Caroli Disease

- Caroli **disease**:
  - Congenital multifocal saccular dilation of large intrahepatic bile ducts in absence of other hepatic abnormalities
- Caroli **syndrome**:
  - More common variant characterized by saccular dilatation of intrahepatic ducts, hepatic fibrosis, and portal hypertension
- Pathology
  - Pathogenesis not completely understood but likely due to arrest or derangement in remodeling of ductal plate
    - » Considered to be variant of fibropolycystic liver disease
    - » Typically autosomal recessive inheritance
  - Frequently associated with polycystic hepatorenal syndrome (usually autosomal recessive), hepatic fibrosis, and medullary sponge kidney
  - Presentation in childhood or young adulthood often due to cholangitis, cholestasis, or progressive hepatic fibrosis

Demographics

- **Age**
  - Condition is present at birth, although patients can be asymptomatic for years
    - » 80% become symptomatic before age of 30
  - Caroli disease: Symptoms usually by 2nd/3rd decades
  - Caroli syndrome: Symptoms can present during infancy or childhood
    - » Concurrent renal anomalies present early in life
- **Gender**
  - M:F = 1:1
- **Epidemiology**
  - Rare disease

Natural History & Prognosis

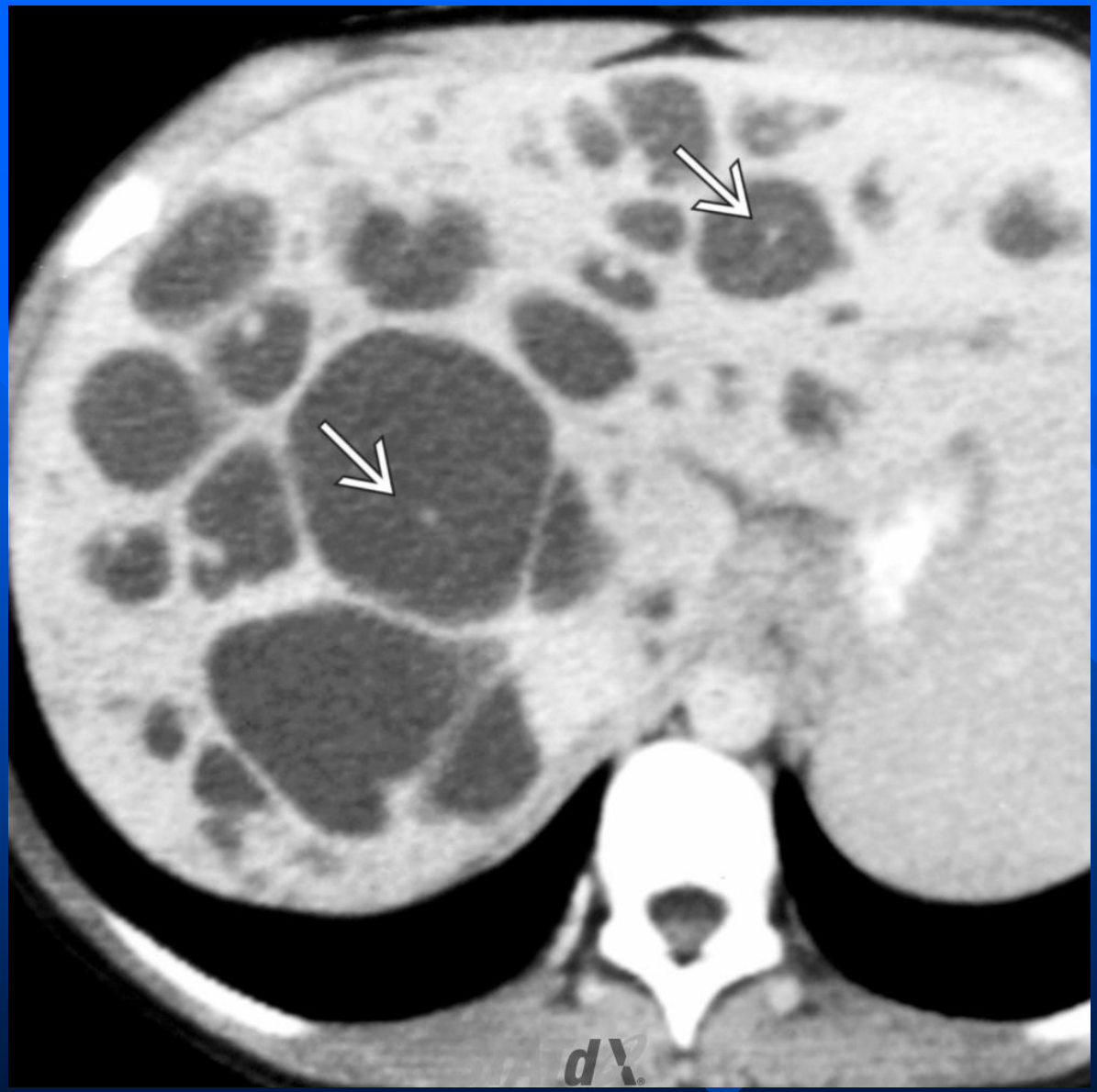
- Complications
  - Recurrent bouts of cholangitis ± hepatic abscesses
    - » Biliary dilatation → stagnation of bile → intraductal sludge/stone formation → cholangitis
  - Stone formation (95%) (usually calcium bilirubinate)
  - Secondary biliary cirrhosis ± portal hypertension
  - Cholangiocarcinoma (up to 7% risk)
- Prognosis
  - Depends on severity of disease and coexisting renal disease
  - Long-term prognosis for Caroli disease is usually poor

