Raising Awareness: Polycystic Kidney Disease

Dana Dendinger

Otterbein University, dana.dendinger@otterbein.edu

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Introduction

A young male patient was recently admitted to the intensive care unit at a local hospital with hypertension, renal failure, and acute renal failure of unknown etiology. A renal ultrasound showed polycystic kidney disease (ADPKD) and prompted further investigation. Upon autopsy and upon testing, it was discovered that the patient had inherited this disease from his mother passed away last year from end stage renal failure related to polycystic kidney disease (PKD). The aforementioned genetic testing is not utilized for PKD because inherited disorders are what prompted further investigation into this patient's condition. Smith (2013) explain that “inherited cystic kidney diseases, autosomal dominant polycystic kidney disease (ADPKD), and autosomal recessive polycystic kidney disease (ARPKD), are the most common monogenic causes of end-stage renal disease (ESRD) in children and adults” (p. 1771). It is estimated that one in a thousand individuals will be diagnosed with PKD in adulthood and one in twenty will be diagnosed with ARPKD (Liu et al, 2012, p. 13). Considering this statistic, patients with PKD may be at a great risk of developing end-stage renal failure. As a healthcare provider, primary or otherwise, it is possible that you may educate and appropriately treat patients in an effort to prevent unnecessary complications related to their condition. The utilization of genetic testing is not standard treatment; however, study into their benefits to patients who are diagnosed with PKD is necessary to order in secondary 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-Gittomer, 2014, p. 17). Srivastava & 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, monophosphorylating, culminating in cytogenesis. Given the dominant nature of this disease, there is a 50 to 50 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 determine genetic mutations for ADPKD early on due to the size and structures of PKD1 or PKD2. Since the detection of the aforementioned genetic mutations can be difficult, genetic testing can be of great benefit. The diagnosis of PKD is based on two factors, a positive family history and the eventual presentation of PKD as an adult, which can be a direct result of age-based symptoms or early diagnosis of PKD1 and PKD2. The renal vessels and obstruct their flow, which ultimately leads to intrauterine and remains irreversible, leading to the activation of the renin-angiotensin-aldosterone system (RAAS), which contributes to the progression of cystic growth, renal fibrosis, increased systemic vascular resistance, and the retention of sodium (Halvorson, Bremmer, & Jacobs, 2010, p. 74). The loss of renal parenchyma and the inability to function nephrons that result from this process is irreversible. The abnormal function of polycystin 1 and 2 also plays a role in vascular dysregulation and increased levels of intracellular calcium and smooth muscle cell infiltration and extracellular matrix defects and contribute to the sizes and structures of PKD 1 and 2 (Bataille, Berland, Fontes, & Burty, 2011, p. 1). During genetic testing, DNA linkage analysis and sequencing/mapping are utilized for the diagnosis of ADPKD (Pei, 2011, p. 19). Once an adult is diagnosed with PKD, it is easier to isolate the genetic mutation and test younger siblings for the same anomalies. ADPKD manifests as fluid-filled cystic dilation of renal tubules. Chang and Ong (2013) state that “renal cysts may be considered as a combination from abnormal cell proliferation, fluid secretion, and intrarenal inflammation results in kidney enlargement and interstitial fibrosis” (p. 524). Current research suggests that the primary reason behind renal cyst formation in both ADPKD and ARPKD is related to defects in cilia-mediated signals (Halvorson, Bremmer, and Jacobs, 2010). The gene products, polycystin 1 and 2, that are affected in ADPKD, regulate the growth of the epithelia in the renal tubules. The genetic mutations associated with PKD affect the formation of the epithelial cells and result in the formation of cysts. The presence of cilia is found on almost all surfaces of nephron cells, which means the cysts have the potential to form on these areas as well. In PKD, the function of the primary cilium is impaired, resulting in the disruption of a number of intracellular signaling cascades that produce decreased motility of the epithelium, increased cell division, increased apoptosis, and loss of regenerative capacity” (Halvorson, Bremmer, & Jacobs, 2010, p. 70). ADPKD and ARPKD present with defects, intracellular and extracellular calcium homeostasis is affected which promotes "alterations of endotheliodependent relaxant and increased systemic blood pressures” (Halvorson, Bremmer, & Jacobs, 2010, p. 70).

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, Gitomer, 2014, p. 177). However, renal failure is an inevitability in PKD, as a primary care provider it is important to educate PKD patients to monitor their blood pressure, including smoking cessation if possible, and to minimize the risk of developing complications that may arise, and diet recommendations. Efforts must be made to promote and maintain the cardiovascular system as well, including smoking cessation if possible, reducing alcohol intake, and appropriate exercise routines. If it is determined that the patient has PKD, it may be necessary to fine-tune medication choices that are made on his or her siblings as well as his or her children if applicable. Renal care is not always easy, and how quickly a patient gets to that point is up to him or her and their primary care provider.

Implications for Nursing Practice

As a primary health care provider it is important to obtain a detailed family history, genetic testing is important for evaluation of the patient, including signs, symptoms, social behaviors, diet, and any medication care provided to PKD patients is education; key topics include exercise, diet, medication compliance, and disease progression. Adequate exercise and an appropriate diet are important components in any disease process. Adherence to the diet is crucial. There should be a diet that is low in cholesterol to reduce cardiovascular risks, as well as a diet that is rich in fruits and vegetables, which will be helpful in the influx of nutrition to the kidneys. However, protein intake should be low in PKD patients to prevent blood pressure, if applicable. Once PKD has progressed to ESRD, patients will likely require dialysis and/or a renal transplant. However, over the last few years, there has been significant clinical trials with regards to slowing the progression of PKD have made advancements in this field (Sitomer, 2014, p. 177).

Conclusion

In the grand scheme of diseases, PKD is considered rare. However, once it is diagnosed the complications that can/cannot 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 educated on the complications that may arise, and diet recommendations. Efforts must be made to promote and maintain the cardiovascular system as well, including smoking cessation if possible, reducing alcohol intake, and appropriate exercise routines. If it is determined that the patient has ADPKD, it may be necessary to fine-tune medication choices that are made on his or her siblings as well as his or her children if applicable. Renal care is not always easy, and how quickly a patient gets to that point is up to him or her and their primary care provider.

References


