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Polycystic Kidney Disease

Caitlin Byrne
byrne1@otterbein.edu

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Caitlin Byrne, BSN-RN
Otterbein University, Westerville, Ohio

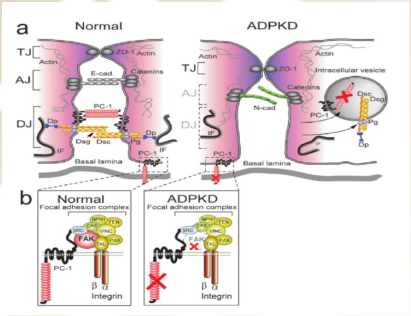
Introduction

Autosomal dominant polycystic kidney disease (ADPKD) and autosomal recessive kidney disease (ARPKD) are two forms of polycystic kidney disease (PKD) and are leading causes of end stage renal disease (ESRD) (Silverman, 2015). PKD can affect both children and adults and is one of the most common hereditary disorders (Silverman, 2015). PKD can lead to other medical complications like hypertension, cardiac disease, and cerebral aneurysm (Silverman, 2015). In PKD there is formation of renal cyst, though cyst can also form on the liver, pancreas, spleen, thyroid, arteries and brain (Silverman, 2015). There is no cure currently for PKD, though treatments for symptoms, secondary medical conditions, undergoing dialysis and kidney transplants are used to manage the disease (Silverman, 2015).

This author currently works at Riverside Methodist hospital on a stroke unit, where PKD is an uncommon diagnosis. Recently a close family friend received the diagnosis of PKD and had to undergo a kidney transplant. With this friend's diagnosis and lack of exposure to patients with PKD, this author chose PKD to become more familiar, gain knowledge and better care for patients with PKD.

Presentation of case or process

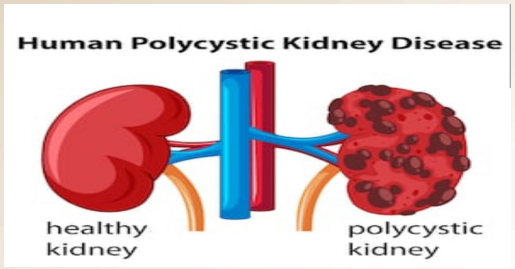
Sam a 42 year old female is concerned with her diagnosis of PKD. Sam's mother and older sister both have PKD and have each received kidney transplants. Due to family genetics, Sam had genetic testing and an ultrasound performed at the age of 18 to determine if she also had PKD, in which cysts were found to confirm her diagnosis. Sam has been symptom free until four months ago in which she has had multiple UTI's and severe flank pain. Sam went to see her PCP, where labs were drawn and another ultrasound performed. Sams creatinine level came back 7mg/dL (normal 0.6-1.2 mg/dL (McCance & Huether, 2018). Along with multiple cyst and an enlarged kidney found on her renal ultrasound. Sam began at home dialysis and was placed on the the kidney transplant list. Sam was also monitored for other complications such as hypertension and cardiac disease, she had frequent visits to her PCP and nephrologist to monitor her kidney function. Sam finally received a kidney transplant, she is now off dialysis and her creatinine is normal.



Bukanov, N.O. (2008, March). [Digital image]. Retrieved July 25, 2019, from https://images.app.goo.gl/Qoqppmw5AK8hvGYdA

Underlying Pathophysiology

•The gene products, polycystin-1 and polycystin-2, encoded on the genes PKD-1 and PKD-2, regulate growth and mechanosensory actions in the renal tubular cilia (Potts & Mousa, 2017).
•Mutations of gene PKD-1 occur on chromosome 16 and PKD-2 on chromosome 4 (Silverman, 2015).
•Mutations cause defects in epithelial cells and cilium leading to cyst formation, obstruction, and destruction of renal parenchyma, interstitial fibrosis, and loss of functional nephrons (McCance & Huether, 2018).
•Most mutations of these genes are caused by nonsense, frameshift, or splice in site alterations (Silverman, 2015).



[Shutterstock Inc.] (n.d). Retrieved July 25, 2019, from https://images.app.goo.gl/tEL99sNVUByg4U7P7

Significance of underlying pathophysiology

•PKD is an inherited disease and makes up for 8-10% of Population with ESRD (Silverman, 2015).
•PKD is a systemic disorder that causes irreversible kidney function, and can cause cyst formation in the liver, pancreas, brain, and arteries (Silverman, 2015).
•Cyst can cause urinary tract infections, flank pain, lead to ESRD and if external cyst exist serious complications such as ruptured cerebral aneurysm can occur (Silverman, 2015).
• Eventually the disease will progress that renal replacement therapy of dialysis is needed and kidney tranplation (Silverman, 2015).

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Signs and Symptoms.

• very few symptoms-some people experience no symptoms at all
•Abdmoinal pain/flank pain is most common
•Hypertension
•Palpable kidneys
•Hematuria
•Frequent UTI's

Nursing Implications

•Obtain a complete medical history and full body assessment
•Encourage low-salt diet, high water intake, and exercise (Tran et al., 2017).
•Monitor blood pressure and provide medications like ACE inhibitors for blood pressure management (Tran et al., 2017).
•Symptom management / pain control including antibiotics for UTI's, and bed rest for gross hematuria (Silverman, 2015).
•Close monitoring, lab draws, and urinalysis for kidney function.
•Provide education/ preparation for renal replacement therapy.
•Provide genetic counseling for patients with children.

Conclusion

PKD is a serious genetic disease and a leading cause of ESRD (Potts & Mousa, 2017). Providing supportive care and symptom management is extremely important throughout the progression of the disease. Genetic counseling for families with PKD can help identify and prepare for the progression and complications PKD entails. Medications, renal replacement therapies, and kidney transplants help slow disease progression and provide disease treatment, though no cure currently exists. Throughout these treaments it is important for health care provides to be supportive and encouragement to all PKD patients.

PKD

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