General Features

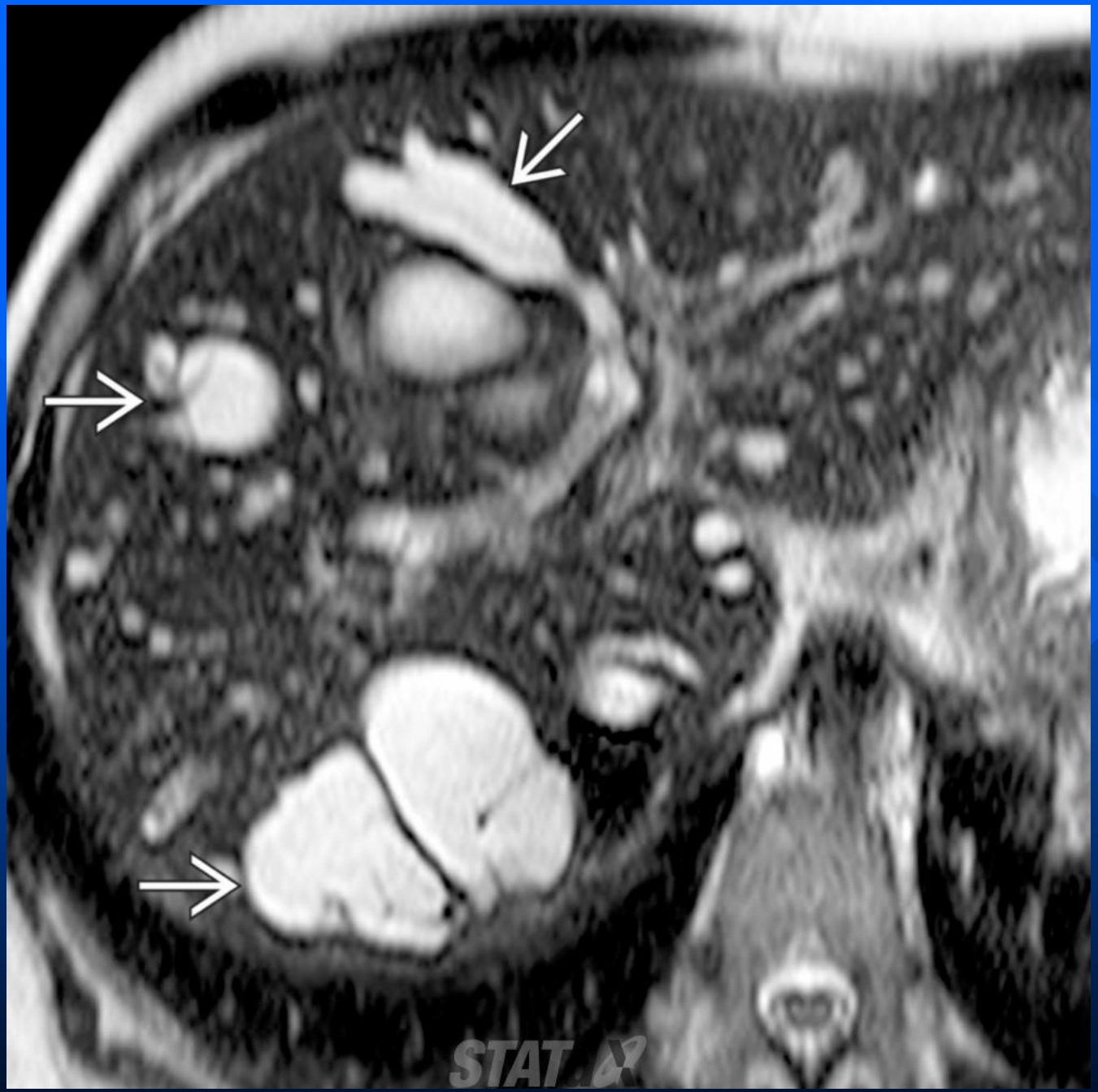
- **Best diagnostic clue**
  - Multiple intrahepatic cysts of varying size that communicate with biliary tree
  - Central dot sign: Enhancing tiny dots (portal radicles) within dilated intrahepatic bile ducts on CECT
- **Location**
  - **Saccular** dilatation of intrahepatic ducts can be diffuse, lobar, or segmental
    - » Caroli syndrome usually involves entire liver diffusely
    - » Caroli disease occasionally can be lobar (usually left lobe) but usually predominates in 1 segment
      - Number of cysts usually < 10 in cases with lobar distribution
- **Size**
  - Cysts can be of variable size (mm to cm)
- **Morphology**
  - Segmental saccular dilatation of large intrahepatic bile ducts separated by normal or dilated bile ducts
    - » Individual cysts are contiguous with biliary tree
  - Size of common bile duct (CBD) is classically thought to be normal
    - » Extrahepatic duct may be dilated in up to 50%, possibly due to prior cholangitis, passage of stones, or concurrent choledochal cyst
  - Caroli syndrome (more common variant) frequently associated with autosomal recessive polycystic kidney disease (ARPKD), medullary sponge kidney, medullary cystic disease

