies**
1) **Reis-Bücklers** corneal dystrophy
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What is the histologic hallmark of Reis-Bücklers and Thiel-Behnke? **Disruption/fragmentation of Bowman’s layer**

---

**Important aside:** When you hear ‘disruption/fragmentation of Bowman’s layer,’ a specific corneal ectatic disorder should come immediately to mind. **Which one? Keratoconus**

**To finish off this aside:** The Cornea book addresses four ectatic conditions. **What are the other three?**
-- Keratoconus
-- Keratoglobus
-- Pellucid marginal degeneration
-- Iatrogenic (ie, post-keratorefractive surgery)
### Epithelial-Stromal *TGFBI* Dystrophies

1. **Reis-Bücklers** corneal dystrophy
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3. Lattice, type 1
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### Stromal Dystrophies

- None listed

### Endothelial Dystrophies

- None listed
**Corneal Dystrophies**

### Epithelial and Subepithelial Dystrophies

**Epithelial-Stromal TGFBI Dystrophies**

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### Stromal Dystrophies

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### Endothelial Dystrophies

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## Corneal Dystrophies

### Epithelial and Subepithelial Dystrophies

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#### Age of onset
- **RBCD**: Childhood
- **TBCD**: Childhood

#### Genetics
- **RBCD**: TGFBI (BIGH3)
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### Epithelial-Stromal *TGFBI* Dystrophies

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### Stromal Dystrophies

### Endothelial Dystrophies
Reis–Bücklers corneal dystrophy: Geographic opacities

Thiel-Benke corneal dystrophy: Honeycomb appearance
### Endothelial Dystrophies

- Reis-Bücklers corneal dystrophy
- Thiel-Behnke corneal dystrophy
- Lattice, type 1
- Lattice, variant types (III, IIIA, I/IIIA, IV)
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- Granular type 2 (Avellino dystrophy)

### Stromal Dystrophies

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### Epithelial-Stromal TGFBI Dystrophies

- 1) Reis-Bücklers corneal dystrophy
- 2) Thiel-Behnke corneal dystrophy

### Epithelial and Subepithelial Dystrophies

- TGFBI (BIGH3)
### Corneal Dystrophies

**Epithelial and Subepithelial Dystrophies**

**Epithelial-Stromal** *TGFBI* Dystrophies

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**Stromal Dystrophies**

- **Age of onset**: Childhood
- **Genetics**: TGFBI (BIGH3)
- **SL appearance**: ‘Geographic’ opacification
- **Painful?**: Yes

**Endothelial Dystrophies**

- **Age of onset**: Childhood
- **Genetics**: TGFBI (BIGH3)
- **SL appearance**: ‘Honeycomb’ opacification
- **Painful?**: Yes
## Epithelial-Stromal TGFBI Dystrophies

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### Epithelial-Stromal *TGFBI* Dystrophies

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3. Lattice, type 1
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5. Granular type 1
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### Stromal Dystrophies

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### Endothelial Dystrophies

- Epithelial
- Stromal
- Subepithelial
### Epithelial-Stromal TGFBI Dystrophies

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**Endothelial Dystrophies**
Corneal Dystrophies

Reis-Bücklers: sheet-like layers

Thiel-Behnke: Sawtooth pattern

Reis-Bücklers

Thiel-Behnke
Corneal Dystrophies

Reis-Bücklers: sheet-like layers
Thiel- Behnke: Sawtooth pattern

The sawtooth pattern in TBCD is also appreciable via anterior-segment OCT
### Corneal Dystrophies

#### Epithelial and Subepithelial Dystrophies

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Corneal Dystrophies

Reis-Bücklers: sheet-like layers

Thiel-Behnke: Sawtooth pattern

Reis-Bücklers: Rod-shaped fibers

Thiel-Behnke: Curly fibers
Corneal Dystrophies

Epithelial and Subepithelial Dystrophies

Epithelial-Stromal TGFB1 Dystrophies

1) **Reis-Bücklers** corneal dystrophy
2) **Thiel-Behnke** corneal dystrophy
3) Lattice, type 1

*Can Reis-Bücklers and Thiel-Behnke be reliably differentiated from one another at the slit lamp?*

*Bowman’s layer dystrophies: Which is which?*

Endothelial Dystrophies
Epithelial-Stromal *TGFBI* Dystrophies

1) **Reis-Bücklers** corneal dystrophy
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*Can Reis-Bücklers and Thiel-Behnke be reliably differentiated from one another at the slit lamp?*  
The BCSC *Cornea* book says doing so is “difficult” (read: no, you can’t)
Epithelial-Stromal \textit{TGFB}I Dystrophies

