Raising Awareness: Polycystic Kidney Disease

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Introduction

A young male patient was recently admitted to the intensive care unit at a local hospital with hypertensive emergency and acute renal failure of unknown etiology. A renal ultrasound showed polycystic kidney disease (PKD). After further investigation, it was discovered that the patient’s mother passed away last year from end stage renal failure related to polycystic kidney disease (PKD). The aforementioned genetics research has led to a recent review of inherited disorders are what prompted further investigation into this case and the recent study into PKD. Liebau and Serra (2013) explain that “inherited cystic kidney diseases, autosomal dominant polycystic kidney disease (ADPKD), and autosomal recessive polycystic kidney disease (ARPKD), are the most common monogenetic causes of end-stage renal disease (ESRD) in children and adults” (p. 1771). It was estimated that one in a thousand individuals will be diagnosed with PKD in adulthood and one in twenty-five of children (Liu et al., 2012, p. 1). Considering this statistic, patients with PKD may be encouraged to educate and appropriately treat patients in an effort to prevent unnecessary damage to their bodies related to the complications of the disease. The utilization of genetic testing is not standard treatment, but a study into its benefits to certain patients can aid in identifying the disease. As a primary health care provider it must be made to decrease risks to either genetic testing is necessary in order to slow disease progression.

Pathophysiology

Eisenberger, et al. (2015) state that “renal cysts are clinically and genetically heterogeneous conditions” (p. 1). ADPKD is the most commonly diagnosed form of PKD and is typically diagnosed during adulthood; it results from a mutation in either of two genes, PKD1, which encodes polycystin 1 (PC1) or PKD2, which encodes polycystin 2 (PC2), both of which are membrane proteins (Reed- Gitomer, 2014, p. 17). Srivastava and Patel (2014) explain that “mutations of PKD 1 (eight-five percent of cases) or PKD 2 (fifteen percent of cases) can lead to signal dysregulation and increased levels of intracellular calcium, monophosphatase, culminating in cytogenesis. Given the dominant nature of transmission, there is at least a fifty percent probability that a child of an affected parent will inherit the disease. A spontaneous mutation causes ADPKD in five percent of cases” (p. 303). It is difficult to define genetic mutations for ADPKD early on due to the size and structures of PKD 1 and 2. Since the discovery of the aforementioned genetic mutations can be difficult, genetic testing is performed to definitively diagnose the condition of PKD is based on two factors, a positive family history and abnormal cystic renal phenotype (Bataille, Berland, Fontes, & Burty, 2011, p. 1). During genetic testing, DNA linkage is performed and based on the results, the DNA is sequenced. A variety of clinical trials with regards to slowing the progression of PKD have made advances in recent years (Sitomer, 2014, p. 177). The enlarging kidneys of PKD will eventually lead to progressive renal failure is an inevitability in PKD, as a primary health care provider it must be made to decrease risks to either genetic testing is necessary in order to slow disease progression.

Clinical Manifestations

The enlarging kidneys of PKD progress over decades, which leads to renal failure in the majority of patients by age sixty (Ma, Tian, Qiu, & Lou, 2013, p. 303). Many patients are asymptomatic until later in life; most are diagnosed between the ages of thirty and forty. The gradual loss of nephrons related to the formation and increasing size of cysts is not usually detectable during the first few decades of life (Srivastava & Patel, 2014, p. 303). The presentation of the disease varies based on the presence of other comorbidities. Early signs or symptoms include:

- flank or abdominal pain
- macroscopic hematuria
- frequent urinary tract infections (UTIs)
- early-onset hypertension

PKD patients are at a higher risk for:
- Renal stones
- Intracranial aneurysms
- Gross hematuria
- Abdominal manifestations, such as cysts that may appear on surrounding organs
- Renal dysfunction
- Cardiovascular diseases associated with hypertension

The preferred diagnostic tools are imaging studies. Ultrasonography is the preferred method due to its cost effectiveness, however magnetic resonance imaging (MRI) or computed tomography (CT) images are more sensitive. Once a patient is diagnosed, the best option is to adopt healthy lifestyle choices in attempt to slow disease progression and minimize the risk of developing other comorbidities

Implications for Nursing Practice

As a primary health care provider it is important to obtain a detailed family history and comprehensively evaluate the patient, including symptoms, signs, social behaviors, diet, and exercise. The most common medical care provided to PKD patients is education; key topics include exercise, nutrition, medication compliance. Adequate exercise and an appropriate diet are important components in almost any disease process; the patient should have a diet that is low in cholesterol to reduce cardiovascular risks, as well as protein intake to slow progression and prevent further renal injury (Maditz, Gigliotti, & Tou, 2013, p. 303). Primary modalities of treatment are related to disease-related symptoms, such as hypertension and pain. It is extremely important for PKD patients to monitor their blood pressure frequently and maintain medication compliance in regards to hypertension, if applicable. Once PKD has progressed to ESRD, patients will likely require hemodialysis and/or a renal transplant. However, over the last few years, clinical trials have been performed in regards to slowing the progression of PKD have made advances in recent years (Sitomer, 2014, p. 177). In the grand scheme of diseases, PKD is considered rare. However, once it is diagnosed the complications that can result from it are life threatening. PKD is frequently misdiagnosed because many of the signs and symptoms that go along with it are similar to other ailments/diseases, such as primary hypertension or a typical UTI. Once a patient is diagnosed with PKD, he or she must be made to decrease risks to either genetic testing is necessary in order to slow disease progression and minimize the risk of developing other comorbidities

References


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