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Autosomal Dominant Polycystic Kidney Disease

Carolynn DeBenedectis

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BIDMC

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Case

- 47 year old female with a history of hypertension, hepatomegaly, early satiety, and some abdominal discomfort.
- Family history is significant for a father who died of renal failure.
- CT abdominal/pelvis was performed for a chief complaint of abdominal discomfort

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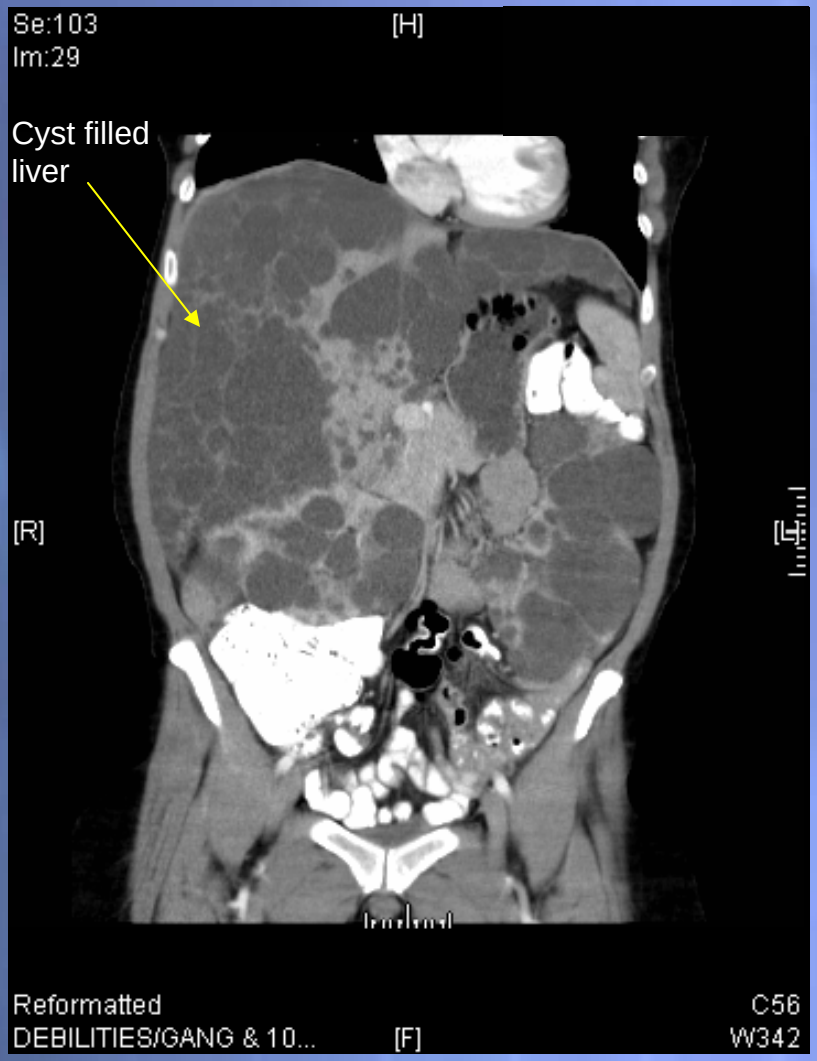
CT Abdominal/Pelvis with IV contrast



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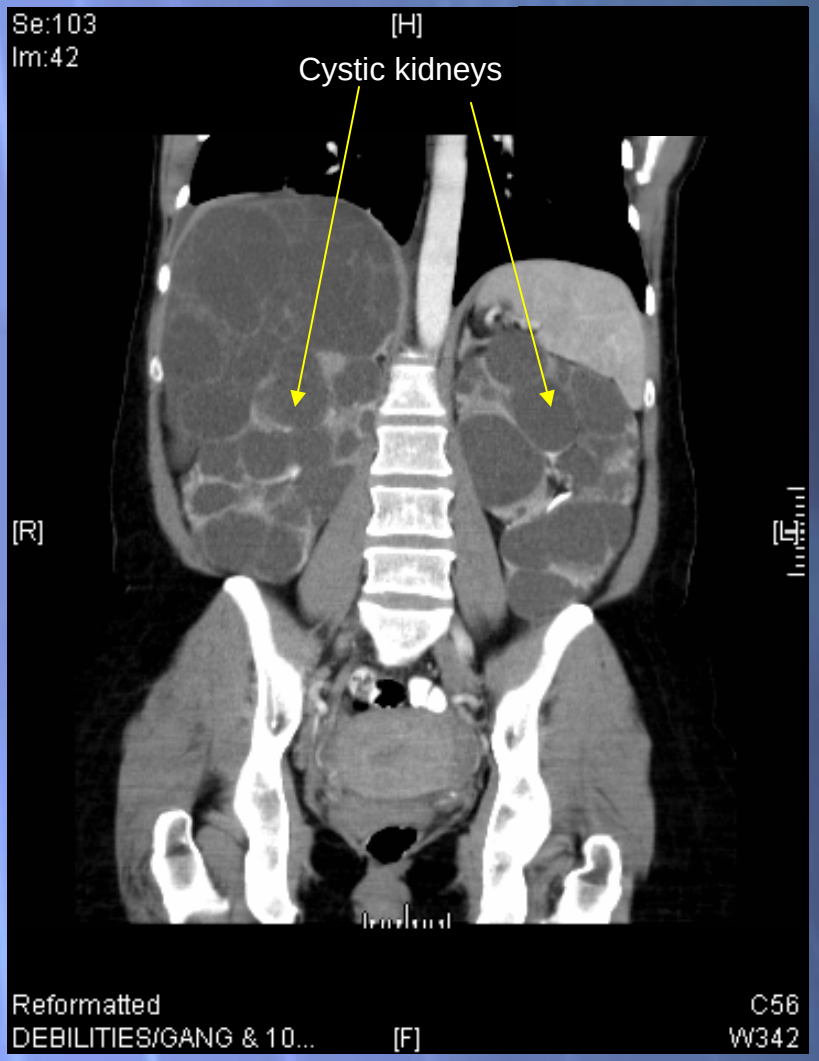