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\textbf{How can they be differentiated clinically?}

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**Corneal Dystrophies**

Epithelial and Subepithelial Dystrophies

Epithelial-Stromal *TGFB1* Dystrophies
1) **Reis-Bücklers** corneal dystrophy
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_Can Reis-Bücklers and Thiel-Behnke be reliably differentiated from one another at the slit lamp?_ The BCSC _Cornea_ book says doing so is “difficult” (read: no, you can’t)

_How can they be differentiated clinically?_ Via anterior-segment OCT, and confocal microscopy

**Bowman’s layer dystrophies: Which is which?**

Endothelial Dystrophies
Epithelial and Subepithelial Dystrophies

**Epithelial-Stromal TGFBI Dystrophies**
1) Reis-Bücklers corneal dystrophy
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3) **Lattice, type 1**
4) **Lattice, variant types (III, IIIA, I/IIIA, IV)**
5) Granular type 1
6) Granular type 2

*Note: The Cornea book lumps together type 1 (aka ‘classic lattice’) and its variants*

**Stromal Dystrophies**

**Endothelial Dystrophies**
Epithelial-Stromal *TGFBI* Dystrophies

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Stromal Dystrophies

At what age does classic lattice type 1 begin to manifest?

**Epithelial-Stromal *TGFBI* Dystrophies**

At what age does classic lattice type 1 begin to manifest?

**Stromal Dystrophies**

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Epithelial and Subepithelial Dystrophies

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**Stromal Dystrophies**

**Endothelial Dystrophies**

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**At what age does classic lattice type 1 begin to manifest?**
Childhood to teens
Epithelial and Subepithelial Dystrophies

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Stromal Dystrophies

Endothelial Dystrophies

*Corneal Dystrophies*

At what age does classic lattice type 1 begin to manifest?
Childhood to teens

How does it present? What is seen at the slit lamp?

Early in the disease, fleck-like central opacities and a diffuse stromal haze are present. Later, myriad crisscrossing refractile lines in the cornea predominate.

Is it painful?
Yes; pts get recurrent epithelial erosions

Does it affect vision?
Surface irregularity and stromal haze often results in decreased vision

What is the hallmark of lattice type 1 on light microscopy?
The presence of amyloid in the subepithelial space and anterior stroma
Epithelial-Stromal Dystrophies

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Epithelial-Stromal *TGFBI* Dystrophies

At what age does classic lattice type 1 begin to manifest?
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Stromal Dystrophies

Endothelial Dystrophies
Lattice corneal dystrophy, type 1 (aka classic lattice). Direct (A) and retroillumination (B) of early lattice corneal dystrophy (LCD) with dots and fine lattice lines.
Epithelial and Subepithelial Dystrophies

Epithelial-Stromal \textit{TGFB1} Dystrophies

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Stromal Dystrophies

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Endothelial Dystrophies
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Epithelial and Subepithelial Dystrophies

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Stromal Dystrophies

Endothelial Dystrophies

At what age does classic lattice type 1 begin to manifest?
Childhood to teens

Why is this condition called ‘lattice’?

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Epithelial and Subepithelial Dystrophies

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Stromal Dystrophies

Endothelial Dystrophies

At what age does classic lattice type 1 begin to manifest?
Childhood to teens

Why is this condition called ‘lattice’?
Because the crisscrossing lines are reminiscent of a lattice structure (eg, like one might encounter in a garden)

diffuse stromal haze are present. Later, myriad crisscrossing refractile lines in the cornea predominate.

Corneal Dystrophies

Corneal Dystrophies

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Epithelial and Subepithelial Dystrophies

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Stromal Dystrophies

At what age does classic lattice type 1 begin to manifest?
Childhood to teens

How does it present? What is seen at the slit lamp?
Early in the disease, fleck-like central opacities and a diffuse stromal haze are present. Later, myriad crisscrossing **re refractile lines in the cornea** predominate.

What word is used to describe the appearance of the lines?

