Galactosemia

- Inability to convert galactose to...
Galactosemia

- Inability to convert galactose to glucose
Galactosemia

- Inability to convert galactose to glucose
- Inheritance: AR
Galactosemia

- Inability to convert galactose to glucose
- Inheritance: AR
Galactosemia

- Inability to convert galactose to glucose
- Inheritance: AR
- Results from a defect in one of the enzymes involved in galactose metabolism
Galactosemia

- Inability to convert galactose to glucose
- Inheritance: AR
- Results from a defect in one of the three enzymes involved in galactose metabolism
- Inability to convert galactose to glucose
- Inheritance: AR
- Results from a defect in one of the three enzymes involved in galactose metabolism
- Classic galactosemia:
  - most vs least common and most vs least severe form
Galactosemia

- Inability to convert galactose to glucose
- Inheritance: AR
- Results from a defect in one of the three enzymes involved in galactose metabolism
- Classic galactosemia:
  - Most common and most severe form
Inability to convert galactose to glucose
- Inheritance: AR
- Results from a defect in one of the three enzymes involved in galactose metabolism
- **Classic galactosemia:**
  - Most common and most severe form
  - Caused by defect in the uridyltransferase enzyme
Inability to convert galactose to glucose
Inheritance: AR
Results from a defect in one of the three enzymes involved in galactose metabolism
Classic galactosemia:
  Most common and most severe form
  Caused by defect in the uridyltransferase enzyme
Inability to convert galactose to glucose

Inheritance: AR

Results from a defect in one of the three enzymes involved in galactose metabolism

Classic galactosemia:

- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
Galactosemia

- Inability to convert galactose to **glucose**
- Inheritance: **AR**
- Results from a defect in one of the **three** enzymes involved in galactose metabolism
- **Classic galactosemia:**
  - **Most** common and **most** severe form
  - Caused by defect in the **uridyltransferase** enzyme
  - 75% develop cataracts within **days to weeks** of birth
Inability to convert galactose to glucose
Inheritance: AR
Results from a defect in one of the three enzymes involved in galactose metabolism

Classic galactosemia:
- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as ‘oil droplet’
Inability to convert galactose to glucose
Inheritance: AR
Results from a defect in one of the three enzymes involved in galactose metabolism

Classic galactosemia:
- Most common and most severe form
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Galactosemia

- Inability to convert galactose to glucose
- Inheritance: AR
- Results from a defect in one of the three enzymes involved in galactose metabolism

**Classic galactosemia:**
- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as ‘oil droplet’

*What simple exam maneuver is the best way to pick up an oil droplet cataract?*
Inability to convert galactose to glucose
Inheritance: AR
Results from a defect in one of the three enzymes involved in galactose metabolism
Classic galactosemia:
  - Most common and most severe form
  - Caused by defect in the uridyltransferase enzyme
  - 75% develop cataracts within days to weeks of birth
    - Starts as ‘oil droplet’

*What simple exam maneuver is the best way to pick up an oil droplet cataract?*
Retroillumination
Galactosemia

Oil-droplet cataract: Retroillumination
Galactosemia: Oil-droplet cataracts
Galactosemia

- Inability to convert galactose to glucose
- Inheritance: AR
- Results from a defect in one of the three enzymes involved in galactose metabolism

*Classic galactosemia:*
- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as ‘oil droplet’; progresses to

```plaintext
classic two-word description
```
Inability to convert galactose to glucose

Inheritance: AR

Results from a defect in one of the three enzymes involved in galactose metabolism

Classic galactosemia:

- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as ‘oil droplet’; progresses to total opacification
Inability to convert galactose to glucose

Inheritance: AR

Results from a defect in one of the three enzymes involved in galactose metabolism

**Classic galactosemia:**

- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as 'oil droplet'; progresses to total opacification

*When you hear oil droplet cataract, three conditions should come to mind. One is galactosemia; what are the other two? (Note: Both are named for the shape of the lens.)*
Inability to convert galactose to glucose

Inheritance: AR

Results from a defect in one of the three enzymes involved in galactose metabolism

Classic galactosemia:

- Most common and most severe form
- Caused by defect in the *uridyltransferase* enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as 'oil droplet'; progresses to total opacification

*When you hear* oil droplet *cataract, three conditions should come to mind. One is* galactosemia; *what are the other two? (Note: Both are named for the shape of the lens.)*

Anterior and posterior
Inability to convert galactose to glucose
Inheritance: AR
Results from a defect in one of the three enzymes involved in galactose metabolism
Classic galactosemia:
- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as 'oil droplet'; progresses to total opacification

When you hear oil droplet cataract, three conditions should come to mind. One is galactosemia; what are the other two? (Note: Both are named for the shape of the lens.)
Anterior and posterior lenticonus
Inability to convert galactose to glucose
Inheritance: AR
Results from a defect in one of the three enzymes involved in galactose metabolism
Classic galactosemia:
- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as 'oil droplet'; progresses to total opacification

When you hear oil droplet cataract, three conditions should come to mind. One is galactosemia; what are the other two? (Note: Both are named for the shape of the lens.)

