Quick Recertification Series

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POLYCYSTIC KIDNEY DISEASE (PKD)

GENERAL FEATURES

- PKD is a genetic disorder that can exhibit systemic manifestations but primarily affects the kidneys.
- It is caused by a gene defect on either chromosome 4 or 16. The chromosome 16 gene is the more common defect and is characterized by a faster progressing disease.
- Autosomal dominant PKD (ADPKD), the most common form of the disease, affects 1:400 to 1:1,000 persons and is often diagnosed in the fourth decade of life.
- Autosomal recessive PKD manifests in childhood, is associated with poorer outcomes, and is significantly less common than the autosomal dominant form (1 in 20,000 persons affected).
- Fifty percent of patients with PKD will have end-stage renal disease (ESRD) by age 60 years.
- Fifty percent to 80% of patients with PKD will have hypertension.
- Other associated disorders include infected renal cysts/renal abscesses, nephrolithiasis, hepatic cysts, cardiac valvular disease, and intracranial aneurysms (4% of younger patients, 10% of older patients).

CLINICAL ASSESSMENT

- History
  - Usually patients present in their 20s to 40s with varying signs and symptoms including abdominal or back pain (secondary to large renal cysts), hematuria, recurrent urinary tract infections (UTIs), hypertension, presence of a palpable abdominal mass, or in acute renal failure.
  - Often kidney cysts are found incidentally in asymptomatic patients on imaging studies.
  - Family history is present in 75% of cases; therefore, a detailed family history is needed with a focus on kidney disorders and intracranial aneurysms.
- Physical examination
  - BP screening and monitoring is important given the high incidence of concomitant hypertension with PKD.
  - Kidneys may be so large that they are palpable.
- Laboratory/radiology studies
  - Urinalysis can show hematuria, proteinuria (usually mild), or signs of UTI. Fat oval bodies are almost pathomneumonic for PKD, being otherwise routinely found only in nephrotic syndrome.
  - Abdominal ultrasonography (US) is the test of choice for diagnosis in symptomatic patients and for screening high-risk patients.
  - CT or MRI scans can be used as a confirmatory test if the US results are unclear.
  - Neuroimaging to screen for intracranial aneurysms is controversial and should be reserved for patients with a family history of intracranial aneurysm. Depending on the size, the risk of intervention is approximately equal to the risk of bleeding.
  - Urography can be beneficial in patients with recalcitrant hematuria or UTIs. With recalcitrant infections, CT urography can help determine if pyelonephritis has progressed to infect the cysts.

DIAGNOSIS

- Diagnostic criteria of ADPKD
  - Two kidney cysts on either kidney before age 30 years
  - Two kidney cysts in one kidney at age 31 to 59 years
  - Four kidney cysts in one kidney after age 60 years

TREATMENT

- Symptomatic treatment includes adequate pain control, management of renal failure, and strict control of hypertension.
- Patients often have gastroenterologic symptoms from the mass effect of the large kidney cysts.
- Tight BP control can help prevent progression to ESRD.
- Diligent diagnosis and treatment of UTIs are essential.
- Renal transplant can be a treatment option for management of ESRD.
- Patients often require bilateral nephrectomy for infection and pain management.

NEPHROTIC SYNDROME

GENERAL FEATURES

- Nephrotic syndrome is defined by the triad of heavy proteinuria (>3 g/24 h), hypoalbuminemia, and peripheral edema.
- It is often the end stage of other renal disease such as glomeulonephritis and diabetic and hypertensive nephropathies.

QUESTION & ANSWER

1. The most common condition accompanying polycystic kidney disease (PKD) is

a. Hypertension
b. Intracranial aneurysm
c. Hepatic cysts
d. Mitral valve prolapse

Answer: a

Explanation: All of the above conditions are associated with PKD, but hypertension is seen in up to 80% of patients. Hypertension screening should be routinely carried out in patients with PKD.