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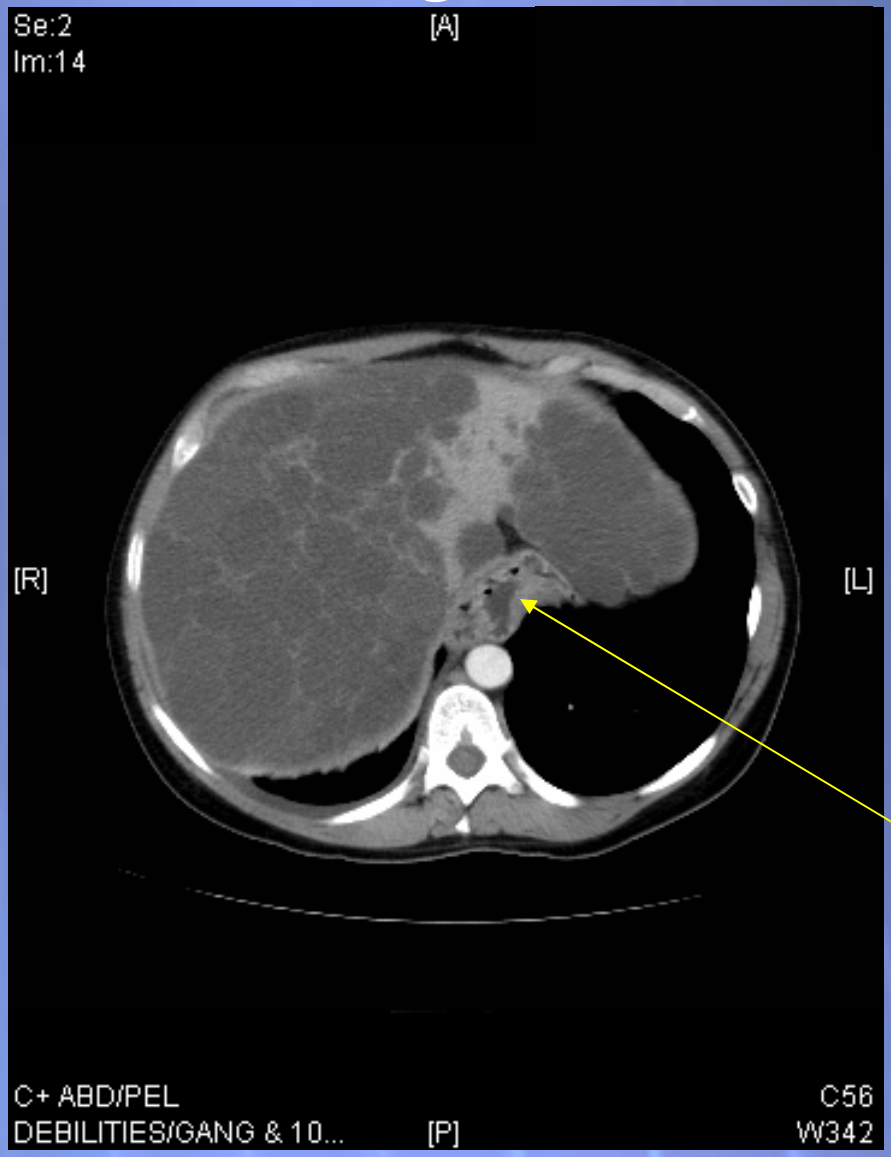
Small sliding hiatal hernia



Stomach protruding into the left hemithorax

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Hiatal Hernias

- Definition – proximal portion of the stomach slips thru the esophageal hiatus and into the thorax
- Sliding type – the gastroesophageal (GE) junction slides into the mediastinum above the diaphragm
  - Most common type
- Paraesophageal type – the GE junction is in the normal location but the gastric fundus "rolls" into the mediastinum
  - Can become infarcted, which is a surgical emergency

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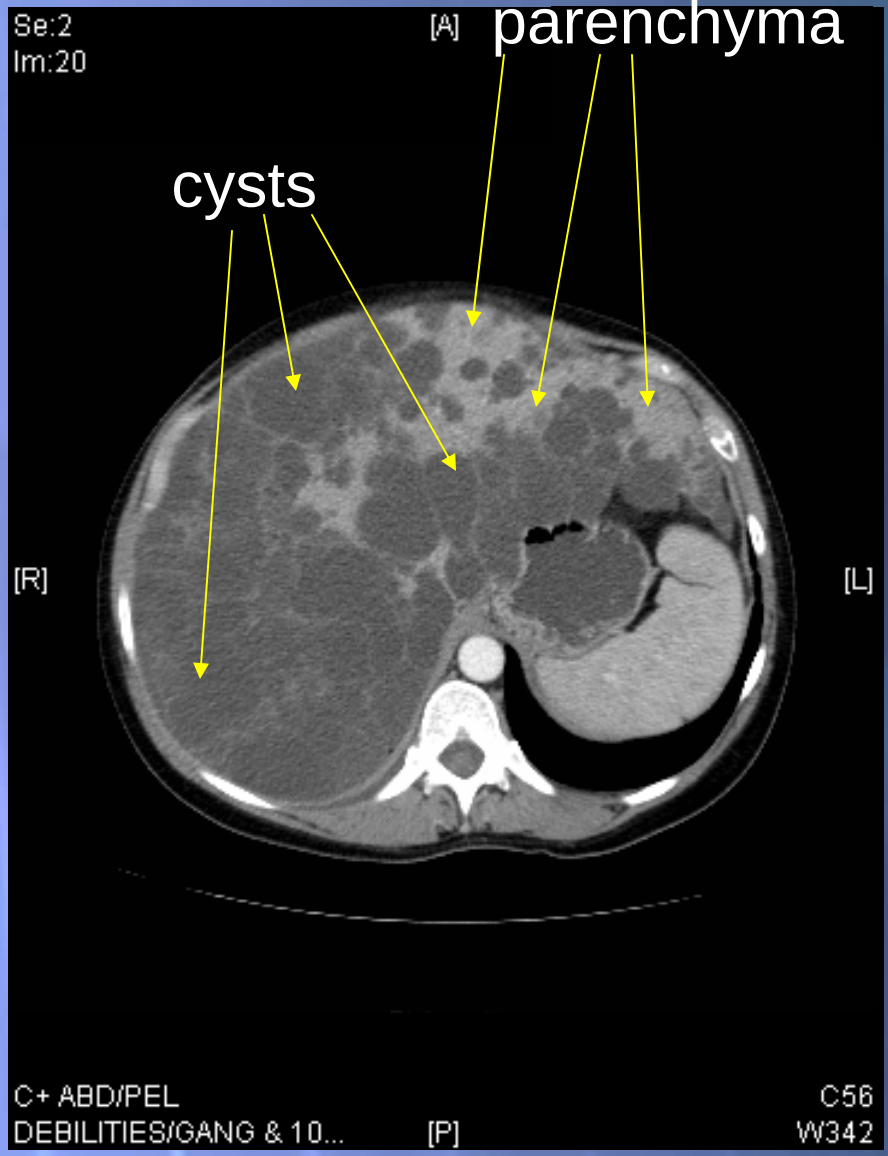
Specific CT findings in the liver