Anterior and posterior lenticonus

In one (unsurprising) word, what is the shape of the affected lens surface in lenticonus?
Inability to convert galactose to glucose
Inheritance: AR
Results from a defect in one of the three enzymes involved in galactose metabolism

Classic galactosemia:
- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as 'oil droplet'; progresses to total opacification

When you hear oil droplet cataract, three conditions should come to mind. One is galactosemia; what are the other two? (Note: Both are named for the shape of the lens.)

Anterior and posterior lenticonus

In one (unsurprising) word, what is the shape of the affected lens surface in lenticonus? ‘Conical.’
Galactosemia

The conical shape of a lenticous lens
Inability to convert galactose to glucose

Inheritance: AR

Results from a defect in one of the three enzymes involved in galactose metabolism

Classic galactosemia:
- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as ‘oil droplet’; progresses to total opacification

When you hear oil droplet cataract, three conditions should come to mind. One is galactosemia; what are the other two? (Note: Both are named for the shape of the lens.)

Anterior and posterior lenticonus

In one (unsurprising) word, what is the shape of the affected lens surface in lenticonus? ‘Conical.’ So, there’s a fundamental difference between the cause of the oil-droplet appearance in lenticonus (= increased central lenticular power) vs that of galactosemia (= the accumulation of galactitol and fluid in the central lens).
Galactosemia

- Inability to convert galactose to glucose
- Inheritance: AR
- Results from a defect in one of the three enzymes involved in galactose metabolism
- **Classic galactosemia:**
  - Most common and most severe form
  - Caused by defect in the uridyltransferase enzyme
  - 75% develop cataracts within days to weeks of birth
    - Starts as 'oil droplet'; progresses to total opacification

When you hear oil droplet cataract, three conditions should come to mind. One is galactosemia; what are the other two? (Note: Both are named for the shape of the lens.)

**Most cases of anterior lenticonus are bilateral and syndromic.** When it is unilateral, it is usually sporadic. With what syndrome is anterior lenticonus strongly associated?

**Anterior and posterior lenticonus**
Inability to convert galactose to glucose

Inheritance: AR

Results from a defect in one of the three enzymes involved in galactose metabolism

Classic galactosemia:
- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as 'oil droplet'; progresses to total opacification

Galactosemia

When you hear oil droplet cataract, three conditions should come to mind. One is galactosemia; what are the other two? (Note: Both are named for the shape of the lens.)

Most cases of anterior lenticonus are bilateral and syndromic

Anterior and posterior lenticonus

Most cases of anterior lenticonus are bilateral and syndromic

allopurinol
- Inability to convert galactose to glucose
- Inheritance: AR
- Results from a defect in one of the three enzymes involved in galactose metabolism
- **Classic galactosemia:**
  - Most common and most severe form
  - Caused by defect in the *uridyltransferase* enzyme
  - 75% develop cataracts within days to weeks of birth
    - Starts as ‘oil droplet’; progresses to total opacification

**Galactosemia**

Most cases of anterior lenticonus are bilateral and syndromic. When it is unilateral, it is usually for the shape of the lens.)

Anterior and posterior lenticonus
Inability to convert galactose to glucose
Inheritance: AR
Results from a defect in one of the three enzymes involved in galactose metabolism
Classic galactosemia:
- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as 'oil droplet'; progresses to total opacification

Most cases of anterior lenticonus are bilateral and syndromic. When it is unilateral, it is usually sporadic.

Anterior and posterior lenticonus
Galactosemia

Anterior lenticonus
Inability to convert galactose to glucose
Inheritance: AR
Results from a defect in one of the three enzymes involved in galactose metabolism

**Classic galactosemia:**
- Most common and most severe form
- Caused by defect in the uridylyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as 'oil droplet'; progresses to total opacification

*Most cases of anterior lenticonus are bilateral and syndromic.* When it is unilateral, it is usually sporadic. With what syndrome is anterior lenticonus strongly associated?

