Case #130

NAME Educational Activities Committee

Case provided by:

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Photos – Image A

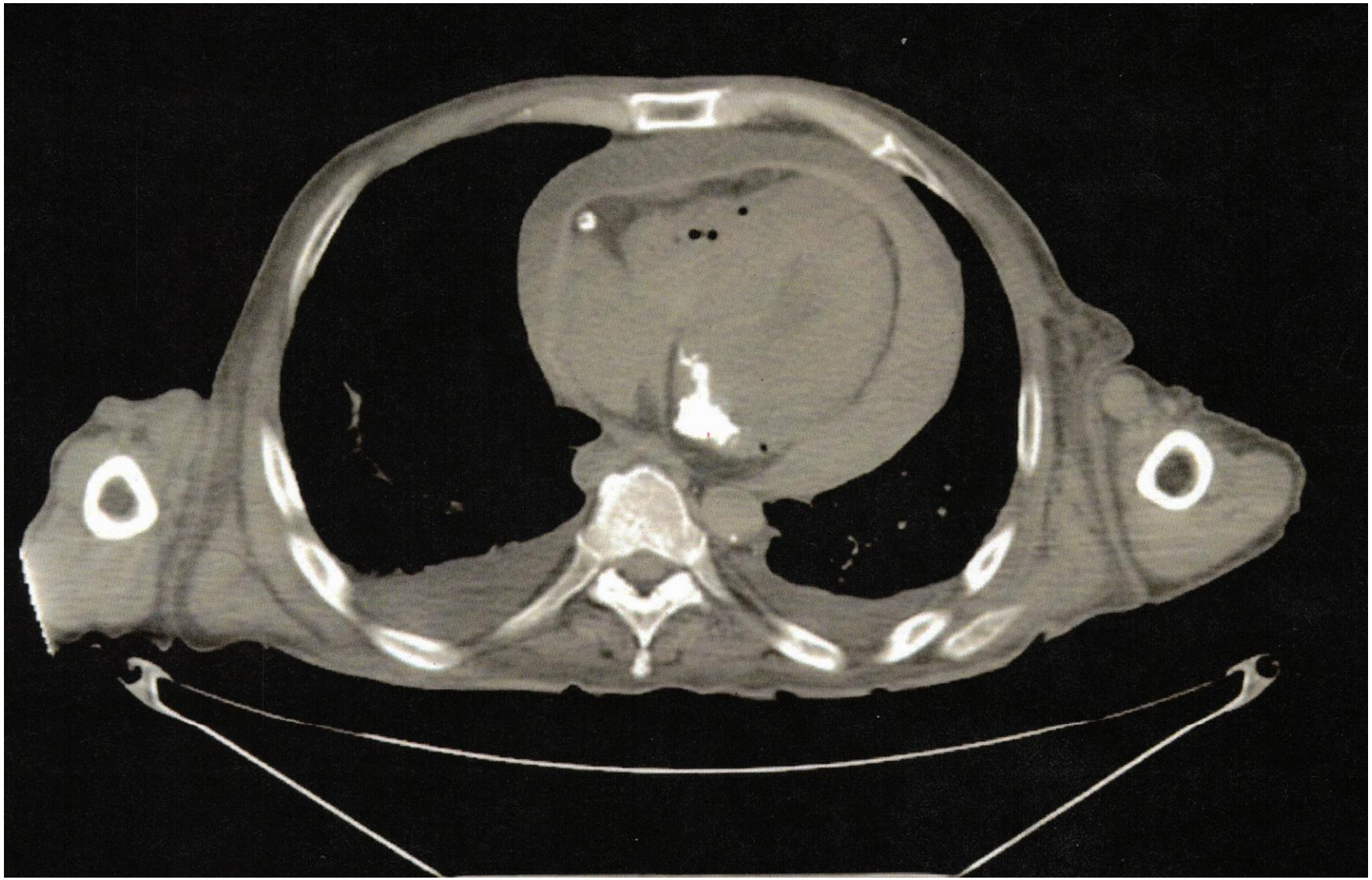
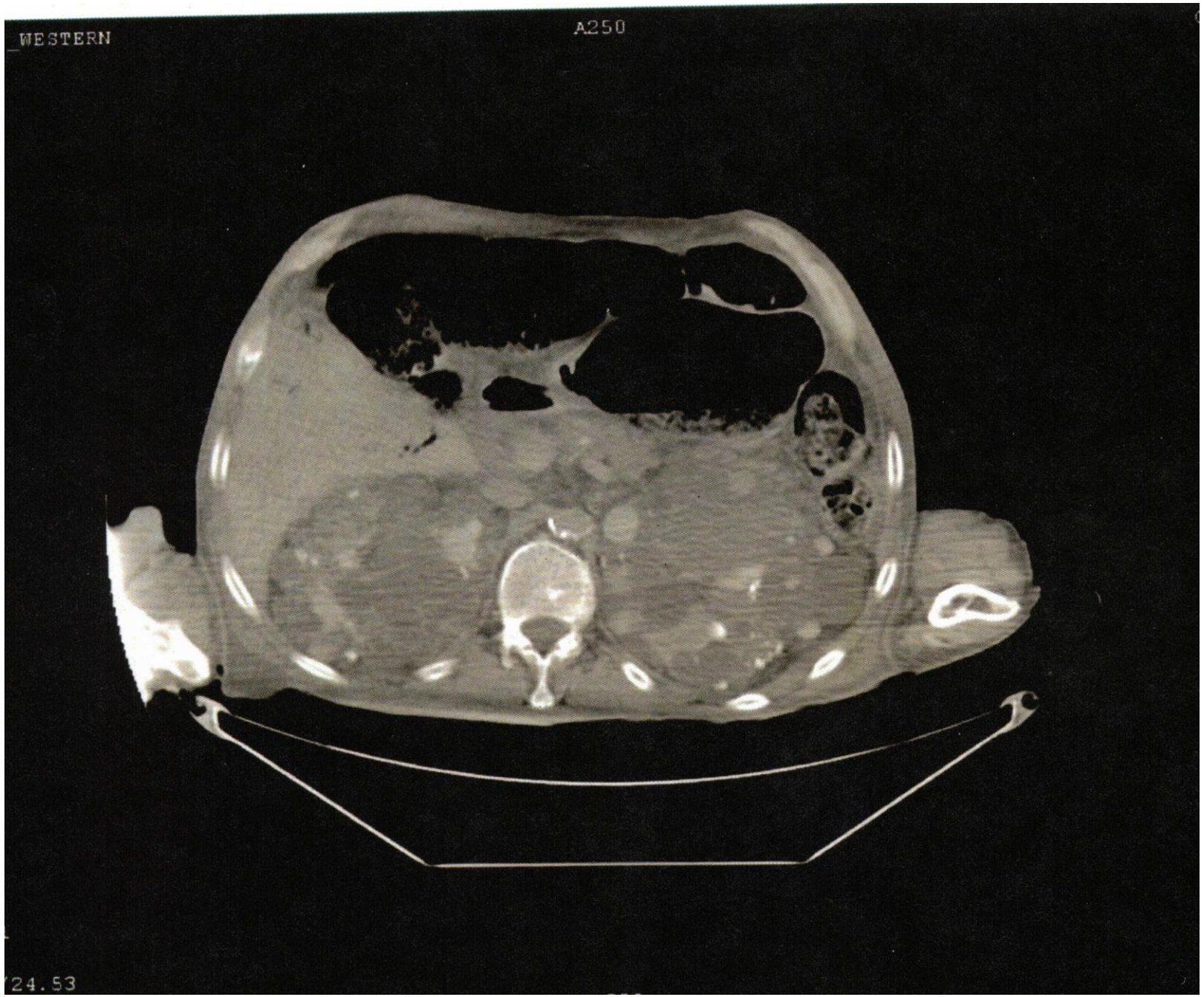