- Multiple fluid-filled cysts are seen throughout and largely replacing the right lobe of the liver

- There is diffuse hypertrophy of the liver parenchyma, with lateral displacement of the stomach wall



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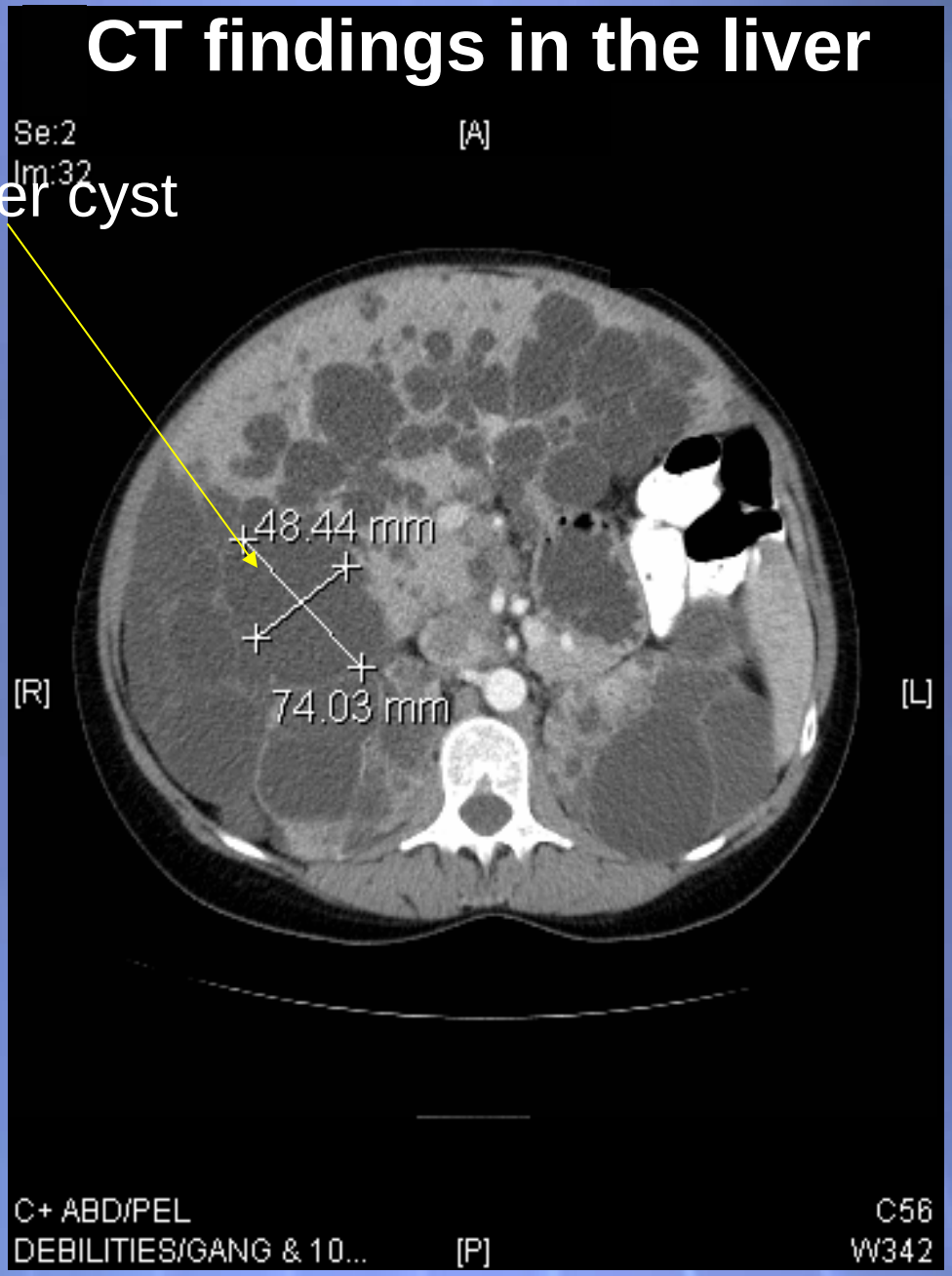
CT findings in the liver

The largest liver cyst



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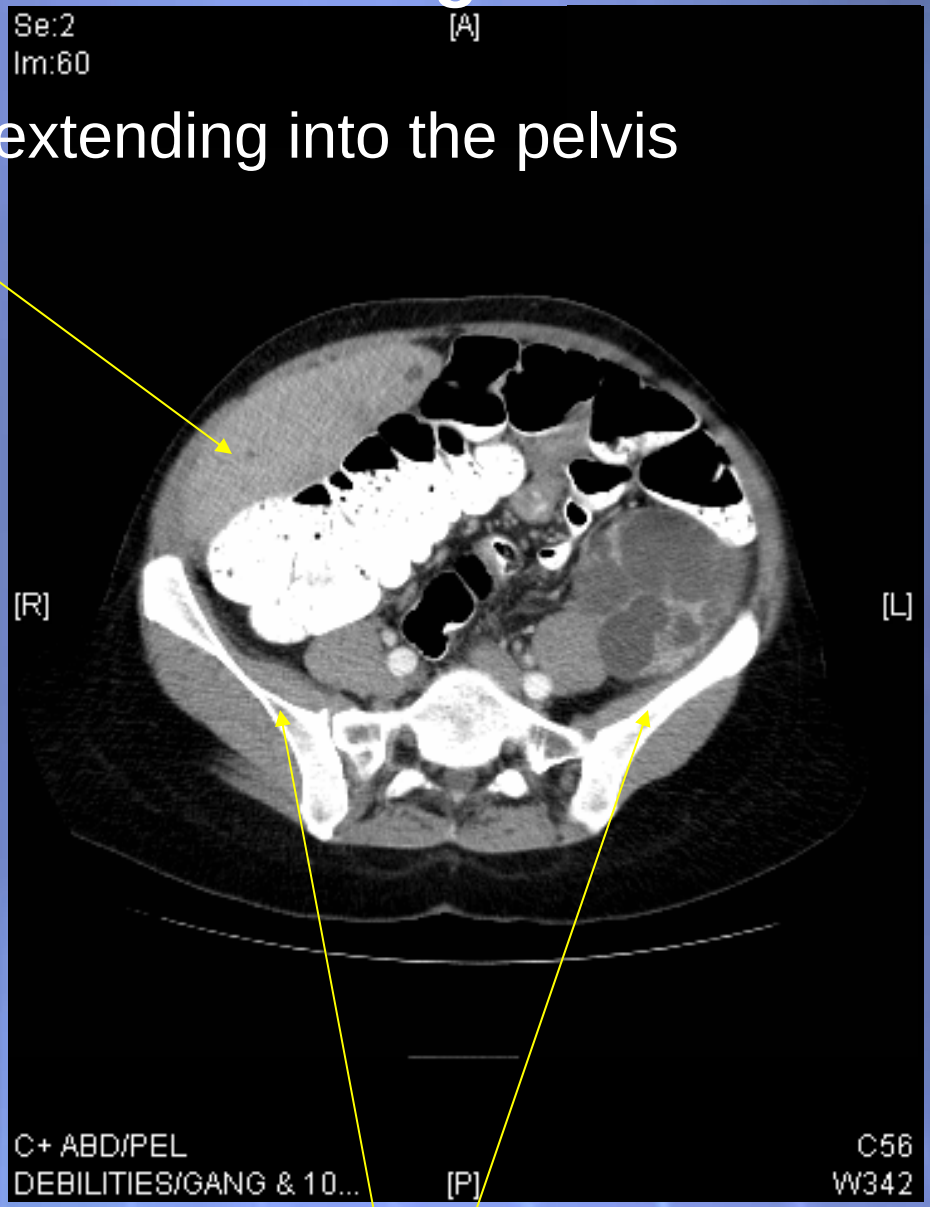
CT findings in the liver