**Anterior** and posterior **lenticonus**
Inability to convert galactose to glucose
Inheritance: AR
Results from a defect in one of the three enzymes involved in galactose metabolism

**Classic galactosemia:**
- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as 'oil droplet'; progresses to total opacification

Most cases of anterior lenticonus are bilateral and syndromic. When it is unilateral, it is usually sporadic. With what syndrome is anterior lenticonus strongly associated? Alport syndrome

**Anterior and posterior lenticonus**

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**Galactosemia**
Inability to convert galactose to glucose
Inheritance: AR
Results from a defect in one of the three enzymes involved in galactose metabolism

**Classic galactosemia:**
- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as ‘oil droplet’; progresses to total opacification

**Galactosemia**

Most cases of anterior lenticonus are bilateral and syndromic. When it is unilateral, it is usually sporadic. With what syndrome is anterior lenticonus strongly associated?
Alport syndrome

Most cases of posterior lenticonus are unilateral vs bilateral and inherited vs sporadic.
Inability to convert galactose to glucose
Inheritance: AR
Results from a defect in one of the three enzymes involved in galactose metabolism

**Classic galactosemia:**
- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as 'oil droplet'; progresses to total opacification

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**Galactosemia**

- Inability to convert galactose to glucose
- Inheritance: AR
- Results from a defect in one of the three enzymes involved in galactose metabolism

**Classic galactosemia:**
- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as 'oil droplet'; progresses to total opacification

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**Anterior and posterior lenticonus**

Most cases of anterior lenticonus are bilateral and syndromic. When it is unilateral, it is usually sporadic. With what syndrome is anterior lenticonus strongly associated? Alport syndrome

Most cases of posterior lenticonus are unilateral and sporadic.
Galactosemia

Posterior lenticonus
Inability to convert galactose to glucose
Inheritance: AR
Results from a defect in one of the three enzymes involved in galactose metabolism

**Classic galactosemia:**
- Most common and most severe form
- Caused by defect in the *uridyltransferase* enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as ‘oil droplet’; progresses to total opacification

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**Galactosemia**

- Most cases of anterior lenticonus are **bilateral** and **syndromic**. *When it is unilateral, it is usually sporadic*. With what syndrome is anterior lenticonus strongly associated? Alport syndrome
- Most cases of posterior lenticonus are **unilateral** and **sporadic**. *When it is bilateral, it is usually** .
Galactosemia

- Inability to convert galactose to glucose
- Inheritance: AR
- Results from a defect in one of the three enzymes involved in galactose metabolism

**Classic galactosemia:**
- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as 'oil droplet'; progresses to total opacification

---

When you hear 'oil droplet cataract', three conditions should come to mind. One is galactosemia; what are the other two? (Note: Both are named for the shape of the lens.)

**Anterior and posterior lenticonus**

Most cases of anterior lenticonus are bilateral and syndromic. When it is unilateral, it is usually sporadic. With what syndrome is anterior lenticonus strongly associated? Alport syndrome

**Posterior lenticonus**

Most cases of posterior lenticonus are unilateral and sporadic. When it is bilateral, it is usually syndromic.
Galactosemia

- Inability to convert galactose to glucose
- Inheritance: AR
- Results from a defect in one of the three enzymes involved in galactose metabolism
- **Classic galactosemia:**
  - Most common and most severe form
  - Caused by defect in the *uridyltransferase* enzyme
  - 75% develop cataracts within days to weeks of birth
    - Starts as 'oil droplet'; progresses to total opacification

---

**Most cases of anterior lenticonus are bilateral and syndromic.** When it is unilateral, it is usually sporadic. With what syndrome is anterior lenticonus strongly associated? Alport syndrome

**Most cases of posterior lenticonus are unilateral and sporadic.** When it is bilateral, it is usually syndromic. With what syndrome is posterior lenticonus strongly associated?
Galactosemia

- Inability to convert galactose to glucose
- Inheritance: AR
- Results from a defect in one of the three enzymes involved in galactose metabolism

**Classic galactosemia:**
- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as 'oil droplet'; progresses to total opacification

---

When you hear oil droplet cataract, three conditions should come to mind. One is galactosemia; what are the other two? (Note: Both are named for the shape of the lens.)