Image B



1. A 67-year-old man was the restrained driver of a motor vehicle that left the roadway at low speed, struck a guardrail and went down an embankment. There was minimal damage to the vehicle. First responders reported no visible external trauma and he was pronounced dead on scene. He was transported to the Medical Examiner's office for examination. No medical history was available at the time of examination.

Postmortem CT demonstrated no fractures. The findings in the images were noted. An autopsy was performed to better classify the reason for the findings in image A to determine natural vs. accidental manner of death, particularly in the setting of the process visible in Image B.

Which of the following additional findings is commonly associated with the disease process in Image B in this case?

- ◯ Atherosclerotic cardiovascular disease
- ◯ Non-traumatic subarachnoid hemorrhage
- ◯ Congenital hepatic fibrosis
- ◯ Aortic transection at the ligamentum arteriosum

Answer...

B. Non-traumatic Subarachnoid hemorrhage (CORRECT ANSWER, 39.45% of responses)

Image A shows a fluid collection around the heart of a density comparable to blood (hemopericardium), which can be both associated with trauma or non-traumatic natural disease processes. An autopsy was performed to determine the source of the fluid collection. An aortic dissection with focal rupture was identified in the ascending aorta just distal to the aortic valve with 800 cc of hemopericardium (no pleural fluid collections). The ascending aorta was dilated from the aortic root to the right subclavian artery, consistent with ascending thoracic aortic aneurysm. The remainder of the arch and descending aorta were unremarkable without any additional abnormalities except focal intimal atheromatous plaques.