Cont

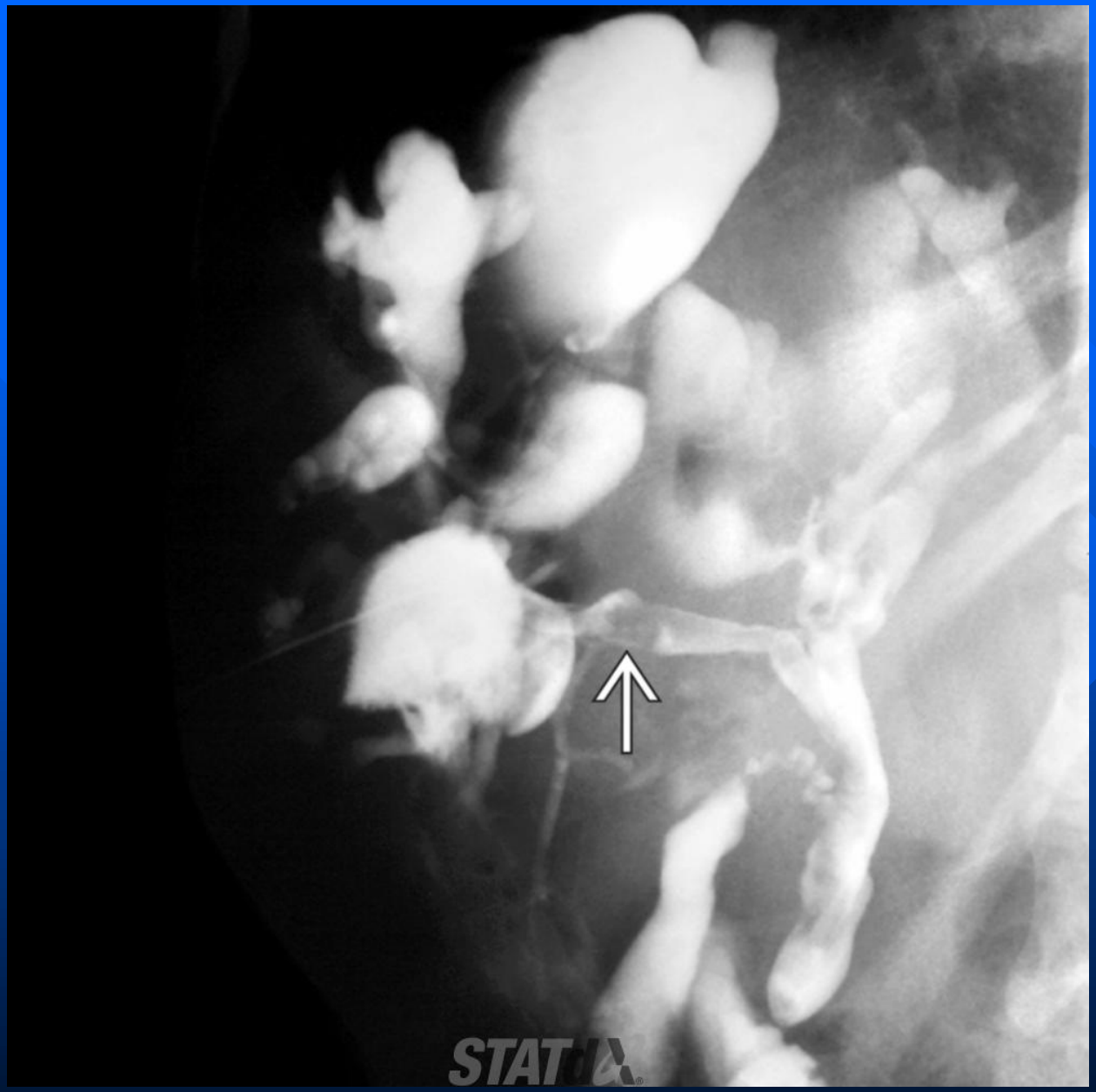
- Considered to be a variant of fibropolycystic liver disease
  - Other variants include congenital hepatic fibrosis, autosomal dominant (ADPKD) and recessive (ARPKD) polycystic liver and kidney disease, biliary hamartomas, and choledochal cyst
- Todani classification of choledochal cysts
  - Caroli disease is classified as type V choledochal cyst: Cystic dilation of intrahepatic bile ducts