Liver extending into the pelvis



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Iliac crests

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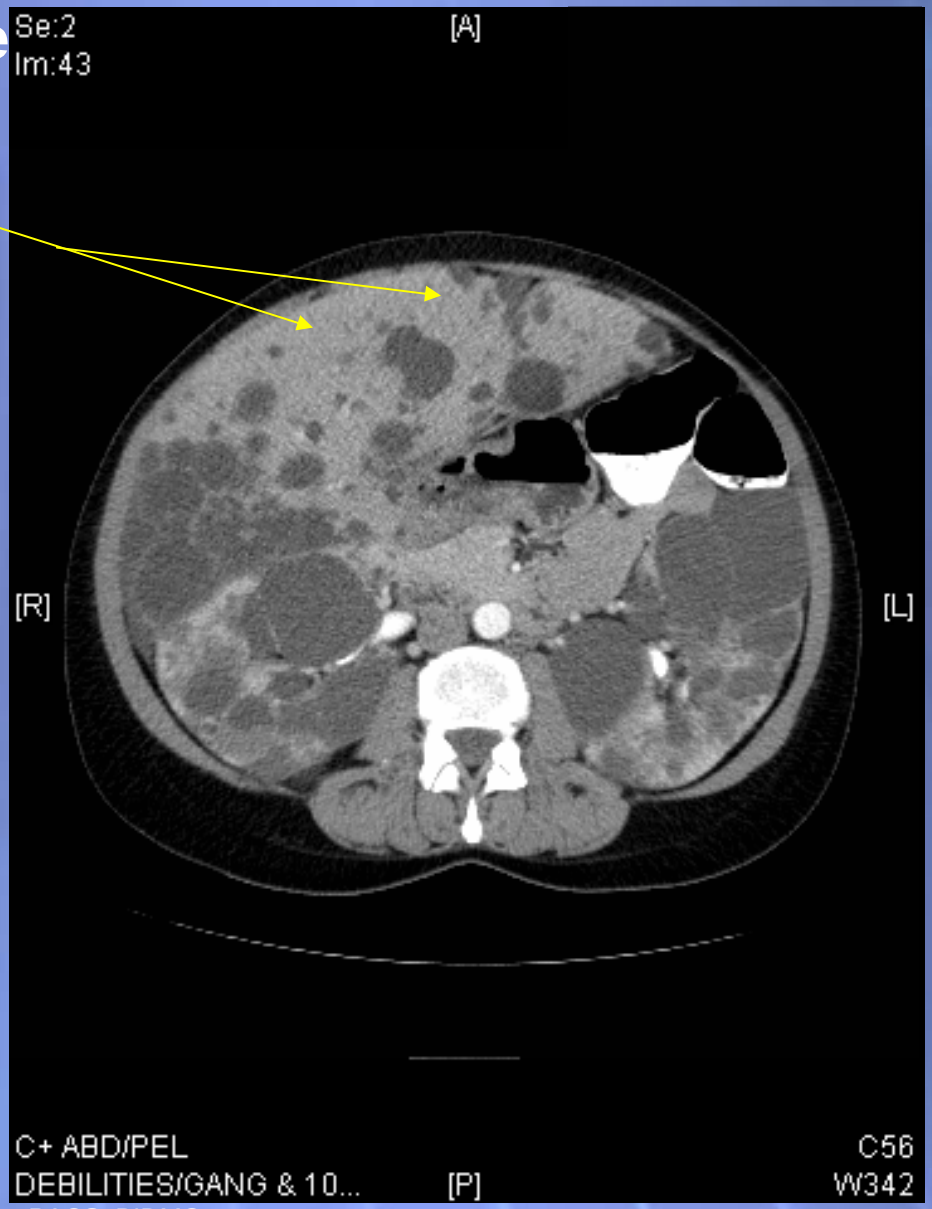
CT findings in the liver

Note the left lobe of the liver is partially clear of cysts (note the amount of liver parenchyma)