Image B demonstrates enlarged kidneys occupying the entire retroperitoneum composed of variable cystic shapes (autosomal dominant polycystic kidney disease). Further imaging in this case demonstrated a transplanted kidney in the right pelvic region and review of medical records obtained after autopsy verified a clinical diagnosis of autosomal dominant polycystic kidney disease (ADPKD).

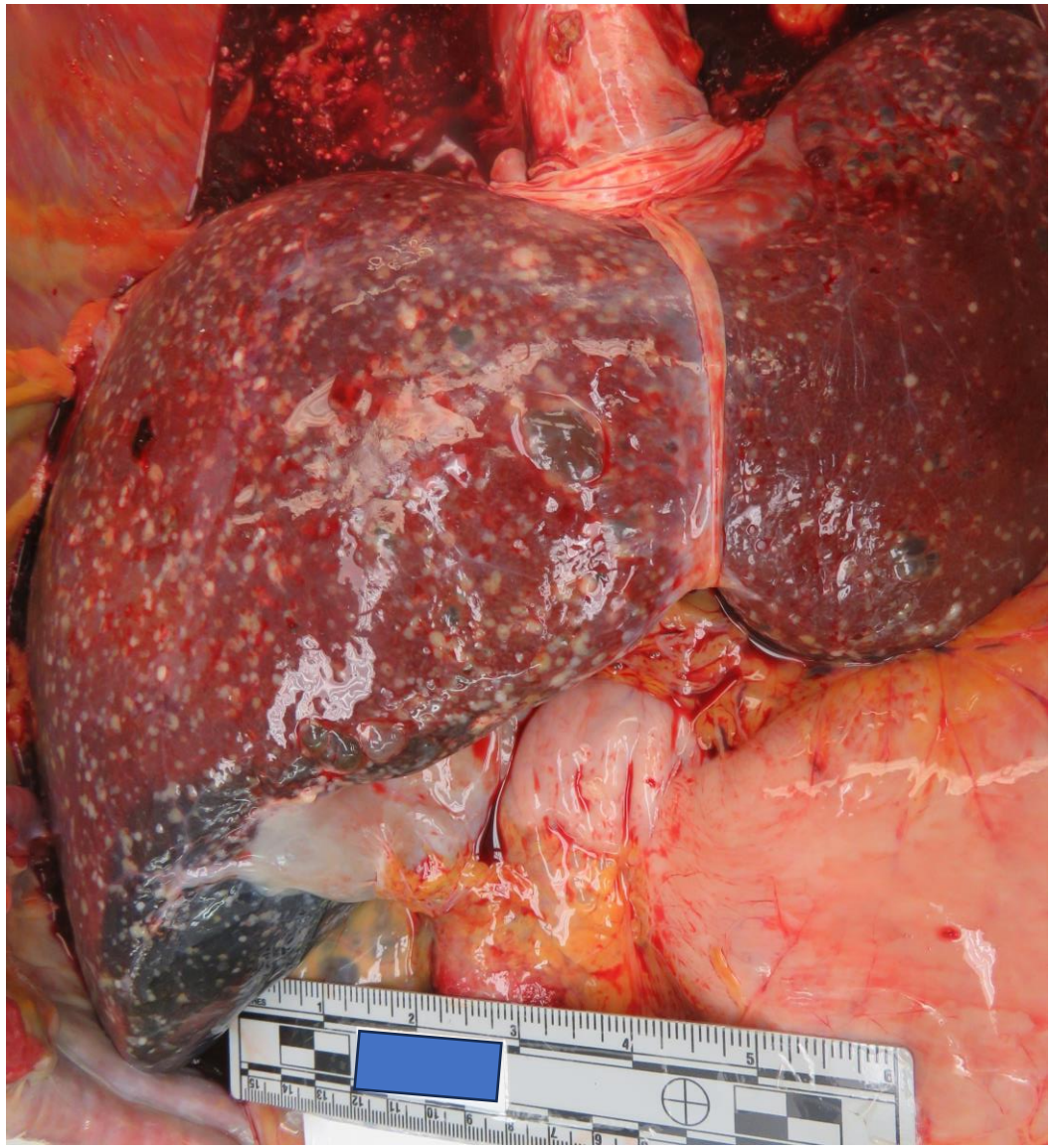
Autosomal dominant polycystic kidney disease is a genetic disorder primarily related to mutations of the PKD1 gene on chromosome 16 or the PKD2 gene on chromosome 4.[1] The PKD1 gene mutations tend to occur earlier with more severe clinical presentations than those of the PKD2 gene. Clinical signs and symptoms can include hypertension, renal dysfunction with proteinuria or hematuria. Secondary urinary tract infections or renal calculi can also occur. Symptoms may also be associated with extrarenal manifestations, including liver or pancreatic cysts, cerebral aneurysm increasing risk for non-traumatic subarachnoid hemorrhage, ascending aortic aneurysms/dissections, heart valve abnormalities, diverticulosis and hernias. While this decedent did not have a cerebral aneurysm, the risk for subarachnoid hemorrhage in these patients is another potential finding on postmortem CT that could also be mistaken for trauma in the setting of a motor vehicle collision.