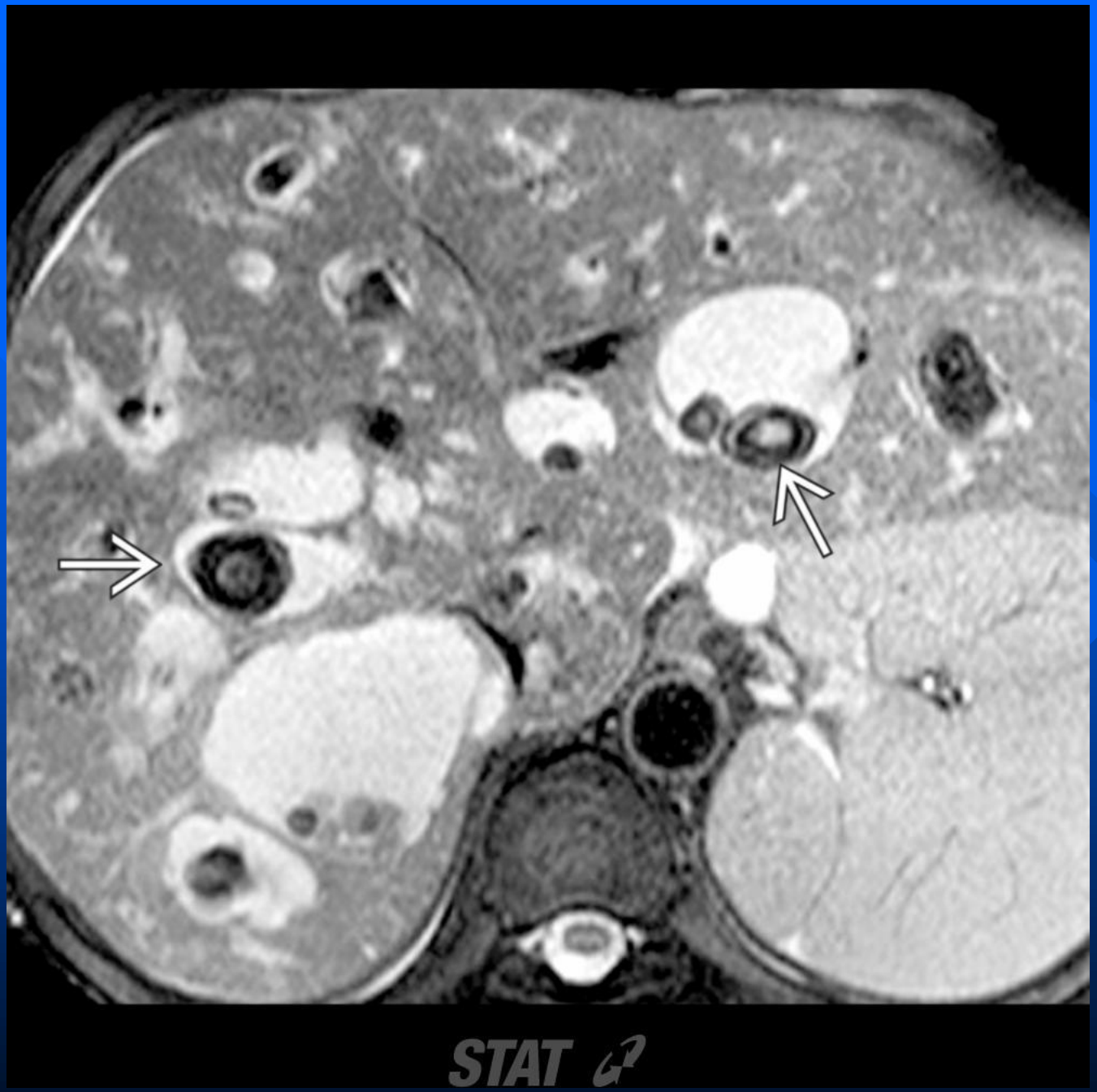
Axial CECT shows massive dilatation of the intrahepatic bile ducts. Note the "central" or "eccentric" dot within many of the cystic structures, representing portal radicles (white solid arrow), a classic imaging finding in Caroli disease