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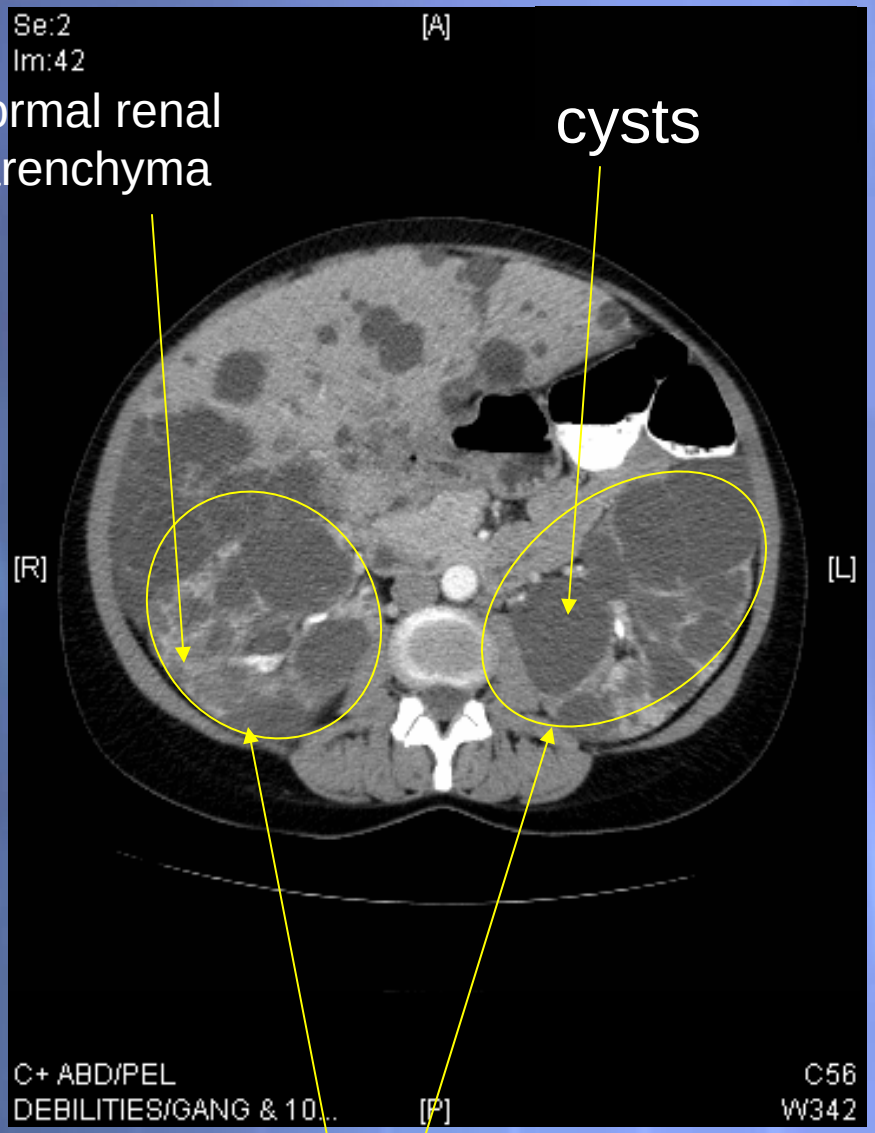
Specific CT findings in the kidneys



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- Multiple large cysts are seen replacing bilateral kidneys.
- Remaining renal parenchyma enhances normally
- There is symmetric excretion of contrast
- No enhancement of any of the visualized cysts
  - Cyst enhancement would indicate cyst rupture, hemorrhage, or infection

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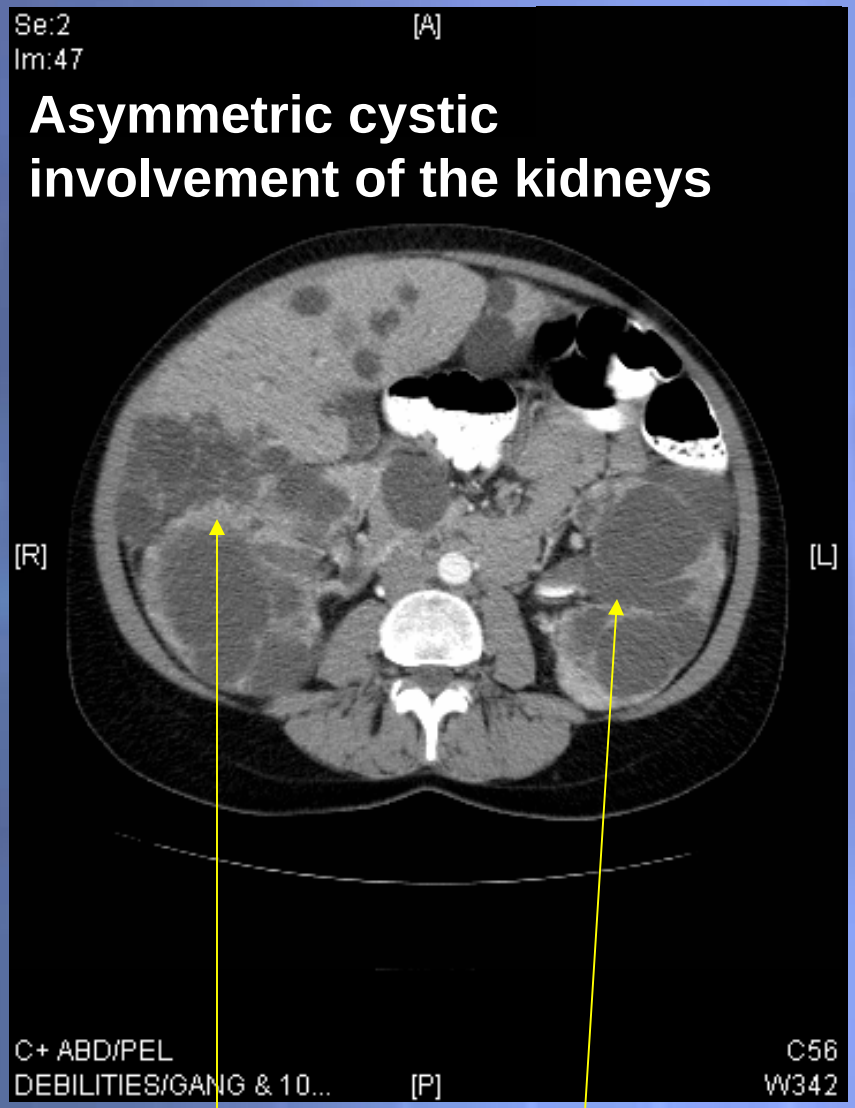