The most common presentation of ADPKD is hypertension, occurring in 50-70% of patients prior to renal function compromise. The pathogenesis of hypertension associated with ADPKD is thought to be due to a combination of renal dysfunction and vascular dysfunction due to abnormal polycystin, which modulates vasodilation[2]. Cerebral aneurysms occur in patients with ADPKD at a rate 4 times that of the general population.[3] Liver cysts can be asymptomatic or present with pain due to infection[4]. The most common valvular abnormalities reported are mitral valve and aortic valve regurgitation.[2]

ADPKD is associated with higher rates of aortic aneurysms and dissections. The vascular and valvular changes associated with ADPKD may be due to polycystin having a role in arterial smooth muscle cells.[5, 6.] Recognizing this risk in patients with ADPKD can add a differential for the finding of a hemopericardium in cases that may or may not be related to trauma.

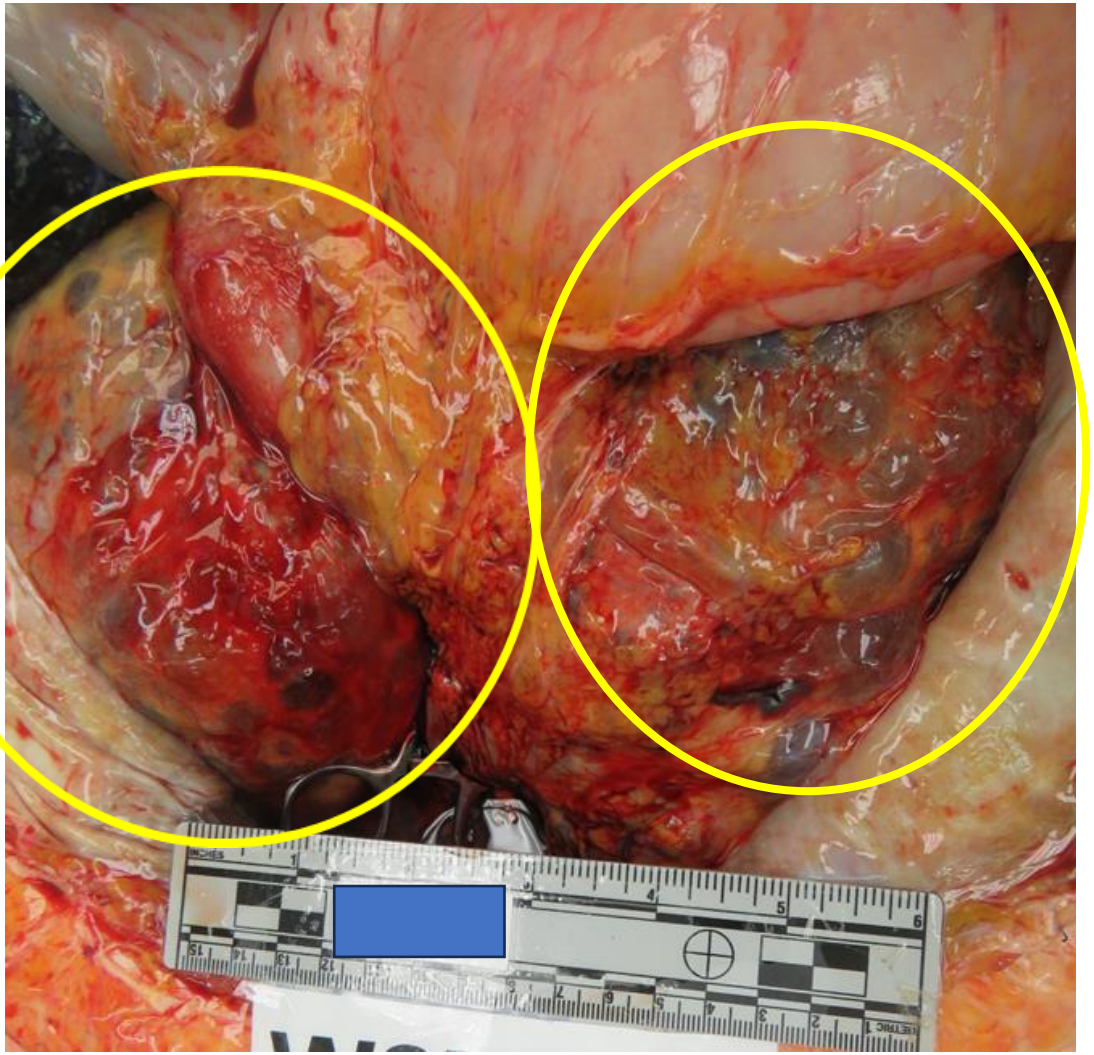
The use of postmortem computed tomography (PMCT) in a forensic setting is becoming more frequent but soft tissue and vascular abnormalities can be difficult to visualize in PMCT. This, in part, is due to the limited ability to perform angiography. There are some findings on unenhanced PMCT that can indicate aortic dissection, including displaced intimal calcification, an intimomedial flap between a true and false lumen and double sedimentation (two distinct levels of sedimentation in the true and false lumen or single sedimentation in one lumen but no the other). Some possible complications in interpreting these signs can include postmortem clot mimicking double sedimentation. Decomposition and collapse of vessels can also hamper interpretation of vascular dissection and in about a quarter of cases reviewed in one report, no clear signs of dissection were evident.[7]

