Short Communication

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Journal of Renal Medicine

2024
Vol.7 No.4:31

Genetic Distinctions between Autosomal Dominant and Autosomal Recessive Polycystic Kidney Disease

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Received date: June 10, 2024, Manuscript No. IPJRM-24-19498; **Editor assigned date:** June 13, 2024, PreQC No. IPJRM-24-19498 (PQ); **Reviewed date:** June 27, 2024, QC No. IPJRM-24-19498; **Revised date:** July 04, 2024, Manuscript No. IPJRM-24-19498 (R); **Published date:** July 11, 2024, DOI: 10.36648/ipjrm.7.4.31

Citation: Kalie D (2024) Genetic Distinctions between Autosomal Dominant and Autosomal Recessive Polycystic Kidney Disease. J Ren Med Vol.7 No.4: 31.

Description

Polycystic Kidney Disease (PKD), which is inherited, is characterized by the formation of numerous kidney cysts. These cysts, which are fluid-filled sacs, can impair kidney function over time, leading to various complications. Polycystic kidney disease is a complex genetic disorder that can have significant consequences for general health and renal health [1]. As though there is currently no cure for PKD, early diagnosis and proper administration might aid in delaying the development of kidney damage and improve quality of life for affected individuals. By raising awareness, promoting research, and advocating for better access to care, we can work towards better understanding and management of this challenging condition [2].

Types of polycystic kidney disease

PKD is primarily caused by genetic mutations that are inherited from one or both parents. Two main types of PKD exist Autosomal Dominant Polycystic Kidney Disease (ADPKD) and Autosomal Recessive Polycystic Kidney Disease (ARPKD). ADPKD is caused by mutations in the *PKD1* or *PKD2* genes, while ARPKD is caused by mutations in the *PKHD1* gene [3]. These genetic mutations lead to abnormal development and proliferation of kidney tubules, resulting in the formation of cysts. Side effects of PKD might differ relying upon the sort and seriousness of the sickness. Common symptoms include abdominal pain, flank pain, blood in the urine, hypertension, and recurrent Urinary Tract Infections (UTIs). As the cysts grow and multiply, they can cause progressive kidney damage, leading to Chronic Kidney Disease (CKD) and eventual kidney failure [4]. Other potential complications of PKD include kidney stones, liver cysts, and aneurysms in the brain. Diagnosis of PKD typically involves a combination of medical history evaluation, physical examination, imaging tests, and genetic testing. Imaging studies such as ultrasound, CT scan, or MRI can visualize the presence of cysts in the kidneys and help confirm the diagnosis Individuals suspected of having a genetic form of PKD or those with a family history of the disease may benefit from genetic testing [5]. Currently, there is no cure for PKD, but treatment aims to manage symptoms, slow the progression of kidney damage, and prevent complications. Pain management strategies may include over-the-counter or prescription pain medications to alleviate abdominal or flank pain. In cases of dialysis, kidney transplantation, or severe kidney failure might be necessary to replace lost kidney function [6].

Management of polycystic kidney disease

Changing one's way of life can help manage PKD and lower the likelihood of complications. Maintaining a healthy diet low in sodium and protein can help manage blood pressure and slow the progression of kidney disease [7]. Regular exercise, weight management and the cessation of smoking are also significant for overall kidney health. Close monitoring of kidney function through regular check-ups and laboratory tests is essential for early detection of complications and timely intervention. Ongoing research efforts are focused on understanding the underlying mechanisms of PKD and developing targeted therapies to halt or reverse disease progression [8]. Clinical trials are underway to evaluate the efficacy of new medications and treatment approaches for PKD. Advances in genetic testing technologies hold potential for early detection of PKD and personalized treatment strategies based on individual genetic profiles. Collaborative efforts between researchers, healthcare providers, and patient advocacy groups are vital for advancing PKD research and improving patient outcomes. Ongoing research efforts are aimed at better understanding the underlying mechanisms of PKD and developing targeted therapies [9]. Clinical trials are consider new treatment approaches, while advances in genetic testing hold potential for early detection and personalized treatment strategies. By raising awareness, by promoting greater access to treatment and funding research, we may strive toward better management and results for individuals living with PKD. Medications like ACE inhibitors or ARBs may be prescribed to control hypertension and protect kidney function, while pain management strategies help alleviate discomfort [10].

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