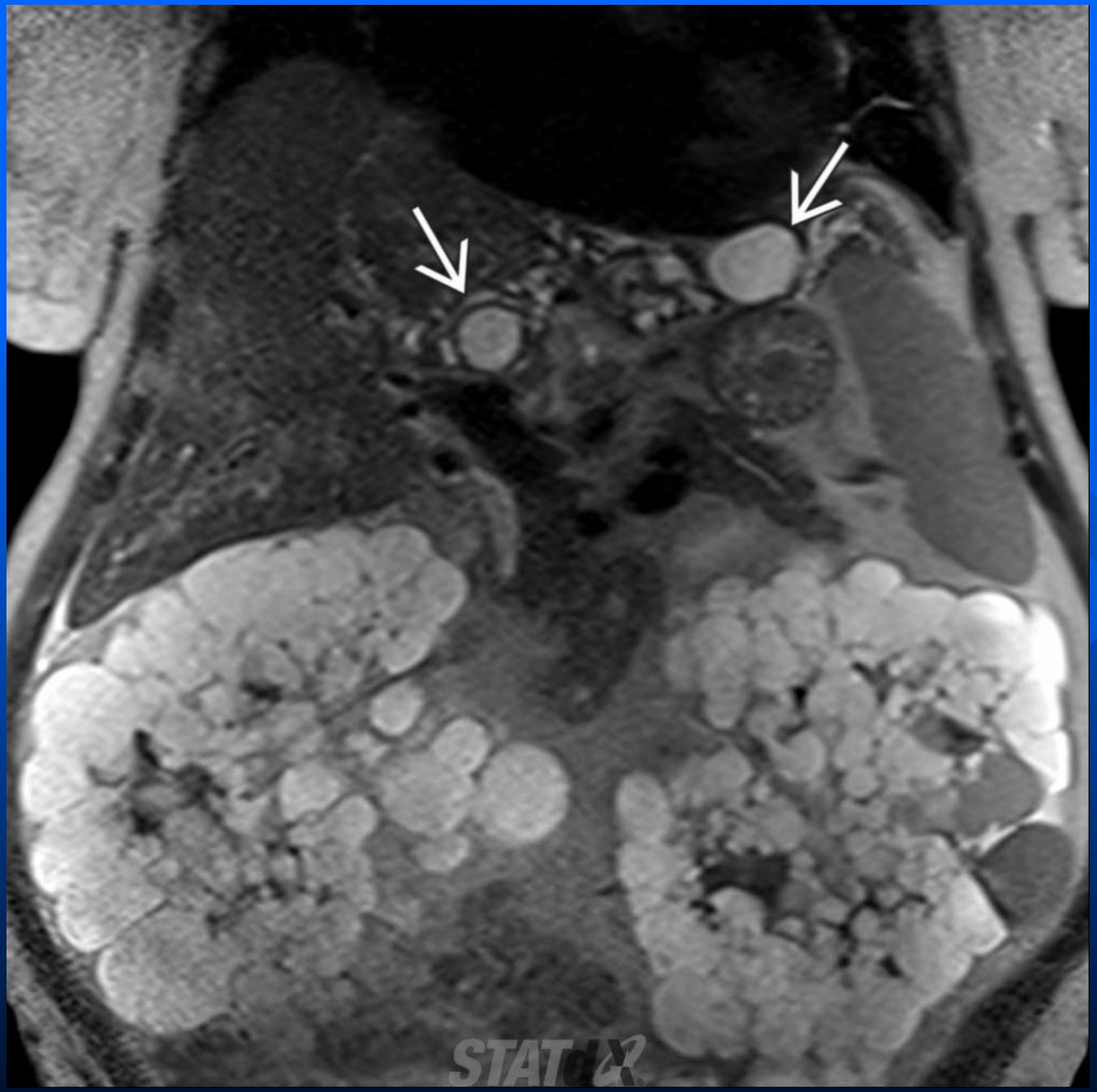
Axial T2WI MR shows gross, irregular dilation of the intrahepatic bile ducts (white solid arrow).