Glasslike

Endothelial Dystrophies
Epithelial and Subepithelial Dystrophies

**Epithelial-Stromal TGFBI Dystrophies**
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**Stromal Dystrophies**

**Endothelial Dystrophies**

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**Corneal Dystrophies**

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*What word is used to describe the appearance of the lines?*
‘Glasslike’
Lattice corneal dystrophy, type 1 (aka classic lattice). C, Subepithelial groundglass haze of the central and inferior cornea, and diffuse lattice lines in advanced disease. D, Dots and paracentral lattice lines in retroillumination.
Epithelial and Subepithelial Dystrophies

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Stromal Dystrophies

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Epithelial and Subepithelial Dystrophies

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Endothelial Dystrophies
Epithelial and Subepithelial Dystrophies

Epithelial-Stromal *TGFBI* Dystrophies
1) Reis-Bücklers corneal dystrophy
2) Thiel-Behnke corneal dystrophy
3) Lattice, type 1
4) Lattice, variant types (III, IIIA, I/IIIA, IV)
5) Granular type 1
6) Granular type 2

Stromal Dystrophies

Endothelial Dystrophies

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**Corneal Dystrophies**

*At what age does classic lattice type 1 begin to manifest?*
Childhood to teens

*How does it present? What is seen at the slit lamp?*
Early in the disease, fleck-like central opacities and a diffuse stromal haze are present. Later, myriad crisscrossing refractile lines in the cornea predominate.

*Is it painful?*
Yes; pts get recurrent epithelial erosions

*Does it affect vision?*
Surface irregularity and stromal haze often results in decreased vision

*What is the hallmark of lattice type 1 on light microscopy?*
The presence of **amyloid** in the subepithelial space and anterior stroma
Lattice corneal dystrophy, type 1 (classic lattice). E, Light microscopy: Congo red prominently stains a continuous layer of amyloid (asterisk) that underlies and partially destroys the Bowman layer and intrastromal amyloid deposits corresponding to lattice lines (arrowheads). F, This same section viewed with polarized light confirms deposits are birefringent and red-green dichroic, thus amyloid. G, In vivo confocal microscopy image shows filaments corresponding to lattice lines within the stroma.
Corneal Dystrophies

Epithelial and Subepithelial Dystrophies

Epithelial-Stromal TGFBI Dystrophies

1) Reis-Bücklers corneal dystrophy
2) Thiel-Behnke corneal dystrophy
3) Lattice, type 1
4) Lattice, variant types (III, IIIA, I/IIIA, IV)
5) Granular type 1
6) Granular type 2

Why is lattice, gelsolin type not grouped with lattice?

Lattice, gelsolin type

Stromal Dystrophies

Endothelial Dystrophies
Epithelial and Subepithelial Dystrophies

Epithelial-Stromal $TGFBI$ Dystrophies
1) Reis-Bücklers corneal dystrophy
2) Thiel-Behnke corneal dystrophy
3) Lattice, type 1
4) Lattice, variant types (III, IIA, I/IIIA, IV)
5) Granular type 1
6) Granular type 2

**Why is lattice, gelsolin type not grouped with lattice?**
Because it is part of a systemic syndrome, it is no longer classified as a corneal dystrophy. (But given its corneal findings, we will touch briefly on it here.)

Stromal Dystrophies

Endothelial Dystrophies
**Corneal Dystrophies**

Epithelial and Subepithelial Dystrophies

**Epithelial-Stromal TGFBI Dystrophies**

1) Reis-Bücklers corneal dystrophy
2) Thiel-Behnke corneal dystrophy
3) Lattice, type 1
4) Lattice, variant types (III, IIIA, I/IIIA, IV)
5) Granular type 1
6) Granular type 2

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**Stromal Dys (aka... syndrome)**

- Why is lattice, gelsolin type *not grouped with lattice*? Because it is part of a *systemic syndrome*, it is no longer classified as a corneal dystrophy. (But given its corneal findings, we will touch briefly on it here.)

- What is the name of the syndrome?

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Endothelial Dystrophies
Corneal Dystrophies

Epithelial and Subepithelial Dystrophies

Epithelial-Stromal *TGFBI* Dystrophies

1) Reis-Bücklers corneal dystrophy
2) Thiel-Behnke corneal dystrophy
3) Lattice, type 1
4) Lattice, variant types (III, IIIA, I/IIIA, IV)
5) Granular type 1
6) Granular type 2

*Why is lattice, gelsolin type not grouped with lattice?*  
Because it is part of a **systemic syndrome**, it is no longer classified as a corneal dystrophy. (But given its corneal findings, we will touch briefly on it here.)

*What is the name of the syndrome?*  
**Meretoja syndrome**

Stromal Dys (aka...**Meretoja syndrome**)
Epithelial and Subepithelial Dystrophies

**Epithelial-Stromal** $TGFBI$ Dystrophies

1) Reis-Bücklers corneal dystrophy
2) Thiel-Behnke corneal dystrophy
3) Lattice, type 1
4) Lattice, variant types (III, IIIA, I/IIIA, IV)
5) Granular type 1
6) Granular type 2

*Why is lattice, gelsolin type not grouped with lattice?*
Because it is part of a **systemic syndrome**, it is no longer classified as a corneal dystrophy. (But given its corneal findings, we will touch briefly on it here.)