- Anterior and posterior lenticonus

Most cases of anterior lenticonus are bilateral and syndromic. When it is unilateral, it is usually sporadic. With what syndrome is anterior lenticonus strongly associated?
- Alport syndrome

Most cases of posterior lenticonus are unilateral and sporadic. When it is bilateral, it is usually syndromic. With what syndrome is posterior lenticonus strongly associated?
- Lowe syndrome
Inability to convert galactose to glucose
Inheritance: AR
Results from a defect in one of the three enzymes involved in galactose metabolism

**Classic galactosemia:**
- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as ‘oil droplet’; progresses to total opacification

When you hear oil droplet cataract, three conditions should come to mind. One is galactosemia; what are the other two? (Note: Both are named for the shape of the lens.)

- Anterior and posterior lenticonus

Most cases of anterior lenticonus are bilateral and syndromic. When it is unilateral, it is usually sporadic. With what syndrome is anterior lenticonus strongly associated?

- Alport syndrome

Most cases of posterior lenticonus are unilateral and sporadic. When it is bilateral, it is usually syndromic. With what syndrome is posterior lenticonus strongly associated?

- Lowe syndrome

Alport and Lowe syndromes are examples of what class of condition?

- Familial oculorenal syndromes

Most are x-linked.

What nonocular symptom is their calling card?

- Hematuria
- Inability to convert galactose to glucose
- Inheritance: AR
- Results from a defect in one of the three enzymes involved in galactose metabolism

**Classic galactosemia:**
- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as 'oil droplet'; progresses to total opacification

When you hear oil droplet cataract, three conditions should come to mind. One is galactosemia; what are the other two? (Note: Both are named for the shape of the lens.)

Anterior and posterior lenticonus

Most cases of posterior lenticonus are unilateral and sporadic. When it is bilateral, it is usually syndromic. With what syndrome is posterior lenticonus strongly associated?

Lowe syndrome

Most cases of anterior lenticonus are bilateral and syndromic. When it is unilateral, it is usually sporadic. With what syndrome is anterior lenticonus strongly associated?

Alport syndrome

Alport and Lowe syndromes are examples of what class of condition?

Familial oculorenal syndromes
Inability to convert galactose to glucose
Inheritance: AR
Results from a defect in one of the three enzymes involved in galactose metabolism

**Classic galactosemia:**
- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as 'oil droplet'; progresses to total opacification

When you hear oil droplet cataract, three conditions should come to mind. One is galactosemia; what are the other two? (Note: Both are named for the shape of the lens.)
- Anterior and posterior lenticonus
  - Most cases of anterior lenticonus are bilateral and syndromic. When it is unilateral, it is usually sporadic. With what syndrome is anterior lenticonus strongly associated?
    - Alport syndrome
  - Most cases of posterior lenticonus are unilateral and sporadic. When it is bilateral, it is usually syndromic. With what syndrome is posterior lenticonus strongly associated?
    - Lowe syndrome

*Alport and Lowe syndromes are examples of what class of condition?*
- Familial oculorenal syndromes

*How are they inherited?*
- Most are X-linked
Inability to convert galactose to glucose
Inheritance: AR
Results from a defect in one of the three enzymes involved in galactose metabolism

Classic galactosemia:
- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth

Alport and Lowe syndromes are examples of what class of condition? Familial oculorenal syndromes
How are they inherited? Most are X-linked

Alport and Lowe syndromes are examples of what class of condition? Familial oculorenal syndromes
How are they inherited? Most are X-linked
Inability to convert galactose to glucose
Inheritance: AR
Results from a defect in one of the three enzymes involved in galactose metabolism

**Classic galactosemia:**
- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as 'oil droplet'; progresses to total opacification

When you hear oil droplet cataract, three conditions should come to mind. One is galactosemia; what are the other two? (Note: Both are named for the shape of the lens.)

**Anterior and posterior lenticonus**
- Most cases of anterior lenticonus are bilateral and syndromic. When it is unilateral, it is usually sporadic.
  - With what syndrome is anterior lenticonus strongly associated?
    - Alport syndrome

**Alport and Lowe syndromes** are examples of what class of condition?
- Familial oculorenal syndromes

How are they inherited?
- Most are X-linked

What nonocular symptom is their calling card?
- Hematuria
Galactosemia

- Inability to convert galactose to glucose
- Inheritance: AR
- Results from a defect in one of the three enzymes involved in galactose metabolism

**Classic galactosemia:**
- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as 'oil droplet'; progresses to total opacification

When you hear oil droplet cataract, three conditions should come to mind. One is galactosemia; what are the other two? (Note: Both are named for the shape of the lens.)

Anterior and posterior lenticonus

Most cases of anterior lenticonus are bilateral and syndromic. When it is unilateral, it is usually sporadic. With what syndrome is anterior lenticonus strongly associated?

