H144 An Uncommon Cause of Pulmonary Embolism

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The goal of this presentation is to increase awareness of uncommon sources/locations of thromboemboli in patients with autosomal dominant polycystic kidney disease.

This presentation will impact the forensic science community by highlighting the importance of examining the inferior vena cava and pelvic vessels in patients with polycystic kidney disease.

Case: A 20-year-old African American female was found unconscious and pulseless in her bedroom. She was last known alive ½ hour prior to discovery. When an ambulance arrived, paramedics intubated her without difficulty and initiated advanced cardiac life support. On arrival at the emergency department, an intraosseous line was placed and the correct position of the endotracheal tube verified. Despite these efforts she remained pulseless, without respirations and blood pressure, and was pronounced dead after 15 minutes.

Prior to the autopsy, the only available medical history indicated that three days before her death she complained of cold-like symptoms and began taking an antibiotic.

The external examination revealed an African American female measuring 5 feet 3 inches in height and weighing 233 pounds. The lower extremities were symmetrical and did not have edema.

The internal examination found large polycystic kidneys occupying the abdominal cavity and extending into the pelvis. The right kidney weighed 1,421 grams and measured 22cm in length and 12cm in width. The left kidney weighed 1,456 grams and measured 19cm in length and 13cm in width (normal range for women — weight: 115-155 grams, length: 11-12cm, width: 5-7cm). The right lung weighed 313 grams and the left lung weighed 299 grams. The pleural surface of the left upper and lower lobes revealed darker red/tan areas respectively measuring 3cm and 3.5cm. The cut surfaces through these areas were consistent with the appearance of pulmonary infarcts.

Opening the pulmonary trunk and the right and left pulmonary arteries revealed thromboemboli which ranged from 1cm to 6cm in length and up to 1cm in width. The proximal right and left pulmonary arteries had adherent thromboemboli. The right and left iliac veins and the inferior vena cava had adherent thrombi. Examination of the veins of the lower extremities was negative for thrombi.

There was mild cardiomegaly (heart weight: 396 grams; LV: 1.6cm) with normal valves and bilateral polycystic ovaries. There were no cysts involving the liver, pancreas, or spleen. There were no aneurysms of the aorta or Circle of Willis. Toxicology was negative for ethanol, cocaine, and heroin. Vitreous electrolytes had a normal postmortem pattern. A nasopharyngeal viral culture was negative.

The patient's mother confirmed that her daughter had polycystic kidney disease and was followed by a physician for her disease but had been putting off going to see her doctor for over a year because she had no complaints, such as fullness in her abdomen, leg pain, and/or swelling.

Autosomal Dominant Polycystic Kidney Disease (ADPKD) is one of the most common inherited disorders and is the most frequent genetic mutation leading to renal failure in adults, accounting for % to 8% of patients on dialysis in the United States. The disease is progressive, can affect multiple organs systems, and is characterized by the formation and enlargement of cysts in the kidneys. Extra-renal cysts are most commonly found in the liver. The clinical features usually begin in the third to fourth decade of life, although cysts can be detected in childhood and *in utero*. Although the presentation of the disease is highly variable, the most common initial complaint is pain and/or discomfort in the abdomen, flank, or back, typically as the result of liver and kidney cysts exerting pressure in these areas.

ADPKD is caused by mutations in one of two genes — PKD1 which codes for polycystin 1 and PKD2 which codes for polycystin 2. Approximately 85%-90% of patients with ADPKD have the PKD1 mutation (ADPKD 1) located on the short arm of chromosome 16 (16p13.3). ADPKD 2 accounts for 10%-15% of cases. The mutation is found on the long arm of chromosome 4 (4q21-22). Individuals with ADPKD 1 have an earlier mean age of onset of end-stage renal disease (53 years), compared to those with ADPKD 2 (74 years).

Two studies have described the causes of death in 217 patients with ADPKD. Prior to 1975 and the use of renal replacement therapy, the average age of death was 51 years, increasing to 59 years by 1995 and 60.5 years by 2009. Cardiac disease is responsible for most deaths, followed by infection and central nervous system causes. A total of eight individuals died as the result of pulmonary embolism. Although detailed information was not available for these patients, there was no thrombosis of the vena cava or renal vein noted in seven of the eight patients.

Only a handful of cases of pulmonary embolism due to thrombosis of the inferior vena cava and/or ileofemoral vessels caused by compression of the infra-hepatic inferior vena cava by renal cysts by have been reported. None of these were fatal and the development of thrombosis of the inferior vena cava was independent of renal function.

The risk factors for the development of a pulmonary embolism are well known. There was no history of coagulation abnormalities, immobility, or cancer in this patient. Although this patient had a BMI of 41.3, placing her in the obese category, it is proposed that thrombosis of the inferior vena cava and iliac veins was a result of extrinsic compression by the kidneys and death was due to pulmonary embolism caused by compression of the inferior vena cava and pelvic vessels due to polycystic kidney disease.

Polycystic Kidney Disease, Pulmonary Embolism, Inferior Vena Cava