*What is the name of the syndrome?*
**Meretoja syndrome**

*What are the systemic findings in Meretoja syndrome?*

**Amyloid deposition in the skin and perineural space leads to dermatochalasis, saggy skin, pendulous ears, and bilateral CN7 palsies. These findings culminate in a characteristic facial appearance known as 'bloodhound facies'**.
Corneal Dystrophies

Epithelial and Subepithelial Dystrophies

Epithelial-Stromal \textit{TGFB1} Dystrophies

1) Reis-Bücklers corneal dystrophy
2) Thiel-Behnke corneal dystrophy
3) Lattice, type 1
4) Lattice, variant types (III, IIIA, I/IIIA, IV)
5) Granular type 1
6) Granular type 2

\textit{Lattice, gelsolin type (aka...Meretoja syndrome)}

Why is lattice, gelsolin type not grouped with lattice? Because it is part of a \textbf{systemic syndrome}, it is no longer classified as a corneal dystrophy. (But given its corneal findings, we will touch briefly on it here.)

What is the name of the syndrome? \textbf{Meretoja syndrome}

What are the systemic findings in Meretoja syndrome? Deposition in the skin and perineural space leads to dermatochalasis, saggy skin, pendulous ears, and bilateral CN7 palsies.

Stromal Dys

Endothelial Dystrophies
Corneal Dystrophies

Epithelial and Subepithelial Dystrophies

Epithelial-Stromal \textit{TGFBI} Dystrophies

1) Reis-Bücklers corneal dystrophy
2) Thiel-Behnke corneal dystrophy
3) Lattice, type 1
4) Lattice, variant types (III, IIIA, I/IIIA, IV)
5) Granular type 1
6) Granular type 2

\textbf{Lattice, gelsolin type}

Stromal Dys (aka...Meretoja syndrome)

\textbf{Why is lattice, gelsolin type not grouped with lattice?}
Because it is part of a \textit{systemic syndrome}, it is no longer classified as a corneal dystrophy. (But given its corneal findings, we will touch briefly on it here.)

\textbf{What is the name of the syndrome?}
\textbf{Meretoja syndrome}

\textbf{What are the systemic findings in Meretoja syndrome?}
Amyloid deposition in the skin and perineural space leads to dermatochalasis, saggy skin, pendulous ears, and bilateral CN7 palsies. These
Epithelial-Stromal \textit{TGFB1} Dystrophies

1) Reis-Bücklers corneal dystrophy
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\textit{Lattice, gelsolin type}

\textbf{Stromal Dys (aka...Meretoja syndrome)}

Why is lattice, gelsolin type not grouped with lattice? Because it is part of a \textbf{systemic syndrome}, it is no longer classified as a corneal dystrophy. (But given its corneal findings, we will touch briefly on it here.)

\textbf{What is the name of the syndrome?}
\textbf{Meretoja syndrome}

\textbf{What are the systemic findings in Meretoja syndrome?}
Amyloid deposition in the skin and perineural space leads to dermatochalasis, saggy skin, pendulous ears, and bilateral CN7 palsies. These findings culminate in a characteristic facial appearance known as 'two words'.
**Epithelial-Stromal** *TGFBI* **Dystrophies**

1) Reis-Bücklers corneal dystrophy  
2) Thiel-Behnke corneal dystrophy  
3) Lattice, type 1  
4) Lattice, variant types (III, IIIA, I/IIIA, IV)  
5) Granular type 1  
6) Granular type 2  

*Lattice, gelsolin type*

**Corneal Dystrophies**

Why is lattice, gelsolin type *not grouped with lattice*? Because it is part of a **systemic syndrome**, it is no longer classified as a corneal dystrophy. (But given its corneal findings, we will touch briefly on it here.)

**What is the name of the syndrome?**  
**Meretoja syndrome**

**What are the systemic findings in Meretoja syndrome?**  
Amyloid deposition in the skin and perineural space leads to dermatochalasis, saggy skin, pendulous ears, and bilateral CN7 palsies. These findings culminate in a characteristic facial appearance known as ‘**bloodhound facies**.’
Meretoja syndrome: ‘Bloodhound facies’
Endothelial and Subepithelial Dystrophies

Epithelial-Stromal TGFBI Dystrophies
1) Reis-Bücklers corneal dystrophy
2) Thiel-Behnke corneal dystrophy
3) Lattice, type 1
4) Lattice, variant types (III, IIIA, IV)
5) Granular type 1
6) Granular type 2

Why is lattice, gelsolin type not grouped with lattice? Because it is part of a systemic syndrome, it is no longer classified as a corneal dystrophy. (But given its corneal findings, we will touch briefly on it here.)