Frontal ERCP shows saccular dilatation of the large intrahepatic ducts as well as multiple ductal filling defects, representing biliary calculi (white solid arrow), a characteristic constellation of findings in Caroli disease.

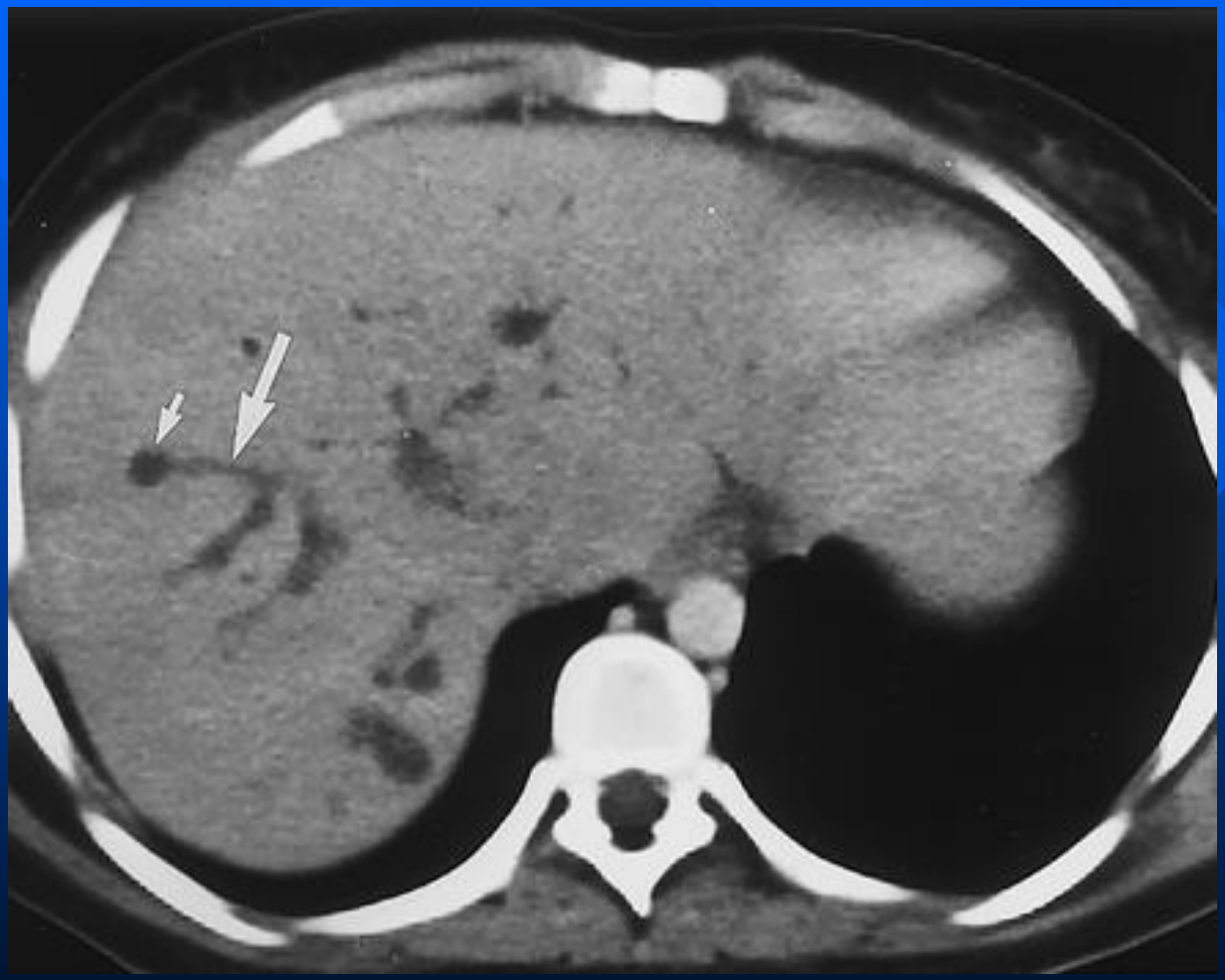


Axial T2WI MR shows saccular dilatation of the intrahepatic bile ducts, many of