Alport syndrome

Most cases of posterior lenticonus are unilateral and sporadic. When it is bilateral, it is usually syndromic. With what syndrome is posterior lenticonus strongly associated?

Lowe syndrome

Alport and Lowe syndromes are examples of what class of condition?

Familial oculorenal syndromes

How are they inherited?

Most are X-linked

What nonocular symptom is their calling card?

Hematuria

Alport and Lowe syndromes are examples of what class of condition?

Familial oculorenal syndromes

How are they inherited?

Most are X-linked

What nonocular symptom is their calling card?

Hematuria
Inability to convert galactose to glucose
Inheritance: AR
Results from a defect in one of the three enzymes involved in galactose metabolism

Classic galactosemia:
- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as 'oil droplet'; progresses to total opacification

Alport and Lowe syndromes are examples of what class of condition? Familial oculorenal syndromes

Of the two relationships (Alport<->anterior lenticus; Lowe<->posterior lenticus), which is stronger?

Most cases of anterior lenticonus are bilateral and syndromic. When it is unilateral, it is usually sporadic.
Most cases of posterior lenticonus are unilateral and sporadic. When it is bilateral, it is usually syndromic. With what syndrome is posterior lenticonus strongly associated?

Alport and Lowe syndromes are examples of what class of condition? Familial oculorenal syndromes

Of the two relationships (Alport<->anterior lenticus; Lowe<->posterior lenticus), which is stronger?

Most cases of anterior lenticonus are bilateral and syndromic. When it is unilateral, it is usually sporadic.
Most cases of posterior lenticonus are unilateral and sporadic. When it is bilateral, it is usually syndromic. With what syndrome is posterior lenticonus strongly associated?

Family oculorenal syndromes

Lowe syndrome

Alport syndrome
Galactosemia

- Inability to convert galactose to glucose
- Inheritance: AR
- Results from a defect in one of the three enzymes involved in galactose metabolism
- Classic galactosemia:
  - Most common and most severe form
  - Caused by defect in the uridyltransferase enzyme
  - 75% develop cataracts within days to weeks of birth
    - Start as 'oil droplet'; progresses to total opacification

When you hear oil droplet cataract, three conditions should come to mind. One is galactosemia; what are the other two? (Note: Both are named for the shape of the lens.)

- Anterior and posterior lenticonus
- Most cases of anterior lenticonus are bilateral and syndromic. When it is unilateral, it is usually sporadic.
- Most cases of anterior lenticonus are not associated with any specific syndrome.
- Most cases of posterior lenticonus are unilateral and sporadic. When it is bilateral, it is usually syndromic.
- With what syndrome is posterior lenticonus strongly associated?
  - Lowe syndrome

Alport and Lowe syndromes are examples of what class of condition?
- Familial oculorenal syndromes

Of the two relationships (Alport<->anterior lenticonus; Lowe<->posterior lenticonus), which is stronger?
- Lowe<->posterior lenticonus. Anterior lenticonus occurs in Alport syndrome in only about 25% of cases.
Galactosemia

- Inability to convert galactose to glucose
- Inheritance: AR
- Results from a defect in one of the three enzymes involved in galactose metabolism

Classic galactosemia:
- Most common and most severe form
- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as 'oil droplet'; progresses to total opacification

When you hear oil droplet cataract, three conditions should come to mind. One is galactosemia; what are the other two? (Note: Both are named for the shape of the lens.)

Anterior and posterior lenticonus

Most cases of anterior lenticonus are bilateral and syndromic. When it is unilateral, it is usually sporadic. With what syndrome is anterior lenticonus strongly associated?

Alport syndrome

Most cases of posterior lenticonus are unilateral and sporadic. When it is bilateral, it is usually syndromic. With what syndrome is posterior lenticonus strongly associated?

Lowe syndrome

Alport and Lowe syndromes are examples of what class of condition?

Familial oculorenal syndromes

Of the two relationships (Alport<->anterior lenticonus; Lowe<->posterior lenticonus), which is stronger?