What is the name of the syndrome? Meretoja syndrome

What are the systemic findings in Meretoja syndrome? Amyloid deposition in the skin and perineural space leads to dermatochalasis, saggy skin, pendulous ears, and bilateral CN7 palsies. These findings culminate in a characteristic facial appearance known as ‘bloodhound facies.’

What about the corneal findings?
Corneal Dystrophies

Epithelial and Subepithelial Dystrophies

Epithelial-Stromal TGFBI Dystrophies
1) Reis-Bücklers corneal dystrophy
2) Thiel-Behnke corneal dystrophy
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Lattice, gelsolin type

Stromal Dys (aka…Meretoja syndrome)

Why is lattice, gelsolin type not grouped with lattice?
Because it is part of a systemic syndrome, it is no longer classified as a corneal dystrophy. (But given its corneal findings, we will touch briefly on it here.)

What is the name of the syndrome?
Meretoja syndrome

What are the systemic findings in Meretoja syndrome?
Amyloid deposition in the skin and perineural space leads to dermatochalasis, saggy skin, pendulous ears, and bilateral CN7 palsies. These findings culminate in a characteristic facial appearance known as ‘bloodhound facies.’

What about the corneal findings?
They are essentially identical to those of lattice type 1
Epithelial and Subepithelial Dystrophies

Epithelial-Stromal *TGFBI* Dystrophies

1) Reis-Bücklers corneal dystrophy
2) Thiel-Behnke corneal dystrophy
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Stromal Dystrophies

Endothelial Dystrophies
Epithelial and Subepithelial Dystrophies

**Epithelial-Stromal TGFBI Dystrophies**

1. Reis-Bücklers corneal dystrophy
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5. **Granular type 1**
6. Granular type 2

**Stromal Dystrophies**

**Endothelial Dystrophies**

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**At what age does GCD1 begin to manifest?**

Early childhood

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**Corneal Dystrophies**
Epithelial and Subepithelial Dystrophies

Epithelial-Stromal TGFBI Dystrophies
1) Reis-Bücklers corneal dystrophy
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Stromal Dystrophies

Endothelial Dystrophies

Corneal Dystrophies

At what age does GCD1 begin to manifest?
Early childhood

How does it present? What is seen at the slit lamp?
At what age does GCD1 begin to manifest?
Early childhood

How does it present? What is seen at the slit lamp?
In early disease, tiny crumblike granules appear.
Granular corneal dystrophy, type 1. A, In a child, early subepithelial verticillate-like opacities are evident by retro and direct illumination. B, With broad slit illumination, stromal deposits are both discrete and confluent, and are axially distributed in anterior stroma.
Epithelial and Subepithelial Dystrophies

Epithelial-Stromal TGFBI Dystrophies

1) Reis-Bücklers corneal dystrophy
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6) Granular type 2

At what age does GCD1 begin to manifest?
Early childhood

How does it present? What is seen at the slit lamp?
In early disease, tiny crumblike granules appear. As the disease progresses, the size and density of the granules increases. The granules never reach as far as the limbus.

Corneal Dystrophies

Stromal Dystrophies

Endothelial Dystrophies
Granular corneal dystrophy, type 1. In an adult, more prominent diffuse granular opacities in the form of “snowfall” are apparent with direct (C) and retroillumination (D).
Corneal Dystrophies

Epithelial and Subepithelial Dystrophies

Epithelial-Stromal TGFBI Dystrophies

1) Reis-Bücklers corneal dystrophy
2) Thiel-Behnke corneal dystrophy
3) Lattice, type 1
4) Lattice, variant types (III, IIIA, I/IIIA, IV)
5) Granular type 1
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At what age does GCD1 begin to manifest?
Early childhood

How does it present? What is seen at the slit lamp?
In early disease, tiny crumblike granules appear. As the disease progresses, the size and density of the granules increases. The granules never reach as far as the limbus.

Is it painful?

Yes; pts get recurrent epithelial erosions

Does it affect vision?
Glare and stromal haze result in decreased vision

What is the histologic hallmark of GCD1 on light microscopy?
The presence of hyaline in the subepithelial space and anterior stroma (just like Reis-Bücklers). It stains bright red with Masson trichrome (just like Reis-Bücklers).