which contain large hypointense calculi (white solid arrow).

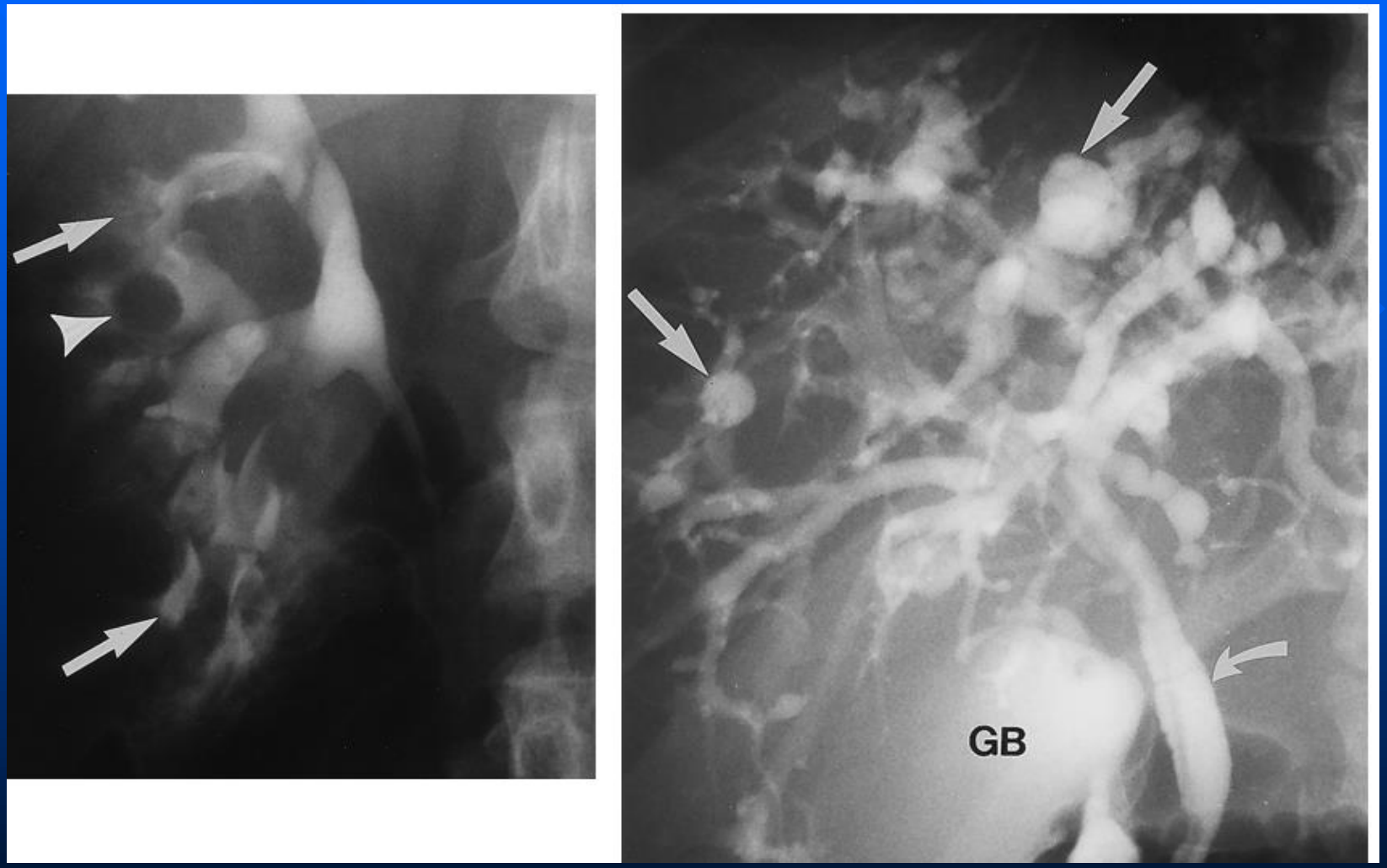


Coronal T2 HASTE MR image demonstrates several cysts (white solid arrow) clustered in the left hepatic lobe in a patient with Caroli syndrome. Note the cystic replacement of the kidneys due to concurrent autosomal dominant polycystic kidney syndrome.

