

A 3-week-old boy is brought to the physician for a 1-week history of jaundice. The parents describe the infant's stool as always being "kind of pale-colored." The patient is exclusively breastfed and has been feeding well. The pregnancy and delivery were uncomplicated. Maternal blood type is O positive, and routine prenatal studies were normal. His temperature is 37.2 C (99 F), pulse is 140/min, respirations are 50/min, and capillary refill is <2 sec. Examination shows jaundice and hepatomegaly. Laboratory results are as follows:

Hemoglobin	15 g/dL
Bilirubin, Total	10.3 mg/dL
Bilirubin, Direct	8.1 mg/dL
Blood type	B positive
Coombs test	Negative

Which of the following is the most likely diagnosis?

- ☐ A. Biliary atresia
- ☐ B. Breast milk jaundice
- ☐ C. Crigler-Najjar syndrome
- ☐ D. Erythroblastosis fetalis
- ☐ E. Gilbert's syndrome
- ☐ F. Physiologic jaundice

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- ✓

☒

A. Biliary atresia [64%]
- ☐

B. Breast milk jaundice [16%]
- ☐

C. Crigler-Najjar syndrome [9%]
- ☐

D. Erythroblastosis fetalis [1%]
- ☐

E. Gilbert's syndrome [6%]
- ☐

F. Physiologic jaundice [3%]

Proceed to Next Item

Explanation:

User Id:

Biliary atresia	
Clinical	<div>Initially well-appearing, followed by development of the following over 1-8 weeks:</div> <ul style="list-style-type: none"><li>Jaundice</li><li>Acholic (pale) stools or dark urine</li></ul>



Proceed to Next Item

Explanation:

User Id: [REDACTED]

Biliary atresia	
Clinical features	<p>Initially well-appearing, followed by development of the following over 1-8 weeks:</p> <ul style="list-style-type: none"> <li>• Jaundice</li> <li>• Acholic (pale) stools or dark urine</li> <li>• Hepatomegaly</li> <li>• Conjugated hyperbilirubinemia</li> <li>• Mild elevation in transaminases</li> </ul>
Diagnosis	<ul style="list-style-type: none"> <li>• Ultrasound: absent or abnormal gallbladder</li> <li>• Hepatobiliary scintigraphy: failure of tracer excretion</li> <li>• Liver biopsy: expanded portal tracts with bile duct obstruction &amp; proliferation; portal tract edema, fibrosis &amp; inflammation</li> <li>• Intraoperative cholangiogram (gold standard): biliary obstruction</li> </ul>
Treatment	<ul style="list-style-type: none"> <li>• Hepatoportoenterostomy (Kasai procedure)</li> <li>• Liver transplant</li> </ul>

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Conjugated hyperbilirubinemia is defined as  $>2$  mg/dL of direct bilirubin or a direct bilirubin fraction that is  $>20\%$  of the total bilirubin level. This neonate's **jaundice**, **light-colored stools**, **hepatomegaly**, and direct (conjugated) hyperbilirubinemia require prompt evaluation for **biliary atresia**. Biliary atresia is caused by progressive obliteration of the extrahepatic biliary ducts connecting the liver to the small bowel. It is the most common indication for pediatric liver transplantation. Newborns are **initially well** but develop **conjugated hyperbilirubinemia** in the first 2 months of life, which manifests as jaundice, acholic stools (due to the absence of biliary pigment), and dark urine (due to renal excretion of bilirubin that cannot reach the small bowel). Without treatment, the liver will become inflamed (eg, hepatomegaly, hepatitis) and eventually fibrose.



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The first step in evaluation is abdominal **ultrasound**, which may show an absent or abnormal gallbladder. Failure of the liver to excrete tracer into the small bowel on **scintigraphy** is highly suggestive of biliary atresia, and the diagnostic gold standard is a **cholangiogram** obtained in the operating room. Once biliary atresia is confirmed, a **Kasai procedure** (hepatportoenterostomy) should be performed. Virtually all patients will require **liver transplantation**, but the Kasai procedure allows time for growth and reduces the morbidity and mortality of hepatic transplant.

**(Choice B)** Breast milk jaundice generally appears in the second week of life. However, the hyperbilirubinemia that occurs is indirect (unconjugated) and may rise as high as 10-30 mg/dL. It is generally a benign condition, but in rare cases phototherapy may be indicated.

**(Choices C and E)** Crigler-Najjar and Gilbert's syndrome are inherited deficiencies of UDP-glucuronyl transferase that result in unconjugated hyperbilirubinemia. In Gilbert's syndrome, the deficiency is mild and patients are asymptomatic or have mild jaundice, particularly during times of stress. In Crigler-Najjar syndrome, the enzyme is absent; patients present early in life and require liver transplantation.