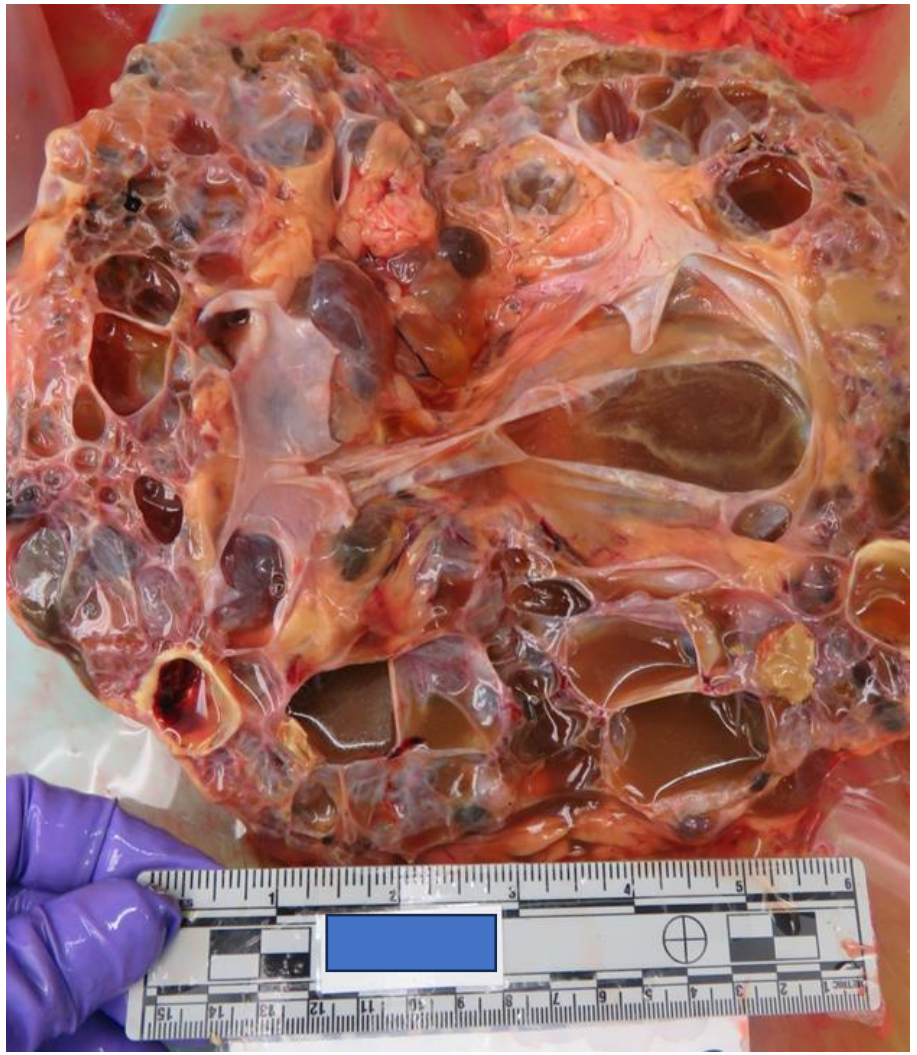
Additional autopsy photos



- Polycystic liver disease is associated with ADPKD. In this particular case, the liver had numerous simple cysts and von Meyenberg complexes (bile duct hamartomas)[3, 8].

Kidneys (in situ and bisected)





Other responses…

A. Atherosclerotic cardiovascular disease (18.63% of responses)

Atherosclerotic cardiovascular disease is a known cause of acute myocardial infarcts which can rupture leading to hemopericardium. This decedent did have some atherosclerotic calcifications in the coronary arteries (seen as white foci in the heart on image A, but this is more related to his overall health and not specifically associated with ADPKD. There were no acute findings of the heart muscle at autopsy and no wall defects.

C. Congenital hepatic fibrosis (14.52% of responses)

Congenital hepatic fibrosis is a known association of autosomal recessive polycystic kidney disease (ARPKD). It is a genetic disorder associated with PKHD1 gene mutations. The gross appearance of kidneys with ARPKD is enlarged but with a “sponge-like” appearance with cystic structures largely focused on the medulla, rather than the large fluid filled cortical cystic spaces seen in ADPKD. This is due to the ARPKD being a ciliopathy affecting the distal convoluted tubules and collecting ducts. [9] Clinically, patients with ARPKD usually present in childhood.

D. Aortic transection at the ligamentum arteriosum (27.40% of responses)

Sudden decelerations seen in motor vehicle collisions can cause tearing of the aorta, typically seen at the level of the ligamentum arteriosum past the arch.[10] The aorta distal to this location is fixed/anchored to the vertebral column and rib cage through adventitia and intercostal branches. Proximal to this point, the ascending aorta and arch are capable of forward or side to side movement during a sudden deceleration or side impact respectively, stretching and possibly tearing the vessel wall in the region of the ligamentum.

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