Caroli's disease

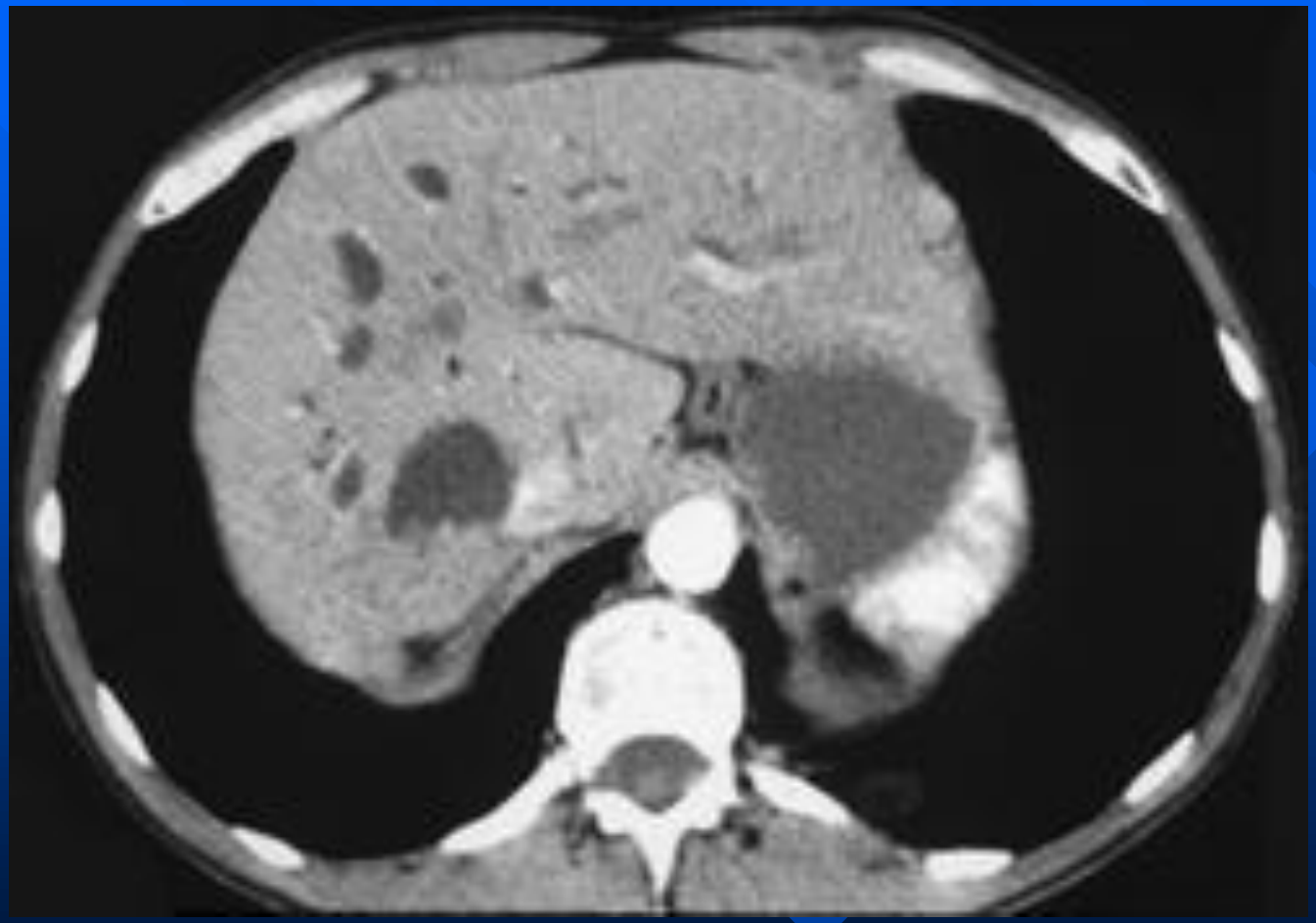


Caroli's disease



dilated renal tubules (arrows) characteristic of renal tubular ectasia

Caroli's Disease



Shunt in long standing cirrohisis in patient with Caroli's