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- The disorder can be the result of either a primary kidney or systemic disease. In adults, the main causes include diabetes and amyloidosis. The most common primary kidney cause is membranous nephropathy.
- In children, most cases (90%) are caused by minimal change disease (also known as *nil disease secondary to IgA nephropathy*), which refers to the normal appearance of the histopathology.
- Albumin is the main protein excreted, but others include clotting factors, transferrin, and hormone/vitamin binding proteins.
- Several other conditions are often present but not included in the diagnostic criteria, including thrombosis and hyperlipidemia.

CLINICAL ASSESSMENT

- History
  - Patients will often report symptoms related to a fluid overload/low protein state, including edema, shortness of breath, weight gain, and/or facial swelling.
  - Patients may report foamy urine as a result of the protein present.
  - Other symptoms can include nonspecific symptoms related to kidney failure, including weakness, fatigue, anorexia, and oliguria.
- Physical examination
  - Focus is on determining the patient's fluid status. Examination findings often show not only peripheral edema but also other signs of fluid overload, including ascites, crackles (rales) on chest auscultation, and evidence of pleural effusion. Uremic pericarditis may occur with a pericardial friction rub and evidence of tamponade such as elevated neck veins. Fluid overload may cause heart failure.
- Laboratory/radiology studies
  - Urinalysis will show heavy (2+) proteinuria. Can be accompanied by lipiduria and hematuria (less common).
  - Serum creatinine levels may be elevated, depending on the underlying cause and degree of disease
  - Serum calcium levels may be low. Be sure to correct serum calcium measurements based on the patients albumin level (ie, calculate corrected calcium).
  - Patients may be vitamin D deficient.

DIAGNOSIS

- Renal biopsy can be helpful if reversible cause is suspected (causes other than diabetes or amyloidosis) and the patient does not have contraindications to biopsy.

TREATMENT

- Treatment and prognosis depends on the underlying condition.
- General considerations include fluid and electrolyte maintenance. Patients will often require fluid restriction and diuresis to avoid fluid overload states.
- Monitor renal function and avoid nephrotoxic medications.
- Either ACE inhibitors or angiotensin receptor blockers can be used in patients with nephrotic syndrome and no contraindications to therapy. In these patients, therapy is geared at reducing proteinuria and delaying disease progression.
- In patients with nephrotic disease secondary to nil disease, corticosteroids are often effective. Recurrent exacerbations can be treated with corticosteroids or immunosuppressive agents. Corticosteroid-responsive patients have good long-term prognosis and rarely develop ESRD.
- Hyperlipidemia should be aggressively treated.
- At extremely low albumin levels, patients can become prothrombotic. Clinicians should maintain a high suspicion for venous thrombosis in patients with nephrotic syndrome, especially those with other venous thrombosis risk factors.
- Children are often started on corticosteroids presumptively without a renal biopsy. They are often corticosteroid-responsive.
- Patients with nephrotic syndrome should be referred for nephrology consultation. JAAPA

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QUESTION & ANSWER

1. A 55-year-old diabetic patient is admitted with anasarca, ascites, and progressing renal failure. During the course of the hospital evaluation, the patient is noted to have symmetric peripheral edema, heavy proteinuria (>3 g/24 h), and hypoalbuminemia. A diagnosis of nephrotic syndrome is made. On hospital day 5, the patient is noted to have significantly worsening pedal edema; the left is worse than the right. Which statement is the most accurate in regards to the patient's worsening lower-extremity edema?

a. This is a common feature of nephrotic syndrome and requires continued observation.
b. The patient should be screened for deep venous thrombosis.
c. The diuretic dose should be increased and daily fluid restriction instituted.
d. The diuretic dose should increased without a daily fluid restriction

Answer: b

Explanation: Nephrotic syndrome is often associated with deep venous thrombosis especially when combined with other risk factors such as hospitalization and/or immobilization. Often patients with nephrotic syndrome will require diuretic therapy and daily fluid restriction to control edema; but in the setting of worsening unilateral edema, venous thrombosis must be ruled out including risk stratification (Wells score) and, possibly, lower-extremity Doppler ultrasonography.