What is the hallmark of GCD1 on electron microscopy?
Rod-shaped fibers (just like Reis-Bücklers)

Stromal Dystrophies

Endothelial Dystrophies
Epithelial and Subepithelial Dystrophies

Epithelial-Stromal TGFBI Dystrophies

1) Reis-Bücklers corneal dystrophy
2) Thiel-Behnke corneal dystrophy
3) Lattice, type 1
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Corneal Dystrophies

At what age does GCD1 begin to manifest?
Early childhood

How does it present? What is seen at the slit lamp?
In early disease, tiny crumblike granules appear. As the disease progresses, the size and density of the granules increases. The granules never reach as far as the limbus.

Is it painful?
Yes; pts get recurrent epithelial erosions

Endothelial Dystrophies
Epithelial and Subepithelial Dystrophies

Epithelial-Stromal TGFBI Dystrophies
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Stromal Dystrophies

Endothelial Dystrophies

**At what age does GCD1 begin to manifest?**
Early childhood

**How does it present? What is seen at the slit lamp?**
In early disease, tiny crumblike granules appear. As the disease progresses, the size and density of the granules increases. The granules never reach as far as the limbus.

**Is it painful?**
Yes; pts get recurrent epithelial erosions

**Does it affect vision?**
Glare and stromal haze result in decreased vision

**What is the histologic hallmark of GCD1 on light microscopy?**
The presence of hyaline in the subepithelial space and anterior stroma (just like Reis-Bücklers). It stains bright red with Masson trichrome (just like Reis-Bücklers).

**What is the hallmark of GCD1 on electron microscopy?**
Rod-shaped fibers (just like Reis-Bücklers)
Epithelial and Subepithelial Dystrophies

Epithelial-Stromal TGFBI Dystrophies
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6) Granular type 2

Stromal Dystrophies

Endothelial Dystrophies

**Corneal Dystrophies**

*At what age does GCD1 begin to manifest?*
Early childhood

*How does it present? What is seen at the slit lamp?*
In early disease, tiny crumblike granules appear. As the disease progresses, the size and density of the granules increases. The granules never reach as far as the limbus.

*Is it painful?*
Yes; pts get recurrent epithelial erosions

*Does it affect vision?*
Glare and stromal haze result in decreased vision
Corneal Dystrophies

Epithelial and Subepithelial Dystrophies

1) Reis-Bücklers corneal dystrophy
2) Thiel-Behnke corneal dystrophy
3) Lattice, type 1
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Epithelial-Stromal TGFBI Dystrophies

Stromal Dystrophies

Endothelial Dystrophies

At what age does GCD1 begin to manifest?
Early childhood

How does it present? What is seen at the slit lamp?
In early disease, tiny crumblike granules appear. As the disease progresses, the size and density of the granules increases. The granules never reach as far as the limbus.

Is it painful?
Yes; pts get recurrent epithelial erosions

Does it affect vision?
Glare and stromal haze result in decreased vision

What is the histologic hallmark of GCD1 on light microscopy?
**Epithelial and Subepithelial Dystrophies**

**Epithelial-Stromal TGFBI Dystrophies**
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**Stromal Dystrophies**

**Endothelial Dystrophies**

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**At what age does GCD1 begin to manifest?**
Early childhood

**How does it present? What is seen at the slit lamp?**
In early disease, tiny crumblike granules appear. As the disease progresses, the size and density of the granules increases. The granules never reach as far as the limbus.

**Is it painful?**
Yes; pts get recurrent epithelial erosions

**Does it affect vision?**
Glare and stromal haze result in decreased vision

**What is the histologic hallmark of GCD1 on light microscopy?**
The presence of **one word** in the subepithelial space and anterior stroma (just like Reis-Bücklers). It stains bright **color** (just like Reis-Bücklers).
Corneal Dystrophies

Epithelial and Subepithelial Dystrophies

Epithelial-Stromal TGFBI Dystrophies

1) Reis-Bücklers corneal dystrophy
2) Thiel-Behnke corneal dystrophy
3) Lattice, type 1
4) Lattice, variant types (III, IIIA, I/IIIA, IV)
5) Granular type 1
6) Granular type 2

Stromal Dystrophies

Endothelial Dystrophies

At what age does GCD1 begin to manifest?
Early childhood

How does it present? What is seen at the slit lamp?
In early disease, tiny crumblike granules appear. As the disease progresses, the size and density of the granules increases. The granules never reach as far as the limbus.