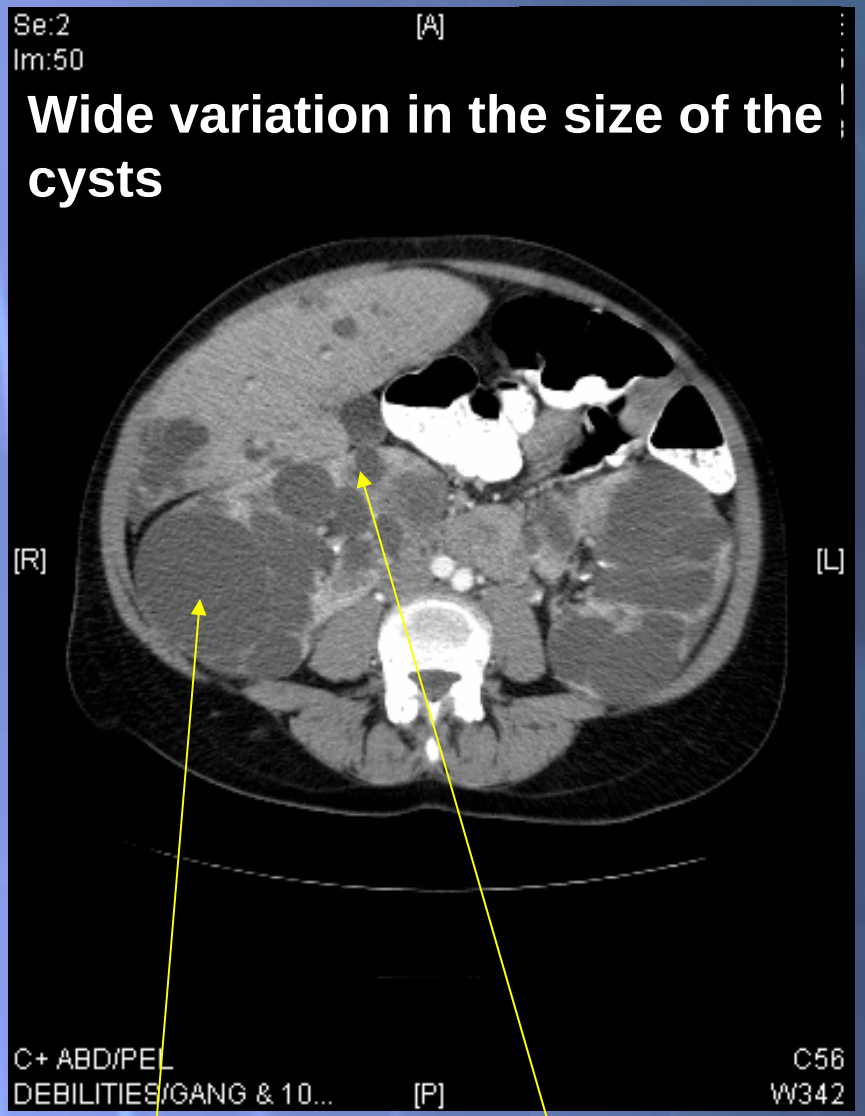

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Kidney Cysts

- Fluid-filled sacs arising from a dilatation in any part of the nephron or collecting duct.
  - Renal cysts are quite common.
  - The incidence of renal cysts increases with age, most people over the age of 60 have one or more simple renal cyst.

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Differential Diagnosis

Nongenetic

- Acquired disorders
  - Simple renal cysts
  - Cysts of the renal sinus
  - Acquired cystic kidney disease (i.e. CRF)
  - Multilocular cysts
  - Hypokalemia-related cysts
- Developmental disorders
  - Medullary sponge kidney
  - Multicystic dysplastic kidney
  - Pyelocalyoceal cysts

Genetic

- Autosomal-dominant
  - Autosomal-dominant polycystic kidney disease (ADPKD)
  - Tuberous sclerosis complex
  - von Hippel-Lindau disease
  - Medullary cystic disease
  - Glomerulocystic kidney disease
- Autosomal-recessive
  - Autosomal-recessive polycystic kidney disease (ARPKD)
  - Nephronophthisis
- X-linked
  - Orofaciodigital syndrome, type I

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Diseases that have renal cysts and liver cysts

- ADPKD
  - Adult onset (4th or 5th decade)
  - Large “bumpy” kidneys (due to renal cysts)
  - Hepatic cysts
  - Autosomal dominant inheritance
- ARPKD
  - Childhood onset (at birth , and die young)
  - Large, smooth kidneys (due to numerous small cysts)
  - Hepatic cysts, congenital hepatic fibrosis, and biliary duct ectasia
  - Autosomal recessive inheritance
- Tuberous sclerosis complex
  - Renal cysts, liver cysts, and liver angiomylipomas
- Nephronophthisis
  - Liver fibrosis, renal cysts, and hepatic cysts

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IMAGING CHARACTERISTICS OF THE MOST COMMON RENAL CYSTIC DISEASES

Disease	Kidney Size	Cyst Size	Cyst Location	Liver
Simple renal cysts	Normal	Variable (mm–10 cm)	All	Normal
Acquired renal cystic disease	Most often small, sometimes large	0.5–2 cm	All	Normal
Medullary sponge kidney	Normal or slightly enlarged	mm	Precalyceal	Normal (most often)
ADPKD	Enlarged	Variable (mm–10 cm)	All	Cysts (most often)
ARPKD	Enlarged	mm increase with age	All	CHF
NPH	Small	mm–2 cm (when present)	Medullary	Normal

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Imaging of Renal Cysts

- Plain abdominal radiography
- IVP
- Radionucleotide imaging
- Ultrasound
- Computed tomography
- MRI
- Renal angiography
- Retrograde pyelography

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Best Modalities for assessing Renal Cysts

- U/S is best for differentiating solid masses from cystic ones
- CT scan is best for finding small cysts and assessing for cystic rupture, infection, or hemorrhage (due to the use of contrast to get differential enhancement of cysts)
- MRI is useful for further characterizing renal cysts and detecting renal neoplasms

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More Information About the Case

- Further research revealed that this patient's father died of renal failure secondary to Autosomal Dominant Polycystic Kidney Disease
- This patient had been diagnosed with and worked up for ADPKD at an outside hospital previously

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ADPKD

- Autosomal dominant hereditary condition characterized by multiple expanding cysts of both kidneys that ultimately destroy the renal parenchyma and cause renal failure
- 100% penetrance and affects 1 of every 400-1000 live births
- The majority of cases are caused by a mutation in the PKD1 gene on chromosome 16, which produces polycystin 1 (involved in cell-to cell interactions)
  - The remainder of cases are caused by a mutation in PKD2 on chromosome 4, which produces polycystin 2 (less severe)
- Has variable expression and does not produce symptoms until adult life (4th or 5th decade of life)

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PKD1 vs. PKD2

PKD1

- Earlier presentation (average age 35)
- Earlier end-stage renal failure (average age 60)
- Earlier death (average age 70)

PKD2

- Later presentation (average age 61)
- Later end-stage renal failure (average age 74)
- Later death (average age 75)

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ADPKD cont.

