Is it time to reassess your patients with progressive cholestasis?

Certain patient presentations of progressive cholestatic liver disease (ChLD) may signal the need for further assessment

Actor portrayal.

Idiopathic Cholestasis

CONSIDER REASSESSING IF signs of cholestasis manifest without apparent cause. Presentation is highly variable, but symptoms can include[1-3]:

- Jaundice
- Pruritus
- Abnormal stools
- Abnormal liver parameters for bilirubin or transaminases*

In addition to clinical symptoms, **MEDICAL HISTORY MAY ALSO REFLECT:**

- **Suffers from long-term symptoms and has not found proper relief**
- **Has sought care without receiving a definitive diagnosis**

Cholestasis With Pruritus or Unusual Presentation

CONSIDER REASSESSING IF your patient is receiving care for another liver disease but has unusual symptoms, including:

- **Small duct PSC[1]**
- **AMA negative PBC[4,5]**
- **NAFLD with pruritus[6]**
- **Lean NAFLD without metabolic syndrome[6]**
- **Lean NASH with pruritus and without metabolic syndrome[6]**

Secondary Cholestasis Triggered by Liver Issue

CONSIDER REASSESSING IF symptoms of cholestatic pruritus arise in patients who have recently experienced liver issues, including:

- **All women with ICP[1]**
- **Drug-induced cholestasis[1]**
- **Hormonal-induced cholestasis triggered by birth control, menopause, etc[1,7]**

History of Complicated Gallstones

CONSIDER REASSESSING IF your patient has a complicated history of gallstones, including:

- **Intrahepatic gallstones[1]**
- **Very strong family history of gallstones and incident at a young age[8,9]**
- **LPAC leading to stones in the gallbladder or liver[10]**

*Including GGT, AST, ALT, or ALP.

ALP=alkaline phosphatase; ALT=alanine aminotransferase; AMA=antimitochondrial antibody; AST=aspartate aminotransferase; GGT=gamma-glutamyl transferase; ICP=intrahepatic cholestasis of pregnancy; LPAC=low phospholipid-associated cholelithiasis; NAFLD=nonalcoholic fatty liver disease; NASH=nonalcoholic steatohepatitis; PBC=primary biliary cholangitis; PSC=primary sclerosing cholangitis.

Could adult progressive familial intrahepatic cholestasis (PFIC) be hiding in your practice?

PFIC is a rare and life-threatening type of cholestatic liver disease with several subtypes. While previously believed to present only in early infancy, PFIC can manifest later in life after a trigger—or patients can experience a long and complicated path to diagnosis.[11]

Diagnosing PFIC can be challenging in adolescent and older patients

REASONS include[11,12]:

- Symptoms are difficult to identify and often overlooked
- Presentation is highly variable and often not considered classical

Identifying PFIC is the first step for providing appropriate treatment

MULTIPLE ASSESSMENTS can be used to help support a clinical diagnosis of PFIC:

- **Common symptoms**, especially pruritus and jaundice, along with elevated serum bile acids and the presence of gastrointestinal symptoms, like diarrhea, are key indicators of PFIC[11]
- **Lab results** with abnormal transaminase levels and high levels of bilirubin, as well as abnormally high levels of serum bile acids, could be predictors of PFIC[13]
- **Imaging** can be used to help rule out other conditions:
  - Performing a cholangiography can rule out extrahepatic conditions[14,15]
  - An ultrasound can identify liver damage progression, extrahepatic causes of cholestasis, and help distinguish PFIC from other forms of cholestasis[14,16]

Genetic testing can reinforce a suspected diagnosis of PFIC, but in some patients, testing can be inconclusive or indeterminate. Support from a geneticist may be required.[13]

There may be more adolescent and adult patients with PFIC in your practice than you think. Confirming a diagnosis is vital to addressing their ChLD.

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