**(Choice D)** Alloimmune hemolytic disease (erythroblastosis fetalis) is characterized by unconjugated hyperbilirubinemia and Coombs-positive hemolytic anemia. It is caused by a mismatch between infant and maternal blood types (eg, Rh disease, ABO incompatibility, or minor blood group antigens). This patient's conjugated hyperbilirubinemia, late onset of jaundice, lack of anemia, and a negative Coombs test are inconsistent with alloimmune hemolysis.

**(Choice F)** Physiologic jaundice is unconjugated hyperbilirubinemia that appears after the first 24 hours of life and resolves within the first week. Conjugated hyperbilirubinemia is always pathologic.



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#### **Educational objective:**

Newborns with conjugated hyperbilirubinemia and hepatomegaly require immediate evaluation for biliary atresia. The first step in evaluation is abdominal ultrasound, which may show an absent or abnormal gallbladder. Early treatment with a Kasai procedure dramatically improves outcomes.

#### **References:**

1. [Conjugated hyperbilirubinemia in children.](#)
2. [Early detection of biliary atresia: past, present & future.](#)



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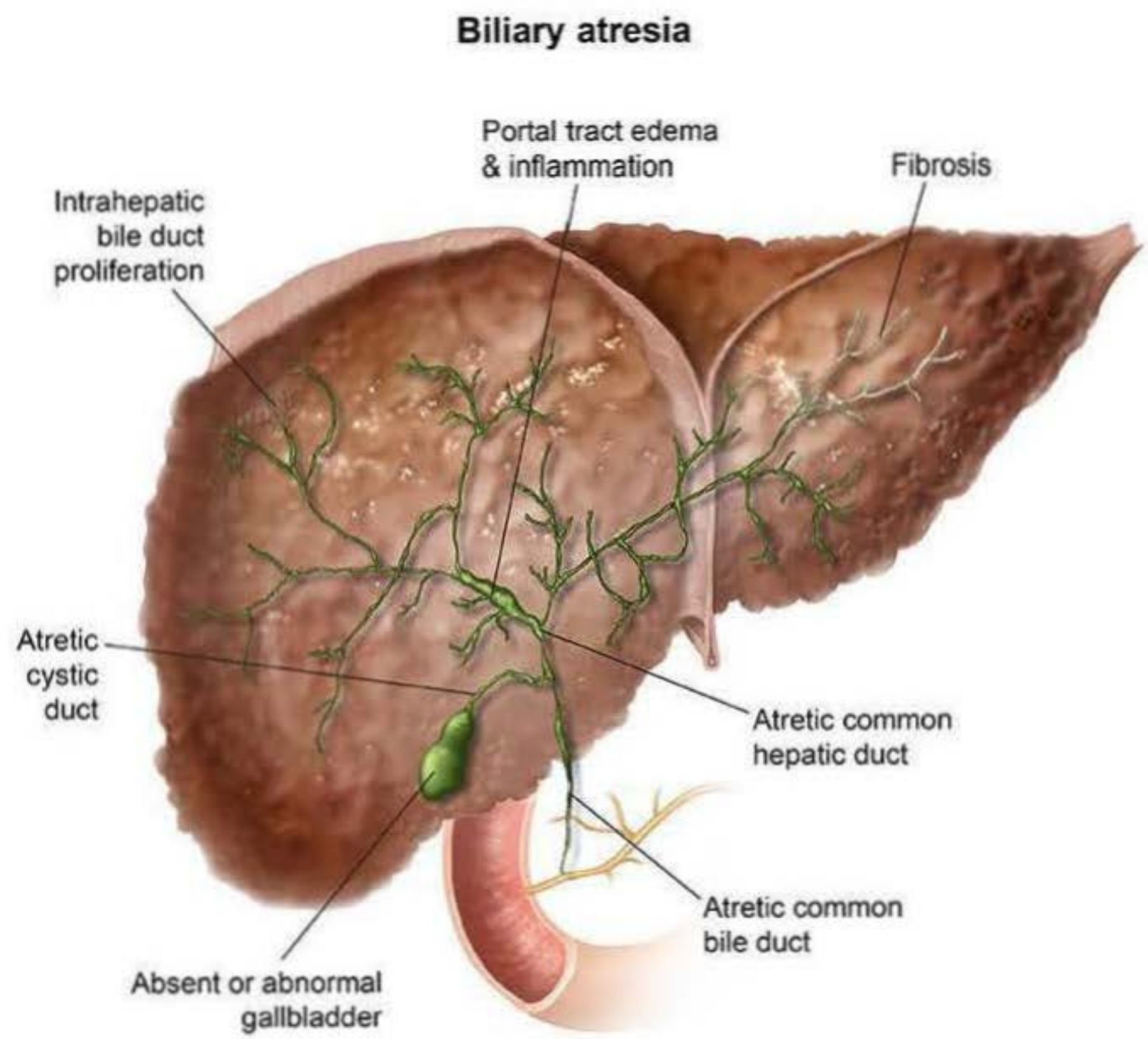
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Media Exhibit

resia



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