- Cysts may also occur within the liver, pancreas, and spleen
  - Cysts are more common in the liver and tend to be less common in the pancreas and spleen
- Hepatic failure does not tend to occur despite extensive infiltration of the liver with cysts
  - However, cysts in the liver and pancreas can lead to obstructive jaundice
- Circle of Willis aneurysms are present in up to 41% of patients that undergo cerebral angiography

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ADPKD and imaging

- The imaging appearance varies with the severity of the disease
- Full-blown disease shows the presence of bilaterally enlarged kidneys with numerous cysts of varying sizes
- Ultrasound demonstrates cysts in the adolescent and young adult, who are usually asymptomatic
- Urography may be normal
- Ultrasound and CT are most sensitive
- MRI is useful for differentiating between simple cysts, hemorrhagic cysts, and neoplasms when CT and U/S are equivocal

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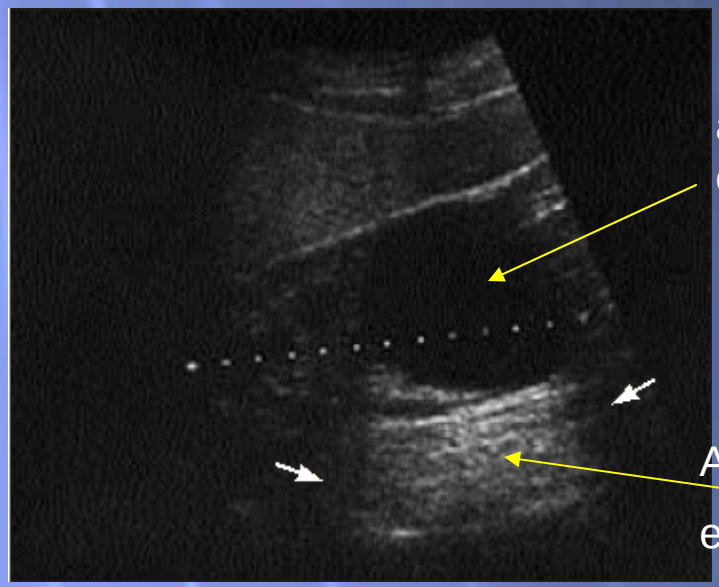
Ultrasonographic Diagnostic Criteria for ADPKD

- The absence of cysts before age 30 does not rule out the diagnosis of ADPKD
- False negative rate is inversely proportional to age
- If U/S diagnosis remains equivocal, CT should be used next (more sensitive for small cysts)

Age	Cysts
15-29	2, uni/bilateral
30-59	2 in ea. kidney
≥60	4 in ea. kidney



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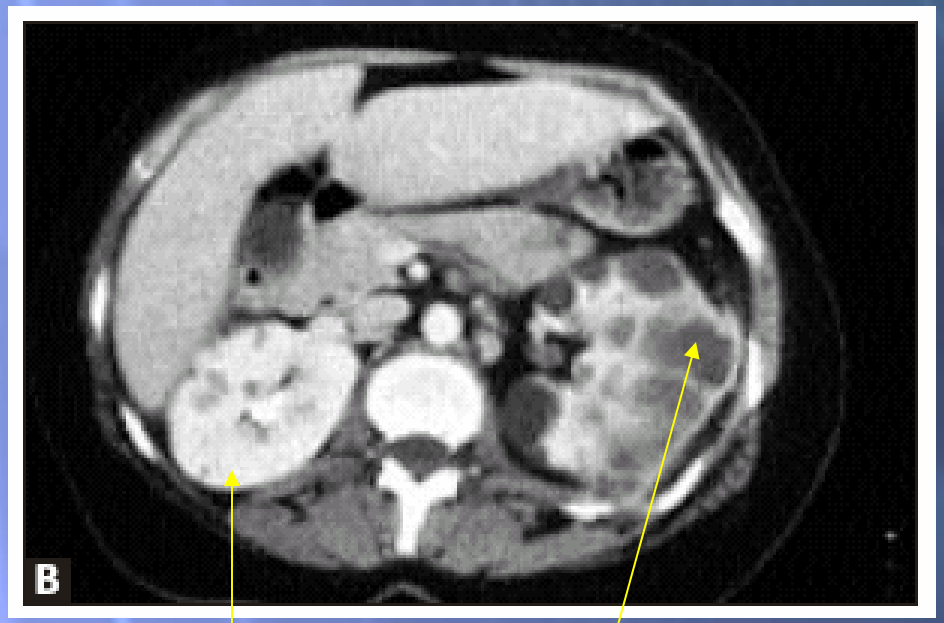
Classic Renal Imaging Findings in ADPKD

- Example of the cystic involvement of the kidneys
- Note the marked asymmetry in the number and size of cysts between the 2 kidneys
- CT is more sensitive at detecting small cysts than U/S



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Small kidney with minimal cystic involvement

Large kidney with multiple cysts of varying size

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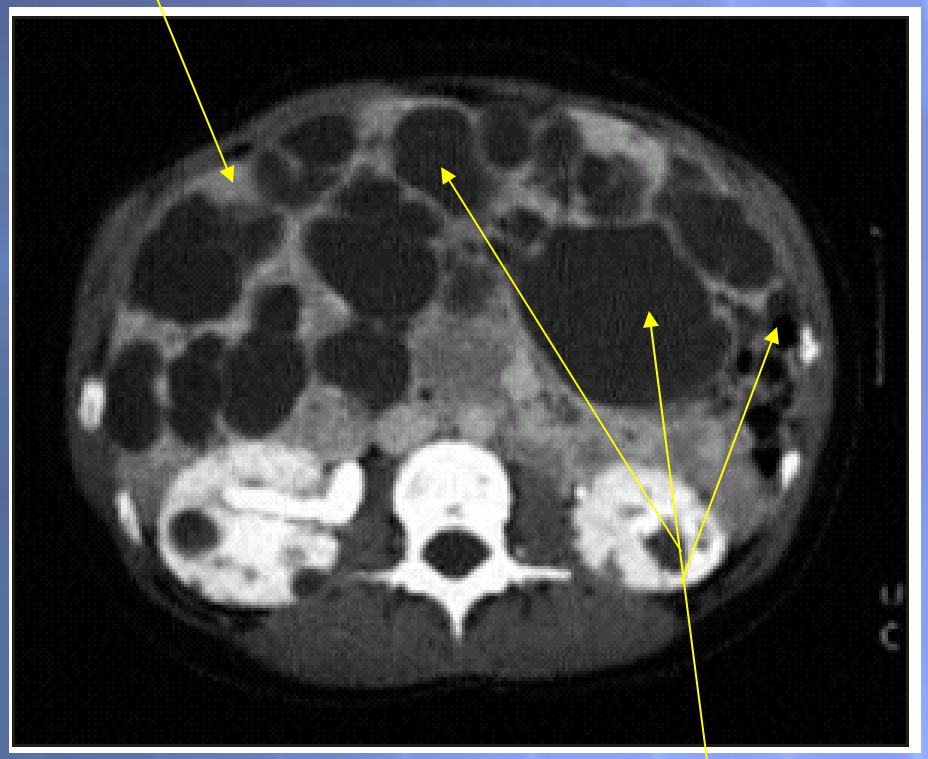
Classic Hepatic Imaging Findings in ADPKD