Is it painful?
Yes; pts get recurrent epithelial erosions

Does it affect vision?
Glare and stromal haze result in decreased vision

What is the histologic hallmark of GCD1 on light microscopy?
The presence of hyaline in the subepithelial space and anterior stroma (just like Reis-Bücklers). It stains bright red with Masson trichrome (just like Reis-Bücklers).
Granular corneal dystrophy, type 1. Light microscopy—Masson trichrome highlights deposits of hyaline at various stromal layers and partial destruction of the Bowman layer (between arrowheads).
Epithelial and Subepithelial Dystrophies

**Epithelial-Stromal TGFBI Dystrophies**
1. Reis-Bücklers corneal dystrophy
2. Thiel-Behnke corneal dystrophy
3. Lattice, type 1
4. Lattice, variant types (III, IIIA, I/IIIA, IV)
5. **Granular type 1**
6. Granular type 2

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**Corneal Dystrophies**

*At what age does GCD1 begin to manifest?*
Early childhood

*How does it present? What is seen at the slit lamp?*
In early disease, tiny crumblike granules appear. As the disease progresses, the size and density of the granules increases. The granules never reach as far as the limbus.

*Is it painful?*
Yes; pts get recurrent epithelial erosions

*Does it affect vision?*
Glare and stromal haze result in decreased vision

*What is the histologic hallmark of GCD1 on light microscopy?*
The presence of **hyaline** in the subepithelial space and anterior stroma (just like Reis-Bücklers). It stains bright **red** with **Masson trichrome** (just like Reis-Bücklers).

*What is the hallmark of GCD1 on electron microscopy?*
**Corneal Dystrophies**

**Epithelial and Subepithelial Dystrophies**

**Epithelial-Stromal TGFBI Dystrophies**
1) Reis-Bücklers corneal dystrophy
2) Thiel-Behnke corneal dystrophy
3) Lattice, type 1
4) Lattice, variant types (III, IIIA, I/IIIA, IV)
5) **Granular type 1**
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**Stromal Dystrophies**

**Endothelial Dystrophies**

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**At what age does GCD1 begin to manifest?**
Early childhood

**How does it present? What is seen at the slit lamp?**
In early disease, tiny crumblike granules appear. As the disease progresses, the size and density of the granules increases. The granules never reach as far as the limbus.

**Is it painful?**
Yes; pts get recurrent epithelial erosions

**Does it affect vision?**
Glare and stromal haze result in decreased vision

**What is the histologic hallmark of GCD1 on light microscopy?**
The presence of **hyaline** in the subepithelial space and anterior stroma (just like Reis-Bücklers). It stains bright **red** with **Masson trichrome** (just like Reis-Bücklers).

**What is the hallmark of GCD1 on electron microscopy?**
(just like Reis-Bücklers)
Epithelial and Subepithelial Dystrophies

Epithelial-Stromal TGFBI Dystrophies
1) Reis-Bücklers corneal dystrophy
2) Thiel-Behnke corneal dystrophy
3) Lattice, type 1
4) Lattice, variant types (II, II A, III, III A, IV)
5) Granular type 1
6) Granular type 2

Stromal Dystrophies

Endothelial Dystrophies

Corneal Dystrophies

At what age does GCD1 begin to manifest?
Early childhood

How does it present? What is seen at the slit lamp?
In early disease, tiny crumblike granules appear. As the disease progresses, the size and density of the granules increases. The granules never reach as far as the limbus.

Is it painful?
Yes; pts get recurrent epithelial erosions

Does it affect vision?
Glare and stromal haze result in decreased vision

What is the histologic hallmark of GCD1 on light microscopy?
The presence of hyaline in the subepithelial space and anterior stroma (just like Reis-Bücklers). It stains bright red with Masson trichrome (just like Reis-Bücklers).

What is the hallmark of GCD1 on electron microscopy?
Rod-shaped fibers (just like Reis-Bücklers)
Corneal Dystrophies

Epithelial and Subepithelial Dystrophies

Epithelial-Stromal TGFBI Dystrophies

1) Reis-Bücklers corneal dystrophy
2) Thiel-Behnke corneal dystrophy
3) Lattice, type 1
4) Lattice, variant types (III, IIIA, I/IIIA, IV)
5) Granular type 1
6) Granular type 2

At what age does GCD1 begin to manifest?
Early childhood

How does it present? What is seen at the slit lamp?
In early disease, tiny crumblike granules appear. As the disease progresses, the size and density of the granules increases. The granules never reach as far as the limbus.