Lowe<->posterior lenticonus. Anterior lenticonus occurs in Alport syndrome in only about 25% of cases.
Galactosemia

- Inability to convert galactose to glucose
- Inheritance: AR
- Results from a defect in one of the three enzymes involved in galactose metabolism
- **Classic galactosemia:**
  - Most common and most severe form
  - Caused by defect in the uridyltransferase enzyme
  - 75% develop cataracts within days to weeks of birth

Other conditions:

**Alport and Lowe syndromes are examples of what class of condition?**
- Familial oculorenal syndromes

Of the two relationships (Alport<->anterior lenticonus; Lowe<->posterior lenticonus), which is stronger?
- Lowe<->posterior lenticonus. Anterior lenticonus occurs in Alport syndrome in only about 25% of cases. OTOH, posterior lenticonus is considered a defining feature of Lowe syndrome.
Galactosemia

- Inability to convert galactose to glucose
- Inheritance: AR
- Results from a defect in one of the three enzymes involved in galactose metabolism
- Classic galactosemia:
  - Most common and most severe form
  - Caused by defect in the uridyltransferase enzyme
  - 75% develop cataracts within days to weeks of birth
    - Starts as ‘oil droplet’; progresses to total opacification

For more on lenticulus and Alport/Lowe syndromes, see slide-set L4

- Caused by defect in the uridyltransferase enzyme
- 75% develop cataracts within days to weeks of birth
  - Alport and Lowe syndromes are examples of what class of condition?
    - Familial oculorenal syndromes

- of the two relationships (Alport<->anterior lenticulus; Lowe<->posterior lenticulus), which is stronger?
  - Lowe<->posterior lenticulus. Anterior lenticulus occurs in Alport syndrome in only about 25% of cases. OTOH, posterior lenticulus is considered a defining feature of Lowe syndrome.

- Most cases of anterior lenticonus are bilateral and syndromic. When it is unilateral, it is usually sporadic. With what syndrome is anterior lenticulus strongly associated?
  - Alport syndrome

- Most cases of posterior lenticonus are unilateral and sporadic. When it is bilateral, it is usually syndromic. With what syndrome is posterior lenticulus strongly associated?
  - Lowe syndrome
Galactosemia

- Inability to convert galactose to glucose
- Inheritance: AR
- Results from a defect in one of the three enzymes involved in galactose metabolism

**Classic galactosemia:**

*What are the systemic manifestations of classic galactosemia?*
- ?
- ?
- ?

75% develop cataracts within days to weeks of birth
- Starts as ‘oil droplet’; progresses to total opacification

What is the prognosis if classic galactosemia goes untreated?
- It is uniformly fatal
- Inability to convert galactose to glucose
- Inheritance: AR
- Results from a defect in one of the three enzymes involved in galactose metabolism

**Classic galactosemia:**

What are the systemic manifestations of classic galactosemia?
- Failure to thrive
- Hepatomegaly with jaundice
- Impaired cognitive development
Galactosemia

- Inability to convert galactose to glucose
- Inheritance: AR
- Results from a defect in one of the three enzymes involved in galactose metabolism

**Classic galactosemia:**

*What are the systemic manifestations of classic galactosemia?*
- Failure to thrive
- Hepatomegaly with jaundice
- Impaired cognitive development

*How soon do these findings begin to manifest?*

- 75% develop cataracts within days to weeks of birth
  - Starts as 'oil droplet'; progresses to total opacification
- It is uniformly fatal
Galactosemia

- Inability to convert galactose to glucose
- Inheritance: AR
- Results from a defect in one of the three enzymes involved in galactose metabolism

**Classic galactosemia:**

*What are the systemic manifestations of classic galactosemia?*

- Failure to thrive
- Hepatomegaly with jaundice
- Impaired cognitive development

*How soon do these findings begin to manifest?*

Within a few weeks after birth

Galactosemia is a genetic disorder that results from a defect in one of the three enzymes involved in galactose metabolism. The classic form of galactosemia is the most common and severe, caused by a defect in the uridyltransferase enzyme. It affects approximately 1 in 40,000 newborns and is characterized by failure to thrive, hepatomegaly with jaundice, and impaired cognitive development. If left untreated, classic galactosemia is uniformly fatal.
Inability to convert galactose to glucose
Inheritance: AR
Results from a defect in one of the three enzymes involved in galactose metabolism

**Classic galactosemia:**

*What are the systemic manifestations of classic galactosemia?*
-- Failure to thrive
-- Hepatomegaly with jaundice
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**Classic galactosemia:**
- Most common and most severe form
- Caused by defect in the *uridyltransferase* enzyme
- 75% develop cataracts within days to weeks of birth
  - Starts as ‘oil droplet’; progresses to total opacification
- To diagnosis classic galactosemia: Check UA for presence of reducing substances **after** milk ingestion
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In the two \textit{non-classic} forms…

- Disease \textit{more/less} severe than in classic disease.
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  - Cataracts present **earlier vs later** in life
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