Atlas of Disease of the Kidney, Schrier

- Note the cystic involvement of the liver
- The presence of liver cysts helps to establish the diagnosis of ADPKD, especially if kidney involvement is mild
- Massive cystic disease of the liver can lead to early satiety, abdominal /diaphragmatic hernia, supine dyspnea, and rarely jaundice.

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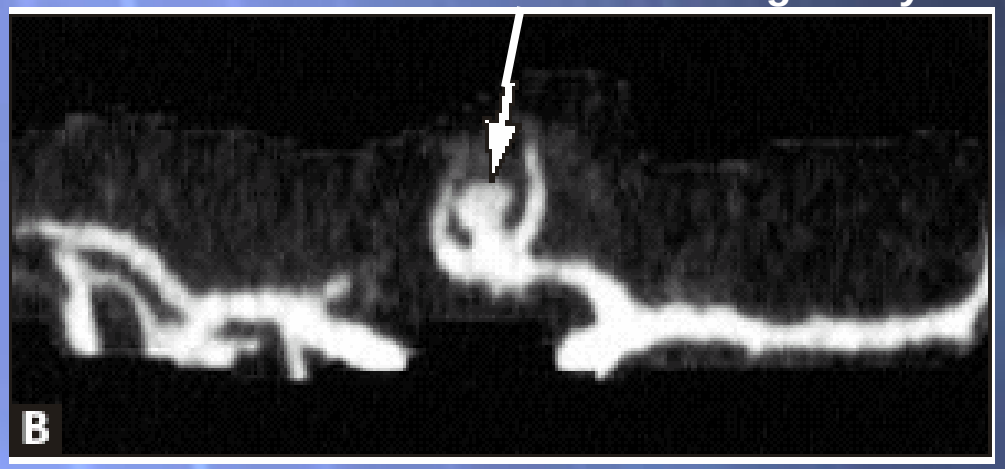
Intracranial aneurysms (ICAs) and ADPKD

- Prevalence of asymptomatic ICA in ADPKD is 8% (1.2% in the general population), increases to 16% with a family history of ICA
- ICA rupture entail a mortality of 30-50% (results in SAH)
- First-line diagnostic procedure for ICA rupture is CT

ICA on the anterior communicating artery



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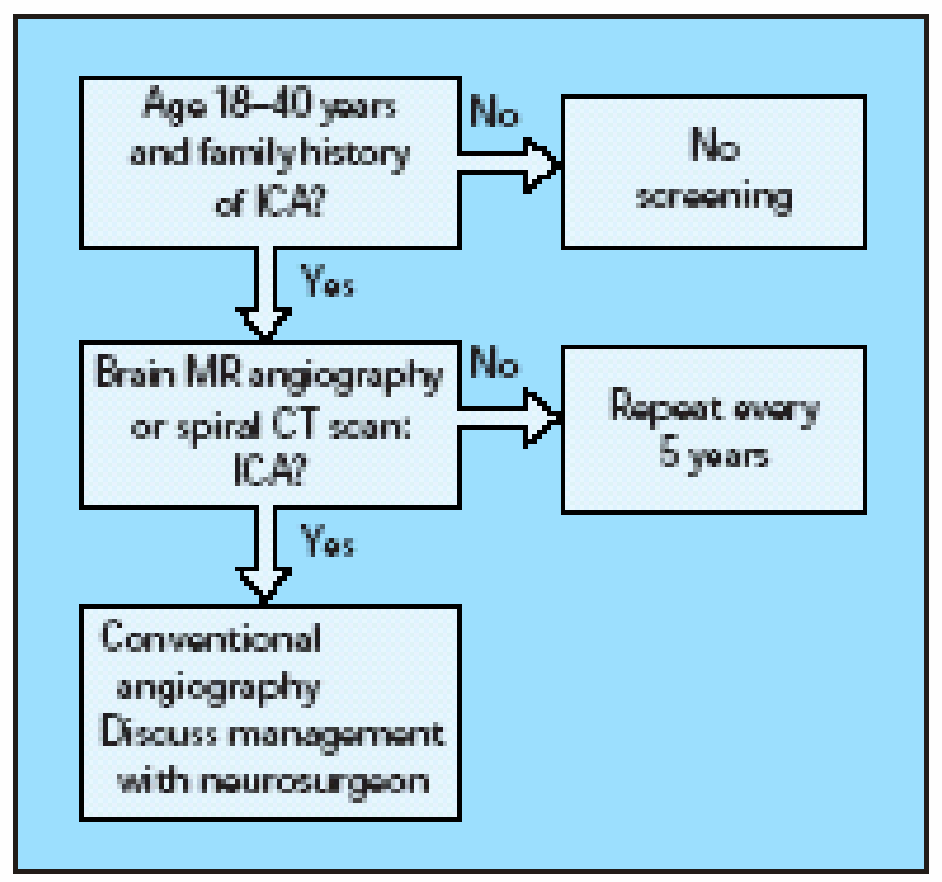
Indications for Screening for ICA in ADPKD



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- Screening in ADPKD for ICA can be achieved with either MRA or spiral CT angiography

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Treatment

- Treatment is largely supportive
  - The associated hypertension can be managed medically
  - Renal failure can be managed with dialysis
  - Kidney transplantation is the only way to “cure” the renal disease

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Summary

- ADPKD is a disease with onset in adulthood
- ADKPD is associated with enlarged, cystic kidneys, hepatic cysts, pancreatic cysts, splenic cysts, and ICAs.
- U/S is the best imaging method for detecting renal cysts, however, CT may be more sensitive for small renal cysts
- It is important to screen ADPKD patients for ICA with spiral CT angiography or MRA in accordance with the appropriate indications.

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