Is it painful?
Yes; pts get recurrent epithelial erosions

Does it affect vision?
Glare and stromal haze result in decreased vision

What is the histologic hallmark of GCD1 on light microscopy?
The presence of hyaline in the subepithelial space and anterior stroma (just like Reis-Bücklers). It stains bright red with Masson trichrome (just like Reis-Bücklers).

What is the hallmark of GCD1 on electron microscopy?
Rod-shaped fibers (just like Reis-Bücklers)

Stromal Dystrophies

Endothelial Dystrophies
Corneal Dystrophies

Epithelial and Subepithelial Dystrophies

Epithelial-Stromal TGFB1 Dystrophies

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6) Granular type 2

By what other names is GCD2 known?

- Combined granular-lattice dystrophy
- Avellino dystrophy

At what age does GCD2 begin to manifest?

Usually in the teen to early-adult years, can be younger

How does it present? What is seen at the slit lamp?

Basically, as a combination of GCD1 and lattice dystrophy: The crumblike granules of GCD1, along with a version of the lattice lines seen in LCD

Is it painful?

Yes; pts get recurrent epithelial erosions

Does it affect vision?

Glare and stromal haze result in decreased vision

What is the histologic hallmark of GCD1 on light microscopy?

The presence of both amyloid and hyaline in the subepithelial space and anterior stroma. The hyaline stains bright red with Masson trichrome; the amyloid stains with Congo Red, birefringes under polarized light.

What is the hallmark of GCD2 on electron microscopy?

Rod-shaped fibers, albeit in reduced numbers

Endothelial Dystrophies

Stromal Dystrophies
Epithelial and Subepithelial Dystrophies

Epithelial-Stromal TGFBI Dystrophies
1) Reis-Bücklers corneal dystrophy
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5) Granular type 1
6) **Granular type 2**

By what other names is GCD2 known?
Combined granular-lattice dystrophy; Avellino dystrophy

Stromal Dystrophies

Endothelial Dystrophies
Epithelial and Subepithelial Dystrophies

Epithelial-Stromal TGFBI Dystrophies

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Corneal Dystrophies

By what other names is GCD2 known?
Combined granular-lattice dystrophy; Avellino dystrophy

At what age does GCD2 begin to manifest?

Epithelial-Stromal TGFBI Dystrophies

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Stromal Dystrophies

Endothelial Dystrophies
Epithelial and Subepithelial Dystrophies

Epithelial-Stromal **TGFBI** Dystrophies

1) Reis-Bücklers corneal dystrophy
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**Corneal Dystrophies**

**By what other names is GCD2 known?**
Combined granular-lattice dystrophy; Avellino dystrophy

**At what age does GCD2 begin to manifest?**
Usually in the teen to early-adult years, can be younger

Stromal Dystrophies

Endothelial Dystrophies
Corneal Dystrophies

Epithelial and Subepithelial Dystrophies

Epithelial-Stromal *TGFBI* Dystrophies

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By what other names is GCD2 known?
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Granular corneal dystrophy, type 2. 13-year-old with a few white dots.
Granular corneal dystrophy, type 2. Direct and retro-illumination views of an older pt demonstrate branching, star-shaped, spiny, and ring-like deposits.
Epithelial-Stromal 

**TGFB1** Dystopathies

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**Epithelial and Subepithelial Dystrophies**
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**Granular corneal dystrophy, type 2.** E, Light microscopy—Masson trichrome stains sub-Bowman and anterior stromal hyaline deposits red (arrowheads). Note that the deeper stromal layers do not have hyaline granules (asterisk). In the deep stroma, small amyloid deposits stain with Congo red (inset).
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--They tend to be shorter

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**How do the lattice lines in GCD2 differ from those in lattice dystrophy?**
--They tend to be shorter
--They tend to be whiter and less ‘glasslike’ (ie, not as refractile)

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Endothelial Dystrophies

How do the lattice lines in GCD2 differ from those in lattice dystrophy?

--They tend to be **shorter**
--They tend to be **whiter** and less ‘glasslike’ (ie, not as refractile)
--They tend not to **amen** (three words) (ie, so they are less reminiscent of an actual lattice structure)
**Corneal Dystrophies**

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These two primarily involve Bowman’s membrane

There are two ‘lattice’ forms

Two are ‘granular’ forms

Stromal Dystrophies

As a way to help remember them, think of the TGFBI dystrophies as consisting of three pairs of conditions: Two Bowman’s dystrophies, two lattice dystrophies, and two granular dystrophies

Endothelial Dystrophies